

Supplementary Appendix

This appendix has been provided by the authors to give readers additional information about their work.

Supplement to: Duncavage EJ, Jacoby MA, Chang GS, et al. Mutation clearance after transplantation for myelodysplastic syndrome. *N Engl J Med* 2018;379:1028-41. DOI: [10.1056/NEJMoa1804714](https://doi.org/10.1056/NEJMoa1804714)

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Supplement to: Duncavage EJ, Jacoby MA, Chang GS, *et al.* Mutation clearance after transplant for myelodysplastic syndrome.

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Supplementary Methods

A. Study Design

All consecutive patients with adequate samples that underwent allogeneic hematopoietic cell transplant (alloHCT), from any source, at Washington University from 11/2002-05/2015 were included in the study. Adequate sample was defined as having sufficient DNA available from the bone marrow and skin pre-alloHCT, and bone marrow on at least day 30 after transplant for sequencing studies. In 58 patients, an additional bone marrow biopsy was performed within a median of 23 days prior to transplant (range, 6-61), with no intervening treatment between biopsy and transplant, and could be used to measure tumor burden at the time of transplant (i.e., pre-transplant sample). In addition, the initial banking sample for 9 patients also met the definition of the pre-transplant time-point (i.e., biopsy was performed within 60 days prior to transplant and had no intervening treatment) and were annotated in Supplementary Table S2. A bone marrow sample was available for 58 patients on day 100 post-alloHCT. The study was approved by the institutional review board at Washington University in St. Louis and was conducted in accordance with the provisions of the Declaration of Helsinki. All of the patients who were enrolled provided written informed consent that explicitly included genome sequencing and data sharing with qualified investigators.

Disease progression was defined prior to sequencing as follows: the presence of $\geq 5\%$ myeloblasts in the marrow or evidence of extramedullary disease; loss of donor chimerism or re-emergence of pre-transplant morphologic abnormalities prompting any intervention by the treating physician (such as withdrawal of immunosuppression, donor lymphocyte infusion, or chemotherapy); or re-emergence of pre-transplant cytogenetic abnormalities. The reviewers were blinded to the results of the sequencing studies. Database lock was 4/16/2017. For progression-free survival, in the absence of progression, and for overall survival, patients were censored at the time they were last known to be alive. Morphologic review for all time points was provided by a board certified hematopathologist. In most cases the blast count was determined by morphological assessment of the Wright Giemsa stained bone marrow aspirate

smears, however in cases where the aspirate was hemodilute or of low technical quality, the blast count was estimated by CD34 and/or CD117 immunohistochemically staining of the bone marrow core depending on the blast phenotype detected by flow cytometry.

Eight patients who had previously undergone ultra-sensitive, error-corrected sequencing prior to transplant and at day 30 post-alloHCT on a previous study were included in the outcome analyses.¹ Their UPNs were as follows: 145094, 147457, 280837, 368402, 435866, 499258, 624702, and 829970.

B. Enhanced Exome Sequencing and Somatic Variant Identification at Initial Banking

We sequenced the exome of 90 patients with a history of MDS, described above, at initial banking. HiSeq2500 sequencing systems (Illumina, CA) were used to sequence DNA, captured using SeqCap EZ Exome v3.0 Kit (Roche, CA) with additional spike-in probes targeted to 285 genes recurrently mutated (or RMGs) in myeloid malignancies, or enhanced exome sequencing (EES).² Exome sequencing data were aligned to the human reference sequence build GRCh37-lite-build37 using bwa³ version 0.5.9 with “-t 4 -q 5” parameters, then merged and de-duplicated using picard version 1.46 (<https://broadinstitute.github.io/picard/>).

Somatic variants in tumor samples were called against normal skin samples we sequenced in EES from the same patient. In detail, single nucleotide variation (SNV) was detected using the union of four variant callers: 1) samtools⁴ version r982 (params: mpileup - BuDS) filtered by var-filter-snv v1 and false-positive-vcf v1 (params: --max-mm-qualsum-diff 100 --bam-readcount-version 0.4 --bam-readcount-min-base-quality 15), then intersected with Somatic Sniper version 1.0.4⁵ (params: -F vcf -G -L -q 1 -Q 15) and processed through false-positive filter v1 (params: --bam-readcount-version 0.4 --bam-readcount-min-base-quality 15 --min-mapping-quality 40 --min-somatic-score 40), 2) VarScan⁶ version 2.3.6 (params: --nobaq --version r982) filtered by varscan-high-confidence filter v1 and processed through false-positive filter v1 (params: --bam-readcount-version 0.4 --bam-readcount-min-base-quality 15), 3) Strelka⁷ version 1.0.11 (params: isSkipDepthFilters = 1), and 4) MuTect version 1.1.4 (--number-of-chunks

50) with COSMIC v54.⁸

For insertions and deletions (INDELs), we used the union of four variant callers: 1) GATK⁹ somatic-indel version 5336, 2) pindel¹⁰ version 0.5 filtered with pindel-somatic-calls v1 and VAF filters v1 (params: --variant-freq-cutoff=0.08), and with pindel-read-support filter v1, 3) VarScan⁶ version 2.3.6 (params: --nobaq --version r982) filtered by varscan-high-confidence-indel v1 and 4) Strelka⁷ version 1.0.11 (params: isSkipDepthFilters = 1).

SNVs and INDELs were further filtered by removing artifacts found in a panel of 905 normal exomes, removing sites that exceeded 1% frequency in the Exome Aggregation Consortium (ExAC) collection,¹¹ and then using a Bayesian classifier (<https://github.com/genome/genome/blob/master/lib/perl/Genome/Model/Tools/Validation/IdentifyOutliers.pm>) and retaining variants classified as somatic with a binomial log-likelihood of at least 3 (params: --llr-cutoff 3). As a result, germline mutations, low-quality mutations, sequencing artifacts, and ambiguous variant calls owing to paralogous DNA sequences and unstable reference sequence contigs were removed.

A total 3,811 somatic variants were called from 90 patients at initial banking in the somatic variation discovery pipeline, described above, including 3,414 SNVs, 257 DELs, and 140 INSS. An average of 42 variant were called per patient (minimum 3 to maximum 644 variants called). Exome sequencing data was deposited in the dbGAP database (phs000159.v9).

C. Ultra-deep error-corrected sequencing

Probes for ultra-deep error-corrected sequencing were designed to target all somatic mutations detected by enhanced exome sequencing (or EES) using the Agilent SureDesign web interface. Genomic DNA was first digested using a mixture of restriction endonucleases found in the Agilent HaloplexHS kit (Agilent Technologies, Santa Clara, CA). Probes (designed to cover both DNA strands), 10bp degenerate barcodes, and sample-specific indexes were synthesized by Agilent Technologies. Following a 16 hour hybridization and magnetic bead enrichment, libraries were amplified for 23 PCR cycles using primers with tailed Illumina sequencing motifs.

Purified libraries were then pooled and sequenced 16 per lane (initial samples and skin) or 8 per lane (Pre, Day 30, and D100) time points on a HiSeq 4000 instrument. Pools were designed to minimize the effect of potential 'index swapping' by including only unique UPNs in each lane; samples from initial time points (high expected VAFs) were excluded from pools with day 30 and day 100 follow-up samples (low expected VAFs). Further, follow-up samples were pooled to avoid including samples in the same pool with the same recurrent somatic mutations when possible (e.g., only one sample with *U2AF1* S34F per pool). Samples were sequenced on an Illumina HiSeq 4000 instrument using 2x150bp reads (Illumina, Carlsbad CA).

Barcoded FASTQ data was demultiplexed using a custom UMI-aware script that merges degenerate barcode information to FASTQ files. Data was then aligned to build GRCh37-lite-build37 using *bwa mem* (version 7.9.a) with default parameters. Aligned BAMs were then analyzed using the Barcrawler pipeline (Duncavage and Abel, <https://github.com/abelhj/gatk>), a GATK locus walker implemented in Java, with the following parameters *-mmq 20 -mbq 20 -minCtBC 3 -dcov 1000000 -discardN 1 -minOffset 3 -maxNM 10*. The resulting list of variants were filtered to include only mutations present in at least 3 unique read families (each with a minimum of 3 members in which the variant was present in >90% of member reads). A background error-rate was further calculated for every targeted position by compiling the number of non-reference bases detected at that position across all cases minus the one with the mutation. For example in UPN 491309, a *TP53* p.Y243C mutation (chr17: 7577580 T->C) was detected with a VAF of 0.54% (80 variant read families out of 14,682 total). The remaining sequenced cases (all cases minus the index case) were then queried at that position and demonstrated the reference base, T, in 3,234,744 unique read families and the variant, C, in 9 total read families. A binomial distribution was then used to determine whether read counts of the observed mutation in the index case were statistically different from background. In the example, 80 out of 14,682 are statistically different from the background rate of the same mutations across all other cases (9 out of 3,234,744) with a P-value of <0.0001. P-value

threshold of <0.0001 was used for all binomial comparisons. Detected mutations with a P-value >0.0001 compared to the background error were excluded from the analysis. Probes were considered validated if the SNV detected by EES was present in a marrow prior to transplant with a VAF of $\geq 1\%$ and had a VAF of $\leq 10\%$ in the normal skin sample and had $\geq 10x$ unique coverage at either the day 30 or day 100 time points. A total of 2,517 SNVs out of 3,414 targeted SNVs passed filter. The majority of positions that failed to validate were due to poor coverage at skin and prior to transplant time points or SNPs that were originally classified as 'somatic' variants based on lower coverage EES data. Indel calls detected by EES were not evaluated by error-corrected sequencing.

There was a wide range in unique coverage obtained across targeted positions in day 30 and day 100 samples, likely due to differences in GC content and the number of probes that could be designed to each targeted region. Therefore, a conservative VAF threshold was chosen to minimize potential variant detection bias due to differences in coverage. A maximum VAF cut-off of 0.5% resulted in $>99\%$ of day 30 and day 100 samples having a unique coverage depth that would allow detection of mutations with a VAF of $\geq 0.5\%$. A single case (day 30, 717045) had lower unique coverage due to limited available DNA. To determine whether the choice of a 0.5% maximum VAF cut-off biased the results, we compared multiple VAF cut off thresholds (2.5%, 1%, 0.1%, and no cut-off [i.e., any detected mutation]) and found that day 30 sequencing results remained a strong predictor of progression (Supplemental Figures S5-S8).

Supplementary Results

A. Treatment with a hypomethylating agent prior to alloHCT is not associated with progression

We previously reported that pre-transplant therapy with azacitidine likely influenced the mutation spectrum (e.g., C to G transversions) and evolution of emergent subclones after alloHCT.¹ This remains a potential mechanism that may affect post-alloHCT progression. We

were not able to address whether putative hypomethylating agent-induced mutations influence progression in the current study because our sequencing strategy was not designed to identify emerging subclones. However, we did examine whether there was an association with therapy prior to transplant and progression in our cohort. When comparing patients who received a hypomethylating agent (e.g., azacitidine or decitabine) at any point pre-transplant versus no therapy, the hazard ratio for progression was 0.73 (95% confidence interval 0.41, 1.30; P=0.28). When compared to patients who received a hypomethylating agent at any point pre-transplant versus a therapy not containing a hypomethylating agent, the hazard ratio for progression was 0.94 (95% confidence interval 0.368, 2.40; P=0.90). While this analysis is limited by the use of non-uniform treatments, doses, and schedules, we did not observe an association with progression whether a patient received therapy prior to transplant, and if that therapy included a hypomethylating agent.

B. Post-alloHCT mutation VAF level is associated with progression

We performed an analysis using max VAF at day 30 post-alloHCT as a continuous variable (i.e., without thresholds) and it remained significantly associated with progression. For each increase of 1% in max VAF at day 30 post-alloHCT, the hazard of progression increases by about 4% (HR=1.04; 95% confidence interval, 1.02 to 1.05; P<0.001).

We also compared the cumulative incidence of progression for patients with a max VAF $\geq 0.5\%$ versus max VAF > 0 but $< 0.5\%$. We found that 18 patients had day 30 VAFs $< 0.5\%$ but $> 0\%$. Six of these 18 patients progressed with a median of 195 days between detection of a day 30 post-alloHCT mutation and progression. The hazard of progression is about 2.5 times greater in patients with day 30 max VAF $\geq 0.5\%$ versus those with detectable but lower max VAF (> 0 and $< 0.5\%$) with a 95% confidence interval of 1.02 to 6.07 (P=0.046).

Supplementary Figure 1. Number and Distribution of Tracked Mutations at the Initial and Pre-transplant Time Points. Panel A shows the distribution of the total number of single nucleotide variant (SNV) mutations validated by error-corrected sequencing at initial sampling for patients with (n=34) or without (n=51) progression or death at one year. Panel B shows the distribution of the total number of SNV mutations validated by error-corrected sequencing at pre-transplant for patients with (n=32) or without (n=35) progression or death at one year. Panel C shows the distribution of mutation VAFs at initial sampling. Panel D shows the distribution of mutation VAFs measured pre-transplant. In Panels A-D, UPN 147457, which had 482 mutations, was omitted for clarity. Nine patients were included in both the initial sampling and pre-transplant data because they had an initial banking sample that also met the definition of the pre-transplant time-point defined in the Supplementary Appendix. They are annotated in Supplementary Table S2.

Supplementary Figure 2. Sequencing Metrics. Panels A and B show box and whiskers plot of coverage depths of non-overlapping 228,531 regions of interest targeted in enhanced exome sequencing. The line represents the median coverage depth; boxes represent upper and lower quartiles; lines represent minimum and maximum coverage values. Bone marrow samples (Panel A) and skin (Panel B, normal DNA source) are denoted by their unique patient number (UPN). Mean tumor coverage of 239x in the exomes and 1,299x over recurrently mutated genes (or RMG) in myeloid malignancies. Panel C and D show sequencing coverage depths obtained for ultra-deep error-corrected sequencing of the skin (Panel C) and bone marrow at initial sampling (Panel D). The total mean tumor coverage was 19,450x and the unique barcode coverage passing filter was 2,450x. Panels E and F show sequencing coverage depths obtained for ultra-deep error corrected sequencing at day 30 (Panel E) and bone marrow and day 100 post-allogeneic hematopoietic cell transplant (Panel F). At day 30 the total mean tumor coverage was 60,930x and the unique barcode coverage passing filter was 2,559x. At day 100

the total mean tumor coverage was 36,705x and the unique barcode coverage passing filter was 3,475x. Panel G shows sequencing coverage depths obtained for ultra-deep error-corrected sequencing for the 58 patients with pre-allogeneic hematopoietic cell transplant samples. The total mean tumor coverage was 13,926x and the unique barcode coverage passing filter was 1,569x.

Supplementary Figure 3. Mutation Clearance of Selected Genes at Day 30 Post-Allogeneic Hematopoietic Cell Transplant.

VAF mutation clearances for recurrently mutated genes. 11 genes are shown, which were identified as mutated 4 or more times in this study including *ASXL1*, *DNMT3A*, *IDH2*, *NRAS*, *PTPN11*, *RUNX1*, *SF3B1*, *SRSF2*, *TET2*, *TP53*, and *U2AF1* (A-K). Shown are initial marrow mutation VAFs and day 30 VAFs of each gene and labeled by UPN and progression and overall day 30 maximum mutation VAF status. Plots on the right highlight mutations with VAFs < 5% at day 30 post-transplant, labeled by the same symbol and color of the patient.

Supplementary Figure 5. Association of Mutation Clearance at Day 30 post-alloHCT with Outcomes. Variant allele frequencies (VAFs) day 30 post-transplant were determined using error-corrected sequencing interrogating single nucleotide variant (SNV) mutations identified by enhanced exome sequencing of samples prior to transplant. Patients are grouped by the presence of at least one VAF $\geq 0.5\%$ (red lines) or all VAFs < 0.5% (blue lines). Risk of progression is shown in Panel A, progression free survival is shown in Panel B. Panels C and D show overall survival based on the presence of at least one VAF $\geq 0.5\%$ (Panel C) and according to whether the patient received reduced-intensity conditioning (RIC, solid lines) or myeloablative conditioning (MAC, dashed line) in Panel D.

Supplementary Figure 5. Association of Mutation Clearance at Day 100 Post-alloHCT with Outcomes. Variant allele frequencies (VAFs) day 100 post-transplant were determined using error-corrected sequencing interrogating single nucleotide variant (SNV) mutations identified by enhanced exome sequencing of samples prior to transplant. Patients are grouped by the presence of at least one VAF $\geq 0.5\%$ (red lines) or all VAFs $< 0.5\%$ (blue lines). Panels B, D, and F show outcomes according to whether the patient received reduced-intensity conditioning (RIC, solid lines) or myeloablative conditioning (MAC, dashed line). Risk of progression (Panels A and B), progression-free survival (Panels C and D), and overall survival (Panels E and F) are shown.

Supplementary Figure 6. Association of Mutation Clearance with Outcomes Using a Variant Allele Frequency Threshold of 0.1%. Variant allele frequencies (VAFs) day 30 post-transplant were determined using error-corrected sequencing interrogating single nucleotide variant (SNV) mutations identified by enhanced exome sequencing of samples prior to transplant. Patients are grouped by the presence of at least one VAF $\geq 0.1\%$ (red lines) or all VAFs $< 0.1\%$ (blue lines). Panels B, D, and F show outcomes according to whether the patient received reduced-intensity conditioning (RIC, solid lines) or myeloablative conditioning (MAC, dashed line). Risk of progression (Panels A and B), progression-free survival (Panels C and D), and overall survival (Panels E and F) are shown.

Supplementary Figure 7. Association of Mutation Clearance with Outcomes Using a Variant Allele Frequency Threshold of 1%. Variant allele frequencies (VAFs) day 30 post-transplant were determined using error-corrected sequencing interrogating single nucleotide variant (SNV) mutations identified by enhanced exome sequencing of samples prior to transplant. Patients are grouped by the presence of at least one VAF $\geq 1\%$ (red lines) or all VAFs $< 1\%$ (blue lines). Panels B, D, and F show outcomes according to whether the patient received reduced-intensity conditioning (RIC, solid lines) or myeloablative conditioning (MAC,

dashed line). Risk of progression (Panels A and B), progression-free survival (Panels C and D), and overall survival (Panels E and F) are shown.

Supplementary Figure 8. Association of Mutation Clearance with Outcomes Using a Variant Allele Frequency Threshold of 2.5%. Variant allele frequencies (VAFs) day 30 post-transplant were determined using error-corrected sequencing interrogating single nucleotide variant (SNV) mutations identified by enhanced exome sequencing of samples prior to transplant. Patients are grouped by the presence of at least one VAF $\geq 2.5\%$ (red lines) or all VAFs $< 2.5\%$ (blue lines). Panels B, D, and F show outcomes according to whether the patient received reduced-intensity conditioning (RIC, solid lines) or myeloablative conditioning (MAC, dashed line). Risk of progression (Panels A and B), progression-free survival (Panels C and D), and overall survival (Panels E and F) are shown.

Supplementary Figure 9. Association of Mutation Clearance with Outcomes Using any Detectable Variant Allele Frequency. Variant allele frequencies (VAFs) day 30 post-transplant were determined using error-corrected sequencing interrogating single nucleotide variant (SNV) mutations identified by enhanced exome sequencing of samples prior to transplant. Patients are grouped by the presence of at least one detectable VAF $> 0\%$ (red lines) or all VAFs $= 0\%$ (blue lines). Panels B, D, and F show outcomes according to whether the patient received reduced-intensity conditioning (RIC, solid lines) or myeloablative conditioning (MAC, dashed line). Risk of progression (Panels A and B), progression-free survival (Panels C and D), and overall survival (Panels E and F) are shown.

Supplementary Figure 10. Association of Mutation Clearance Determined by a 40 Gene-Panel. Only the variant allele frequencies (VAFs) of 40 genes recurrently mutated in myeloid malignancies were used to assess mutation clearance at day 30 post-transplant. VAFs were

determined using error-corrected sequencing interrogating single nucleotide variants identified by enhanced exome sequencing of samples prior to transplant. Panel A shows the cumulative incidence of progression in patients grouped by the presence of at least one VAF $\geq 0.5\%$ (red lines) or all VAFs $< 0.5\%$ (blue lines). Panel B show progression-free survival. Panels C and D show overall survival grouped by VAF (Panel C) and according to whether the patient received reduced-intensity conditioning (RIC, solid lines) or myeloablative conditioning (MAC, dashed line) in Panel D.

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Figure S1. Number and Distribution of Tracked Mutations at the Initial and Pre-transplant Time Points

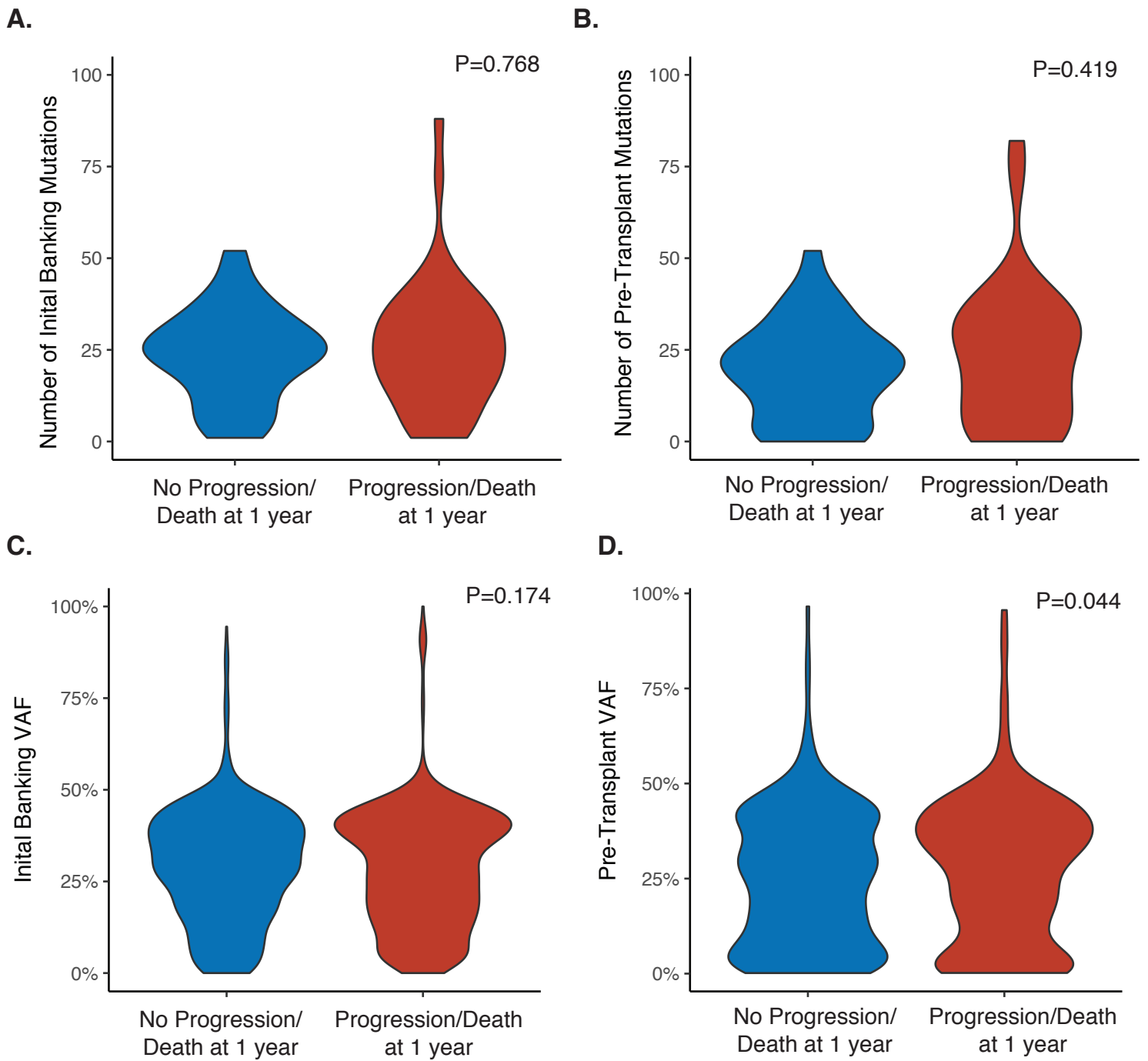
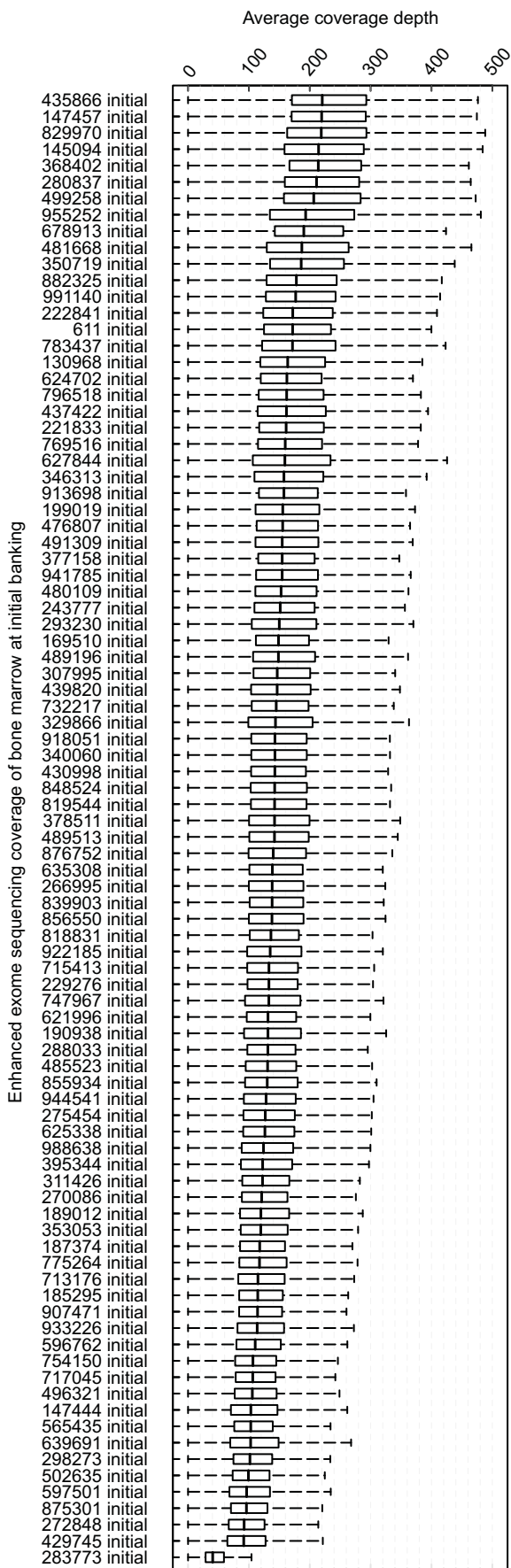


Figure S2. Sequencing Metrics

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B.

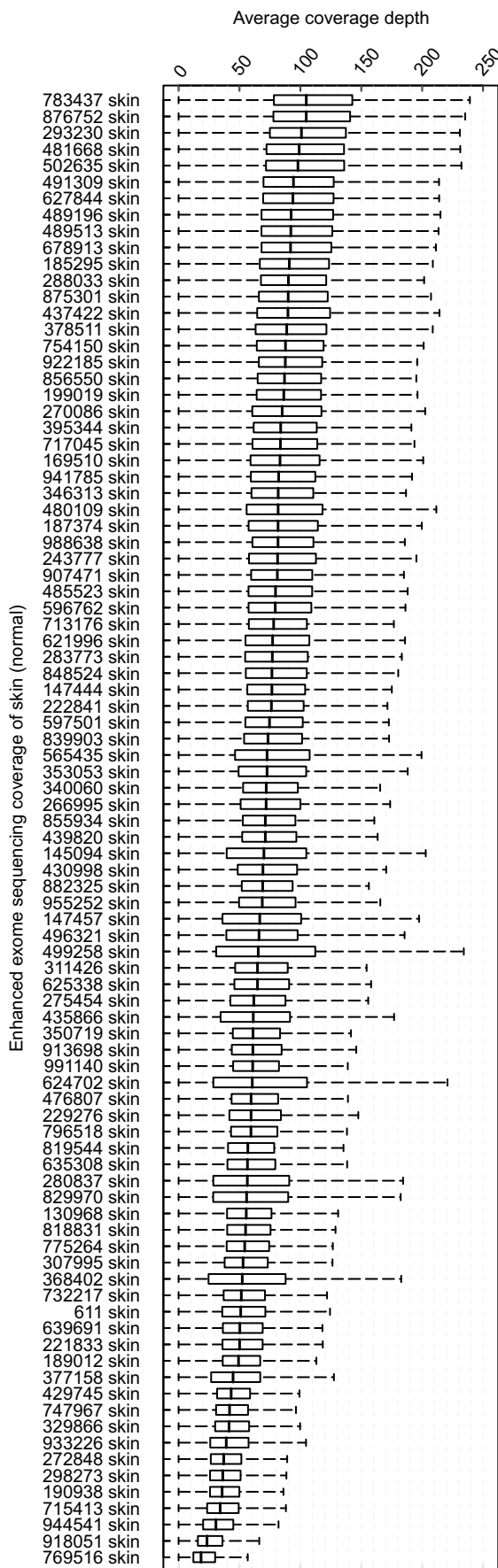
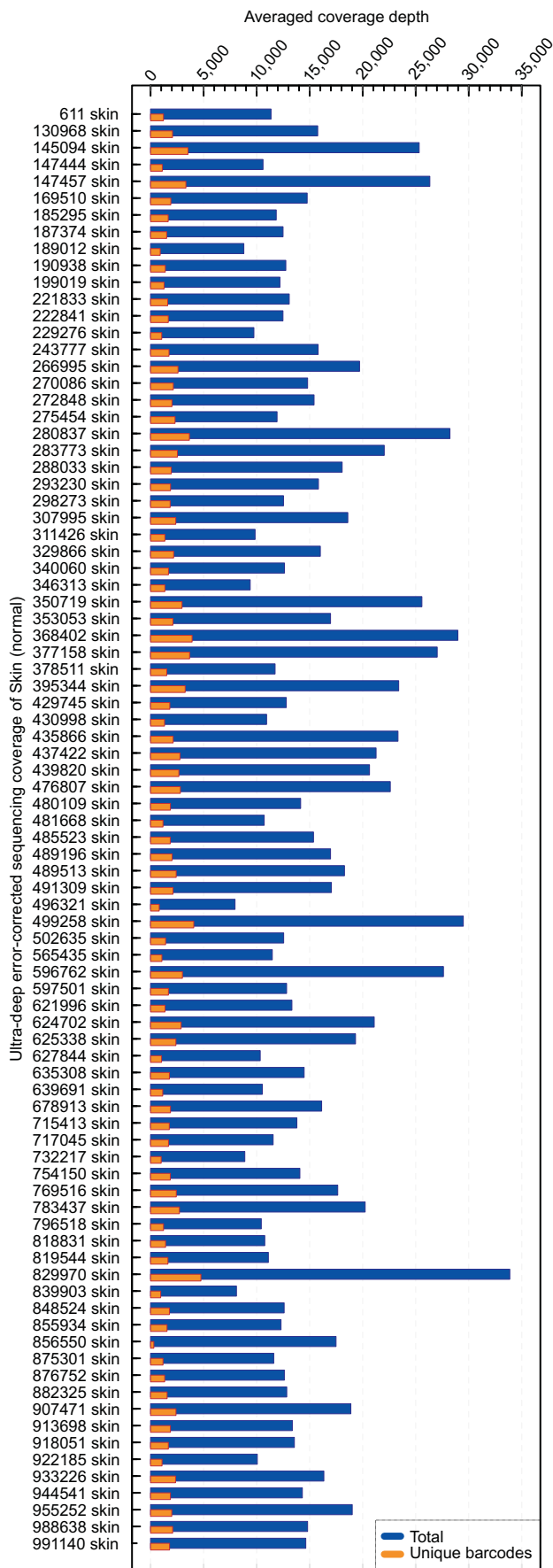


Figure S2. Sequencing Metrics

C.



D.

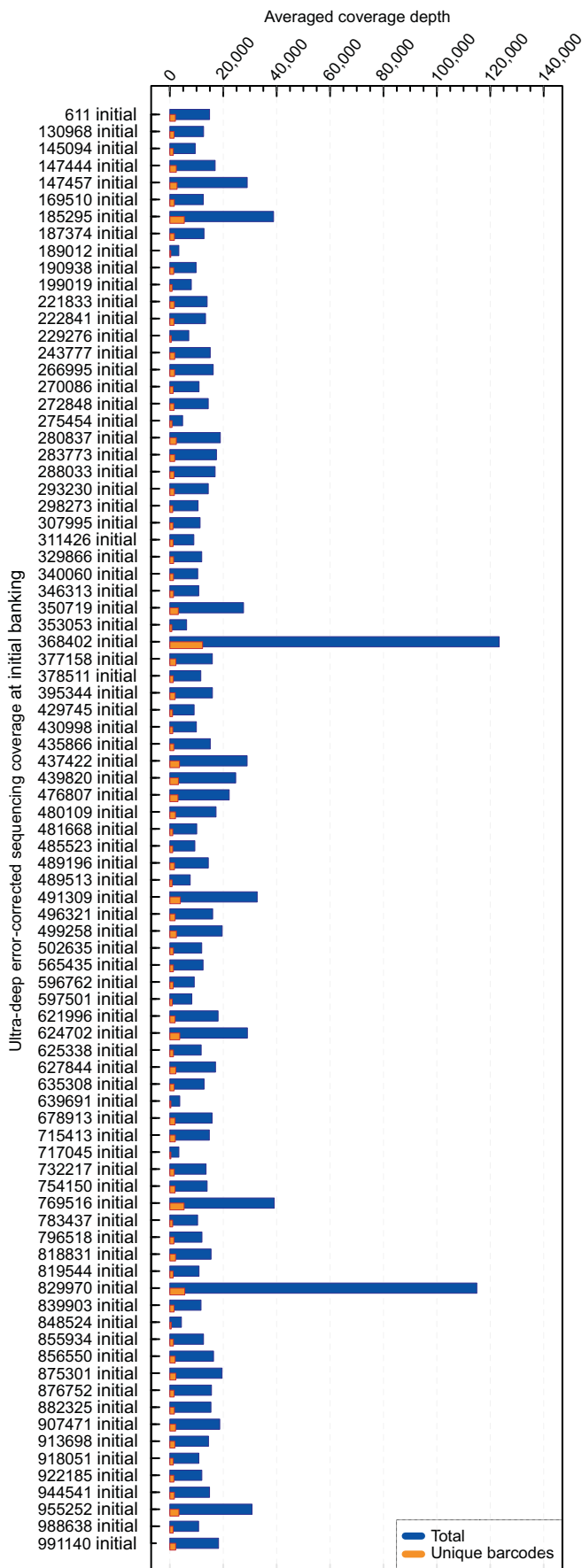
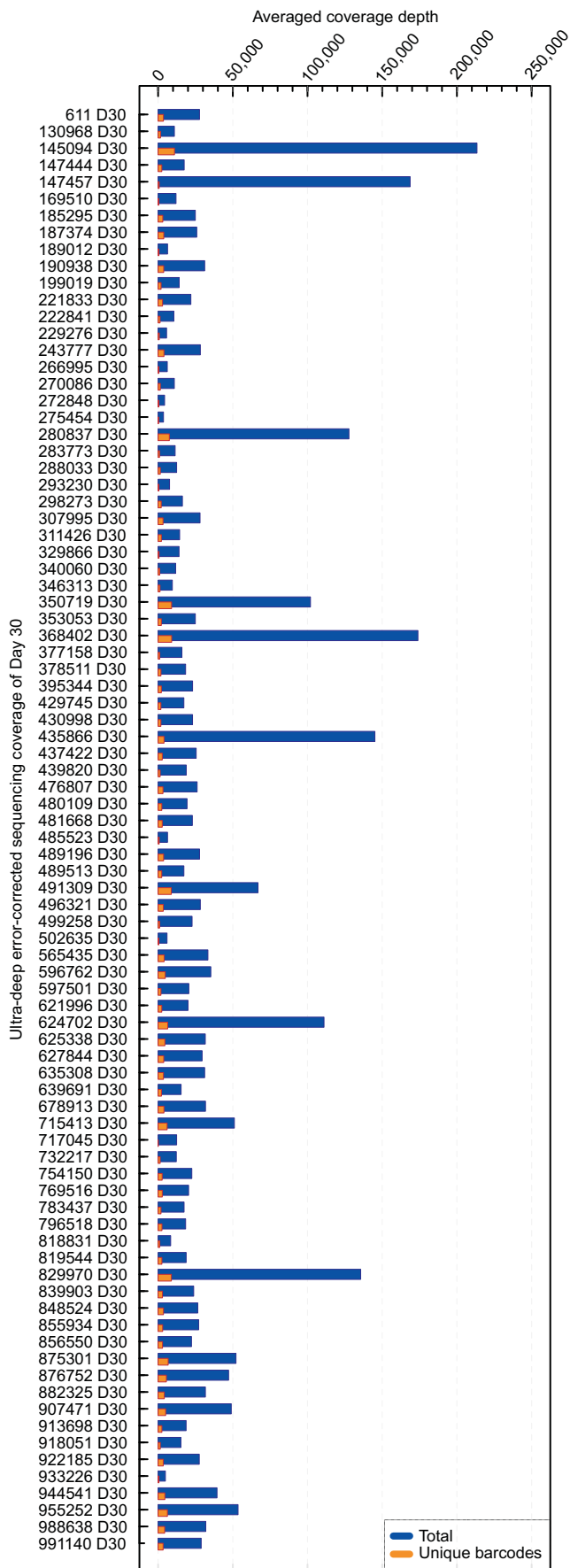


Figure S2. Sequencing Metrics

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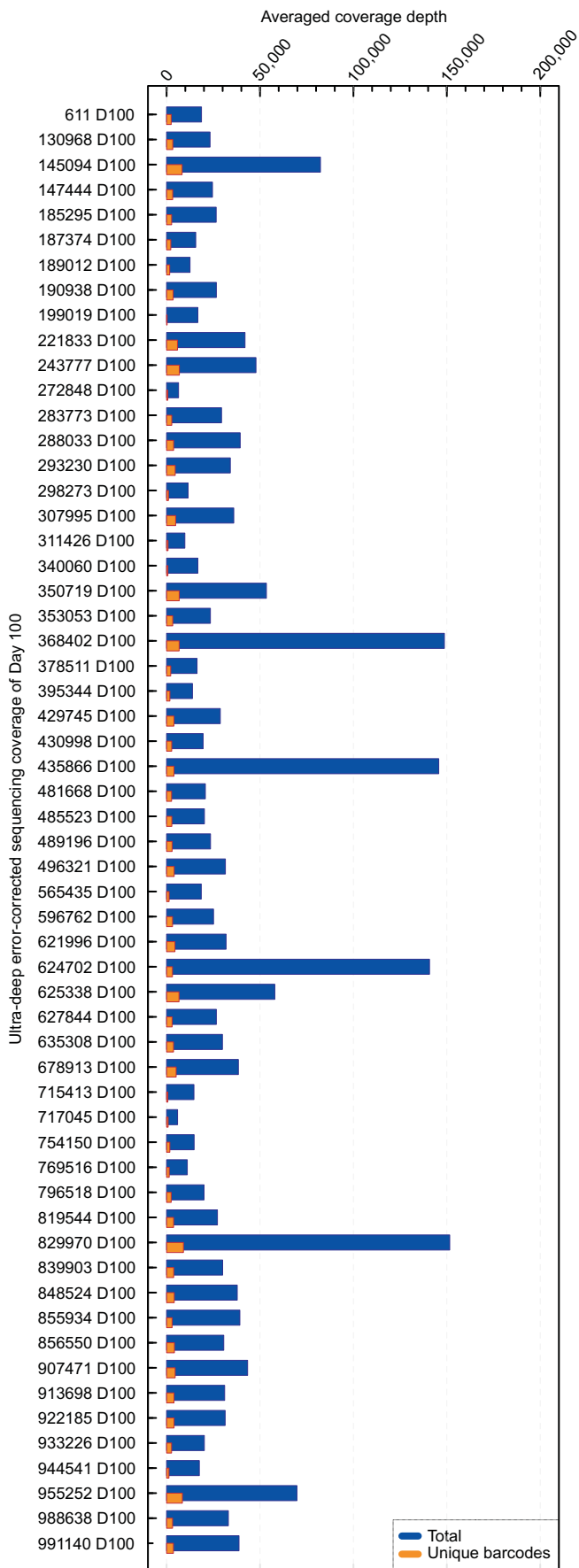


Figure S2. Sequencing Metrics

G.

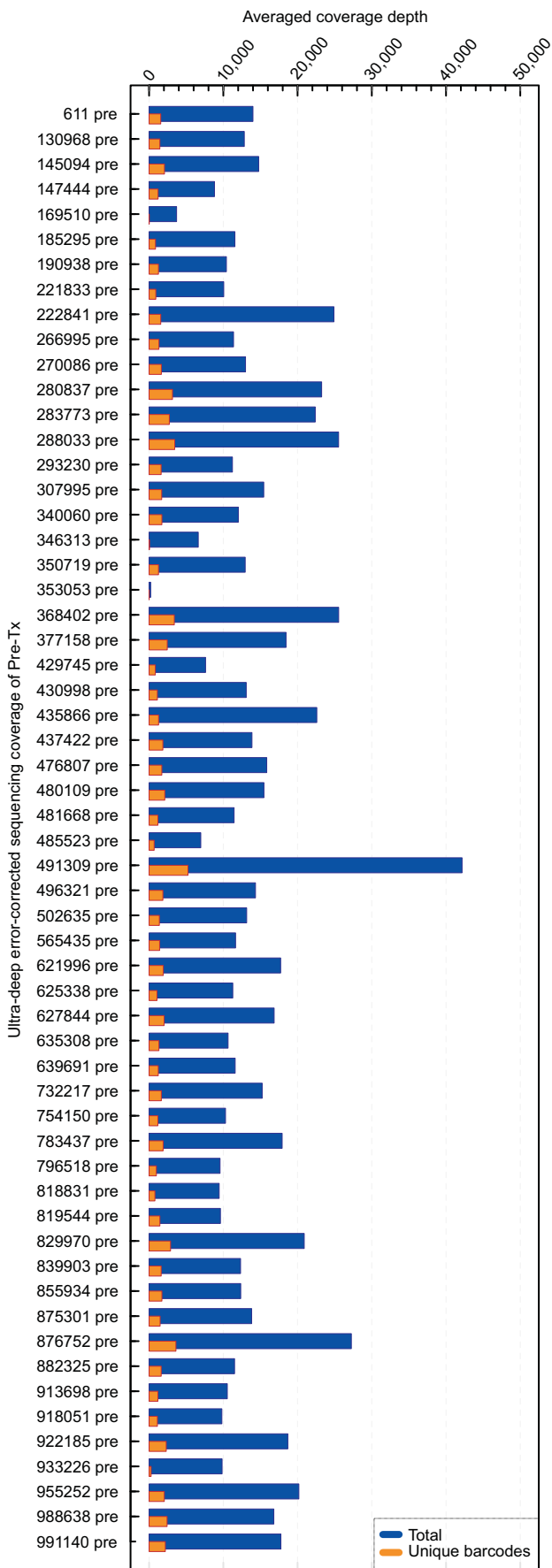
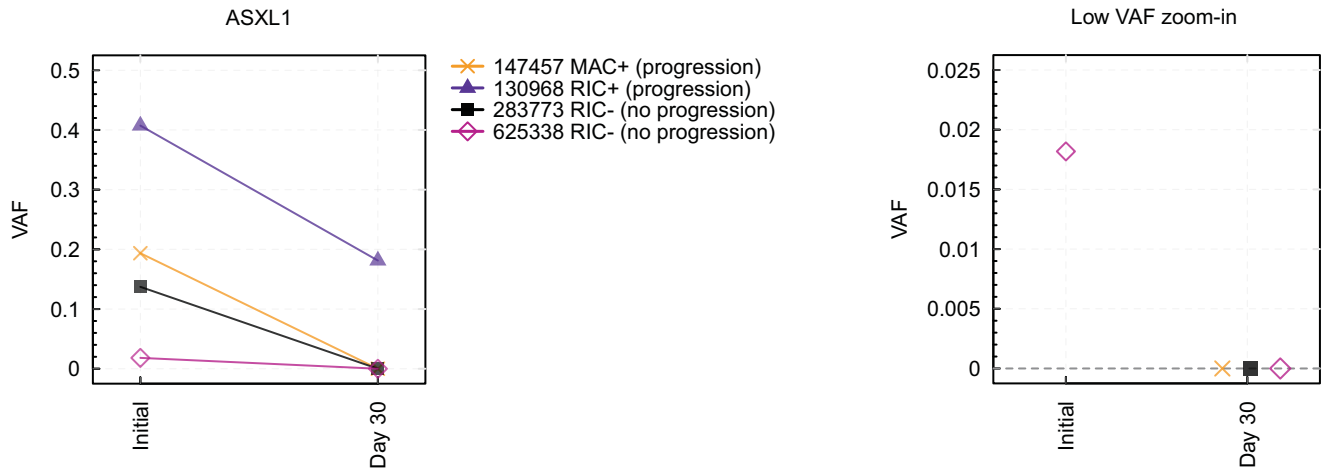
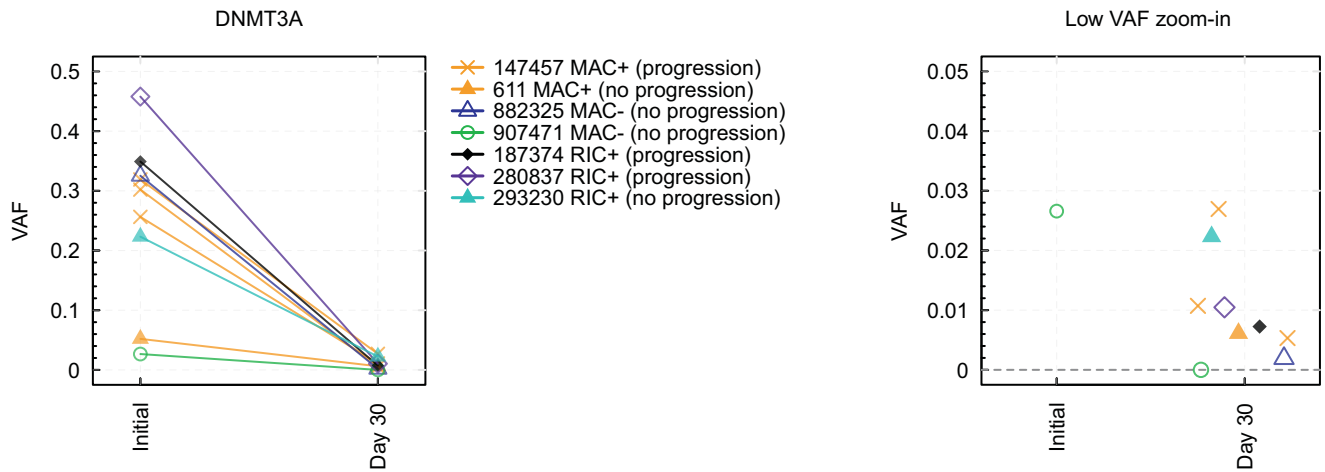


Figure S3. Mutation Clearance of Selected Genes at Day 30 Post-Allogeneic Hematopoietic Cell Transplant

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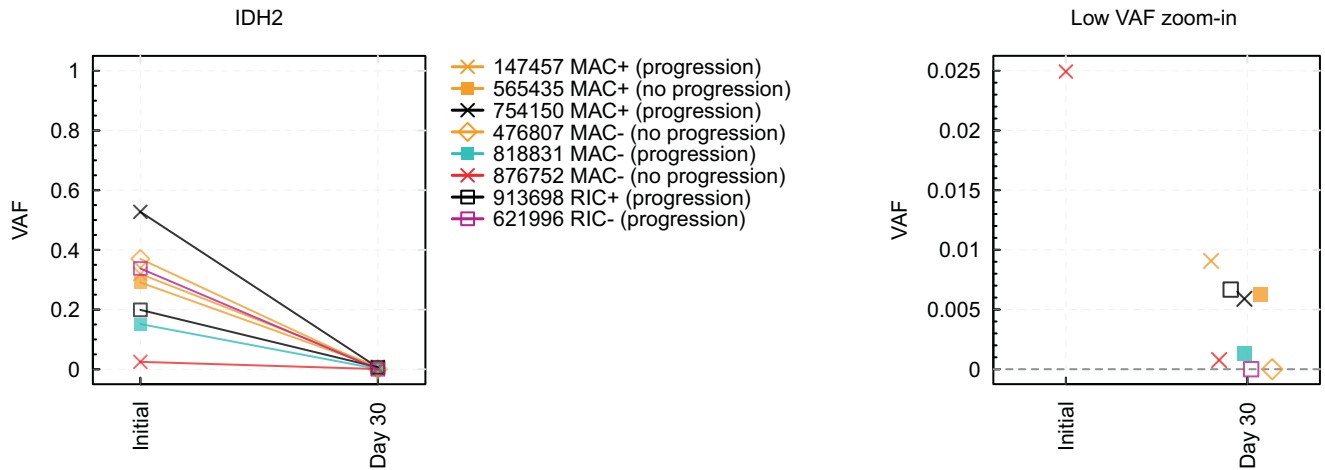
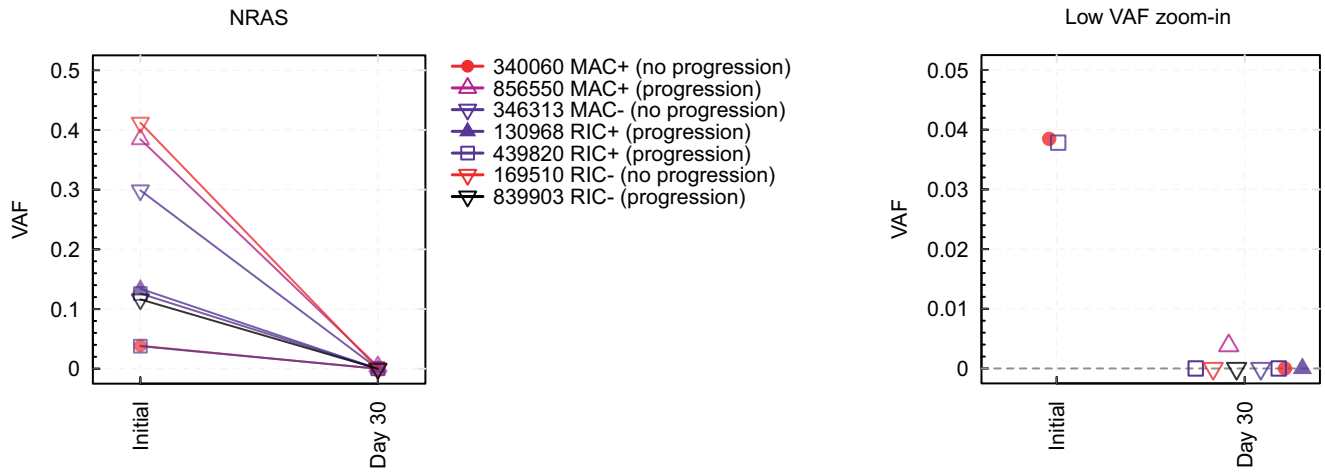
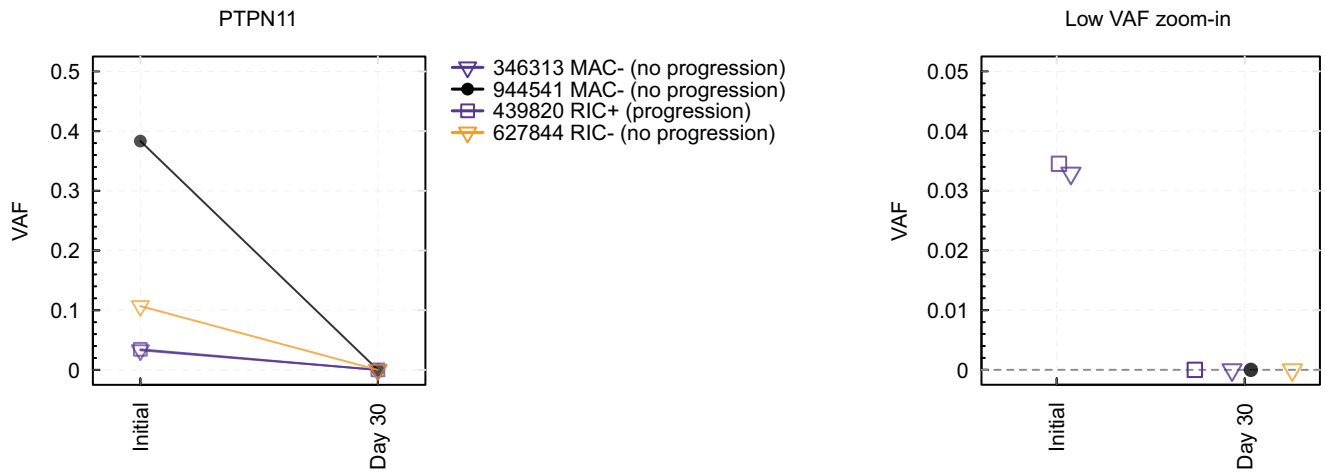


Figure S3. Mutation Clearance of Selected Genes at Day 30 Post-Allogeneic Hematopoietic Cell Transplant

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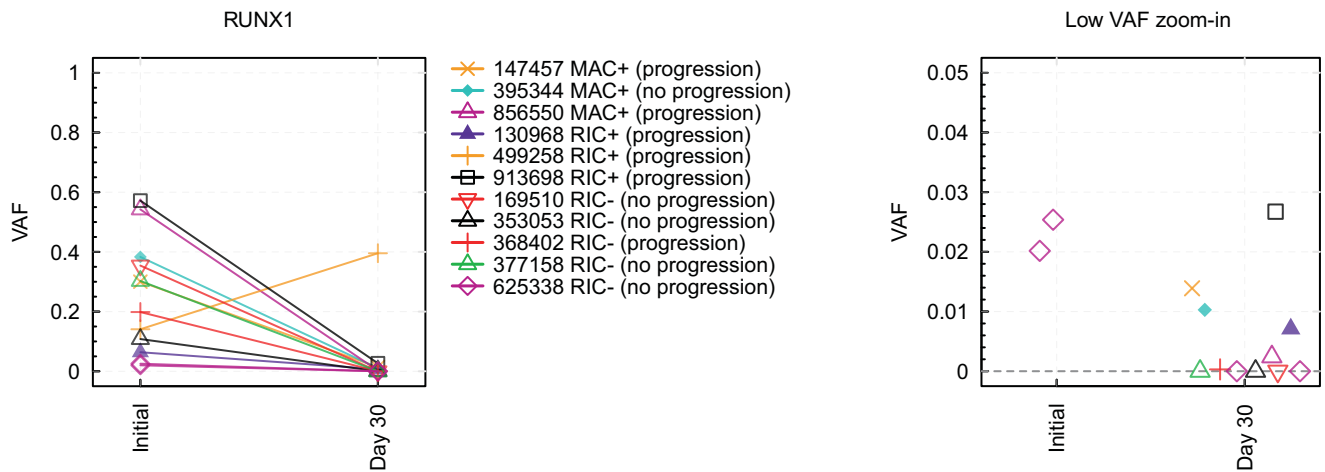
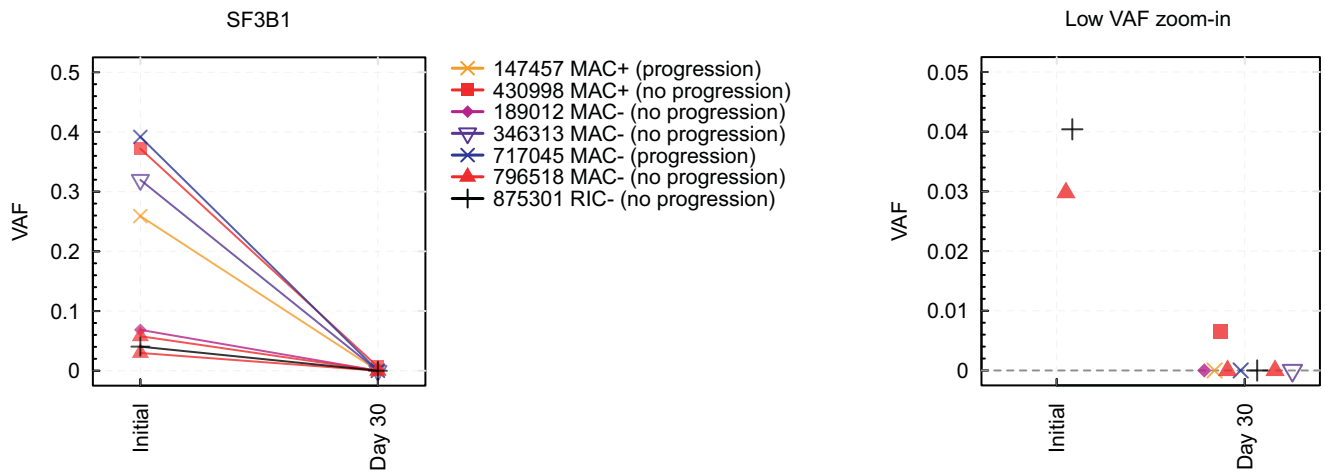
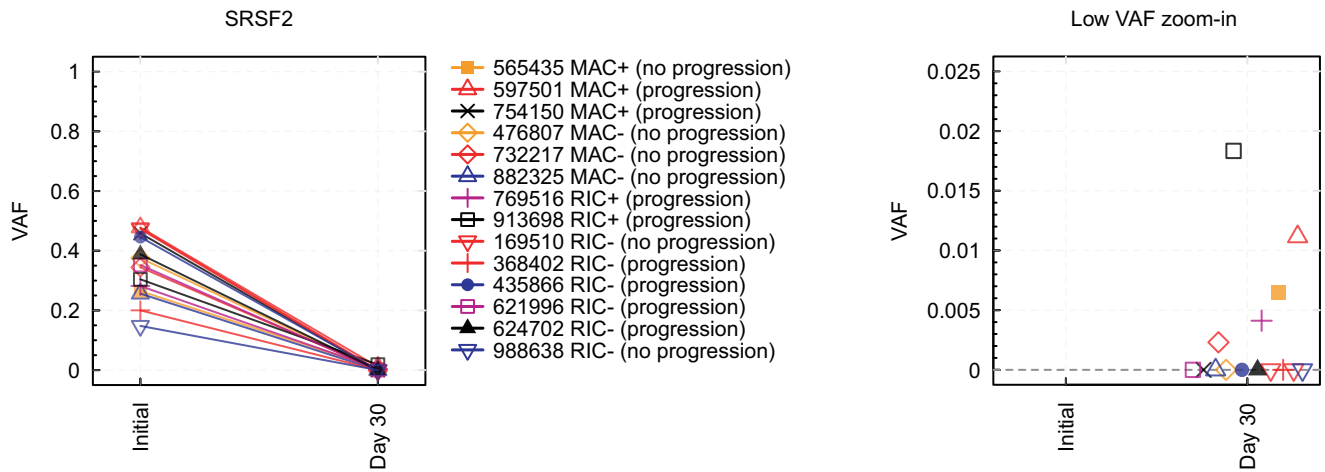


Figure S3. Mutation Clearance of Selected Genes at Day 30 Post-Allogeneic Hematopoietic Cell Transplant

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H.



I.

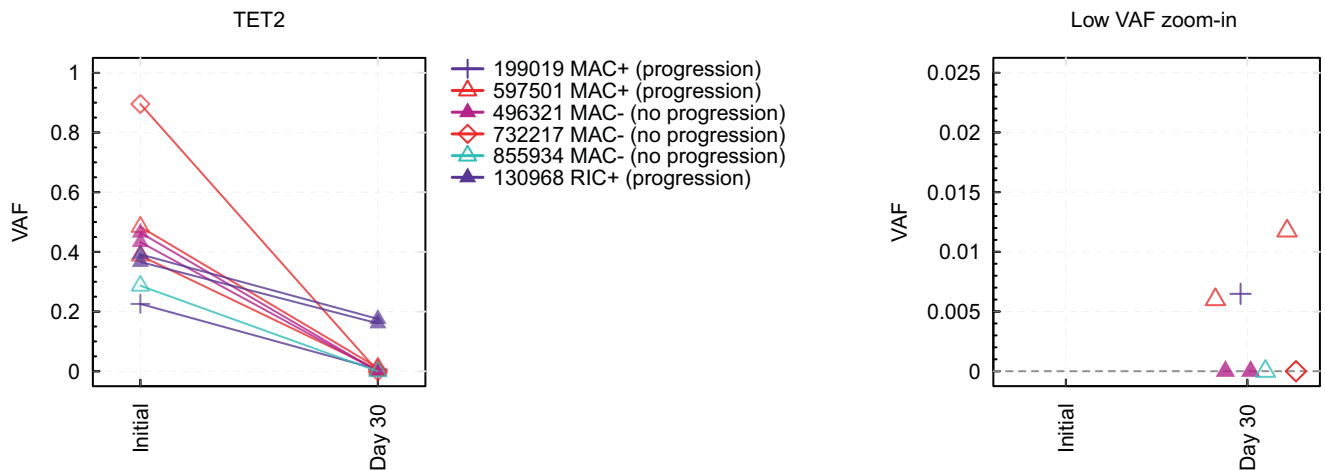
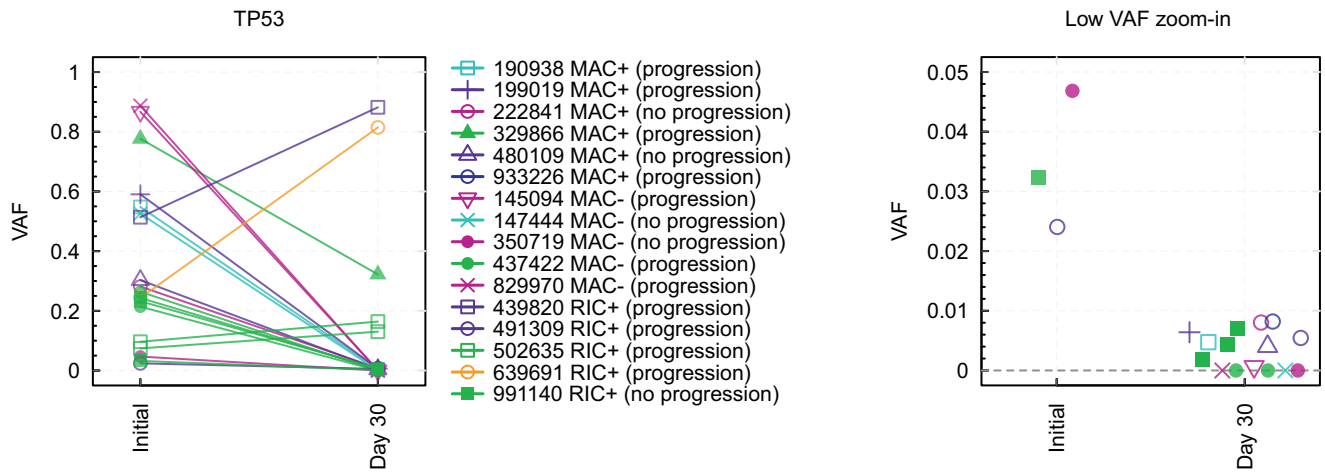


Figure S3. Mutation Clearance of Selected Genes at Day 30 Post-Allogeneic Hematopoietic Cell Transplant

J.



K.

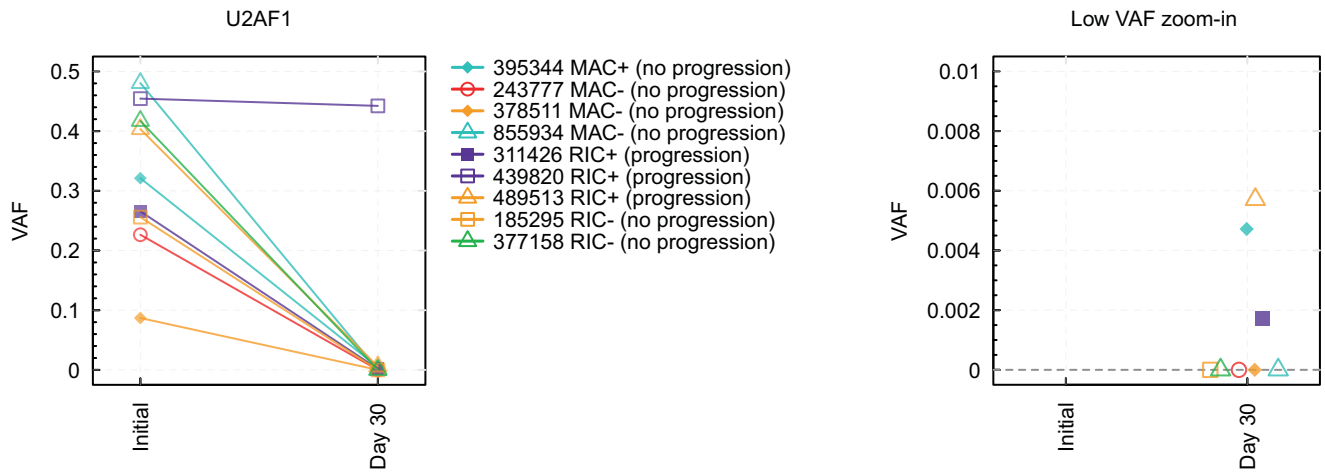
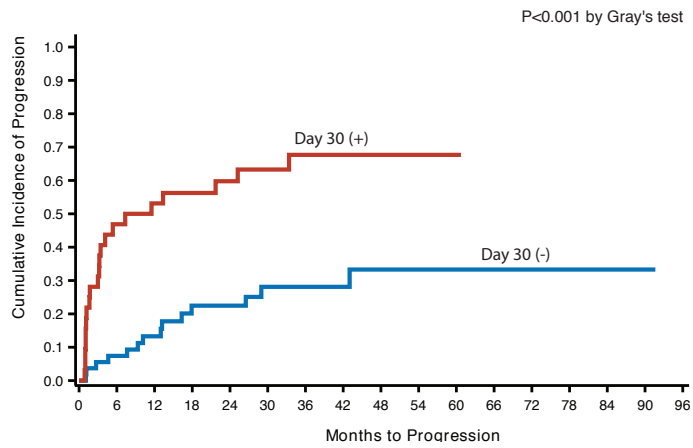
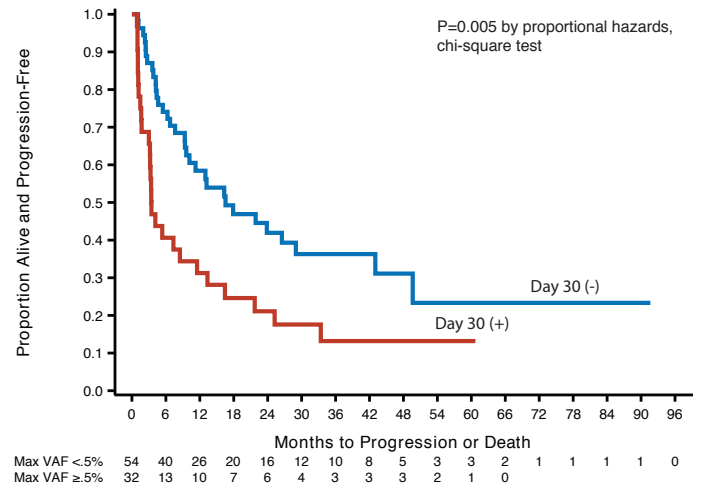


Figure S4. Association of Mutation Clearance at Day 30 post-alloHCT with Outcomes

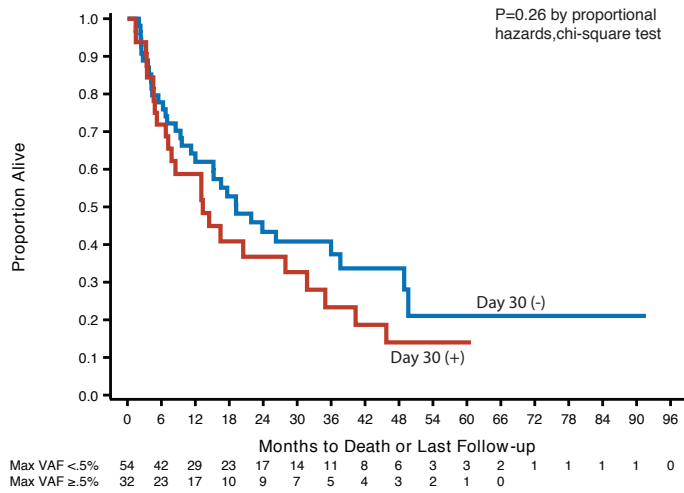
A.



B.



C.



D.

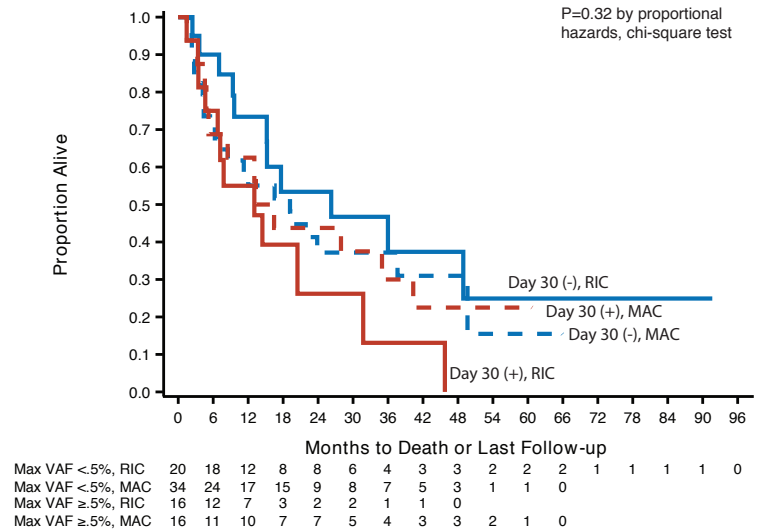


Figure S5. Association of Mutation Clearance at Day 100 post-alloHCT with Outcomes

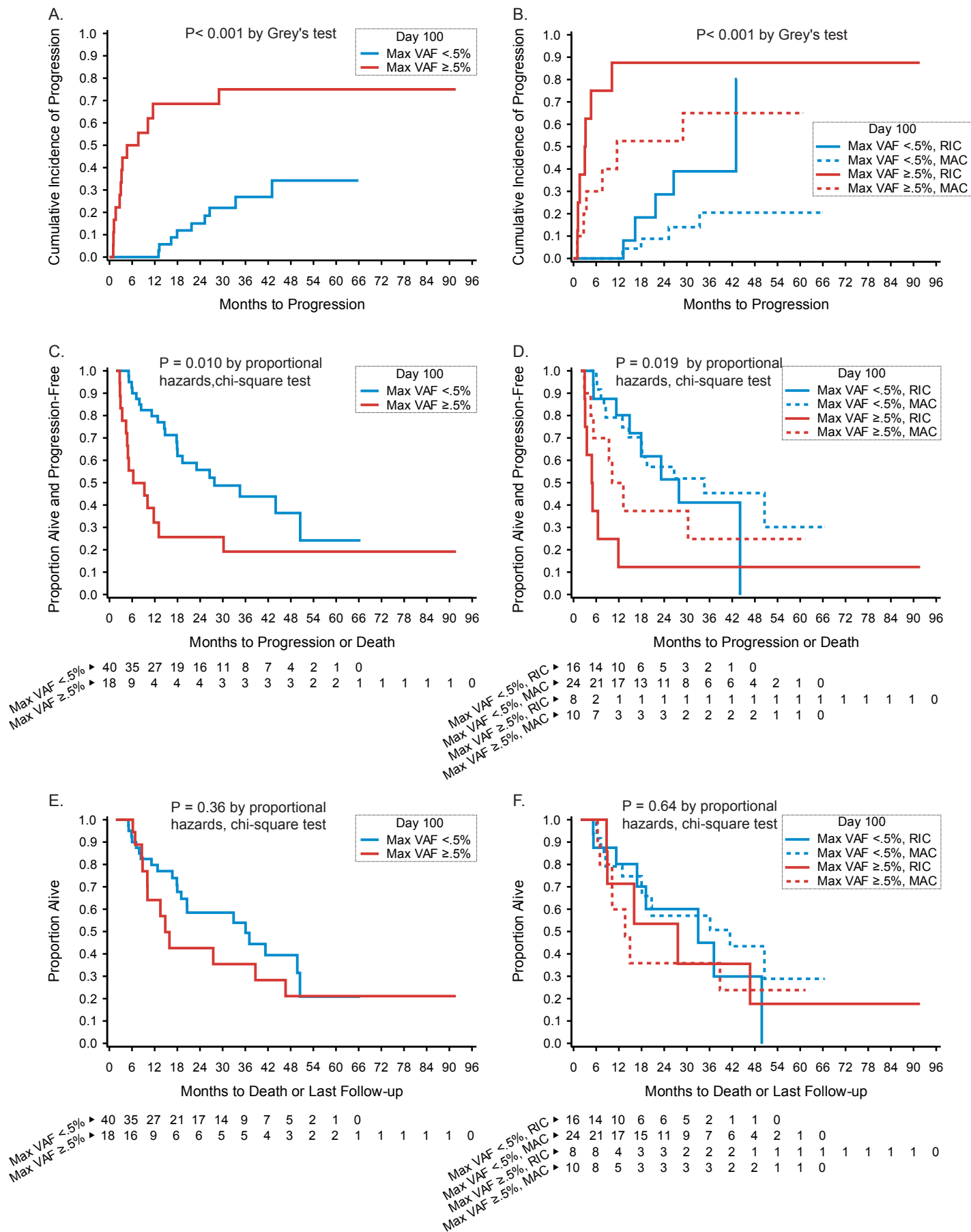


Figure S6. Association of Mutation Clearance with Outcomes Using a Variant Allele Frequency Threshold of 0.1%

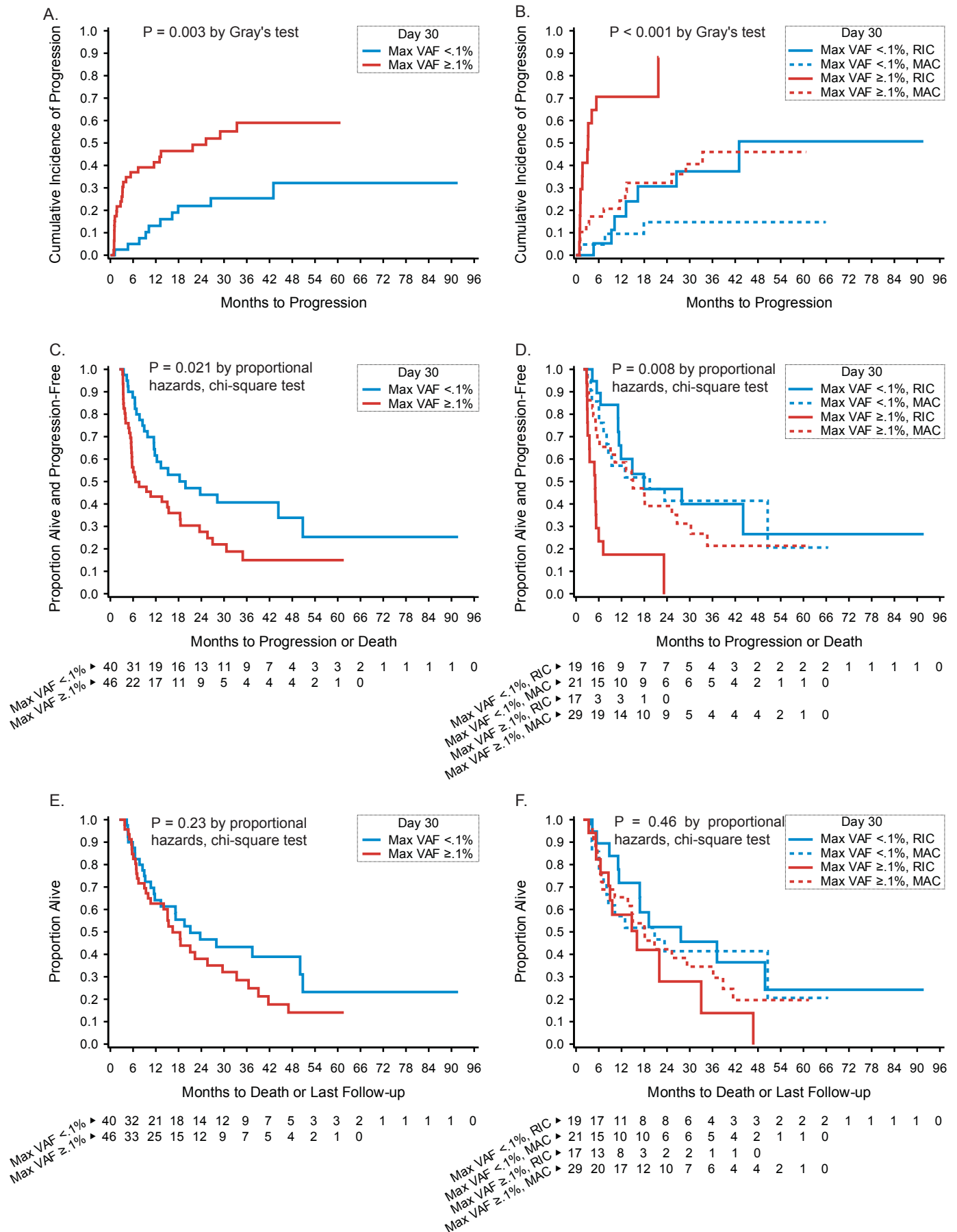


Figure S7. Association of Mutation Clearance with Outcomes Using a Variant Allele Frequency Threshold of 1%

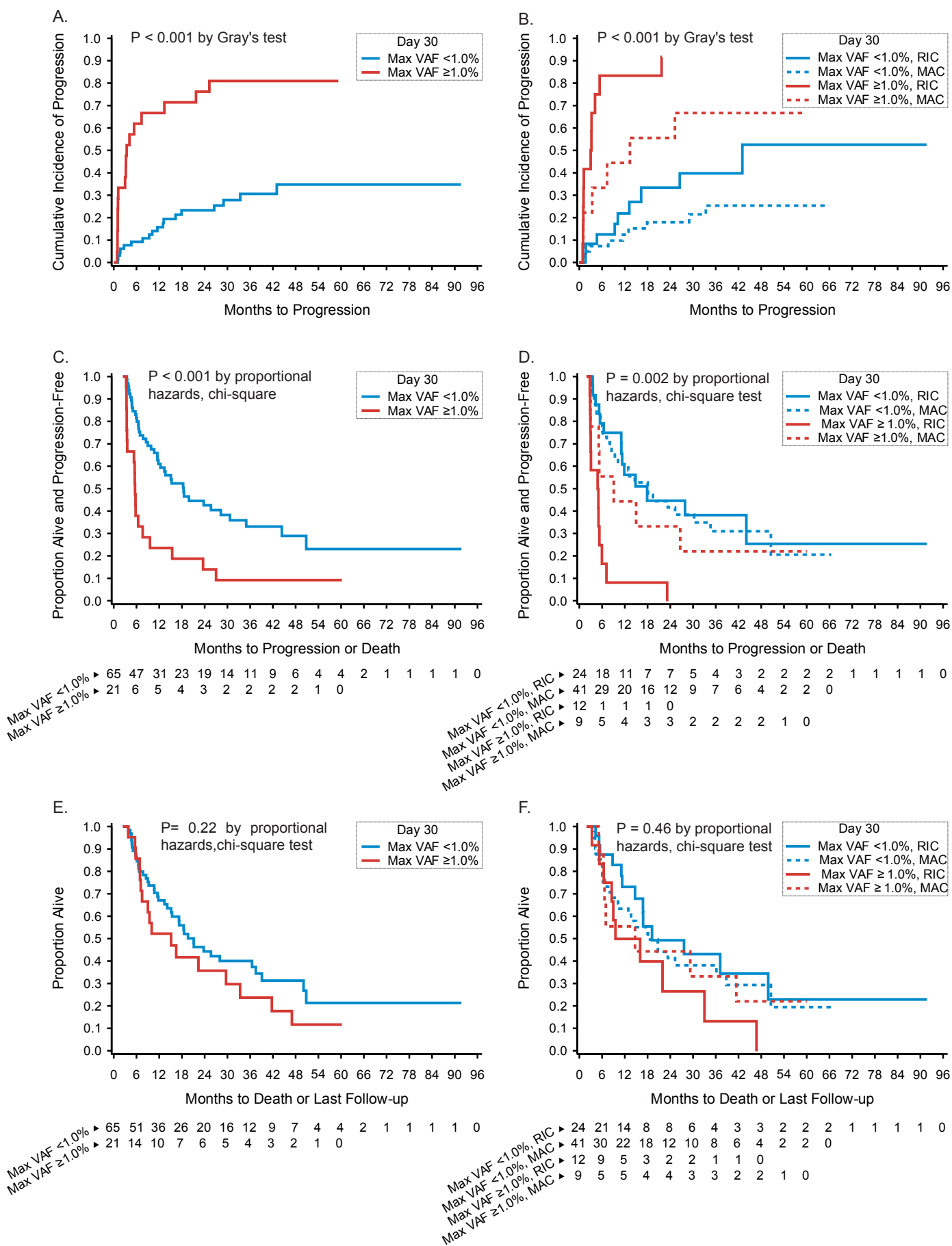


Figure S8. Association of Mutation Clearance with Outcomes Using a Variant Allele Frequency Threshold of 2.5%^{5%}

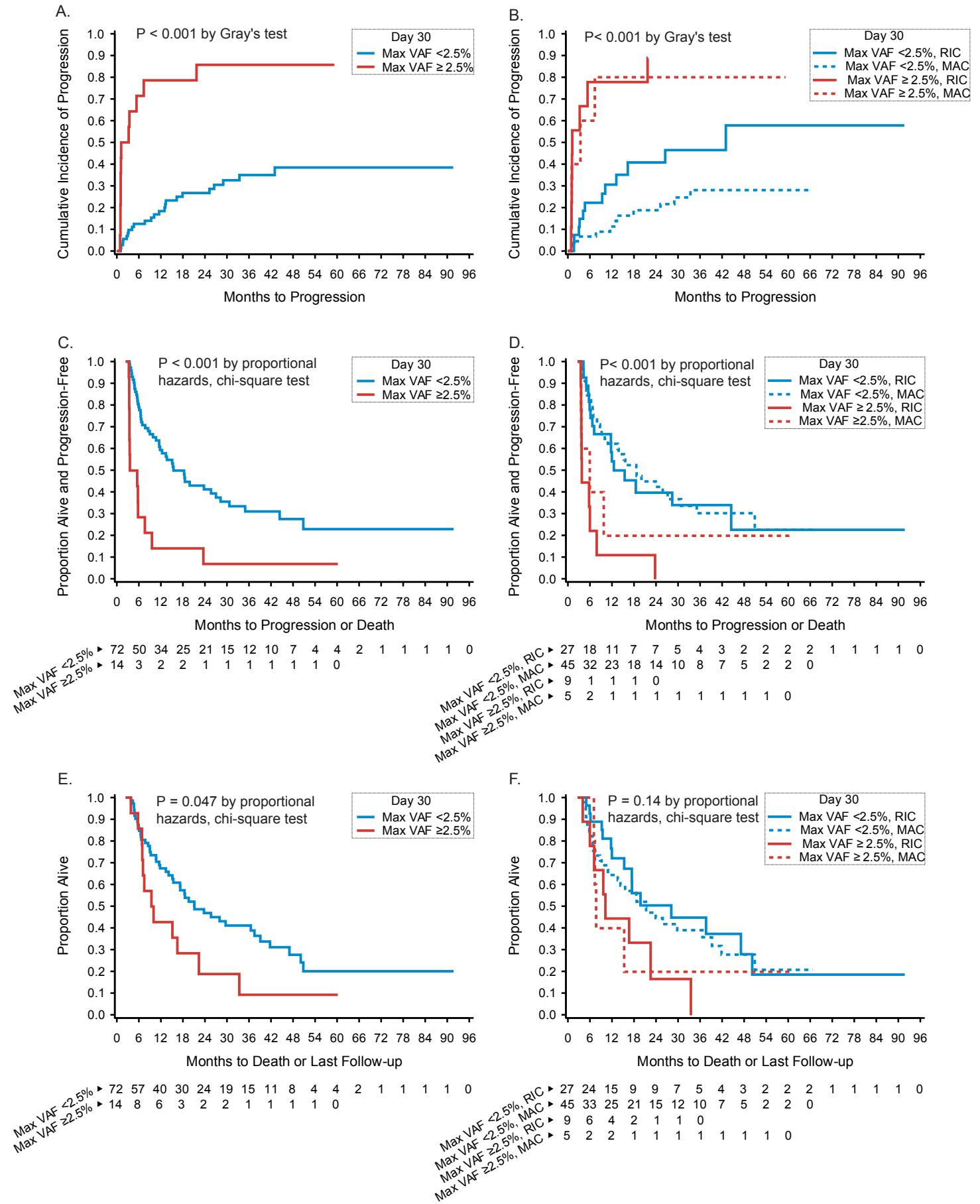


Figure S9. Association of Mutation Clearance with Outcomes Using Any Detectable Variant Allele Frequency

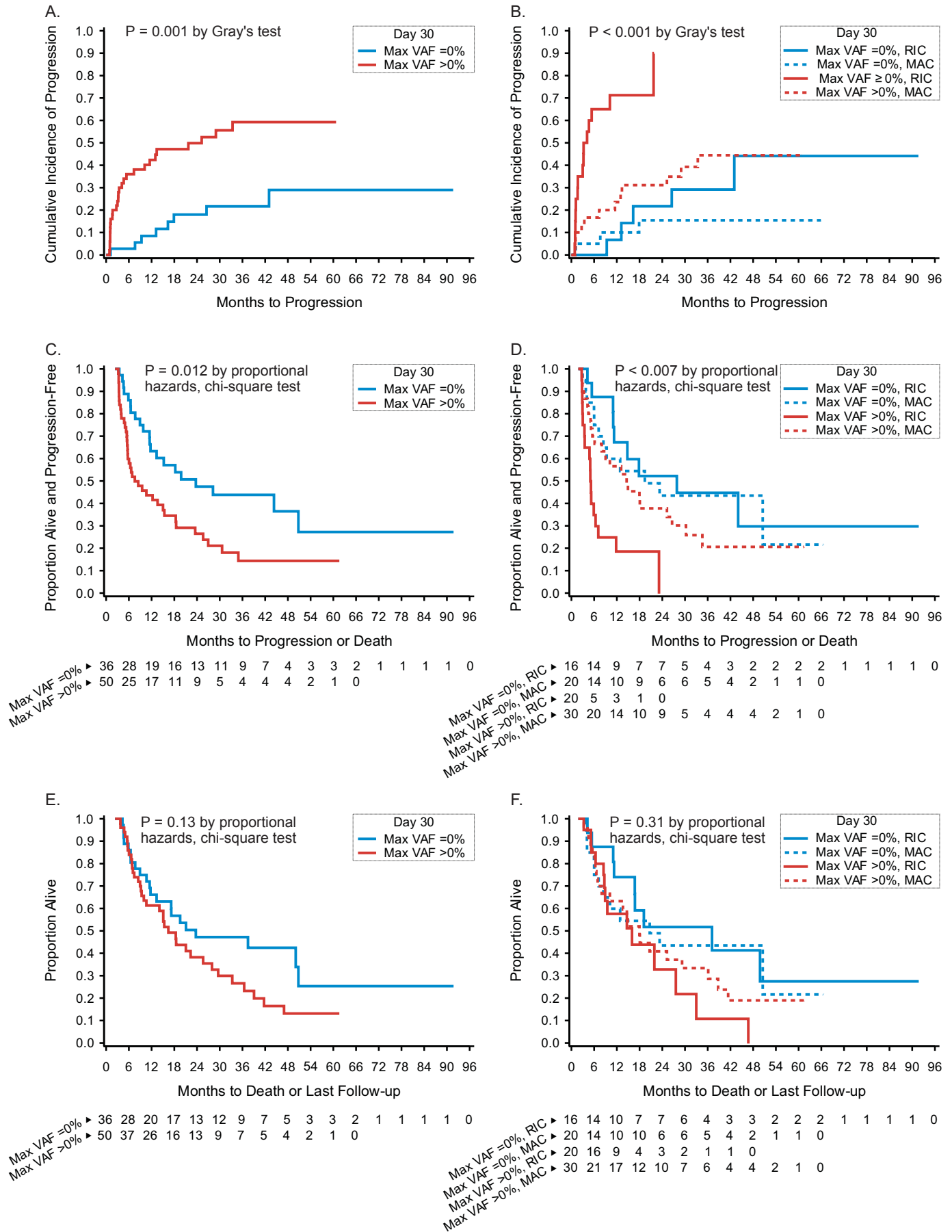
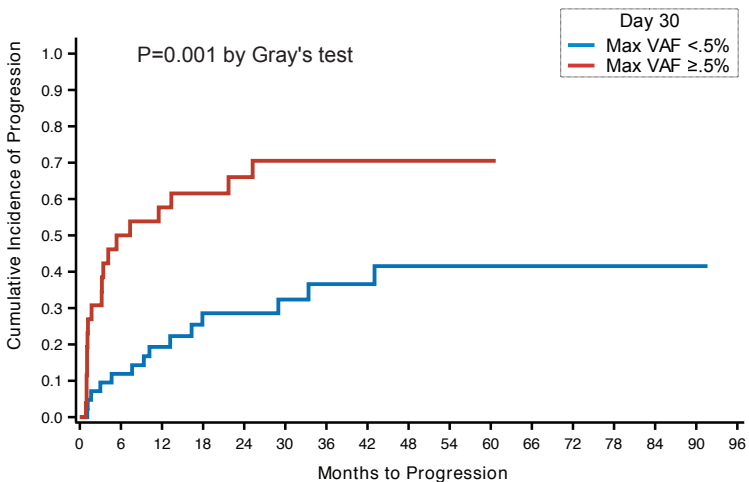
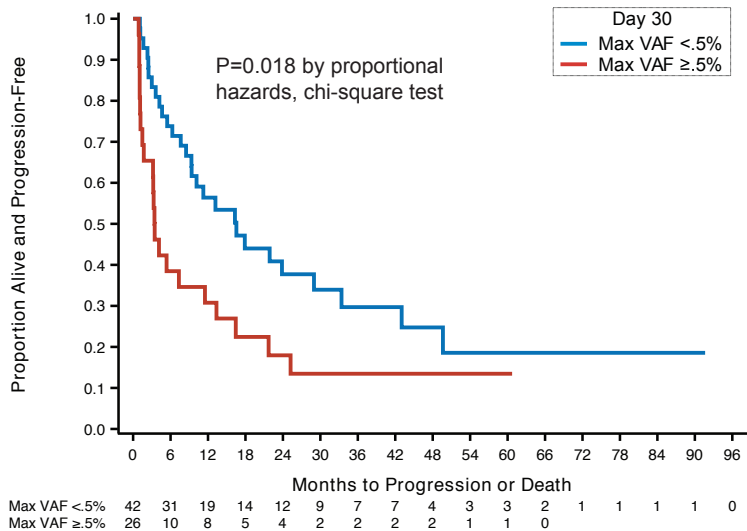


Figure S10. Association of Overall Survival with Mutation Clearance Determined by a 40 Gene-Panel

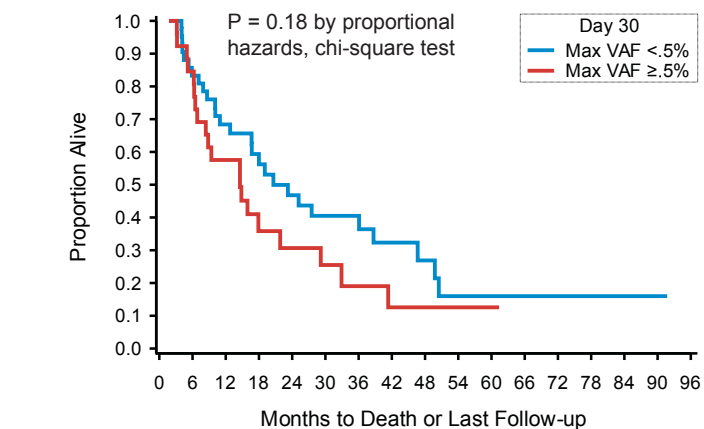
A.



B.

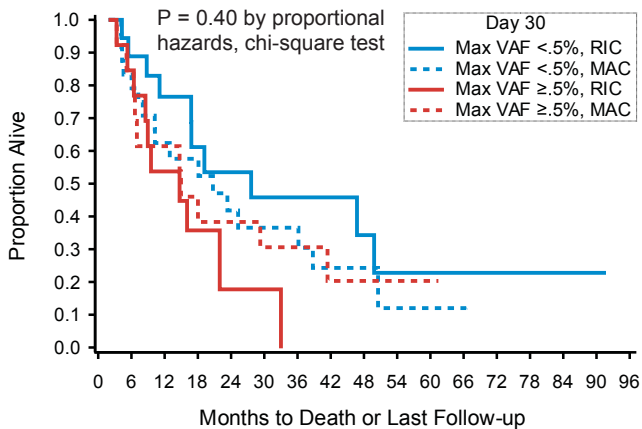


C.



| | | | | | | | | | | | | | | | | | | |
|--------------|---|----|----|----|----|----|----|---|---|---|---|---|---|---|---|---|---|---|
| Max VAF < 5% | ▶ | 42 | 34 | 22 | 17 | 14 | 11 | 9 | 8 | 5 | 3 | 3 | 2 | 1 | 1 | 1 | 1 | 0 |
| Max VAF ≥ 5% | ▶ | 26 | 18 | 14 | 7 | 6 | 4 | 3 | 2 | 2 | 1 | 1 | 0 | | | | | |

D.



| | | | | | | | | | | | | | | | | | | |
|-------------------|---|----|----|----|----|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Max VAF < 5%, RIC | ▶ | 18 | 16 | 11 | 7 | 7 | 5 | 4 | 4 | 3 | 2 | 2 | 2 | 1 | 1 | 1 | 1 | 0 |
| Max VAF < 5%, MAC | ▶ | 24 | 18 | 11 | 10 | 7 | 6 | 5 | 4 | 2 | 1 | 1 | 0 | | | | | |
| Max VAF ≥ 5%, RIC | ▶ | 13 | 10 | 6 | 2 | 1 | 1 | 0 | | | | | | | | | | |
| Max VAF ≥ 5%, MAC | ▶ | 13 | 8 | 8 | 5 | 5 | 3 | 3 | 2 | 2 | 1 | 1 | 0 | | | | | |

Table S1. Recurrently Mutated Genes in AML and MDS

| ENSG ID | Gene symbol | Chr | Start | End | Strand |
|-----------------|--------------------|------------|--------------|------------|---------------|
| ENSG00000078369 | <i>GNB1</i> | 1 | 1716729 | 1822495 | - |
| ENSG00000120952 | <i>PRAMEF2</i> | 1 | 12916941 | 12921764 | + |
| ENSG00000065526 | <i>SPEN</i> | 1 | 16174359 | 16266955 | + |
| ENSG00000058453 | <i>CROCC</i> | 1 | 17222232 | 17299474 | + |
| ENSG00000142661 | <i>MYOM3</i> | 1 | 24382525 | 24438665 | - |
| ENSG00000162521 | <i>RBBP4</i> | 1 | 33116749 | 33151812 | + |
| ENSG00000188910 | <i>GJB3</i> | 1 | 35246790 | 35251970 | + |
| ENSG00000054118 | <i>THRAP3</i> | 1 | 36690017 | 36770958 | + |
| ENSG00000183317 | <i>EPHA10</i> | 1 | 38179561 | 38230824 | - |
| ENSG00000162434 | <i>JAK1</i> | 1 | 65298906 | 65432187 | - |
| ENSG00000162654 | <i>GBP4</i> | 1 | 89646831 | 89664633 | - |
| ENSG00000203914 | <i>HSP90B3P</i> | 1 | 92100568 | 92109633 | + |
| ENSG00000213281 | <i>NRAS</i> | 1 | 115247078 | 115259515 | - |
| ENSG00000143373 | <i>ZNF687</i> | 1 | 151254094 | 151264381 | + |
| ENSG00000143631 | <i>FLG</i> | 1 | 152274651 | 152297679 | - |
| ENSG00000160691 | <i>SHC1</i> | 1 | 154934774 | 154946959 | - |
| ENSG00000196189 | <i>SEMA4A</i> | 1 | 156117157 | 156147543 | + |
| ENSG00000162733 | <i>DDR2</i> | 1 | 162601163 | 162750255 | + |
| ENSG00000034971 | <i>MYOC</i> | 1 | 171604557 | 171621811 | - |
| ENSG00000198216 | <i>CACNA1E</i> | 1 | 181382295 | 181770716 | + |
| ENSG00000143341 | <i>HMCN1</i> | 1 | 185703683 | 186160085 | + |
| ENSG00000162670 | <i>BRINP3</i> | 1 | 190066792 | 190446759 | - |
| ENSG00000134369 | <i>NAV1</i> | 1 | 201592411 | 201796097 | + |
| ENSG00000084674 | <i>APOB</i> | 2 | 21224301 | 21266945 | - |
| ENSG00000119772 | <i>DNMT3A</i> | 2 | 25455496 | 25565459 | - |
| ENSG00000091106 | <i>NLRC4</i> | 2 | 32449518 | 32490923 | - |
| ENSG00000115808 | <i>STRN</i> | 2 | 37070783 | 37193615 | - |
| ENSG00000068878 | <i>PSME4</i> | 2 | 54091204 | 54197977 | - |
| ENSG00000135636 | <i>DYSF</i> | 2 | 71680753 | 71913898 | + |
| ENSG00000168702 | <i>LRP1B</i> | 2 | 140988992 | 142889270 | - |
| ENSG00000144285 | <i>SCN1A</i> | 2 | 166845670 | 166930149 | - |
| ENSG00000115524 | <i>SF3B1</i> | 2 | 198256698 | 198299815 | - |
| ENSG00000138413 | <i>IDH1</i> | 2 | 209100951 | 209130798 | - |
| ENSG00000078018 | <i>MAP2</i> | 2 | 210288771 | 210598842 | + |
| ENSG00000054356 | <i>PTPRN</i> | 2 | 220154345 | 220174370 | - |
| ENSG00000072195 | <i>SPEG</i> | 2 | 220299568 | 220363009 | + |
| ENSG00000204120 | <i>GIGYF2</i> | 2 | 233562009 | 233725287 | + |
| ENSG00000085978 | <i>ATG16L1</i> | 2 | 234118697 | 234204320 | + |
| ENSG00000168334 | <i>XIRP1</i> | 3 | 39224701 | 39234087 | - |
| ENSG00000008300 | <i>CELSR3</i> | 3 | 48673896 | 48700348 | - |
| ENSG00000164061 | <i>BSN</i> | 3 | 49591922 | 49708982 | + |
| ENSG00000157445 | <i>CACNA2D3</i> | 3 | 54156574 | 55108584 | + |
| ENSG00000163618 | <i>CADPS</i> | 3 | 62384021 | 62861064 | - |
| ENSG00000114861 | <i>FOXP1</i> | 3 | 71003844 | 71633140 | - |
| ENSG00000175161 | <i>CADM2</i> | 3 | 85008132 | 86117951 | + |
| ENSG00000145103 | <i>ILDR1</i> | 3 | 121706170 | 121741051 | - |
| ENSG00000173175 | <i>ADCY5</i> | 3 | 123001143 | 123168605 | - |
| ENSG00000114626 | <i>ABTB1</i> | 3 | 127391778 | 127399763 | + |
| ENSG00000179348 | <i>GATA2</i> | 3 | 128198265 | 128212030 | - |
| ENSG00000118007 | <i>STAG1</i> | 3 | 136055077 | 136471220 | - |
| ENSG00000090402 | <i>SI</i> | 3 | 164696686 | 164796283 | - |

| | | | | | |
|-----------------|-----------------|---|-----------|-----------|---|
| ENSG00000136527 | <i>TRA2B</i> | 3 | 185632356 | 185655924 | - |
| ENSG00000038219 | <i>BOD1L1</i> | 4 | 13570362 | 13629347 | - |
| ENSG00000047365 | <i>ARAP2</i> | 4 | 36067620 | 36246131 | - |
| ENSG00000075539 | <i>FRYL</i> | 4 | 48499378 | 48782339 | - |
| ENSG00000072201 | <i>LNK1</i> | 4 | 54325468 | 54567572 | - |
| ENSG00000134853 | <i>PDGFRA</i> | 4 | 55095264 | 55164414 | + |
| ENSG00000157404 | <i>KIT</i> | 4 | 55524085 | 55606881 | + |
| ENSG00000128052 | <i>KDR</i> | 4 | 55944426 | 55991762 | - |
| ENSG00000118777 | <i>ABCG2</i> | 4 | 89011416 | 89152474 | - |
| ENSG00000168769 | <i>TET2</i> | 4 | 106067032 | 106200973 | + |
| ENSG00000145362 | <i>ANK2</i> | 4 | 113739239 | 114304896 | + |
| ENSG00000198589 | <i>LRBA</i> | 4 | 151185594 | 151936879 | - |
| ENSG00000197410 | <i>DCHS2</i> | 4 | 155155527 | 155412930 | - |
| ENSG00000039139 | <i>DNAH5</i> | 5 | 13690437 | 13944652 | - |
| ENSG00000131711 | <i>MAP1B</i> | 5 | 71403061 | 71505397 | + |
| ENSG00000164309 | <i>CMYA5</i> | 5 | 78985659 | 79096063 | + |
| ENSG00000038427 | <i>VCAN</i> | 5 | 82767284 | 82878122 | + |
| ENSG00000164176 | <i>EDIL3</i> | 5 | 83236373 | 83680611 | - |
| ENSG00000120733 | <i>KDM3B</i> | 5 | 137688285 | 137772717 | + |
| ENSG00000081842 | <i>PCDHA6</i> | 5 | 140206816 | 140391929 | + |
| ENSG00000239389 | <i>PCDHA13</i> | 5 | 140261793 | 140391929 | + |
| ENSG00000171815 | <i>PCDHB1</i> | 5 | 140430961 | 140433547 | + |
| ENSG00000146001 | <i>PCDHB18</i> | 5 | 140613936 | 140617101 | + |
| ENSG00000182578 | <i>CSF1R</i> | 5 | 149432854 | 149492935 | - |
| ENSG00000019582 | <i>CD74</i> | 5 | 149781200 | 149792332 | - |
| ENSG00000132911 | <i>NMUR2</i> | 5 | 151771093 | 151784840 | - |
| ENSG00000118322 | <i>ATP10B</i> | 5 | 159990127 | 160279219 | - |
| ENSG00000134516 | <i>DOCK2</i> | 5 | 169064251 | 169510386 | + |
| ENSG00000181163 | <i>NPM1</i> | 5 | 170814708 | 170838141 | + |
| ENSG00000113739 | <i>STC2</i> | 5 | 172741716 | 172756506 | - |
| ENSG00000183258 | <i>DDX41</i> | 5 | 176938578 | 176944470 | - |
| ENSG00000112739 | <i>PRPF4B</i> | 6 | 4021501 | 4065217 | + |
| ENSG00000095951 | <i>HIVEP1</i> | 6 | 12008995 | 12165232 | + |
| ENSG00000112137 | <i>PHACTR1</i> | 6 | 12717833 | 13288075 | + |
| ENSG00000137411 | <i>VAR2</i> | 6 | 30881982 | 30894233 | + |
| ENSG00000197283 | <i>SYNGAP1</i> | 6 | 33387847 | 33421466 | + |
| ENSG00000096433 | <i>ITPR3</i> | 6 | 33588522 | 33664351 | + |
| ENSG00000112624 | <i>GLTSCR1L</i> | 6 | 42714696 | 42836296 | + |
| ENSG00000146216 | <i>TTBK1</i> | 6 | 43211222 | 43255997 | + |
| ENSG00000170927 | <i>PKHD1</i> | 6 | 51480098 | 51952423 | - |
| ENSG00000151914 | <i>DST</i> | 6 | 56322785 | 56819413 | - |
| ENSG00000079841 | <i>RIMS1</i> | 6 | 72596406 | 73112508 | + |
| ENSG00000111799 | <i>COL12A1</i> | 6 | 75794042 | 75915767 | - |
| ENSG00000146247 | <i>PHIP</i> | 6 | 79644136 | 79788011 | - |
| ENSG00000164418 | <i>GRIK2</i> | 6 | 101846664 | 102517958 | + |
| ENSG00000112333 | <i>NR2E1</i> | 6 | 108487215 | 108510013 | + |
| ENSG00000213079 | <i>SCAF8</i> | 6 | 155054459 | 155230002 | + |
| ENSG00000164619 | <i>BMPER</i> | 7 | 33944523 | 34195484 | + |
| ENSG00000002746 | <i>HECW1</i> | 7 | 43152198 | 43602938 | + |
| ENSG00000146648 | <i>EGFR</i> | 7 | 55086714 | 55324313 | + |
| ENSG00000187391 | <i>MAGI2</i> | 7 | 77646374 | 79082890 | - |
| ENSG00000075213 | <i>SEMA3A</i> | 7 | 83587659 | 84122040 | - |
| ENSG00000198822 | <i>GRM3</i> | 7 | 86273224 | 86494200 | + |
| ENSG00000066923 | <i>STAG3</i> | 7 | 99775186 | 99819111 | + |
| ENSG00000154415 | <i>PPP1R3A</i> | 7 | 113516832 | 113715975 | - |

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|-----------------|------------------|----|-----------|-----------|---|
| ENSG00000105976 | <i>MET</i> | 7 | 116312444 | 116438440 | + |
| ENSG00000179603 | <i>GRM8</i> | 7 | 126078652 | 126893348 | - |
| ENSG00000128594 | <i>LRRC4</i> | 7 | 127667124 | 127672160 | - |
| ENSG00000128578 | <i>STRIP2</i> | 7 | 129073731 | 129128240 | + |
| ENSG00000269955 | <i>LUC7L2</i> | 7 | 139026106 | 139107345 | + |
| ENSG00000157764 | <i>BRAF</i> | 7 | 140419127 | 140624564 | - |
| ENSG00000197448 | <i>GSTK1</i> | 7 | 142941186 | 142967947 | + |
| ENSG00000055130 | <i>CUL1</i> | 7 | 148395006 | 148498202 | + |
| ENSG00000106462 | <i>EZH2</i> | 7 | 148504475 | 148581414 | - |
| ENSG00000055118 | <i>KCNH2</i> | 7 | 150642049 | 150675403 | - |
| ENSG00000055609 | <i>KMT2C</i> | 7 | 151832007 | 152133628 | - |
| ENSG00000183117 | <i>CSMD1</i> | 8 | 2792875 | 4852328 | - |
| ENSG00000164741 | <i>DLC1</i> | 8 | 12940870 | 13372396 | - |
| ENSG00000215262 | <i>KCNU1</i> | 8 | 36641842 | 36793643 | + |
| ENSG00000248522 | <i>SBF1P1</i> | 8 | 56362069 | 56367699 | - |
| ENSG00000121005 | <i>CRISPLD1</i> | 8 | 75896843 | 75946793 | + |
| ENSG00000079102 | <i>RUNX1T1</i> | 8 | 92971152 | 93107443 | - |
| ENSG00000205038 | <i>PKHD1L1</i> | 8 | 110374706 | 110543500 | + |
| ENSG00000164796 | <i>CSMD3</i> | 8 | 113235157 | 114449328 | - |
| ENSG00000164754 | <i>RAD21</i> | 8 | 117858173 | 117887105 | - |
| ENSG00000136997 | <i>MYC</i> | 8 | 128747680 | 128753674 | + |
| ENSG00000226807 | <i>MROH5</i> | 8 | 142443929 | 142517330 | - |
| ENSG00000227184 | <i>EPPK1</i> | 8 | 144935822 | 144947434 | - |
| ENSG00000096968 | <i>JAK2</i> | 9 | 4985033 | 5128183 | + |
| ENSG00000122735 | <i>DNAI1</i> | 9 | 34457412 | 34520982 | + |
| ENSG00000083067 | <i>TRPM3</i> | 9 | 73149949 | 74061820 | - |
| ENSG00000165119 | <i>HNRNPK</i> | 9 | 86582998 | 86595569 | - |
| ENSG00000185920 | <i>PTCH1</i> | 9 | 98205262 | 98279339 | - |
| ENSG00000197816 | <i>CCDC180</i> | 9 | 100000779 | 100139569 | + |
| ENSG00000041982 | <i>TNC</i> | 9 | 117782805 | 117880486 | - |
| ENSG00000106804 | <i>C5</i> | 9 | 123714613 | 123837452 | - |
| ENSG00000107147 | <i>KCNT1</i> | 9 | 138594031 | 138684992 | + |
| ENSG00000148408 | <i>CACNA1B</i> | 9 | 140772241 | 141019076 | + |
| ENSG00000095787 | <i>WAC</i> | 10 | 28821422 | 28911862 | + |
| ENSG00000204176 | <i>SYT15</i> | 10 | 46955444 | 46971400 | - |
| ENSG00000138336 | <i>TET1</i> | 10 | 70320117 | 70454239 | + |
| ENSG00000107731 | <i>UNC5B</i> | 10 | 72972298 | 73062621 | + |
| ENSG00000148602 | <i>LRIT1</i> | 10 | 85991276 | 86001217 | - |
| ENSG00000182771 | <i>GRID1</i> | 10 | 87359312 | 88126250 | - |
| ENSG00000171862 | <i>PTEN</i> | 10 | 89622870 | 89731687 | + |
| ENSG00000138193 | <i>PLCE1</i> | 10 | 95753746 | 96088149 | + |
| ENSG00000166024 | <i>R3HCC1L</i> | 10 | 99894381 | 100004654 | + |
| ENSG00000107593 | <i>PKD2L1</i> | 10 | 102047903 | 102090243 | - |
| ENSG00000229256 | <i>ST13P13</i> | 10 | 104975466 | 104976557 | + |
| ENSG00000108055 | <i>SMC3</i> | 10 | 112327449 | 112364394 | + |
| ENSG00000165813 | <i>C10orf118</i> | 10 | 115880621 | 115934364 | - |
| ENSG00000117983 | <i>MUC5B</i> | 11 | 1142474 | 1288726 | + |
| ENSG00000110328 | <i>GALNT18</i> | 11 | 11292421 | 11643561 | - |
| ENSG00000129173 | <i>E2F8</i> | 11 | 19245614 | 19263389 | - |
| ENSG00000182255 | <i>KCNA4</i> | 11 | 30031765 | 30038488 | - |
| ENSG00000184937 | <i>WT1</i> | 11 | 32409321 | 32457176 | - |
| ENSG00000149480 | <i>MTA2</i> | 11 | 62360675 | 62369303 | - |
| ENSG00000168066 | <i>SF1</i> | 11 | 64532078 | 64546258 | - |
| ENSG00000175591 | <i>P2RY2</i> | 11 | 72929344 | 72947397 | + |
| ENSG00000074266 | <i>EED</i> | 11 | 85955815 | 85989781 | + |

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|-----------------|-----------------|----|-----------|-----------|---|
| ENSG00000165325 | <i>CCDC67</i> | 11 | 93063883 | 93171636 | + |
| ENSG00000149972 | <i>CNTN5</i> | 11 | 98891871 | 100227473 | + |
| ENSG00000187240 | <i>DYNC2H1</i> | 11 | 102980160 | 103350591 | + |
| ENSG00000149295 | <i>DRD2</i> | 11 | 113280317 | 113346001 | - |
| ENSG00000110395 | <i>CBL</i> | 11 | 119076990 | 119178859 | + |
| ENSG00000149403 | <i>GRIK4</i> | 11 | 120530971 | 120857132 | + |
| ENSG00000170953 | <i>OR8B12</i> | 11 | 124412618 | 124413550 | - |
| ENSG00000111642 | <i>CHD4</i> | 12 | 6679248 | 6716551 | - |
| ENSG00000139083 | <i>ETV6</i> | 12 | 11802788 | 12048336 | + |
| ENSG00000111276 | <i>CDKN1B</i> | 12 | 12867992 | 12875305 | + |
| ENSG00000133703 | <i>KRAS</i> | 12 | 25358180 | 25403854 | - |
| ENSG0000013573 | <i>DDX11</i> | 12 | 31226779 | 31257725 | + |
| ENSG00000167548 | <i>KMT2D</i> | 12 | 49412758 | 49453557 | - |
| ENSG00000110844 | <i>PRPF40B</i> | 12 | 49962001 | 50038449 | + |
| ENSG00000185640 | <i>KRT79</i> | 12 | 53215231 | 53228079 | - |
| ENSG00000123411 | <i>IKZF4</i> | 12 | 56414689 | 56432219 | + |
| ENSG00000139540 | <i>SLC39A5</i> | 12 | 56623820 | 56631630 | + |
| ENSG00000179295 | <i>PTPN11</i> | 12 | 112856536 | 112947717 | + |
| ENSG00000171435 | <i>KSR2</i> | 12 | 117890817 | 118406060 | - |
| ENSG00000073060 | <i>SCARB1</i> | 12 | 125262174 | 125348519 | - |
| ENSG00000198033 | <i>TUBA3C</i> | 13 | 19747910 | 19755992 | - |
| ENSG00000122025 | <i>FLT3</i> | 13 | 28577411 | 28674729 | - |
| ENSG00000102755 | <i>FLT1</i> | 13 | 28874489 | 29069265 | - |
| ENSG00000132938 | <i>MTUS2</i> | 13 | 29598748 | 30080084 | + |
| ENSG00000133119 | <i>RFC3</i> | 13 | 34392186 | 34540695 | + |
| ENSG00000133083 | <i>DCLK1</i> | 13 | 36343122 | 36705464 | - |
| ENSG00000150893 | <i>FREM2</i> | 13 | 39261173 | 39461268 | + |
| ENSG00000139687 | <i>RB1</i> | 13 | 48877887 | 49056122 | + |
| ENSG00000083520 | <i>DIS3</i> | 13 | 73329540 | 73356344 | - |
| ENSG00000169508 | <i>GPR183</i> | 13 | 99946784 | 99959749 | - |
| ENSG00000102452 | <i>NALCN</i> | 13 | 101706130 | 102068843 | - |
| ENSG00000184497 | <i>TMEM255B</i> | 13 | 114462216 | 114514926 | + |
| ENSG00000183087 | <i>GAS6</i> | 13 | 114523522 | 114567046 | - |
| ENSG00000257115 | <i>OR11H12</i> | 14 | 19377594 | 19378574 | + |
| ENSG00000054690 | <i>PLEKHH1</i> | 14 | 68000008 | 68056255 | + |
| ENSG00000021645 | <i>NRXN3</i> | 14 | 78870093 | 80330762 | + |
| ENSG00000185070 | <i>FLRT2</i> | 14 | 85996488 | 86094270 | + |
| ENSG00000152315 | <i>KCNK13</i> | 14 | 90528108 | 90652199 | + |
| ENSG00000211897 | <i>IGHG3</i> | 14 | 106230841 | 106237742 | - |
| ENSG00000198838 | <i>RYR3</i> | 15 | 33603177 | 34158303 | + |
| ENSG00000188659 | <i>FAM154B</i> | 15 | 82555140 | 82577268 | + |
| ENSG00000136383 | <i>ALPK3</i> | 15 | 85359911 | 85416713 | + |
| ENSG00000170776 | <i>AKAP13</i> | 15 | 85923871 | 86292586 | + |
| ENSG00000140538 | <i>NTRK3</i> | 15 | 88402982 | 88799978 | - |
| ENSG00000182054 | <i>IDH2</i> | 15 | 90626277 | 90645736 | - |
| ENSG00000103313 | <i>MEFV</i> | 16 | 3292028 | 3306627 | - |
| ENSG00000157106 | <i>SMG1</i> | 16 | 18816175 | 18937726 | - |
| ENSG00000149926 | <i>FAM57B</i> | 16 | 30035748 | 30042194 | - |
| ENSG00000070915 | <i>SLC12A3</i> | 16 | 56899119 | 56949762 | + |
| ENSG00000067955 | <i>CBFB</i> | 16 | 67063050 | 67134961 | + |
| ENSG00000039523 | <i>FAM65A</i> | 16 | 67562720 | 67580689 | + |
| ENSG00000102974 | <i>CTCF</i> | 16 | 67596310 | 67673086 | + |
| ENSG00000039068 | <i>CDH1</i> | 16 | 68771128 | 68869451 | + |
| ENSG00000157423 | <i>HYDIN</i> | 16 | 70841289 | 71264569 | - |
| ENSG00000140839 | <i>CLEC18B</i> | 16 | 74442529 | 74455649 | - |

| | | | | | |
|-----------------|-----------------|----|----------|----------|---|
| ENSG00000152910 | <i>CNTNAP4</i> | 16 | 76311176 | 76593135 | + |
| ENSG00000166473 | <i>PKD1L2</i> | 16 | 81134484 | 81253975 | - |
| ENSG00000158545 | <i>ZC3H18</i> | 16 | 88636789 | 88698374 | + |
| ENSG00000187741 | <i>FANCA</i> | 16 | 89803957 | 89883065 | - |
| ENSG00000187624 | <i>C17orf97</i> | 17 | 260118 | 264367 | + |
| ENSG00000174231 | <i>PRPF8</i> | 17 | 1553923 | 1588176 | - |
| ENSG00000141510 | <i>TP53</i> | 17 | 7565257 | 7590863 | - |
| ENSG00000264424 | <i>MYH4</i> | 17 | 10346607 | 10372876 | - |
| ENSG00000007174 | <i>DNAH9</i> | 17 | 11501748 | 11873485 | + |
| ENSG00000141027 | <i>NCOR1</i> | 17 | 15932471 | 16121499 | - |
| ENSG00000196712 | <i>NF1</i> | 17 | 29421945 | 29705949 | + |
| ENSG00000178691 | <i>SUZ12</i> | 17 | 30264044 | 30328064 | + |
| ENSG00000171345 | <i>KRT19</i> | 17 | 39679869 | 39684641 | - |
| ENSG00000073670 | <i>ADAM11</i> | 17 | 42836568 | 42859214 | + |
| ENSG00000120071 | <i>KANSL1</i> | 17 | 44107282 | 44270166 | - |
| ENSG00000006283 | <i>CACNA1G</i> | 17 | 48638429 | 48704835 | + |
| ENSG00000141200 | <i>KIF2B</i> | 17 | 51900239 | 51902573 | + |
| ENSG00000180891 | <i>CUEDC1</i> | 17 | 55940337 | 56032684 | - |
| ENSG00000265206 | <i>MIR142</i> | 17 | 56408593 | 56408679 | - |
| ENSG00000109066 | <i>TMEM104</i> | 17 | 72772622 | 72835922 | + |
| ENSG00000161547 | <i>SRSF2</i> | 17 | 74730197 | 74733413 | - |
| ENSG00000173821 | <i>RNF213</i> | 17 | 78234665 | 78370085 | + |
| ENSG00000152217 | <i>SETBP1</i> | 18 | 42260138 | 42648475 | + |
| ENSG00000167306 | <i>MYO5B</i> | 18 | 47349101 | 47721384 | - |
| ENSG00000197561 | <i>ELANE</i> | 19 | 851014 | 856242 | + |
| ENSG00000181143 | <i>MUC16</i> | 19 | 8959520 | 9092018 | - |
| ENSG00000080573 | <i>COL5A3</i> | 19 | 10070237 | 10121147 | - |
| ENSG00000105397 | <i>TYK2</i> | 19 | 10461204 | 10491248 | - |
| ENSG00000179218 | <i>CALR</i> | 19 | 13049414 | 13055304 | + |
| ENSG00000105639 | <i>JAK3</i> | 19 | 17935591 | 17958870 | - |
| ENSG00000130518 | <i>KIAA1683</i> | 19 | 18367906 | 18385319 | - |
| ENSG00000105701 | <i>FKBP8</i> | 19 | 18642568 | 18654383 | - |
| ENSG00000245848 | <i>CEBPA</i> | 19 | 33790840 | 33793470 | - |
| ENSG00000126249 | <i>PDCD2L</i> | 19 | 34895303 | 34917073 | + |
| ENSG00000196218 | <i>RYR1</i> | 19 | 38924340 | 39078204 | + |
| ENSG00000090920 | <i>FCGBP</i> | 19 | 40353963 | 40440533 | - |
| ENSG00000105429 | <i>MEGF8</i> | 19 | 42829761 | 42882921 | + |
| ENSG00000063244 | <i>U2AF2</i> | 19 | 56165512 | 56186081 | + |
| ENSG00000171456 | <i>ASXL1</i> | 20 | 30946153 | 31027122 | + |
| ENSG00000088305 | <i>DNMT3B</i> | 20 | 31350191 | 31397162 | + |
| ENSG00000196090 | <i>PTPRT</i> | 20 | 40701392 | 41818610 | - |
| ENSG00000087460 | <i>GNAS</i> | 20 | 57414773 | 57486247 | + |
| ENSG00000075043 | <i>KCNQ2</i> | 20 | 62037542 | 62103993 | - |
| ENSG00000159216 | <i>RUNX1</i> | 21 | 36160098 | 37357047 | - |
| ENSG00000171587 | <i>DSCAM</i> | 21 | 41382926 | 42219065 | - |
| ENSG00000160201 | <i>U2AF1</i> | 21 | 44513066 | 44527697 | - |
| ENSG00000099954 | <i>CECR2</i> | 22 | 17840837 | 18033845 | + |
| ENSG00000100038 | <i>TOP3B</i> | 22 | 21075651 | 22337213 | - |
| ENSG00000099995 | <i>SF3A1</i> | 22 | 30727977 | 30752936 | - |
| ENSG00000146950 | <i>SHROOM2</i> | X | 9754496 | 9917483 | + |
| ENSG00000169249 | <i>ZRSR2</i> | X | 15808595 | 15841383 | + |
| ENSG00000102098 | <i>SCML2</i> | X | 18257433 | 18372847 | - |
| ENSG00000185448 | <i>FAM47A</i> | X | 34147873 | 34150428 | - |
| ENSG00000183337 | <i>BCOR</i> | X | 39909068 | 40036582 | - |
| ENSG00000124486 | <i>USP9X</i> | X | 40944888 | 41095832 | + |

| | | | | | |
|-----------------|------------------|---|-----------|-----------|---|
| ENSG00000147050 | <i>KDM6A</i> | X | 44732423 | 44971847 | + |
| ENSG00000102145 | <i>GATA1</i> | X | 48644962 | 48652716 | + |
| ENSG00000072501 | <i>SMC1A</i> | X | 53401070 | 53449677 | - |
| ENSG00000102043 | <i>MTMR8</i> | X | 63487961 | 63615311 | - |
| ENSG00000184634 | <i>MED12</i> | X | 70338406 | 70362304 | + |
| ENSG00000215093 | <i>EEF1A1P29</i> | X | 85415268 | 85416363 | - |
| ENSG00000204071 | <i>TCEAL6</i> | X | 101394933 | 101397942 | - |
| ENSG00000196507 | <i>TCEAL3</i> | X | 102862379 | 102884618 | + |
| ENSG00000177485 | <i>ZBTB33</i> | X | 119384607 | 119392251 | + |
| ENSG00000101972 | <i>STAG2</i> | X | 123094062 | 123556514 | + |
| ENSG00000171054 | <i>OR13H1</i> | X | 130677950 | 130679030 | + |
| ENSG00000156531 | <i>PHF6</i> | X | 133507283 | 133562822 | + |
| ENSG00000156920 | <i>GPR112</i> | X | 135383122 | 135499047 | + |
| ENSG00000067842 | <i>ATP2B3</i> | X | 152783134 | 152848397 | + |

All genome coordinates use NCBI Build 37.

Chr, chromosome.

Table S3. Somatic Mutations (SNVs and indels) by Exome Sequencing

| UPN | Chr | Start | Stop | Reference | Variant | Var Type | Gene Symbol | Transcript_ID | Strand | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal | Normal | Tumor | | | |
|--------|-----|-----------|-----------|-----------|---------|----------|--------------|-----------------|--------|-----------------------------|-----------------|-------------------|-------|-----------|-----------|------------|-----------|-----------|-----------|
| | | | | | | | | | | | | | | Ref count | Var Count | Normal VAF | Ref Count | Var Count | Tumor VAF |
| 611 | 1 | 53547545 | 53547545 | C | T | SNP | PODN | ENST00000312553 | 1 | intronic | c.1806-108 | e10-108 | tier3 | 19 | 0 | 0.000 | 52 | 21 | 0.288 |
| 611 | 1 | 63153953 | 63153953 | C | T | SNP | DOCK7 | ENST00000251157 | -1 | 5_prime_untranslated_region | c.-18 | NULL | tier4 | 14 | 0 | 0.000 | 25 | 19 | 0.432 |
| 611 | 2 | 25464481 | 25464481 | G | A | SNP | DNMT3A | ENST00000264709 | -1 | nonsense | c.2032 | p.Q678* | tier1 | 292 | 4 | 0.014 | 1146 | 70 | 0.057 |
| 611 | 3 | 31495819 | 31495819 | G | A | SNP | THRAP3P1 | ENST00000411753 | 1 | 3_prime_untranslated_region | c.*31495819 | NULL | tier3 | 462 | 0 | 0.000 | 769 | 311 | 0.288 |
| 611 | 3 | 64644370 | 64644370 | G | A | SNP | ADAMTS9 | ENST00000498707 | -1 | silent | c.777 | p.S259 | tier1 | 79 | 2 | 0.025 | 140 | 68 | 0.327 |
| 611 | 3 | 130720537 | 130720537 | T | A | SNP | ATP2C1 | ENST00000428331 | 1 | 3_prime_untranslated_region | c.*343 | NULL | tier4 | 42 | 0 | 0.000 | 159 | 91 | 0.364 |
| 611 | 4 | 187540539 | 187540539 | C | T | SNP | FAT1 | ENST00000441802 | -1 | missense | c.7201 | p.E2401K | tier1 | 33 | 1 | 0.029 | 94 | 36 | 0.277 |
| 611 | 6 | 33037020 | 33037020 | G | A | SNP | HLA-DPA1 | ENST00000419277 | -1 | missense | c.404 | p.T135I | tier1 | 57 | 0 | 0.000 | 138 | 77 | 0.358 |
| 611 | 6 | 43206066 | 43206066 | C | G | SNP | TTBK1 | ENST00000259750 | 1 | 5_prime_flanking_region | c.-8333 | NULL | tier4 | 58 | 0 | 0.000 | 177 | 49 | 0.217 |
| 611 | 7 | 45753309 | 45753309 | C | T | SNP | ADCY1 | ENST00000297323 | 1 | silent | c.3075 | p.H1025 | tier1 | 91 | 2 | 0.021 | 92 | 56 | 0.378 |
| 611 | 7 | 100549883 | 100549883 | G | A | SNP | LOC100131514 | ENST00000379458 | 1 | missense | c.464 | p.S155N | tier1 | 19 | 0 | 0.000 | 51 | 28 | 0.350 |
| 611 | 8 | 17944578 | 17944578 | C | T | SNP | LOC101929066 | ENST00000499554 | 1 | rna | NULL | NULL | tier1 | 85 | 0 | 0.000 | 182 | 79 | 0.303 |
| 611 | 8 | 72127533 | 72127533 | C | T | SNP | EYA1 | ENST00000340726 | -1 | intronic | c.1597+89 | e14+89 | tier3 | 74 | 2 | 0.026 | 192 | 62 | 0.244 |
| 611 | 8 | 116424437 | 116424437 | G | C | SNP | TRPS1 | ENST00000395715 | -1 | 3_prime_untranslated_region | c.*1814 | NULL | tier3 | 26 | 0 | 0.000 | 69 | 29 | 0.287 |
| 611 | 8 | 140768932 | 140768932 | C | T | SNP | TRAPPC9 | ENST00000389328 | -1 | intronic | c.3350-24487 | e22-24487 | tier3 | 64 | 1 | 0.015 | 97 | 35 | 0.263 |
| 611 | 9 | 103338826 | 103338826 | C | T | SNP | TMEFF1 | ENST00000374879 | -1 | nonsense | c.1087 | p.R363* | tier1 | 51 | 0 | 0.000 | 95 | 43 | 0.312 |
| 611 | 9 | 131502394 | 131502394 | G | A | SNP | ZER1 | ENST00000291900 | 1 | silent | c.1858 | p.L620 | tier1 | 31 | 0 | 0.000 | 76 | 22 | 0.225 |
| 611 | 11 | 6648113 | 6648113 | C | T | SNP | DCHS1 | ENST00000299441 | -1 | missense | c.6157 | p.V2053M | tier1 | 58 | 0 | 0.000 | 93 | 32 | 0.254 |
| 611 | 11 | 25004690 | 25004690 | A | G | SNP | LUZP2 | ENST00000336930 | 1 | missense | c.616 | p.M206V | tier1 | 58 | 0 | 0.000 | 121 | 18 | 0.130 |
| 611 | 11 | 125298875 | 125298875 | C | T | SNP | PKNOX2 | ENST00000298282 | 1 | intronic | c.937-33 | e8-33 | tier2 | 44 | 1 | 0.022 | 91 | 46 | 0.336 |
| 611 | 12 | 53553483 | 53553483 | T | C | SNP | CSAD | ENST00000267085 | -1 | missense | c.1313 | p.N438S | tier1 | 23 | 0 | 0.000 | 90 | 33 | 0.266 |
| 611 | 14 | 23072764 | 23072764 | T | A | SNP | ABHD4 | ENST00000539344 | 1 | 3_prime_untranslated_region | c.*23072764 | NULL | tier3 | 86 | 1 | 0.011 | 160 | 61 | 0.276 |
| 611 | 14 | 104216205 | 104216205 | G | A | SNP | PPP1R13B | ENST00000202556 | -1 | missense | c.895 | p.R299C | tier1 | 57 | 0 | 0.000 | 203 | 67 | 0.248 |
| 611 | 15 | 63615709 | 63615709 | A | - | DEL | CA12 | ENST00000178638 | -1 | 3_prime_untranslated_region | c.*2775 | NULL | tier2 | 45 | 1 | 0.022 | 77 | 24 | 0.238 |
| 611 | 15 | 66008017 | 66008017 | G | A | SNP | DENND4A | ENST00000443035 | -1 | intronic | c.1808-56 | e12-56 | tier4 | 34 | 0 | 0.000 | 99 | 42 | 0.296 |
| 611 | 16 | 3071729 | 3071729 | G | A | SNP | TNFRSF12A | ENST00000326577 | 1 | intronic | c.334+39 | e3+39 | tier2 | 27 | 0 | 0.000 | 76 | 28 | 0.269 |
| 611 | 17 | 3661144 | 3661144 | G | A | SNP | ITGAE | ENST00000263087 | -1 | silent | c.876 | p.L292 | tier1 | 56 | 0 | 0.000 | 142 | 43 | 0.232 |
| 611 | 17 | 5077452 | 5077452 | T | C | SNP | USP6 | ENST00000250066 | 1 | 3_prime_untranslated_region | c.*1179 | NULL | tier3 | 76 | 0 | 0.000 | 360 | 131 | 0.267 |
| 611 | 17 | 26087697 | 26087697 | G | T | SNP | NOS2 | ENST00000313735 | -1 | missense | c.2962 | p.P988T | tier1 | 160 | 2 | 0.012 | 318 | 166 | 0.342 |
| 611 | 17 | 29553477 | 29553478 | - | C | INS | NF1 | ENST00000358273 | 1 | frame_shift_ins | c.2026_2027 | p.I679fs | tier1 | 207 | 0 | 0.000 | 653 | 174 | 0.210 |
| 611 | 18 | 42531913 | 42531913 | G | A | SNP | SETBP1 | ENST00000282030 | 1 | missense | c.2608 | p.G870S | tier1 | 445 | 4 | 0.009 | 798 | 358 | 0.310 |
| 611 | 19 | 39787541 | 39787541 | C | T | SNP | IFNL1 | ENST00000333625 | 1 | intronic | c.249+19 | e2+19 | tier3 | 19 | 0 | 0.000 | 86 | 35 | 0.289 |
| 611 | 21 | 33684165 | 33684165 | A | G | SNP | MRAP | ENST00000303645 | 1 | missense | c.377 | p.E126G | tier1 | 30 | 0 | 0.000 | 78 | 29 | 0.271 |
| 611 | 22 | 21480989 | 21480989 | G | C | SNP | POM121L7 | ENST00000329949 | -1 | missense | c.937 | p.P313A | tier1 | 49 | 0 | 0.000 | 111 | 40 | 0.261 |
| 611 | 22 | 23922123 | 23922123 | G | A | SNP | IGLL1 | ENST00000330377 | -1 | intronic | c.206+49 | e1+49 | tier2 | 48 | 2 | 0.040 | 125 | 75 | 0.373 |
| 611 | X | 9757836 | 9757836 | C | T | SNP | SHROOM2 | ENST00000380913 | 1 | intronic | c.165+3086 | e1+3086 | tier3 | 98 | 0 | 0.000 | 203 | 45 | 0.182 |
| 130968 | 1 | 6101941 | 6101941 | T | A | SNP | KCNAB2 | ENST00000164247 | 1 | splice_region | c.119+9 | e2+9 | tier2 | 117 | 9 | 0.071 | 176 | 130 | 0.425 |
| 130968 | 1 | 115258747 | 115258747 | C | T | SNP | NRAS | ENST00000369535 | -1 | missense | c.35 | p.G12D | tier1 | 290 | 0 | 0.000 | 656 | 113 | 0.141 |
| 130968 | 1 | 154960473 | 154960473 | C | A | SNP | FLAD1 | ENST00000492620 | 1 | rna | NULL | NULL | tier1 | 32 | 0 | 0.000 | 96 | 22 | 0.186 |
| 130968 | 1 | 160144523 | 160144523 | C | T | SNP | ATP1A4 | ENST00000368081 | 1 | missense | c.2297 | p.T766M | tier1 | 59 | 0 | 0.000 | 75 | 41 | 0.350 |
| 130968 | 2 | 76975737 | 76975737 | G | A | SNP | LRRTM4 | ENST00000409911 | -1 | 3_prime_untranslated_region | c.*84 | NULL | tier3 | 61 | 0 | 0.000 | 110 | 32 | 0.222 |
| 130968 | 2 | 101554339 | 101554339 | C | T | SNP | NPAS2 | ENST00000542504 | 1 | intronic | c.558+35 | e5+35 | tier3 | 42 | 0 | 0.000 | 113 | 27 | 0.193 |
| 130968 | 3 | 121100282 | 121100282 | C | T | SNP | STXBP5L | ENST00000273666 | 1 | silent | c.2562 | p.F854 | tier1 | 89 | 2 | 0.022 | 233 | 38 | 0.140 |
| 130968 | 3 | 140281105 | 140281105 | C | T | SNP | CLSTN2 | ENST00000458420 | 1 | nonsense | c.2167 | p.R723* | tier1 | 55 | 0 | 0.000 | 159 | 38 | 0.193 |
| 130968 | 3 | 141595701 | 141595701 | A | G | SNP | ATP1B3 | ENST00000286371 | 1 | missense | c.58 | p.I20V | tier1 | 4 | 0 | 0.000 | 5 | 18 | 0.783 |
| 130968 | 3 | 150877579 | 150877579 | A | G | SNP | MED12L | ENST00000474524 | 1 | intronic | c.838-40 | e7-40 | tier3 | 48 | 0 | 0.000 | 132 | 21 | 0.137 |
| 130968 | 4 | 57389885 | 57389885 | T | G | SNP | ARL9 | ENST00000360096 | 1 | missense | c.215 | p.I72S | tier1 | 97 | 2 | 0.020 | 153 | 99 | 0.391 |
| 130968 | 4 | 106157023 | 106157023 | C | T | SNP | TET2 | ENST00000380013 | 1 | nonsense | c.1924 | p.Q642* | tier1 | 413 | 7 | 0.017 | 712 | 422 | 0.372 |
| 130968 | 4 | 106164913 | 106164913 | C | A | SNP | TET2 | ENST00000380013 | 1 | missense | c.3781 | p.R1261S | tier1 | 214 | 16 | 0.070 | 378 | 265 | 0.412 |
| 130968 | 4 | 187541079 | 187541079 | C | T | SNP | FAT1 | ENST00000441802 | -1 | missense | c.6661 | p.G2221R | tier1 | 55 | 0 | 0.000 | 99 | 28 | 0.221 |
| 130968 | 6 | 27840011 | 27840011 | T | A | SNP | HIST1H3I | ENST00000328488 | -1 | missense | c.83 | p.K28M | tier1 | 65 | 0 | 0.000 | 135 | 25 | 0.156 |
| 130968 | 6 | 56366476 | 56366476 | C | T | SNP | DST | ENST00000370754 | -1 | missense | c.19489 | p.A6497T | tier1 | 260 | 11 | 0.041 | 578 | 402 | 0.410 |
| 130968 | 6 | 123869713 | 123869713 | G | A | SNP | TRDN | ENST00000398178 | -1 | missense | c.277 | p.R93C | tier1 | 41 | 0 | 0.000 | 106 | 19 | 0.152 |
| 130968 | 7 | 30692952 | 30692953 | CC | - | DEL | CRHR2 | ENST00000348438 | -1 | 3_prime_untranslated_region | c.*124 | NULL | tier3 | 26 | 0 | 0.000 | 37 | 13 | 0.260 |
| 130968 | 7 | 30692957 | 30692965 | CTGGAGAGC | - | DEL | CRHR2 | ENST00000348438 | -1 | 3_prime_untranslated_region | c.*119 | NULL | tier3 | 28 | 0 | 0.000 | 38 | 13 | 0.255 |
| 130968 | 7 | 45140023 | 45140031 | GACCTAGAA | - | DEL | TBRG4 | ENST00000258770 | -1 | splice_site_del | c.1795-6_1795-1 | e10-1 | tier1 | 49 | 0 | 0.000 | 73 | 19 | 0.207 |
| 130968 | 7 | 135282052 | 135282052 | A | - | DEL | NUP205 | ENST00000462316 | 1 | 3_prime_untranslated_region | c.*135282052 | NULL | tier3 | 49 | 0 | 0.000 | 141 | 25 | 0.151 |
| 130968 | 10 | 126365553 | 126365553 | G | A | SNP | FAM53B | ENST00000337318 | -1 | intronic | c.906+4623 | e3+4623 | tier3 | 95 | 0 | 0.000 | 214 | 21 | 0.089 |
| 130968 | 11 | 121384952 | 121384952 | A | G | SNP | SORL1 | ENST00000260197 | 1 | missense | c.1133 | p.E378G | tier1 | 58 | 3 | 0.049 | 115 | 60 | 0.343 |
| 130968 | 12 | 51752848 | 51752848 | C | T | SNP | GALNT6 | ENST00000356317 | -1 | intronic | c.1368+68 | e6+68 | tier3 | 40 | 3 | 0.070 | 71 | 55 | 0.437 |
| 13 | | | | | | | | | | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|---------|----|-----|------------------------|-----------------|----|-----------------------------|------------------------|-----------|-------|-----|-----|-------|------|-----|-------|
| 130968 | 21 | 36252865 | 36252865 | C | T | SNP | <i>RUNX1</i> | ENST00000300305 | -1 | missense | c.497 | p.R166Q | tier1 | 743 | 7 | 0.009 | 1885 | 112 | 0.056 |
| 130968 | 21 | 39673268 | 39673268 | C | T | SNP | <i>KCNJ15</i> | ENST00000328656 | 1 | 3_prime_untranslated_region | c.*957 | NULL | tier3 | 39 | 1 | 0.025 | 43 | 40 | 0.482 |
| 130968 | X | 37028271 | 37028271 | G | A | SNP | <i>FAM47C</i> | ENST00000358047 | 1 | silent | c.1788 | p.P596 | tier1 | 144 | 1 | 0.007 | 218 | 83 | 0.276 |
| 130968 | X | 39913253 | 39913253 | G | - | DEL | <i>BCOR</i> | ENST00000378444 | -1 | frame_shift_del | c.4862 | p.P1621fs | tier1 | 267 | 6 | 0.022 | 604 | 232 | 0.278 |
| 130968 | X | 107512279 | 107512279 | C | T | SNP | <i>COL4A6</i> | ENST00000461897 | -1 | rna | NULL | NULL | tier1 | 40 | 0 | 0.000 | 60 | 32 | 0.348 |
| 130968 | X | 123197009 | 123197015 | CTCAGTA | - | DEL | <i>STAG2</i> | ENST00000218089 | 1 | frame_shift_del | c.1775_1781 | p.Q593fs | tier1 | 84 | 0 | 0.000 | 391 | 46 | 0.105 |
| 145094 | 1 | 16073346 | 16073346 | C | T | SNP | <i>TMEM82</i> | ENST00000375782 | 1 | intronic | c.758-16 | e5-16 | tier3 | 38 | 2 | 0.050 | 318 | 275 | 0.463 |
| 145094 | 1 | 186084343 | 186084343 | G | A | SNP | <i>HMCN1</i> | ENST00000271588 | 1 | intronic | c.11405-47 | e75-47 | tier3 | 181 | 1 | 0.006 | 442 | 331 | 0.428 |
| 145094 | 1 | 200880497 | 200880497 | C | T | SNP | <i>C1orf106</i> | ENST00000367342 | 1 | intronic | c.1215-84 | e9-84 | tier2 | 72 | 0 | 0.000 | 200 | 149 | 0.427 |
| 145094 | 1 | 206945563 | 206945563 | C | G | SNP | <i>IL10</i> | ENST00000423557 | -1 | intronic | c.165+53 | e1+53 | tier3 | 44 | 0 | 0.000 | 87 | 63 | 0.420 |
| 145094 | 2 | 116520209 | 116520209 | C | T | SNP | <i>DPP10</i> | ENST00000393147 | 1 | intronic | c.1125+23 | e12+23 | tier3 | 63 | 1 | 0.016 | 93 | 84 | 0.475 |
| 145094 | 2 | 122004343 | 122004343 | C | G | SNP | <i>TFCP2L1</i> | ENST00000263707 | -1 | intronic | c.657+51 | e6+51 | tier3 | 21 | 0 | 0.000 | 80 | 54 | 0.403 |
| 145094 | 2 | 233651863 | 233651863 | G | A | SNP | <i>GIGYF2</i> | ENST00000373566 | 1 | missense | c.602 | p.R201K | tier1 | 256 | 0 | 0.000 | 502 | 463 | 0.479 |
| 145094 | 4 | 48563681 | 48563681 | G | C | SNP | <i>FRYL</i> | ENST00000358350 | -1 | intronic | c.3697-28 | e30-28 | tier3 | 126 | 2 | 0.016 | 416 | 358 | 0.462 |
| 145094 | 4 | 54880073 | 54880073 | T | C | SNP | <i>CHIC2</i> | ENST00000263921 | -1 | splice_region | c.388-10 | e5-10 | tier3 | 95 | 0 | 0.000 | 149 | 122 | 0.450 |
| 145094 | 5 | 98868390 | 98868390 | A | G | SNP | <i>GUSBP8</i> | ENST00000502652 | 1 | 3_prime_untranslated_region | c.*98868390 | NULL | tier3 | 204 | 4 | 0.019 | 38 | 24 | 0.387 |
| 145094 | 6 | 131211458 | 131211458 | G | A | SNP | <i>EPB41L2</i> | ENST00000337057 | -1 | missense | c.1636 | p.R546W | tier1 | 81 | 0 | 0.000 | 196 | 154 | 0.440 |
| 145094 | 7 | 2962258 | 2962258 | G | A | SNP | <i>CARD11</i> | ENST00000396946 | -1 | splice_region | c.2269+10 | e16+10 | tier3 | 20 | 0 | 0.000 | 84 | 56 | 0.400 |
| 145094 | 7 | 128520034 | 128520034 | G | A | SNP | <i>KCP</i> | ENST00000297801 | -1 | 5_prime_untranslated_region | c.-128520034 | NULL | tier2 | 30 | 0 | 0.000 | 60 | 32 | 0.348 |
| 145094 | 8 | 30702831 | 30702831 | C | G | SNP | <i>TEX15</i> | ENST00000256246 | -1 | missense | c.3703 | p.V1235L | tier1 | 85 | 0 | 0.000 | 99 | 70 | 0.414 |
| 145094 | 8 | 86573767 | 86573767 | C | T | SNP | <i>REXO1L1</i> | ENST00000379010 | -1 | missense | c.1960 | p.V654I | tier1 | 133 | 0 | 0.000 | 1637 | 135 | 0.076 |
| 145094 | 8 | 134239796 | 134239796 | A | G | SNP | <i>WISP1</i> | ENST00000250160 | 1 | missense | c.947 | p.K316R | tier1 | 48 | 0 | 0.000 | 105 | 91 | 0.464 |
| 145094 | 9 | 35079180 | 35079180 | T | G | SNP | <i>FANCG</i> | ENST00000378643 | -1 | missense | c.143 | p.E48A | tier1 | 26 | 0 | 0.000 | 94 | 84 | 0.472 |
| 145094 | 9 | 126029252 | 126029252 | C | - | DEL | <i>STRBP</i> | ENST00000348403 | -1 | intronic | c.1+1463 | e0+1463 | tier3 | 38 | 0 | 0.000 | 62 | 45 | 0.421 |
| 145094 | 9 | 131115881 | 131115881 | G | A | SNP | <i>SLC27A4</i> | ENST00000300456 | 1 | intronic | c.1324+61 | e8+61 | tier3 | 20 | 0 | 0.000 | 102 | 105 | 0.507 |
| 145094 | 10 | 18829133 | 18829133 | G | C | SNP | <i>CACNB2</i> | ENST00000324631 | 1 | 3_prime_untranslated_region | c.*480 | NULL | tier3 | 88 | 0 | 0.000 | 93 | 95 | 0.505 |
| 145094 | 10 | 75526485 | 75526485 | A | G | SNP | <i>SEC24C</i> | ENST00000339365 | 1 | intronic | c.1800-33 | e12-33 | tier3 | 78 | 0 | 0.000 | 100 | 107 | 0.517 |
| 145094 | 11 | 12183811 | 12183811 | C | T | SNP | <i>MICAL2</i> | ENST00000256194 | 1 | nonsense | c.109 | p.R37* | tier1 | 81 | 0 | 0.000 | 197 | 165 | 0.456 |
| 145094 | 12 | 120638934 | 120638934 | C | G | SNP | <i>RPLP0</i> | ENST00000228306 | -1 | 5_prime_untranslated_region | c.-348 | NULL | tier2 | 81 | 0 | 0.000 | 302 | 302 | 0.499 |
| 145094 | 15 | 45266191 | 45266191 | A | G | SNP | <i>C15orf43</i> | ENST00000340827 | 1 | intronic | c.523+38 | e6+38 | tier2 | 70 | 0 | 0.000 | 118 | 98 | 0.454 |
| 145094 | 17 | 7577120 | 7577120 | C | T | SNP | <i>TP53</i> | ENST00000269305 | -1 | missense | c.818 | p.R273H | tier1 | 195 | 5 | 0.025 | 66 | 803 | 0.923 |
| 145094 | 17 | 29552142 | 29552142 | - | G | INS | <i>NF1</i> | ENST00000358273 | 1 | frame_shift_ins | c.1875_1876 | p.L625fs | tier1 | 163 | 0 | 0.000 | 91 | 316 | 0.776 |
| 145094 | 17 | 29552143 | 29552143 | C | A | SNP | <i>NF1</i> | ENST00000358273 | 1 | missense | c.1876 | p.L626I | tier1 | 163 | 2 | 0.012 | 53 | 336 | 0.818 |
| 145094 | 17 | 45234327 | 45234327 | C | T | SNP | <i>CDC27</i> | ENST00000531206 | -1 | missense | c.794 | p.G265D | tier1 | 126 | 0 | 0.000 | 299 | 19 | 0.060 |
| 145094 | 21 | 44324363 | 44324363 | G | A | SNP | <i>NDUFV3</i> | ENST00000354250 | 1 | missense | c.1241 | p.G414E | tier1 | 47 | 1 | 0.021 | 96 | 77 | 0.445 |
| 145094 | X | 18845595 | 18845595 | A | T | SNP | <i>PPEF1</i> | ENST00000361511 | 1 | missense | c.1952 | p.N651I | tier1 | 61 | 1 | 0.016 | 4 | 106 | 0.964 |
| 147444 | 1 | 34164429 | 34164429 | G | T | SNP | <i>CSMD2</i> | ENST00000373381 | -1 | missense | c.3849 | p.S1283R | tier1 | 80 | 0 | 0.000 | 126 | 38 | 0.232 |
| 147444 | 1 | 38262323 | 38262323 | C | A | SNP | <i>MANEAL</i> | ENST00000373045 | 1 | intronic | c.661-93 | e3-93 | tier3 | 39 | 0 | 0.000 | 55 | 19 | 0.253 |
| 147444 | 1 | 89480937 | 89480937 | G | A | SNP | <i>GBP3</i> | ENST00000370481 | -1 | intronic | c.318+33 | e2+33 | tier3 | 190 | 0 | 0.000 | 225 | 72 | 0.242 |
| 147444 | 1 | 120612014 | 120612014 | C | A | SNP | <i>NOTCH2</i> | ENST00000256646 | -1 | missense | c.7 | p.A3S | tier1 | 41 | 0 | 0.000 | 87 | 16 | 0.155 |
| 147444 | 1 | 163306613 | 163306613 | C | T | SNP | <i>NUF2</i> | ENST00000271452 | 1 | missense | c.410 | p.T137M | tier1 | 84 | 1 | 0.012 | 67 | 17 | 0.202 |
| 147444 | 1 | 201016781 | 201016781 | C | T | SNP | <i>CACNA1S</i> | ENST00000362061 | -1 | intronic | c.4442-27 | e37-27 | tier3 | 57 | 0 | 0.000 | 70 | 22 | 0.239 |
| 147444 | 1 | 226061960 | 226061960 | G | A | SNP | <i>TMEM63A</i> | ENST00000366835 | -1 | intronic | c.266+28 | e2+28 | tier3 | 30 | 0 | 0.000 | 50 | 17 | 0.254 |
| 147444 | 2 | 21229034 | 21229034 | T | - | DEL | <i>APOB</i> | ENST00000233242 | -1 | frame_shift_del | c.10706 | p.N3569fs | tier1 | 776 | 0 | 0.000 | 783 | 262 | 0.251 |
| 147444 | 2 | 42119915 | 42119915 | C | A | SNP | <i>LOC388942</i> | ENST00000398796 | 1 | rna | NULL | NULL | tier1 | 39 | 0 | 0.000 | 45 | 10 | 0.182 |
| 147444 | 2 | 42936013 | 42936013 | G | T | SNP | <i>MTA3</i> | ENST00000405094 | 1 | splice_site | c.1303-1 | e14-1 | tier1 | 166 | 0 | 0.000 | 151 | 52 | 0.256 |
| 147444 | 2 | 128262356 | 128262356 | T | C | SNP | <i>IWS1</i> | ENST00000295321 | -1 | missense | c.1123 | p.M375V | tier1 | 89 | 0 | 0.000 | 70 | 29 | 0.293 |
| 147444 | 2 | 138000072 | 138000072 | C | T | SNP | <i>THSD7B</i> | ENST00000272643 | 1 | silent | c.2196 | p.L732 | tier1 | 157 | 0 | 0.000 | 217 | 77 | 0.262 |
| 147444 | 2 | 216972458 | 216972458 | C | G | SNP | <i>XRCC5</i> | ENST00000392133 | 1 | intronic | c.1+37 | e0+37 | tier2 | 57 | 0 | 0.000 | 70 | 21 | 0.231 |
| 147444 | 3 | 158306755 | 158306755 | A | T | SNP | <i>MLF1</i> | ENST00000392822 | 1 | intronic | c.96-3468 | e2-3468 | tier4 | 82 | 0 | 0.000 | 54 | 25 | 0.317 |
| 147444 | 4 | 74352793 | 74352793 | A | - | DEL | <i>AFM</i> | ENST00000226355 | 1 | frame_shift_del | c.592 | p.V199fs | tier1 | 83 | 0 | 0.000 | 84 | 35 | 0.294 |
| 147444 | 4 | 100761543 | 100761543 | C | A | SNP | <i>DAPP1</i> | ENST00000512369 | 1 | missense | c.322 | p.H108N | tier1 | 92 | 0 | 0.000 | 61 | 12 | 0.164 |
| 147444 | 4 | 149358015 | 149358015 | C | T | SNP | <i>NR3C2</i> | ENST00000355292 | -1 | splice_site | c.1-1 | e1-1 | tier1 | 63 | 0 | 0.000 | 86 | 23 | 0.209 |
| 147444 | 5 | 1331845 | 1331845 | C | T | SNP | <i>CLPTM1L</i> | ENST00000503042 | -1 | 5_prime_untranslated_region | c.-1331845 | NULL | tier3 | 30 | 0 | 0.000 | 42 | 14 | 0.246 |
| 147444 | 5 | 38464253 | 38464253 | C | T | SNP | <i>EGFLAM</i> | ENST00000354891 | 1 | 3_prime_untranslated_region | c.*165 | NULL | tier3 | 21 | 0 | 0.000 | 30 | 12 | 0.286 |
| 147444 | 5 | 39331903 | 39331903 | C | T | SNP | <i>C9</i> | ENST00000263408 | -1 | missense | c.490 | p.G164R | tier1 | 80 | 1 | 0.012 | 80 | 33 | 0.292 |
| 147444 | 6 | 39841127 | 39841127 | G | A | SNP | <i>DAAM2</i> | ENST00000274867 | 1 | missense | c.1158 | p.M386I | tier1 | 34 | 0 | 0.000 | 32 | 16 | 0.333 |
| 147444 | 6 | 72086766 | 72086766 | A | G | SNP | <i>LINC00472</i> | ENST00000413945 | -1 | intronic | c.NULL | NULL | tier3 | 80 | 0 | 0.000 | 79 | 15 | 0.160 |
| 147444 | 6 | 153603445 | 153603445 | C | T | SNP | <i>ENSG00000218426</i> | ENST00000492974 | -1 | 5_prime_untranslated_region | c.-153603445 | NULL | tier2 | 93 | 0 | 0.000 | 87 | 36 | 0.293 |
| 147444 | 6 | 167571516 | 167571516 | G | A | SNP | <i>GPR31</i> | ENST00000366834 | -1 | 5_prime_untranslated_region | c.-197 | NULL | tier3 | 92 | 0 | 0.000 | 91 | 31 | 0.254 |
| 147444 | 7 | 57522854 | 57522854 | A | T | SNP | <i>ZNF716</i> | ENST00000420713 | 1 | missense | c.242 | p.E81V | tier1 | 144 | 0 | 0.000 | 66 | 23 | 0.258 |
| 147444 | 7 | 100190385 | 100190385 | C | T | SNP | <i>FBXO24</i> | ENST00000427939 | 1 | intronic | c.673-21 | e5-21 | tier3 | 102 | 0 | 0.000 | 89 | 62 | 0.408 |
| 147444 | 7 | 157650971 | 157650971 | - | CT | INS | <i>PTPRN2</i> | ENST00000404321 | -1 | intronic | c.1857+40393_1857+4039 | e12+40393 | tier3 | 52 | 1 | 0.019 | 36 | 11 | 0.234 |
| 147444 | 9 | 33557962 | 33557962 | T | C | SNP | <i>ANKRD18B</i> | ENST00000290943 | 1 | intronic | c.2145-94 | e12-94 | tier3 | 85 | 0 | 0.000 | 59 | 14 | 0.192 |
| 147444 | 9 | 43628454 | 43628454 | G | A | SNP | <i>SPATA31A6</i> | ENST00000332857 | -1 | intronic | c.309-76 | e4-76 | tier3 | 120 | 0 | 0.000 | 135 | 44 | 0.246 |
| 147444 | 10 | 24762962 | 24762962 | C | T | SNP | <i>KIAA1217</i> | ENST00000376454 | 1 | missense | c.1652 | p.A551V | tier1 | 63 | 2 | 0.031 | 109 | 27 | 0.199 |
| 147444 | 10 | 46148449 | 46148449 | A | T | SNP | <i>ZFAND4</i> | ENST00000344646 | -1 | nonsense | c.243 | p.Y81* | tier1 | 39 | 0</ | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|---|---|-----|-----------------|-----------------|----|-----------------------------|--------------|----------|-------|------|---|-------|------|------|-------|
| 147444 | 16 | 4384836 | 4384836 | C | T | SNP | GLIS2 | ENST00000262366 | 1 | missense | c.380 | p.P127L | tier1 | 97 | 0 | 0.000 | 120 | 35 | 0.224 |
| 147444 | 16 | 68721437 | 68721437 | G | A | SNP | CDH3 | ENST00000264012 | 1 | silent | c.1593 | p.T531 | tier1 | 41 | 0 | 0.000 | 45 | 36 | 0.444 |
| 147444 | 17 | 6902088 | 6902088 | T | A | SNP | ALOX12 | ENST00000251535 | 1 | missense | c.474 | p.D158E | tier1 | 116 | 0 | 0.000 | 147 | 31 | 0.174 |
| 147444 | 17 | 7578212 | 7578212 | G | A | SNP | TP53 | ENST00000269305 | -1 | nonsense | c.637 | p.R213* | tier1 | 1150 | 2 | 0.002 | 981 | 1049 | 0.515 |
| 147444 | 17 | 42981590 | 42981590 | C | T | SNP | FAM187A | ENST00000331733 | 1 | silent | c.393 | p.D131 | tier1 | 39 | 0 | 0.000 | 49 | 13 | 0.210 |
| 147444 | 17 | 67079228 | 67079228 | T | C | SNP | ABCA6 | ENST00000284425 | -1 | intronic | c.4476-74 | e35-74 | tier3 | 110 | 0 | 0.000 | 93 | 31 | 0.250 |
| 147444 | 19 | 15511862 | 15511862 | A | G | SNP | AKAP8L | ENST00000397410 | -1 | splice_region | c.817-8 | e6-8 | tier3 | 41 | 0 | 0.000 | 80 | 21 | 0.208 |
| 147444 | 22 | 29886361 | 29886361 | C | T | SNP | NEFH | ENST00000310624 | 1 | missense | c.2732 | p.P911L | tier1 | 39 | 0 | 0.000 | 46 | 16 | 0.258 |
| 147444 | X | 49857254 | 49857254 | C | T | SNP | CLCN5 | ENST00000376088 | 1 | 3_prime_untranslated_region | c.*378 | NULL | tier3 | 70 | 1 | 0.014 | 33 | 15 | 0.313 |
| 147457 | 1 | 809179 | 809179 | G | A | SNP | ENSG00000234711 | ENST00000415481 | 1 | 3_prime_untranslated_region | c.*809179 | NULL | tier2 | 69 | 0 | 0.000 | 523 | 203 | 0.279 |
| 147457 | 1 | 1275514 | 1275514 | G | A | SNP | DVL1 | ENST00000378888 | -1 | silent | c.813 | p.N271 | tier1 | 38 | 1 | 0.025 | 174 | 77 | 0.307 |
| 147457 | 1 | 1861568 | 1861568 | G | A | SNP | C1orf222 | ENST00000493964 | -1 | silent | c.1068 | p.F356 | tier1 | 35 | 0 | 0.000 | 212 | 88 | 0.293 |
| 147457 | 1 | 6021933 | 6021933 | C | T | SNP | NPHP4 | ENST00000378156 | -1 | silent | c.594 | p.A198 | tier1 | 29 | 0 | 0.000 | 143 | 57 | 0.285 |
| 147457 | 1 | 6532561 | 6532561 | C | T | SNP | PLEKHG5 | ENST00000537245 | -1 | intronic | c.1317+26 | e11+26 | tier2 | 23 | 0 | 0.000 | 110 | 39 | 0.262 |
| 147457 | 1 | 9790439 | 9790439 | C | T | SNP | CLSTN1 | ENST00000477264 | -1 | 5_prime_untranslated_region | c.-9790439 | NULL | tier2 | 35 | 1 | 0.028 | 270 | 116 | 0.299 |
| 147457 | 1 | 12908093 | 12908093 | C | T | SNP | HNRNPCL1 | ENST00000317869 | -1 | missense | c.50 | p.R17H | tier1 | 490 | 1 | 0.002 | 1500 | 165 | 0.099 |
| 147457 | 1 | 15701093 | 15701093 | G | A | SNP | FHAD1 | ENST00000481324 | 1 | missense | c.526 | p.A176T | tier1 | 29 | 0 | 0.000 | 139 | 55 | 0.284 |
| 147457 | 1 | 21605671 | 21605671 | C | T | SNP | ECE1 | ENST00000374893 | -1 | intronic | c.280+13 | e3+13 | tier3 | 16 | 0 | 0.000 | 215 | 97 | 0.311 |
| 147457 | 1 | 22852716 | 22852716 | G | A | SNP | ZBTB40 | ENST00000375647 | 1 | missense | c.3547 | p.V1183M | tier1 | 20 | 0 | 0.000 | 195 | 75 | 0.278 |
| 147457 | 1 | 22902834 | 22902834 | G | A | SNP | EPHA8 | ENST00000166244 | 1 | missense | c.284 | p.R95Q | tier1 | 83 | 0 | 0.000 | 211 | 64 | 0.230 |
| 147457 | 1 | 24385364 | 24385364 | G | A | SNP | MYOM3 | ENST00000330966 | -1 | intronic | c.4059+50 | e35+50 | tier3 | 109 | 0 | 0.000 | 485 | 204 | 0.296 |
| 147457 | 1 | 24410953 | 24410953 | C | T | SNP | MYOM3 | ENST00000475306 | -1 | 5_prime_untranslated_region | c.-24410953 | NULL | tier3 | 227 | 3 | 0.013 | 635 | 306 | 0.325 |
| 147457 | 1 | 24671443 | 24671443 | C | T | SNP | GRHL3 | ENST00000350501 | 1 | intronic | c.1454+22 | e12+22 | tier3 | 21 | 0 | 0.000 | 94 | 42 | 0.309 |
| 147457 | 1 | 26773790 | 26773790 | G | A | SNP | DHDDS | ENST00000360009 | 1 | intronic | c.441-260 | e5-260 | tier4 | 191 | 5 | 0.025 | 142 | 49 | 0.257 |
| 147457 | 1 | 27220835 | 27220835 | G | A | SNP | GPATCH3 | ENST00000361720 | -1 | missense | c.943 | p.R315W | tier1 | 60 | 0 | 0.000 | 180 | 90 | 0.333 |
| 147457 | 1 | 27708286 | 27708286 | C | T | SNP | CD164L2 | ENST00000374030 | -1 | silent | c.357 | p.P119 | tier1 | 49 | 0 | 0.000 | 164 | 85 | 0.341 |
| 147457 | 1 | 29365837 | 29365837 | G | A | SNP | EPB41 | ENST00000343067 | 1 | missense | c.1535 | p.R512Q | tier1 | 91 | 0 | 0.000 | 271 | 103 | 0.275 |
| 147457 | 1 | 29609283 | 29609283 | C | T | SNP | PTPRU | ENST00000345512 | 1 | missense | c.1964 | p.A655V | tier1 | 26 | 1 | 0.037 | 143 | 75 | 0.343 |
| 147457 | 1 | 31186218 | 31186218 | G | A | SNP | MATN1 | ENST00000373765 | -1 | 3_prime_untranslated_region | c.*211 | NULL | tier3 | 92 | 1 | 0.011 | 219 | 81 | 0.270 |
| 147457 | 1 | 32090677 | 32090677 | G | A | SNP | HCRTR1 | ENST00000373706 | 1 | missense | c.1045 | p.A349T | tier1 | 23 | 0 | 0.000 | 113 | 54 | 0.321 |
| 147457 | 1 | 37316001 | 37316001 | C | T | SNP | GRIK3 | ENST00000373091 | -1 | missense | c.1237 | p.G413R | tier1 | 33 | 0 | 0.000 | 188 | 71 | 0.274 |
| 147457 | 1 | 43618426 | 43618426 | C | T | SNP | FAM183A | ENST00000335282 | 1 | intronic | c.194-73 | e3-73 | tier3 | 34 | 2 | 0.056 | 115 | 56 | 0.328 |
| 147457 | 1 | 43897426 | 43897426 | C | T | SNP | SZT2 | ENST00000562955 | 1 | nonsense | c.4957 | p.R1653* | tier1 | 24 | 0 | 0.000 | 107 | 47 | 0.305 |
| 147457 | 1 | 46206734 | 46206734 | C | T | SNP | IPP | ENST00000396478 | -1 | missense | c.563 | p.R188Q | tier1 | 101 | 0 | 0.000 | 175 | 68 | 0.280 |
| 147457 | 1 | 52826134 | 52826134 | C | T | SNP | CC2D1B | ENST00000371586 | -1 | missense | c.565 | p.E189K | tier1 | 20 | 0 | 0.000 | 242 | 128 | 0.344 |
| 147457 | 1 | 54670978 | 54670978 | G | A | SNP | MRPL37 | ENST00000360840 | 1 | missense | c.541 | p.V181M | tier1 | 41 | 0 | 0.000 | 107 | 48 | 0.310 |
| 147457 | 1 | 57085113 | 57085113 | C | T | SNP | ENSG00000225475 | ENST00000442812 | -1 | 5_prime_untranslated_region | c.-57085113 | NULL | tier3 | 233 | 2 | 0.009 | 451 | 206 | 0.313 |
| 147457 | 1 | 74905278 | 74905278 | C | T | SNP | FPGT-TNNI3K | ENST00000370891 | 1 | silent | c.2286 | p.G762 | tier1 | 109 | 0 | 0.000 | 169 | 76 | 0.309 |
| 147457 | 1 | 78031830 | 78031830 | G | A | SNP | ZZZ3 | ENST00000370801 | -1 | missense | c.2503 | p.R835W | tier1 | 147 | 1 | 0.007 | 282 | 133 | 0.321 |
| 147457 | 1 | 110217523 | 110217523 | C | T | SNP | GSTM2 | ENST00000241337 | 1 | 3_prime_untranslated_region | c.*65 | NULL | tier3 | 16 | 0 | 0.000 | 149 | 64 | 0.301 |
| 147457 | 1 | 116609238 | 116609238 | C | T | SNP | SLC22A15 | ENST00000369503 | 1 | missense | c.1463 | p.P488L | tier1 | 132 | 0 | 0.000 | 222 | 95 | 0.300 |
| 147457 | 1 | 149914970 | 149914970 | C | T | SNP | OTUD7B | ENST00000369135 | -1 | 3_prime_untranslated_region | c.*786 | NULL | tier3 | 36 | 0 | 0.000 | 100 | 49 | 0.329 |
| 147457 | 1 | 151507600 | 151507600 | G | A | SNP | CGN | ENST00000271636 | 1 | missense | c.2987 | p.R996Q | tier1 | 52 | 0 | 0.000 | 133 | 67 | 0.333 |
| 147457 | 1 | 155019719 | 155019719 | C | T | SNP | DCST1 | ENST00000295542 | 1 | missense | c.1543 | p.R515W | tier1 | 78 | 1 | 0.013 | 288 | 125 | 0.303 |
| 147457 | 1 | 155584452 | 155584452 | C | T | SNP | MSTO1 | ENST00000245564 | 1 | 3_prime_untranslated_region | c.*388 | NULL | tier3 | 48 | 1 | 0.020 | 203 | 91 | 0.309 |
| 147457 | 1 | 156437379 | 156437379 | G | A | SNP | MEF2D | ENST00000464356 | -1 | 5_prime_untranslated_region | c.-156437379 | NULL | tier3 | 58 | 2 | 0.033 | 382 | 175 | 0.314 |
| 147457 | 1 | 156713349 | 156713349 | G | A | SNP | HDGF | ENST00000477306 | -1 | 5_prime_untranslated_region | c.-156713349 | NULL | tier3 | 24 | 1 | 0.040 | 135 | 75 | 0.356 |
| 147457 | 1 | 160604556 | 160604556 | G | A | SNP | SLAMF1 | ENST00000302035 | -1 | missense | c.547 | p.H183Y | tier1 | 53 | 1 | 0.019 | 230 | 124 | 0.348 |
| 147457 | 1 | 162492585 | 162492585 | C | T | SNP | UHMK1 | ENST00000489294 | 1 | 3_prime_untranslated_region | c.*245 | NULL | tier3 | 47 | 0 | 0.000 | 106 | 47 | 0.307 |
| 147457 | 1 | 165182958 | 165182958 | G | A | SNP | LMX1A | ENST00000294816 | -1 | missense | c.589 | p.R197C | tier1 | 120 | 0 | 0.000 | 220 | 83 | 0.273 |
| 147457 | 1 | 171308645 | 171308645 | C | T | SNP | TOP1P1 | ENST00000404238 | 1 | 3_prime_untranslated_region | c.*171308645 | NULL | tier3 | 127 | 5 | 0.038 | 261 | 121 | 0.316 |
| 147457 | 1 | 172410952 | 172410952 | G | A | SNP | PIGC | ENST00000258324 | -1 | missense | c.811 | p.R271C | tier1 | 122 | 0 | 0.000 | 184 | 77 | 0.294 |
| 147457 | 1 | 175014844 | 175014844 | G | A | SNP | TNN | ENST00000239462 | 1 | 5_prime_flanking_region | c.-31711 | NULL | tier2 | 85 | 0 | 0.000 | 101 | 54 | 0.346 |
| 147457 | 1 | 186010193 | 186010193 | G | A | SNP | HMCN1 | ENST00000271588 | 1 | missense | c.6229 | p.D2077N | tier1 | 111 | 3 | 0.026 | 374 | 166 | 0.307 |
| 147457 | 1 | 200183198 | 200183198 | C | T | SNP | FAM58BP | ENST00000424019 | 1 | 3_prime_untranslated_region | c.*200183198 | NULL | tier2 | 34 | 1 | 0.029 | 242 | 88 | 0.267 |
| 147457 | 1 | 201168470 | 201168470 | C | T | SNP | IGFN1 | ENST00000335211 | 1 | missense | c.376 | p.R126W | tier1 | 40 | 1 | 0.024 | 144 | 75 | 0.343 |
| 147457 | 1 | 201184183 | 201184183 | G | A | SNP | IGFN1 | ENST00000335211 | 1 | missense | c.8896 | p.V2966M | tier1 | 16 | 0 | 0.000 | 134 | 56 | 0.293 |
| 147457 | 1 | 201983055 | 201983055 | G | A | SNP | ELF3 | ENST00000359651 | 1 | missense | c.904 | p.V302I | tier1 | 18 | 0 | 0.000 | 113 | 52 | 0.313 |
| 147457 | 1 | 205064086 | 205064086 | C | T | SNP | RBBP5 | ENST00000264515 | -1 | silent | c.1503 | p.P501 | tier1 | 92 | 0 | 0.000 | 224 | 72 | 0.243 |
| 147457 | 1 | 213058620 | 213058620 | G | A | SNP | FLVCR1 | ENST00000366971 | 1 | intronic | c.1093-15 | e5-15 | tier3 | 150 | 1 | 0.007 | 217 | 98 | 0.311 |
| 147457 | 1 | 215298011 | 215298011 | G | A | SNP | KCNK2 | ENST00000444842 | 1 | silent | c.393 | p.P131 | tier1 | 103 | 2 | 0.019 | 216 | 85 | 0.282 |
| 147457 | 1 | 220340567 | 220340567 | G | A | SNP | RAB3GAP2 | ENST00000358951 | -1 | intronic | c.3087+68 | e26+68 | tier3 | 59 | 1 | 0.017 | 67 | 32 | 0.323 |
| 147457 | 1 | 228033658 | 228033658 | G | A | SNP | PRSS38 | ENST00000366757 | 1 | missense | c.730 | p.D244N | tier1 | 22 | 0 | 0.000 | 70 | 24 | 0.255 |
| 147457 | 1 | 232944031 | 232944031 | G | A | SNP | KIAA1383 | ENST00000418460 | 1 | 3_prime_untranslated_region | c.*118 | NULL | tier3 | 115 | 0 | 0.000 | 187 | 75 | 0.286 |
| 147457 | 1 | 236898908 | 236898908 | G | A | SNP | ACTN2 | ENST00000366578 | 1 | intronic | c.698-27 | e8-27 | tier3 | 48 | 0 | 0.000 | 62 | 29 | 0.319 |
| 147457 | 1 | 244230451 | 244230451 | C | T | SNP | ENSG00000237759 | ENST00000440148 | 1 | 5_prime_flanking_region | c.-40216 | NULL | tier3 | 53 | 2 | 0.036 | 162 | 66 | 0.290 |
| 147457 | 2 | 1271318 | 1271318 | C | T | SNP | SNTG2 | ENST00000308624 | 1 | missense | c.1259 | p.T420M | tier1 | 48 | 0 | 0.000 | 112 | 46 | 0.291 |
| 147457 | 2 | 1652185 | 1652185 | C | T | SNP | PXDN | ENST00000252804 | -1 | missense | c.3367 | p.G1123R | tier1 | 22 | 0 | 0.000 | 153 | 69 | 0.308 |
| 147457 | 2 | 3469478 | 3469478 | C | T | SNP | | | | | | | | | | | | | |

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|--------|---|-----------|-----------|---|---|-----|------------------------|-----------------|----|-----------------------------|--------------|-----------|-------|-----|---|-------|------|-----|-------|
| 147457 | 2 | 25466800 | 25466800 | G | A | SNP | <i>DNMT3A</i> | ENST00000264709 | -1 | missense | c.1903 | p.R635W | tier1 | 165 | 2 | 0.012 | 704 | 291 | 0.292 |
| 147457 | 2 | 25469932 | 25469932 | G | A | SNP | <i>DNMT3A</i> | ENST00000264709 | -1 | silent | c.1110 | p.Y370 | tier1 | 143 | 1 | 0.007 | 570 | 249 | 0.304 |
| 147457 | 2 | 26177162 | 26177162 | G | A | SNP | <i>KIF3C</i> | ENST00000264712 | -1 | silent | c.1863 | p.N621 | tier1 | 25 | 0 | 0.000 | 165 | 46 | 0.217 |
| 147457 | 2 | 26644243 | 26644243 | G | A | SNP | <i>CCDC164</i> | ENST00000288710 | 1 | missense | c.331 | p.E111K | tier1 | 120 | 3 | 0.024 | 190 | 89 | 0.319 |
| 147457 | 2 | 26800492 | 26800492 | G | A | SNP | <i>C2orf70</i> | ENST00000329615 | 1 | missense | c.457 | p.V153I | tier1 | 28 | 0 | 0.000 | 185 | 71 | 0.276 |
| 147457 | 2 | 27261955 | 27261955 | C | T | SNP | <i>TMEM214</i> | ENST00000238788 | 1 | missense | c.1448 | p.T483M | tier1 | 13 | 0 | 0.000 | 145 | 75 | 0.339 |
| 147457 | 2 | 27310227 | 27310227 | C | T | SNP | <i>KHK</i> | ENST00000260598 | 1 | splice_region | c.92+8 | e1+8 | tier2 | 34 | 0 | 0.000 | 181 | 60 | 0.248 |
| 147457 | 2 | 27592008 | 27592008 | G | A | SNP | <i>EIF2B4</i> | ENST00000347454 | -1 | missense | c.283 | p.R95W | tier1 | 25 | 0 | 0.000 | 83 | 37 | 0.308 |
| 147457 | 2 | 27801023 | 27801023 | G | A | SNP | <i>C2orf16</i> | ENST00000408964 | 1 | silent | c.1584 | p.T528 | tier1 | 120 | 0 | 0.000 | 263 | 94 | 0.263 |
| 147457 | 2 | 56403145 | 56403145 | C | T | SNP | <i>LOC100129434</i> | ENST00000432793 | -1 | 5_prime_untranslated_region | c.-56403145 | NULL | tier2 | 94 | 2 | 0.021 | 157 | 74 | 0.320 |
| 147457 | 2 | 58459235 | 58459235 | G | A | SNP | <i>FANCL</i> | ENST00000233741 | -1 | missense | c.109 | p.H37Y | tier1 | 61 | 1 | 0.016 | 143 | 63 | 0.306 |
| 147457 | 2 | 61331050 | 61331050 | G | A | SNP | <i>KIAA1841</i> | ENST00000356719 | 1 | silent | c.1428 | p.P476 | tier1 | 97 | 1 | 0.010 | 244 | 104 | 0.298 |
| 147457 | 2 | 70504464 | 70504464 | C | T | SNP | <i>PCYOX1</i> | ENST00000433351 | 1 | silent | c.1458 | p.N486 | tier1 | 35 | 1 | 0.028 | 95 | 45 | 0.321 |
| 147457 | 2 | 73829499 | 73829499 | C | T | SNP | <i>ALMS1</i> | ENST00000264448 | 1 | splice_region | c.12295+4 | e20+4 | tier2 | 26 | 0 | 0.000 | 164 | 65 | 0.283 |
| 147457 | 2 | 74173887 | 74173887 | G | A | SNP | <i>DGUOK</i> | ENST00000264093 | 1 | missense | c.297 | p.M99I | tier1 | 62 | 2 | 0.031 | 219 | 83 | 0.275 |
| 147457 | 2 | 74867224 | 74867224 | C | T | SNP | <i>C2orf65</i> | ENST00000290536 | -1 | missense | c.179 | p.R60H | tier1 | 58 | 3 | 0.049 | 178 | 62 | 0.257 |
| 147457 | 2 | 75158001 | 75158001 | C | T | SNP | <i>ENSG00000204792</i> | ENST00000377469 | 1 | rna | NULL | NULL | tier1 | 38 | 0 | 0.000 | 83 | 44 | 0.347 |
| 147457 | 2 | 91800800 | 91800800 | G | A | SNP | <i>ENSG00000233991</i> | ENST00000443031 | 1 | 3_prime_untranslated_region | c.*91800800 | NULL | tier2 | 213 | 0 | 0.000 | 1207 | 71 | 0.056 |
| 147457 | 2 | 98732125 | 98732125 | G | A | SNP | <i>VWA3B</i> | ENST00000451075 | 1 | missense | c.8 | p.R3H | tier1 | 96 | 0 | 0.000 | 138 | 65 | 0.320 |
| 147457 | 2 | 99342686 | 99342686 | G | A | SNP | <i>MGAT4A</i> | ENST00000495056 | -1 | 5_prime_untranslated_region | c.-99342686 | NULL | tier2 | 121 | 1 | 0.008 | 232 | 101 | 0.303 |
| 147457 | 2 | 120397368 | 120397368 | C | T | SNP | <i>PCDP1</i> | ENST00000443972 | 1 | missense | c.821 | p.P274L | tier1 | 106 | 2 | 0.018 | 203 | 92 | 0.311 |
| 147457 | 2 | 131704217 | 131704217 | C | T | SNP | <i>ARHGEF4</i> | ENST00000409359 | 1 | missense | c.3004 | p.R1002C | tier1 | 32 | 0 | 0.000 | 168 | 60 | 0.263 |
| 147457 | 2 | 132384417 | 132384417 | G | A | SNP | <i>POTEKP</i> | ENST00000397487 | 1 | 3_prime_untranslated_region | c.*132384417 | NULL | tier3 | 49 | 1 | 0.020 | 374 | 145 | 0.279 |
| 147457 | 2 | 141459402 | 141459402 | G | A | SNP | <i>LRP1B</i> | ENST00000389484 | -1 | silent | c.6315 | p.N2105 | tier1 | 142 | 0 | 0.000 | 385 | 144 | 0.272 |
| 147457 | 2 | 160983105 | 160983105 | G | A | SNP | <i>ITGB6</i> | ENST00000283249 | -1 | silent | c.1668 | p.G556 | tier1 | 16 | 0 | 0.000 | 106 | 46 | 0.303 |
| 147457 | 2 | 170134318 | 170134318 | C | T | SNP | <i>LRP2</i> | ENST00000263816 | -1 | missense | c.1709 | p.R570H | tier1 | 94 | 0 | 0.000 | 185 | 76 | 0.290 |
| 147457 | 2 | 179613743 | 179613743 | C | T | SNP | <i>TTN</i> | ENST00000360870 | -1 | missense | c.13384 | p.E4462K | tier1 | 115 | 1 | 0.009 | 171 | 79 | 0.316 |
| 147457 | 2 | 198274685 | 198274685 | C | T | SNP | <i>SF3B1</i> | ENST00000335508 | -1 | missense | c.713 | p.R238H | tier1 | 88 | 3 | 0.033 | 603 | 239 | 0.284 |
| 147457 | 2 | 201619547 | 201619547 | C | T | SNP | <i>AOX2P</i> | ENST00000470911 | 1 | 3_prime_untranslated_region | c.*201619547 | NULL | tier3 | 128 | 1 | 0.008 | 200 | 84 | 0.296 |
| 147457 | 2 | 203948117 | 203948117 | G | A | SNP | <i>NBEAL1</i> | ENST00000449802 | 1 | missense | c.860 | p.R287H | tier1 | 83 | 0 | 0.000 | 199 | 89 | 0.309 |
| 147457 | 2 | 213886807 | 213886807 | G | A | SNP | <i>IKZF2</i> | ENST00000434687 | -1 | missense | c.622 | p.R208C | tier1 | 89 | 2 | 0.022 | 113 | 53 | 0.317 |
| 147457 | 2 | 215798769 | 215798769 | C | T | SNP | <i>ABCA12</i> | ENST00000272895 | -1 | intronic | c.7680+33 | e52+33 | tier3 | 52 | 0 | 0.000 | 148 | 71 | 0.324 |
| 147457 | 2 | 216807787 | 216807787 | C | T | SNP | <i>MREG</i> | ENST00000423087 | -1 | 3_prime_untranslated_region | c.*1799 | NULL | tier3 | 167 | 3 | 0.018 | 255 | 105 | 0.291 |
| 147457 | 2 | 218938006 | 218938006 | G | T | SNP | <i>RUFY4</i> | ENST00000374155 | 1 | missense | c.427 | p.R143C | tier1 | 99 | 1 | 0.010 | 355 | 138 | 0.279 |
| 147457 | 2 | 219538351 | 219538351 | C | A | SNP | <i>STK36</i> | ENST00000295709 | 1 | missense | c.88 | p.V30M | tier1 | 62 | 0 | 0.000 | 138 | 53 | 0.275 |
| 147457 | 2 | 219544699 | 219544699 | C | T | SNP | <i>STK36</i> | ENST00000295709 | 1 | silent | c.1032 | p.L344 | tier1 | 29 | 0 | 0.000 | 119 | 68 | 0.364 |
| 147457 | 2 | 220312930 | 220312930 | G | T | SNP | <i>SPEG</i> | ENST00000312358 | 1 | silent | c.1050 | p.T350 | tier1 | 179 | 1 | 0.006 | 726 | 274 | 0.274 |
| 147457 | 2 | 226447163 | 226447163 | C | A | SNP | <i>NYAP2</i> | ENST00000272907 | 1 | missense | c.1030 | p.V344M | tier1 | 25 | 0 | 0.000 | 98 | 31 | 0.223 |
| 147457 | 2 | 226447731 | 226447731 | G | A | SNP | <i>NYAP2</i> | ENST00000272907 | 1 | missense | c.1598 | p.R533H | tier1 | 16 | 0 | 0.000 | 174 | 96 | 0.356 |
| 147457 | 2 | 227872842 | 227872842 | C | T | SNP | <i>COL4A4</i> | ENST00000396625 | -1 | silent | c.4701 | p.A1567 | tier1 | 17 | 0 | 0.000 | 219 | 81 | 0.270 |
| 147457 | 2 | 227915822 | 227915822 | G | A | SNP | <i>COL4A4</i> | ENST00000396625 | -1 | silent | c.3021 | p.Y1007 | tier1 | 59 | 1 | 0.017 | 169 | 100 | 0.372 |
| 147457 | 2 | 231307770 | 231307770 | G | A | SNP | <i>SP100</i> | ENST00000340126 | 1 | missense | c.226 | p.E76K | tier1 | 132 | 1 | 0.008 | 204 | 92 | 0.311 |
| 147457 | 2 | 238245159 | 238245159 | C | T | SNP | <i>COL6A3</i> | ENST00000295550 | -1 | missense | c.8584 | p.V2862I | tier1 | 218 | 4 | 0.018 | 291 | 115 | 0.283 |
| 147457 | 2 | 238461018 | 238461018 | C | T | SNP | <i>MLPH</i> | ENST00000264605 | 1 | nonsense | c.1714 | p.R572* | tier1 | 89 | 0 | 0.000 | 194 | 81 | 0.295 |
| 147457 | 2 | 241517127 | 241517127 | C | T | SNP | <i>RNPEPL1</i> | ENST00000270357 | 1 | missense | c.1303 | p.R435C | tier1 | 25 | 0 | 0.000 | 297 | 125 | 0.296 |
| 147457 | 3 | 3137142 | 3137142 | C | T | SNP | <i>IL5RA</i> | ENST00000256452 | -1 | intronic | c.710-14 | e6-14 | tier3 | 55 | 0 | 0.000 | 105 | 34 | 0.245 |
| 147457 | 3 | 9768381 | 9768381 | C | T | SNP | <i>CPNE9</i> | ENST00000273027 | 1 | missense | c.572 | p.S191L | tier1 | 59 | 0 | 0.000 | 144 | 70 | 0.326 |
| 147457 | 3 | 11468460 | 11468460 | C | T | SNP | <i>ATG7</i> | ENST00000354449 | 1 | intronic | c.2079+60 | e17+60 | tier3 | 128 | 0 | 0.000 | 191 | 91 | 0.323 |
| 147457 | 3 | 11487941 | 11487941 | G | A | SNP | <i>ATG7</i> | ENST00000354449 | 1 | intronic | c.2079+19541 | e17+19541 | tier3 | 127 | 0 | 0.000 | 202 | 72 | 0.263 |
| 147457 | 3 | 13916529 | 13916529 | G | A | SNP | <i>WNT7A</i> | ENST00000285018 | -1 | silent | c.213 | p.D71 | tier1 | 35 | 0 | 0.000 | 94 | 48 | 0.338 |
| 147457 | 3 | 14552987 | 14552987 | G | A | SNP | <i>GRIP2</i> | ENST00000273083 | -1 | 5_prime_untranslated_region | c.-14552987 | NULL | tier3 | 28 | 0 | 0.000 | 132 | 67 | 0.335 |
| 147457 | 3 | 14862667 | 14862667 | C | T | SNP | <i>FGD5</i> | ENST00000285046 | 1 | missense | c.2089 | p.R697W | tier1 | 81 | 1 | 0.012 | 245 | 127 | 0.341 |
| 147457 | 3 | 16269593 | 16269593 | C | T | SNP | <i>GALNTL2</i> | ENST00000339732 | 1 | 3_prime_untranslated_region | c.*586 | NULL | tier3 | 60 | 0 | 0.000 | 123 | 45 | 0.268 |
| 147457 | 3 | 24231711 | 24231711 | G | T | SNP | <i>THRB</i> | ENST00000356447 | -1 | missense | c.137 | p.R46H | tier1 | 70 | 2 | 0.028 | 100 | 44 | 0.306 |
| 147457 | 3 | 32811381 | 32811381 | C | A | SNP | <i>CNOT10</i> | ENST00000454516 | 1 | silent | c.2187 | p.A729 | tier1 | 140 | 4 | 0.028 | 223 | 101 | 0.310 |
| 147457 | 3 | 46450139 | 46450139 | C | T | SNP | <i>CCRL2</i> | ENST00000357392 | 1 | missense | c.605 | p.P202L | tier1 | 65 | 1 | 0.015 | 176 | 61 | 0.256 |
| 147457 | 3 | 48266702 | 48266702 | G | T | SNP | <i>CAMP</i> | ENST00000296435 | 1 | intronic | c.391-81 | e4-81 | tier2 | 70 | 0 | 0.000 | 266 | 111 | 0.291 |
| 147457 | 3 | 48694445 | 48694445 | C | A | SNP | <i>CELSR3</i> | ENST00000544264 | -1 | missense | c.4085 | p.A1362V | tier1 | 136 | 1 | 0.007 | 553 | 227 | 0.290 |
| 147457 | 3 | 49455325 | 49455325 | C | T | SNP | <i>AMT</i> | ENST00000273588 | -1 | missense | c.959 | p.R320H | tier1 | 20 | 0 | 0.000 | 149 | 60 | 0.287 |
| 147457 | 3 | 52559275 | 52559275 | G | A | SNP | <i>NT5DC2</i> | ENST00000422318 | -1 | missense | c.1156 | p.R386C | tier1 | 27 | 0 | 0.000 | 126 | 40 | 0.241 |
| 147457 | 3 | 68781240 | 68781240 | C | T | SNP | <i>FAM19A4</i> | ENST00000295569 | -1 | 3_prime_untranslated_region | c.*1053 | NULL | tier3 | 47 | 0 | 0.000 | 88 | 38 | 0.302 |
| 147457 | 3 | 69987598 | 69987598 | C | T | SNP | <i>MITF</i> | ENST00000394348 | 1 | 3_prime_untranslated_region | c.*69987598 | NULL | tier3 | 133 | 1 | 0.008 | 290 | 82 | 0.220 |
| 147457 | 3 | 70014355 | 70014355 | C | T | SNP | <i>MITF</i> | ENST00000448226 | 1 | missense | c.1537 | p.R513W | tier1 | 26 | 0 | 0.000 | 71 | 25 | 0.260 |
| 147457 | 3 | 101050880 | 101050880 | C | T | SNP | <i>SENP7</i> | ENST00000394095 | -1 | missense | c.2647 | p.V883I | tier1 | 163 | 0 | 0.000 | 270 | 106 | 0.282 |
| 147457 | 3 | 101308992 | 101308992 | G | A | SNP | <i>PCNP</i> | ENST00000465366 | 1 | 3_prime_untranslated_region | c.*101308992 | NULL | tier3 | 123 | 2 | 0.016 | 147 | 53 | 0.265 |
| 147457 | 3 | 112991437 | 112991437 | A | G | SNP | <i>BOC</i> | ENST00000273395 | 1 | missense | c.848 | p.N283S | tier1 | 9 | 0 | 0.000 | 80 | 88 | 0.524 |
| 147457 | 3 | 122329604 | 122329604 | G | A | SNP | <i>PARP15</i> | ENST00000464300 | 1 | intronic | c.543+27 | e3+27 | tier3 | 51 | 0 | 0.000 | 156 | | |

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|--------|---|-----------|-----------|---|---|-----|------------------------|-----------------|----|-----------------------------|--------------|-----------|-------|-----|---|-------|------|-----|-------|
| 147457 | 3 | 194081495 | 194081495 | G | A | SNP | <i>LRRC15</i> | ENST00000439944 | -1 | missense | c.296 | p.T99M | tier1 | 22 | 0 | 0.000 | 99 | 39 | 0.281 |
| 147457 | 3 | 195509143 | 195509143 | G | A | SNP | <i>MUC4</i> | ENST00000463781 | -1 | missense | c.9308 | p.T3103M | tier1 | 226 | 2 | 0.009 | 1333 | 87 | 0.061 |
| 147457 | 3 | 196387275 | 196387275 | C | T | SNP | <i>LRRC33</i> | ENST00000328557 | 1 | missense | c.761 | p.T254M | tier1 | 17 | 0 | 0.000 | 147 | 74 | 0.335 |
| 147457 | 3 | 196751232 | 196751232 | G | A | SNP | <i>MF12</i> | ENST00000296350 | -1 | silent | c.429 | p.P143 | tier1 | 24 | 0 | 0.000 | 175 | 81 | 0.316 |
| 147457 | 4 | 1332229 | 1332229 | G | A | SNP | <i>MAEA</i> | ENST00000303400 | 1 | missense | c.919 | p.G307S | tier1 | 33 | 0 | 0.000 | 274 | 120 | 0.305 |
| 147457 | 4 | 2941321 | 2941321 | G | A | SNP | <i>NOP14</i> | ENST00000416614 | -1 | missense | c.2251 | p.R751W | tier1 | 18 | 0 | 0.000 | 116 | 57 | 0.330 |
| 147457 | 4 | 4317442 | 4317442 | C | T | SNP | <i>ZBTB49</i> | ENST00000337872 | 1 | nonsense | c.1456 | p.R486* | tier1 | 73 | 0 | 0.000 | 124 | 67 | 0.349 |
| 147457 | 4 | 8304175 | 8304175 | G | A | SNP | <i>HTRA3</i> | ENST00000307358 | 1 | intronic | c.1052-15 | e7-15 | tier3 | 30 | 0 | 0.000 | 93 | 44 | 0.319 |
| 147457 | 4 | 9828063 | 9828063 | G | A | SNP | <i>SLC2A9</i> | ENST00000264784 | -1 | silent | c.1581 | p.L527 | tier1 | 91 | 1 | 0.011 | 131 | 54 | 0.292 |
| 147457 | 4 | 15691400 | 15691400 | G | A | SNP | <i>FAM200B</i> | ENST00000504137 | 1 | 3_prime_untranslated_region | c.*15691400 | NULL | tier4 | 99 | 2 | 0.020 | 172 | 73 | 0.298 |
| 147457 | 4 | 17868743 | 17868743 | C | T | SNP | <i>LCORL</i> | ENST00000326877 | -1 | intronic | c.776+18948 | e6+18948 | tier3 | 44 | 0 | 0.000 | 103 | 44 | 0.299 |
| 147457 | 4 | 42509034 | 42509034 | G | A | SNP | <i>ATP8A1</i> | ENST00000381668 | -1 | silent | c.2085 | p.I695 | tier1 | 135 | 0 | 0.000 | 193 | 66 | 0.255 |
| 147457 | 4 | 44177057 | 44177057 | C | T | SNP | <i>KCTD8</i> | ENST00000360029 | -1 | missense | c.1172 | p.R391H | tier1 | 197 | 3 | 0.015 | 247 | 91 | 0.268 |
| 147457 | 4 | 48165752 | 48165752 | G | A | SNP | <i>TEC</i> | ENST00000381501 | -1 | missense | c.704 | p.T235M | tier1 | 113 | 3 | 0.026 | 238 | 85 | 0.262 |
| 147457 | 4 | 57384937 | 57384937 | G | A | SNP | <i>ARL9</i> | ENST00000360096 | 1 | missense | c.302 | p.R101Q | tier1 | 146 | 0 | 0.000 | 233 | 87 | 0.271 |
| 147457 | 4 | 69048233 | 69048233 | C | T | SNP | <i>FTLP10</i> | ENST00000503647 | 1 | 3_prime_untranslated_region | c.*69048233 | NULL | tier3 | 48 | 0 | 0.000 | 281 | 115 | 0.290 |
| 147457 | 4 | 71099798 | 71099798 | G | A | SNP | <i>FDCSP</i> | ENST00000317987 | 1 | missense | c.152 | p.R51H | tier1 | 190 | 1 | 0.005 | 161 | 55 | 0.255 |
| 147457 | 4 | 85758110 | 85758110 | C | T | SNP | <i>WDFY3</i> | ENST00000295888 | -1 | missense | c.548 | p.R183Q | tier1 | 136 | 3 | 0.022 | 255 | 124 | 0.326 |
| 147457 | 4 | 88293939 | 88293939 | G | A | SNP | <i>HSD17B11</i> | ENST00000358290 | -1 | missense | c.479 | p.T160M | tier1 | 195 | 2 | 0.010 | 302 | 110 | 0.267 |
| 147457 | 4 | 119461591 | 119461591 | G | A | SNP | <i>CEP170P1</i> | ENST00000412784 | 1 | intronic | c.NULL | NULL | tier3 | 52 | 1 | 0.019 | 88 | 48 | 0.353 |
| 147457 | 4 | 122846298 | 122846298 | G | A | SNP | <i>TRPC3</i> | ENST00000379645 | -1 | nonsense | c.1051 | p.R351* | tier1 | 74 | 0 | 0.000 | 103 | 64 | 0.383 |
| 147457 | 4 | 162421108 | 162421108 | C | T | SNP | <i>FSTL5</i> | ENST00000306100 | -1 | intronic | c.1458+60 | e11+60 | tier3 | 161 | 0 | 0.000 | 184 | 84 | 0.313 |
| 147457 | 4 | 166009582 | 166009582 | C | T | SNP | <i>TMEM192</i> | ENST00000306480 | -1 | intronic | c.574+38 | e4+38 | tier3 | 111 | 2 | 0.018 | 184 | 85 | 0.316 |
| 147457 | 4 | 187630302 | 187630302 | C | T | SNP | <i>FAT1</i> | ENST00000260147 | -1 | missense | c.680 | p.R227H | tier1 | 34 | 0 | 0.000 | 104 | 59 | 0.362 |
| 147457 | 4 | 189026057 | 189026057 | C | T | SNP | <i>TRIML2</i> | ENST00000512729 | -1 | silent | c.69 | p.S23 | tier1 | 112 | 0 | 0.000 | 127 | 61 | 0.323 |
| 147457 | 5 | 770160 | 770160 | C | T | SNP | <i>ZDHHC11</i> | ENST00000424784 | -1 | intronic | c.1239+25854 | e12+25854 | tier3 | 87 | 0 | 0.000 | 705 | 123 | 0.148 |
| 147457 | 5 | 1293693 | 1293693 | C | T | SNP | <i>TERT</i> | ENST00000310581 | -1 | silent | c.1308 | p.A436 | tier1 | 126 | 0 | 0.000 | 750 | 334 | 0.307 |
| 147457 | 5 | 1338034 | 1338034 | G | A | SNP | <i>CLPTM1L</i> | ENST00000320895 | -1 | silent | c.663 | p.R221 | tier1 | 31 | 0 | 0.000 | 118 | 45 | 0.276 |
| 147457 | 5 | 1483662 | 1483662 | G | A | SNP | <i>LPCAT1</i> | ENST00000283415 | -1 | intronic | c.668-61 | e6-61 | tier3 | 48 | 0 | 0.000 | 233 | 74 | 0.240 |
| 147457 | 5 | 5463371 | 5463371 | G | A | SNP | <i>KIAA0947</i> | ENST00000296564 | 1 | silent | c.3924 | p.T1308 | tier1 | 31 | 0 | 0.000 | 80 | 31 | 0.279 |
| 147457 | 5 | 6602599 | 6602599 | C | T | SNP | <i>NSUN2</i> | ENST00000264670 | -1 | missense | c.1972 | p.V658M | tier1 | 84 | 3 | 0.035 | 162 | 86 | 0.347 |
| 147457 | 5 | 13762777 | 13762777 | C | T | SNP | <i>DNAH5</i> | ENST00000265104 | -1 | intronic | c.10281+54 | e60+54 | tier3 | 76 | 1 | 0.013 | 294 | 135 | 0.315 |
| 147457 | 5 | 13931209 | 13931209 | C | T | SNP | <i>DNAH5</i> | ENST00000265104 | -1 | splice_region | c.192+10 | e2+10 | tier3 | 223 | 0 | 0.000 | 660 | 28 | 0.041 |
| 147457 | 5 | 31995780 | 31995780 | C | T | SNP | <i>PDZD2</i> | ENST00000282493 | 1 | silent | c.1077 | p.H359 | tier1 | 77 | 3 | 0.038 | 236 | 104 | 0.304 |
| 147457 | 5 | 34190535 | 34190535 | C | T | SNP | <i>ENSG00000215156</i> | ENST00000332686 | 1 | 3_prime_untranslated_region | c.*34190535 | NULL | tier3 | 636 | 6 | 0.009 | 4301 | 196 | 0.043 |
| 147457 | 5 | 35910093 | 35910093 | C | T | SNP | <i>CAPSL</i> | ENST00000397366 | -1 | missense | c.400 | p.E134K | tier1 | 155 | 4 | 0.025 | 236 | 74 | 0.239 |
| 147457 | 5 | 41149449 | 41149449 | G | A | SNP | <i>C6</i> | ENST00000263413 | -1 | silent | c.2517 | p.D839 | tier1 | 106 | 2 | 0.019 | 133 | 46 | 0.257 |
| 147457 | 5 | 79646877 | 79646877 | C | T | SNP | <i>CRSP8P</i> | ENST00000514280 | -1 | 5_prime_untranslated_region | c.-79646877 | NULL | tier3 | 107 | 1 | 0.009 | 498 | 173 | 0.257 |
| 147457 | 5 | 129521471 | 129521471 | G | A | SNP | <i>CHSY3</i> | ENST00000305031 | 1 | missense | c.2636 | p.R879Q | tier1 | 28 | 0 | 0.000 | 92 | 47 | 0.338 |
| 147457 | 5 | 140221632 | 140221632 | C | T | SNP | <i>PCDHAC1</i> | ENST00000531613 | 1 | silent | c.726 | p.F242 | tier1 | 316 | 1 | 0.003 | 877 | 53 | 0.057 |
| 147457 | 5 | 140308059 | 140308059 | C | T | SNP | <i>PCDHAC1</i> | ENST00000253807 | 1 | missense | c.1582 | p.R528W | tier1 | 57 | 0 | 0.000 | 180 | 69 | 0.277 |
| 147457 | 5 | 140720521 | 140720521 | C | T | SNP | <i>PCDHGA2</i> | ENST00000394576 | 1 | silent | c.1983 | p.T661 | tier1 | 42 | 0 | 0.000 | 432 | 199 | 0.315 |
| 147457 | 5 | 140751057 | 140751057 | G | A | SNP | <i>PCDHGB3</i> | ENST00000576222 | 1 | missense | c.1096 | p.V366I | tier1 | 78 | 2 | 0.025 | 189 | 70 | 0.270 |
| 147457 | 5 | 145610359 | 145610359 | C | T | SNP | <i>RBM27</i> | ENST00000265271 | 1 | silent | c.729 | p.I243 | tier1 | 93 | 0 | 0.000 | 159 | 66 | 0.293 |
| 147457 | 5 | 146236055 | 146236055 | C | T | SNP | <i>PPP2R2B</i> | ENST00000394410 | -1 | splice_region | c.37+7 | e1+7 | tier3 | 110 | 1 | 0.009 | 147 | 69 | 0.319 |
| 147457 | 5 | 147929769 | 147929769 | G | A | SNP | <i>HTR4</i> | ENST00000360693 | -1 | missense | c.83 | p.T28M | tier1 | 69 | 0 | 0.000 | 217 | 79 | 0.267 |
| 147457 | 5 | 149584221 | 149584221 | C | T | SNP | <i>SLC6A7</i> | ENST00000230671 | 1 | intronic | c.1432+27 | e11+27 | tier3 | 17 | 0 | 0.000 | 142 | 62 | 0.304 |
| 147457 | 5 | 153434852 | 153434852 | C | T | SNP | <i>MFAP3</i> | ENST00000436816 | 1 | 3_prime_untranslated_region | c.*1579 | NULL | tier3 | 89 | 0 | 0.000 | 119 | 44 | 0.270 |
| 147457 | 5 | 159665575 | 159665575 | G | A | SNP | <i>FABP6</i> | ENST00000393980 | 1 | intronic | c.481-38 | e6-38 | tier3 | 45 | 0 | 0.000 | 143 | 49 | 0.253 |
| 147457 | 5 | 173534383 | 173534383 | C | T | SNP | <i>HMP19</i> | ENST00000303177 | 1 | missense | c.391 | p.R131C | tier1 | 75 | 2 | 0.026 | 253 | 117 | 0.316 |
| 147457 | 5 | 175915965 | 175915965 | G | A | SNP | <i>FAF2</i> | ENST00000261942 | 1 | nonsense | c.281 | p.W94* | tier1 | 100 | 4 | 0.039 | 155 | 52 | 0.251 |
| 147457 | 5 | 176308809 | 176308809 | G | A | SNP | <i>HK3</i> | ENST00000292432 | -1 | silent | c.2277 | p.I759 | tier1 | 38 | 0 | 0.000 | 172 | 91 | 0.346 |
| 147457 | 5 | 178359183 | 178359183 | G | A | SNP | <i>ZFP2</i> | ENST00000361362 | 1 | missense | c.869 | p.R290Q | tier1 | 73 | 0 | 0.000 | 184 | 71 | 0.278 |
| 147457 | 6 | 3127540 | 3127540 | C | T | SNP | <i>BPHL</i> | ENST00000380379 | 1 | silent | c.276 | p.V92 | tier1 | 78 | 2 | 0.025 | 138 | 80 | 0.365 |
| 147457 | 6 | 25727357 | 25727357 | G | A | SNP | <i>HIST1H2BA</i> | ENST00000274764 | 1 | missense | c.221 | p.R74H | tier1 | 39 | 2 | 0.049 | 98 | 41 | 0.295 |
| 147457 | 6 | 27217008 | 27217008 | C | T | SNP | <i>PRSS16</i> | ENST00000230582 | 1 | missense | c.467 | p.A156V | tier1 | 54 | 1 | 0.018 | 115 | 46 | 0.286 |
| 147457 | 6 | 31555086 | 31555086 | C | T | SNP | <i>LST1</i> | ENST00000396117 | 1 | missense | c.10 | p.R4W | tier1 | 59 | 0 | 0.000 | 173 | 80 | 0.314 |
| 147457 | 6 | 31913016 | 31913016 | G | A | SNP | <i>C2</i> | ENST00000299367 | 1 | missense | c.2141 | p.R714H | tier1 | 94 | 1 | 0.011 | 272 | 126 | 0.314 |
| 147457 | 6 | 33554497 | 33554497 | C | T | SNP | <i>GGNBP1</i> | ENST00000374458 | 1 | missense | c.148 | p.R50C | tier1 | 28 | 0 | 0.000 | 175 | 73 | 0.294 |
| 147457 | 6 | 33990389 | 33990389 | C | T | SNP | <i>GRM4</i> | ENST00000374181 | -1 | 3_prime_untranslated_region | c.*209 | NULL | tier3 | 14 | 0 | 0.000 | 79 | 44 | 0.358 |
| 147457 | 6 | 42985429 | 42985429 | C | T | SNP | <i>KLHDC3</i> | ENST00000244670 | 1 | missense | c.41 | p.T14M | tier1 | 58 | 1 | 0.017 | 162 | 94 | 0.367 |
| 147457 | 6 | 52522264 | 52522264 | C | T | SNP | <i>TMEM14A</i> | ENST00000211314 | 1 | 5_prime_flanking_region | c.-19637 | NULL | tier3 | 35 | 0 | 0.000 | 147 | 70 | 0.320 |
| 147457 | 6 | 55270421 | 55270421 | G | A | SNP | <i>GFRAL</i> | ENST00000340465 | 1 | 3_prime_flanking_region | c.*3770 | NULL | tier3 | 104 | 2 | 0.019 | 164 | 102 | 0.382 |
| 147457 | 6 | 56919602 | 56919602 | C | T | SNP | <i>KIAA1586</i> | ENST00000370733 | 1 | missense | c.2305 | p.R769W | tier1 | 195 | 6 | 0.030 | 301 | 129 | 0.300 |
| 147457 | 6 | 75831004 | 75831004 | G | A | SNP | <i>COL12A1</i> | ENST00000322507 | -1 | intronic | c.7086+14 | e43+14 | tier3 | 201 | 0 | 0.000 | 682 | 28 | 0.039 |
| 147457 | 6 | 105726111 | 105726111 | C | T | SNP | <i>PREP</i> | ENST00000369110 | -1 | missense | c.2041 | p.G681R | tier1 | 18 | 0 | 0.000 | 105 | 41 | 0.281 |
| 147457 | 6 | 108985689 | 108985689 | G | A | SNP | <i>FOXO3</i> | ENST00000343882 | 1 | silent | c.1653 | p.S551 | tier1 | 19 | 0 | 0.000 | 131 | 70 | 0.348 |
| 147457 | 6 | 1367 | | | | | | | | | | | | | | | | | |

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|--------|---|-----------|-----------|---|---|-----|-----------------|-----------------|----|-----------------------------|--------------|----------|-------|-----|---|-------|------|-----|-------|
| 147457 | 7 | 2474107 | 2474107 | C | T | SNP | CHST12 | ENST00000258711 | 1 | 3_prime_untranslated_region | c.*588 | NULL | tier4 | 56 | 1 | 0.018 | 90 | 34 | 0.270 |
| 147457 | 7 | 6005211 | 6005211 | C | T | SNP | RSPH10B | ENST00000337579 | -1 | intronic | c.330+57 | e2+57 | tier3 | 15 | 0 | 0.000 | 59 | 32 | 0.352 |
| 147457 | 7 | 20762660 | 20762660 | G | A | SNP | ABCB5 | ENST00000258738 | 1 | missense | c.1108 | p.V370I | tier1 | 103 | 0 | 0.000 | 110 | 51 | 0.317 |
| 147457 | 7 | 21906048 | 21906048 | C | T | SNP | DNAH11 | ENST00000328843 | 1 | intronic | c.11518-40 | e72-40 | tier3 | 47 | 0 | 0.000 | 124 | 39 | 0.239 |
| 147457 | 7 | 42005169 | 42005169 | C | T | SNP | GLI3 | ENST00000395925 | -1 | missense | c.3502 | p.G1168R | tier1 | 54 | 1 | 0.018 | 334 | 108 | 0.244 |
| 147457 | 7 | 45813153 | 45813153 | C | T | SNP | ENSG00000212450 | ENST00000391148 | 1 | 5_prime_flanking_region | c.-16031 | NULL | tier3 | 879 | 4 | 0.005 | 1789 | 86 | 0.046 |
| 147457 | 7 | 48139220 | 48139220 | C | T | SNP | UPP1 | ENST00000331803 | 1 | intronic | c.45-47 | e2-47 | tier3 | 101 | 0 | 0.000 | 156 | 49 | 0.238 |
| 147457 | 7 | 51454298 | 51454298 | G | A | SNP | ENSG00000228897 | ENST00000444843 | -1 | 5_prime_untranslated_region | c.-51454298 | NULL | tier3 | 67 | 1 | 0.015 | 812 | 282 | 0.257 |
| 147457 | 7 | 75609676 | 75609676 | C | T | SNP | POR | ENST00000461988 | 1 | missense | c.386 | p.P129L | tier1 | 20 | 0 | 0.000 | 110 | 40 | 0.265 |
| 147457 | 7 | 76610187 | 76610187 | G | A | SNP | ENSG00000186704 | ENST00000425797 | 1 | 3_prime_untranslated_region | c.*76610187 | NULL | tier3 | 58 | 0 | 0.000 | 346 | 137 | 0.283 |
| 147457 | 7 | 92855290 | 92855290 | C | T | SNP | HEPACAM2 | ENST00000394468 | -1 | intronic | c.79+391 | e1+391 | tier3 | 59 | 0 | 0.000 | 69 | 34 | 0.330 |
| 147457 | 7 | 94293941 | 94293941 | G | A | SNP | PEG10 | ENST00000482108 | 1 | 3_prime_untranslated_region | c.*95 | NULL | tier3 | 44 | 2 | 0.044 | 131 | 49 | 0.271 |
| 147457 | 7 | 97872988 | 97872988 | C | T | SNP | TECPR1 | ENST00000379795 | -1 | intronic | c.532-84 | e4-84 | tier3 | 20 | 0 | 0.000 | 172 | 60 | 0.259 |
| 147457 | 7 | 97920701 | 97920701 | G | A | SNP | BRI3 | ENST00000297290 | 1 | 3_prime_untranslated_region | c.*146 | NULL | tier3 | 39 | 0 | 0.000 | 99 | 38 | 0.277 |
| 147457 | 7 | 98258817 | 98258817 | G | A | SNP | NPTX2 | ENST00000265634 | 1 | 3_prime_untranslated_region | c.*876 | NULL | tier3 | 149 | 0 | 0.000 | 211 | 73 | 0.257 |
| 147457 | 7 | 100730606 | 100730606 | G | A | SNP | TRIM56 | ENST00000306085 | 1 | missense | c.13 | p.G5R | tier1 | 25 | 0 | 0.000 | 176 | 70 | 0.282 |
| 147457 | 7 | 105662648 | 105662648 | C | T | SNP | CDHR3 | ENST00000468477 | 1 | missense | c.236 | p.T79M | tier1 | 105 | 1 | 0.009 | 216 | 90 | 0.292 |
| 147457 | 7 | 123594538 | 123594538 | G | A | SNP | SPAM1 | ENST00000340011 | 1 | missense | c.914 | p.R305H | tier1 | 98 | 0 | 0.000 | 103 | 43 | 0.295 |
| 147457 | 7 | 133894966 | 133894966 | C | T | SNP | LRGUK | ENST00000285928 | 1 | intronic | c.1843+8638 | e15+8638 | tier3 | 12 | 0 | 0.000 | 126 | 77 | 0.379 |
| 147457 | 7 | 134632353 | 134632353 | C | T | SNP | CALD1 | ENST00000361388 | 1 | missense | c.940 | p.R314C | tier1 | 46 | 0 | 0.000 | 313 | 129 | 0.291 |
| 147457 | 7 | 138522637 | 138522637 | C | T | SNP | TMEM213 | ENST00000413208 | 1 | nonsense | c.178 | p.R60* | tier1 | 44 | 0 | 0.000 | 87 | 61 | 0.412 |
| 147457 | 7 | 139801866 | 139801866 | C | T | SNP | JHDM1D | ENST00000397560 | -1 | missense | c.1523 | p.R508Q | tier1 | 202 | 1 | 0.005 | 197 | 83 | 0.296 |
| 147457 | 7 | 140156546 | 140156546 | G | A | SNP | MKRN1 | ENST00000255977 | -1 | missense | c.892 | p.R298C | tier1 | 46 | 0 | 0.000 | 134 | 69 | 0.340 |
| 147457 | 7 | 143010124 | 143010124 | C | T | SNP | HINT1P1 | ENST00000464866 | -1 | 5_prime_untranslated_region | c.-143010124 | NULL | tier3 | 36 | 0 | 0.000 | 79 | 33 | 0.295 |
| 147457 | 7 | 150772740 | 150772740 | G | A | SNP | SLC4A2 | ENST00000413384 | 1 | missense | c.3349 | p.V1117M | tier1 | 18 | 0 | 0.000 | 125 | 66 | 0.346 |
| 147457 | 7 | 150939682 | 150939682 | C | T | SNP | SMARCD3 | ENST00000262188 | -1 | missense | c.464 | p.R155Q | tier1 | 32 | 0 | 0.000 | 150 | 65 | 0.302 |
| 147457 | 8 | 8185940 | 8185940 | G | A | SNP | SGK223 | ENST00000330777 | -1 | silent | c.2352 | p.S784 | tier1 | 36 | 0 | 0.000 | 154 | 78 | 0.336 |
| 147457 | 8 | 8235576 | 8235576 | G | A | SNP | SGK223 | ENST00000330777 | -1 | nonsense | c.343 | p.R115* | tier1 | 153 | 2 | 0.013 | 363 | 141 | 0.279 |
| 147457 | 8 | 10464473 | 10464473 | C | T | SNP | RP1L1 | ENST00000382483 | -1 | missense | c.7135 | p.G2379R | tier1 | 36 | 0 | 0.000 | 103 | 55 | 0.346 |
| 147457 | 8 | 11223131 | 11223131 | G | A | SNP | TDH | ENST00000525246 | 1 | 3_prime_untranslated_region | c.*11223131 | NULL | tier3 | 77 | 1 | 0.013 | 160 | 72 | 0.310 |
| 147457 | 8 | 12232826 | 12232826 | G | A | SNP | ENSG00000254423 | ENST00000532111 | -1 | 5_prime_untranslated_region | c.-12232826 | NULL | tier3 | 89 | 1 | 0.011 | 587 | 139 | 0.192 |
| 147457 | 8 | 21927047 | 21927047 | G | A | SNP | EPB49 | ENST00000381455 | 1 | intronic | c.211+18 | e3+18 | tier3 | 21 | 0 | 0.000 | 160 | 75 | 0.319 |
| 147457 | 8 | 27094400 | 27094400 | C | T | SNP | STMN4 | ENST00000350889 | -1 | missense | c.614 | p.R205Q | tier1 | 41 | 0 | 0.000 | 105 | 47 | 0.309 |
| 147457 | 8 | 32505766 | 32505766 | G | A | SNP | NRG1 | ENST00000520502 | 1 | missense | c.530 | p.R177H | tier1 | 91 | 0 | 0.000 | 197 | 92 | 0.317 |
| 147457 | 8 | 38109809 | 38109809 | A | C | SNP | DDHD2 | ENST00000397166 | 1 | splice_region | c.1617+4 | e12+4 | tier3 | 121 | 1 | 0.008 | 158 | 58 | 0.267 |
| 147457 | 8 | 41791093 | 41791093 | C | T | SNP | KAT6A | ENST00000265713 | -1 | missense | c.4645 | p.G1549S | tier1 | 33 | 1 | 0.029 | 139 | 58 | 0.294 |
| 147457 | 8 | 48955608 | 48955608 | G | A | SNP | UBE2V2 | ENST00000523111 | 1 | missense | c.32 | p.R11H | tier1 | 243 | 1 | 0.004 | 282 | 91 | 0.244 |
| 147457 | 8 | 61192308 | 61192308 | G | A | SNP | CA8 | ENST00000317995 | -1 | nonsense | c.232 | p.R78* | tier1 | 78 | 0 | 0.000 | 158 | 57 | 0.265 |
| 147457 | 8 | 88249205 | 88249205 | C | T | SNP | CNBD1 | ENST00000518476 | 1 | silent | c.636 | p.N212 | tier1 | 105 | 2 | 0.019 | 158 | 81 | 0.339 |
| 147457 | 8 | 92998437 | 92998437 | G | A | SNP | RUNX1T1 | ENST00000521751 | -1 | missense | c.293 | p.T98M | tier1 | 226 | 0 | 0.000 | 684 | 26 | 0.037 |
| 147457 | 8 | 125592809 | 125592809 | G | A | SNP | MTSS1 | ENST00000325064 | -1 | splice_region | c.461-6 | e7-6 | tier3 | 56 | 0 | 0.000 | 180 | 92 | 0.338 |
| 147457 | 8 | 128753030 | 128753030 | G | A | SNP | MYC | ENST00000377970 | 1 | silent | c.1191 | p.P397 | tier1 | 50 | 1 | 0.020 | 140 | 47 | 0.247 |
| 147457 | 8 | 130867827 | 130867827 | G | A | SNP | FAM49B | ENST00000401979 | -1 | intronic | c.438+30 | e4+30 | tier3 | 94 | 2 | 0.021 | 114 | 54 | 0.321 |
| 147457 | 8 | 143618381 | 143618381 | C | T | SNP | BAI1 | ENST00000323289 | 1 | missense | c.3604 | p.R1202W | tier1 | 62 | 0 | 0.000 | 548 | 236 | 0.300 |
| 147457 | 8 | 143783019 | 143783019 | C | T | SNP | LY6K | ENST00000561179 | 1 | silent | c.279 | p.D93 | tier1 | 135 | 1 | 0.007 | 203 | 76 | 0.271 |
| 147457 | 8 | 144620573 | 144620573 | G | A | SNP | ZC3H3 | ENST00000262577 | -1 | missense | c.964 | p.R322W | tier1 | 59 | 0 | 0.000 | 206 | 85 | 0.292 |
| 147457 | 8 | 144946163 | 144946163 | C | T | SNP | EPPK1 | ENST00000525985 | -1 | missense | c.1259 | p.R420H | tier1 | 75 | 0 | 0.000 | 324 | 155 | 0.323 |
| 147457 | 9 | 368135 | 368135 | G | A | SNP | DOCK8 | ENST00000453981 | 1 | silent | c.1797 | p.P599 | tier1 | 61 | 0 | 0.000 | 191 | 95 | 0.311 |
| 147457 | 9 | 34551979 | 34551979 | G | A | SNP | CNTFR | ENST00000351266 | -1 | 3_prime_untranslated_region | c.*90 | NULL | tier2 | 105 | 1 | 0.009 | 312 | 113 | 0.265 |
| 147457 | 9 | 35664015 | 35664015 | G | A | SNP | C9orf100 | ENST00000378387 | -1 | missense | c.463 | p.R155W | tier1 | 48 | 1 | 0.020 | 155 | 77 | 0.332 |
| 147457 | 9 | 38577921 | 38577921 | C | T | SNP | ANKRD18A | ENST00000399703 | -1 | silent | c.2472 | p.S824 | tier1 | 266 | 4 | 0.015 | 513 | 229 | 0.308 |
| 147457 | 9 | 39176062 | 39176062 | C | T | SNP | CNTNAP3 | ENST00000297668 | -1 | missense | c.955 | p.G319R | tier1 | 467 | 4 | 0.009 | 683 | 36 | 0.050 |
| 147457 | 9 | 43709787 | 43709787 | C | T | SNP | CNTNAP3B | ENST00000377564 | 1 | intronic | c.196+27 | e2+27 | tier3 | 255 | 5 | 0.019 | 628 | 289 | 0.315 |
| 147457 | 9 | 45120270 | 45120270 | G | A | SNP | ENSG00000231838 | ENST00000417850 | 1 | 5_prime_flanking_region | c.-44308 | NULL | tier3 | 30 | 0 | 0.000 | 177 | 69 | 0.279 |
| 147457 | 9 | 66500729 | 66500729 | G | A | SNP | CDK2AP2P2 | ENST00000478512 | 1 | 3_prime_untranslated_region | c.*66500729 | NULL | tier3 | 96 | 1 | 0.010 | 1004 | 101 | 0.091 |
| 147457 | 9 | 78973784 | 78973784 | C | T | SNP | PCSK5 | ENST00000545128 | 1 | silent | c.5529 | p.D1843 | tier1 | 67 | 1 | 0.015 | 185 | 107 | 0.365 |
| 147457 | 9 | 82333659 | 82333659 | G | A | SNP | TLE4 | ENST00000376520 | 1 | missense | c.1459 | p.A487T | tier1 | 73 | 2 | 0.027 | 169 | 72 | 0.299 |
| 147457 | 9 | 95872991 | 95872991 | C | T | SNP | C9orf89 | ENST00000466409 | 1 | missense | c.292 | p.R98C | tier1 | 19 | 0 | 0.000 | 224 | 106 | 0.320 |
| 147457 | 9 | 97847849 | 97847849 | G | A | SNP | C9orf3 | ENST00000375315 | 1 | intronic | c.2460-1115 | e16-1115 | tier3 | 43 | 2 | 0.044 | 89 | 42 | 0.321 |
| 147457 | 9 | 99711889 | 99711889 | C | T | SNP | HIATL2 | ENST00000375223 | -1 | missense | c.343 | p.G115S | tier1 | 83 | 3 | 0.035 | 229 | 105 | 0.313 |
| 147457 | 9 | 103339628 | 103339628 | G | A | SNP | TMEFF1 | ENST00000374879 | 1 | 3_prime_untranslated_region | c.*746 | NULL | tier2 | 154 | 1 | 0.007 | 237 | 85 | 0.263 |
| 147457 | 9 | 104124794 | 104124794 | G | A | SNP | BAAT | ENST00000259407 | -1 | silent | c.1173 | p.H391 | tier1 | 60 | 0 | 0.000 | 107 | 65 | 0.376 |
| 147457 | 9 | 112017768 | 112017768 | G | A | SNP | EPB41L4B | ENST00000374566 | -1 | intronic | c.1169+23 | e11+23 | tier3 | 54 | 0 | 0.000 | 154 | 58 | 0.271 |
| 147457 | 9 | 116050499 | 116050499 | C | T | SNP | PRPF4 | ENST00000374198 | 1 | missense | c.980 | p.A327V | tier1 | 78 | 0 | 0.000 | 160 | 59 | 0.269 |
| 147457 | 9 | 117087344 | 117087344 | C | T | SNP | ORM1 | ENST00000259396 | 1 | missense | c.452 | p.T151M | tier1 | 31 | 0 | 0.000 | 158 | 58 | 0.269 |
| 147457 | 9 | 117143546 | 117143546 | C | T | SNP | AKNA | ENST00000307564 | -1 | missense | c.68 | p.R23H | tier1 | 60 | 0 | 0.000 | 212 | 77 | 0.266 |
| 147457 | 9 | 123370046 | 123370046 | C | T | SNP | MEGF9 | ENST00000426959 | -1 | missense | c.1441 | p.D481N | tier1 | 85 | 1 | 0.012 | 224 | 79 | 0.259 |
| 147457 | 9 | 127572232 | 127572232 | G | A | SNP | OLFML2A | ENST00000373580 | 1 | silent | c.1500 | p.A500 | tier1 | 21 | 0 | 0.000 | 153 | 51 | 0.249 |
| 147457 | 9 | 131394806 | 131394806 | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|---|---|-----|------------------------|-----------------|----|-----------------------------|-------------|----------|-------|-----|---|-------|-----|-----|-------|
| 147457 | 10 | 11805425 | 11805425 | C | T | SNP | <i>ECHDC3</i> | ENST00000379215 | 1 | missense | c.794 | p.T265M | tier1 | 26 | 1 | 0.037 | 214 | 118 | 0.354 |
| 147457 | 10 | 21827795 | 21827795 | C | T | SNP | <i>MLLT10</i> | ENST00000307729 | 1 | missense | c.194 | p.P65L | tier1 | 109 | 1 | 0.009 | 172 | 70 | 0.289 |
| 147457 | 10 | 25890983 | 25890983 | G | A | SNP | <i>GPR158</i> | ENST00000376351 | 1 | 3_prime_untranslated_region | c.*2780 | NULL | tier2 | 80 | 0 | 0.000 | 123 | 52 | 0.294 |
| 147457 | 10 | 27048066 | 27048066 | G | A | SNP | <i>ABI1</i> | ENST00000355394 | -1 | nonsense | c.1006 | p.R336* | tier1 | 125 | 2 | 0.016 | 189 | 75 | 0.284 |
| 147457 | 10 | 28023604 | 28023604 | G | A | SNP | <i>MKX</i> | ENST00000375790 | -1 | missense | c.619 | p.R207W | tier1 | 104 | 1 | 0.010 | 225 | 78 | 0.257 |
| 147457 | 10 | 29773728 | 29773728 | G | A | SNP | <i>SVIL</i> | ENST00000355867 | -1 | silent | c.4812 | p.Y1604 | tier1 | 202 | 2 | 0.010 | 287 | 114 | 0.284 |
| 147457 | 10 | 29813480 | 29813480 | C | T | SNP | <i>SVIL</i> | ENST00000355867 | -1 | missense | c.2507 | p.R836Q | tier1 | 117 | 4 | 0.033 | 173 | 71 | 0.290 |
| 147457 | 10 | 30654823 | 30654823 | C | T | SNP | <i>ENSG00000241621</i> | ENST00000340929 | -1 | 5_prime_untranslated_region | c.-30654823 | NULL | tier3 | 96 | 0 | 0.000 | 546 | 236 | 0.302 |
| 147457 | 10 | 34671634 | 34671634 | C | T | SNP | <i>PARD3</i> | ENST00000374789 | -1 | silent | c.1233 | p.P411 | tier1 | 142 | 0 | 0.000 | 132 | 69 | 0.342 |
| 147457 | 10 | 46210567 | 46210567 | G | A | SNP | <i>ENSG00000237840</i> | ENST00000426241 | -1 | intronic | c.NULL | NULL | tier3 | 296 | 2 | 0.007 | 713 | 211 | 0.228 |
| 147457 | 10 | 47747160 | 47747160 | G | A | SNP | <i>ENSG00000244599</i> | ENST00000445847 | 1 | 3_prime_untranslated_region | c.*47747160 | NULL | tier3 | 68 | 1 | 0.015 | 210 | 90 | 0.300 |
| 147457 | 10 | 64967543 | 64967543 | C | T | SNP | <i>JMJD1C</i> | ENST00000399262 | -1 | missense | c.3886 | p.G1296R | tier1 | 79 | 0 | 0.000 | 127 | 52 | 0.291 |
| 147457 | 10 | 86004953 | 86004953 | C | T | SNP | <i>RGR</i> | ENST00000359452 | 1 | intronic | c.79+28 | e1+28 | tier3 | 11 | 0 | 0.000 | 84 | 56 | 0.397 |
| 147457 | 10 | 87379729 | 87379729 | G | A | SNP | <i>GRID1</i> | ENST00000327946 | -1 | missense | c.2255 | p.T752M | tier1 | 188 | 3 | 0.016 | 648 | 297 | 0.314 |
| 147457 | 10 | 87482902 | 87482902 | C | T | SNP | <i>GRID1</i> | ENST00000327946 | -1 | splice_region | c.1859-4 | e12-4 | tier2 | 44 | 0 | 0.000 | 203 | 98 | 0.325 |
| 147457 | 10 | 87966282 | 87966282 | G | A | SNP | <i>GRID1</i> | ENST00000327946 | -1 | missense | c.359 | p.P120L | tier1 | 134 | 1 | 0.007 | 510 | 257 | 0.335 |
| 147457 | 10 | 99019280 | 99019280 | C | T | SNP | <i>ARHGAP19</i> | ENST00000453547 | -1 | missense | c.719 | p.R240H | tier1 | 133 | 0 | 0.000 | 235 | 116 | 0.331 |
| 147457 | 10 | 100022513 | 100022513 | G | A | SNP | <i>LOXL4</i> | ENST00000260702 | -1 | silent | c.264 | p.Y88 | tier1 | 38 | 0 | 0.000 | 280 | 121 | 0.302 |
| 147457 | 10 | 103870964 | 103870964 | G | A | SNP | <i>LDB1</i> | ENST00000425280 | -1 | intronic | c.173+49 | e3+49 | tier3 | 81 | 3 | 0.036 | 144 | 62 | 0.300 |
| 147457 | 10 | 104245343 | 104245343 | C | T | SNP | <i>ACTR1A</i> | ENST00000369905 | -1 | intronic | c.440+23 | e5+23 | tier3 | 59 | 1 | 0.017 | 197 | 73 | 0.269 |
| 147457 | 10 | 104628872 | 104628872 | C | T | SNP | <i>C10orf32</i> | ENST00000299353 | 1 | intronic | c.321-690 | e6-690 | tier4 | 80 | 2 | 0.024 | 100 | 27 | 0.213 |
| 147457 | 10 | 106160477 | 106160477 | C | T | SNP | <i>CCDC147</i> | ENST00000369704 | 1 | missense | c.1855 | p.R619W | tier1 | 56 | 0 | 0.000 | 121 | 59 | 0.326 |
| 147457 | 10 | 116008560 | 116008560 | C | T | SNP | <i>VWA2</i> | ENST00000392982 | 1 | intronic | c.52+36 | e1+36 | tier3 | 140 | 1 | 0.007 | 167 | 82 | 0.329 |
| 147457 | 10 | 118969094 | 118969094 | G | A | SNP | <i>KCNK18</i> | ENST00000334549 | 1 | missense | c.439 | p.V147I | tier1 | 90 | 1 | 0.011 | 165 | 70 | 0.298 |
| 147457 | 10 | 123976222 | 123976222 | G | A | SNP | <i>TACC2</i> | ENST00000334433 | 1 | silent | c.7425 | p.A2475 | tier1 | 31 | 0 | 0.000 | 156 | 66 | 0.297 |
| 147457 | 10 | 124402685 | 124402685 | G | A | SNP | <i>DMBT1</i> | ENST00000368915 | 1 | missense | c.7400 | p.R2467H | tier1 | 64 | 0 | 0.000 | 245 | 86 | 0.259 |
| 147457 | 10 | 135011989 | 135011989 | C | T | SNP | <i>KNDC1</i> | ENST00000368572 | 1 | silent | c.2055 | p.N685 | tier1 | 20 | 0 | 0.000 | 367 | 142 | 0.278 |
| 147457 | 11 | 1215595 | 1215595 | G | A | SNP | <i>MUC5AC</i> | ENST00000358378 | 1 | splice_region | c.NULL | NULL | tier2 | 13 | 0 | 0.000 | 205 | 106 | 0.339 |
| 147457 | 11 | 1316941 | 1316941 | C | T | SNP | <i>TOLLIP</i> | ENST00000317204 | -1 | silent | c.117 | p.A39 | tier1 | 21 | 0 | 0.000 | 264 | 104 | 0.282 |
| 147457 | 11 | 1997630 | 1997630 | C | T | SNP | <i>MRPL23</i> | ENST00000397297 | 1 | intronic | c.497+3642 | e5+3642 | tier3 | 77 | 3 | 0.038 | 708 | 273 | 0.277 |
| 147457 | 11 | 6048700 | 6048700 | C | T | SNP | <i>OR56A1</i> | ENST00000316650 | -1 | missense | c.235 | p.V79M | tier1 | 49 | 0 | 0.000 | 301 | 134 | 0.307 |
| 147457 | 11 | 6232207 | 6232207 | G | A | SNP | <i>C11orf42</i> | ENST00000316375 | 1 | missense | c.937 | p.D313N | tier1 | 36 | 1 | 0.027 | 199 | 95 | 0.323 |
| 147457 | 11 | 10500213 | 10500213 | C | T | SNP | <i>AMPD3</i> | ENST00000396553 | 1 | missense | c.362 | p.T121M | tier1 | 34 | 0 | 0.000 | 131 | 87 | 0.399 |
| 147457 | 11 | 16036588 | 16036588 | C | T | SNP | <i>SOX6</i> | ENST00000352083 | -1 | silent | c.1632 | p.T544 | tier1 | 152 | 3 | 0.019 | 154 | 60 | 0.280 |
| 147457 | 11 | 16823374 | 16823374 | G | A | SNP | <i>PLEKHHA7</i> | ENST00000448080 | -1 | splice_region | c.2158-10 | e16-10 | tier3 | 29 | 0 | 0.000 | 156 | 50 | 0.243 |
| 147457 | 11 | 17659970 | 17659970 | C | T | SNP | <i>OTOG</i> | ENST00000399391 | 1 | intronic | c.7832-28 | e47-28 | tier3 | 25 | 0 | 0.000 | 90 | 41 | 0.313 |
| 147457 | 11 | 17801160 | 17801160 | C | T | SNP | <i>KCNC1</i> | ENST00000265969 | 1 | silent | c.1662 | p.Y554 | tier1 | 37 | 0 | 0.000 | 216 | 84 | 0.280 |
| 147457 | 11 | 26558879 | 26558879 | G | A | SNP | <i>ANO3</i> | ENST00000256737 | 1 | intronic | c.977-74 | e10-74 | tier3 | 103 | 3 | 0.028 | 130 | 62 | 0.323 |
| 147457 | 11 | 43515299 | 43515299 | C | T | SNP | <i>TTC17</i> | ENST0000039989 | 1 | intronic | c.3295-24 | e24-24 | tier3 | 87 | 3 | 0.033 | 185 | 71 | 0.277 |
| 147457 | 11 | 44253924 | 44253924 | C | T | SNP | <i>EXT2</i> | ENST00000395673 | 1 | missense | c.1783 | p.R595W | tier1 | 77 | 3 | 0.038 | 182 | 75 | 0.292 |
| 147457 | 11 | 45877647 | 45877647 | C | T | SNP | <i>CRY2</i> | ENST00000496571 | 1 | 3_prime_untranslated_region | c.*45877647 | NULL | tier3 | 42 | 1 | 0.023 | 139 | 85 | 0.380 |
| 147457 | 11 | 45950277 | 45950277 | C | T | SNP | <i>GYLTL1B</i> | ENST00000325468 | 1 | missense | c.2047 | p.R683C | tier1 | 45 | 2 | 0.043 | 301 | 103 | 0.255 |
| 147457 | 11 | 46741344 | 46741344 | C | T | SNP | <i>F2</i> | ENST00000311907 | 1 | nonsense | c.172 | p.R58* | tier1 | 47 | 2 | 0.041 | 243 | 145 | 0.374 |
| 147457 | 11 | 46819420 | 46819420 | G | A | SNP | <i>CKAP5</i> | ENST00000415402 | -1 | missense | c.1273 | p.R425C | tier1 | 174 | 1 | 0.006 | 218 | 77 | 0.260 |
| 147457 | 11 | 49897778 | 49897778 | C | A | SNP | <i>OR4R3P</i> | ENST00000525084 | 1 | 5_prime_flanking_region | c.-46823 | NULL | tier3 | 251 | 3 | 0.012 | 424 | 188 | 0.307 |
| 147457 | 11 | 55322719 | 55322719 | C | T | SNP | <i>OR4C15</i> | ENST00000314644 | 1 | nonsense | c.937 | p.R313* | tier1 | 100 | 2 | 0.020 | 139 | 69 | 0.330 |
| 147457 | 11 | 55761663 | 55761663 | G | T | SNP | <i>OR5F1</i> | ENST00000278409 | -1 | missense | c.439 | p.G147R | tier1 | 48 | 0 | 0.000 | 158 | 74 | 0.319 |
| 147457 | 11 | 55839272 | 55839272 | C | A | SNP | <i>OR5J1P</i> | ENST00000313544 | 1 | 3_prime_untranslated_region | c.*55839272 | NULL | tier3 | 23 | 0 | 0.000 | 56 | 23 | 0.291 |
| 147457 | 11 | 59623312 | 59623312 | G | A | SNP | <i>TCN1</i> | ENST00000257264 | -1 | intronic | c.937+30 | e6+30 | tier3 | 67 | 0 | 0.000 | 112 | 38 | 0.253 |
| 147457 | 11 | 61295431 | 61295431 | C | T | SNP | <i>SYT7</i> | ENST00000263846 | -1 | missense | c.578 | p.R193Q | tier1 | 60 | 0 | 0.000 | 151 | 66 | 0.301 |
| 147457 | 11 | 62571274 | 62571274 | G | A | SNP | <i>NXF1</i> | ENST00000294172 | -1 | nonsense | c.205 | p.R69* | tier1 | 37 | 0 | 0.000 | 97 | 41 | 0.297 |
| 147457 | 11 | 62751103 | 62751103 | G | A | SNP | <i>SLC22A6</i> | ENST00000540654 | -1 | missense | c.511 | p.R171C | tier1 | 19 | 0 | 0.000 | 199 | 102 | 0.338 |
| 147457 | 11 | 64882321 | 64882321 | C | T | SNP | <i>TM7SF2</i> | ENST00000528026 | 1 | 3_prime_untranslated_region | c.*64882321 | NULL | tier3 | 24 | 0 | 0.000 | 153 | 55 | 0.264 |
| 147457 | 11 | 64882420 | 64882420 | C | T | SNP | <i>TM7SF2</i> | ENST00000528802 | 1 | missense | c.242 | p.T81M | tier1 | 20 | 0 | 0.000 | 128 | 43 | 0.252 |
| 147457 | 11 | 65305479 | 65305479 | C | T | SNP | <i>SCYL1</i> | ENST00000533862 | 1 | missense | c.2036 | p.P679L | tier1 | 15 | 0 | 0.000 | 97 | 55 | 0.362 |
| 147457 | 11 | 65427692 | 65427692 | C | T | SNP | <i>RELA</i> | ENST00000526738 | -1 | 5_prime_untranslated_region | c.-65427692 | NULL | tier3 | 35 | 0 | 0.000 | 141 | 49 | 0.258 |
| 147457 | 11 | 66103134 | 66103134 | C | T | SNP | <i>RIN1</i> | ENST00000530745 | -1 | missense | c.221 | p.R74Q | tier1 | 31 | 0 | 0.000 | 208 | 77 | 0.269 |
| 147457 | 11 | 66633819 | 66633819 | C | T | SNP | <i>PC</i> | ENST00000393958 | -1 | missense | c.1024 | p.V342I | tier1 | 37 | 0 | 0.000 | 319 | 115 | 0.263 |
| 147457 | 11 | 67209188 | 67209188 | G | A | SNP | <i>CORO1B</i> | ENST00000545016 | -1 | missense | c.470 | p.P157L | tier1 | 22 | 0 | 0.000 | 142 | 53 | 0.270 |
| 147457 | 11 | 67795455 | 67795455 | C | T | SNP | <i>ALDH3B1</i> | ENST00000007633 | 1 | 3_prime_untranslated_region | c.*67795455 | NULL | tier3 | 36 | 2 | 0.053 | 356 | 166 | 0.317 |
| 147457 | 11 | 68701368 | 68701368 | G | A | SNP | <i>IGHMBP2</i> | ENST00000255078 | 1 | silent | c.1524 | p.S508 | tier1 | 84 | 1 | 0.012 | 296 | 154 | 0.342 |
| 147457 | 11 | 68822767 | 68822767 | G | A | SNP | <i>TPCN2</i> | ENST00000294309 | 1 | missense | c.376 | p.E126K | tier1 | 32 | 0 | 0.000 | 98 | 39 | 0.285 |
| 147457 | 11 | 72540399 | 72540399 | C | T | SNP | <i>ATG16L2</i> | ENST00000540222 | 1 | missense | c.1156 | p.R386C | tier1 | 28 | 1 | 0.035 | 316 | 147 | 0.314 |
| 147457 | 11 | 74559446 | 74559446 | G | A | SNP | <i>XRR1</i> | ENST00000340360 | -1 | missense | c.1418 | p.T473M | tier1 | 28 | 0 | 0.000 | 70 | 33 | 0.320 |
| 147457 | 11 | 78565287 | 78565287 | G | A | SNP | <i>ODZ4</i> | ENST00000278550 | -1 | missense | c.1543 | p.R515C | tier1 | 76 | 3 | 0.038 | 257 | 104 | 0.288 |
| 147457 | 11 | 99827584 | 99827584 | G | A | SNP | <i>CNTN5</i> | ENST00000279463 | 1 | silent | c.720 | p.A240 | tier1 | 245 | 4 | 0.016 | 575 | 250 | 0.303 |
| 147457 | 11 | 104872776 | 104872776 | G | A | SNP | <i>CASP5</i> | ENST00000393141 | -1 | silent | c.735 | p.D245 | tier1 | 73 | 0 | | | | |

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|--------|----|-----------|-----------|---|---|-----|-----------------|-----------------|----|-----------------------------|-------------|----------|-------|-----|---|-------|-----|-----|-------|
| 147457 | 11 | 123909293 | 123909293 | G | A | SNP | OR10G7 | ENST00000330487 | -1 | missense | c.416 | p.S139L | tier1 | 68 | 0 | 0.000 | 204 | 117 | 0.363 |
| 147457 | 12 | 3390547 | 3390547 | C | T | SNP | TSPAN9 | ENST00000011898 | 1 | intronic | c.564+52 | e5+52 | tier2 | 19 | 0 | 0.000 | 183 | 66 | 0.264 |
| 147457 | 12 | 4722773 | 4722773 | G | A | SNP | DYRK4 | ENST00000010132 | 1 | missense | c.1417 | p.G473R | tier1 | 67 | 2 | 0.029 | 169 | 82 | 0.323 |
| 147457 | 12 | 5154521 | 5154521 | G | A | SNP | KCNA5 | ENST00000252321 | 1 | missense | c.1208 | p.R403H | tier1 | 43 | 0 | 0.000 | 112 | 54 | 0.325 |
| 147457 | 12 | 7125761 | 7125761 | G | T | SNP | LPCAT3 | ENST00000261407 | -1 | 5_prime_untranslated_region | c.-33 | NULL | tier2 | 28 | 0 | 0.000 | 205 | 80 | 0.281 |
| 147457 | 12 | 7922788 | 7922788 | C | T | SNP | NANOGNB | ENST00000382119 | 1 | silent | c.312 | p.P104 | tier1 | 90 | 0 | 0.000 | 193 | 80 | 0.292 |
| 147457 | 12 | 10659128 | 10659128 | G | A | SNP | ENSG00000180574 | ENST00000538173 | 1 | silent | c.627 | p.A209 | tier1 | 224 | 3 | 0.013 | 295 | 128 | 0.303 |
| 147457 | 12 | 16713647 | 16713647 | G | A | SNP | LMO3 | ENST00000541295 | -1 | intronic | c.261-175 | e3-175 | tier2 | 45 | 0 | 0.000 | 123 | 70 | 0.363 |
| 147457 | 12 | 18644489 | 18644489 | C | T | SNP | PIK3C2G | ENST00000538779 | 1 | silent | c.2790 | p.H930 | tier1 | 74 | 0 | 0.000 | 137 | 60 | 0.303 |
| 147457 | 12 | 31135422 | 31135422 | C | T | SNP | TSPAN11 | ENST00000261177 | 1 | intronic | c.457-45 | e5-45 | tier2 | 69 | 1 | 0.014 | 216 | 85 | 0.282 |
| 147457 | 12 | 40758824 | 40758824 | G | A | SNP | LRRK2 | ENST00000298910 | 1 | silent | c.7362 | p.S2454 | tier1 | 185 | 2 | 0.011 | 172 | 87 | 0.336 |
| 147457 | 12 | 40820395 | 40820395 | C | T | SNP | MUC19 | ENST00000425730 | 1 | nonsense | c.1060 | p.R354* | tier1 | 93 | 0 | 0.000 | 183 | 78 | 0.299 |
| 147457 | 12 | 41337805 | 41337805 | C | T | SNP | CNTN1 | ENST00000347616 | 1 | nonsense | c.1516 | p.R506* | tier1 | 45 | 0 | 0.000 | 30 | 23 | 0.434 |
| 147457 | 12 | 47630017 | 47630017 | C | T | SNP | FAM113B | ENST00000432328 | 1 | missense | c.1171 | p.R391W | tier1 | 29 | 0 | 0.000 | 85 | 38 | 0.309 |
| 147457 | 12 | 48723716 | 48723716 | C | T | SNP | H1FNT | ENST00000335017 | 1 | silent | c.642 | p.D214 | tier1 | 15 | 0 | 0.000 | 204 | 119 | 0.368 |
| 147457 | 12 | 49719830 | 49719830 | C | T | SNP | TROAP | ENST00000257909 | 1 | intronic | c.634-29 | e5-29 | tier3 | 112 | 1 | 0.009 | 241 | 74 | 0.235 |
| 147457 | 12 | 56607793 | 56607793 | G | A | SNP | RNF41 | ENST00000345093 | -1 | silent | c.39 | p.D13 | tier1 | 47 | 0 | 0.000 | 174 | 59 | 0.252 |
| 147457 | 12 | 65230315 | 65230315 | G | A | SNP | TBC1D30 | ENST00000229088 | 1 | missense | c.1129 | p.V377I | tier1 | 133 | 1 | 0.007 | 152 | 77 | 0.336 |
| 147457 | 12 | 111345179 | 111345179 | G | A | SNP | CCDC63 | ENST00000308208 | 1 | missense | c.1591 | p.D531N | tier1 | 18 | 0 | 0.000 | 109 | 44 | 0.288 |
| 147457 | 12 | 117653072 | 117653072 | C | T | SNP | NOS1 | ENST00000317775 | -1 | 3_prime_untranslated_region | c.*42 | NULL | tier3 | 48 | 0 | 0.000 | 205 | 91 | 0.307 |
| 147457 | 12 | 119583293 | 119583293 | G | A | SNP | SRRM4 | ENST00000267260 | 1 | silent | c.879 | p.S293 | tier1 | 30 | 1 | 0.032 | 150 | 98 | 0.392 |
| 147457 | 12 | 123001856 | 123001856 | G | A | SNP | RSRC2 | ENST00000331738 | -1 | missense | c.520 | p.R174W | tier1 | 141 | 1 | 0.007 | 215 | 112 | 0.342 |
| 147457 | 12 | 123201344 | 123201344 | G | A | SNP | HCAR3 | ENST00000528880 | -1 | 5_prime_untranslated_region | c.-60 | NULL | tier3 | 55 | 1 | 0.018 | 174 | 55 | 0.240 |
| 147457 | 12 | 124175149 | 124175149 | G | A | SNP | TCTN2 | ENST00000303372 | 1 | missense | c.961 | p.V321I | tier1 | 86 | 1 | 0.012 | 150 | 66 | 0.306 |
| 147457 | 12 | 124422048 | 124422048 | C | T | SNP | CCDC92 | ENST00000238156 | -1 | missense | c.553 | p.V185M | tier1 | 19 | 0 | 0.000 | 100 | 58 | 0.367 |
| 147457 | 12 | 131498748 | 131498748 | G | A | SNP | GPR133 | ENST00000261654 | 1 | missense | c.1336 | p.A446T | tier1 | 36 | 1 | 0.027 | 98 | 44 | 0.310 |
| 147457 | 12 | 133181273 | 133181273 | G | A | SNP | ENSG00000204583 | ENST00000376608 | -1 | intronic | c.NULL | NULL | tier2 | 14 | 0 | 0.000 | 123 | 62 | 0.330 |
| 147457 | 13 | 21562125 | 21562125 | C | T | SNP | LATS2 | ENST00000382592 | -1 | silent | c.1794 | p.S598 | tier1 | 204 | 2 | 0.010 | 278 | 143 | 0.339 |
| 147457 | 13 | 23928871 | 23928871 | G | A | SNP | SACS | ENST00000382292 | -1 | missense | c.1880 | p.T627M | tier1 | 28 | 0 | 0.000 | 81 | 44 | 0.349 |
| 147457 | 13 | 24410420 | 24410420 | G | A | SNP | MIPEP | ENST00000382172 | -1 | nonsense | c.1612 | p.R538* | tier1 | 136 | 4 | 0.029 | 205 | 104 | 0.337 |
| 147457 | 13 | 28133957 | 28133957 | A | - | DEL | LNX2 | ENST00000316334 | -1 | intronic | c.1368+22 | e5+22 | tier2 | 49 | 0 | 0.000 | 142 | 30 | 0.174 |
| 147457 | 13 | 39450219 | 39450219 | G | A | SNP | FREM2 | ENST00000280481 | 1 | missense | c.8342 | p.R2781H | tier1 | 144 | 0 | 0.000 | 447 | 175 | 0.280 |
| 147457 | 13 | 45857726 | 45857726 | C | T | SNP | GTF2F2 | ENST00000340473 | 1 | 3_prime_untranslated_region | c.*30 | NULL | tier3 | 21 | 0 | 0.000 | 86 | 29 | 0.250 |
| 147457 | 13 | 58299227 | 58299227 | C | T | SNP | PCDH17 | ENST00000377918 | 1 | silent | c.3279 | p.S1093 | tier1 | 50 | 0 | 0.000 | 116 | 76 | 0.396 |
| 147457 | 13 | 95118904 | 95118904 | G | A | SNP | DCT | ENST00000446125 | -1 | missense | c.604 | p.R202C | tier1 | 60 | 1 | 0.016 | 168 | 58 | 0.257 |
| 147457 | 13 | 111329450 | 111329450 | G | A | SNP | CARS2 | ENST00000257347 | -1 | missense | c.656 | p.A219V | tier1 | 39 | 0 | 0.000 | 196 | 75 | 0.274 |
| 147457 | 13 | 113838700 | 113838700 | G | A | SNP | PCID2 | ENST00000246505 | -1 | silent | c.807 | p.Y269 | tier1 | 164 | 4 | 0.024 | 214 | 91 | 0.298 |
| 147457 | 14 | 21552150 | 21552150 | C | T | SNP | ARHGEF40 | ENST00000298694 | 1 | missense | c.3730 | p.R1244W | tier1 | 35 | 0 | 0.000 | 416 | 179 | 0.301 |
| 147457 | 14 | 21861926 | 21861926 | C | T | SNP | CHD8 | ENST00000399982 | -1 | missense | c.6028 | p.E2010K | tier1 | 30 | 0 | 0.000 | 138 | 73 | 0.346 |
| 147457 | 14 | 38261600 | 38261600 | G | A | SNP | TTC6 | ENST00000553443 | 1 | missense | c.3689 | p.R1230Q | tier1 | 140 | 2 | 0.014 | 229 | 71 | 0.237 |
| 147457 | 14 | 45664649 | 45664649 | G | A | SNP | FANCM | ENST00000267430 | 1 | intronic | c.5341-726 | e21-726 | tier3 | 47 | 0 | 0.000 | 80 | 31 | 0.279 |
| 147457 | 14 | 52471285 | 52471285 | C | T | SNP | C14orf166 | ENST00000261700 | 1 | 3_prime_untranslated_region | c.*51 | NULL | tier3 | 76 | 0 | 0.000 | 132 | 66 | 0.333 |
| 147457 | 14 | 57713531 | 57713531 | C | T | SNP | EXOC5 | ENST00000413566 | -1 | missense | c.168 | p.M56I | tier1 | 89 | 1 | 0.011 | 137 | 51 | 0.271 |
| 147457 | 14 | 68047847 | 68047847 | G | A | SNP | PLEKHH1 | ENST00000329153 | 1 | intronic | c.3284+92 | e22+92 | tier3 | 42 | 0 | 0.000 | 233 | 92 | 0.283 |
| 147457 | 14 | 76259462 | 76259462 | G | A | SNP | TLL5 | ENST00000298832 | 1 | intronic | c.3171+19 | e26+19 | tier3 | 19 | 0 | 0.000 | 80 | 51 | 0.389 |
| 147457 | 14 | 91211188 | 91211188 | C | T | SNP | TTC7B | ENST00000357056 | -1 | missense | c.524 | p.C175Y | tier1 | 71 | 1 | 0.014 | 171 | 77 | 0.308 |
| 147457 | 14 | 91700696 | 91700696 | G | A | SNP | GPR68 | ENST00000238699 | -1 | silent | c.729 | p.T243 | tier1 | 32 | 1 | 0.030 | 250 | 118 | 0.320 |
| 147457 | 14 | 92136153 | 92136153 | C | T | SNP | CATSPERB | ENST00000256343 | -1 | splice_region | c.1287+5 | e13+5 | tier3 | 92 | 0 | 0.000 | 142 | 63 | 0.304 |
| 147457 | 14 | 93681299 | 93681299 | G | A | SNP | UBR7 | ENST00000013070 | 1 | missense | c.466 | p.V156I | tier1 | 104 | 3 | 0.028 | 181 | 81 | 0.309 |
| 147457 | 14 | 93706769 | 93706769 | G | A | SNP | BTBD7 | ENST00000334746 | -1 | 3_prime_untranslated_region | c.*1850 | NULL | tier2 | 152 | 1 | 0.007 | 290 | 24 | 0.076 |
| 147457 | 14 | 102801015 | 102801015 | G | A | SNP | ZNF839 | ENST00000442396 | 1 | intronic | c.1509+32 | e4+32 | tier3 | 124 | 1 | 0.008 | 185 | 59 | 0.241 |
| 147457 | 14 | 105419206 | 105419206 | G | A | SNP | AHNAK2 | ENST00000333244 | -1 | missense | c.2582 | p.P861L | tier1 | 160 | 1 | 0.006 | 778 | 202 | 0.206 |
| 147457 | 15 | 23313159 | 23313159 | G | A | SNP | HERC2P2 | ENST00000560464 | -1 | 5_prime_untranslated_region | c.-23313159 | NULL | tier2 | 81 | 2 | 0.024 | 386 | 186 | 0.325 |
| 147457 | 15 | 23606950 | 23606950 | C | T | SNP | ENSG00000261739 | ENST00000562295 | 1 | intronic | c.1179+11 | e13+11 | tier2 | 45 | 0 | 0.000 | 283 | 132 | 0.317 |
| 147457 | 15 | 25349836 | 25349836 | C | T | SNP | SNORD116-28 | ENST00000516123 | 1 | rna | NULL | NULL | tier1 | 115 | 0 | 0.000 | 218 | 102 | 0.319 |
| 147457 | 15 | 28517461 | 28517461 | C | T | SNP | HERC2 | ENST00000261609 | -1 | missense | c.983 | p.R328H | tier1 | 160 | 1 | 0.006 | 819 | 61 | 0.069 |
| 147457 | 15 | 33358756 | 33358756 | G | A | SNP | FMN1 | ENST00000334528 | -1 | missense | c.1330 | p.R444W | tier1 | 59 | 0 | 0.000 | 144 | 61 | 0.298 |
| 147457 | 15 | 33936693 | 33936693 | C | T | SNP | RYR3 | ENST00000389232 | 1 | silent | c.3738 | p.N1246 | tier1 | 145 | 2 | 0.014 | 481 | 250 | 0.341 |
| 147457 | 15 | 40099429 | 40099429 | C | T | SNP | GPR176 | ENST00000561100 | -1 | missense | c.203 | p.R68H | tier1 | 78 | 0 | 0.000 | 232 | 82 | 0.261 |
| 147457 | 15 | 40532849 | 40532849 | C | T | SNP | ENSG00000259288 | ENST00000558658 | 1 | missense | c.131 | p.T44M | tier1 | 26 | 0 | 0.000 | 308 | 135 | 0.304 |
| 147457 | 15 | 40583762 | 40583762 | C | T | SNP | PLCB2 | ENST00000558588 | -1 | 3_prime_untranslated_region | c.*205 | NULL | tier2 | 16 | 0 | 0.000 | 63 | 39 | 0.382 |
| 147457 | 15 | 40993282 | 40993282 | C | T | SNP | RAD51 | ENST00000382643 | 1 | silent | c.108 | p.N36 | tier1 | 96 | 5 | 0.050 | 176 | 64 | 0.267 |
| 147457 | 15 | 44673070 | 44673070 | G | A | SNP | CASC4 | ENST00000299957 | 1 | missense | c.968 | p.R323H | tier1 | 208 | 3 | 0.014 | 298 | 109 | 0.268 |
| 147457 | 15 | 52622645 | 52622645 | C | T | SNP | MYO5A | ENST00000358212 | -1 | missense | c.4460 | p.R1487Q | tier1 | 146 | 0 | 0.000 | 252 | 77 | 0.234 |
| 147457 | 15 | 63964722 | 63964722 | G | A | SNP | HERC1 | ENST00000443617 | -1 | missense | c.8018 | p.P2673L | tier1 | 48 | 0 | 0.000 | 157 | 66 | 0.296 |
| 147457 | 15 | 64218300 | 64218300 | C | T | SNP | DAPK2 | ENST00000557867 | -1 | 5_prime_untranslated_region | c.-64218300 | NULL | tier2 | 28 | 0 | 0.000 | 67 | 28 | 0.295 |
| 147457 | 15 | 66181422 | 66181422 | C | T | SNP | RAB11A | ENST00000261890 | 1 | 3_prime_untranslated_region | c.*1244 | NULL | tier3 | 72 | 0 | 0.000 | 91 | 35 | 0.273 |
| 147457 | 15 | 68480403 | 68480403 | A | - | DEL | PIAS1 | ENST00000563996 | 1 | 3_prime_untranslated_region | c.*474 | NULL | tier4 | 22 | 0 | 0.000 | 47 | 24 | 0.338 |
| 147457 | 15 | 68603671 | 68603671 | C | T | SNP | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|---|---------|-----|------------------------|-----------------|-----|-----------------------------|-----------------|----------|-------|-----|---|-------|------|-----|-------|
| 147457 | 15 | 83931848 | 83931848 | G | A | SNP | <i>BNC1</i> | ENST00000345382 | -1 | missense | c.2155 | p.R719C | tier1 | 58 | 1 | 0.017 | 128 | 54 | 0.297 |
| 147457 | 15 | 85805937 | 85805937 | C | T | SNP | <i>ENSG00000218052</i> | ENST00000561209 | -1 | 5_prime_untranslated_region | c.-85805937 | NULL | tier3 | 30 | 1 | 0.032 | 269 | 131 | 0.328 |
| 147457 | 15 | 90192982 | 90192982 | G | A | SNP | <i>KIF7</i> | ENST00000394412 | -1 | silent | c.519 | p.R173 | tier1 | 18 | 0 | 0.000 | 62 | 43 | 0.410 |
| 147457 | 15 | 90631934 | 90631934 | C | T | SNP | <i>IDH2</i> | ENST00000330062 | -1 | missense | c.419 | p.R140Q | tier1 | 173 | 1 | 0.006 | 705 | 334 | 0.321 |
| 147457 | 15 | 91577490 | 91577490 | G | A | SNP | <i>ENSG00000214432</i> | ENST00000557804 | 1 | intronic | c.NULL | NULL | tier3 | 84 | 0 | 0.000 | 322 | 152 | 0.321 |
| 147457 | 15 | 99512919 | 99512919 | C | T | SNP | <i>PGPEP1L</i> | ENST00000378919 | -1 | intronic | c.145-39 | e3-39 | tier3 | 27 | 0 | 0.000 | 181 | 82 | 0.311 |
| 147457 | 15 | 99671249 | 99671249 | C | T | SNP | <i>SYNM</i> | ENST00000336292 | 1 | missense | c.2681 | p.A894V | tier1 | 38 | 0 | 0.000 | 184 | 66 | 0.264 |
| 147457 | 15 | 100821502 | 100821502 | C | T | SNP | <i>ADAMTS17</i> | ENST00000268070 | -1 | missense | c.721 | p.D241N | tier1 | 78 | 0 | 0.000 | 381 | 175 | 0.314 |
| 147457 | 15 | 102292626 | 102292626 | G | A | SNP | <i>ENSG00000259660</i> | ENST00000561463 | 1 | intronic | c.NULL | NULL | tier3 | 57 | 0 | 0.000 | 272 | 122 | 0.309 |
| 147457 | 16 | 593258 | 593258 | C | T | SNP | <i>SOLH</i> | ENST00000219611 | 1 | intronic | c.1-3559 | e1-3559 | tier3 | 31 | 0 | 0.000 | 317 | 152 | 0.324 |
| 147457 | 16 | 721798 | 721799 | - | CCCCCTG | INS | <i>MSLN</i> | ENST00000446427 | 1 | intronic | c.1+28495_28496 | e0+28495 | tier4 | 19 | 0 | 0.000 | 84 | 33 | 0.282 |
| 147457 | 16 | 1255229 | 1255229 | C | T | SNP | <i>CACNA1H</i> | ENST00000348261 | 1 | missense | c.2567 | p.P856L | tier1 | 47 | 0 | 0.000 | 170 | 78 | 0.312 |
| 147457 | 16 | 1397840 | 1397840 | C | T | SNP | <i>BAIAP3</i> | ENST00000324385 | 1 | intronic | c.3130+18 | e31+18 | tier2 | 25 | 0 | 0.000 | 161 | 88 | 0.352 |
| 147457 | 16 | 2129600 | 2129600 | G | A | SNP | <i>TSC2</i> | ENST00000219476 | 1 | silent | c.3327 | p.P1109 | tier1 | 26 | 1 | 0.037 | 318 | 145 | 0.313 |
| 147457 | 16 | 4431260 | 4431260 | C | T | SNP | <i>VASN</i> | ENST00000304735 | 1 | missense | c.382 | p.R128C | tier1 | 28 | 0 | 0.000 | 182 | 74 | 0.289 |
| 147457 | 16 | 11001032 | 11001032 | C | T | SNP | <i>CITA</i> | ENST00000324288 | 1 | silent | c.1683 | p.D561 | tier1 | 27 | 0 | 0.000 | 161 | 62 | 0.276 |
| 147457 | 16 | 11121344 | 11121344 | G | A | SNP | <i>ENSG00000229917</i> | ENST00000425129 | 1 | 3_prime_untranslated_region | c.*11121344 | NULL | tier3 | 148 | 2 | 0.013 | 213 | 97 | 0.312 |
| 147457 | 16 | 15457736 | 15457736 | G | A | SNP | <i>LOC642778</i> | ENST00000360151 | -1 | missense | c.833 | p.A278V | tier1 | 321 | 0 | 0.000 | 1467 | 151 | 0.093 |
| 147457 | 16 | 28914739 | 28914739 | C | T | SNP | <i>ATP2A1</i> | ENST00000564112 | 1 | missense | c.46 | p.R16C | tier1 | 122 | 3 | 0.024 | 234 | 107 | 0.313 |
| 147457 | 16 | 31519174 | 31519174 | G | A | SNP | <i>C16orf58</i> | ENST00000327237 | -1 | missense | c.326 | p.S109F | tier1 | 28 | 0 | 0.000 | 220 | 80 | 0.266 |
| 147457 | 16 | 33740957 | 33740957 | C | T | SNP | <i>ENSG00000260781</i> | ENST00000567025 | -1 | 5_prime_flanking_region | c.-1323 | NULL | tier3 | 333 | 2 | 0.006 | 1132 | 225 | 0.166 |
| 147457 | 16 | 46638006 | 46638006 | G | A | SNP | <i>SHCBP1</i> | ENST00000303383 | -1 | missense | c.973 | p.R325C | tier1 | 35 | 0 | 0.000 | 88 | 44 | 0.333 |
| 147457 | 16 | 56397961 | 56397961 | G | A | SNP | <i>AMFR</i> | ENST00000290649 | -1 | silent | c.1656 | p.G552 | tier1 | 22 | 0 | 0.000 | 97 | 53 | 0.353 |
| 147457 | 16 | 57752349 | 57752349 | G | A | SNP | <i>CCDC135</i> | ENST00000360716 | 1 | missense | c.1168 | p.D390N | tier1 | 122 | 1 | 0.008 | 267 | 112 | 0.296 |
| 147457 | 16 | 58550812 | 58550812 | G | A | SNP | <i>SETD6</i> | ENST00000219315 | 1 | missense | c.772 | p.A258T | tier1 | 59 | 0 | 0.000 | 161 | 54 | 0.251 |
| 147457 | 16 | 67469912 | 67469912 | G | A | SNP | <i>HSD11B2</i> | ENST00000326152 | 1 | silent | c.531 | p.A177 | tier1 | 29 | 0 | 0.000 | 138 | 60 | 0.303 |
| 147457 | 16 | 67573993 | 67573993 | G | A | SNP | <i>FAM65A</i> | ENST00000422602 | 1 | missense | c.512 | p.R171H | tier1 | 94 | 2 | 0.021 | 491 | 217 | 0.307 |
| 147457 | 16 | 83378566 | 83378566 | C | T | SNP | <i>CDH13</i> | ENST00000566620 | 1 | missense | c.736 | p.R246W | tier1 | 43 | 0 | 0.000 | 141 | 65 | 0.316 |
| 147457 | 16 | 83520057 | 83520057 | G | A | SNP | <i>CDH13</i> | ENST00000566620 | 1 | intronic | c.782-25 | e7-25 | tier3 | 77 | 1 | 0.013 | 198 | 51 | 0.204 |
| 147457 | 16 | 83994327 | 83994327 | G | A | SNP | <i>OSGIN1</i> | ENST00000361711 | 1 | missense | c.358 | p.V120M | tier1 | 29 | 0 | 0.000 | 220 | 79 | 0.264 |
| 147457 | 16 | 84529437 | 84529437 | G | A | SNP | <i>KIAA1609</i> | ENST00000343629 | -1 | missense | c.236 | p.A79V | tier1 | 25 | 0 | 0.000 | 106 | 46 | 0.301 |
| 147457 | 16 | 88747899 | 88747899 | G | A | SNP | <i>SNAI3</i> | ENST00000332281 | -1 | silent | c.300 | p.A100 | tier1 | 66 | 0 | 0.000 | 440 | 176 | 0.285 |
| 147457 | 16 | 89178494 | 89178494 | T | C | SNP | <i>ACSF3</i> | ENST00000317447 | 1 | splice_region | c.823-6 | e3-6 | tier3 | 79 | 1 | 0.013 | 241 | 30 | 0.111 |
| 147457 | 17 | 3526660 | 3526660 | G | A | SNP | <i>SHPK</i> | ENST00000225519 | -1 | missense | c.620 | p.T207M | tier1 | 21 | 0 | 0.000 | 159 | 78 | 0.329 |
| 147457 | 17 | 6377943 | 6377943 | G | A | SNP | <i>PITPNM3</i> | ENST00000262483 | -1 | intronic | c.1086-19 | e10-19 | tier2 | 15 | 0 | 0.000 | 251 | 122 | 0.277 |
| 147457 | 17 | 7492394 | 7492394 | G | A | SNP | <i>MPDU1</i> | ENST00000301597 | 1 | 3_prime_untranslated_region | c.*586 | NULL | tier2 | 16 | 0 | 0.000 | 135 | 75 | 0.357 |
| 147457 | 17 | 7506344 | 7506344 | C | T | SNP | <i>FXR2</i> | ENST00000250113 | -1 | intronic | c.450-24 | e6-24 | tier2 | 91 | 0 | 0.000 | 107 | 58 | 0.352 |
| 147457 | 17 | 8216386 | 8216386 | C | T | SNP | <i>ARHGEF15</i> | ENST00000361926 | 1 | missense | c.748 | p.R250C | tier1 | 26 | 0 | 0.000 | 306 | 120 | 0.281 |
| 147457 | 17 | 9632002 | 9632002 | G | A | SNP | <i>USP43</i> | ENST00000285199 | 1 | missense | c.3067 | p.V1023I | tier1 | 20 | 0 | 0.000 | 172 | 62 | 0.265 |
| 147457 | 17 | 10322056 | 10322056 | G | A | SNP | <i>MYH8</i> | ENST00000252173 | -1 | silent | c.417 | p.P139 | tier1 | 121 | 0 | 0.000 | 378 | 164 | 0.303 |
| 147457 | 17 | 10404747 | 10404747 | G | A | SNP | <i>MYH1</i> | ENST00000226207 | -1 | missense | c.3418 | p.R1140C | tier1 | 150 | 0 | 0.000 | 520 | 235 | 0.310 |
| 147457 | 17 | 11631100 | 11631100 | C | T | SNP | <i>DNAH9</i> | ENST00000262442 | 1 | intronic | c.5710-35 | e28-35 | tier2 | 123 | 5 | 0.039 | 306 | 124 | 0.288 |
| 147457 | 17 | 16330812 | 16330812 | C | T | SNP | <i>TRPV2</i> | ENST00000338560 | 1 | missense | c.1301 | p.T434M | tier1 | 32 | 0 | 0.000 | 186 | 85 | 0.314 |
| 147457 | 17 | 16344404 | 16344404 | G | A | SNP | <i>C17orf76-AS1</i> | ENST00000460249 | 1 | 3_prime_untranslated_region | c.*16344404 | NULL | tier2 | 79 | 0 | 0.000 | 206 | 81 | 0.281 |
| 147457 | 17 | 16981370 | 16981370 | C | T | SNP | <i>MPRIP</i> | ENST00000395811 | 1 | missense | c.247 | p.R83C | tier1 | 38 | 0 | 0.000 | 168 | 78 | 0.317 |
| 147457 | 17 | 18149237 | 18149237 | C | T | SNP | <i>FLII</i> | ENST00000465046 | -1 | 5_prime_untranslated_region | c.-18149237 | NULL | tier4 | 21 | 0 | 0.000 | 186 | 83 | 0.307 |
| 147457 | 17 | 18996977 | 18996977 | T | G | SNP | <i>LOC100132472</i> | ENST00000428928 | 1 | missense | c.315 | p.S105R | tier1 | 49 | 1 | 0.020 | 537 | 220 | 0.291 |
| 147457 | 17 | 19315938 | 19315938 | G | A | SNP | <i>RNF112</i> | ENST00000453070 | 1 | missense | c.79 | p.D27N | tier1 | 14 | 0 | 0.000 | 110 | 69 | 0.386 |
| 147457 | 17 | 26574933 | 26574933 | G | A | SNP | <i>ENSG00000197825</i> | ENST00000355177 | -1 | 5_prime_flanking_region | c.-44102 | NULL | tier2 | 23 | 0 | 0.000 | 101 | 56 | 0.357 |
| 147457 | 17 | 26817411 | 26817411 | C | T | SNP | <i>SLC13A2</i> | ENST00000444914 | 1 | silent | c.318 | p.L106 | tier1 | 50 | 0 | 0.000 | 172 | 79 | 0.312 |
| 147457 | 17 | 27013676 | 27013676 | G | A | SNP | <i>SUPT6H</i> | ENST00000314616 | 1 | missense | c.2569 | p.A857T | tier1 | 47 | 1 | 0.021 | 171 | 59 | 0.257 |
| 147457 | 17 | 28395764 | 28395764 | C | T | SNP | <i>EFCAB5</i> | ENST00000394835 | 1 | intronic | c.2737+9045 | e14+9045 | tier2 | 51 | 0 | 0.000 | 212 | 90 | 0.298 |
| 147457 | 17 | 29533403 | 29533403 | C | T | SNP | <i>NF1</i> | ENST00000358273 | 1 | intronic | c.1392+14 | e12+14 | tier2 | 184 | 8 | 0.042 | 395 | 168 | 0.298 |
| 147457 | 17 | 31320043 | 31320043 | C | T | SNP | <i>SPACA3</i> | ENST00000269053 | 1 | intronic | c.34+1053 | e1+1053 | tier2 | 81 | 1 | 0.012 | 301 | 103 | 0.254 |
| 147457 | 17 | 33328217 | 33328217 | G | A | SNP | <i>LIG3</i> | ENST00000378526 | 1 | intronic | c.2332-59 | e16-59 | tier4 | 23 | 0 | 0.000 | 78 | 30 | 0.278 |
| 147457 | 17 | 34312816 | 34312816 | G | A | SNP | <i>CCL14</i> | ENST00000435911 | -1 | missense | c.95 | p.P32L | tier1 | 87 | 2 | 0.023 | 184 | 67 | 0.267 |
| 147457 | 17 | 36622583 | 36622583 | G | A | SNP | <i>ARHGAP23</i> | ENST00000431231 | 1 | missense | c.659 | p.R220Q | tier1 | 38 | 0 | 0.000 | 344 | 161 | 0.318 |
| 147457 | 17 | 36689701 | 36689701 | G | A | SNP | <i>SRCIN1</i> | ENST00000264659 | -1 | intronic | c.3418-35 | e19-35 | tier2 | 20 | 0 | 0.000 | 217 | 141 | 0.394 |
| 147457 | 17 | 37075080 | 37075080 | C | T | SNP | <i>LASP1</i> | ENST00000318008 | 1 | 3_prime_untranslated_region | c.*49 | NULL | tier2 | 33 | 0 | 0.000 | 150 | 58 | 0.279 |
| 147457 | 17 | 38146349 | 38146349 | C | T | SNP | <i>PSMD3</i> | ENST00000264639 | 1 | nonsense | c.880 | p.R294* | tier1 | 49 | 1 | 0.020 | 224 | 86 | 0.277 |
| 147457 | 17 | 39740595 | 39740595 | C | T | SNP | <i>KRT14</i> | ENST00000167586 | -1 | missense | c.679 | p.E227K | tier1 | 116 | 3 | 0.025 | 419 | 156 | 0.271 |
| 147457 | 17 | 39959603 | 39959603 | G | A | SNP | <i>LEPREL4</i> | ENST00000465097 | -1 | missense | c.134 | p.T45M | tier1 | 26 | 0 | 0.000 | 162 | 69 | 0.299 |
| 147457 | 17 | 40063711 | 40063711 | C | T | SNP | <i>ACLY</i> | ENST00000401700 | -1 | missense | c.893 | p.R298Q | tier1 | 32 | 0 | 0.000 | 201 | 111 | 0.356 |
| 147457 | 17 | 41026210 | 41026210 | C | T | SNP | <i>ENSG00000260105</i> | ENST00000570207 | 1 | 3_prime_untranslated_region | c.*41026210 | NULL | tier2 | 109 | 0 | 0.000 | 322 | 106 | 0.247 |
| 147457 | 17 | 44144025 | 44144025 | G | A | SNP | <i>KANSL1</i> | ENST00000262419 | -1 | nonsense | c.1726 | p.R576* | tier1 | 196 | 2 | 0.010 | 616 | 273 | 0.307 |
| 147457 | 17 | 48434597 | 48434597 | G | A | SNP | <i>XYLT2</i> | ENST00000017003 | 1 | missense | c.1925 | p.R642Q | tier1 | 39 | 0 | 0.000 | 380 | 161 | 0.296 |
| 147457 | 17 | 52993020 | 52993020 | C | T | SNP | <i>TOM1L1</i> | ENST00000575882 | 1</ | | | | | | | | | | |

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|--------|----|----------|----------|---|---|-----|-----------|-----------------|----|-----------------------------|-------------|----------|-------|-----|---|-------|-----|-----|-------|
| 147457 | 17 | 76167847 | 76167847 | C | T | SNP | SYNGR2 | ENST00000225777 | 1 | missense | c.505 | p.R169C | tier1 | 18 | 0 | 0.000 | 148 | 53 | 0.264 |
| 147457 | 17 | 76803689 | 76803689 | C | T | SNP | USP36 | ENST00000312010 | -1 | silent | c.1437 | p.P479 | tier1 | 14 | 0 | 0.000 | 54 | 31 | 0.365 |
| 147457 | 17 | 77081692 | 77081692 | C | T | SNP | ENGASE | ENST00000300682 | 1 | splice_region | c.1701-10 | e13-10 | tier3 | 18 | 0 | 0.000 | 101 | 41 | 0.289 |
| 147457 | 17 | 77083117 | 77083117 | G | A | SNP | ENGASE | ENST00000300682 | 1 | 3_prime_untranslated_region | c.*686 | NULL | tier3 | 24 | 1 | 0.040 | 175 | 90 | 0.340 |
| 147457 | 17 | 78247071 | 78247071 | G | A | SNP | RNF213 | ENST00000508628 | 1 | silent | c.129 | p.S43 | tier1 | 281 | 6 | 0.021 | 830 | 332 | 0.285 |
| 147457 | 18 | 5397381 | 5397381 | G | A | SNP | EPB41L3 | ENST00000341928 | -1 | silent | c.2517 | p.P839 | tier1 | 27 | 0 | 0.000 | 95 | 42 | 0.302 |
| 147457 | 18 | 6837314 | 6837314 | C | T | SNP | ARHGAP28 | ENST00000400091 | 1 | silent | c.444 | p.A148 | tier1 | 89 | 0 | 0.000 | 243 | 88 | 0.265 |
| 147457 | 18 | 11868565 | 11868565 | G | A | SNP | GNAL | ENST00000334049 | 1 | missense | c.934 | p.A312T | tier1 | 67 | 1 | 0.015 | 168 | 62 | 0.268 |
| 147457 | 18 | 13072806 | 13072806 | C | T | SNP | CEP192 | ENST00000506447 | 1 | nonsense | c.5401 | p.R1801* | tier1 | 166 | 2 | 0.012 | 197 | 98 | 0.332 |
| 147457 | 18 | 19024347 | 19024347 | C | T | SNP | GREB1L | ENST00000424526 | 1 | missense | c.1370 | p.T457M | tier1 | 129 | 0 | 0.000 | 231 | 78 | 0.252 |
| 147457 | 18 | 19780629 | 19780629 | C | T | SNP | GATA6 | ENST00000269216 | 1 | missense | c.1631 | p.P544L | tier1 | 54 | 1 | 0.018 | 178 | 98 | 0.355 |
| 147457 | 18 | 24126926 | 24126926 | G | A | SNP | KCTD1 | ENST00000417602 | -1 | silent | c.1575 | p.D525 | tier1 | 20 | 0 | 0.000 | 97 | 43 | 0.307 |
| 147457 | 18 | 29046693 | 29046693 | G | A | SNP | DSG3 | ENST00000257189 | 1 | missense | c.1612 | p.V538I | tier1 | 53 | 3 | 0.054 | 124 | 48 | 0.278 |
| 147457 | 18 | 33605730 | 33605730 | G | A | SNP | RPRD1A | ENST00000357384 | -1 | intronic | c.789+1133 | e6+1133 | tier2 | 71 | 0 | 0.000 | 138 | 55 | 0.285 |
| 147457 | 18 | 34340699 | 34340699 | C | T | SNP | FHOD3 | ENST00000257209 | 1 | silent | c.4029 | p.D1343 | tier1 | 28 | 0 | 0.000 | 145 | 59 | 0.288 |
| 147457 | 18 | 47527556 | 47527556 | C | T | SNP | MYO5B | ENST00000285039 | -1 | intronic | c.612+69 | e5+69 | tier2 | 75 | 0 | 0.000 | 498 | 212 | 0.298 |
| 147457 | 19 | 648900 | 648900 | C | T | SNP | RNF126 | ENST00000340092 | -1 | missense | c.652 | p.V218I | tier1 | 94 | 2 | 0.021 | 333 | 185 | 0.357 |
| 147457 | 19 | 806592 | 806592 | C | T | SNP | PTBP1 | ENST00000356948 | 1 | intronic | c.1119+36 | e10+36 | tier2 | 56 | 0 | 0.000 | 187 | 100 | 0.347 |
| 147457 | 19 | 1046266 | 1046266 | G | A | SNP | ABCA7 | ENST00000263094 | 1 | missense | c.1483 | p.D495N | tier1 | 53 | 0 | 0.000 | 472 | 164 | 0.257 |
| 147457 | 19 | 3976654 | 3976654 | G | A | SNP | EEF2 | ENST00000309311 | -1 | silent | c.2475 | p.F825 | tier1 | 37 | 0 | 0.000 | 342 | 124 | 0.264 |
| 147457 | 19 | 4491450 | 4491450 | C | T | SNP | HDFFRP2 | ENST00000301284 | 1 | intronic | c.490-113 | e5-113 | tier3 | 25 | 0 | 0.000 | 132 | 59 | 0.309 |
| 147457 | 19 | 4511513 | 4511513 | G | A | SNP | PLIN4 | ENST00000301286 | -1 | missense | c.2417 | p.T806M | tier1 | 54 | 0 | 0.000 | 368 | 48 | 0.115 |
| 147457 | 19 | 4930798 | 4930798 | C | T | SNP | UHRF1 | ENST00000398240 | 1 | missense | c.518 | p.T173M | tier1 | 29 | 0 | 0.000 | 169 | 62 | 0.268 |
| 147457 | 19 | 7267653 | 7267653 | C | T | SNP | INSR | ENST00000302850 | -1 | missense | c.355 | p.A119T | tier1 | 24 | 0 | 0.000 | 90 | 35 | 0.280 |
| 147457 | 19 | 7620282 | 7620282 | G | A | SNP | PNPLA6 | ENST00000414982 | 1 | intronic | c.2966+29 | e24+29 | tier2 | 12 | 0 | 0.000 | 68 | 43 | 0.387 |
| 147457 | 19 | 8027941 | 8027941 | C | T | SNP | ELAVL1 | ENST00000407627 | -1 | 3_prime_untranslated_region | c.*426 | NULL | tier3 | 75 | 3 | 0.039 | 147 | 66 | 0.310 |
| 147457 | 19 | 8138001 | 8138001 | G | A | SNP | FBN3 | ENST00000270509 | -1 | missense | c.7883 | p.T2628M | tier1 | 39 | 0 | 0.000 | 283 | 123 | 0.303 |
| 147457 | 19 | 8191296 | 8191296 | C | T | SNP | FBN3 | ENST00000270509 | -1 | intronic | c.2554+56 | e19+56 | tier3 | 39 | 1 | 0.025 | 367 | 147 | 0.285 |
| 147457 | 19 | 8617070 | 8617070 | C | T | SNP | MYO1F | ENST00000338257 | -1 | intronic | c.505-22 | e7-22 | tier3 | 32 | 0 | 0.000 | 111 | 39 | 0.258 |
| 147457 | 19 | 10099957 | 10099957 | G | A | SNP | COL5A3 | ENST00000264828 | -1 | intronic | c.2091+41 | e26+41 | tier3 | 109 | 2 | 0.018 | 460 | 190 | 0.292 |
| 147457 | 19 | 10753529 | 10753529 | C | T | SNP | SLC44A2 | ENST00000335757 | 1 | intronic | c.2014+402 | e21+402 | tier2 | 47 | 1 | 0.021 | 190 | 67 | 0.260 |
| 147457 | 19 | 13007031 | 13007031 | G | A | SNP | GCDH | ENST00000222214 | 1 | silent | c.648 | p.S216 | tier1 | 66 | 1 | 0.015 | 305 | 119 | 0.280 |
| 147457 | 19 | 13409955 | 13409955 | C | T | SNP | CACNA1A | ENST00000418012 | -1 | missense | c.2504 | p.R835H | tier1 | 89 | 4 | 0.043 | 413 | 140 | 0.253 |
| 147457 | 19 | 13441256 | 13441256 | C | T | SNP | CACNA1A | ENST00000418012 | -1 | intronic | c.1256-106 | e10-106 | tier4 | 41 | 0 | 0.000 | 75 | 29 | 0.279 |
| 147457 | 19 | 13930271 | 13930271 | G | A | SNP | ZSWIM4 | ENST00000254323 | 1 | silent | c.1674 | p.F558 | tier1 | 36 | 0 | 0.000 | 270 | 111 | 0.291 |
| 147457 | 19 | 14071785 | 14071785 | G | A | SNP | DCAF15 | ENST00000254337 | 1 | missense | c.1781 | p.R594Q | tier1 | 22 | 0 | 0.000 | 213 | 92 | 0.302 |
| 147457 | 19 | 14211653 | 14211653 | C | T | SNP | PRKACA | ENST00000308677 | -1 | missense | c.404 | p.R135Q | tier1 | 26 | 0 | 0.000 | 136 | 58 | 0.299 |
| 147457 | 19 | 14218255 | 14218255 | C | T | SNP | PRKACA | ENST00000308677 | -1 | intronic | c.47-34 | e2-34 | tier3 | 105 | 1 | 0.009 | 172 | 91 | 0.346 |
| 147457 | 19 | 15760007 | 15760007 | G | A | SNP | CYP4F3 | ENST00000221307 | 1 | missense | c.563 | p.R188H | tier1 | 97 | 0 | 0.000 | 344 | 143 | 0.293 |
| 147457 | 19 | 16855367 | 16855367 | G | A | SNP | NWD1 | ENST00000379808 | 1 | missense | c.334 | p.E112K | tier1 | 39 | 0 | 0.000 | 180 | 96 | 0.348 |
| 147457 | 19 | 17893967 | 17893967 | C | T | SNP | FCHO1 | ENST00000252771 | 1 | silent | c.2079 | p.A693 | tier1 | 32 | 1 | 0.030 | 124 | 50 | 0.287 |
| 147457 | 19 | 20308488 | 20308488 | G | A | SNP | ZNF486 | ENST00000335117 | 1 | silent | c.969 | p.T323 | tier1 | 133 | 2 | 0.015 | 219 | 96 | 0.304 |
| 147457 | 19 | 23927554 | 23927554 | C | T | SNP | ZNF681 | ENST00000402377 | -1 | silent | c.798 | p.S266 | tier1 | 107 | 1 | 0.009 | 209 | 56 | 0.211 |
| 147457 | 19 | 33487078 | 33487078 | C | T | SNP | RHPN2 | ENST00000254260 | -1 | missense | c.1274 | p.R425Q | tier1 | 27 | 0 | 0.000 | 153 | 66 | 0.301 |
| 147457 | 19 | 34925778 | 34925778 | C | T | SNP | UBA2 | ENST00000246548 | 1 | nonsense | c.364 | p.R122* | tier1 | 89 | 0 | 0.000 | 195 | 69 | 0.260 |
| 147457 | 19 | 38954130 | 38954130 | C | T | SNP | RYR1 | ENST00000359596 | 1 | missense | c.2645 | p.A882V | tier1 | 131 | 0 | 0.000 | 670 | 240 | 0.263 |
| 147457 | 19 | 40398322 | 40398322 | G | A | SNP | FCGBP | ENST00000221347 | -1 | silent | c.6645 | p.N2215 | tier1 | 170 | 1 | 0.006 | 770 | 207 | 0.212 |
| 147457 | 19 | 42091848 | 42091848 | G | A | SNP | CEACAM21 | ENST00000401445 | 1 | missense | c.850 | p.G284S | tier1 | 49 | 0 | 0.000 | 169 | 75 | 0.305 |
| 147457 | 19 | 42525580 | 42525580 | G | A | SNP | GRIK5 | ENST00000301218 | -1 | missense | c.1744 | p.R582C | tier1 | 72 | 0 | 0.000 | 332 | 120 | 0.266 |
| 147457 | 19 | 44891009 | 44891009 | C | T | SNP | ZFP112 | ENST00000330997 | -1 | silent | c.1398 | p.A466 | tier1 | 112 | 1 | 0.009 | 247 | 109 | 0.306 |
| 147457 | 19 | 47910756 | 47910756 | C | T | SNP | MEIS3 | ENST00000559524 | -1 | missense | c.926 | p.R309Q | tier1 | 23 | 0 | 0.000 | 228 | 84 | 0.269 |
| 147457 | 19 | 48956016 | 48956016 | G | A | SNP | GRWD1 | ENST00000253237 | 1 | missense | c.1075 | p.V359I | tier1 | 34 | 0 | 0.000 | 248 | 94 | 0.275 |
| 147457 | 19 | 49113254 | 49113254 | C | T | SNP | FAM83E | ENST00000263266 | -1 | missense | c.637 | p.V213M | tier1 | 30 | 1 | 0.032 | 168 | 72 | 0.300 |
| 147457 | 19 | 49526129 | 49526129 | G | A | SNP | CGB | ENST00000357383 | -1 | 3_prime_untranslated_region | c.*14 | NULL | tier2 | 34 | 0 | 0.000 | 185 | 95 | 0.339 |
| 147457 | 19 | 50563467 | 50563467 | C | T | SNP | FLJ26850 | ENST00000527209 | 1 | intronic | c.NULL | NULL | tier3 | 60 | 0 | 0.000 | 95 | 38 | 0.286 |
| 147457 | 19 | 51128500 | 51128500 | G | A | SNP | SYT3 | ENST00000338916 | -1 | silent | c.1626 | p.A542 | tier1 | 36 | 0 | 0.000 | 240 | 137 | 0.363 |
| 147457 | 19 | 53208386 | 53208386 | C | T | SNP | ZNF611 | ENST00000319783 | -1 | missense | c.1922 | p.R641H | tier1 | 173 | 2 | 0.011 | 393 | 153 | 0.280 |
| 147457 | 19 | 54204449 | 54204449 | C | T | SNP | MIR521-2 | ENST00000384818 | 1 | 5_prime_flanking_region | c.-15399 | NULL | tier3 | 114 | 5 | 0.042 | 310 | 111 | 0.264 |
| 147457 | 19 | 54760964 | 54760964 | C | T | SNP | LILRB5 | ENST00000463460 | -1 | 5_prime_untranslated_region | c.-54760964 | NULL | tier3 | 32 | 0 | 0.000 | 191 | 83 | 0.303 |
| 147457 | 19 | 55710221 | 55710221 | G | A | SNP | PTPRH | ENST00000376350 | -1 | missense | c.1480 | p.R494C | tier1 | 29 | 0 | 0.000 | 207 | 95 | 0.315 |
| 147457 | 19 | 55870015 | 55870015 | G | A | SNP | FAM71E2 | ENST00000424985 | -1 | missense | c.2221 | p.R741C | tier1 | 37 | 0 | 0.000 | 150 | 70 | 0.318 |
| 147457 | 19 | 55943894 | 55943894 | G | A | SNP | SHISA7 | ENST00000376325 | -1 | 3_prime_untranslated_region | c.*629 | NULL | tier3 | 44 | 0 | 0.000 | 118 | 44 | 0.272 |
| 147457 | 19 | 57133465 | 57133465 | C | T | SNP | ZNF71 | ENST00000328070 | 1 | silent | c.810 | p.Y270 | tier1 | 32 | 0 | 0.000 | 311 | 142 | 0.313 |
| 147457 | 19 | 59061551 | 59061551 | G | A | SNP | TRIM28 | ENST00000253024 | 1 | missense | c.2231 | p.R744H | tier1 | 17 | 0 | 0.000 | 120 | 54 | 0.310 |
| 147457 | 20 | 2842746 | 2842746 | G | A | SNP | VPS16 | ENST00000380445 | 1 | silent | c.1041 | p.A347 | tier1 | 49 | 0 | 0.000 | 176 | 64 | 0.266 |
| 147457 | 20 | 3147452 | 3147452 | C | T | SNP | ProSAPIP1 | ENST00000329152 | -1 | missense | c.358 | p.G120R | tier1 | 21 | 0 | 0.000 | 111 | 53 | 0.321 |
| 147457 | 20 | 4705892 | 4705892 | C | T | SNP | PRND | ENST00000305817 | 1 | 3_prime_untranslated_region | c.*164 | NULL | tier3 | 31 | 0 | 0.000 | 114 | 58 | 0.337 |
| 147457 | 20 | 4707604 | 4707604 | G | A | SNP | PRND | ENST00000305817 | 1 | 3_prime_untranslated_region | c.*1876 | NULL | tier3 | 63 | 3 | 0.046 | 132 | 62 | 0.318 |
| 147457 | 20 | 18805846 | 18805846 | C | T | SNP | C20orf78 | ENST00000278779 | -1 | intronic | c.116+64 | e2+64 | | | | | | | |

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|--------|----|-----------|-----------|---|---|-----|-----------------|-----------------|----|-----------------------------|-----------------|----------|-------|-----|----|-------|-----|-----|-------|
| 147457 | 20 | 40118424 | 40118424 | G | A | SNP | CHD6 | ENST00000440697 | -1 | missense | c.782 | p.T261M | tier1 | 45 | 0 | 0.000 | 187 | 80 | 0.300 |
| 147457 | 20 | 40122259 | 40122259 | C | T | SNP | CHD6 | ENST00000440697 | -1 | missense | c.341 | p.R114H | tier1 | 74 | 0 | 0.000 | 118 | 46 | 0.281 |
| 147457 | 20 | 44050134 | 44050134 | C | T | SNP | PIGT | ENST00000279036 | 1 | missense | c.1145 | p.P382L | tier1 | 22 | 0 | 0.000 | 100 | 44 | 0.306 |
| 147457 | 20 | 49620923 | 49620923 | C | T | SNP | KCNG1 | ENST00000371571 | -1 | missense | c.1195 | p.E399K | tier1 | 30 | 0 | 0.000 | 189 | 72 | 0.275 |
| 147457 | 20 | 53208320 | 53208320 | C | T | SNP | DOK5 | ENST00000262593 | 1 | missense | c.575 | p.T192M | tier1 | 154 | 2 | 0.013 | 245 | 133 | 0.352 |
| 147457 | 20 | 60704165 | 60704165 | G | A | SNP | LSM14B | ENST00000253001 | 1 | intronic | c.661-1175 | e5-1175 | tier3 | 41 | 0 | 0.000 | 141 | 76 | 0.347 |
| 147457 | 20 | 60910153 | 60910153 | G | A | SNP | LAMA5 | ENST00000252999 | -1 | silent | c.2406 | p.H802 | tier1 | 25 | 0 | 0.000 | 303 | 132 | 0.303 |
| 147457 | 20 | 62037970 | 62037970 | G | A | SNP | KCNQ2 | ENST00000354587 | -1 | 3_prime_untranslated_region | c.*27 | NULL | tier2 | 35 | 0 | 0.000 | 177 | 59 | 0.250 |
| 147457 | 20 | 62055538 | 62055538 | C | T | SNP | KCNQ2 | ENST00000354587 | -1 | silent | c.1209 | p.P403 | tier1 | 82 | 2 | 0.024 | 387 | 162 | 0.295 |
| 147457 | 21 | 31744090 | 31744090 | G | A | SNP | KRTAP13-2 | ENST00000399889 | -1 | missense | c.442 | p.R148C | tier1 | 52 | 1 | 0.019 | 156 | 78 | 0.333 |
| 147457 | 21 | 33318470 | 33318470 | G | A | SNP | HUNK | ENST00000270112 | 1 | missense | c.733 | p.D245N | tier1 | 54 | 0 | 0.000 | 136 | 58 | 0.297 |
| 147457 | 21 | 33696887 | 33696887 | C | T | SNP | URB1 | ENST00000382751 | -1 | missense | c.5147 | p.R1716Q | tier1 | 104 | 1 | 0.010 | 249 | 111 | 0.308 |
| 147457 | 21 | 34945659 | 34945659 | G | A | SNP | SON | ENST00000356577 | 1 | missense | c.6931 | p.V2311I | tier1 | 160 | 2 | 0.012 | 270 | 87 | 0.243 |
| 147457 | 21 | 36206893 | 36206893 | G | A | SNP | RUNX1 | ENST00000300305 | -1 | missense | c.619 | p.R207W | tier1 | 134 | 1 | 0.007 | 755 | 326 | 0.301 |
| 147457 | 21 | 43547924 | 43547924 | G | A | SNP | UMODL1 | ENST00000408989 | 1 | missense | c.4057 | p.G1353R | tier1 | 113 | 0 | 0.000 | 200 | 91 | 0.313 |
| 147457 | 21 | 43984766 | 43984766 | G | A | SNP | SLC37A1 | ENST00000352133 | 1 | intronic | c.1136-52 | e13-52 | tier3 | 16 | 0 | 0.000 | 168 | 75 | 0.309 |
| 147457 | 21 | 46363698 | 46363698 | G | A | SNP | FAM207A | ENST00000291634 | 1 | missense | c.229 | p.V77I | tier1 | 34 | 0 | 0.000 | 160 | 74 | 0.315 |
| 147457 | 21 | 47766132 | 47766132 | C | T | SNP | PCNT | ENST00000359568 | 1 | splice_region | c.720+10 | e4+10 | tier3 | 43 | 1 | 0.023 | 149 | 59 | 0.282 |
| 147457 | 22 | 21022153 | 21022153 | G | A | SNP | FAM108A5P | ENST00000411545 | -1 | 5_prime_untranslated_region | c.-21022153 | NULL | tier2 | 34 | 0 | 0.000 | 424 | 172 | 0.289 |
| 147457 | 22 | 21349265 | 21349265 | G | A | SNP | LZTR1 | ENST00000215739 | 1 | missense | c.1892 | p.R631Q | tier1 | 37 | 0 | 0.000 | 264 | 118 | 0.308 |
| 147457 | 22 | 23077532 | 23077532 | C | T | SNP | IGLV2-18 | ENST00000390310 | 1 | silent | c.309 | p.D103 | tier1 | 37 | 0 | 0.000 | 233 | 89 | 0.276 |
| 147457 | 22 | 24649098 | 24649098 | G | A | SNP | POM121L9P | ENST00000414583 | 1 | 3_prime_untranslated_region | c.*24649098 | NULL | tier3 | 88 | 0 | 0.000 | 551 | 256 | 0.317 |
| 147457 | 22 | 25598652 | 25598652 | C | T | SNP | CRYBB3 | ENST00000215855 | 1 | silent | c.87 | p.Y29 | tier1 | 49 | 0 | 0.000 | 172 | 54 | 0.239 |
| 147457 | 22 | 37319436 | 37319436 | C | T | SNP | CSF2RB | ENST00000262825 | 1 | intronic | c.200+27 | e2+27 | tier3 | 17 | 0 | 0.000 | 134 | 89 | 0.399 |
| 147457 | 22 | 37892522 | 37892522 | C | T | SNP | CARD10 | ENST00000251973 | -1 | missense | c.1993 | p.E665K | tier1 | 15 | 0 | 0.000 | 114 | 62 | 0.352 |
| 147457 | 22 | 38062929 | 38062929 | G | A | SNP | PDXP | ENST00000215904 | 1 | 3_prime_untranslated_region | c.*1051 | NULL | tier2 | 97 | 3 | 0.030 | 148 | 61 | 0.292 |
| 147457 | 22 | 39358204 | 39358204 | C | T | SNP | APOBEC3A | ENST00000249116 | 1 | missense | c.571 | p.R191W | tier1 | 34 | 0 | 0.000 | 178 | 80 | 0.309 |
| 147457 | 22 | 46125385 | 46125385 | A | A | SNP | ATXN10 | ENST00000252934 | 1 | missense | c.809 | p.R270Q | tier1 | 67 | 2 | 0.029 | 152 | 62 | 0.290 |
| 147457 | 22 | 50728653 | 50728653 | C | T | SNP | PLXNB2 | ENST00000359337 | -1 | missense | c.361 | p.A121T | tier1 | 22 | 0 | 0.000 | 186 | 83 | 0.309 |
| 147457 | 22 | 50900128 | 50900128 | C | T | SNP | SBF1 | ENST00000337034 | -1 | missense | c.2693 | p.R898H | tier1 | 50 | 0 | 0.000 | 291 | 131 | 0.309 |
| 147457 | 22 | 50962222 | 50962222 | C | T | SNP | SCO2 | ENST00000252785 | -1 | missense | c.619 | p.V207M | tier1 | 48 | 2 | 0.040 | 206 | 80 | 0.280 |
| 147457 | 22 | 51182575 | 51182575 | C | T | SNP | ACR | ENST00000216139 | 1 | missense | c.652 | p.R218C | tier1 | 59 | 2 | 0.033 | 273 | 109 | 0.285 |
| 147457 | X | 2406917 | 2406917 | C | T | SNP | ZBED1 | ENST00000381218 | -1 | missense | c.1844 | p.R615H | tier1 | 41 | 0 | 0.000 | 282 | 118 | 0.293 |
| 147457 | X | 13765025 | 13765025 | G | A | SNP | OFD1 | ENST00000340096 | 1 | missense | c.781 | p.V261I | tier1 | 293 | 4 | 0.014 | 415 | 190 | 0.312 |
| 147457 | X | 20070889 | 20070889 | C | T | SNP | MAP7D2 | ENST00000379643 | -1 | intronic | c.595+107 | e5+107 | tier3 | 67 | 0 | 0.000 | 144 | 58 | 0.283 |
| 147457 | X | 29973688 | 29973688 | G | A | SNP | IL1RAPL1 | ENST00000302196 | 1 | silent | c.1842 | p.T614 | tier1 | 98 | 2 | 0.020 | 185 | 80 | 0.302 |
| 147457 | X | 29974566 | 29974566 | G | A | SNP | IL1RAPL1 | ENST00000378993 | 1 | 3_prime_untranslated_region | c.*629 | NULL | tier3 | 154 | 1 | 0.007 | 371 | 139 | 0.272 |
| 147457 | X | 34961163 | 34961163 | G | A | SNP | FAM47B | ENST00000329357 | 1 | missense | c.215 | p.R72H | tier1 | 183 | 4 | 0.021 | 504 | 240 | 0.323 |
| 147457 | X | 41530801 | 41530801 | C | T | SNP | CASK | ENST00000378163 | -1 | intronic | c.430-18 | e6-18 | tier4 | 231 | 4 | 0.017 | 520 | 212 | 0.289 |
| 147457 | X | 49032048 | 49032048 | G | A | SNP | PRICKLE3 | ENST00000376317 | -1 | nonsense | c.1822 | p.R608* | tier1 | 47 | 2 | 0.041 | 326 | 129 | 0.283 |
| 147457 | X | 51639618 | 51639618 | C | T | SNP | MAGED1 | ENST00000375695 | 1 | silent | c.1035 | p.G345 | tier1 | 35 | 0 | 0.000 | 340 | 138 | 0.288 |
| 147457 | X | 69595890 | 69595890 | C | T | SNP | KIF4A | ENST00000374403 | 1 | intronic | c.1924-60 | e17-60 | tier3 | 103 | 0 | 0.000 | 153 | 47 | 0.233 |
| 147457 | X | 70823895 | 70823895 | C | T | SNP | ACRC | ENST00000373695 | 1 | silent | c.768 | p.D256Q | tier1 | 121 | 1 | 0.008 | 363 | 118 | 0.244 |
| 147457 | X | 70836264 | 70836264 | C | T | SNP | XCRCR3 | ENST00000373691 | -1 | missense | c.1199 | p.R400Q | tier1 | 30 | 1 | 0.032 | 278 | 116 | 0.292 |
| 147457 | X | 71427464 | 71427464 | C | A | SNP | ERCC6L | ENST00000334463 | -1 | missense | c.1153 | p.V385L | tier1 | 85 | 1 | 0.012 | 148 | 54 | 0.267 |
| 147457 | X | 79281277 | 79281277 | G | A | SNP | TBX22 | ENST00000373294 | 1 | splice_site | c.633+1 | e4+1 | tier1 | 54 | 0 | 0.000 | 218 | 87 | 0.284 |
| 147457 | X | 84525966 | 84525966 | C | T | SNP | ZNF711 | ENST00000395402 | 1 | missense | c.1442 | p.P481L | tier1 | 119 | 1 | 0.008 | 236 | 105 | 0.308 |
| 147457 | X | 103316458 | 103316458 | C | T | SNP | H2BFWT | ENST00000217926 | -1 | 5_prime_flanking_region | c.-48226 | NULL | tier3 | 288 | 4 | 0.014 | 491 | 187 | 0.275 |
| 147457 | X | 105144665 | 105144665 | C | T | SNP | NRK | ENST00000428173 | 1 | missense | c.766 | p.P256S | tier1 | 123 | 0 | 0.000 | 125 | 52 | 0.294 |
| 147457 | X | 120181966 | 120181966 | G | A | SNP | GLUD2 | ENST00000328078 | 1 | missense | c.428 | p.R143H | tier1 | 168 | 2 | 0.012 | 398 | 194 | 0.327 |
| 147457 | X | 123215311 | 123215311 | C | T | SNP | STAG2 | ENST00000218089 | 1 | nonsense | c.2857 | p.R953* | tier1 | 402 | 0 | 0.000 | 644 | 273 | 0.297 |
| 147457 | X | 128929233 | 128929233 | G | A | SNP | ENSG00000240143 | ENST00000432513 | 1 | intronic | c.NULL | NULL | tier3 | 85 | 0 | 0.000 | 142 | 57 | 0.286 |
| 147457 | X | 129190113 | 129190113 | C | T | SNP | BCORL1 | ENST00000303743 | 1 | 3_prime_untranslated_region | c.*2 | NULL | tier3 | 40 | 0 | 0.000 | 142 | 42 | 0.228 |
| 147457 | X | 129318354 | 129318354 | C | T | SNP | RAB33A | ENST00000257017 | 1 | silent | c.354 | p.D118 | tier1 | 58 | 0 | 0.000 | 193 | 84 | 0.303 |
| 147457 | X | 129658059 | 129658059 | G | A | SNP | ENSG00000229702 | ENST00000458525 | -1 | 5_prime_untranslated_region | c.-129658059 | NULL | tier2 | 41 | 0 | 0.000 | 208 | 97 | 0.317 |
| 147457 | X | 135961560 | 135961560 | C | G | SNP | RBMX | ENST00000320676 | -1 | missense | c.27 | p.K9N | tier1 | 101 | 0 | 0.000 | 147 | 13 | 0.081 |
| 147457 | X | 138678815 | 138678815 | G | A | SNP | MCF2 | ENST00000370578 | -1 | nonsense | c.2605 | p.R869* | tier1 | 239 | 1 | 0.004 | 280 | 112 | 0.285 |
| 147457 | X | 152808398 | 152808398 | G | A | SNP | ATP2B3 | ENST00000263519 | 1 | intronic | c.791-103 | e5-103 | tier3 | 47 | 1 | 0.021 | 172 | 70 | 0.288 |
| 147457 | X | 153660704 | 153660704 | C | T | SNP | ATP6AP1 | ENST00000369762 | 1 | silent | c.456 | p.L152 | tier1 | 31 | 0 | 0.000 | 221 | 109 | 0.329 |
| 147457 | X | 153696165 | 153696165 | G | A | SNP | PLXNA3 | ENST00000369682 | 1 | missense | c.3641 | p.R1214Q | tier1 | 19 | 0 | 0.000 | 120 | 56 | 0.316 |
| 169510 | 1 | 115258745 | 115258745 | C | G | SNP | NRAS | ENST00000369535 | -1 | missense | c.37 | p.G13R | tier1 | 447 | 15 | 0.033 | 489 | 323 | 0.397 |
| 169510 | 1 | 197168802 | 197168802 | C | T | SNP | ZBTB41 | ENST00000367405 | -1 | missense | c.802 | p.D268N | tier1 | 51 | 3 | 0.056 | 40 | 26 | 0.394 |
| 169510 | 2 | 39053173 | 39053173 | C | T | SNP | DHX57 | ENST00000295373 | -1 | missense | c.2830 | p.G944R | tier1 | 52 | 1 | 0.019 | 68 | 46 | 0.404 |
| 169510 | 2 | 70458050 | 70458051 | - | A | INS | TIA1 | ENST00000433529 | -1 | intronic | c.124-65_124-64 | e3-64 | tier3 | 102 | 0 | 0.000 | 172 | 15 | 0.080 |
| 169510 | 2 | 73316541 | 73316541 | G | A | SNP | RAB11FIP5 | ENST00000493523 | -1 | rna | NULL | NULL | tier1 | 60 | 5 | 0.077 | 54 | 42 | 0.438 |
| 169510 | 3 | 184922434 | 184922434 | C | G | SNP | EHHADH | ENST00000231887 | -1 | missense | c.680 | p.C227S | tier1 | 57 | 3 | 0.050 | 68 | 42 | 0.382 |
| 169510 | 3 | 192874515 | 192874515 | G | A | SNP | VEZF1P1 | ENST00000423931 | -1 | 5_prime_untranslated_region | c.-192874515 | NULL | tier2 | 124 | 9 | 0.068 | 171 | 131 | 0.434 |
| 169510 | 4 | 96044976 | 96044976 | C | G | SNP | BMPR1B | ENST00000440890 | 1 | missense | c.455 | p.P152R | tier1 | 109 | 3 | 0.027 | 63 | 45 | 0.417 |
| 169510 | 4 | 170483082 | 170483082 | C | A | SNP | NEK1 | ENST00000 | | | | | | | | | | | |

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|--------|----|-----------|-----------|---|-------|-----|--------------------|-----------------|----|-----------------------------|-------------|----------|-------|-----|----|-------|-----|-----|-------|
| 169510 | 8 | 9634628 | 9634628 | A | G | SNP | <i>TNKS</i> | ENST00000310430 | 1 | 3_prime_untranslated_region | c.*382 | NULL | tier3 | 90 | 6 | 0.062 | 73 | 60 | 0.451 |
| 169510 | 8 | 17206503 | 17206503 | G | A | SNP | <i>MTMR7</i> | ENST00000180173 | -1 | missense | c.556 | p.R186W | tier1 | 112 | 5 | 0.043 | 111 | 74 | 0.400 |
| 169510 | 9 | 8485187 | 8485187 | C | T | SNP | <i>PTPRD</i> | ENST00000356435 | -1 | intronic | c.3153+40 | | tier3 | 67 | 2 | 0.029 | 57 | 51 | 0.468 |
| 169510 | 9 | 99691238 | 99691238 | G | C | SNP | <i>NUTM2G</i> | ENST00000354649 | 1 | 5_prime_untranslated_region | c.-69 | NULL | tier3 | 90 | 0 | 0.000 | 185 | 41 | 0.181 |
| 169510 | 9 | 139848309 | 139848309 | C | G | SNP | <i>LCN12</i> | ENST00000371633 | -1 | missense | c.341 | p.A114G | tier1 | 82 | 5 | 0.056 | 139 | 91 | 0.396 |
| 169510 | 10 | 42832502 | 42832503 | - | TATCG | INS | <i>LOC441666</i> | ENST00000609841 | -1 | rna | NULL | NULL | tier1 | 264 | 1 | 0.004 | 263 | 14 | 0.051 |
| 169510 | 10 | 42832522 | 42832522 | C | A | SNP | <i>LOC441666</i> | ENST00000609841 | -1 | rna | NULL | NULL | tier1 | 258 | 2 | 0.008 | 261 | 15 | 0.053 |
| 169510 | 10 | 102703909 | 102703909 | T | G | SNP | <i>FAM178A</i> | ENST00000370269 | 1 | intronic | c.2757+24 | e12+24 | tier3 | 57 | 1 | 0.017 | 44 | 35 | 0.443 |
| 169510 | 11 | 1085942 | 1085942 | G | A | SNP | <i>MUC2</i> | ENST00000441003 | 1 | missense | c.2782 | p.E928K | tier1 | 45 | 4 | 0.082 | 41 | 36 | 0.468 |
| 169510 | 11 | 6191332 | 6191332 | C | T | SNP | <i>OR52B2</i> | ENST00000530810 | -1 | silent | c.225 | p.L75 | tier1 | 91 | 7 | 0.071 | 97 | 69 | 0.416 |
| 169510 | 12 | 149069 | 149069 | G | A | SNP | <i>FAM138D</i> | ENST00000320165 | -1 | rna | NULL | NULL | tier1 | 187 | 9 | 0.046 | 312 | 172 | 0.355 |
| 169510 | 12 | 22625197 | 22625197 | A | T | SNP | <i>C2CD5</i> | ENST00000333957 | -1 | intronic | c.2169+23 | e17+23 | tier3 | 211 | 9 | 0.041 | 182 | 123 | 0.402 |
| 169510 | 12 | 58215300 | 58215300 | C | T | SNP | <i>CTDSP2</i> | ENST00000398073 | -1 | 3_prime_untranslated_region | c.*2085 | NULL | tier3 | 27 | 1 | 0.036 | 38 | 27 | 0.403 |
| 169510 | 12 | 81655879 | 81655879 | G | C | SNP | <i>PPFIA2</i> | ENST00000549396 | -1 | intronic | c.3716-60 | e30-60 | tier3 | 54 | 2 | 0.036 | 54 | 36 | 0.400 |
| 169510 | 12 | 110783211 | 110783211 | T | A | SNP | <i>ATP2A2</i> | ENST00000539276 | 1 | intronic | c.2741+24 | e18+24 | tier3 | 44 | 2 | 0.044 | 69 | 47 | 0.405 |
| 169510 | 14 | 21216226 | 21216226 | C | G | SNP | <i>EDDM3A</i> | ENST00000326842 | 1 | 3_prime_untranslated_region | c.*43 | NULL | tier3 | 44 | 0 | 0.000 | 89 | 39 | 0.305 |
| 169510 | 15 | 85383164 | 85383164 | C | T | SNP | <i>ALPK3</i> | ENST00000258888 | 1 | silent | c.1260 | p.P420 | tier1 | 389 | 36 | 0.084 | 508 | 424 | 0.453 |
| 169510 | 16 | 32070612 | 32070612 | A | C | SNP | <i>IGHV1OR16-3</i> | ENST00000568138 | 1 | 3_prime_untranslated_region | c.*32070612 | NULL | tier2 | 213 | 1 | 0.005 | 355 | 18 | 0.048 |
| 169510 | 16 | 57250830 | 57250830 | A | T | SNP | <i>RSPRY1</i> | ENST00000394420 | 1 | missense | c.784 | p.T262S | tier1 | 134 | 5 | 0.036 | 110 | 105 | 0.486 |
| 169510 | 16 | 89597025 | 89597025 | C | T | SNP | <i>SPG7</i> | ENST00000268704 | 1 | intronic | c.862-66 | e7-66 | tier3 | 42 | 1 | 0.023 | 52 | 38 | 0.422 |
| 169510 | 17 | 7414546 | 7414546 | G | C | SNP | <i>POLR2A</i> | ENST00000322644 | 1 | missense | c.3826 | p.V1276L | tier1 | 59 | 3 | 0.048 | 60 | 58 | 0.492 |
| 169510 | 17 | 7832533 | 7832533 | C | T | SNP | <i>KCNAB3</i> | ENST00000303790 | -1 | missense | c.221 | p.R74Q | tier1 | 77 | 4 | 0.049 | 73 | 54 | 0.252 |
| 169510 | 17 | 10352234 | 10352234 | G | A | SNP | <i>MYH4</i> | ENST00000255381 | -1 | nonsense | c.4312 | p.R1438* | tier1 | 667 | 29 | 0.042 | 704 | 508 | 0.419 |
| 169510 | 17 | 49835349 | 49835349 | C | T | SNP | <i>CA10</i> | ENST00000340813 | -1 | intronic | c.298-10171 | e4-10171 | tier3 | 96 | 2 | 0.020 | 106 | 70 | 0.398 |
| 169510 | 17 | 73997619 | 73997619 | A | G | SNP | <i>CDK3</i> | ENST00000425876 | 1 | missense | c.113 | p.D38G | tier1 | 74 | 3 | 0.039 | 82 | 55 | 0.402 |
| 169510 | 17 | 74733199 | 74733199 | G | T | SNP | <i>SRSF2</i> | ENST00000359995 | -1 | missense | c.44 | p.S15Y | tier1 | 254 | 23 | 0.083 | 327 | 310 | 0.486 |
| 169510 | 17 | 74733200 | 74733200 | A | C | SNP | <i>SRSF2</i> | ENST00000359995 | -1 | missense | c.43 | p.S15A | tier1 | 258 | 21 | 0.075 | 328 | 310 | 0.486 |
| 169510 | 19 | 15996791 | 15996791 | C | T | SNP | <i>CYP4F2</i> | ENST00000221700 | -1 | missense | c.1058 | p.R353H | tier1 | 133 | 9 | 0.063 | 127 | 96 | 0.431 |
| 169510 | 19 | 50753076 | 50753076 | G | A | SNP | <i>MYH14</i> | ENST00000262269 | 1 | missense | c.1652 | p.R551Q | tier1 | 80 | 6 | 0.070 | 95 | 76 | 0.442 |
| 169510 | 20 | 48600334 | 48600334 | C | T | SNP | <i>SNAI1</i> | ENST00000244050 | 1 | intronic | c.83-27 | e2-27 | tier2 | 40 | 0 | 0.000 | 110 | 19 | 0.147 |
| 169510 | 20 | 49575622 | 49575622 | G | T | SNP | <i>MOCS3</i> | ENST00000244051 | 1 | silent | c.243 | p.G81 | tier1 | 18 | 1 | 0.053 | 19 | 19 | 0.500 |
| 169510 | 21 | 36231782 | 36231782 | C | T | SNP | <i>RUNX1</i> | ENST00000300305 | -1 | missense | c.602 | p.R201Q | tier1 | 719 | 55 | 0.071 | 843 | 712 | 0.457 |
| 169510 | X | 123176411 | 123176411 | A | G | SNP | <i>STAG2</i> | ENST00000218089 | 1 | splice_region | c.386-8 | e5-8 | tier3 | 275 | 37 | 0.119 | 24 | 240 | 0.899 |
| 185295 | 1 | 12032977 | 12032977 | C | T | SNP | <i>PLOD1</i> | ENST00000376369 | 1 | missense | c.2092 | p.P698S | tier1 | 113 | 0 | 0.000 | 115 | 49 | 0.299 |
| 185295 | 1 | 167341159 | 167341159 | A | T | SNP | <i>POU2F1</i> | ENST00000367866 | 1 | missense | c.288 | p.K96N | tier1 | 105 | 0 | 0.000 | 86 | 28 | 0.246 |
| 185295 | 1 | 176675464 | 176675464 | G | A | SNP | <i>PAPPA2</i> | ENST00000367662 | 1 | intronic | c.3366-31 | e9-31 | tier3 | 106 | 2 | 0.019 | 72 | 36 | 0.333 |
| 185295 | 1 | 220830533 | 220830533 | C | T | SNP | <i>MARK1</i> | ENST00000366917 | 1 | intronic | c.1989-599 | e17-599 | tier3 | 239 | 0 | 0.000 | 185 | 75 | 0.289 |
| 185295 | 1 | 244218466 | 244218466 | C | A | SNP | <i>ZBTB18</i> | ENST00000358704 | 1 | missense | c.1390 | p.R464S | tier1 | 33 | 0 | 0.000 | 34 | 15 | 0.306 |
| 185295 | 2 | 56611386 | 56611386 | C | T | SNP | <i>CCDC85A</i> | ENST00000407595 | 1 | intronic | c.1573-15 | e6-15 | tier3 | 149 | 1 | 0.007 | 100 | 9 | 0.083 |
| 185295 | 4 | 20529829 | 20529829 | G | A | SNP | <i>SLIT2</i> | ENST00000504154 | 1 | intronic | c.1463-743 | e16-743 | tier3 | 96 | 0 | 0.000 | 76 | 25 | 0.248 |
| 185295 | 4 | 69344614 | 69344614 | G | A | SNP | <i>TMPPRS11E</i> | ENST00000305363 | 1 | missense | c.1015 | p.A339T | tier1 | 95 | 0 | 0.000 | 79 | 25 | 0.238 |
| 185295 | 4 | 159618713 | 159618713 | A | T | SNP | <i>ETFDH</i> | ENST00000511912 | 1 | missense | c.834 | p.L278F | tier1 | 37 | 1 | 0.026 | 45 | 16 | 0.262 |
| 185295 | 4 | 169195149 | 169195149 | T | C | SNP | <i>DDX60</i> | ENST00000393743 | -1 | missense | c.2390 | p.Y797C | tier1 | 99 | 0 | 0.000 | 68 | 18 | 0.209 |
| 185295 | 6 | 5144542 | 5144542 | T | C | SNP | <i>LYRM4</i> | ENST00000330636 | -1 | intronic | c.208-34817 | e3-34817 | tier3 | 45 | 0 | 0.000 | 29 | 15 | 0.341 |
| 185295 | 6 | 13365713 | 13365713 | C | T | SNP | <i>GFOD1</i> | ENST00000379287 | -1 | silent | c.435 | p.V145 | tier1 | 53 | 0 | 0.000 | 41 | 16 | 0.281 |
| 185295 | 6 | 157150512 | 157150512 | G | A | SNP | <i>ARID1B</i> | ENST00000367148 | 1 | missense | c.1694 | p.R565Q | tier1 | 105 | 0 | 0.000 | 75 | 21 | 0.219 |
| 185295 | 7 | 124404234 | 124404234 | C | T | SNP | <i>GPR37</i> | ENST00000303921 | -1 | missense | c.797 | p.C266Y | tier1 | 77 | 0 | 0.000 | 85 | 34 | 0.286 |
| 185295 | 7 | 146805221 | 146805221 | T | - | DEL | <i>CNTNAP2</i> | ENST00000361727 | 1 | intronic | c.551-18 | e5-18 | tier3 | 72 | 2 | 0.027 | 73 | 21 | 0.223 |
| 185295 | 8 | 70533457 | 70533457 | G | A | SNP | <i>SULF1</i> | ENST00000260128 | 1 | missense | c.1565 | p.R522Q | tier1 | 71 | 0 | 0.000 | 66 | 35 | 0.347 |
| 185295 | 8 | 145737271 | 145737271 | C | G | SNP | <i>RECQL4</i> | ENST00000428558 | -1 | intronic | c.3393+23 | e20+23 | tier3 | 50 | 0 | 0.000 | 39 | 28 | 0.418 |
| 185295 | 9 | 97380079 | 97380079 | C | T | SNP | <i>FBP1</i> | ENST00000375326 | -1 | missense | c.397 | p.V133I | tier1 | 121 | 1 | 0.008 | 94 | 54 | 0.365 |
| 185295 | 10 | 75510973 | 75510973 | G | - | DEL | <i>SEC24C</i> | ENST00000339365 | 1 | frame_shift_del | c.280 | p.P95fs | tier1 | 53 | 0 | 0.000 | 61 | 13 | 0.176 |
| 185295 | 10 | 88414661 | 88414661 | C | T | SNP | <i>OPN4</i> | ENST00000372071 | 1 | missense | c.121 | p.R41W | tier1 | 79 | 0 | 0.000 | 93 | 40 | 0.301 |
| 185295 | 11 | 17663639 | 17663639 | C | A | SNP | <i>OTOG</i> | ENST00000399391 | 1 | intronic | c.8332-35 | e52-35 | tier3 | 59 | 0 | 0.000 | 64 | 36 | 0.360 |
| 185295 | 12 | 49719635 | 49719635 | C | T | SNP | <i>TROAP</i> | ENST00000257909 | 1 | missense | c.601 | p.R201W | tier1 | 65 | 0 | 0.000 | 81 | 25 | 0.236 |
| 185295 | 12 | 65272304 | 65272304 | C | T | SNP | <i>TBC1D30</i> | ENST00000542120 | 1 | 3_prime_untranslated_region | c.*2736 | NULL | tier3 | 75 | 0 | 0.000 | 86 | 41 | 0.323 |
| 185295 | 12 | 124427322 | 124427322 | A | C | SNP | <i>CCDC92</i> | ENST00000238156 | -1 | nonsense | c.192 | p.Y64* | tier1 | 119 | 0 | 0.000 | 76 | 46 | 0.371 |
| 185295 | 13 | 48547335 | 48547335 | G | A | SNP | <i>SUCLA2</i> | ENST00000378654 | -1 | intronic | c.663+64 | e5+64 | tier3 | 96 | 0 | 0.000 | 104 | 37 | 0.262 |
| 185295 | 13 | 52684711 | 52684711 | G | T | SNP | <i>NEK5</i> | ENST00000355568 | -1 | silent | c.315 | p.I105 | tier1 | 84 | 0 | 0.000 | 84 | 43 | 0.339 |
| 185295 | 14 | 96552991 | 96552991 | C | T | SNP | <i>C14orf132</i> | ENST00000553764 | 1 | rna | NULL | NULL | tier1 | 46 | 0 | 0.000 | 42 | 22 | 0.344 |
| 185295 | 15 | 50926721 | 50926721 | T | C | SNP | <i>TRPM7</i> | ENST00000313478 | -1 | missense | c.865 | p.I289V | tier1 | 157 | 0 | 0.000 | 174 | 83 | 0.323 |
| 185295 | 15 | 78207447 | 78207447 | T | A | SNP | <i>LOC645752</i> | ENST00000563349 | -1 | rna | NULL | NULL | tier1 | 159 | 0 | 0.000 | 127 | 60 | 0.321 |
| 185295 | 15 | 85343270 | 85343270 | C | G | SNP | <i>ZNF592</i> | ENST00000299927 | 1 | intronic | c.3273+62 | e7+62 | tier3 | 42 | 0 | 0.000 | 42 | 26 | 0.382 |
| 185295 | 17 | 10545798 | 10545798 | C | T | SNP | <i>MYH3</i> | ENST00000226209 | -1 | silent | c.1824 | p.G608 | tier1 | 204 | 1 | 0.005 | 199 | 52 | 0.207 |
| 185295 | 17 | 43008929 | 43008929 | C | T | SNP | <i>KIF18B</i> | ENST00000339151 | -1 | intronic | c.1553+41 | e10+41 | tier3 | 38 | 0 | 0.000 | 19 | 12 | 0.387 |
| 185295 | 17 | 62658267 | 62658267 | C | A | SNP | <i>SMURF2</i> | ENST00000262435 | -1 | 5_prime_flanking_region | c.-269 | NULL | tier4 | 6 | 0 | 0.000 | 1 | 5 | 0.833 |
| 185295 | 18 | 60572437 | 60572437 | G | T | SNP | <i>PHLPP1</i> | ENST00000262719 | 1 | intronic | c.2648-20 | e8-20 | tier3 | 135 | 0 | 0.000 | 119 | 9 | 0.070 |
| 185295 | 21 | 44524456 | 4 | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|-----|---|-----|-----------------|-----------------|----|-----------------------------|--------------|-----------|-------|-----|---|-------|------|-----|-------|
| 187374 | 1 | 44467034 | 44467034 | C | G | SNP | <i>SLC6A9</i> | ENST00000360584 | -1 | intronic | c.1419+28 | e9+28 | tier3 | 85 | 4 | 0.045 | 63 | 43 | 0.406 |
| 187374 | 1 | 47512254 | 47512254 | G | A | SNP | <i>CYP4X1</i> | ENST00000371901 | 1 | missense | c.1189 | p.G397R | tier1 | 65 | 0 | 0.000 | 73 | 43 | 0.371 |
| 187374 | 1 | 176564033 | 176564033 | C | T | SNP | <i>PAPPA2</i> | ENST00000367662 | 1 | silent | c.1293 | p.T431 | tier1 | 79 | 1 | 0.013 | 59 | 27 | 0.314 |
| 187374 | 1 | 179526224 | 179526224 | G | A | SNP | <i>NPHS2</i> | ENST00000367615 | -1 | silent | c.676 | p.L226 | tier1 | 120 | 2 | 0.016 | 127 | 70 | 0.355 |
| 187374 | 2 | 25463295 | 25463295 | T | A | SNP | <i>DNMT3A</i> | ENST00000264709 | -1 | missense | c.2198 | p.E733V | tier1 | 514 | 3 | 0.006 | 499 | 303 | 0.376 |
| 187374 | 2 | 37458814 | 37458814 | C | A | SNP | <i>CEBPZ</i> | ENST00000234170 | -1 | 5_prime_untranslated_region | c.-104 | NULL | tier2 | 73 | 0 | 0.000 | 90 | 42 | 0.316 |
| 187374 | 2 | 42280542 | 42280542 | G | A | SNP | <i>PKDCC</i> | ENST00000485578 | 1 | 3_prime_untranslated_region | c.*42280542 | NULL | tier3 | 39 | 0 | 0.000 | 37 | 25 | 0.403 |
| 187374 | 2 | 196673523 | 196673523 | C | A | SNP | <i>DNAH7</i> | ENST00000312428 | -1 | missense | c.9966 | p.W3322C | tier1 | 92 | 0 | 0.000 | 116 | 48 | 0.293 |
| 187374 | 2 | 207570559 | 207570559 | G | A | SNP | <i>DYTN</i> | ENST00000452335 | -1 | missense | c.335 | p.A112V | tier1 | 106 | 2 | 0.018 | 111 | 61 | 0.355 |
| 187374 | 3 | 49700929 | 49700929 | C | - | DEL | <i>BSN</i> | ENST00000296452 | 1 | frame_shift_del | c.11338 | p.Q3780fs | tier1 | 829 | 4 | 0.005 | 651 | 329 | 0.336 |
| 187374 | 3 | 133667500 | 133667500 | C | T | SNP | <i>SLCO2A1</i> | ENST00000310926 | -1 | missense | c.985 | p.V329I | tier1 | 60 | 1 | 0.016 | 52 | 20 | 0.278 |
| 187374 | 4 | 89352470 | 89352470 | T | - | DEL | <i>HERC6</i> | ENST00000264346 | 1 | intronic | c.2250+13 | e17+13 | tier3 | 69 | 1 | 0.014 | 85 | 15 | 0.150 |
| 187374 | 6 | 49676860 | 49676860 | A | G | SNP | <i>CRISP2</i> | ENST00000339139 | -1 | missense | c.50 | p.L17S | tier1 | 102 | 0 | 0.000 | 76 | 56 | 0.421 |
| 187374 | 7 | 139083345 | 139083345 | A | T | SNP | <i>LUC7L2</i> | ENST00000541515 | 1 | nonsense | c.355 | p.R119* | tier1 | 394 | 3 | 0.008 | 251 | 149 | 0.373 |
| 187374 | 9 | 101751401 | 101751401 | G | T | SNP | <i>COL15A1</i> | ENST00000375001 | 1 | intronic | c.724-59 | e5-59 | tier3 | 63 | 0 | 0.000 | 49 | 23 | 0.319 |
| 187374 | 9 | 131708388 | 131708388 | C | A | SNP | <i>DOLK</i> | ENST00000372586 | -1 | nonsense | c.1195 | p.R399* | tier1 | 37 | 1 | 0.026 | 30 | 23 | 0.434 |
| 187374 | 9 | 132980111 | 132980111 | G | T | SNP | <i>NCS1</i> | ENST00000372398 | 1 | missense | c.90 | p.W30C | tier1 | 58 | 3 | 0.049 | 74 | 34 | 0.315 |
| 187374 | 10 | 25465090 | 25465090 | T | T | SNP | <i>GPR158</i> | ENST00000376351 | 1 | silent | c.741 | p.G247 | tier1 | 78 | 0 | 0.000 | 62 | 37 | 0.374 |
| 187374 | 11 | 36601073 | 36601073 | C | C | SNP | <i>RAG1</i> | ENST00000299440 | 1 | 3_prime_untranslated_region | c.*3087 | NULL | tier2 | 161 | 0 | 0.000 | 75 | 28 | 0.269 |
| 187374 | 11 | 70779207 | 70779207 | C | T | SNP | <i>SHANK2</i> | ENST00000338508 | -1 | intronic | c.1072+71 | e14+71 | tier4 | 27 | 0 | 0.000 | 14 | 20 | 0.588 |
| 187374 | 11 | 93439939 | 93439939 | C | A | SNP | <i>KIAA1731</i> | ENST00000411936 | 1 | missense | c.5625 | p.D1875E | tier1 | 95 | 1 | 0.010 | 102 | 49 | 0.325 |
| 187374 | 11 | 93826848 | 93826848 | T | C | SNP | <i>HEPHL1</i> | ENST00000315765 | 1 | intronic | c.2434+42 | e13+42 | tier2 | 46 | 0 | 0.000 | 27 | 22 | 0.449 |
| 187374 | 12 | 53699798 | 53699798 | T | G | SNP | <i>C12orf10</i> | ENST00000549488 | 1 | missense | c.107 | p.V36G | tier1 | 70 | 1 | 0.014 | 68 | 29 | 0.299 |
| 187374 | 12 | 56811467 | 56811467 | G | A | SNP | <i>TIMELESS</i> | ENST00000557589 | -1 | 5_prime_untranslated_region | c.-56811467 | NULL | tier3 | 45 | 0 | 0.000 | 86 | 37 | 0.301 |
| 187374 | 12 | 106631792 | 106631792 | CAA | - | DEL | <i>CKAP4</i> | ENST00000378026 | -1 | 3_prime_untranslated_region | c.*1010 | NULL | tier4 | 51 | 0 | 0.000 | 53 | 9 | 0.145 |
| 187374 | 13 | 73334857 | 73334857 | C | T | SNP | <i>DIS3</i> | ENST00000377767 | -1 | intronic | c.2671-68 | e20-68 | tier3 | 242 | 1 | 0.004 | 159 | 84 | 0.344 |
| 187374 | 16 | 32187496 | 32187496 | A | - | DEL | <i>HERC2P4</i> | ENST00000433784 | -1 | 5_prime_untranslated_region | c.-32187496 | NULL | tier3 | 17 | 0 | 0.000 | 7 | 8 | 0.533 |
| 187374 | 17 | 8011913 | 8011913 | G | A | SNP | <i>ALOXE3</i> | ENST00000318227 | -1 | splice_region | c.1959-6 | e13-6 | tier3 | 71 | 1 | 0.014 | 64 | 33 | 0.337 |
| 187374 | 17 | 13399643 | 13399643 | C | T | SNP | <i>HS3ST3A1</i> | ENST00000284110 | -1 | silent | c.1092 | p.L364 | tier1 | 88 | 0 | 0.000 | 82 | 46 | 0.359 |
| 187374 | 17 | 21319856 | 21319856 | G | A | SNP | <i>KCNJ12</i> | ENST00000331718 | 1 | missense | c.1202 | p.R401Q | tier1 | 87 | 0 | 0.000 | 94 | 17 | 0.153 |
| 187374 | 17 | 27936061 | 27936061 | C | T | SNP | <i>ANKRD13B</i> | ENST00000394859 | 1 | intronic | c.566-43 | e6-43 | tier3 | 64 | 1 | 0.015 | 46 | 22 | 0.324 |
| 187374 | 17 | 38926332 | 38926332 | C | T | SNP | <i>KRT26</i> | ENST00000335552 | -1 | missense | c.724 | p.V242M | tier1 | 119 | 1 | 0.008 | 121 | 70 | 0.367 |
| 187374 | 17 | 43530828 | 43530828 | C | G | SNP | <i>PLEKHM1</i> | ENST00000430334 | -1 | missense | c.2390 | p.C797S | tier1 | 80 | 0 | 0.000 | 63 | 17 | 0.213 |
| 187374 | 19 | 648180 | 648180 | G | A | SNP | <i>RNF126</i> | ENST00000292363 | -1 | missense | c.884 | p.S295L | tier1 | 81 | 0 | 0.000 | 82 | 57 | 0.407 |
| 187374 | 19 | 17471347 | 17471347 | C | T | SNP | <i>PLVAP</i> | ENST00000252590 | -1 | missense | c.1312 | p.A438T | tier1 | 51 | 0 | 0.000 | 85 | 36 | 0.295 |
| 187374 | 20 | 44037609 | 44037609 | C | T | SNP | <i>DBNDD2</i> | ENST00000372717 | 1 | missense | c.308 | p.A103V | tier1 | 37 | 1 | 0.026 | 50 | 23 | 0.315 |
| 187374 | 21 | 35467626 | 35467626 | G | A | SNP | <i>SLC5A3</i> | ENST00000381151 | 1 | silent | c.129 | p.G43 | tier1 | 112 | 1 | 0.009 | 103 | 49 | 0.322 |
| 187374 | 21 | 43412853 | 43412853 | G | C | SNP | <i>ZBTB21</i> | ENST00000310826 | -1 | missense | c.1352 | p.A451G | tier1 | 27 | 0 | 0.000 | 33 | 19 | 0.365 |
| 187374 | 22 | 20133674 | 20133674 | C | T | SNP | <i>ZDHC8</i> | ENST00000334554 | 1 | 3_prime_untranslated_region | c.*751 | NULL | tier3 | 92 | 1 | 0.011 | 79 | 32 | 0.288 |
| 187374 | X | 110489305 | 110489305 | C | T | SNP | <i>CAPN6</i> | ENST00000324068 | -1 | 3_prime_untranslated_region | c.*500 | NULL | tier3 | 28 | 0 | 0.000 | 18 | 25 | 0.581 |
| 189012 | 1 | 16973159 | 16973159 | A | T | SNP | <i>MST1P2</i> | ENST00000334429 | 1 | 3_prime_untranslated_region | c.*16973159 | NULL | tier3 | 128 | 1 | 0.008 | 265 | 24 | 0.083 |
| 189012 | 2 | 122368824 | 122368824 | T | G | SNP | <i>CLASP1</i> | ENST00000263710 | -1 | intronic | c.-1-5068 | e1-5068 | tier4 | 45 | 0 | 0.000 | 127 | 27 | 0.175 |
| 189012 | 2 | 198266834 | 198266834 | T | C | SNP | <i>SF3B1</i> | ENST00000335508 | -1 | missense | c.2098 | p.K700E | tier1 | 251 | 0 | 0.000 | 598 | 48 | 0.074 |
| 189012 | 2 | 210707164 | 210707164 | C | T | SNP | <i>UNC80</i> | ENST00000439458 | 1 | missense | c.3454 | p.R1152C | tier1 | 51 | 0 | 0.000 | 99 | 20 | 0.168 |
| 189012 | 3 | 172674651 | 172674651 | C | T | SNP | <i>SPATA16</i> | ENST00000351008 | -1 | intronic | c.934-37 | e5-37 | tier3 | 26 | 0 | 0.000 | 68 | 22 | 0.244 |
| 189012 | 4 | 39293321 | 39293321 | T | G | SNP | <i>RFC1</i> | ENST00000381897 | -1 | missense | c.3109 | p.N1037H | tier1 | 79 | 0 | 0.000 | 146 | 27 | 0.156 |
| 189012 | 5 | 79095309 | 79095309 | G | A | SNP | <i>CMYA5</i> | ENST00000446378 | 1 | missense | c.12080 | p.S4027N | tier1 | 397 | 0 | 0.000 | 933 | 79 | 0.078 |
| 189012 | 5 | 131911487 | 131911487 | G | A | SNP | <i>RAD50</i> | ENST00000265335 | 1 | missense | c.232 | p.V78M | tier1 | 118 | 0 | 0.000 | 133 | 26 | 0.164 |
| 189012 | 5 | 170232684 | 170232684 | C | - | DEL | <i>GABRP</i> | ENST00000265294 | 1 | intronic | c.542-36 | e6-36 | tier3 | 65 | 1 | 0.015 | 147 | 41 | 0.218 |
| 189012 | 6 | 56335881 | 56335881 | T | C | SNP | <i>DST</i> | ENST00000370754 | -1 | intronic | c.22228+23 | e95+23 | tier3 | 309 | 6 | 0.019 | 434 | 115 | 0.210 |
| 189012 | 6 | 158994487 | 158994487 | G | A | SNP | <i>TMEM181</i> | ENST00000367090 | 1 | missense | c.455 | p.R152H | tier1 | 35 | 0 | 0.000 | 77 | 16 | 0.172 |
| 189012 | 8 | 140768641 | 140768641 | T | G | SNP | <i>TRAPPC9</i> | ENST00000389328 | -1 | intronic | c.3350-24196 | e22-24196 | tier3 | 50 | 0 | 0.000 | 127 | 18 | 0.122 |
| 189012 | 11 | 47753670 | 47753670 | T | C | SNP | <i>FNBP4</i> | ENST00000528388 | -1 | rna | NULL | NULL | tier1 | 47 | 0 | 0.000 | 60 | 14 | 0.189 |
| 189012 | 11 | 66106152 | 66106152 | C | T | SNP | <i>BRMS1</i> | ENST00000530238 | -1 | 5_prime_untranslated_region | c.-66106152 | NULL | tier3 | 56 | 0 | 0.000 | 131 | 17 | 0.115 |
| 189012 | 11 | 119177652 | 119177652 | A | - | DEL | <i>CBL</i> | ENST00000264033 | 1 | 3_prime_untranslated_region | c.*7161 | NULL | tier2 | 38 | 0 | 0.000 | 67 | 14 | 0.173 |
| 189012 | 12 | 8376028 | 8376028 | C | T | SNP | <i>FAM90A1</i> | ENST00000307435 | -1 | intronic | c.432+17 | e3+17 | tier3 | 691 | 1 | 0.001 | 1696 | 61 | 0.035 |
| 189012 | 14 | 20853200 | 20853200 | T | C | SNP | <i>TEP1</i> | ENST00000262715 | -1 | silent | c.3051 | p.Q1017 | tier1 | 40 | 0 | 0.000 | 72 | 15 | 0.172 |
| 189012 | 18 | 53253420 | 53253420 | C | G | SNP | <i>TCF4</i> | ENST00000398339 | -1 | intronic | c.379-837 | e4-837 | tier4 | 27 | 0 | 0.000 | 74 | 23 | 0.237 |
| 189012 | X | 20083381 | 20083381 | A | T | SNP | <i>MAP7D2</i> | ENST00000379643 | -1 | intronic | c.131-458 | e2-458 | tier4 | 60 | 0 | 0.000 | 117 | 19 | 0.138 |
| 190938 | 1 | 17320299 | 17320299 | A | C | SNP | <i>ATP13A2</i> | ENST00000326735 | -1 | missense | c.1574 | p.V525G | tier1 | 14 | 0 | 0.000 | 20 | 17 | 0.460 |
| 190938 | 1 | 38510268 | 38510268 | T | - | DEL | <i>POU3F1</i> | ENST00000373012 | -1 | 3_prime_untranslated_region | c.*792 | NULL | tier4 | 87 | 2 | 0.023 | 217 | 34 | 0.136 |
| 190938 | 2 | 61411861 | 61411861 | A | G | SNP | <i>AHSA2</i> | ENST00000357022 | 1 | missense | c.1 | p.M1V | tier1 | 50 | 0 | 0.000 | 131 | 70 | 0.348 |
| 190938 | 2 | 97833261 | 97833261 | T | A | SNP | <i>ANKRD36</i> | ENST00000420699 | 1 | intronic | c.1532-53 | e21-53 | tier3 | 75 | 0 | 0.000 | 582 | 66 | 0.102 |
| 190938 | 2 | 130832517 | 130832517 | T | C | SNP | <i>POTEF</i> | ENST00000357462 | -1 | missense | c.2528 | p.Y843C | tier1 | 105 | 0 | 0.000 | 190 | 86 | 0.311 |
| 190938 | 3 | 11406161 | 11406161 | G | T | SNP | <i>ATG7</i> | ENST00000354449 | 1 | missense | c.1828 | p.D610Y | tier1 | 48 | 0 | 0.000 | 110 | 79 | 0.418 |
| 190938 | 4 | 79394576 | 79394576 | G | T | SNP | <i>FRAS1</i> | ENST00000264895 | 1 | intronic | c.7523-16 | e53-16 | tier3 | 49 | 0 | 0.000 | 134 | 68 | 0.337 |
| 190938 | 4 | 101386736 | 101386736 | C | T | SNP | <i>EMCN</i> | ENST00000296420 | -1 | intronic | c.260-40 | e4-40 | tier3 | 19 | 0 | 0.000 | 96 | 77 | 0.445 |
| 19 | | | | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|---|----|-----|------------------------|-----------------|----|-----------------------------|-------------------|-----------|-------|-----|----|-------|------|------|-------|
| 190938 | 11 | 60230585 | 60230585 | A | T | SNP | <i>MS4A1</i> | ENST00000345732 | 1 | silent | c.270 | p.G90 | tier1 | 27 | 0 | 0.000 | 62 | 26 | 0.296 |
| 190938 | 13 | 42803590 | 42803590 | G | A | SNP | <i>DGKH</i> | ENST00000337343 | 1 | 3_prime_untranslated_region | c.*266 | NULL | tier2 | 28 | 0 | 0.000 | 68 | 51 | 0.429 |
| 190938 | 14 | 54416601 | 54416602 | - | AT | INS | <i>BMP4</i> | ENST00000245451 | -1 | 3_prime_untranslated_region | c.*149 | NULL | tier4 | 66 | 1 | 0.015 | 276 | 44 | 0.138 |
| 190938 | 15 | 43572011 | 43572011 | G | C | SNP | <i>TGM7</i> | ENST00000452443 | -1 | missense | c.1490 | p.A497G | tier1 | 89 | 0 | 0.000 | 136 | 82 | 0.374 |
| 190938 | 17 | 1028614 | 1028614 | G | A | SNP | <i>ABR</i> | ENST00000302538 | -1 | silent | c.150 | p.I50 | tier1 | 84 | 0 | 0.000 | 49 | 54 | 0.519 |
| 190938 | 17 | 7578190 | 7578190 | T | C | SNP | <i>TP53</i> | ENST00000269305 | -1 | missense | c.659 | p.Y220C | tier1 | 797 | 7 | 0.009 | 495 | 636 | 0.561 |
| 190938 | 19 | 50826916 | 50826916 | C | T | SNP | <i>KCNC3</i> | ENST00000477616 | -1 | missense | c.1294 | p.V432M | tier1 | 18 | 0 | 0.000 | 21 | 13 | 0.382 |
| 190938 | 19 | 51958846 | 51958846 | G | A | SNP | <i>SIGLEC8</i> | ENST00000321424 | -1 | silent | c.877 | p.L293 | tier1 | 33 | 0 | 0.000 | 60 | 35 | 0.368 |
| 190938 | 20 | 58381127 | 58381127 | C | G | SNP | <i>PHACTR3</i> | ENST00000371015 | 1 | silent | c.1206 | p.L402 | tier1 | 76 | 0 | 0.000 | 106 | 59 | 0.358 |
| 190938 | X | 124455660 | 124455660 | C | G | SNP | <i>ENSG00000263886</i> | ENST00000580355 | 1 | 3_prime_flanking_region | c.*33968 | NULL | tier3 | 39 | 0 | 0.000 | 30 | 70 | 0.700 |
| 199019 | 1 | 1688824 | 1688824 | A | - | DEL | <i>NADK</i> | ENST00000344463 | -1 | intronic | c.576-75 | e5-75 | tier3 | 123 | 1 | 0.008 | 182 | 22 | 0.108 |
| 199019 | 1 | 1747229 | 1747229 | T | C | SNP | <i>GNB1</i> | ENST00000378609 | -1 | missense | c.169 | p.K57E | tier1 | 636 | 12 | 0.019 | 623 | 296 | 0.321 |
| 199019 | 1 | 100602624 | 100602624 | A | G | SNP | <i>TRMT13</i> | ENST00000370141 | 1 | missense | c.244 | p.R82G | tier1 | 104 | 1 | 0.010 | 176 | 126 | 0.415 |
| 199019 | 1 | 104260547 | 104260547 | G | A | SNP | <i>AMY1P1</i> | ENST00000440059 | 1 | intronic | c.NULL | NULL | tier3 | 81 | 2 | 0.024 | 137 | 53 | 0.279 |
| 199019 | 1 | 248402867 | 248402867 | G | T | SNP | <i>OR2M4</i> | ENST00000306687 | 1 | missense | c.637 | p.V213F | tier1 | 71 | 0 | 0.000 | 125 | 62 | 0.332 |
| 199019 | 2 | 95942786 | 95942786 | C | T | SNP | <i>PROM2</i> | ENST00000317620 | 1 | missense | c.749 | p.A250V | tier1 | 51 | 2 | 0.038 | 48 | 17 | 0.262 |
| 199019 | 2 | 100015184 | 100015184 | G | C | SNP | <i>EIF5B</i> | ENST00000470023 | 1 | 3_prime_untranslated_region | c.*100015184 | NULL | tier3 | 96 | 3 | 0.030 | 111 | 38 | 0.255 |
| 199019 | 2 | 212288901 | 212288901 | C | T | SNP | <i>ERBB4</i> | ENST00000342788 | -1 | missense | c.2845 | p.V949I | tier1 | 136 | 3 | 0.022 | 188 | 61 | 0.245 |
| 199019 | 2 | 219267899 | 219267899 | C | T | SNP | <i>CTDSP1</i> | ENST00000273062 | 1 | intronic | c.471+49 | e5+49 | tier3 | 82 | 0 | 0.000 | 108 | 50 | 0.317 |
| 199019 | 3 | 154002358 | 154002358 | T | - | DEL | <i>DHX36</i> | ENST00000496811 | -1 | intronic | c.2292+14 | e19+14 | tier3 | 73 | 0 | 0.000 | 126 | 15 | 0.106 |
| 199019 | 4 | 106156453 | 106156453 | G | T | SNP | <i>TET2</i> | ENST00000380013 | 1 | nonsense | c.1354 | p.E452* | tier1 | 640 | 32 | 0.048 | 846 | 413 | 0.328 |
| 199019 | 4 | 106196304 | 106196304 | A | T | SNP | <i>TET2</i> | ENST00000380013 | 1 | missense | c.4637 | p.Q1546L | tier1 | 741 | 5 | 0.007 | 1064 | 110 | 0.093 |
| 199019 | 5 | 7302322 | 7302322 | G | A | SNP | <i>ENSG00000205976</i> | ENST00000512854 | -1 | intronic | c.NULL | NULL | tier3 | 175 | 4 | 0.022 | 199 | 115 | 0.363 |
| 199019 | 7 | 57529430 | 57529430 | A | C | SNP | <i>ZNF716</i> | ENST00000420713 | 1 | silent | c.1263 | p.S421 | tier1 | 81 | 0 | 0.000 | 112 | 33 | 0.228 |
| 199019 | 8 | 38835514 | 38835514 | C | T | SNP | <i>HTRA4</i> | ENST00000302495 | 1 | missense | c.814 | p.R272W | tier1 | 70 | 0 | 0.000 | 84 | 40 | 0.323 |
| 199019 | 11 | 46747390 | 46747390 | C | T | SNP | <i>F2</i> | ENST00000311907 | 1 | intronic | c.560-19 | e7-19 | tier4 | 23 | 0 | 0.000 | 31 | 20 | 0.392 |
| 199019 | 11 | 65062076 | 65062076 | G | A | SNP | <i>POLA2</i> | ENST00000265465 | 1 | silent | c.1413 | p.L471 | tier1 | 74 | 0 | 0.000 | 114 | 29 | 0.203 |
| 199019 | 11 | 71949104 | 71949104 | G | A | SNP | <i>INPL1</i> | ENST00000298229 | 1 | missense | c.3571 | p.G1191R | tier1 | 139 | 0 | 0.000 | 209 | 13 | 0.058 |
| 199019 | 11 | 89018012 | 89018012 | G | T | SNP | <i>TYR</i> | ENST00000263321 | 1 | missense | c.1256 | p.G419V | tier1 | 171 | 2 | 0.012 | 178 | 71 | 0.285 |
| 199019 | 12 | 49426412 | 49426413 | - | T | INS | <i>KMT2D</i> | ENST00000301067 | -1 | frame_shift_ins | c.12076_12075 | p.Q4025fs | tier1 | 789 | 16 | 0.020 | 1091 | 388 | 0.262 |
| 199019 | 12 | 119844492 | 119844492 | A | - | DEL | <i>CCDC60</i> | ENST00000327554 | 1 | intronic | c.91-21996 | e2-21996 | tier3 | 53 | 0 | 0.000 | 77 | 14 | 0.154 |
| 199019 | 12 | 124241407 | 124241407 | G | A | SNP | <i>ATP6VOA2</i> | ENST00000330342 | 1 | missense | c.2339 | p.R780H | tier1 | 93 | 4 | 0.041 | 107 | 47 | 0.305 |
| 199019 | 14 | 106357572 | 106357572 | A | G | SNP | <i>IGHD6-19</i> | ENST00000390574 | -1 | silent | c.6 | p.Y2 | tier1 | 342 | 5 | 0.014 | 387 | 117 | 0.232 |
| 199019 | 15 | 50897271 | 50897271 | C | T | SNP | <i>TRPM7</i> | ENST00000313478 | -1 | missense | c.2780 | p.S927N | tier1 | 47 | 0 | 0.000 | 97 | 49 | 0.336 |
| 199019 | 16 | 4736332 | 4736332 | G | A | SNP | <i>MGRN1</i> | ENST00000399577 | 1 | 3_prime_untranslated_region | c.*32 | NULL | tier2 | 134 | 3 | 0.022 | 151 | 65 | 0.301 |
| 199019 | 17 | 7578445 | 7578445 | A | T | SNP | <i>TP53</i> | ENST00000269305 | -1 | missense | c.485 | p.I162N | tier1 | 987 | 46 | 0.044 | 601 | 1092 | 0.645 |
| 199019 | 17 | 61898424 | 61898424 | A | G | SNP | <i>FTSJ3</i> | ENST00000427159 | -1 | silent | c.1938 | p.D646 | tier1 | 79 | 2 | 0.025 | 98 | 33 | 0.252 |
| 199019 | 17 | 72589154 | 72589154 | T | C | SNP | <i>C17orf77</i> | ENST00000328023 | 1 | 3_prime_untranslated_region | c.*237 | NULL | tier3 | 106 | 1 | 0.009 | 117 | 53 | 0.312 |
| 199019 | 17 | 75209492 | 75209492 | G | A | SNP | <i>SEC14L1</i> | ENST00000392476 | 1 | missense | c.1960 | p.D654N | tier1 | 62 | 0 | 0.000 | 93 | 22 | 0.191 |
| 199019 | X | 46513121 | 46513121 | A | T | SNP | <i>SLC9A7</i> | ENST00000328306 | -1 | missense | c.1067 | p.F356Y | tier1 | 179 | 3 | 0.017 | 184 | 54 | 0.227 |
| 199019 | X | 132437274 | 132437274 | C | T | SNP | <i>GPC4</i> | ENST00000370828 | -1 | missense | c.1388 | p.R463H | tier1 | 70 | 0 | 0.000 | 119 | 52 | 0.304 |
| 221833 | 1 | 3566445 | 3566445 | A | C | SNP | <i>WRAP73</i> | ENST00000270708 | -1 | intronic | c.69+50 | e1+50 | tier2 | 25 | 1 | 0.039 | 48 | 56 | 0.539 |
| 221833 | 2 | 59482372 | 59482372 | G | A | SNP | <i>LOC101927285</i> | ENST00000409590 | 1 | rna | NULL | NULL | tier1 | 103 | 1 | 0.010 | 281 | 107 | 0.276 |
| 221833 | 2 | 74742861 | 74742861 | T | A | SNP | <i>TLX2</i> | ENST00000233638 | 1 | missense | c.502 | p.Q168K | tier1 | 55 | 1 | 0.018 | 64 | 156 | 0.709 |
| 221833 | 2 | 86923062 | 86923062 | C | A | SNP | <i>CHMP3</i> | ENST00000439940 | -1 | intronic | c.1-7614 | e1-7614 | tier4 | 17 | 0 | 0.000 | 37 | 76 | 0.673 |
| 221833 | 2 | 113190918 | 113190918 | C | G | SNP | <i>RGPD8</i> | ENST00000302558 | -1 | intronic | c.72+41 | e1+41 | tier4 | 51 | 2 | 0.037 | 111 | 44 | 0.282 |
| 221833 | 2 | 198638852 | 198638852 | G | A | SNP | <i>ENSG00000222017</i> | ENST00000409845 | 1 | 3_prime_untranslated_region | c.*71 | NULL | tier3 | 43 | 1 | 0.023 | 131 | 40 | 0.234 |
| 221833 | 3 | 38766591 | 38766591 | C | A | SNP | <i>SCN10A</i> | ENST00000449082 | -1 | intronic | c.3228+74 | e17+74 | tier3 | 54 | 0 | 0.000 | 87 | 69 | 0.442 |
| 221833 | 3 | 57616622 | 57616622 | G | A | SNP | <i>DENND6A</i> | ENST00000311128 | -1 | intronic | c.1407+30 | e16+30 | tier3 | 39 | 1 | 0.025 | 119 | 102 | 0.462 |
| 221833 | 3 | 97806175 | 97806175 | C | T | SNP | <i>ORSAC2</i> | ENST00000358642 | 1 | silent | c.159 | p.N53 | tier1 | 81 | 4 | 0.047 | 107 | 83 | 0.437 |
| 221833 | 3 | 195690286 | 195690286 | C | G | SNP | <i>SDHAP1</i> | ENST00000354559 | -1 | rna | NULL | NULL | tier1 | 209 | 3 | 0.014 | 906 | 64 | 0.066 |
| 221833 | 4 | 6050724 | 6050725 | - | T | INS | <i>JAKMIP1</i> | ENST00000409021 | -1 | intronic | c.1963-76_1963-75 | e15-75 | tier3 | 35 | 0 | 0.000 | 108 | 23 | 0.176 |
| 221833 | 4 | 106156548 | 106156548 | T | - | DEL | <i>TET2</i> | ENST00000380013 | 1 | frame_shift_del | c.1449 | p.C484fs | tier1 | 400 | 4 | 0.010 | 689 | 497 | 0.419 |
| 221833 | 4 | 106164859 | 106164859 | A | - | DEL | <i>TET2</i> | ENST00000380013 | 1 | frame_shift_del | c.3727 | p.K1243fs | tier1 | 276 | 7 | 0.025 | 392 | 339 | 0.464 |
| 221833 | 4 | 115898324 | 115898324 | A | G | SNP | <i>NDST4</i> | ENST00000264363 | -1 | intronic | c.1066+19 | e2+19 | tier3 | 46 | 3 | 0.061 | 86 | 59 | 0.407 |
| 221833 | 5 | 13901373 | 13901373 | C | T | SNP | <i>DNAH5</i> | ENST00000265104 | -1 | silent | c.2040 | p.A680 | tier1 | 370 | 8 | 0.021 | 569 | 456 | 0.444 |
| 221833 | 6 | 33381539 | 33381539 | G | A | SNP | <i>PHF1</i> | ENST00000374516 | 1 | missense | c.614 | p.R205Q | tier1 | 53 | 1 | 0.019 | 69 | 77 | 0.524 |
| 221833 | 7 | 36396735 | 36396735 | G | A | SNP | <i>KIAA0895</i> | ENST00000297063 | -1 | missense | c.643 | p.P215S | tier1 | 44 | 0 | 0.000 | 55 | 49 | 0.471 |
| 221833 | 7 | 63032656 | 63032656 | G | A | SNP | <i>TNRC18P2</i> | ENST00000417666 | -1 | intronic | c.NULL | NULL | tier2 | 112 | 0 | 0.000 | 289 | 86 | 0.229 |
| 221833 | 7 | 74482588 | 74482588 | C | T | SNP | <i>WBSR16</i> | ENST00000329959 | -1 | missense | c.472 | p.V158M | tier1 | 15 | 0 | 0.000 | 35 | 27 | 0.436 |
| 221833 | 9 | 9091069 | 9091069 | G | A | SNP | <i>RPS26P3</i> | ENST00000470205 | 1 | 3_prime_untranslated_region | c.*9091069 | NULL | tier3 | 53 | 0 | 0.000 | 137 | 19 | 0.122 |
| 221833 | 9 | 138642911 | 138642911 | G | A | SNP | <i>KCNT1</i> | ENST00000298480 | 1 | intronic | c.434+24 | e4+24 | tier3 | 163 | 4 | 0.024 | 234 | 210 | 0.473 |
| 221833 | 11 | 63671439 | 63671439 | C | T | SNP | <i>MARK2</i> | ENST00000402010 | 1 | intronic | c.1515-19 | e15-19 | tier3 | 35 | 2 | 0.054 | 58 | 41 | 0.414 |
| 221833 | 12 | 53578786 | 53578786 | C | T | SNP | <i>ZNF740</i> | ENST00000416904 | 1 | missense | c.121 | p.R41W | tier1 | 21 | 0 | 0.000 | 43 | 16 | 0.271 |
| 221833 | 14 | 60395157 | 60395157 | C | T | SNP | <i>LRRC9</i> | ENST00000254271 | 1 | missense | c.220 | p.P74S | tier1 | 76 | 0 | 0.000 | 153 | 123 | 0.444 |
| 221833 | 15 | 29488779 | 29488779 | G | A | SNP | <i>FAM189A1</i> | ENST00000560021 | -1 | 5_prime_untranslated_region | c.-29488779 | NULL | tier3 | 21 | 0 | 0.000 | 43 | 26 | 0.377 |
| 221833 | 16 | 67650720 | 67650720 | G | A | SNP | <i>CTCF</i> | ENST00000264010 | 1 | missense | c.1025 | p.R342H | tier1 | 279 | 2 | 0.007 | 624 | 444 | 0.415 |
| 2 | | | | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|---|-----|-----|------------------------|-----------------|----|-----------------------------|-------------------|----------------------|----------|-------|-----|-------|-------|-----|-------|-------|
| 222841 | 2 | 91885956 | 91885956 | A | C | SNP | <i>ENSG00000230572</i> | ENST00000436174 | -1 | intronic | c.NULL | NULL | tier3 | 421 | 4 | 0.009 | 1163 | 50 | 0.041 | |
| 222841 | 3 | 17418001 | 17418002 | - | A | INS | <i>TBC1D5</i> | ENST00000253692 | -1 | intronic | c.701+15_701+14 | e8+15 | tier3 | 78 | 0 | 0.000 | 207 | 22 | 0.096 | |
| 222841 | 3 | 49571621 | 49571621 | T | - | DEL | <i>DAG1</i> | ENST00000308775 | -1 | 3_prime_untranslated_region | c.*989 | NULL | tier3 | 46 | 0 | 0.000 | 63 | 14 | 0.182 | |
| 222841 | 3 | 62142833 | 62142833 | C | A | SNP | <i>PTPRG</i> | ENST00000474889 | 1 | missense | c.775 | p.P259T | tier1 | 85 | 1 | 0.012 | 206 | 26 | 0.112 | |
| 222841 | 3 | 195453104 | 195453104 | G | A | SNP | <i>MUC20</i> | ENST00000447234 | 1 | missense | c.1630 | p.V544I | tier1 | 152 | 0 | 0.000 | 234 | 20 | 0.079 | |
| 222841 | 4 | 8601068 | 8601068 | G | A | SNP | <i>CPZ</i> | ENST00000360986 | 1 | intronic | c.89-112 | e2-112 | tier3 | 137 | 0 | 0.000 | 280 | 25 | 0.082 | |
| 222841 | 4 | 78694321 | 78694321 | C | T | SNP | <i>CNOT6L</i> | ENST00000264903 | -1 | splice_site | c.315-1 | e4-1 | tier1 | 180 | 0 | 0.000 | 341 | 53 | 0.134 | |
| 222841 | 5 | 71496184 | 71496184 | C | G | SNP | <i>MAP1B</i> | ENST00000296755 | 1 | silent | c.7002 | p.T2334 | tier1 | 371 | 1 | 0.003 | 767 | 51 | 0.062 | |
| 222841 | 6 | 101215150 | 101215150 | G | C | SNP | <i>ASCC3</i> | ENST00000369162 | -1 | nonsense | c.1467 | p.Y489* | tier1 | 199 | 1 | 0.005 | 489 | 28 | 0.054 | |
| 222841 | 6 | 117631399 | 117631399 | G | C | SNP | <i>ROS1</i> | ENST00000368508 | -1 | silent | c.6279 | p.T2093 | tier1 | 145 | 0 | 0.000 | 267 | 16 | 0.057 | |
| 222841 | 7 | 72814623 | 72814623 | C | T | SNP | <i>FZD9</i> | ENST00000344575 | 1 | 5_prime_flanking_region | c.-33715 | NULL | tier3 | 210 | 1 | 0.005 | 345 | 25 | 0.068 | |
| 222841 | 8 | 19694580 | 19694580 | C | G | SNP | <i>INTS10</i> | ENST00000520670 | 1 | missense | c.17 | p.S6C | tier1 | 113 | 0 | 0.000 | 246 | 21 | 0.079 | |
| 222841 | 8 | 22548343 | 22548343 | C | T | SNP | <i>EGR3</i> | ENST00000317216 | -1 | silent | c.807 | p.P269 | tier1 | 276 | 0 | 0.000 | 562 | 25 | 0.043 | |
| 222841 | 8 | 140768059 | 140768059 | A | G | SNP | <i>TRAPPC9</i> | ENST00000389328 | -1 | intronic | c.3350-23614 | e22-23614 | tier3 | 92 | 1 | 0.011 | 159 | 22 | 0.122 | |
| 222841 | 10 | 42949783 | 42949783 | C | A | SNP | <i>CCNYL2</i> | ENST00000426433 | -1 | 5_prime_untranslated_region | c.-42949783 | NULL | tier3 | 97 | 4 | 0.039 | 137 | 38 | 0.216 | |
| 222841 | 10 | 73497534 | 73497534 | C | G | SNP | <i>C10orf105</i> | ENST00000398786 | -1 | 5_prime_untranslated_region | c.-21440 | NULL | tier3 | 294 | 0 | 0.000 | 505 | 26 | 0.049 | |
| 222841 | 11 | 32444548 | 32444548 | C | G | SNP | <i>WT1</i> | ENST00000332351 | -1 | intronic | c.872+4954 | e3+4954 | tier3 | 220 | 0 | 0.000 | 491 | 33 | 0.063 | |
| 222841 | 12 | 49170094 | 49170094 | G | A | SNP | <i>ADCY6</i> | ENST00000307885 | -1 | silent | c.1575 | p.N525 | tier1 | 412 | 2 | 0.005 | 742 | 31 | 0.040 | |
| 222841 | 12 | 109604677 | 109604677 | C | G | SNP | <i>ACACB</i> | ENST00000338432 | 1 | missense | c.665 | p.S222W | missense | tier1 | 109 | 0 | 0.000 | 193 | 17 | 0.081 |
| 222841 | 13 | 36410159 | 36410159 | A | T | SNP | <i>DCLK1</i> | ENST00000360631 | -1 | intronic | c.1229+11 | e7+11 | tier3 | 246 | 0 | 0.000 | 666 | 50 | 0.070 | |
| 222841 | 16 | 81412314 | 81412314 | G | C | SNP | <i>GAN</i> | ENST00000568107 | 1 | 3_prime_untranslated_region | c.*1113 | NULL | tier3 | 107 | 0 | 0.000 | 297 | 21 | 0.066 | |
| 222841 | 17 | 7577568 | 7577568 | C | T | SNP | <i>TP53</i> | ENST00000269305 | -1 | missense | c.713 | p.C238Y | missense | tier1 | 803 | 19 | 0.023 | 873 | 482 | 0.355 |
| 222841 | 17 | 10350502 | 10350502 | C | A | SNP | <i>MYH4</i> | ENST00000255381 | -1 | missense | c.4997 | p.R1666I | missense | tier1 | 417 | 2 | 0.005 | 927 | 47 | 0.048 |
| 222841 | 17 | 10401122 | 10401122 | G | C | SNP | <i>MYH1</i> | ENST00000226207 | -1 | missense | c.4294 | p.L1432V | missense | tier1 | 323 | 0 | 0.000 | 628 | 21 | 0.032 |
| 222841 | 19 | 9176322 | 9176322 | G | C | SNP | <i>OR1M1</i> | ENST00000429566 | 1 | 5_prime_flanking_region | c.-27599 | NULL | missense | tier3 | 199 | 0 | 0.000 | 620 | 26 | 0.040 |
| 222841 | 22 | 40055124 | 40055124 | G | C | SNP | <i>CACNA1I</i> | ENST00000336649 | 1 | splice_region | c.2348+3 | e15+3 | missense | tier2 | 263 | 0 | 0.000 | 428 | 21 | 0.047 |
| 222841 | X | 137714472 | 137714472 | A | G | SNP | <i>FGF13</i> | ENST00000315930 | -1 | 3_prime_untranslated_region | c.*539 | NULL | missense | tier3 | 94 | 0 | 0.000 | 169 | 15 | 0.082 |
| 222841 | X | 152801875 | 152801875 | G | A | SNP | <i>ATP2B3</i> | ENST00000263519 | -1 | missense | c.170 | p.S57N | missense | tier1 | 500 | 0 | 0.000 | 883 | 39 | 0.042 |
| 229276 | 1 | 15903 | 15904 | - | C | INS | <i>WASH7P</i> | ENST00000438504 | -1 | splice_site_ins | c.NULL | c.NULL | missense | tier1 | 7 | 0 | 0.000 | 2 | 12 | 0.857 |
| 229276 | 1 | 17084269 | 17084269 | G | A | SNP | <i>MST1L</i> | ENST00000455405 | -1 | rna | NULL | NULL | missense | tier1 | 141 | 3 | 0.021 | 378 | 42 | 0.100 |
| 229276 | 1 | 186647324 | 186647324 | T | - | DEL | <i>PTGS2</i> | ENST00000367468 | -1 | intronic | c.457+69 | e4+69 | missense | tier3 | 33 | 0 | 0.000 | 73 | 18 | 0.198 |
| 229276 | 1 | 201172932 | 201172932 | C | T | SNP | <i>IGFN1</i> | ENST00000335211 | 1 | missense | c.814 | p.R272C | missense | tier1 | 71 | 0 | 0.000 | 116 | 26 | 0.183 |
| 229276 | 2 | 231974064 | 231974065 | - | A | INS | <i>HTR2B</i> | ENST00000258400 | -1 | frame_shift_ins | c.613_612 | p.I204fs | missense | tier1 | 61 | 0 | 0.000 | 125 | 19 | 0.132 |
| 229276 | 3 | 38739967 | 38739967 | G | A | SNP | <i>SCN10A</i> | ENST00000449082 | -1 | missense | c.4744 | p.R1582C | missense | tier1 | 74 | 0 | 0.000 | 195 | 30 | 0.133 |
| 229276 | 3 | 53736702 | 53736702 | C | T | SNP | <i>CACNA1D</i> | ENST00000288139 | 1 | missense | c.1255 | p.R419W | missense | tier1 | 76 | 2 | 0.026 | 165 | 33 | 0.167 |
| 229276 | 3 | 68684184 | 68684184 | C | T | SNP | <i>ENSG00000244640</i> | ENST00000461635 | -1 | 5_prime_flanking_region | c.-677 | NULL | missense | tier4 | 65 | 0 | 0.000 | 109 | 26 | 0.193 |
| 229276 | 4 | 15055756 | 15055756 | A | G | SNP | <i>CPEB2</i> | ENST00000538197 | 1 | silent | c.2376 | p.E792 | missense | tier1 | 102 | 1 | 0.010 | 229 | 46 | 0.167 |
| 229276 | 4 | 36214995 | 36214995 | T | C | SNP | <i>ARAP2</i> | ENST00000303965 | -1 | intronic | c.965-54 | e3-54 | missense | tier3 | 173 | 1 | 0.006 | 256 | 33 | 0.114 |
| 229276 | 4 | 39233719 | 39233720 | - | A | INS | <i>WDR19</i> | ENST00000399820 | 1 | intronic | c.2143-63_2143-62 | e19-62 | missense | tier3 | 43 | 0 | 0.000 | 78 | 14 | 0.152 |
| 229276 | 4 | 39928393 | 39928393 | A | T | SNP | <i>PDS5A</i> | ENST00000303538 | -1 | splice_site | c.429+2 | e3+2 | missense | tier1 | 195 | 1 | 0.005 | 312 | 20 | 0.059 |
| 229276 | 4 | 95202758 | 95202758 | T | A | SNP | <i>SMARCAD1</i> | ENST00000359052 | 1 | intronic | c.2732+17 | e20+17 | missense | tier3 | 64 | 0 | 0.000 | 85 | 12 | 0.124 |
| 229276 | 5 | 55237459 | 55237459 | A | C | SNP | <i>IL6ST</i> | ENST00000336909 | -1 | silent | c.2208 | p.S736 | missense | tier1 | 77 | 1 | 0.013 | 162 | 26 | 0.138 |
| 229276 | 5 | 60170374 | 60170374 | A | C | SNP | <i>ERCC8</i> | ENST00000462279 | -1 | rna | NULL | NULL | missense | tier1 | 129 | 0 | 0.000 | 200 | 33 | 0.142 |
| 229276 | 6 | 30593399 | 30593399 | C | A | SNP | <i>MRPS18B</i> | ENST00000259873 | 1 | missense | c.602 | p.P201Q | missense | tier1 | 60 | 0 | 0.000 | 116 | 36 | 0.237 |
| 229276 | 6 | 52883236 | 52883236 | C | T | SNP | <i>ICK</i> | ENST00000350082 | -1 | silent | c.555 | p.A185 | missense | tier1 | 99 | 0 | 0.000 | 196 | 33 | 0.144 |
| 229276 | 6 | 76022886 | 76022886 | G | A | SNP | <i>FILIP1</i> | ENST00000237172 | -1 | missense | c.2662 | p.P888S | missense | tier1 | 88 | 0 | 0.000 | 183 | 32 | 0.149 |
| 229276 | 6 | 88125414 | 88125414 | C | T | SNP | <i>C6orf165</i> | ENST00000369562 | 1 | silent | c.294 | p.L98 | missense | tier1 | 169 | 0 | 0.000 | 273 | 45 | 0.142 |
| 229276 | 9 | 139997605 | 139997605 | A | G | SNP | <i>MAN1B1</i> | ENST00000536349 | 1 | 3_prime_untranslated_region | c.*139997605 | NULL | missense | tier3 | 81 | 1 | 0.012 | 173 | 42 | 0.194 |
| 229276 | 9 | 139997644 | 139997644 | A | G | SNP | <i>MAN1B1</i> | ENST00000536349 | 1 | 3_prime_untranslated_region | c.*139997644 | NULL | missense | tier3 | 75 | 3 | 0.039 | 141 | 50 | 0.254 |
| 229276 | 10 | 15177301 | 15177301 | A | G | SNP | <i>NMT2</i> | ENST00000378165 | -1 | silent | c.363 | p.F121 | missense | tier1 | 30 | 0 | 0.000 | 38 | 14 | 0.269 |
| 229276 | 12 | 12006417 | 12006417 | - | T | INS | <i>ETV6</i> | ENST00000396373 | 1 | frame_shift_ins | c.385_386 | p.S131fs | missense | tier1 | 379 | 7 | 0.018 | 618 | 110 | 0.151 |
| 229276 | 12 | 13229047 | 13229047 | A | T | SNP | <i>KIAA1467</i> | ENST00000197268 | 1 | missense | c.1612 | p.N538Y | missense | tier1 | 110 | 1 | 0.009 | 203 | 25 | 0.109 |
| 229276 | 12 | 64842427 | 64842427 | T | A | SNP | <i>XPOT</i> | ENST00000332707 | 1 | 3_prime_untranslated_region | c.*516 | NULL | missense | tier4 | 120 | 0 | 0.000 | 133 | 27 | 0.169 |
| 229276 | 12 | 112657189 | 112657189 | C | T | SNP | <i>HECTD4</i> | ENST00000377560 | -1 | splice_site | c.6558+1 | e43+1 | missense | tier1 | 35 | 0 | 0.000 | 66 | 17 | 0.205 |
| 229276 | 12 | 124082411 | 124082411 | G | T | SNP | <i>TMED2</i> | ENST00000262225 | 1 | 3_prime_untranslated_region | c.*1135 | NULL | missense | tier3 | 33 | 0 | 0.000 | 64 | 14 | 0.175 |
| 229276 | 17 | 29553477 | 29553478 | - | C | INS | <i>NF1</i> | ENST00000358273 | 1 | frame_shift_ins | c.2026_2027 | p.I679fs | missense | tier1 | 301 | 4 | 0.013 | 489 | 85 | 0.148 |
| 229276 | 19 | 36125240 | 36125240 | G | A | SNP | <i>RBM42</i> | ENST00000262633 | 1 | missense | c.1100 | p.S367N | missense | tier1 | 42 | 0 | 0.000 | 102 | 25 | 0.197 |
| 229276 | 21 | 36259295 | 36259316 | CAGCGCCGGCGTCC GGGGCGCC CCACGCCGG | - | DEL | <i>RUNX1</i> | ENST00000300305 | -1 | frame_shift_del | c.196_175 | p.G59fs | missense | tier1 | 460 | 0 | 0.000 | 933 | 41 | 0.042 |
| 229276 | X | 2835999 | 2836007 | - | DEL | DEL | <i>ARSD</i> | ENST00000381154 | -1 | in_frame_del | c.709_701 | p.AGV234in_frame_del | missense | tier1 | 31 | 0 | 0.000 | 59 | 14 | 0.192 |
| 229276 | X | 39934092 | 39934092 | G | - | DEL | <i>BCOR</i> | ENST00000378444 | -1 | frame_shift_del | c.507 | p.D169fs | missense | tier1 | 289 | 9 | 0.030 | 396 | 133 | 0.251 |
| 229276 | X | 123179001 | 123179001 | A | G | SNP | <i>STAG2</i> | ENST00000218089 | 1 | intronic | c.463-13 | e6-13 | missense | tier2 | 158 | 2 | 0.013 | 184 | 75 | 0.287 |
| 243777 | 1 | 3428641 | 3428641 | T | C | SNP | <i>MEGF6</i> | ENST00000356575 | -1 | missense | c.905 | p.N302S | missense | tier1 | 89 | 0 | 0.000 | 155 | 16 | 0.093 |
| 243777 | 1 | 24383863 | 24383863 | C | T | SNP | <i>MYOM3</i> | ENST00000330966 | -1 | silent | c.4314 | p.K1438 | missense | tier1 | 274 | 9 | 0.032 | 435 | 137 | 0.238 |
| 243777 | 1 | 152082466 | 152082466 | A | C | SNP | <i>TCHH</i> | ENST00000368804 | -1 | missense | c.3227 | p.L1076R | missense | tier1 | 125 | 1 | 0.008 | 146 | 16 | 0.099 |
| 243777 | 1 | 230921753 | 230921753 | A | G | SNP | <i>CAPN9</i> | ENST00000271971 | 1 | missense | c.1508 | p.D503G | missense | tier1 | 208 | 0 | 0.000 | 337 | 16 | 0.045 |
| 243777 | 2 | 103 | | | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|-----|---------------|-----|------------------------|-----------------|----|-----------------------------|-------------------|-------------------|-------|-----|----|-------|------|-----|-------|
| 243777 | 6 | 42985377 | 42985377 | G | A | SNP | <i>KLHDC3</i> | ENST00000326974 | 1 | nonsense | c.275 | p.W92* | tier1 | 63 | 0 | 0.000 | 137 | 21 | 0.133 |
| 243777 | 6 | 64290808 | 64290808 | A | A | SNP | <i>PTP4A1</i> | ENST00000370651 | 1 | 3_prime_untranslated_region | c.*729 | NULL | tier3 | 100 | 0 | 0.000 | 158 | 39 | 0.197 |
| 243777 | 7 | 135263622 | 135263622 | C | G | SNP | <i>NUP205</i> | ENST00000285968 | 1 | missense | c.1001 | p.A334V | tier1 | 151 | 5 | 0.032 | 216 | 55 | 0.202 |
| 243777 | 8 | 101522892 | 101522892 | A | - | DEL | <i>ANKRD46</i> | ENST00000520552 | -1 | intronic | c.637-25 | e4-25 | tier3 | 38 | 0 | 0.000 | 48 | 10 | 0.172 |
| 243777 | 8 | 117859879 | 117859879 | G | A | SNP | <i>RAD21</i> | ENST00000297338 | -1 | nonsense | c.1756 | p.R586* | tier1 | 810 | 5 | 0.006 | 1188 | 130 | 0.099 |
| 243777 | 9 | 108125237 | 108125237 | G | A | SNP | <i>SLC44A1</i> | ENST00000374720 | 1 | missense | c.1036 | p.V346I | tier1 | 123 | 0 | 0.000 | 203 | 50 | 0.198 |
| 243777 | 10 | 54076396 | 54076396 | T | C | SNP | <i>DKK1</i> | ENST00000373970 | 1 | silent | c.630 | p.C210 | tier1 | 71 | 1 | 0.014 | 75 | 33 | 0.306 |
| 243777 | 10 | 135350563 | 135350563 | G | T | SNP | <i>CYP2E1</i> | ENST00000252945 | 1 | splice_region | c.968-4 | e7-4 | tier3 | 51 | 0 | 0.000 | 85 | 19 | 0.183 |
| 243777 | 11 | 67552796 | 67552796 | G | T | SNP | <i>ALG1L8P</i> | ENST00000533887 | 1 | 3_prime_untranslated_region | c.*67552796 | NULL | tier3 | 97 | 0 | 0.000 | 146 | 46 | 0.240 |
| 243777 | 11 | 71175151 | 71175151 | C | A | SNP | <i>NADSYN1</i> | ENST00000319023 | 1 | missense | c.370 | p.R124S | tier1 | 40 | 0 | 0.000 | 39 | 17 | 0.304 |
| 243777 | 11 | 108188267 | 108188267 | T | - | DEL | <i>ATM</i> | ENST00000278616 | 1 | intronic | c.6347+19 | e42+19 | tier3 | 39 | 0 | 0.000 | 69 | 13 | 0.159 |
| 243777 | 11 | 132081872 | 132081872 | T | A | SNP | <i>NTM</i> | ENST00000539799 | 1 | intronic | c.401-44 | e4-44 | tier3 | 85 | 2 | 0.023 | 91 | 25 | 0.216 |
| 243777 | 12 | 11992117 | 11992117 | G | A | SNP | <i>ETV6</i> | ENST00000396373 | 1 | nonsense | c.207 | p.W69* | tier1 | 566 | 3 | 0.005 | 876 | 122 | 0.122 |
| 243777 | 16 | 12297010 | 12297010 | T | G | SNP | <i>SNX29</i> | ENST00000306030 | 1 | intronic | c.523+3470 | e6+3470 | tier3 | 265 | 3 | 0.011 | 510 | 36 | 0.066 |
| 243777 | 16 | 51175096 | 51175096 | G | A | SNP | <i>SALL1</i> | ENST00000251020 | -1 | missense | c.1037 | p.A346V | tier1 | 77 | 1 | 0.013 | 97 | 32 | 0.248 |
| 243777 | 17 | 10305158 | 10305158 | - | T | INS | <i>MYH8</i> | ENST00000403437 | -1 | intronic | c.2689-57_2689-56 | e21-56 | tier3 | 168 | 2 | 0.012 | 223 | 46 | 0.171 |
| 243777 | 17 | 10451010 | 10451010 | G | A | SNP | <i>MYH2</i> | ENST00000245503 | -1 | intronic | c.204+24 | e1+24 | tier3 | 156 | 2 | 0.013 | 268 | 63 | 0.190 |
| 243777 | 17 | 62399159 | 62399160 | - | T | INS | <i>RPL31P57</i> | ENST00000461109 | -1 | 5_prime_flanking_region | c.-13109 | NULL | tier3 | 85 | 0 | 0.000 | 103 | 37 | 0.264 |
| 243777 | 17 | 78313118 | 78313118 | C | G | SNP | <i>RNF213</i> | ENST00000582970 | 1 | missense | c.4951 | p.L1651V | tier1 | 565 | 18 | 0.031 | 806 | 234 | 0.225 |
| 243777 | 18 | 42531917 | 42531917 | T | C | SNP | <i>SETBP1</i> | ENST00000282030 | 1 | missense | c.2612 | p.I871T | tier1 | 545 | 3 | 0.006 | 951 | 30 | 0.031 |
| 243777 | 18 | 77896475 | 77896475 | A | T | SNP | <i>ADNP2</i> | ENST00000262198 | 1 | missense | c.3179 | p.D1060V | tier1 | 123 | 5 | 0.039 | 159 | 77 | 0.326 |
| 243777 | 19 | 1525633 | 1525633 | C | T | SNP | <i>PLK5</i> | ENST00000334770 | 1 | 5_prime_untranslated_region | c.-1363 | NULL | tier2 | 92 | 0 | 0.000 | 119 | 62 | 0.343 |
| 243777 | 19 | 51358189 | 51358189 | T | A | SNP | <i>KLK3</i> | ENST00000326003 | 1 | 5_prime_untranslated_region | c.-23 | NULL | tier3 | 81 | 1 | 0.012 | 100 | 43 | 0.301 |
| 243777 | 19 | 57722941 | 57722941 | A | G | SNP | <i>ZNF264</i> | ENST00000263095 | 1 | missense | c.476 | p.D159G | tier1 | 89 | 0 | 0.000 | 143 | 21 | 0.128 |
| 243777 | 20 | 3214593 | 3214593 | C | T | SNP | <i>SLC4A11</i> | ENST00000380059 | -1 | silent | c.708 | p.R236 | tier1 | 67 | 1 | 0.015 | 95 | 34 | 0.264 |
| 243777 | 20 | 31022762 | 31022762 | C | - | DEL | <i>ASXL1</i> | ENST00000375687 | 1 | frame_shift_del | c.2247 | p.V751fs | tier1 | 718 | 15 | 0.021 | 939 | 327 | 0.258 |
| 243777 | 21 | 9755905 | 9755905 | A | G | SNP | - | - | - | - | - | - | tier3 | 31 | 0 | 0.000 | 72 | 33 | 0.314 |
| 243777 | 21 | 44514777 | 44514777 | T | G | SNP | <i>U2AF1</i> | ENST00000291552 | -1 | missense | c.470 | p.Q157P | tier1 | 314 | 5 | 0.016 | 477 | 121 | 0.202 |
| 266995 | 1 | 24417462 | 24417462 | C | A | SNP | <i>MYOM3</i> | ENST00000330966 | -1 | missense | c.1260 | p.W420C | tier1 | 329 | 1 | 0.003 | 901 | 40 | 0.043 |
| 266995 | 1 | 181706618 | 181706618 | G | A | SNP | <i>CACNA1E</i> | ENST00000367573 | 1 | intronic | c.3423-43 | e23-43 | tier3 | 354 | 0 | 0.000 | 511 | 43 | 0.077 |
| 266995 | 2 | 152267722 | 152267722 | T | - | DEL | <i>RIF1</i> | ENST00000243326 | 1 | intronic | c.105-52 | e2-52 | tier2 | 20 | 0 | 0.000 | 12 | 7 | 0.368 |
| 266995 | 3 | 14239517 | 14239518 | - | T | INS | <i>LSM3</i> | ENST00000306024 | 1 | intronic | c.229-19_229-18 | e4-18 | tier2 | 88 | 0 | 0.000 | 114 | 13 | 0.102 |
| 266995 | 3 | 39230412 | 39230412 | C | T | SNP | <i>XIRP1</i> | ENST00000340369 | -1 | silent | c.525 | p.L175 | tier1 | 530 | 2 | 0.004 | 988 | 64 | 0.061 |
| 266995 | 3 | 100463779 | 100463779 | A | G | SNP | <i>TFG</i> | ENST00000240851 | 1 | splice_region | c.820+4 | e6+4 | tier3 | 107 | 1 | 0.009 | 185 | 21 | 0.102 |
| 266995 | 3 | 137981440 | 137981440 | A | - | DEL | <i>ARMC8</i> | ENST00000469044 | 1 | intronic | c.1135-1141 | e13-1141 | tier4 | 58 | 0 | 0.000 | 60 | 9 | 0.130 |
| 266995 | 6 | 132067939 | 132067939 | A | G | SNP | <i>ENPP3</i> | ENST00000357639 | 1 | missense | c.2471 | p.E824G | tier1 | 140 | 0 | 0.000 | 200 | 16 | 0.074 |
| 266995 | 7 | 129028858 | 129028858 | T | - | DEL | <i>AHCYL2</i> | ENST00000491079 | 1 | 3_prime_untranslated_region | c.*129028858 | NULL | tier3 | 86 | 1 | 0.012 | 104 | 14 | 0.119 |
| 266995 | 8 | 80948699 | 80948699 | C | A | SNP | <i>TPD52</i> | ENST00000518937 | -1 | 3_prime_untranslated_region | c.*1652 | NULL | tier3 | 112 | 0 | 0.000 | 106 | 9 | 0.078 |
| 266995 | 8 | 139736829 | 139736829 | G | A | SNP | <i>COL22A1</i> | ENST00000303045 | -1 | intronic | c.2247+29 | e24+29 | tier3 | 65 | 0 | 0.000 | 91 | 17 | 0.157 |
| 266995 | 10 | 95720906 | 95720906 | G | A | SNP | <i>PIPSL</i> | ENST00000480546 | -1 | rna | NULL | NULL | tier1 | 161 | 0 | 0.000 | 270 | 18 | 0.063 |
| 266995 | 12 | 25398262 | 25398262 | G | G | SNP | <i>KRAS</i> | ENST00000256078 | -1 | missense | c.57 | p.L19F | tier1 | 478 | 0 | 0.000 | 750 | 46 | 0.058 |
| 266995 | 12 | 54369181 | 54369181 | C | C | SNP | <i>HOXC11</i> | ENST00000546378 | 1 | missense | c.899 | p.G300A | tier1 | 184 | 0 | 0.000 | 339 | 18 | 0.050 |
| 266995 | 12 | 133050261 | 133050261 | G | A | SNP | <i>MUC8</i> | ENST00000595994 | -1 | silent | c.183 | p.L61 | tier1 | 130 | 0 | 0.000 | 129 | 12 | 0.085 |
| 266995 | 13 | 101762936 | 101762936 | G | A | SNP | <i>NALCN</i> | ENST00000251127 | -1 | intronic | c.2364+34 | e19+34 | tier2 | 390 | 2 | 0.005 | 454 | 47 | 0.094 |
| 266995 | 13 | 101910940 | 101910940 | C | G | SNP | <i>NALCN</i> | ENST00000251127 | -1 | intronic | c.1135-15 | e10-15 | tier3 | 234 | 0 | 0.000 | 381 | 34 | 0.081 |
| 266995 | 15 | 101844339 | 101844339 | C | G | SNP | <i>ENSG00000259172</i> | ENST00000558838 | 1 | rna | NULL | NULL | tier1 | 114 | 0 | 0.000 | 179 | 17 | 0.087 |
| 266995 | 16 | 81941970 | 81941970 | C | A | SNP | <i>PLCG2</i> | ENST00000359376 | 1 | intronic | c.1558-51 | e16-51 | tier3 | 113 | 0 | 0.000 | 199 | 15 | 0.070 |
| 266995 | 18 | 14830201 | 14830201 | C | G | SNP | <i>MIR3156-2</i> | ENST00000581101 | 1 | rna | NULL | NULL | tier1 | 243 | 0 | 0.000 | 247 | 12 | 0.046 |
| 266995 | 19 | 9057573 | 9057573 | G | A | SNP | <i>MUC16</i> | ENST00000397910 | -1 | missense | c.29873 | p.T9958I | tier1 | 734 | 0 | 0.000 | 1536 | 68 | 0.042 |
| 266995 | X | 107811978 | 107811978 | G | A | SNP | <i>COL4A5</i> | ENST00000328300 | 1 | intronic | c.322-11 | e6-11 | tier3 | 220 | 0 | 0.000 | 320 | 28 | 0.080 |
| 270086 | 6 | 56473262 | 56473262 | G | A | SNP | <i>DST</i> | ENST00000370754 | -1 | missense | c.6065 | p.T2022I | tier1 | 777 | 0 | 0.000 | 1084 | 44 | 0.039 |
| 270086 | 7 | 114563154 | 114563154 | C | A | SNP | <i>MDFIC</i> | ENST00000393486 | 1 | silent | c.66 | p.G22 | tier1 | 6 | 0 | 0.000 | 1 | 8 | 0.889 |
| 270086 | 9 | 35906348 | 35906350 | CTG | - | DEL | <i>HRCT1</i> | ENST00000354323 | 1 | in_frame_del | c.64_66 | p.L25in_frame_del | tier1 | 74 | 0 | 0.000 | 89 | 11 | 0.110 |
| 270086 | 14 | 23567883 | 23567883 | A | T | SNP | <i>C14orf119</i> | ENST00000319074 | 1 | 3_prime_untranslated_region | c.*593 | NULL | tier4 | 86 | 0 | 0.000 | 124 | 13 | 0.095 |
| 270086 | 19 | 633529 | 633530 | - | GCGCCGCCGCCGC | INS | <i>POLRMT</i> | ENST00000588649 | -1 | 5_prime_untranslated_region | c.-17 | NULL | tier2 | 16 | 0 | 0.000 | 16 | 10 | 0.385 |
| 270086 | 19 | 4817310 | 4817310 | A | C | SNP | <i>TICAM1</i> | ENST00000248244 | -1 | silent | c.1080 | p.P360 | tier1 | 186 | 2 | 0.011 | 256 | 24 | 0.085 |
| 270086 | 21 | 14469103 | 14469103 | G | A | SNP | <i>ZNF355P</i> | ENST00000427301 | -1 | 5_prime_untranslated_region | c.-14469103 | NULL | tier3 | 347 | 0 | 0.000 | 347 | 15 | 0.041 |
| 272848 | 1 | 145684679 | 145684679 | G | A | SNP | <i>RNF115</i> | ENST00000369291 | 1 | splice_site | c.667+1 | e7+1 | tier1 | 48 | 2 | 0.039 | 118 | 60 | 0.337 |
| 272848 | 2 | 65497525 | 65497525 | A | G | SNP | <i>ACTR2</i> | ENST00000260641 | 1 | 3_prime_untranslated_region | c.*1657 | NULL | tier3 | 46 | 0 | 0.000 | 62 | 43 | 0.410 |
| 272848 | 2 | 90109121 | 90109121 | G | A | SNP | <i>IGKV1D-17</i> | ENST00000483379 | 1 | 5_prime_flanking_region | c.-12537 | NULL | tier3 | 49 | 0 | 0.000 | 81 | 46 | 0.362 |
| 272848 | 2 | 144159094 | 144159094 | A | T | SNP | <i>ARHGAP15</i> | ENST00000295095 | 1 | intronic | c.475-34076 | e6-34076 | tier2 | 19 | 0 | 0.000 | 78 | 36 | 0.316 |
| 272848 | 3 | 37095520 | 37095520 | A | G | SNP | <i>LRRFIP2</i> | ENST00000336686 | -1 | intronic | c.2056-68 | e27-68 | tier3 | 29 | 0 | 0.000 | 26 | 28 | 0.519 |
| 272848 | 3 | 39453515 | 39453515 | A | G | SNP | <i>RPSA</i> | ENST00000301821 | 1 | silent | c.756 | p.V252 | tier1 | 62 | 0 | 0.000 | 118 | 17 | 0.126 |
| 272848 | 3 | 41722946 | 41722946 | G | - | DEL | <i>ULK4</i> | ENST00000301831 | -1 | intronic | c.2978+53 | e28+53 | tier3 | 40 | 0 | 0.000 | 57 | 32 | 0.360 |
| 272848 | 4 | 3891295 | 3891295 | C | T | SNP | <i>OR7E163P</i> | ENST00000505943 | -1 | 5_prime_untranslated_region | c.-3891295 | NULL | tier3 | 43 | 0 | 0.000 | 67 | 35 | 0.343 |
| 272848 | 4 | 85654560 | 85654560 | T | A | SNP | <i>WDFY3</i> | ENST00000295888 | -1 | missense | c.7196 | p.E2399V | tier1 | 48 | 0 | 0.000 | 47 | 49 | 0.510 |
| 272848 | 4 | 140619263 | 140619263 | T | A | SNP | <i>MGST2</i> | ENST00000265498 | 1 | intronic | c.229+2842 | e3+2842 | tier3 | 50 | 0 | 0.000 | 105 | 16 | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|--------------------------|---|-----|-----------------|-----------------|----|-----------------------------|-------------------|-----------|-------|-----|---|-------|------|-----|-------|
| 272848 | 8 | 100523442 | 100523442 | G | C | SNP | VPS13B | ENST00000358544 | 1 | missense | c.4410 | p.E1470D | tier1 | 47 | 0 | 0.000 | 70 | 59 | 0.457 |
| 272848 | 9 | 35480204 | 35480204 | C | T | SNP | ATP8B5P | ENST00000329395 | 1 | rna | NULL | NULL | tier1 | 44 | 0 | 0.000 | 18 | 23 | 0.561 |
| 272848 | 9 | 66457014 | 66457014 | C | T | SNP | LOC100966870 | ENST00000424345 | 1 | 5_prime_flanking_region | c.-275 | NULL | tier4 | 45 | 0 | 0.000 | 69 | 14 | 0.169 |
| 272848 | 9 | 115932843 | 115932843 | T | C | SNP | FKBP15 | ENST00000238256 | -1 | missense | c.2725 | p.N909D | tier1 | 55 | 1 | 0.018 | 84 | 25 | 0.227 |
| 272848 | 9 | 136634584 | 136634584 | C | T | SNP | VAV2 | ENST00000371850 | -1 | missense | c.2389 | p.G797S | tier1 | 39 | 0 | 0.000 | 59 | 36 | 0.379 |
| 272848 | 10 | 69695996 | 69695996 | T | C | SNP | HERC4 | ENST00000395198 | -1 | splice_region | c.2596-4 | e21-4 | tier4 | 37 | 0 | 0.000 | 58 | 44 | 0.431 |
| 272848 | 10 | 99660362 | 99660362 | C | T | SNP | CRTAC1 | ENST00000370597 | -1 | intronic | c.1133+898 | e8+898 | tier3 | 248 | 2 | 0.008 | 252 | 256 | 0.503 |
| 272848 | 11 | 9081984 | 9081984 | G | C | SNP | SCUBE2 | ENST00000457346 | -1 | missense | c.938 | p.T313S | tier1 | 50 | 1 | 0.020 | 54 | 32 | 0.372 |
| 272848 | 11 | 51411669 | 51411669 | C | T | SNP | OR4A5 | ENST00000319760 | -1 | missense | c.727 | p.V243I | tier1 | 40 | 0 | 0.000 | 60 | 17 | 0.218 |
| 272848 | 11 | 85988219 | 85988219 | A | G | SNP | EED | ENST00000525244 | 1 | 3_prime_untranslated_region | c.*85988219 | NULL | tier3 | 62 | 0 | 0.000 | 169 | 148 | 0.467 |
| 272848 | 12 | 58159282 | 58159282 | C | T | SNP | CYP27B1 | ENST00000546609 | -1 | missense | c.299 | p.R100Q | tier1 | 47 | 0 | 0.000 | 40 | 31 | 0.437 |
| 272848 | 15 | 41634594 | 41634594 | T | A | SNP | NUSAP1 | ENST00000559596 | 1 | nonsense | c.104 | p.L35* | tier1 | 35 | 0 | 0.000 | 69 | 44 | 0.389 |
| 272848 | 16 | 16272771 | 16272771 | C | T | SNP | ABCC6 | ENST00000205557 | -1 | missense | c.2299 | p.V767I | tier1 | 50 | 0 | 0.000 | 80 | 17 | 0.175 |
| 272848 | 16 | 30793418 | 30793418 | T | - | DEL | ZNF629 | ENST00000262525 | -1 | frame_shift_del | c.2231 | p.K744fs | tier1 | 45 | 0 | 0.000 | 43 | 35 | 0.449 |
| 272848 | 16 | 31090611 | 31090611 | C | T | SNP | ZNF646 | ENST00000300850 | 1 | missense | c.2966 | p.A989V | tier1 | 59 | 0 | 0.000 | 74 | 52 | 0.413 |
| 272848 | 16 | 53529201 | 53529201 | C | T | SNP | AKTIP | ENST00000300245 | -1 | missense | c.286 | p.V96M | tier1 | 30 | 0 | 0.000 | 57 | 41 | 0.418 |
| 272848 | 16 | 87870085 | 87870085 | C | T | SNP | SLC7A5 | ENST00000261622 | -1 | intronic | c.1290+19 | e8+19 | tier3 | 49 | 0 | 0.000 | 65 | 25 | 0.278 |
| 272848 | 17 | 33476023 | 33476023 | G | A | SNP | UNC45B | ENST00000268876 | 1 | missense | c.200 | p.S67Y | tier1 | 32 | 0 | 0.000 | 41 | 24 | 0.369 |
| 272848 | 18 | 42531907 | 42531907 | C | A | SNP | SETBP1 | ENST00000282030 | 1 | missense | c.2602 | p.D868N | tier1 | 367 | 0 | 0.000 | 609 | 133 | 0.179 |
| 272848 | 18 | 42531913 | 42531913 | G | T | SNP | SETBP1 | ENST00000282030 | 1 | missense | c.2608 | p.G870C | tier1 | 367 | 0 | 0.000 | 527 | 189 | 0.264 |
| 272848 | 19 | 4310495 | 4310495 | G | A | SNP | FSD1 | ENST00000221856 | 1 | missense | c.392 | p.R131Q | tier1 | 36 | 0 | 0.000 | 56 | 42 | 0.429 |
| 272848 | 19 | 19351506 | 19351506 | G | C | SNP | NCAN | ENST00000252575 | 1 | intronic | c.3492+12 | e11+12 | tier3 | 40 | 0 | 0.000 | 41 | 29 | 0.414 |
| 272848 | 21 | 46957834 | 46957834 | C | T | SNP | SLC19A1 | ENST00000311124 | -1 | missense | c.40 | p.V14M | tier1 | 53 | 0 | 0.000 | 58 | 38 | 0.396 |
| 272848 | X | 12739552 | 12739552 | G | T | SNP | FRMPD4 | ENST00000380682 | 1 | 3_prime_untranslated_region | c.*900 | NULL | tier3 | 19 | 0 | 0.000 | 29 | 15 | 0.341 |
| 272848 | X | 77224984 | 77224984 | T | C | SNP | PGAM4 | ENST00000458128 | -1 | missense | c.152 | p.E51G | tier1 | 81 | 0 | 0.000 | 55 | 36 | 0.391 |
| 272848 | X | 120008710 | 120008710 | T | G | SNP | CT47B1 | ENST00000371311 | -1 | intronic | c.775+40 | e1+40 | tier2 | 74 | 0 | 0.000 | 78 | 42 | 0.347 |
| 272848 | X | 145701712 | 145701712 | G | A | SNP | ENSG00000231686 | ENST00000445173 | -1 | 5_prime_untranslated_region | c.-145701712 | NULL | tier2 | 50 | 0 | 0.000 | 39 | 30 | 0.435 |
| 275454 | 1 | 1920435 | 1920435 | A | - | DEL | C1orf222 | ENST00000434971 | -1 | intronic | c.68-23 | e2-23 | tier3 | 95 | 0 | 0.000 | 165 | 18 | 0.098 |
| 275454 | 5 | 37212598 | 37212598 | A | - | DEL | C5orf42 | ENST00000425232 | -1 | intronic | c.2920+1063 | e15+1063 | tier3 | 43 | 0 | 0.000 | 82 | 14 | 0.146 |
| 275454 | 5 | 65475096 | 65475096 | T | - | DEL | SREK1 | ENST00000284041 | 1 | rna | NULL | NULL | tier1 | 81 | 0 | 0.000 | 126 | 15 | 0.106 |
| 275454 | 5 | 131607200 | 131607200 | C | T | SNP | PDLIM4 | ENST00000462597 | 1 | 3_prime_untranslated_region | c.*131607200 | NULL | tier2 | 131 | 0 | 0.000 | 234 | 15 | 0.060 |
| 275454 | 5 | 161579342 | 161579342 | G | A | SNP | GABRG2 | ENST00000356592 | 1 | intronic | c.1152+583 | e9+583 | tier3 | 151 | 0 | 0.000 | 286 | 17 | 0.056 |
| 275454 | 12 | 107237614 | 107237615 | - | T | INS | RIC8B | ENST00000392837 | 1 | intronic | c.1066-16_1066-15 | e6-15 | tier3 | 73 | 1 | 0.014 | 117 | 23 | 0.164 |
| 275454 | 12 | 123107166 | 123107166 | A | - | DEL | KNTC1 | ENST00000333479 | 1 | intronic | c.6515+12 | e61+12 | tier3 | 54 | 0 | 0.000 | 77 | 16 | 0.172 |
| 275454 | 14 | 19807134 | 19807134 | G | T | SNP | OR11H13P | ENST00000359695 | -1 | 5_prime_untranslated_region | c.-19807134 | NULL | tier3 | 618 | 1 | 0.002 | 1105 | 29 | 0.026 |
| 275454 | 18 | 14809914 | 14809914 | C | G | SNP | ANKRD30B | ENST00000358984 | 1 | intronic | c.2313+1336 | e25+1336 | tier3 | 206 | 0 | 0.000 | 352 | 19 | 0.051 |
| 280837 | 1 | 1669668 | 1669668 | G | C | SNP | SLC35E2 | ENST00000246421 | -1 | intronic | c.586+92 | e3+92 | tier2 | 17 | 0 | 0.000 | 127 | 97 | 0.433 |
| 280837 | 1 | 27861349 | 27861349 | T | G | SNP | AHDC1 | ENST00000482400 | -1 | 5_prime_untranslated_region | c.-27861349 | NULL | tier3 | 32 | 1 | 0.030 | 96 | 43 | 0.309 |
| 280837 | 1 | 28605680 | 28605680 | G | A | SNP | SESN2 | ENST00000253063 | 1 | silent | c.1284 | p.V428 | tier1 | 29 | 0 | 0.000 | 103 | 86 | 0.453 |
| 280837 | 1 | 71998450 | 71998450 | C | T | SNP | NEGR1 | ENST00000357731 | -1 | intronic | c.940+60050 | e6+60050 | tier3 | 63 | 2 | 0.031 | 123 | 70 | 0.363 |
| 280837 | 1 | 151675014 | 151675014 | A | C | SNP | CELF3 | ENST00000470688 | -1 | 5_prime_untranslated_region | c.-151675014 | NULL | tier2 | 87 | 2 | 0.022 | 199 | 35 | 0.150 |
| 280837 | 1 | 161976283 | 161976283 | G | A | SNP | OLFML2B | ENST00000294794 | -1 | intronic | c.547-20 | e4-20 | tier3 | 47 | 4 | 0.078 | 32 | 37 | 0.507 |
| 280837 | 2 | 25467497 | 25467497 | G | A | SNP | DNMT3A | ENST00000264709 | -1 | nonsense | c.1579 | p.Q527* | tier1 | 122 | 5 | 0.039 | 622 | 489 | 0.437 |
| 280837 | 2 | 131453866 | 131453866 | T | C | SNP | ENSG00000240253 | ENST00000425151 | 1 | intronic | c.NULL | NULL | tier3 | 42 | 1 | 0.023 | 74 | 62 | 0.456 |
| 280837 | 2 | 234184342 | 234184342 | G | A | SNP | ATG16L1 | ENST00000392017 | 1 | intronic | c.954+918 | e9+918 | tier4 | 50 | 1 | 0.020 | 119 | 40 | 0.252 |
| 280837 | 3 | 118913221 | 118913221 | C | T | SNP | UPK1B | ENST00000264234 | 1 | silent | c.624 | p.G208 | tier1 | 59 | 0 | 0.000 | 103 | 100 | 0.490 |
| 280837 | 4 | 1949812 | 1949812 | A | C | SNP | WHSC1 | ENST00000398261 | 1 | 3_prime_untranslated_region | c.*5684 | NULL | tier3 | 49 | 0 | 0.000 | 67 | 65 | 0.492 |
| 280837 | 4 | 71888174 | 71888174 | G | T | SNP | DCK | ENST00000286648 | 1 | missense | c.298 | p.A100S | tier1 | 88 | 2 | 0.022 | 121 | 102 | 0.453 |
| 280837 | 4 | 76817552 | 76817552 | G | C | SNP | PPEF2 | ENST00000511880 | -1 | 5_prime_untranslated_region | c.-74 | NULL | tier2 | 21 | 0 | 0.000 | 90 | 71 | 0.438 |
| 280837 | 4 | 109769882 | 109769882 | C | T | SNP | COL25A1 | ENST00000399132 | -1 | intronic | c.1434+33 | e26+33 | tier3 | 80 | 0 | 0.000 | 92 | 58 | 0.384 |
| 280837 | 5 | 34190834 | 34190834 | G | A | SNP | ENSG00000215156 | ENST00000332686 | 1 | 3_prime_untranslated_region | c.*34190834 | NULL | tier3 | 525 | 2 | 0.004 | 6549 | 156 | 0.023 |
| 280837 | 5 | 36958149 | 36958156 | GTCTTCTT | - | DEL | NIPBL | ENST00000282516 | 1 | intronic | c.231-57_231-50 | e3-50 | tier3 | 32 | 0 | 0.000 | 33 | 14 | 0.298 |
| 280837 | 5 | 36958159 | 36958178 | GAATCAGTCACCATT TTAAT | - | DEL | NIPBL | ENST00000282516 | 1 | intronic | c.231-47_231-28 | e3-28 | tier3 | 33 | 0 | 0.000 | 34 | 14 | 0.292 |
| 280837 | 5 | 74712651 | 74712651 | C | A | SNP | COL4A3BP | ENST00000380494 | -1 | intronic | c.1221+50 | e8+50 | tier3 | 177 | 2 | 0.011 | 148 | 111 | 0.429 |
| 280837 | 6 | 33136506 | 33136506 | C | T | SNP | COL11A2 | ENST00000341947 | -1 | missense | c.3883 | p.E1295K | tier1 | 31 | 0 | 0.000 | 192 | 144 | 0.427 |
| 280837 | 6 | 36698561 | 36698561 | C | A | SNP | P116 | ENST00000536757 | 1 | intronic | c.1+17544 | e0+17544 | tier4 | 13 | 0 | 0.000 | 40 | 40 | 0.500 |
| 280837 | 7 | 1055741 | 1055741 | A | G | SNP | C7orf50 | ENST00000357429 | -1 | intronic | c.130-5962 | e2-5962 | tier4 | 25 | 0 | 0.000 | 93 | 35 | 0.273 |
| 280837 | 7 | 1055742 | 1055742 | G | C | SNP | C7orf50 | ENST00000357429 | -1 | intronic | c.130-5963 | e2-5963 | tier4 | 26 | 0 | 0.000 | 91 | 37 | 0.287 |
| 280837 | 7 | 75050891 | 75050891 | T | C | SNP | POM121C | ENST00000257665 | -1 | missense | c.3370 | p.T1124A | tier1 | 23 | 0 | 0.000 | 33 | 31 | 0.477 |
| 280837 | 7 | 128553132 | 128553132 | T | C | SNP | ENSG00000224163 | ENST00000433622 | -1 | 5_prime_untranslated_region | c.-128553132 | NULL | tier3 | 23 | 0 | 0.000 | 23 | 20 | 0.465 |
| 280837 | 7 | 150714308 | 150714308 | C | T | SNP | ATG9B | ENST00000377974 | -1 | 5_prime_untranslated_region | c.-150714308 | NULL | tier2 | 33 | 0 | 0.000 | 107 | 42 | 0.280 |
| 280837 | 7 | 157509090 | 157509090 | A | G | SNP | PTPRN2 | ENST00000404321 | -1 | intronic | c.1858-33461 | e13-33461 | tier3 | 27 | 0 | 0.000 | 52 | 15 | 0.224 |
| 280837 | 7 | 157509091 | 157509091 | C | T | SNP | PTPRN2 | ENST00000404321 | -1 | intronic | c.1858-33462 | e13-33462 | tier3 | 27 | 0 | 0.000 | 54 | 19 | 0.260 |
| 280837 | 7 | 157651592 | 157651592 | C | T | SNP | PTPRN2 | ENST00000404321 | -1 | intronic | c.1857+39773 | e12+39773 | tier3 | 80 | 1 | 0.012 | 325 | 43 | 0.117 |
| 280837 | 8 | 131129068 | 131129068 | G | T | SNP | ASAP1 | ENST00000343135 | -1 | intronic | c.2005+58 | e21+58 | tier3 | 80 | 0 | 0.000 | 128 | 116 | 0.474 |
| 280837 | 9 | 139903977 | 139903977 | C | A | SNP | ABCA2 | ENST00000355090 | -1 | intronic | c.6824+16 | e44+16 | tier2 | 33 | 0 | 0.000 | 191 | 175 | 0.478 |
| 280837 | 10 | 38651191 | 38651191 | G | A | SNP | HSD17B7P2 | ENST00000494540 | 1 | 3_prime_untranslated_region | c.*38651191 | NULL | tier3 | 142 | 2 | 0.014 | 173 | 151 | 0.466 |
| 280837 | 10 | 68139043 | 68139043 | T | C | SNP | CTNNA3 | ENST00000373744 | -1 | silent | c.1599 | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|---|-----|-----|-----------------|-----------------|----|-----------------------------|-------------------|----------|-------|------|----|-------|------|-----|-------|
| 280837 | 12 | 50344754 | 50344754 | G | A | SNP | AQP2 | ENST00000199280 | 1 | silent | c.141 | p.A47 | tier1 | 30 | 0 | 0.000 | 143 | 120 | 0.456 |
| 280837 | 17 | 1563288 | 1563288 | T | A | SNP | PRPF8 | ENST00000304992 | -1 | missense | c.4793 | p.D1598V | tier1 | 87 | 1 | 0.011 | 479 | 400 | 0.454 |
| 280837 | 17 | 27210235 | 27210235 | C | T | SNP | FLOT2 | ENST00000394906 | -1 | silent | c.402 | p.T134 | tier1 | 19 | 1 | 0.050 | 125 | 112 | 0.473 |
| 280837 | 17 | 73910078 | 73910078 | C | T | SNP | FBF1 | ENST00000389570 | -1 | silent | c.3003 | p.Q1001 | tier1 | 9 | 0 | 0.000 | 150 | 123 | 0.447 |
| 280837 | 18 | 5891961 | 5891961 | G | G | SNP | TMEM200C | ENST00000383490 | -1 | missense | c.102 | p.R34S | tier1 | 23 | 0 | 0.000 | 84 | 56 | 0.400 |
| 280837 | 19 | 9001863 | 9001863 | C | A | SNP | MUC16 | ENST00000380947 | -1 | nonsense | c.904 | p.R302* | tier1 | 87 | 1 | 0.011 | 376 | 334 | 0.468 |
| 280837 | 19 | 18553578 | 18553578 | C | T | SNP | ELL | ENST00000262809 | -1 | 3_prime_untranslated_region | c.*1984 | NULL | tier3 | 583 | 0 | 0.000 | 1133 | 27 | 0.023 |
| 280837 | 19 | 51890473 | 51890473 | C | T | SNP | LIM2 | ENST00000221973 | -1 | silent | c.225 | p.A75 | tier1 | 28 | 0 | 0.000 | 91 | 58 | 0.389 |
| 280837 | 20 | 30409206 | 30409206 | C | T | SNP | MYLK2 | ENST00000375985 | 1 | intronic | c.474-36 | e3-36 | tier3 | 18 | 0 | 0.000 | 80 | 56 | 0.412 |
| 280837 | X | 129147939 | 129147939 | G | A | SNP | BCORL1 | ENST00000303743 | 1 | missense | c.1191 | p.M397I | tier1 | 9 | 0 | 0.000 | 30 | 208 | 0.822 |
| 283773 | 1 | 33476435 | 33476435 | C | A | SNP | AK2 | ENST00000373449 | -1 | splice_site | c.695-1 | e7-1 | tier1 | 108 | 0 | 0.000 | 83 | 9 | 0.098 |
| 283773 | 1 | 89236209 | 89236209 | G | T | SNP | PKN2 | ENST00000370521 | 1 | intronic | c.622+57 | e4+57 | tier3 | 53 | 0 | 0.000 | 22 | 6 | 0.214 |
| 283773 | 1 | 201180702 | 201180702 | G | C | SNP | IGFN1 | ENST00000335211 | 1 | silent | c.6681 | p.G2227 | tier1 | 273 | 0 | 0.000 | 120 | 7 | 0.055 |
| 283773 | 1 | 211836204 | 211836204 | G | A | SNP | NEK2 | ENST00000366999 | -1 | 3_prime_untranslated_region | c.*564 | NULL | tier3 | 31 | 0 | 0.000 | 27 | 9 | 0.250 |
| 283773 | 2 | 20423047 | 20423048 | - | A | INS | SDC1 | ENST00000482879 | -1 | rna | NULL | NULL | tier1 | 133 | 1 | 0.008 | 50 | 7 | 0.123 |
| 283773 | 2 | 84668259 | 84668259 | A | - | DEL | SUCLG1 | ENST00000393868 | -1 | intronic | c.532-31 | e5-31 | tier3 | 56 | 0 | 0.000 | 52 | 10 | 0.161 |
| 283773 | 2 | 182399497 | 182399498 | - | TT | INS | ITGA4 | ENST00000397033 | 1 | intronic | c.2884-46_2884-45 | e27-45 | tier3 | 7 | 0 | 0.000 | 3 | 7 | 0.700 |
| 283773 | 2 | 220082366 | 220082366 | A | - | DEL | ABCB6 | ENST00000265316 | -1 | intronic | c.687+26 | e2+26 | tier4 | 51 | 1 | 0.019 | 33 | 10 | 0.233 |
| 283773 | 5 | 132031958 | 132031958 | T | DEL | DEL | KIF3A | ENST00000378735 | -1 | 3_prime_untranslated_region | c.*368 | NULL | tier3 | 48 | 0 | 0.000 | 35 | 9 | 0.205 |
| 283773 | 5 | 139905660 | 139905660 | C | A | SNP | ANKHD1 | ENST00000297183 | 1 | missense | c.4572 | p.F1524L | tier1 | 115 | 0 | 0.000 | 27 | 4 | 0.129 |
| 283773 | 6 | 100404004 | 100404004 | G | T | SNP | MCHR2 | ENST00000281806 | -1 | missense | c.20 | p.S7Y | tier1 | 108 | 0 | 0.000 | 22 | 4 | 0.154 |
| 283773 | 7 | 86547738 | 86547738 | A | G | SNP | KIAA1324L | ENST00000450689 | -1 | intronic | c.1593+20 | e12+20 | tier3 | 85 | 0 | 0.000 | 17 | 4 | 0.191 |
| 283773 | 7 | 127013340 | 127013340 | T | - | DEL | ZNF800 | ENST00000265827 | -1 | intronic | c.1994+56 | e4+56 | tier3 | 50 | 0 | 0.000 | 15 | 5 | 0.250 |
| 283773 | 8 | 101522892 | 101522892 | A | - | DEL | ANKRD46 | ENST00000520552 | -1 | intronic | c.637-25 | e4-25 | tier3 | 41 | 0 | 0.000 | 14 | 5 | 0.263 |
| 283773 | 9 | 20923717 | 20923717 | G | A | SNP | FOCAD | ENST00000338382 | 1 | missense | c.2911 | p.V971I | tier1 | 99 | 0 | 0.000 | 24 | 6 | 0.200 |
| 283773 | 10 | 33619598 | 33619598 | G | T | SNP | NRP1 | ENST00000265371 | -1 | intronic | c.248+38 | e2+38 | tier2 | 139 | 0 | 0.000 | 33 | 4 | 0.108 |
| 283773 | 10 | 124359634 | 124359634 | A | C | SNP | DMBT1 | ENST00000338354 | 1 | intronic | c.3310+25 | e27+25 | tier3 | 184 | 1 | 0.005 | 140 | 15 | 0.097 |
| 283773 | 11 | 559313 | 559313 | C | - | DEL | ENSG00000254815 | ENST00000527620 | 1 | rna | NULL | NULL | tier1 | 66 | 0 | 0.000 | 15 | 4 | 0.211 |
| 283773 | 12 | 124144573 | 124144573 | T | - | DEL | GTF2H3 | ENST00000543341 | 1 | intronic | c.821-20 | e12-20 | tier3 | 83 | 1 | 0.012 | 34 | 8 | 0.191 |
| 283773 | 14 | 72171435 | 72171435 | C | T | SNP | SIPA1L1 | ENST00000555818 | 1 | splice_region | c.3647-3 | e12-3 | tier3 | 105 | 0 | 0.000 | 25 | 4 | 0.138 |
| 283773 | 15 | 42503954 | 42503954 | A | - | DEL | TMEM87A | ENST00000389834 | -1 | splice_region_del | c.1627-7 | e20-7 | tier3 | 56 | 0 | 0.000 | 22 | 6 | 0.214 |
| 283773 | 16 | 4796967 | 4796967 | G | A | SNP | C16orf71 | ENST00000299320 | 1 | silent | c.1221 | p.E407 | tier1 | 115 | 0 | 0.000 | 40 | 5 | 0.111 |
| 283773 | 19 | 2121672 | 2121672 | C | T | SNP | AP3D1 | ENST00000355272 | -1 | intronic | c.1101+61 | e12+61 | tier3 | 53 | 0 | 0.000 | 20 | 11 | 0.355 |
| 283773 | 20 | 31023112 | 31023112 | T | G | SNP | ASXL1 | ENST00000375687 | 1 | nonsense | c.2597 | p.L866* | tier1 | 1114 | 3 | 0.003 | 346 | 99 | 0.222 |
| 283773 | 20 | 31451686 | 31451686 | G | T | SNP | EFCAB8 | ENST00000400522 | 1 | intronic | c.42+46 | e1+46 | tier3 | 69 | 0 | 0.000 | 39 | 6 | 0.133 |
| 283773 | 21 | 33919279 | 33919279 | A | - | DEL | RNA5SP490 | ENST00000517141 | 1 | 5_prime_flanking_region | c.-16106 | NULL | tier3 | 54 | 0 | 0.000 | 34 | 7 | 0.171 |
| 283773 | 22 | 22114660 | 22114661 | - | T | INS | MAPK1 | ENST00000215832 | -1 | 3_prime_untranslated_region | c.*8833 | NULL | tier3 | 39 | 0 | 0.000 | 21 | 6 | 0.222 |
| 283773 | 22 | 29939380 | 29939380 | G | T | SNP | THOC5 | ENST00000397871 | -1 | intronic | c.354+38 | e3+38 | tier3 | 48 | 0 | 0.000 | 19 | 5 | 0.208 |
| 283773 | 22 | 50439189 | 50439189 | C | - | DEL | IL17REL | ENST00000341280 | -1 | frame_shift_del | c.213 | p.Q72fs | tier1 | 32 | 0 | 0.000 | 7 | 4 | 0.364 |
| 283773 | X | 70339865 | 70339865 | T | A | SNP | MED12 | ENST00000333646 | 1 | missense | c.398 | p.V133D | tier1 | 755 | 3 | 0.004 | 301 | 15 | 0.048 |
| 283773 | X | 76709731 | 76709731 | C | T | SNP | FGF16 | ENST00000439435 | 1 | silent | c.84 | p.S28 | tier1 | 145 | 0 | 0.000 | 52 | 5 | 0.088 |
| 283773 | X | 106788960 | 106788960 | T | C | SNP | FRMPD3-AS1 | ENST00000415252 | -1 | rna | NULL | NULL | tier1 | 149 | 0 | 0.000 | 32 | 4 | 0.111 |
| 288033 | 1 | 146491965 | 146491965 | G | C | SNP | NBPF13P | ENST00000444680 | -1 | intronic | c.NULL | NULL | tier3 | 123 | 2 | 0.016 | 240 | 27 | 0.101 |
| 288033 | 1 | 146492064 | 146492064 | C | T | SNP | NBPF13P | ENST00000444680 | -1 | intronic | c.NULL | NULL | tier3 | 75 | 1 | 0.013 | 81 | 14 | 0.143 |
| 288033 | 2 | 33614218 | 33614218 | G | T | SNP | LTBP1 | ENST00000354476 | 1 | intronic | c.4715-33 | e32-33 | tier3 | 91 | 2 | 0.022 | 81 | 91 | 0.529 |
| 288033 | 2 | 122369100 | 122369100 | T | G | SNP | CLASP1 | ENST00000263710 | -1 | intronic | c.1-5344 | e1-5344 | tier4 | 93 | 1 | 0.011 | 135 | 18 | 0.118 |
| 288033 | 2 | 183903211 | 183903211 | G | A | SNP | NCKAP1 | ENST00000360982 | -1 | 5_prime_untranslated_region | c.-384 | NULL | tier2 | 8 | 0 | 0.000 | 0 | 7 | 1.000 |
| 288033 | 3 | 126219009 | 126219009 | C | A | SNP | UROC1 | ENST00000290868 | -1 | intronic | c.1146-12 | e12-12 | tier3 | 106 | 3 | 0.028 | 79 | 63 | 0.444 |
| 288033 | 4 | 103611794 | 103611794 | G | A | SNP | MANBA | ENST00000226578 | -1 | missense | c.808 | p.P270S | tier1 | 102 | 3 | 0.029 | 78 | 74 | 0.484 |
| 288033 | 7 | 100839218 | 100839218 | C | T | SNP | MOGAT3 | ENST00000379423 | -1 | missense | c.832 | p.A278T | tier1 | 83 | 0 | 0.000 | 90 | 11 | 0.109 |
| 288033 | 9 | 45120089 | 45120089 | G | T | SNP | ENSG00000231838 | ENST00000417850 | 1 | 5_prime_flanking_region | c.-44489 | NULL | tier3 | 34 | 0 | 0.000 | 31 | 11 | 0.262 |
| 288033 | 9 | 90503384 | 90503384 | G | C | SNP | SPATA31E1 | ENST00000325643 | 1 | missense | c.3982 | p.V1328L | tier1 | 42 | 1 | 0.023 | 19 | 27 | 0.587 |
| 288033 | 9 | 136918842 | 136918842 | T | C | SNP | BRD3 | ENST00000433041 | -1 | 5_prime_untranslated_region | c.-243 | NULL | tier3 | 74 | 1 | 0.013 | 65 | 33 | 0.337 |
| 288033 | 11 | 1997318 | 1997318 | T | C | SNP | MRPL23 | ENST00000397297 | 1 | intronic | c.497+3330 | e5+3330 | tier3 | 325 | 7 | 0.021 | 404 | 36 | 0.081 |
| 288033 | 12 | 64474024 | 64474024 | A | - | DEL | SRGAP1 | ENST00000355086 | 1 | intronic | c.1324-42 | e10-42 | tier4 | 89 | 2 | 0.022 | 62 | 13 | 0.173 |
| 288033 | 12 | 85285921 | 85285921 | A | - | DEL | SLC6A15 | ENST00000266682 | -1 | 5_prime_untranslated_region | c.-22 | NULL | tier4 | 40 | 0 | 0.000 | 37 | 8 | 0.178 |
| 288033 | 12 | 133201355 | 133201355 | C | T | SNP | POLE | ENST00000320574 | -1 | silent | c.6789 | p.Q2263 | tier1 | 147 | 0 | 0.000 | 187 | 16 | 0.079 |
| 288033 | 14 | 106330469 | 106330469 | T | C | SNP | IGHJ4 | ENST00000461719 | -1 | missense | c.2 | p.Y1C | tier1 | 393 | 0 | 0.000 | 412 | 16 | 0.037 |
| 288033 | 15 | 85327071 | 85327071 | C | T | SNP | ZNF592 | ENST00000299927 | 1 | missense | c.1165 | p.R389W | tier1 | 79 | 0 | 0.000 | 87 | 13 | 0.130 |
| 288033 | 17 | 10350420 | 10350420 | C | T | SNP | MYH4 | ENST00000255381 | -1 | silent | c.5079 | p.R1693 | tier1 | 607 | 16 | 0.026 | 495 | 445 | 0.473 |
| 288033 | 17 | 67178774 | 67178774 | A | C | SNP | ABCA10 | ENST00000269081 | -1 | intronic | c.2631+42 | e19+42 | tier3 | 32 | 0 | 0.000 | 33 | 9 | 0.118 |
| 288033 | 19 | 13941346 | 13941346 | C | G | SNP | ZSWIM4 | ENST00000254323 | 1 | missense | c.2452 | p.P818A | tier1 | 28 | 0 | 0.000 | 23 | 27 | 0.540 |
| 288033 | 19 | 33793252 | 33793253 | - | G | INS | CEBPA | ENST00000498907 | -1 | frame_shift_ins | c.69_68 | p.H24fs | tier1 | 811 | 9 | 0.011 | 957 | 184 | 0.161 |
| 288033 | 20 | 43723594 | 43723594 | A | T | SNP | KCNS1 | ENST00000306117 | -1 | missense | c.168 | p.S500T | tier1 | 46 | 0 | 0.000 | 47 | 44 | 0.484 |
| 288033 | 20 | 57768618 | 57768618 | G | A | SNP | ZNF831 | ENST00000371030 | 1 | silent | c.2544 | p.T848 | tier1 | 39 | 2 | 0.049 | 50 | 46 | 0.479 |
| 288033 | 21 | 42838094 | 42838094 | A | - | DEL | TMPRSS2 | ENST00000398585 | -1 | intronic | c.1579-14 | e14-14 | tier3 | 58 | 0 | 0.000 | 71 | 10 | 0.124 |
| 288033 | 22 | 41742111 | 41742111 | C | T | SNP | ZC3H7B | ENST00000351589 | 1 | missense | c.1564 | p.R522C | tier1 | 55 | 2 | 0.035 | 44 | 47 | 0.517 |
| 288033 | 22 | 50960184 | 50960184 | G | A | SNP | NCAPH2 | ENST00000299821 | 1 | missense | c.1015 | p.V339M | tier1 | 76 | 3 | 0.038 | 66 | 82 | 0.554 |
| 288033 | X | 107512391 | 107512391 | A | - | DEL | COL4A6 | ENST00000461897 | -1 | splice_region_del | c.NULL | NULL | tier3 | 107 | 0 | 0.000 | 106 | 11 | 0.0 |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-----|------|-----|-----------------|-----------------|----|-----------------------------|-------------------|----------|-------|------|---|-------|------|-----|-------|
| 293230 | 5 | 5462936 | 5462936 | T | C | SNP | KIAA0947 | ENST00000296564 | 1 | silent | c.3489 | p.T1163 | tier1 | 108 | 1 | 0.009 | 156 | 21 | 0.119 |
| 293230 | 5 | 67522723 | 67522723 | G | T | SNP | PIK3R1 | ENST00000396611 | 1 | missense | c.220 | p.V74L | tier1 | 182 | 0 | 0.000 | 242 | 14 | 0.055 |
| 293230 | 6 | 125146129 | 125146129 | G | T | SNP | NKAIN2 | ENST00000368417 | 1 | 3_prime_untranslated_region | c.*1754 | NULL | tier3 | 44 | 1 | 0.022 | 118 | 31 | 0.208 |
| 293230 | 6 | 159653093 | 159653093 | G | C | SNP | FNDC1 | ENST00000297267 | 1 | missense | c.1549 | p.G517R | tier1 | 87 | 2 | 0.023 | 87 | 18 | 0.171 |
| 293230 | 9 | 138641963 | 138641963 | G | A | SNP | KCNT1 | ENST00000298480 | 1 | missense | c.274 | p.V92I | tier1 | 651 | 5 | 0.008 | 771 | 60 | 0.072 |
| 293230 | 12 | 56844914 | 56844914 | G | A | SNP | MIP | ENST00000257979 | -1 | 3_prime_untranslated_region | c.*150 | NULL | tier3 | 43 | 0 | 0.000 | 42 | 9 | 0.173 |
| 293230 | 12 | 133159733 | 133159733 | C | T | SNP | FBRSL1 | ENST00000434748 | 1 | missense | c.2507 | p.A836V | tier1 | 8 | 0 | 0.000 | 3 | 7 | 0.700 |
| 293230 | 15 | 34130377 | 34130377 | C | T | SNP | RYR3 | ENST00000389232 | 1 | nonsense | c.12196 | p.R4066* | tier1 | 1002 | 0 | 0.000 | 1296 | 74 | 0.054 |
| 293230 | 15 | 91044236 | 91044236 | G | A | SNP | IQGAP1 | ENST00000268182 | 1 | 3_prime_untranslated_region | c.*896 | NULL | tier3 | 223 | 0 | 0.000 | 365 | 21 | 0.054 |
| 293230 | 16 | 1116377 | 1116377 | C | T | SNP | ENSG00000261720 | ENST00000564390 | 1 | rna | NULL | NULL | tier1 | 82 | 2 | 0.024 | 55 | 14 | 0.194 |
| 293230 | 16 | 12297010 | 12297010 | T | G | SNP | SNX29 | ENST00000306030 | 1 | intronic | c.523+3470 | e6+3470 | tier3 | 356 | 8 | 0.022 | 513 | 47 | 0.084 |
| 293230 | 16 | 67962332 | 67962332 | G | C | SNP | PSKH1 | ENST00000291041 | 1 | 3_prime_untranslated_region | c.*787 | NULL | tier2 | 88 | 0 | 0.000 | 85 | 22 | 0.202 |
| 293230 | 19 | 7995988 | 7995988 | A | - | DEL | TIMM44 | ENST00000270538 | -1 | intronic | c.1038+35 | e10+35 | tier3 | 22 | 0 | 0.000 | 30 | 13 | 0.302 |
| 293230 | X | 72224144 | 72224145 | - | C | INS | PABPC1L2B | ENST00000373521 | 1 | 3_prime_untranslated_region | c.*60 | NULL | tier2 | 14 | 0 | 0.000 | 4 | 5 | 0.556 |
| 298273 | 1 | 46159297 | 46159297 | C | T | SNP | TMEM69 | ENST00000372025 | 1 | missense | c.464 | p.A155V | tier1 | 49 | 1 | 0.020 | 125 | 100 | 0.443 |
| 298273 | 1 | 151262327 | 151262328 | - | C | INS | ZNF687 | ENST00000324048 | 1 | frame_shift_ins | c.2808_2809 | p.R938fs | tier1 | 382 | 1 | 0.003 | 514 | 321 | 0.384 |
| 298273 | 1 | 183498545 | 183498545 | G | T | SNP | SMG7 | ENST00000507469 | 1 | silent | c.720 | p.V240 | tier1 | 65 | 0 | 0.000 | 97 | 63 | 0.394 |
| 298273 | 2 | 101099723 | 101099723 | G | A | SNP | NMS | ENST00000376865 | 1 | nonsense | c.459 | p.W153* | tier1 | 39 | 0 | 0.000 | 73 | 53 | 0.421 |
| 298273 | 3 | 14106117 | 14106117 | T | C | SNP | TPRXL | ENST00000326972 | 1 | silent | c.441 | p.S147 | tier1 | 48 | 0 | 0.000 | 94 | 19 | 0.168 |
| 298273 | 3 | 46759141 | 46759141 | C | T | SNP | PRSS50 | ENST00000315170 | -1 | intronic | c.107-14 | e2-14 | tier2 | 79 | 0 | 0.000 | 82 | 58 | 0.414 |
| 298273 | 3 | 50339933 | 50339933 | C | T | SNP | HYAL1 | ENST00000266031 | -1 | missense | c.455 | p.R152Q | tier1 | 46 | 0 | 0.000 | 49 | 22 | 0.306 |
| 298273 | 3 | 58792080 | 58792081 | - | T | INS | C3orf67 | ENST00000482387 | -1 | intronic | c.1862+40_1862+39 | e11+40 | tier3 | 15 | 0 | 0.000 | 70 | 39 | 0.358 |
| 298273 | 3 | 158407931 | 158407931 | T | - | DEL | GFM1 | ENST00000486715 | 1 | intronic | c.1910-21 | e16-21 | tier3 | 25 | 0 | 0.000 | 50 | 15 | 0.231 |
| 298273 | 5 | 31405700 | 31405701 | - | A | INS | DROSHA | ENST00000344624 | -1 | intronic | c.3994+83_3994+82 | e32+83 | tier3 | 21 | 0 | 0.000 | 61 | 21 | 0.256 |
| 298273 | 5 | 140596188 | 140596188 | G | A | SNP | PCDHB13 | ENST00000341948 | 1 | 3_prime_untranslated_region | c.*96 | NULL | tier3 | 24 | 0 | 0.000 | 54 | 35 | 0.393 |
| 298273 | 5 | 170837543 | 170837544 | - | TCTG | INS | NPM1 | ENST00000296930 | 1 | frame_shift_ins | c.859_860 | p.W288fs | tier1 | 108 | 2 | 0.018 | 283 | 133 | 0.320 |
| 298273 | 5 | 179150788 | 179150789 | - | T | INS | CANX | ENST00000415618 | 1 | splice_region_ins | c.1623+8_1623+7 | e12+8 | tier3 | 35 | 0 | 0.000 | 83 | 18 | 0.178 |
| 298273 | 6 | 10397893 | 10397894 | - | A | INS | TFAP2A | ENST00000379604 | -1 | 3_prime_untranslated_region | c.*757 | NULL | tier3 | 16 | 0 | 0.000 | 26 | 16 | 0.381 |
| 298273 | 6 | 152461140 | 152461140 | C | T | SNP | SYNE1 | ENST00000265368 | -1 | missense | c.25403 | p.R8468H | tier1 | 68 | 0 | 0.000 | 52 | 50 | 0.490 |
| 298273 | 7 | 64275471 | 64275471 | T | - | DEL | ZNF138 | ENST00000359735 | 1 | intronic | c.1+49 | e0+49 | tier3 | 31 | 0 | 0.000 | 72 | 21 | 0.226 |
| 298273 | 7 | 131071805 | 131071805 | T | G | SNP | MKLN1 | ENST00000352689 | 1 | intronic | c.169-74 | e3-74 | tier3 | 9 | 0 | 0.000 | 27 | 31 | 0.535 |
| 298273 | 10 | 94773927 | 94773928 | - | T | INS | EXOC6 | ENST00000371547 | 1 | intronic | c.2144-24_2144-23 | e21-23 | tier3 | 12 | 0 | 0.000 | 35 | 29 | 0.453 |
| 298273 | 10 | 112340661 | 112340661 | G | A | SNP | SMC3 | ENST00000361804 | 1 | splice_site | c.430-1 | e8-1 | tier1 | 196 | 0 | 0.000 | 654 | 30 | 0.044 |
| 298273 | 11 | 1092457 | 1092457 | A | C | SNP | MUC2 | ENST00000441003 | 1 | missense | c.4276 | p.T1426P | tier1 | 188 | 3 | 0.016 | 425 | 38 | 0.082 |
| 298273 | 11 | 134605529 | 134605529 | A | G | SNP | ENSG00000251226 | ENST00000513405 | 1 | rna | NULL | NULL | tier1 | 120 | 1 | 0.008 | 222 | 35 | 0.136 |
| 298273 | 13 | 49073878 | 49073878 | C | T | SNP | RCBTB2 | ENST00000430805 | -1 | silent | c.1278 | p.S426 | tier1 | 41 | 0 | 0.000 | 50 | 45 | 0.474 |
| 298273 | 14 | 21841578 | 21841578 | A | G | SNP | SUPT16H | ENST00000216297 | -1 | silent | c.78 | p.D26 | tier1 | 24 | 0 | 0.000 | 75 | 44 | 0.367 |
| 298273 | 17 | 1563288 | 1563288 | T | C | SNP | PRPF8 | ENST00000304992 | -1 | missense | c.4793 | p.D1598G | tier1 | 266 | 2 | 0.007 | 432 | 319 | 0.423 |
| 298273 | 17 | 35503592 | 35503592 | A | G | SNP | ACACA | ENST00000353139 | -1 | intronic | c.5679+3196 | e45+3196 | tier3 | 36 | 2 | 0.053 | 32 | 21 | 0.396 |
| 298273 | 17 | 66873661 | 66873661 | G | A | SNP | ABCA8 | ENST00000430352 | -1 | missense | c.4198 | p.R1400W | tier1 | 91 | 0 | 0.000 | 93 | 70 | 0.429 |
| 298273 | 19 | 8436034 | 8436034 | C | T | SNP | ANGPTL4 | ENST00000301455 | 1 | silent | c.756 | p.H252 | tier1 | 91 | 1 | 0.011 | 154 | 109 | 0.414 |
| 298273 | 19 | 36015866 | 36015866 | C | A | SNP | SBSN | ENST00000518157 | -1 | intronic | c.610-40 | e3-40 | tier3 | 66 | 0 | 0.000 | 74 | 68 | 0.479 |
| 298273 | 20 | 1106550 | 1106550 | G | A | SNP | PSMF1 | ENST00000333082 | 1 | intronic | c.282+257 | e2+257 | tier3 | 34 | 0 | 0.000 | 66 | 68 | 0.508 |
| 298273 | 21 | 10934043 | 10934043 | G | A | SNP | TPTE | ENST00000361285 | -1 | silent | c.934 | p.L312 | tier1 | 244 | 0 | 0.000 | 810 | 126 | 0.135 |
| 298273 | 21 | 43533838 | 43533838 | G | A | SNP | UMODL1 | ENST00000408989 | -1 | missense | c.2644 | p.V882M | tier1 | 33 | 0 | 0.000 | 47 | 48 | 0.505 |
| 298273 | 22 | 20710809 | 20710809 | C | G | SNP | FAM230A | ENST00000434783 | 1 | silent | c.2541 | p.T847 | tier1 | 156 | 1 | 0.006 | 394 | 31 | 0.073 |
| 298273 | X | 22196368 | 22196370 | TGT | - | DEL | PHEX | ENST00000379374 | 1 | intronic | c.1483-22_1483-20 | e14-20 | tier2 | 24 | 0 | 0.000 | 18 | 49 | 0.731 |
| 298273 | X | 135761723 | 135761724 | - | A | INS | ARHGEF6 | ENST00000250617 | -1 | frame_shift_ins | c.1801_1800 | p.R600fs | tier1 | 34 | 0 | 0.000 | 28 | 83 | 0.748 |
| 307995 | 1 | 34052117 | 34052117 | G | A | SNP | CSMD2 | ENST00000373381 | -1 | silent | c.7038 | p.P2346 | tier1 | 82 | 0 | 0.000 | 212 | 48 | 0.185 |
| 307995 | 1 | 121137613 | 121137613 | T | A | SNP | SRGAP2-AS1 | ENST00000437515 | -1 | intronic | c.NULL | NULL | tier3 | 47 | 1 | 0.019 | 67 | 17 | 0.185 |
| 307995 | 1 | 159174836 | 159174836 | C | T | SNP | DARC | ENST00000368122 | 1 | intronic | c.211+66 | e1+66 | tier2 | 35 | 0 | 0.000 | 95 | 20 | 0.172 |
| 307995 | 1 | 207218940 | 207218940 | G | T | SNP | YOD1 | ENST00000315927 | -1 | 3_prime_untranslated_region | c.*3425 | NULL | tier3 | 66 | 0 | 0.000 | 123 | 18 | 0.126 |
| 307995 | 2 | 27325149 | 27325149 | C | G | SNP | CGREF1 | ENST00000467539 | -1 | 5_prime_untranslated_region | c.-27325149 | NULL | tier2 | 30 | 0 | 0.000 | 82 | 25 | 0.234 |
| 307995 | 2 | 43986084 | 43986084 | A | G | SNP | PLEKHH2 | ENST00000490038 | 1 | missense | c.253 | p.T85A | tier1 | 41 | 0 | 0.000 | 135 | 23 | 0.144 |
| 307995 | 2 | 61510631 | 61510631 | T | C | SNP | USP34 | ENST00000398571 | -1 | intronic | c.4833-47 | e36-47 | tier3 | 103 | 0 | 0.000 | 179 | 43 | 0.193 |
| 307995 | 2 | 138738953 | 138738953 | C | T | SNP | HNMT | ENST00000329366 | 1 | silent | c.358 | p.L120 | tier1 | 84 | 0 | 0.000 | 171 | 37 | 0.178 |
| 307995 | 2 | 207310122 | 207310122 | T | C | SNP | ADAM23 | ENST00000264377 | 1 | silent | c.306 | p.S102 | tier1 | 45 | 0 | 0.000 | 97 | 31 | 0.242 |
| 307995 | 3 | 9517257 | 9517257 | G | T | SNP | SETD5 | ENST00000402198 | 1 | nonsense | c.3811 | p.G1271* | tier1 | 32 | 0 | 0.000 | 84 | 27 | 0.243 |
| 307995 | 3 | 30702951 | 30702951 | G | A | SNP | TGFB2 | ENST00000359013 | 1 | intronic | c.530-10179 | e5-10179 | tier4 | 65 | 0 | 0.000 | 98 | 17 | 0.148 |
| 307995 | 3 | 126180517 | 126180517 | G | A | SNP | ZXDC | ENST00000389709 | -1 | missense | c.1988 | p.P663L | tier1 | 27 | 0 | 0.000 | 58 | 24 | 0.293 |
| 307995 | 4 | 107154095 | 107154095 | C | A | SNP | TBCK | ENST00000273980 | -1 | missense | c.1639 | p.G547C | tier1 | 59 | 1 | 0.017 | 67 | 15 | 0.183 |
| 307995 | 5 | 5235176 | 5235176 | C | T | SNP | ADAMTS16 | ENST00000274181 | 1 | missense | c.1900 | p.L634F | tier1 | 69 | 0 | 0.000 | 250 | 47 | 0.158 |
| 307995 | 5 | 14364017 | 14364017 | C | A | SNP | TRIO | ENST00000344204 | 1 | silent | c.2568 | p.V856 | tier1 | 69 | 0 | 0.000 | 118 | 34 | 0.222 |
| 307995 | 5 | 140174742 | 140174742 | C | T | SNP | PCDHA2 | ENST00000526136 | 1 | missense | c.193 | p.R65W | tier1 | 338 | 0 | 0.000 | 906 | 127 | 0.123 |
| 307995 | 6 | 32713784 | 32713784 | C | A | SNP | HLA-DQA2 | ENST00000374940 | 1 | missense | c.548 | p.A183D | tier1 | 137 | 5 | 0.035 | 371 | 81 | 0.178 |
| 307995 | 6 | 38743672 | 38743672 | G | A | SNP | DNAH8 | ENST00000359357 | 1 | missense | c.1256 | p.R419K | tier1 | 48 | 0 | 0.000 | 155 | 27 | 0.148 |
| 307995 | 7 | 29689532 | 29689532 | T | A | SNP | ZNRF2P2 | ENST00000442865 | -1 | intronic | c.NULL | NULL | tier2 | 69 | 0 | 0.000 | 187 | 49 | 0.206 |
| 307995 | 7 | 98639905 | 98639905 | T | - | DEL | SMURF1 | ENST00000361125 | -1 | intronic | c.1335-50 | e13-50 | tier4 | 29 | 0 | 0.000 | 53 | 16 | 0.232 |
| 307995 | 9 | 38414100 | 38414100 | C | T | SNP | IGFBPL1 | ENST00000377694 | -1 | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-----------|----|-----|--------------|-----------------|----|-----------------------------|-----------------|----------------------|-------|------|----|-------|------|-----|-------|
| 307995 | 12 | 9095076 | 9095076 | C | G | SNP | M6PR | ENST00000000412 | -1 | missense | c.647 | p.G216A | tier1 | 57 | 0 | 0.000 | 129 | 20 | 0.133 |
| 307995 | 12 | 50027356 | 50027356 | G | A | SNP | PRPF40B | ENST00000548825 | 1 | intronic | c.580+26 | e8+26 | tier3 | 496 | 0 | 0.000 | 1312 | 317 | 0.194 |
| 307995 | 12 | 52715016 | 52715016 | G | T | SNP | KRT83 | ENST00000293670 | -1 | missense | c.104 | p.A35D | tier1 | 102 | 0 | 0.000 | 219 | 73 | 0.249 |
| 307995 | 12 | 52891238 | 52891238 | T | A | SNP | KRT6C | ENST00000252250 | -1 | 5_prime_flanking_region | c.-23717 | NULL | tier3 | 185 | 0 | 0.000 | 390 | 106 | 0.213 |
| 307995 | 12 | 109519720 | 109519720 | C | T | SNP | USP30 | ENST00000257548 | 1 | intronic | c.781-18 | e9-18 | tier3 | 44 | 0 | 0.000 | 120 | 22 | 0.155 |
| 307995 | 14 | 19378051 | 19378051 | G | C | SNP | OR11H12 | ENST00000550708 | 1 | missense | c.458 | p.G153A | tier1 | 1326 | 0 | 0.000 | 3626 | 167 | 0.044 |
| 307995 | 14 | 94945375 | 94945375 | G | A | SNP | SERPINA9 | ENST00000539349 | -1 | intronic | c.NULL | NULL | tier3 | 133 | 0 | 0.000 | 261 | 54 | 0.170 |
| 307995 | 15 | 45725117 | 45725118 | - | T | INS | C15orf48 | ENST00000344300 | 1 | intronic | c.184-29_184-28 | e4-28 | tier3 | 73 | 1 | 0.014 | 114 | 20 | 0.149 |
| 307995 | 16 | 49315688 | 49315688 | G | T | SNP | CBLN1 | ENST00000219197 | -1 | 5_prime_untranslated_region | c.-312 | NULL | tier4 | 13 | 0 | 0.000 | 34 | 23 | 0.404 |
| 307995 | 17 | 61986092 | 61986092 | G | C | SNP | GH2 | ENST00000332800 | -1 | 5_prime_flanking_region | c.-26931 | NULL | tier3 | 135 | 0 | 0.000 | 233 | 57 | 0.196 |
| 307995 | 18 | 50683690 | 50683690 | C | T | SNP | DCC | ENST00000442544 | 1 | intronic | c.1262-36 | e8-36 | tier3 | 94 | 0 | 0.000 | 153 | 39 | 0.203 |
| 307995 | 20 | 47247205 | 47247205 | C | T | SNP | PREX1 | ENST00000371941 | -1 | intronic | c.4593+61 | e36+61 | tier3 | 42 | 0 | 0.000 | 147 | 28 | 0.160 |
| 307995 | 20 | 56082765 | 56082765 | C | T | SNP | CTCF1 | ENST00000432255 | -1 | silent | c.1326 | p.P442 | tier1 | 54 | 0 | 0.000 | 105 | 31 | 0.226 |
| 307995 | 20 | 58349240 | 58349240 | G | A | SNP | PHACTR3 | ENST00000371015 | 1 | intronic | c.927-58 | e7-58 | tier3 | 83 | 0 | 0.000 | 159 | 37 | 0.189 |
| 307995 | 21 | 36252994 | 36252995 | - | CC | INS | RUNX1 | ENST00000300305 | -1 | frame_shift_ins | c.368_367 | p.D123fs | tier1 | 833 | 0 | 0.000 | 1991 | 374 | 0.158 |
| 307995 | 22 | 20171118 | 20171118 | C | T | SNP | LINC00896 | ENST00000423736 | 1 | 5_prime_flanking_region | c.-22982 | NULL | tier3 | 27 | 0 | 0.000 | 104 | 38 | 0.268 |
| 307995 | X | 39911641 | 39911641 | C | T | SNP | BCOR | ENST00000378444 | -1 | nonsense | c.4989 | p.W1663* | tier1 | 189 | 0 | 0.000 | 371 | 135 | 0.266 |
| 307995 | X | 53574723 | 53574723 | G | T | SNP | HUWE1 | ENST00000262854 | -1 | missense | c.10547 | p.A3516D | tier1 | 41 | 0 | 0.000 | 87 | 36 | 0.293 |
| 307995 | X | 151336800 | 151336800 | C | T | SNP | GABRA3 | ENST00000370314 | -1 | missense | c.1379 | p.R460H | tier1 | 75 | 0 | 0.000 | 158 | 62 | 0.282 |
| 311426 | 1 | 15987264 | 15987264 | T | C | SNP | RSC1A1 | ENST00000345034 | 1 | silent | c.901 | p.L301 | tier1 | 31 | 0 | 0.000 | 54 | 41 | 0.432 |
| 311426 | 1 | 23660085 | 23660085 | T | C | SNP | HNRNPR | ENST00000374616 | -1 | missense | c.424 | p.T142A | tier1 | 74 | 2 | 0.026 | 105 | 69 | 0.397 |
| 311426 | 1 | 26795865 | 26795865 | A | G | SNP | DHDDS | ENST00000360009 | 1 | 3_prime_untranslated_region | c.*243 | NULL | tier3 | 77 | 3 | 0.038 | 69 | 67 | 0.489 |
| 311426 | 1 | 47904000 | 47904000 | G | A | SNP | FOXD2 | ENST00000334793 | 1 | missense | c.193 | p.A65T | tier1 | 6 | 0 | 0.000 | 3 | 9 | 0.750 |
| 311426 | 1 | 211433418 | 211433418 | T | A | SNP | RCOR3 | ENST00000367005 | 1 | 5_prime_untranslated_region | c.-40 | NULL | tier4 | 65 | 1 | 0.015 | 58 | 38 | 0.392 |
| 311426 | 3 | 158380318 | 158380318 | C | T | SNP | GFM1 | ENST00000486715 | 1 | intronic | c.1324-99 | e11-99 | tier3 | 28 | 1 | 0.035 | 34 | 21 | 0.382 |
| 311426 | 5 | 228288 | 228288 | C | T | SNP | SDHA | ENST00000514027 | 1 | 3_prime_untranslated_region | c.*228288 | NULL | tier3 | 161 | 0 | 0.000 | 154 | 10 | 0.061 |
| 311426 | 5 | 68849514 | 68849514 | T | G | SNP | OCLN | ENST00000514370 | 1 | rna | NULL | NULL | tier1 | 72 | 3 | 0.040 | 104 | 88 | 0.458 |
| 311426 | 5 | 108691731 | 108691731 | A | - | DEL | PJA2 | ENST00000361189 | -1 | splice_region_del | c.1653-4 | e6-4 | tier3 | 35 | 0 | 0.000 | 57 | 17 | 0.230 |
| 311426 | 5 | 118824832 | 118824832 | G | A | SNP | HSD17B4 | ENST00000256216 | 1 | intronic | c.623-55 | e9-55 | tier3 | 55 | 2 | 0.035 | 52 | 33 | 0.388 |
| 311426 | 5 | 156659265 | 156659265 | A | G | SNP | ITK | ENST00000422843 | 1 | intronic | c.714-85 | e8-85 | tier3 | 51 | 2 | 0.038 | 59 | 42 | 0.416 |
| 311426 | 6 | 43481400 | 43481400 | A | G | SNP | YIPF3 | ENST00000372417 | -1 | 5_prime_untranslated_region | c.-43481400 | NULL | tier3 | 20 | 0 | 0.000 | 30 | 17 | 0.362 |
| 311426 | 6 | 75798722 | 75798722 | C | - | DEL | COL12A1 | ENST00000322507 | -1 | intronic | c.9010+100 | e63+100 | tier4 | 132 | 2 | 0.015 | 118 | 14 | 0.106 |
| 311426 | 6 | 96651060 | 96651060 | G | A | SNP | FUT9 | ENST00000302103 | 1 | missense | c.29 | p.R10H | tier1 | 66 | 0 | 0.000 | 89 | 54 | 0.375 |
| 311426 | 7 | 100643736 | 100643736 | T | C | SNP | MUC12 | ENST00000536621 | 1 | missense | c.9892 | p.S3298P | tier1 | 462 | 5 | 0.011 | 756 | 61 | 0.075 |
| 311426 | 7 | 120390342 | 120390342 | C | A | SNP | KCND2 | ENST00000331113 | 1 | 3_prime_untranslated_region | c.*2430 | NULL | tier2 | 30 | 0 | 0.000 | 30 | 20 | 0.400 |
| 311426 | 7 | 141791778 | 141791778 | G | A | SNP | MGAM | ENST00000475668 | 1 | silent | c.6876 | p.R2292 | tier1 | 142 | 5 | 0.034 | 102 | 73 | 0.417 |
| 311426 | 7 | 151945554 | 151945554 | G | A | SNP | KMT2C | ENST00000355193 | -1 | silent | c.1965 | p.V655 | tier1 | 318 | 17 | 0.051 | 467 | 332 | 0.416 |
| 311426 | 8 | 2857551 | 2857551 | C | T | SNP | CSMD1 | ENST00000520002 | -1 | missense | c.8135 | p.R2712Q | tier1 | 703 | 23 | 0.032 | 607 | 490 | 0.446 |
| 311426 | 9 | 35301169 | 35301169 | T | C | SNP | UNC13B | ENST00000378496 | 1 | intronic | c.761+5242 | e8+5242 | tier3 | 57 | 2 | 0.034 | 95 | 56 | 0.371 |
| 311426 | 9 | 113086243 | 113086243 | A | G | SNP | TXNDC8 | ENST00000374507 | -1 | silent | c.234 | p.H78 | tier1 | 109 | 5 | 0.044 | 95 | 61 | 0.389 |
| 311426 | 10 | 123727090 | 123727090 | A | T | SNP | NSMCE4A | ENST00000481320 | -1 | 5_prime_untranslated_region | c.-123727090 | NULL | tier3 | 57 | 0 | 0.000 | 81 | 62 | 0.434 |
| 311426 | 11 | 247379 | 247379 | G | A | SNP | PSMD13 | ENST00000431206 | 1 | missense | c.505 | p.G169R | tier1 | 145 | 5 | 0.033 | 152 | 90 | 0.372 |
| 311426 | 11 | 10831943 | 10831944 | - | A | INS | LOC101928053 | ENST00000499765 | 1 | intronic | c.NULL | NULL | tier3 | 31 | 0 | 0.000 | 39 | 23 | 0.371 |
| 311426 | 11 | 75378554 | 75378554 | C | T | SNP | MAP6 | ENST00000304771 | -1 | silent | c.861 | p.R287 | tier1 | 16 | 0 | 0.000 | 2 | 3 | 0.600 |
| 311426 | 11 | 125616169 | 125616169 | G | A | SNP | PATE1 | ENST00000305738 | 1 | 5_prime_flanking_region | c.-31 | NULL | tier3 | 77 | 5 | 0.061 | 71 | 50 | 0.413 |
| 311426 | 12 | 13714497 | 13714497 | C | A | SNP | GRIN2B | ENST00000609686 | -1 | 3_prime_untranslated_region | c.*1220 | NULL | tier3 | 162 | 5 | 0.030 | 127 | 126 | 0.496 |
| 311426 | 12 | 78334060 | 78334060 | C | T | SNP | NAV3 | ENST00000549369 | 1 | rna | NULL | NULL | tier1 | 61 | 2 | 0.032 | 44 | 39 | 0.470 |
| 311426 | 12 | 106853134 | 106853134 | A | T | SNP | POLR3B | ENST00000228347 | 1 | missense | c.2558 | p.D853V | tier1 | 71 | 2 | 0.027 | 89 | 65 | 0.422 |
| 311426 | 13 | 26114392 | 26114392 | T | C | SNP | ATP8A2 | ENST00000381655 | 1 | intronic | c.582-65 | e8-65 | tier3 | 77 | 0 | 0.000 | 67 | 51 | 0.432 |
| 311426 | 14 | 99988655 | 99988655 | G | A | SNP | CCDC85C | ENST00000380243 | -1 | intronic | c.868-78 | e3-78 | tier3 | 68 | 2 | 0.029 | 57 | 47 | 0.452 |
| 311426 | 15 | 48065413 | 48065413 | C | T | SNP | SEMA6D | ENST00000316364 | 1 | 3_prime_untranslated_region | c.*1431 | NULL | tier3 | 48 | 0 | 0.000 | 63 | 42 | 0.400 |
| 311426 | 15 | 72037471 | 72037471 | G | A | SNP | THSD4 | ENST00000261862 | 1 | missense | c.1933 | p.V645M | tier1 | 67 | 4 | 0.056 | 40 | 36 | 0.474 |
| 311426 | 16 | 90162620 | 90162620 | T | G | SNP | TUBB8P7 | ENST00000564451 | 1 | rna | NULL | NULL | tier1 | 190 | 0 | 0.000 | 263 | 15 | 0.054 |
| 311426 | 19 | 16145115 | 16145115 | C | A | SNP | LINC00905 | ENST00000397365 | 1 | intronic | c.NULL | NULL | tier3 | 44 | 1 | 0.022 | 40 | 34 | 0.453 |
| 311426 | 19 | 19767521 | 19767521 | C | T | SNP | ATP13A1 | ENST00000357324 | -1 | missense | c.1031 | p.R344H | tier1 | 34 | 1 | 0.029 | 50 | 32 | 0.390 |
| 311426 | 21 | 44524456 | 44524456 | G | A | SNP | U2AF1 | ENST00000291552 | -1 | missense | c.101 | p.S34F | tier1 | 373 | 17 | 0.044 | 387 | 388 | 0.499 |
| 311426 | X | 138698653 | 138698653 | A | - | DEL | MCF2 | ENST00000370578 | -1 | intronic | c.1435-21 | e12-21 | tier3 | 79 | 1 | 0.013 | 113 | 17 | 0.131 |
| 329866 | 1 | 55129373 | 55129373 | A | C | SNP | MROH7-TTC4 | ENST00000414150 | 1 | intronic | c.1232-1467 | e2-1467 | tier4 | 17 | 0 | 0.000 | 37 | 21 | 0.362 |
| 329866 | 1 | 89615249 | 89615249 | C | T | SNP | GBP7 | ENST00000294671 | -1 | missense | c.878 | p.G293E | tier1 | 256 | 0 | 0.000 | 708 | 287 | 0.288 |
| 329866 | 1 | 244715862 | 244715862 | A | G | SNP | C1orf101 | ENST00000366531 | 1 | missense | c.322 | p.T108A | tier1 | 24 | 0 | 0.000 | 58 | 35 | 0.376 |
| 329866 | 2 | 238436052 | 238436052 | C | T | SNP | MLPH | ENST00000264605 | 1 | silent | c.913 | p.L305 | tier1 | 51 | 0 | 0.000 | 113 | 67 | 0.370 |
| 329866 | 3 | 56037242 | 56037242 | A | - | DEL | ERC2 | ENST00000288221 | -1 | intronic | c.2061+3967 | e9+3967 | tier3 | 24 | 0 | 0.000 | 73 | 38 | 0.342 |
| 329866 | 3 | 75467466 | 75467466 | G | A | SNP | ALG1L6P | ENST00000515248 | 1 | intronic | c.NULL | NULL | tier3 | 84 | 0 | 0.000 | 275 | 25 | 0.083 |
| 329866 | 3 | 112190167 | 112190167 | C | T | SNP | BTLA | ENST00000334529 | -1 | missense | c.439 | p.E147K | tier1 | 65 | 2 | 0.030 | 172 | 107 | 0.382 |
| 329866 | 3 | 127381208 | 127381208 | G | A | SNP | PODXL2 | ENST00000342480 | 1 | intronic | c.1206+54 | e4+54 | tier3 | 21 | 0 | 0.000 | 57 | 32 | 0.356 |
| 329866 | 3 | 132363753 | 132363753 | A | - | DEL | ACAD11 | ENST00000264990 | -1 | intronic | c.150-13 | e2-13 | tier3 | 42 | 0 | 0.000 | 107 | 27 | 0.202 |
| 329866 | 3 | 164700861 | 164700861 | G | A | SNP | SI | ENST00000264382 | -1 | intronic | c.5198-22 | e45-22 | tier3 | 197 | 0 | 0.000 | 347 | 154 | 0.307 |
| 329866 | 4 | 88536147 | 88536155 | ACAGCAGCA | A | DEL | DSPP | ENST00000282478 | 1 | in_frame_del | c.2333_2341 | p.NSS781in_frame_del | tier1 | 107 | 0 | 0.000 | 309 | 28 | 0.083 |
| 329866 | 4 | 104007719 | 104007719 | A | - | DEL | BDH2 | ENST00000296424 | -1 | intronic | c.3 | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-----|------|-----|-----------------|-----------------|----|-----------------------------|-------------------|--------------------|-------|-----|---|-------|------|------|-------|
| 329866 | 8 | 140768860 | 140768860 | A | G | SNP | TRAPPC9 | ENST00000389328 | -1 | intronic | c.3350-24415 | e22-24415 | tier3 | 67 | 0 | 0.000 | 317 | 32 | 0.091 |
| 329866 | 9 | 35076941 | 35076941 | C | T | SNP | FANCG | ENST00000378643 | -1 | intronic | c.777+27 | e6+27 | tier3 | 40 | 0 | 0.000 | 100 | 46 | 0.315 |
| 329866 | 9 | 36204051 | 36204051 | C | G | SNP | CLTA | ENST00000464497 | 1 | intronic | c.374-14 | e4-14 | tier3 | 34 | 0 | 0.000 | 83 | 59 | 0.416 |
| 329866 | 9 | 68433510 | 68433510 | C | G | SNP | ENSG00000215548 | ENST00000376334 | -1 | 5_prime_untranslated_region | c.-68433510 | NULL | tier3 | 83 | 0 | 0.000 | 204 | 26 | 0.113 |
| 329866 | 10 | 112360315 | 112360316 | - | T | INS | SMC3 | ENST00000361804 | -1 | intronic | c.2535+11_2535+10 | e22+11 | tier3 | 135 | 1 | 0.007 | 278 | 26 | 0.086 |
| 329866 | 11 | 20178072 | 20178072 | C | T | SNP | DBX1 | ENST00000227256 | -1 | silent | c.837 | p.K279 | tier1 | 67 | 1 | 0.015 | 203 | 106 | 0.343 |
| 329866 | 12 | 22211533 | 22211533 | C | G | SNP | CMAS | ENST00000229329 | 1 | missense | c.592 | p.P198A | tier1 | 86 | 0 | 0.000 | 187 | 55 | 0.227 |
| 329866 | 12 | 22211534 | 22211534 | C | T | SNP | CMAS | ENST00000229329 | 1 | missense | c.593 | p.P198L | tier1 | 86 | 0 | 0.000 | 189 | 55 | 0.225 |
| 329866 | 12 | 57037304 | 57037304 | T | C | SNP | ATP5B | ENST00000552959 | -1 | missense | c.484 | p.S162G | tier1 | 32 | 0 | 0.000 | 88 | 46 | 0.343 |
| 329866 | 12 | 114832571 | 114832571 | G | A | SNP | TBX5 | ENST00000310346 | -1 | missense | c.638 | p.A213V | tier1 | 43 | 0 | 0.000 | 113 | 55 | 0.327 |
| 329866 | 16 | 50750582 | 50750583 | - | T | INS | NOD2 | ENST00000300589 | 1 | splice_site_ins | c.2546+1_2546+1 | e5+1 | tier1 | 47 | 0 | 0.000 | 120 | 34 | 0.221 |
| 329866 | 16 | 89776108 | 89776108 | G | A | SNP | VPS9D1 | ENST00000389386 | -1 | intronic | c.1388+77 | e11+77 | tier3 | 20 | 0 | 0.000 | 52 | 40 | 0.435 |
| 329866 | 17 | 7577581 | 7577581 | A | C | SNP | TP53 | ENST00000269305 | -1 | missense | c.700 | p.Y234D | tier1 | 658 | 0 | 0.000 | 326 | 1371 | 0.806 |
| 329866 | 17 | 40342300 | 40342300 | C | A | SNP | GHDC | ENST00000436923 | -1 | missense | c.1366 | p.V456F | tier1 | 63 | 0 | 0.000 | 88 | 44 | 0.331 |
| 329866 | 19 | 7796670 | 7796670 | G | - | DEL | CLEC4G | ENST00000328853 | -1 | intronic | c.56-13 | e2-13 | tier3 | 25 | 0 | 0.000 | 49 | 27 | 0.355 |
| 329866 | 20 | 29623180 | 29623180 | T | C | SNP | FRG1B | ENST00000482423 | 1 | rna | NULL | NULL | tier1 | 618 | 3 | 0.005 | 2127 | 56 | 0.026 |
| 329866 | 20 | 62326771 | 62326771 | G | C | SNP | RTEL1 | ENST00000318100 | 1 | missense | c.3590 | p.G1197A | tier1 | 188 | 0 | 0.000 | 385 | 174 | 0.311 |
| 329866 | 21 | 40823790 | 40823790 | C | T | SNP | SH3BGR | ENST00000333634 | 1 | 5_prime_untranslated_region | c.-44 | NULL | tier3 | 44 | 0 | 0.000 | 121 | 69 | 0.363 |
| 340060 | 1 | 115258747 | 115258747 | C | T | SNP | NRAS | ENST00000369535 | -1 | missense | c.35 | p.G12D | tier1 | 534 | 5 | 0.009 | 614 | 32 | 0.050 |
| 340060 | 1 | 207888661 | 207888661 | T | C | SNP | CR1L | ENST00000294997 | 1 | nonstop | c.1165 | p.*389R | tier1 | 223 | 0 | 0.000 | 354 | 16 | 0.043 |
| 340060 | 2 | 55808823 | 55808825 | AAG | - | DEL | SMEK2 | ENST00000272313 | -1 | in_frame_del | c.1245_1243 | p.L415in_frame_del | tier1 | 44 | 1 | 0.022 | 86 | 26 | 0.232 |
| 340060 | 2 | 105897181 | 105897181 | C | A | SNP | TGFBRAP1 | ENST00000258449 | -1 | splice_site | c.1122-1 | e5-1 | tier1 | 42 | 1 | 0.023 | 55 | 29 | 0.345 |
| 340060 | 3 | 58368237 | 58368237 | A | T | SNP | PXK | ENST00000356151 | 1 | splice_region | c.202-4 | e4-4 | tier3 | 50 | 0 | 0.000 | 88 | 38 | 0.302 |
| 340060 | 4 | 24422954 | 24422954 | A | T | SNP | ENSG00000222262 | ENST00000410330 | -1 | 5_prime_flanking_region | c.-30 | NULL | tier4 | 70 | 1 | 0.014 | 162 | 75 | 0.314 |
| 340060 | 4 | 141786725 | 141786725 | A | T | SNP | RNF150 | ENST00000515673 | -1 | 3_prime_untranslated_region | c.*2690 | NULL | tier3 | 58 | 0 | 0.000 | 133 | 23 | 0.147 |
| 340060 | 5 | 31267727 | 31267727 | C | A | SNP | CDH6 | ENST00000265071 | 1 | missense | c.147 | p.N49K | tier1 | 102 | 0 | 0.000 | 164 | 13 | 0.073 |
| 340060 | 5 | 122941019 | 122941019 | G | T | SNP | C5NK1G3 | ENST00000360683 | 1 | intronic | c.1290-14 | e12-14 | tier3 | 78 | 0 | 0.000 | 134 | 21 | 0.132 |
| 340060 | 5 | 134063500 | 134063500 | A | - | DEL | SEC24A | ENST00000398844 | 1 | 3_prime_untranslated_region | c.*2716 | NULL | tier3 | 55 | 0 | 0.000 | 163 | 21 | 0.114 |
| 340060 | 6 | 108882438 | 108882438 | C | T | SNP | FOXO3 | ENST00000343882 | 1 | silent | c.27 | p.A9 | tier1 | 18 | 0 | 0.000 | 5 | 5 | 0.500 |
| 340060 | 7 | 43548582 | 43548582 | G | A | SNP | HECW1 | ENST00000395891 | 1 | missense | c.3881 | p.G1294D | tier1 | 665 | 1 | 0.002 | 881 | 43 | 0.047 |
| 340060 | 9 | 139997711 | 139997711 | T | G | SNP | MAN1B1 | ENST00000536349 | 1 | 3_prime_untranslated_region | c.*139997711 | NULL | tier3 | 118 | 1 | 0.008 | 162 | 18 | 0.099 |
| 340060 | 10 | 112356156 | 112356156 | G | T | SNP | SMC3 | ENST00000361804 | 1 | missense | c.1964 | p.G655V | tier1 | 434 | 1 | 0.002 | 732 | 95 | 0.115 |
| 340060 | 11 | 32429666 | 32429666 | G | A | SNP | WT1 | ENST00000332351 | -1 | intronic | c.1002-8076 | e6-8076 | tier4 | 6 | 0 | 0.000 | 6 | 12 | 0.667 |
| 340060 | 12 | 43827516 | 43827516 | C | A | SNP | ADAMTS20 | ENST00000389420 | -1 | intronic | c.2720+532 | e19+532 | tier4 | 52 | 0 | 0.000 | 63 | 16 | 0.203 |
| 340060 | 12 | 72067980 | 72067980 | A | T | SNP | THAP2 | ENST00000308086 | 1 | splice_region | c.72-3 | e2-3 | tier2 | 67 | 0 | 0.000 | 136 | 15 | 0.098 |
| 340060 | 13 | 28592642 | 28592642 | C | A | SNP | FLT3 | ENST00000380982 | -1 | missense | c.2503 | p.D835Y | tier1 | 457 | 1 | 0.002 | 757 | 43 | 0.054 |
| 340060 | 14 | 58038541 | 58038541 | A | C | SNP | SLC35F4 | ENST00000339762 | -1 | intronic | c.1041+71 | e5+71 | tier3 | 41 | 0 | 0.000 | 80 | 15 | 0.158 |
| 340060 | 15 | 85805938 | 85805938 | G | A | SNP | ENSG00000218052 | ENST00000561209 | -1 | 5_prime_untranslated_region | c.-85805938 | NULL | tier3 | 124 | 0 | 0.000 | 210 | 16 | 0.071 |
| 340060 | 16 | 15227776 | 15227776 | G | A | SNP | PKD1P6 | ENST00000540502 | -1 | rna | NULL | NULL | tier1 | 49 | 1 | 0.020 | 63 | 29 | 0.315 |
| 340060 | 17 | 61995194 | 61995194 | C | G | SNP | GH1 | ENST00000323322 | -1 | missense | c.382 | p.V128L | tier1 | 96 | 0 | 0.000 | 164 | 23 | 0.122 |
| 340060 | 18 | 30908069 | 30908069 | T | G | SNP | CCDC178 | ENST00000406524 | -1 | intronic | c.880-4472 | e9-4472 | tier4 | 74 | 0 | 0.000 | 323 | 31 | 0.088 |
| 346313 | 1 | 24294362 | 24294362 | A | T | SNP | SRSF10 | ENST00000374452 | -1 | 3_prime_untranslated_region | c.*54 | NULL | tier4 | 117 | 4 | 0.033 | 55 | 14 | 0.203 |
| 346313 | 1 | 89353241 | 89353241 | G | C | SNP | GTF2B | ENST00000370500 | -1 | intronic | c.18-191 | e2-191 | tier3 | 58 | 0 | 0.000 | 34 | 155 | 0.816 |
| 346313 | 1 | 115258744 | 115258744 | C | T | SNP | NRAS | ENST00000369535 | -1 | missense | c.38 | p.G13D | tier1 | 456 | 2 | 0.004 | 520 | 253 | 0.327 |
| 346313 | 1 | 159038320 | 159038320 | T | - | DEL | AIM2 | ENST00000368130 | -1 | intronic | c.396+38 | e2+38 | tier3 | 100 | 0 | 0.000 | 114 | 63 | 0.356 |
| 346313 | 1 | 185815276 | 185815276 | G | A | SNP | HMCN1 | ENST00000271588 | 1 | intronic | c.339+48 | e2+48 | tier3 | 319 | 1 | 0.003 | 437 | 286 | 0.395 |
| 346313 | 1 | 237494338 | 237494338 | G | A | SNP | RYR2 | ENST00000360064 | 1 | intronic | c.270+52 | e4+52 | tier3 | 130 | 0 | 0.000 | 139 | 94 | 0.403 |
| 346313 | 2 | 79914651 | 79914651 | C | T | SNP | CTNNA2 | ENST00000361291 | 1 | intronic | c.204+35867 | e2+35867 | tier3 | 116 | 2 | 0.017 | 124 | 57 | 0.313 |
| 346313 | 2 | 95976762 | 95976762 | G | A | SNP | KCNIP3 | ENST00000475491 | 1 | 3_prime_untranslated_region | c.*95976762 | NULL | tier3 | 43 | 0 | 0.000 | 37 | 21 | 0.362 |
| 346313 | 2 | 97493997 | 97493997 | C | T | SNP | CNNM3 | ENST00000465224 | 1 | 3_prime_untranslated_region | c.*97493997 | NULL | tier3 | 35 | 0 | 0.000 | 42 | 16 | 0.276 |
| 346313 | 2 | 103148926 | 103148926 | G | A | SNP | SLC9A4 | ENST00000295269 | 1 | missense | c.2176 | p.G726S | tier1 | 87 | 2 | 0.022 | 103 | 65 | 0.376 |
| 346313 | 2 | 130796690 | 130796690 | C | T | SNP | LOC440905 | ENST00000325390 | -1 | intronic | c.NULL | NULL | tier3 | 89 | 1 | 0.011 | 197 | 27 | 0.121 |
| 346313 | 2 | 198267359 | 198267359 | C | A | SNP | SF3B1 | ENST00000335508 | -1 | missense | c.1998 | p.K666N | tier1 | 739 | 6 | 0.008 | 786 | 514 | 0.395 |
| 346313 | 3 | 124180873 | 124180873 | A | G | SNP | KALRN | ENST00000360013 | 1 | intronic | c.3915+70 | e24+70 | tier3 | 108 | 0 | 0.000 | 127 | 74 | 0.366 |
| 346313 | 3 | 157363624 | 157363624 | A | T | SNP | C3orf55 | ENST00000461040 | 1 | nonsense | c.151 | p.K51* | tier1 | 111 | 2 | 0.018 | 117 | 56 | 0.324 |
| 346313 | 3 | 185362401 | 185362401 | C | T | SNP | IGF2BP2 | ENST00000382199 | -1 | 3_prime_untranslated_region | c.*918 | NULL | tier3 | 45 | 0 | 0.000 | 32 | 23 | 0.397 |
| 346313 | 4 | 1389054 | 1389055 | - | CA | INS | CRIPAK | ENST00000324803 | 1 | frame_shift_ins | c.755_756 | p.C254fs | tier1 | 107 | 1 | 0.009 | 126 | 20 | 0.137 |
| 346313 | 4 | 47888007 | 47888007 | T | C | SNP | NFXL1 | ENST00000381538 | -1 | missense | c.1553 | p.Y518C | tier1 | 75 | 0 | 0.000 | 64 | 41 | 0.387 |
| 346313 | 5 | 128365287 | 128365287 | G | A | SNP | SLC27A6 | ENST00000262462 | 1 | missense | c.1570 | p.G524R | tier1 | 53 | 0 | 0.000 | 196 | 108 | 0.354 |
| 346313 | 5 | 170837545 | 170837546 | - | TGTT | INS | NPM1 | ENST00000296930 | 1 | frame_shift_ins | c.861_862 | p.W287fs | tier1 | 229 | 0 | 0.000 | 641 | 247 | 0.278 |
| 346313 | 5 | 180659955 | 180659955 | C | T | SNP | TRIM41 | ENST00000515223 | 1 | 3_prime_untranslated_region | c.*180659955 | NULL | tier3 | 69 | 0 | 0.000 | 77 | 38 | 0.330 |
| 346313 | 7 | 112077307 | 112077307 | A | C | SNP | IFRD1 | ENST00000443101 | 1 | 5_prime_untranslated_region | c.-18567 | NULL | tier4 | 94 | 0 | 0.000 | 129 | 100 | 0.435 |
| 346313 | 7 | 141794384 | 141794384 | G | A | SNP | MGAM | ENST00000549489 | 1 | intronic | c.4619-36 | e38-36 | tier3 | 46 | 4 | 0.080 | 27 | 66 | 0.710 |
| 346313 | 8 | 33356644 | 33356644 | T | C | SNP | TTI2 | ENST00000360742 | -1 | 3_prime_untranslated_region | c.*47 | NULL | tier3 | 98 | 0 | 0.000 | 120 | 55 | 0.314 |
| 346313 | 8 | 39111964 | 39111964 | C | T | SNP | ADAM32 | ENST00000379907 | 1 | missense | c.1934 | p.S645L | tier1 | 142 | 0 | 0.000 | 139 | 103 | 0.426 |
| 346313 | 8 | 56362027 | 56362027 | G | A | SNP | XKR4 | ENST00000327381 | 1 | intronic | c.1007-73813 | e3-73813 | tier2 | 413 | 0 | 0.000 | 331 | 281 | 0.456 |
| 346313 | 9 | 2058252 | 2058252 | A | T | SNP | SMARCA2 | ENST00000349721 | 1 | intronic | c.1348-39 | e7-39 | tier3 | 91 | 0 | 0.000 | 131 | 54 | 0.292 |
| 346313 | 9 | 84548900 | 84548900 | C | A | SNP | SPATA31D4 | ENST00000341875 | 1 | rna | NULL | NULL | tier1 | 75 | 0 | 0.000 | 86 | 57 | 0.399 |
| | | | | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|---|----|-----|-----------------|-----------------|----|-----------------------------|---------------|----------|-------|------|---|-------|------|-----|-------|
| 346313 | 11 | 74880778 | 74880778 | C | T | SNP | SLCO2B1 | ENST00000289575 | 1 | silent | c.750 | p.R250 | tier1 | 50 | 0 | 0.000 | 39 | 35 | 0.473 |
| 346313 | 12 | 8073897 | 8073897 | G | A | SNP | SLC2A3 | ENST00000469295 | -1 | 5_prime_untranslated_region | c.-8073897 | NULL | tier3 | 135 | 1 | 0.007 | 213 | 18 | 0.078 |
| 346313 | 12 | 22606832 | 22606832 | G | C | SNP | C2CD5 | ENST00000333957 | -1 | missense | c.2869 | p.Q957E | tier1 | 84 | 0 | 0.000 | 99 | 59 | 0.373 |
| 346313 | 12 | 112926910 | 112926910 | G | C | SNP | PTPN11 | ENST00000351677 | 1 | missense | c.1530 | p.Q510H | tier1 | 758 | 0 | 0.000 | 1191 | 33 | 0.027 |
| 346313 | 12 | 133384599 | 133384599 | C | T | SNP | GOLGA3 | ENST00000204726 | -1 | silent | c.1056 | p.A352 | tier1 | 102 | 0 | 0.000 | 85 | 50 | 0.368 |
| 346313 | 13 | 95363388 | 95363388 | G | A | SNP | SOX21 | ENST00000376945 | -1 | 3_prime_untranslated_region | c.*85 | NULL | tier2 | 52 | 0 | 0.000 | 53 | 34 | 0.391 |
| 346313 | 13 | 99030086 | 99030086 | C | T | SNP | FARP1 | ENST00000376586 | 1 | missense | c.410 | p.A137V | tier1 | 64 | 0 | 0.000 | 69 | 36 | 0.343 |
| 346313 | 14 | 38274022 | 38274022 | A | T | SNP | TTC6 | ENST00000478811 | 1 | splice_region | c.357+3 | e3+3 | tier3 | 167 | 1 | 0.006 | 191 | 110 | 0.365 |
| 346313 | 14 | 70442537 | 70442537 | G | A | SNP | SMOC1 | ENST00000361956 | 1 | splice_region | c.478+6 | e4+6 | tier2 | 107 | 0 | 0.000 | 94 | 44 | 0.319 |
| 346313 | 14 | 101429460 | 101429460 | A | T | SNP | SNORD114-7 | ENST00000362520 | 1 | rna | NULL | NULL | tier1 | 30 | 0 | 0.000 | 67 | 41 | 0.380 |
| 346313 | 15 | 92397297 | 92397298 | - | GT | INS | SLCO3A1 | ENST00000318445 | 1 | frame_shift_ins | c.159_160 | p.G53fs | tier1 | 25 | 0 | 0.000 | 34 | 14 | 0.292 |
| 346313 | 16 | 790001 | 790001 | G | A | SNP | NARFL | ENST00000540986 | -1 | 5_prime_untranslated_region | c.-3603 | NULL | tier3 | 27 | 0 | 0.000 | 47 | 34 | 0.420 |
| 346313 | 17 | 45664777 | 45664777 | T | C | SNP | NPEPPS | ENST00000322157 | 1 | intronic | c.1095+67 | e9+67 | tier3 | 88 | 0 | 0.000 | 104 | 61 | 0.370 |
| 346313 | 19 | 35991219 | 35991219 | T | C | SNP | DMKN | ENST00000469960 | -1 | 5_prime_untranslated_region | c.-35991219 | NULL | tier3 | 50 | 0 | 0.000 | 51 | 27 | 0.346 |
| 346313 | 19 | 38782452 | 38782452 | C | T | SNP | SPINT2 | ENST00000585357 | 1 | 3_prime_untranslated_region | c.*38782452 | NULL | tier3 | 72 | 0 | 0.000 | 73 | 34 | 0.315 |
| 346313 | 20 | 29633902 | 29633902 | C | A | SNP | FRG1B | ENST00000278882 | 1 | missense | c.541 | p.P181T | tier1 | 95 | 0 | 0.000 | 537 | 41 | 0.071 |
| 346313 | 21 | 11039295 | 11039295 | C | G | SNP | BAGE2 | ENST00000470054 | -1 | rna | NULL | NULL | tier1 | 2708 | 1 | 0.000 | 5435 | 378 | 0.065 |
| 346313 | 21 | 45744779 | 45744779 | T | G | SNP | PFKL | ENST00000403390 | 1 | missense | c.1997 | p.I666S | tier1 | 85 | 0 | 0.000 | 61 | 31 | 0.333 |
| 346313 | X | 19037686 | 19037686 | A | T | SNP | GPR64 | ENST00000479496 | -1 | 5_prime_untranslated_region | c.-19037686 | NULL | tier4 | 46 | 0 | 0.000 | 76 | 15 | 0.163 |
| 346313 | X | 123164976 | 123164977 | - | T | INS | STAG2 | ENST00000218089 | 1 | splice_site_ins | c.288+1_288+1 | e3+1 | tier1 | 293 | 0 | 0.000 | 142 | 477 | 0.771 |
| 346313 | Y | 28654460 | 28654460 | C | T | SNP | TPTE2P4 | ENST00000258589 | 1 | intronic | c.NULL | NULL | tier3 | 84 | 0 | 0.000 | 39 | 102 | 0.713 |
| 350719 | 17 | 7577568 | 7577568 | C | T | SNP | TP53 | ENST00000269305 | -1 | missense | c.713 | p.C238Y | tier1 | 494 | 0 | 0.000 | 1426 | 85 | 0.056 |
| 350719 | 18 | 15272098 | 15272098 | C | A | SNP | BNIP3P3 | ENST00000577336 | -1 | 5_prime_flanking_region | c.-24102 | NULL | tier3 | 72 | 2 | 0.027 | 367 | 71 | 0.162 |
| 350719 | 19 | 46214702 | 46214702 | A | T | SNP | FBXO46 | ENST00000317683 | -1 | 3_prime_untranslated_region | c.*240 | NULL | tier2 | 87 | 1 | 0.011 | 214 | 28 | 0.114 |
| 353053 | 1 | 20881411 | 20881412 | - | C | INS | FAM43B | ENST00000332947 | 1 | 3_prime_untranslated_region | c.*955 | NULL | tier3 | 45 | 0 | 0.000 | 87 | 18 | 0.171 |
| 353053 | 1 | 20981993 | 20981993 | A | G | SNP | DDOST | ENST00000375048 | -1 | missense | c.542 | p.L181P | tier1 | 43 | 0 | 0.000 | 75 | 16 | 0.176 |
| 353053 | 1 | 196887311 | 196887311 | T | C | SNP | CFHR4 | ENST00000367416 | 1 | intronic | c.1538-29 | e10-29 | tier2 | 68 | 0 | 0.000 | 83 | 11 | 0.117 |
| 353053 | 1 | 205801794 | 205801794 | G | A | SNP | PM20D1 | ENST00000367136 | -1 | missense | c.1217 | p.A406V | tier1 | 165 | 0 | 0.000 | 256 | 29 | 0.102 |
| 353053 | 1 | 222833276 | 222833276 | A | G | SNP | MIA3 | ENST00000344441 | 1 | missense | c.4903 | p.M1635V | tier1 | 62 | 0 | 0.000 | 98 | 19 | 0.162 |
| 353053 | 5 | 14713807 | 14713807 | G | A | SNP | ANKH | ENST00000284268 | -1 | intronic | c.1142-31 | e10-31 | tier3 | 56 | 0 | 0.000 | 97 | 20 | 0.171 |
| 353053 | 7 | 139094357 | 139094357 | C | T | SNP | LUC7L2 | ENST00000541515 | 1 | nonsense | c.934 | p.R312* | tier1 | 449 | 0 | 0.000 | 741 | 63 | 0.078 |
| 353053 | 9 | 135073547 | 135073547 | C | T | SNP | NTNG2 | ENST00000360670 | 1 | silent | c.408 | p.D136 | tier1 | 88 | 0 | 0.000 | 111 | 11 | 0.090 |
| 353053 | 11 | 75141266 | 75141266 | C | T | SNP | KLHL35 | ENST00000539798 | -1 | missense | c.409 | p.G137S | tier1 | 6 | 0 | 0.000 | 5 | 12 | 0.706 |
| 353053 | 12 | 102291646 | 102291646 | C | T | SNP | DRAM1 | ENST00000258534 | 1 | missense | c.137 | p.T46M | tier1 | 85 | 0 | 0.000 | 145 | 15 | 0.094 |
| 353053 | 12 | 102576308 | 102576308 | T | - | DEL | PARPBP | ENST00000358383 | 1 | intronic | c.1185-19 | e8-19 | tier3 | 55 | 1 | 0.018 | 112 | 23 | 0.170 |
| 353053 | 13 | 98634738 | 98634738 | T | - | DEL | IPO5 | ENST00000261574 | 1 | intronic | c.145-19 | e3-19 | tier3 | 63 | 1 | 0.016 | 95 | 18 | 0.159 |
| 353053 | 13 | 113538580 | 113538580 | C | A | SNP | ATP11A | ENST00000375630 | 1 | 3_prime_untranslated_region | c.*2202 | NULL | tier2 | 153 | 0 | 0.000 | 156 | 12 | 0.071 |
| 353053 | 15 | 23689650 | 23689650 | G | A | SNP | GOLGA6L2 | ENST00000312015 | -1 | splice_region | c.214-3 | e3-3 | tier3 | 149 | 0 | 0.000 | 277 | 32 | 0.104 |
| 353053 | 15 | 45392025 | 45392025 | G | A | SNP | DUOX2 | ENST00000389039 | -1 | nonsense | c.3250 | p.R1084* | tier1 | 65 | 0 | 0.000 | 102 | 16 | 0.136 |
| 353053 | 17 | 16723414 | 16723414 | C | T | SNP | ENSG00000226145 | ENST00000417510 | -1 | splice_site | c.NULL | NULL | tier1 | 190 | 0 | 0.000 | 221 | 20 | 0.083 |
| 353053 | 21 | 36164611 | 36164611 | C | A | SNP | RUNX1 | ENST00000300305 | -1 | nonsense | c.1264 | p.E422* | tier1 | 706 | 1 | 0.001 | 833 | 106 | 0.113 |
| 353053 | X | 102840637 | 102840637 | T | G | SNP | TCEAL4 | ENST00000372629 | 1 | missense | c.268 | p.W90G | tier1 | 76 | 2 | 0.026 | 88 | 19 | 0.176 |
| 353053 | X | 133547986 | 133547986 | A | G | SNP | PHF6 | ENST00000394292 | 1 | missense | c.722 | p.Y241C | tier1 | 392 | 0 | 0.000 | 601 | 84 | 0.123 |
| 368402 | 1 | 154215669 | 154215669 | G | A | SNP | UBAP2L | ENST00000466173 | 1 | 3_prime_untranslated_region | c.*154215669 | NULL | tier3 | 79 | 1 | 0.013 | 228 | 59 | 0.206 |
| 368402 | 1 | 216723715 | 216723715 | C | T | SNP | ESRRG | ENST00000408911 | -1 | intronic | c.862+13846 | e5+13846 | tier2 | 118 | 0 | 0.000 | 281 | 62 | 0.181 |
| 368402 | 2 | 11702607 | 11702607 | A | G | SNP | GREB1 | ENST00000234142 | 1 | missense | c.176 | p.N59S | tier1 | 27 | 0 | 0.000 | 206 | 62 | 0.231 |
| 368402 | 2 | 20460043 | 20460043 | C | A | SNP | PUM2 | ENST00000361078 | -1 | intronic | c.2355+55 | e14+55 | tier2 | 51 | 1 | 0.019 | 86 | 20 | 0.189 |
| 368402 | 2 | 179596201 | 179596201 | T | A | SNP | TTN | ENST00000342992 | -1 | silent | c.13560 | p.L4520 | tier1 | 79 | 0 | 0.000 | 213 | 36 | 0.145 |
| 368402 | 3 | 38418449 | 38418449 | T | C | SNP | XYLB | ENST00000207870 | 1 | silent | c.1161 | p.I387 | tier1 | 107 | 2 | 0.018 | 189 | 47 | 0.198 |
| 368402 | 4 | 22421553 | 22421553 | C | T | SNP | GPR125 | ENST00000334304 | -1 | intronic | c.1809+956 | e12+956 | tier3 | 97 | 1 | 0.010 | 120 | 30 | 0.200 |
| 368402 | 4 | 140432050 | 140432050 | T | C | SNP | SETD7 | ENST00000274031 | -1 | 3_prime_untranslated_region | c.*767 | NULL | tier3 | 126 | 0 | 0.000 | 191 | 41 | 0.177 |
| 368402 | 4 | 166915437 | 166915437 | A | T | SNP | TLL1 | ENST00000061240 | 1 | intronic | c.362-96 | e4-96 | tier3 | 188 | 0 | 0.000 | 184 | 48 | 0.206 |
| 368402 | 5 | 37239010 | 37239010 | C | - | DEL | C5orf42 | ENST00000425232 | -1 | frame_shift_del | c.887 | p.S296fs | tier1 | 124 | 1 | 0.008 | 351 | 82 | 0.189 |
| 368402 | 5 | 115319116 | 115319116 | G | A | SNP | AQPEP | ENST00000357872 | 1 | missense | c.828 | p.M276I | tier1 | 77 | 1 | 0.013 | 183 | 35 | 0.160 |
| 368402 | 5 | 156924056 | 156924056 | C | T | SNP | ADAM19 | ENST00000517374 | -1 | missense | c.152 | p.S51N | tier1 | 22 | 0 | 0.000 | 169 | 57 | 0.252 |
| 368402 | 7 | 44105132 | 44105132 | G | A | SNP | PGAM2 | ENST00000297283 | -1 | 5_prime_untranslated_region | c.-4 | NULL | tier3 | 8 | 0 | 0.000 | 67 | 80 | 0.544 |
| 368402 | 9 | 78973756 | 78973756 | G | A | SNP | PCSK5 | ENST00000545128 | 1 | missense | c.5501 | p.R1834Q | tier1 | 44 | 0 | 0.000 | 195 | 52 | 0.210 |
| 368402 | 9 | 79319663 | 79319663 | G | A | SNP | PRUNE2 | ENST00000428286 | -1 | intronic | c.6436+14 | e1+14 | tier2 | 108 | 0 | 0.000 | 237 | 49 | 0.171 |
| 368402 | 9 | 79319664 | 79319664 | G | T | SNP | PRUNE2 | ENST00000428286 | -1 | intronic | c.6436+13 | e1+13 | tier2 | 107 | 0 | 0.000 | 236 | 49 | 0.172 |
| 368402 | 10 | 115963109 | 115963109 | C | A | SNP | TDRD1 | ENST00000251864 | 1 | intronic | c.802-38 | e7-38 | tier3 | 114 | 0 | 0.000 | 124 | 40 | 0.244 |
| 368402 | 12 | 124826497 | 124826497 | T | C | SNP | NCOR2 | ENST00000356219 | -1 | missense | c.5081 | p.N1694S | tier1 | 42 | 0 | 0.000 | 379 | 94 | 0.198 |
| 368402 | 12 | 132505857 | 132505857 | C | T | SNP | EP400 | ENST00000333577 | 1 | missense | c.4789 | p.R1597C | tier1 | 38 | 0 | 0.000 | 334 | 66 | 0.165 |
| 368402 | 16 | 34290568 | 34290568 | G | G | SNP | CCNYL3 | ENST00000407030 | 1 | 3_prime_untranslated_region | c.*34290568 | NULL | tier3 | 88 | 2 | 0.022 | 138 | 41 | 0.228 |
| 368402 | 17 | 74732959 | 74732959 | C | C | SNP | SRSF2 | ENST00000392485 | -1 | missense | c.284 | p.P95R | tier1 | 43 | 0 | 0.000 | 439 | 118 | 0.212 |
| 368402 | 17 | 78700994 | 78700994 | C | T | SNP | RPTOR | ENST00000306801 | 1 | intronic | c.508-3366 | e5-3366 | tier3 | 44 | 0 | 0.000 | 435 | 103 | 0.191 |
| 368402 | 17 | 79563240 | 79563240 | C | T | SNP | NPLOC4 | ENST00000374747 | -1 | missense | c.1022 | p.C341Y | tier1 | 47 | 0 | 0.000 | 177 | 38 | 0.177 |
| 368402 | 19 | 9025669 | 9025669 | G | A | SNP | MUC16 | ENST00000397910 | -1 | intronic | c.36796-11 | e15-11 | tier3 | 204 | 1 | 0.005 | 1554 | 72 | 0.044 |
| 368402 | 21 | 36231802 | 36231802 | T | A | SNP | RUNX1 | ENST00000300305 | -1 | missense | c.582 | p.K194N | tier1 | 228 | 1 | 0.004 | 1359 | 318 | 0.190 |
| 368402 | 21 | 36259137 | 36259137 | T | C | SNP | RUNX1 | ENST0000 | | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-----------------------------|----|-----|-----------------|-----------------|----|-----------------------------|-----------------|----------|-------|-----|---|-------|------|-----|-------|
| 377158 | 1 | 226413278 | 226413278 | C | A | SNP | MIXL1 | ENST00000366810 | 1 | missense | c.464 | p.P155Q | tier1 | 58 | 3 | 0.049 | 116 | 92 | 0.442 |
| 377158 | 3 | 1415578 | 1415578 | C | T | SNP | CNTN6 | ENST00000350110 | 1 | intronic | c.1946-30 | e15-30 | tier3 | 53 | 0 | 0.000 | 178 | 110 | 0.382 |
| 377158 | 4 | 123260564 | 123260564 | G | A | SNP | KIAA1109 | ENST00000264501 | 1 | splice_site | c.12352+1 | e70+1 | tier1 | 14 | 0 | 0.000 | 85 | 79 | 0.482 |
| 377158 | 4 | 156135650 | 156135650 | C | T | SNP | NPY2R | ENST00000329476 | 1 | missense | c.559 | p.R187W | tier1 | 33 | 0 | 0.000 | 48 | 30 | 0.385 |
| 377158 | 4 | 186920705 | 186920705 | G | C | SNP | SORBS2 | ENST00000284776 | -1 | 5_prime_flanking_region | c.-337354 | NULL | tier3 | 199 | 3 | 0.015 | 432 | 41 | 0.087 |
| 377158 | 5 | 140532032 | 140532032 | G | C | SNP | PCDH6 | ENST00000231136 | 1 | missense | c.2194 | p.G732R | tier1 | 188 | 1 | 0.005 | 214 | 172 | 0.444 |
| 377158 | 5 | 141233621 | 141233621 | C | T | SNP | PCDH1 | ENST00000287008 | -1 | missense | c.3700 | p.E1234K | tier1 | 123 | 1 | 0.008 | 173 | 129 | 0.427 |
| 377158 | 7 | 44268486 | 44268486 | C | T | SNP | CAMK2B | ENST00000395749 | -1 | silent | c.1377 | p.R459 | tier1 | 79 | 0 | 0.000 | 88 | 56 | 0.386 |
| 377158 | 7 | 76162997 | 76162997 | C | T | SNP | UPK3B | ENST00000419923 | 1 | intronic | c.963+18224 | e4+18224 | tier2 | 183 | 0 | 0.000 | 293 | 195 | 0.400 |
| 377158 | 7 | 100636426 | 100636426 | C | T | SNP | MUC12 | ENST00000536621 | 1 | missense | c.2582 | p.T861M | tier1 | 17 | 0 | 0.000 | 33 | 38 | 0.535 |
| 377158 | 7 | 102257060 | 102257060 | G | A | SNP | RASA4 | ENST00000462172 | -1 | 5_prime_untranslated_region | c.-7253 | NULL | tier2 | 191 | 0 | 0.000 | 490 | 58 | 0.106 |
| 377158 | 7 | 129083907 | 129083907 | G | A | SNP | STRIP2 | ENST00000249344 | 1 | missense | c.242 | p.R81K | tier1 | 160 | 1 | 0.006 | 530 | 407 | 0.433 |
| 377158 | 8 | 1616604 | 1616604 | C | T | SNP | DLGAP2 | ENST00000520901 | 1 | missense | c.1730 | p.T577M | tier1 | 103 | 4 | 0.037 | 72 | 65 | 0.471 |
| 377158 | 8 | 93029456 | 93029456 | G | A | SNP | RUNX1T1 | ENST00000436581 | -1 | missense | c.257 | p.T86M | tier1 | 418 | 0 | 0.000 | 693 | 480 | 0.409 |
| 377158 | 9 | 68438490 | 68438490 | T | C | SNP | ENSG00000215548 | ENST00000376334 | -1 | intronic | c.NULL | NULL | tier3 | 134 | 2 | 0.015 | 719 | 65 | 0.083 |
| 377158 | 9 | 68438491 | 68438491 | T | C | SNP | ENSG00000215548 | ENST00000376334 | -1 | intronic | c.NULL | NULL | tier3 | 134 | 2 | 0.015 | 720 | 64 | 0.082 |
| 377158 | 10 | 125521468 | 125521468 | C | A | SNP | CPXM2 | ENST00000241305 | -1 | missense | c.1697 | p.R566L | tier1 | 128 | 0 | 0.000 | 116 | 96 | 0.449 |
| 377158 | 11 | 93796883 | 93796883 | G | C | SNP | HEPHL1 | ENST00000315765 | 1 | missense | c.625 | p.E209Q | tier1 | 79 | 2 | 0.025 | 137 | 57 | 0.294 |
| 377158 | 11 | 132177495 | 132177495 | A | G | SNP | NTM | ENST00000539799 | 1 | intronic | c.527-88 | e5-88 | tier3 | 74 | 1 | 0.013 | 109 | 68 | 0.384 |
| 377158 | 12 | 2794921 | 2794921 | G | A | SNP | CACNA1C | ENST00000399634 | 1 | missense | c.5806 | p.E1936K | tier1 | 98 | 1 | 0.010 | 194 | 21 | 0.097 |
| 377158 | 12 | 53439709 | 53439709 | C | T | SNP | LOC283335 | ENST00000546566 | -1 | intronic | c.NULL | NULL | tier3 | 87 | 0 | 0.000 | 176 | 113 | 0.391 |
| 377158 | 14 | 20216351 | 20216351 | C | T | SNP | OR4Q3 | ENST00000331723 | 1 | silent | c.765 | p.C255 | tier1 | 27 | 0 | 0.000 | 48 | 32 | 0.400 |
| 377158 | 14 | 38296537 | 38296537 | G | A | SNP | TTC6 | ENST00000478811 | 1 | missense | c.1457 | p.G486E | tier1 | 11 | 0 | 0.000 | 97 | 61 | 0.386 |
| 377158 | 14 | 80328831 | 80328831 | - | A | INS | NRXN3 | ENST00000428277 | 1 | 3_prime_untranslated_region | c.*524 | NULL | tier4 | 29 | 0 | 0.000 | 69 | 27 | 0.281 |
| 377158 | 14 | 94750258 | 94750258 | A | T | SNP | SERPINA10 | ENST00000261994 | -1 | 3_prime_untranslated_region | c.*44 | NULL | tier3 | 47 | 1 | 0.021 | 85 | 43 | 0.336 |
| 377158 | 15 | 20362902 | 20362902 | T | - | DEL | BMS1P15 | ENST00000554269 | 1 | intronic | c.NULL | NULL | tier3 | 376 | 1 | 0.003 | 794 | 289 | 0.267 |
| 377158 | 16 | 67242195 | 67242195 | C | T | SNP | LRRC29 | ENST00000341546 | -1 | intronic | c.285+20 | e2+20 | tier3 | 66 | 0 | 0.000 | 90 | 68 | 0.430 |
| 377158 | 17 | 13399610 | 13399610 | G | A | SNP | HS3ST3A1 | ENST00000284110 | -1 | silent | c.1125 | p.I375 | tier1 | 157 | 0 | 0.000 | 166 | 134 | 0.447 |
| 377158 | 17 | 37901877 | 37901877 | C | A | SNP | GRB7 | ENST00000445327 | 1 | intronic | c.1278+86 | e11+86 | tier3 | 47 | 0 | 0.000 | 103 | 79 | 0.432 |
| 377158 | 17 | 47490642 | 47490642 | A | G | SNP | PHB | ENST00000300408 | -1 | 5_prime_untranslated_region | c.-16 | NULL | tier3 | 53 | 0 | 0.000 | 58 | 60 | 0.509 |
| 377158 | 17 | 48653205 | 48653205 | G | A | SNP | CACNA1G | ENST00000359106 | 1 | missense | c.1442 | p.S481N | tier1 | 397 | 2 | 0.005 | 469 | 348 | 0.425 |
| 377158 | 19 | 12221114 | 12221114 | T | C | SNP | ENSG00000188474 | ENST00000339302 | 1 | splice_region | c.1-6 | e1-6 | tier3 | 84 | 0 | 0.000 | 132 | 85 | 0.390 |
| 377158 | 19 | 16008165 | 16008165 | G | A | SNP | CYP4F2 | ENST00000221700 | -1 | intronic | c.198+59 | e1+59 | tier3 | 66 | 3 | 0.043 | 69 | 61 | 0.459 |
| 377158 | 19 | 33792822 | 33792822 | C | A | SNP | CEBPA | ENST00000498907 | -1 | nonsense | c.499 | p.E167* | tier1 | 451 | 1 | 0.002 | 795 | 70 | 0.081 |
| 377158 | 20 | 31022403 | 31022425 | CACCACTGCCATAGA GAGGCGGC | - | DEL | ASXL1 | ENST00000375687 | 1 | frame_shift_del | c.1888_1910 | p.H633fs | tier1 | 648 | 5 | 0.008 | 1110 | 152 | 0.120 |
| 377158 | 20 | 31022417 | 31022417 | A | - | DEL | ASXL1 | ENST00000375687 | 1 | frame_shift_del | c.1902 | p.E635fs | tier1 | 641 | 4 | 0.006 | 892 | 115 | 0.114 |
| 377158 | 20 | 33573852 | 33573852 | T | C | SNP | MYH7B | ENST00000262873 | 1 | intronic | c.932-51 | e12-51 | tier3 | 16 | 0 | 0.000 | 22 | 20 | 0.476 |
| 377158 | 21 | 36231773 | 36231773 | C | T | SNP | RUNX1 | ENST00000300305 | -1 | missense | c.611 | p.R204Q | tier1 | 773 | 3 | 0.004 | 951 | 594 | 0.384 |
| 377158 | 21 | 39672197 | 39672197 | T | C | SNP | KCNJ15 | ENST00000328656 | 1 | silent | c.1014 | p.D338 | tier1 | 19 | 0 | 0.000 | 39 | 24 | 0.381 |
| 377158 | 21 | 44514777 | 44514777 | T | G | SNP | U2AF1 | ENST00000291552 | -1 | missense | c.470 | p.Q157P | tier1 | 272 | 1 | 0.004 | 448 | 282 | 0.386 |
| 377158 | 21 | 44838927 | 44838927 | G | A | SNP | SIK1 | ENST00000270162 | -1 | missense | c.1436 | p.T479I | tier1 | 46 | 0 | 0.000 | 42 | 36 | 0.462 |
| 377158 | 22 | 34875057 | 34875057 | C | T | SNP | - | - | - | - | - | - | tier3 | 25 | 0 | 0.000 | 50 | 31 | 0.383 |
| 377158 | 22 | 40425476 | 40425476 | G | A | SNP | FAM83F | ENST00000333407 | 1 | intronic | c.1454-44 | e5-44 | tier3 | 79 | 0 | 0.000 | 95 | 61 | 0.391 |
| 377158 | X | 54948751 | 54948751 | G | A | SNP | TRO | ENST00000452830 | 1 | 5_prime_untranslated_region | c.-347 | NULL | tier3 | 48 | 0 | 0.000 | 84 | 48 | 0.364 |
| 377158 | X | 62974526 | 62974526 | G | A | SNP | ARHGGEF9 | ENST00000495564 | -1 | rna | NULL | NULL | tier1 | 168 | 0 | 0.000 | 179 | 93 | 0.341 |
| 377158 | X | 152826317 | 152826317 | C | A | SNP | ATP2B3 | ENST00000263519 | 1 | missense | c.3023 | p.T1008N | tier1 | 385 | 1 | 0.003 | 544 | 223 | 0.290 |
| 378511 | 1 | 38185727 | 38185727 | C | T | SNP | EPHA10 | ENST00000427468 | -1 | missense | c.2416 | p.G806S | tier1 | 311 | 1 | 0.003 | 326 | 208 | 0.389 |
| 378511 | 1 | 103471831 | 103471831 | T | A | SNP | COL11A1 | ENST00000358392 | -1 | missense | c.1760 | p.K587I | tier1 | 151 | 0 | 0.000 | 142 | 72 | 0.335 |
| 378511 | 1 | 151542094 | 151542095 | - | TT | INS | TUFT1 | ENST00000538902 | 1 | intronic | c.538-39_538-38 | e7-38 | tier3 | 75 | 0 | 0.000 | 85 | 64 | 0.430 |
| 378511 | 1 | 159505705 | 159505705 | G | C | SNP | OR10J5 | ENST00000334857 | -1 | missense | c.93 | p.F31L | tier1 | 77 | 0 | 0.000 | 105 | 33 | 0.239 |
| 378511 | 1 | 215990362 | 215990362 | T | C | SNP | USH2A | ENST00000366943 | -1 | missense | c.9547 | p.I3183V | tier1 | 97 | 0 | 0.000 | 80 | 112 | 0.583 |
| 378511 | 1 | 218458652 | 218458652 | C | T | SNP | RRP15 | ENST00000366932 | 1 | 5_prime_untranslated_region | c.-7 | NULL | tier2 | 152 | 0 | 0.000 | 210 | 73 | 0.258 |
| 378511 | 2 | 10797867 | 10797867 | C | T | SNP | NOL10 | ENST00000381685 | -1 | splice_site | c.906+1 | e11+1 | tier1 | 112 | 1 | 0.009 | 121 | 51 | 0.295 |
| 378511 | 2 | 89246763 | 89246763 | C | A | SNP | IGKV7-3 | ENST00000357453 | -1 | 5_prime_flanking_region | c.-31869 | NULL | tier3 | 173 | 1 | 0.006 | 124 | 88 | 0.415 |
| 378511 | 2 | 127805801 | 127805801 | G | A | SNP | BIN1 | ENST00000462958 | -1 | 5_prime_untranslated_region | c.-127805801 | NULL | tier3 | 130 | 0 | 0.000 | 114 | 86 | 0.430 |
| 378511 | 2 | 191366422 | 191366422 | T | A | SNP | MFSD6 | ENST00000281416 | 1 | 3_prime_untranslated_region | c.*1478 | NULL | tier3 | 84 | 0 | 0.000 | 103 | 57 | 0.356 |
| 378511 | 4 | 70826715 | 70826715 | G | T | SNP | CSN2 | ENST00000353151 | -1 | 5_prime_untranslated_region | c.-2 | NULL | tier3 | 100 | 0 | 0.000 | 99 | 60 | 0.377 |
| 378511 | 4 | 85562062 | 85562062 | C | T | SNP | CDS1 | ENST00000295887 | 1 | silent | c.951 | p.F317 | tier1 | 93 | 0 | 0.000 | 67 | 51 | 0.432 |
| 378511 | 4 | 100239105 | 100239105 | T | G | SNP | ADH1B | ENST00000504498 | -1 | rna | NULL | NULL | tier1 | 74 | 0 | 0.000 | 83 | 41 | 0.331 |
| 378511 | 4 | 150509056 | 150509056 | A | G | SNP | ENSG00000234828 | ENST00000511993 | -1 | intronic | c.NULL | NULL | tier3 | 94 | 0 | 0.000 | 159 | 80 | 0.335 |
| 378511 | 5 | 134059300 | 134059300 | T | C | SNP | SEC24A | ENST00000398844 | 1 | missense | c.3107 | p.I1036T | tier1 | 236 | 0 | 0.000 | 222 | 119 | 0.347 |
| 378511 | 5 | 140033542 | 140033542 | A | G | SNP | IK | ENST00000417647 | 1 | missense | c.424 | p.K142E | tier1 | 180 | 0 | 0.000 | 170 | 106 | 0.381 |
| 378511 | 5 | 140719581 | 140719581 | A | C | SNP | PCDHGA2 | ENST00000394576 | 1 | missense | c.1043 | p.Y348S | tier1 | 76 | 0 | 0.000 | 68 | 46 | 0.404 |
| 378511 | 5 | 145616829 | 145616829 | T | C | SNP | RBM27 | ENST00000265271 | 1 | intronic | c.1145-32 | e8-32 | tier3 | 92 | 0 | 0.000 | 64 | 44 | 0.407 |
| 378511 | 6 | 23855602 | 23855602 | A | G | SNP | SPTLC1P2 | ENST00000407089 | -1 | 3_prime_flanking_region | c.*1324 | NULL | tier3 | 229 | 0 | 0.000 | 215 | 104 | 0.326 |
| 378511 | 6 | 122777603 | 122777603 | T | C | SNP | SERINC1 | ENST00000339697 | -1 | intronic | c.371+23 | e3+23 | tier3 | 109 | 1 | 0.009 | 120 | 53 | 0.306 |
| 378511 | 7 | 128291564 | 128291564 | C | G | SNP | ENSG00000243302 | ENST00000465971 | 1 | 3_prime_untranslated_region | c.*128291564 | NULL | tier3 | 60 | 1 | 0.016 | 35 | 10 | 0.222 |
| 378511 | 8 | 7450151 | 7450151 | A | T | SNP | OR7E157P | ENST00000494376 | 1 | 3_prime_untranslated_region | c.*7450151 | NULL | tier2 | 311 | 0 | 0.000 | 442 | 61 | 0.121 |
| 378511 | 8 | 22477862 | 22477862 | T | C | SN | | | | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|----|-----|-----|------------------------|-----------------|----|-----------------------------|-----------------|----------|-------|-----|----|-------|-----|-----|-------|
| 378511 | 11 | 18955245 | 18955245 | C | A | SNP | <i>ENSG00000255244</i> | ENST00000528646 | 1 | 5_prime_flanking_region | c.-1287 | NULL | tier3 | 146 | 0 | 0.000 | 191 | 21 | 0.099 |
| 378511 | 11 | 20805389 | 20805389 | T | G | SNP | <i>NELL1</i> | ENST00000357134 | 1 | intronic | c.335+13 | e3+13 | tier3 | 89 | 0 | 0.000 | 82 | 46 | 0.359 |
| 378511 | 11 | 34186853 | 34186853 | G | A | SNP | <i>ABTB2</i> | ENST00000435224 | -1 | silent | c.1806 | p.G602 | tier1 | 52 | 0 | 0.000 | 63 | 18 | 0.222 |
| 378511 | 11 | 66152452 | 66152452 | G | T | SNP | <i>NPAS4</i> | ENST00000311034 | 1 | 5_prime_flanking_region | c.-36199 | NULL | tier4 | 42 | 0 | 0.000 | 75 | 53 | 0.414 |
| 378511 | 11 | 71939384 | 71939384 | G | A | SNP | <i>INPPL1</i> | ENST00000298229 | 1 | splice_region | c.247-8 | e3-8 | tier3 | 83 | 0 | 0.000 | 67 | 38 | 0.362 |
| 378511 | 11 | 72492296 | 72492296 | C | T | SNP | <i>STARD10</i> | ENST00000334805 | -1 | 5_prime_untranslated_region | c.-70 | NULL | tier2 | 48 | 0 | 0.000 | 46 | 33 | 0.418 |
| 378511 | 11 | 119025065 | 119025065 | T | C | SNP | <i>ABCG4</i> | ENST00000307417 | 1 | missense | c.452 | p.M151T | tier1 | 54 | 0 | 0.000 | 53 | 36 | 0.405 |
| 378511 | 11 | 129739492 | 129739492 | G | A | SNP | <i>NFRKB</i> | ENST00000524794 | -1 | missense | c.3503 | p.A1168V | tier1 | 86 | 0 | 0.000 | 61 | 36 | 0.371 |
| 378511 | 12 | 12038950 | 12038951 | - | T | INS | <i>ETV6</i> | ENST00000396373 | 1 | frame_shift_ins | c.1243_1244 | p.L416fs | tier1 | 647 | 0 | 0.000 | 577 | 264 | 0.314 |
| 378511 | 12 | 52182517 | 52182517 | T | A | SNP | <i>SCN8A</i> | ENST00000354534 | 1 | silent | c.4266 | p.A1422 | tier1 | 71 | 0 | 0.000 | 68 | 43 | 0.387 |
| 378511 | 12 | 133768616 | 133768616 | T | C | SNP | <i>ZNF268</i> | ENST00000228289 | 1 | intronic | c.457+27 | e4+27 | tier3 | 88 | 0 | 0.000 | 61 | 32 | 0.344 |
| 378511 | 13 | 95361880 | 95361880 | T | C | SNP | <i>LINC00391</i> | ENST00000433569 | -1 | 5_prime_flanking_region | c.-6764 | NULL | tier2 | 135 | 2 | 0.015 | 224 | 145 | 0.393 |
| 378511 | 14 | 73587919 | 73587919 | A | T | SNP | <i>RBM25</i> | ENST00000261973 | 1 | 3_prime_untranslated_region | c.*1406 | NULL | tier3 | 241 | 1 | 0.004 | 385 | 18 | 0.045 |
| 378511 | 15 | 25928387 | 25928387 | G | A | SNP | <i>ATP10A</i> | ENST00000356865 | -1 | intronic | c.3492+46 | e17+46 | tier3 | 40 | 0 | 0.000 | 36 | 28 | 0.438 |
| 378511 | 15 | 44090170 | 44090170 | G | A | SNP | <i>SERINC4</i> | ENST00000249714 | -1 | silent | c.60 | p.S20 | tier1 | 93 | 0 | 0.000 | 98 | 46 | 0.319 |
| 378511 | 17 | 7910348 | 7910348 | T | C | SNP | <i>GUCY2D</i> | ENST00000254854 | 1 | intronic | c.1379-30 | e4-30 | tier3 | 30 | 0 | 0.000 | 29 | 29 | 0.500 |
| 378511 | 17 | 39551774 | 39551774 | A | G | SNP | <i>KRT31</i> | ENST00000251645 | -1 | silent | c.690 | p.S230 | tier1 | 114 | 0 | 0.000 | 108 | 43 | 0.285 |
| 378511 | 17 | 59557555 | 59557555 | C | G | SNP | <i>TBX4</i> | ENST00000393853 | 1 | missense | c.896 | p.A299G | tier1 | 75 | 0 | 0.000 | 84 | 48 | 0.364 |
| 378511 | 17 | 65924700 | 65924700 | A | C | SNP | <i>BPTF</i> | ENST00000321892 | 1 | missense | c.6359 | p.Q2120P | tier1 | 146 | 3 | 0.020 | 129 | 75 | 0.368 |
| 378511 | 18 | 60999185 | 60999185 | T | C | SNP | <i>KDSR</i> | ENST00000406396 | -1 | intronic | c.880-51 | e10-51 | tier3 | 55 | 0 | 0.000 | 44 | 29 | 0.397 |
| 378511 | 18 | 61570598 | 61570598 | G | T | SNP | <i>SERPINB2</i> | ENST00000299502 | 1 | 3_prime_untranslated_region | c.*59 | NULL | tier3 | 75 | 0 | 0.000 | 86 | 43 | 0.333 |
| 378511 | 19 | 51648220 | 51648220 | G | A | SNP | <i>SIGLEC7</i> | ENST00000317643 | 1 | splice_region | c.760+4 | e3+4 | tier3 | 63 | 0 | 0.000 | 74 | 38 | 0.336 |
| 378511 | 20 | 11906923 | 11906923 | T | G | SNP | <i>BTBD3</i> | ENST00000378226 | 1 | 3_prime_untranslated_region | c.*2609 | NULL | tier2 | 96 | 0 | 0.000 | 140 | 68 | 0.325 |
| 378511 | 21 | 44514777 | 44514777 | T | C | SNP | <i>U2AF1</i> | ENST00000291552 | -1 | missense | c.470 | p.Q157R | tier1 | 399 | 1 | 0.003 | 496 | 38 | 0.071 |
| 378511 | 22 | 24401850 | 24401850 | C | T | SNP | <i>GSTTP2</i> | ENST00000466426 | -1 | rna | NULL | NULL | tier1 | 68 | 0 | 0.000 | 78 | 59 | 0.431 |
| 378511 | 22 | 32162607 | 32162607 | G | T | SNP | <i>DEPDC5</i> | ENST00000266091 | 1 | missense | c.316 | p.D106Y | tier1 | 246 | 1 | 0.004 | 271 | 177 | 0.395 |
| 378511 | X | 26179440 | 26179440 | C | T | SNP | <i>MAGEB6P1</i> | ENST00000416929 | 1 | 3_prime_untranslated_region | c.*26179440 | NULL | tier3 | 77 | 0 | 0.000 | 37 | 74 | 0.667 |
| 395344 | 1 | 25611201 | 25611201 | G | A | SNP | <i>RHD</i> | ENST00000328664 | 1 | missense | c.286 | p.G96S | tier1 | 111 | 0 | 0.000 | 72 | 67 | 0.475 |
| 395344 | 1 | 53972518 | 53972518 | C | A | SNP | <i>GLIS1</i> | ENST00000312233 | -1 | intronic | c.1706-69 | e8-69 | tier3 | 27 | 0 | 0.000 | 21 | 19 | 0.475 |
| 395344 | 2 | 3652556 | 3652556 | T | - | DEL | <i>COLEC11</i> | ENST00000418971 | 1 | intronic | c.172+496 | e2+496 | tier3 | 99 | 0 | 0.000 | 133 | 14 | 0.095 |
| 395344 | 2 | 47748558 | 47748558 | C | A | SNP | <i>KCNK12</i> | ENST00000327876 | -1 | missense | c.781 | p.D261Y | tier1 | 147 | 5 | 0.033 | 93 | 87 | 0.483 |
| 395344 | 2 | 131520055 | 131520055 | C | T | SNP | <i>AMER3</i> | ENST00000321420 | 1 | missense | c.410 | p.P137L | tier1 | 84 | 1 | 0.012 | 56 | 28 | 0.333 |
| 395344 | 2 | 141130704 | 141130704 | T | A | SNP | <i>LRP1B</i> | ENST00000389484 | -1 | missense | c.10641 | p.R3547S | tier1 | 378 | 5 | 0.013 | 344 | 254 | 0.424 |
| 395344 | 2 | 167759975 | 167759975 | G | A | SNP | <i>XIRP2</i> | ENST00000409195 | 1 | 5_prime_untranslated_region | c.-18 | NULL | tier2 | 43 | 0 | 0.000 | 33 | 31 | 0.484 |
| 395344 | 4 | 5990869 | 5990870 | CT | - | DEL | <i>C4orf50</i> | ENST00000531445 | -1 | frame_shift_del | c.630_629 | p.E210fs | tier1 | 74 | 1 | 0.013 | 86 | 15 | 0.149 |
| 395344 | 4 | 134073863 | 134073863 | C | T | SNP | <i>PCDH10</i> | ENST00000264360 | 1 | silent | c.2568 | p.I856 | tier1 | 63 | 0 | 0.000 | 52 | 32 | 0.381 |
| 395344 | 5 | 140431040 | 140431040 | G | A | SNP | <i>PCDHB1</i> | ENST00000306549 | 1 | 5_prime_untranslated_region | c.-16 | NULL | tier3 | 475 | 5 | 0.010 | 418 | 46 | 0.099 |
| 395344 | 5 | 162932633 | 162932633 | C | T | SNP | <i>MAT2B</i> | ENST00000321757 | 1 | 5_prime_untranslated_region | c.-60 | NULL | tier2 | 265 | 0 | 0.000 | 207 | 12 | 0.055 |
| 395344 | 5 | 176730908 | 176730908 | C | A | SNP | <i>PRELID1</i> | ENST00000502670 | 1 | rna | NULL | NULL | tier1 | 47 | 0 | 0.000 | 39 | 27 | 0.409 |
| 395344 | 7 | 1056023 | 1056023 | A | G | SNP | <i>C7orf50</i> | ENST00000357429 | -1 | intronic | c.130-6244 | e2-6244 | tier4 | 29 | 0 | 0.000 | 30 | 11 | 0.268 |
| 395344 | 7 | 1484979 | 1484979 | G | C | SNP | <i>MICALL2</i> | ENST00000297508 | -1 | missense | c.727 | p.P243A | tier1 | 191 | 1 | 0.005 | 133 | 96 | 0.417 |
| 395344 | 7 | 2598908 | 2598908 | T | C | SNP | <i>IQCE</i> | ENST00000402050 | 1 | intronic | c.36+57 | e1+57 | tier2 | 15 | 0 | 0.000 | 7 | 7 | 0.467 |
| 395344 | 8 | 124383531 | 124383531 | C | T | SNP | <i>ATAD2</i> | ENST00000287394 | -1 | missense | c.584 | p.R195H | tier1 | 80 | 0 | 0.000 | 119 | 36 | 0.232 |
| 395344 | 11 | 3041385 | 3041385 | C | T | SNP | <i>CARS</i> | ENST00000380525 | -1 | intronic | c.1266+65 | e11+65 | tier3 | 48 | 0 | 0.000 | 39 | 29 | 0.427 |
| 395344 | 11 | 103043939 | 103043939 | C | A | SNP | <i>DYNC2H1</i> | ENST00000398093 | 1 | missense | c.5463 | p.D1821E | tier1 | 494 | 4 | 0.008 | 522 | 389 | 0.427 |
| 395344 | 12 | 42866304 | 42866304 | C | T | SNP | <i>PRICKLE1</i> | ENST00000345127 | -1 | missense | c.15 | p.M5I | tier1 | 51 | 1 | 0.019 | 29 | 23 | 0.442 |
| 395344 | 13 | 95095724 | 95095724 | G | A | SNP | <i>DCT</i> | ENST00000446125 | -1 | silent | c.1446 | p.D482 | tier1 | 134 | 4 | 0.029 | 113 | 74 | 0.394 |
| 395344 | 13 | 107822358 | 107822358 | G | T | SNP | <i>FAM155A</i> | ENST00000375915 | -1 | 3_prime_untranslated_region | c.*487 | NULL | tier4 | 164 | 2 | 0.012 | 229 | 19 | 0.077 |
| 395344 | 14 | 59931931 | 59931931 | T | G | SNP | <i>GPR135</i> | ENST00000395116 | -1 | missense | c.14 | p.Q5P | tier1 | 6 | 0 | 0.000 | 1 | 7 | 0.875 |
| 395344 | 14 | 97033430 | 97033431 | - | A | INS | <i>PAPOLA</i> | ENST00000216277 | 1 | 3_prime_untranslated_region | c.*2043 | NULL | tier3 | 78 | 0 | 0.000 | 101 | 13 | 0.114 |
| 395344 | 16 | 32914951 | 32914951 | C | A | SNP | <i>IGHV3OR16-15</i> | ENST00000565463 | 1 | 3_prime_untranslated_region | c.*32914951 | NULL | tier3 | 168 | 0 | 0.000 | 125 | 86 | 0.408 |
| 395344 | 19 | 5686425 | 5686425 | A | - | DEL | <i>HSD11B1L</i> | ENST00000423665 | 1 | splice_site_del | c.205-2 | e3-2 | tier1 | 128 | 0 | 0.000 | 99 | 40 | 0.288 |
| 395344 | 19 | 36216047 | 36216047 | T | C | SNP | <i>KMT2B</i> | ENST00000222270 | 1 | intronic | c.3528+59 | e10+59 | tier3 | 58 | 0 | 0.000 | 56 | 28 | 0.333 |
| 395344 | 20 | 31022441 | 31022442 | - | G | INS | <i>ASXL1</i> | ENST00000375687 | 1 | frame_shift_ins | c.1926_1927 | p.G645fs | tier1 | 677 | 7 | 0.010 | 624 | 294 | 0.320 |
| 395344 | 21 | 36252865 | 36252865 | C | T | SNP | <i>RUNX1</i> | ENST00000300305 | -1 | missense | c.497 | p.R166Q | tier1 | 922 | 3 | 0.003 | 767 | 568 | 0.425 |
| 395344 | 21 | 44524456 | 44524456 | G | A | SNP | <i>U2AF1</i> | ENST00000291552 | -1 | missense | c.101 | p.S34F | tier1 | 456 | 23 | 0.048 | 457 | 297 | 0.393 |
| 395344 | 22 | 19452777 | 19452777 | T | A | SNP | <i>UFD1L</i> | ENST00000263202 | -1 | missense | c.443 | p.N148I | tier1 | 85 | 2 | 0.023 | 86 | 55 | 0.390 |
| 429745 | 1 | 1078202 | 1078202 | A | SNP | SNP | <i>LOC254099</i> | ENST00000416774 | 1 | rna | NULL | NULL | tier1 | 53 | 0 | 0.000 | 62 | 42 | 0.404 |
| 429745 | 1 | 78189035 | 78189035 | T | C | SNP | <i>USP33</i> | ENST00000357428 | -1 | missense | c.1463 | p.Q488R | tier1 | 42 | 0 | 0.000 | 29 | 22 | 0.431 |
| 429745 | 1 | 103400670 | 103400670 | C | T | SNP | <i>COL11A1</i> | ENST00000358392 | -1 | splice_site | c.3475-1 | e45-1 | tier1 | 85 | 0 | 0.000 | 172 | 19 | 0.100 |
| 429745 | 1 | 193018974 | 193018974 | G | T | SNP | <i>UCHL5</i> | ENST00000367451 | -1 | missense | c.148 | p.H50N | tier1 | 64 | 0 | 0.000 | 87 | 76 | 0.466 |
| 429745 | 1 | 197021696 | 197021696 | T | A | SNP | <i>F13B</i> | ENST00000367412 | -1 | intronic | c.1555+68 | e9+68 | tier3 | 34 | 1 | 0.029 | 37 | 20 | 0.351 |
| 429745 | 1 | 240071866 | 240071866 | C | T | SNP | <i>CHRM3</i> | ENST00000255380 | 1 | missense | c.1115 | p.P372L | tier1 | 16 | 0 | 0.000 | 25 | 20 | 0.444 |
| 429745 | 2 | 152267897 | 152267898 | - | T | INS | <i>RIF1</i> | ENST00000243326 | 1 | intronic | c.183+45_183+44 | e2+45 | tier4 | 24 | 0 | 0.000 | 32 | 12 | 0.273 |
| 429745 | 2 | 234231616 | 234231616 | G | A | SNP | <i>SAG</i> | ENST00000409110 | 1 | missense | c.400 | p.V134M | tier1 | 90 | 0 | 0.000 | 105 | 51 | 0.325 |
| 429745 | 3 | 38010850 | 38010850 | G | T | SNP | <i>CTDSPL</i> | ENST00000273179 | 1 | intronic | c.426+1477 | e5+1477 | tier3 | 54 | 0 | 0.000 | 99 | 71 | 0.410 |
| 429745 | 3 | 197807179 | 197807179 | C | T | SNP | <i>ANKRD18DP</i> | ENST00000335478 | -1 | splice_region | c.NULL | NULL | tier2 | 24 | 0 | 0.000 | 44 | 22 | 0.333 |
| 429745 | 4 | 146046124 | | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|---|--------------|-----|------------------------|-----------------|----|-----------------------------|-----------------|----------|-------|-----|---|-------|------|-----|-------|
| 429745 | 6 | 32521236 | 32521236 | C | T | SNP | <i>HLA-DRB6</i> | ENST00000411500 | -1 | intronic | c.NULL | NULL | tier3 | 35 | 0 | 0.000 | 74 | 19 | 0.204 |
| 429745 | 6 | 32521237 | 32521237 | A | G | SNP | <i>HLA-DRB6</i> | ENST00000411500 | -1 | intronic | c.NULL | NULL | tier3 | 35 | 0 | 0.000 | 74 | 19 | 0.204 |
| 429745 | 7 | 53854870 | 53854870 | C | T | SNP | <i>ENSG00000232418</i> | ENST00000426503 | 1 | 3_prime_untranslated_region | c.*53854870 | NULL | tier2 | 84 | 0 | 0.000 | 86 | 56 | 0.394 |
| 429745 | 7 | 103206001 | 103206002 | - | A | INS | <i>RELN</i> | ENST00000424685 | -1 | splice_region_ins | c.4937-4_4937-3 | e34-3 | tier3 | 19 | 0 | 0.000 | 27 | 13 | 0.325 |
| 429745 | 9 | 114412150 | 114412150 | C | - | DEL | <i>DNAJC25</i> | ENST00000313525 | 1 | frame_shift_del | c.907 | p.H303fs | tier1 | 14 | 0 | 0.000 | 18 | 18 | 0.500 |
| 429745 | 10 | 11376350 | 11376350 | T | G | SNP | <i>CELF2</i> | ENST00000450189 | 1 | 3_prime_untranslated_region | c.*5334 | NULL | tier3 | 41 | 0 | 0.000 | 29 | 22 | 0.431 |
| 429745 | 10 | 35300792 | 35300792 | T | C | SNP | <i>CUL2</i> | ENST00000537177 | -1 | missense | c.2147 | p.N716S | tier1 | 56 | 1 | 0.018 | 62 | 62 | 0.500 |
| 429745 | 10 | 111967928 | 111967928 | C | - | DEL | <i>MXI1</i> | ENST00000332674 | 1 | intronic | c.274+88 | e1+88 | tier2 | 77 | 0 | 0.000 | 180 | 19 | 0.096 |
| 429745 | 11 | 62302518 | 62302518 | C | T | SNP | <i>AHNAK</i> | ENST00000378024 | -1 | nonsense | c.299 | p.W100* | tier1 | 50 | 0 | 0.000 | 53 | 26 | 0.329 |
| 429745 | 11 | 65271780 | 65271780 | T | - | DEL | <i>MALAT1</i> | ENST00000534336 | 1 | rna | NULL | NULL | tier1 | 28 | 0 | 0.000 | 22 | 11 | 0.333 |
| 429745 | 11 | 67020214 | 67020214 | G | A | SNP | <i>KDM2A</i> | ENST00000529006 | 1 | missense | c.2818 | p.A940T | tier1 | 40 | 1 | 0.024 | 68 | 36 | 0.346 |
| 429745 | 11 | 67565182 | 67565182 | C | T | SNP | <i>FAM86C2P</i> | ENST00000528684 | -1 | 5_prime_untranslated_region | c.-67565182 | NULL | tier3 | 138 | 2 | 0.014 | 149 | 110 | 0.423 |
| 429745 | 12 | 102074329 | 102074329 | G | A | SNP | <i>MYBPC1</i> | ENST00000361466 | 1 | intronic | c.3433+22 | e29+22 | tier3 | 27 | 0 | 0.000 | 28 | 21 | 0.429 |
| 429745 | 12 | 109569189 | 109569189 | G | T | SNP | <i>ACACB</i> | ENST00000539864 | 1 | 5_prime_untranslated_region | c.-8097 | NULL | tier3 | 38 | 0 | 0.000 | 23 | 21 | 0.477 |
| 429745 | 14 | 31405745 | 31405745 | C | A | SNP | <i>STRN3</i> | ENST00000357479 | -1 | missense | c.802 | p.G268C | tier1 | 71 | 0 | 0.000 | 76 | 29 | 0.274 |
| 429745 | 14 | 64921502 | 64921502 | A | G | SNP | <i>MTHFD1</i> | ENST00000216605 | 1 | missense | c.2627 | p.E876G | tier1 | 41 | 0 | 0.000 | 95 | 17 | 0.152 |
| 429745 | 14 | 81950742 | 81950742 | C | A | SNP | <i>SEL1L</i> | ENST00000336735 | -1 | splice_site | c.1874-1 | e19-1 | tier1 | 41 | 0 | 0.000 | 49 | 17 | 0.258 |
| 429745 | 17 | 26817851 | 26817851 | C | T | SNP | <i>SLC13A2</i> | ENST00000444914 | 1 | silent | c.648 | p.N216 | tier1 | 33 | 1 | 0.029 | 39 | 31 | 0.443 |
| 429745 | 17 | 45913390 | 45913390 | T | A | SNP | <i>LRRC46</i> | ENST00000269025 | 1 | splice_region | c.383-9 | e6-9 | tier3 | 41 | 0 | 0.000 | 60 | 47 | 0.439 |
| 429745 | 17 | 62924761 | 62924761 | A | G | SNP | <i>ENSG00000264057</i> | ENST00000579873 | 1 | 3_prime_untranslated_region | c.*62924761 | NULL | tier2 | 52 | 0 | 0.000 | 152 | 20 | 0.116 |
| 429745 | 17 | 65871717 | 65871717 | T | G | SNP | <i>BPTF</i> | ENST00000321892 | 1 | missense | c.1910 | p.L637R | tier1 | 27 | 0 | 0.000 | 39 | 25 | 0.391 |
| 429745 | 17 | 74018124 | 74018124 | C | A | SNP | <i>EVPL</i> | ENST00000301607 | -1 | intronic | c.646-15 | e7-15 | tier2 | 13 | 0 | 0.000 | 22 | 14 | 0.389 |
| 429745 | 18 | 40849590 | 40849590 | A | T | SNP | <i>SYT4</i> | ENST00000255224 | -1 | 3_prime_untranslated_region | c.*716 | NULL | tier3 | 66 | 0 | 0.000 | 82 | 64 | 0.438 |
| 429745 | 19 | 11019894 | 11019894 | C | T | SNP | <i>CARM1</i> | ENST00000327064 | 1 | intronic | c.558+11 | e4+11 | tier3 | 21 | 0 | 0.000 | 34 | 20 | 0.370 |
| 429745 | 19 | 48643310 | 48643310 | G | C | SNP | <i>LIG1</i> | ENST00000263274 | -1 | silent | c.1005 | p.L335 | tier1 | 13 | 0 | 0.000 | 41 | 39 | 0.488 |
| 429745 | 20 | 23860305 | 23860305 | C | T | SNP | <i>CST5</i> | ENST00000304710 | -1 | nonsense | c.9 | p.W3* | tier1 | 14 | 0 | 0.000 | 12 | 12 | 0.500 |
| 429745 | 21 | 36252919 | 36252920 | - | TAGCATTTCACG | INS | <i>RUNX1</i> | ENST00000300305 | -1 | frame_shift_ins | c.443_442 | p.T148fs | tier1 | 681 | 0 | 0.000 | 1118 | 73 | 0.061 |
| 429745 | X | 39931644 | 39931644 | A | T | SNP | <i>BCOR</i> | ENST00000378444 | -1 | nonsense | c.2955 | p.Y985* | tier1 | 320 | 2 | 0.006 | 423 | 313 | 0.424 |
| 430998 | 1 | 24421386 | 24421386 | C | T | SNP | <i>MYOM3</i> | ENST00000330966 | -1 | silent | c.888 | p.S296 | tier1 | 202 | 1 | 0.005 | 404 | 46 | 0.102 |
| 430998 | 1 | 32162944 | 32162944 | T | C | SNP | <i>COL16A1</i> | ENST00000271069 | -1 | intronic | c.658-43 | e6-43 | tier4 | 31 | 1 | 0.031 | 32 | 24 | 0.949 |
| 430998 | 2 | 15533730 | 15533730 | G | - | DEL | <i>NBAS</i> | ENST00000281513 | -1 | intronic | c.3257+621 | e28+621 | tier4 | 105 | 1 | 0.009 | 159 | 18 | 0.102 |
| 430998 | 2 | 42553171 | 42553171 | T | G | SNP | <i>EML4</i> | ENST00000318522 | 1 | intronic | c.2243-123 | e21-123 | tier3 | 103 | 2 | 0.019 | 106 | 42 | 0.284 |
| 430998 | 2 | 196851848 | 196851848 | T | C | SNP | <i>DNAH7</i> | ENST00000312428 | -1 | missense | c.1696 | p.I566V | tier1 | 72 | 3 | 0.040 | 84 | 39 | 0.317 |
| 430998 | 2 | 198266834 | 198266834 | T | C | SNP | <i>SF3B1</i> | ENST00000335508 | -1 | missense | c.2098 | p.K700E | tier1 | 274 | 4 | 0.014 | 424 | 268 | 0.386 |
| 430998 | 3 | 46486788 | 46486788 | C | T | SNP | <i>LTF</i> | ENST00000231751 | -1 | silent | c.1497 | p.T499 | tier1 | 35 | 2 | 0.053 | 51 | 36 | 0.414 |
| 430998 | 3 | 184557400 | 184557400 | G | T | SNP | <i>VPS8</i> | ENST00000424463 | 1 | 3_prime_untranslated_region | c.*851 | NULL | tier3 | 104 | 3 | 0.028 | 102 | 63 | 0.382 |
| 430998 | 4 | 20574880 | 20574880 | A | G | SNP | <i>SLIT2</i> | ENST00000273739 | 1 | missense | c.3124 | p.K1042E | tier1 | 120 | 0 | 0.000 | 186 | 13 | 0.065 |
| 430998 | 5 | 93217240 | 93217240 | C | T | SNP | <i>FAM172A</i> | ENST00000395965 | -1 | missense | c.722 | p.R241H | tier1 | 98 | 0 | 0.000 | 144 | 12 | 0.077 |
| 430998 | 6 | 123786098 | 123786098 | G | A | SNP | <i>TRDN</i> | ENST00000398178 | -1 | missense | c.884 | p.P295L | tier1 | 92 | 1 | 0.011 | 145 | 78 | 0.350 |
| 430998 | 8 | 11188922 | 11188922 | T | C | SNP | <i>SLC35G5</i> | ENST00000382435 | 1 | missense | c.307 | p.W103R | tier1 | 80 | 0 | 0.000 | 182 | 17 | 0.085 |
| 430998 | 9 | 6534811 | 6534811 | G | A | SNP | <i>GLDC</i> | ENST00000321612 | -1 | intronic | c.2839-23 | e24-23 | tier3 | 54 | 1 | 0.018 | 62 | 37 | 0.374 |
| 430998 | 10 | 105831879 | 105831879 | C | T | SNP | <i>COL17A1</i> | ENST00000353479 | -1 | intronic | c.416-42 | e7-42 | tier3 | 30 | 0 | 0.000 | 50 | 38 | 0.432 |
| 430998 | 11 | 89606363 | 89606363 | C | G | SNP | <i>TRIM64B</i> | ENST00000329862 | -1 | intronic | c.758+73 | e4+73 | tier3 | 686 | 7 | 0.010 | 1004 | 217 | 0.177 |
| 430998 | 12 | 3662052 | 3662052 | C | T | SNP | <i>PRMT8</i> | ENST00000543701 | 1 | 3_prime_untranslated_region | c.*3662052 | NULL | tier3 | 42 | 3 | 0.067 | 70 | 54 | 0.436 |
| 430998 | 12 | 88525006 | 88525006 | A | - | DEL | <i>CEP290</i> | ENST00000309041 | -1 | intronic | c.442-11 | e6-11 | tier4 | 48 | 0 | 0.000 | 66 | 12 | 0.154 |
| 430998 | 13 | 79894878 | 79894878 | A | - | DEL | <i>RBM26</i> | ENST00000438724 | -1 | intronic | c.2863-33 | e21-33 | tier4 | 60 | 1 | 0.016 | 81 | 19 | 0.190 |
| 430998 | 14 | 106391086 | 106391086 | C | T | SNP | <i>KIAA0125</i> | ENST00000482999 | 1 | rna | NULL | NULL | tier1 | 151 | 4 | 0.026 | 211 | 183 | 0.463 |
| 430998 | 16 | 1251965 | 1251965 | C | T | SNP | <i>CACNA1H</i> | ENST00000348261 | 1 | silent | c.1515 | p.R505 | tier1 | 24 | 1 | 0.040 | 44 | 26 | 0.371 |
| 430998 | 16 | 70044642 | 70044642 | G | A | SNP | <i>PDXDC2P</i> | ENST00000325845 | -1 | intronic | c.NULL | NULL | tier3 | 111 | 0 | 0.000 | 186 | 21 | 0.101 |
| 430998 | 17 | 10403963 | 10403963 | G | A | SNP | <i>MYH1</i> | ENST00000226207 | -1 | missense | c.3845 | p.A1282V | tier1 | 74 | 0 | 0.000 | 198 | 25 | 0.111 |
| 430998 | 17 | 47676686 | 47676686 | T | A | SNP | <i>SPOP</i> | ENST00000347630 | -1 | 3_prime_untranslated_region | c.*1054 | NULL | tier4 | 92 | 0 | 0.000 | 138 | 14 | 0.092 |
| 430998 | 17 | 48267657 | 48267657 | G | A | SNP | <i>COL1A1</i> | ENST00000225964 | -1 | intronic | c.2451+31 | e35+31 | tier3 | 89 | 2 | 0.022 | 106 | 53 | 0.333 |
| 430998 | 17 | 66146458 | 66146458 | A | G | SNP | <i>LRRC37A16P</i> | ENST00000586022 | -1 | 5_prime_untranslated_region | c.-66146458 | NULL | tier3 | 162 | 6 | 0.036 | 217 | 113 | 0.342 |
| 430998 | 19 | 1806137 | 1806137 | C | T | SNP | <i>ATP8B3</i> | ENST00000539485 | -1 | missense | c.709 | p.V237M | tier1 | 30 | 0 | 0.000 | 34 | 22 | 0.393 |
| 430998 | 19 | 12767474 | 12767474 | C | T | SNP | <i>MAN2B1</i> | ENST00000593686 | -1 | nonsense | c.165 | p.W55* | tier1 | 95 | 0 | 0.000 | 155 | 13 | 0.077 |
| 430998 | 19 | 22941441 | 22941441 | G | T | SNP | <i>ZNF99</i> | ENST00000397104 | -1 | missense | c.997 | p.P333T | tier1 | 144 | 6 | 0.040 | 141 | 80 | 0.362 |
| 430998 | 19 | 54722897 | 54722897 | C | T | SNP | <i>LILRB3</i> | ENST00000407860 | -1 | intronic | c.1477+101 | e9+101 | tier3 | 69 | 0 | 0.000 | 121 | 16 | 0.116 |
| 430998 | 22 | 40719779 | 40719779 | C | A | SNP | <i>TNRC6B</i> | ENST00000454349 | 1 | 3_prime_untranslated_region | c.*534 | NULL | tier4 | 43 | 0 | 0.000 | 127 | 25 | 0.162 |
| 430998 | 22 | 47075722 | 47075722 | T | C | SNP | <i>GRAMD4</i> | ENST00000361034 | 1 | 3_prime_flanking_region | c.*2574 | NULL | tier2 | 36 | 2 | 0.053 | 49 | 42 | 0.462 |
| 430998 | X | 54319632 | 54319632 | C | T | SNP | <i>WNK3</i> | ENST00000354646 | -1 | missense | c.1822 | p.V608I | tier1 | 97 | 2 | 0.020 | 43 | 112 | 0.718 |
| 430998 | X | 134992384 | 134992384 | T | C | SNP | <i>SAGE1</i> | ENST00000324447 | 1 | intronic | c.1864+55 | e14+55 | tier3 | 21 | 0 | 0.000 | 16 | 59 | 0.776 |
| 435866 | 1 | 98511783 | 98511783 | C | T | SNP | <i>MIR137HG</i> | ENST00000424528 | -1 | 5_prime_untranslated_region | c.-98511783 | NULL | tier4 | 89 | 1 | 0.011 | 243 | 33 | 0.120 |
| 435866 | 2 | 230127480 | 230127480 | C | T | SNP | <i>PID1</i> | ENST00000392054 | -1 | silent | c.42 | p.S14 | tier1 | 28 | 0 | 0.000 | 75 | 35 | 0.318 |
| 435866 | 3 | 45718805 | 45718805 | C | T | SNP | <i>LIMD1</i> | ENST00000273317 | 1 | 3_prime_untranslated_region | c.*254 | NULL | tier3 | 46 | 2 | 0.042 | 101 | 70 | 0.409 |
| 435866 | 3 | 185978207 | 185978207 | T | C | SNP | <i>DGKG</i> | ENST00000265022 | -1 | intronic | c.1424+21 | e15+21 | tier3 | 170 | 1 | 0.006 | 230 | 153 | 0.396 |
| 435866 | 4 | 66356333 | 66356333 | G | A | SNP | <i>EPHA5</i> | ENST00000273854 | -1 | silent | c.1164 | p.D388 | tier1 | 44 | 0 | 0.000 | 115 | 110 | 0.489 |
| 435866 | 5 | 138658293 | 138658293 | A | T | SNP | <i>MATR3</i> | ENST00000361059 | 1 | missense | c.1785 | p.R595S | tier1 | 57 | 0 | 0.000 | 112 | 78 | 0.411 |
| 435866 | 5 | | | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|------------|---|-----|------------------------|-----------------|----|-----------------------------|-------------------|----------|-------|------|---|-------|------|-----|-------|
| 435866 | 8 | 68024182 | 68024182 | G | A | SNP | <i>CSPP1</i> | ENST00000389042 | 1 | intronic | c.1226-25 | e12-25 | tier3 | 51 | 1 | 0.019 | 109 | 101 | 0.481 |
| 435866 | 8 | 139727925 | 139727925 | C | A | SNP | <i>COL22A1</i> | ENST00000303045 | -1 | missense | c.2517 | p.K839N | tier1 | 151 | 1 | 0.007 | 245 | 193 | 0.441 |
| 435866 | 10 | 81921715 | 81921715 | G | T | SNP | <i>ANXA11</i> | ENST00000265447 | -1 | silent | c.1156 | p.R386 | tier1 | 50 | 0 | 0.000 | 206 | 143 | 0.410 |
| 435866 | 10 | 120810716 | 120810716 | A | G | SNP | <i>EIF3A</i> | ENST00000369144 | -1 | missense | c.2314 | p.Y772H | tier1 | 72 | 0 | 0.000 | 154 | 127 | 0.450 |
| 435866 | 12 | 12006446 | 12006447 | CT | - | DEL | <i>ETV6</i> | ENST00000396373 | 1 | frame_shift_del | c.414_415 | p.S139fs | tier1 | 149 | 0 | 0.000 | 366 | 247 | 0.403 |
| 435866 | 14 | 20020000 | 20020000 | C | T | SNP | <i>POTEM</i> | ENST00000439503 | -1 | missense | c.221 | p.G74E | tier1 | 263 | 0 | 0.000 | 1591 | 170 | 0.096 |
| 435866 | 15 | 28420657 | 28420657 | C | T | SNP | <i>HERC2</i> | ENST00000261609 | -1 | splice_site | c.9831+1 | e63+1 | tier1 | 49 | 0 | 0.000 | 311 | 260 | 0.455 |
| 435866 | 16 | 3341019 | 3341019 | C | T | SNP | <i>ZNF263</i> | ENST00000219069 | 1 | 3_prime_untranslated_region | c.*461 | NULL | tier2 | 37 | 0 | 0.000 | 69 | 41 | 0.373 |
| 435866 | 16 | 87870242 | 87870242 | C | T | SNP | <i>SLC7A5</i> | ENST00000261622 | -1 | silent | c.1152 | p.T384 | tier1 | 40 | 0 | 0.000 | 95 | 90 | 0.487 |
| 435866 | 17 | 18967192 | 18967192 | T | C | SNP | <i>SNORD3B-1</i> | ENST00000571722 | -1 | 5_prime_untranslated_region | c.-18967192 | NULL | tier2 | 161 | 0 | 0.000 | 608 | 148 | 0.195 |
| 435866 | 17 | 40049260 | 40049260 | A | G | SNP | <i>ACLY</i> | ENST00000401700 | -1 | intronic | c.1763+26 | e15+26 | tier2 | 34 | 0 | 0.000 | 86 | 79 | 0.479 |
| 435866 | 17 | 74732959 | 74732959 | G | T | SNP | <i>SRSF2</i> | ENST00000392485 | -1 | missense | c.284 | p.P95H | tier1 | 49 | 2 | 0.039 | 140 | 101 | 0.417 |
| 435866 | 18 | 61226995 | 61226995 | C | G | SNP | <i>SERPINB12</i> | ENST00000269491 | 1 | intronic | c.384+44 | e3+44 | tier2 | 37 | 0 | 0.000 | 73 | 61 | 0.455 |
| 435866 | 20 | 14830634 | 14830634 | A | G | SNP | <i>MACROD2</i> | ENST00000463861 | 1 | 3_prime_untranslated_region | c.*14830634 | NULL | tier3 | 116 | 0 | 0.000 | 167 | 126 | 0.430 |
| 435866 | 22 | 20715907 | 20715916 | CAGGGCATCG | - | DEL | <i>ENSG00000188280</i> | ENST00000434783 | 1 | intronic | c.NULL | NULL | tier2 | 70 | 0 | 0.000 | 627 | 79 | 0.112 |
| 435866 | 22 | 38884063 | 38884063 | T | C | SNP | <i>DDX17</i> | ENST00000396821 | -1 | missense | c.1505 | p.Y502C | tier1 | 42 | 0 | 0.000 | 85 | 71 | 0.455 |
| 435866 | X | 79971691 | 79971691 | C | G | SNP | <i>BRWD3</i> | ENST00000373275 | -1 | missense | c.2290 | p.E764Q | tier1 | 78 | 0 | 0.000 | 115 | 90 | 0.439 |
| 435866 | X | 105450024 | 105450024 | C | T | SNP | <i>MUM1L1</i> | ENST00000337685 | 1 | missense | c.599 | p.S200L | tier1 | 60 | 2 | 0.032 | 102 | 74 | 0.421 |
| 437422 | 1 | 22186507 | 22186507 | G | T | SNP | <i>HSPG2</i> | ENST00000374695 | -1 | intronic | c.5015-12 | e41-12 | tier3 | 38 | 0 | 0.000 | 51 | 13 | 0.203 |
| 437422 | 1 | 27736195 | 27736195 | T | C | SNP | <i>WASF2</i> | ENST00000430629 | -1 | missense | c.1330 | p.I444V | tier1 | 168 | 0 | 0.000 | 222 | 40 | 0.153 |
| 437422 | 1 | 84641417 | 84641417 | T | - | DEL | <i>PRKACB</i> | ENST00000370685 | 1 | intronic | c.188-3443 | e2-3443 | tier3 | 58 | 0 | 0.000 | 109 | 14 | 0.114 |
| 437422 | 1 | 145014784 | 145014784 | C | G | SNP | <i>PDE4DIP</i> | ENST00000313382 | -1 | intronic | c.228+1076 | e3+1076 | tier3 | 75 | 0 | 0.000 | 121 | 19 | 0.136 |
| 437422 | 1 | 167802237 | 167802237 | C | A | SNP | <i>ADCY10</i> | ENST00000367851 | -1 | missense | c.3581 | p.S1194I | tier1 | 78 | 0 | 0.000 | 102 | 28 | 0.215 |
| 437422 | 1 | 190067860 | 190067860 | C | T | SNP | <i>BRINP3</i> | ENST00000367462 | -1 | missense | c.1589 | p.R530H | tier1 | 724 | 4 | 0.006 | 906 | 416 | 0.315 |
| 437422 | 1 | 208061976 | 208061976 | C | T | SNP | <i>CD34</i> | ENST00000310833 | -1 | intronic | c.972+51 | e7+51 | tier3 | 73 | 3 | 0.040 | 96 | 37 | 0.278 |
| 437422 | 2 | 5838174 | 5838174 | A | T | SNP | <i>SOX11</i> | ENST00000322002 | 1 | 3_prime_untranslated_region | c.*3995 | NULL | tier2 | 118 | 0 | 0.000 | 225 | 17 | 0.070 |
| 437422 | 2 | 75105821 | 75105821 | G | A | SNP | <i>HK2</i> | ENST00000290573 | 1 | silent | c.1038 | p.K346 | tier1 | 43 | 0 | 0.000 | 60 | 29 | 0.326 |
| 437422 | 2 | 196602340 | 196602340 | C | T | SNP | <i>SLC39A10</i> | ENST00000359634 | 1 | 3_prime_untranslated_region | c.*2575 | NULL | tier3 | 100 | 0 | 0.000 | 189 | 16 | 0.078 |
| 437422 | 3 | 39453515 | 39453515 | A | G | SNP | <i>RPSA</i> | ENST00000301821 | 1 | silent | c.756 | p.V252 | tier1 | 130 | 1 | 0.008 | 200 | 19 | 0.086 |
| 437422 | 3 | 167006710 | 167006710 | C | G | SNP | <i>ZBBX</i> | ENST00000307529 | -1 | missense | c.1941 | p.L647F | tier1 | 118 | 1 | 0.008 | 161 | 75 | 0.314 |
| 437422 | 4 | 120327397 | 120327397 | C | T | SNP | <i>LOC101926979</i> | ENST00000508519 | 1 | 5_prime_flanking_region | c.-48597 | NULL | tier2 | 126 | 1 | 0.008 | 170 | 22 | 0.115 |
| 437422 | 4 | 166416855 | 166416855 | C | T | SNP | <i>CPE</i> | ENST00000402744 | 1 | intronic | c.1332+26 | e8+26 | tier3 | 67 | 0 | 0.000 | 118 | 29 | 0.197 |
| 437422 | 5 | 121786643 | 121786643 | C | G | SNP | <i>SNCAIP</i> | ENST00000379533 | 1 | missense | c.2242 | p.P748A | tier1 | 36 | 1 | 0.027 | 47 | 18 | 0.277 |
| 437422 | 5 | 133898014 | 133898014 | G | A | SNP | <i>PHF15</i> | ENST00000395003 | 1 | missense | c.809 | p.G270E | tier1 | 90 | 0 | 0.000 | 131 | 57 | 0.298 |
| 437422 | 6 | 31239544 | 31239544 | G | T | SNP | <i>HLA-C</i> | ENST00000415537 | -1 | missense | c.173 | p.A58E | tier1 | 167 | 2 | 0.012 | 237 | 26 | 0.099 |
| 437422 | 7 | 6193554 | 6193554 | C | T | SNP | <i>USP42</i> | ENST00000306177 | 1 | missense | c.2369 | p.A790V | tier1 | 52 | 1 | 0.019 | 65 | 31 | 0.323 |
| 437422 | 7 | 77885784 | 77885784 | C | T | SNP | <i>MAGI2</i> | ENST00000354212 | -1 | missense | c.1523 | p.R508H | tier1 | 640 | 2 | 0.003 | 1123 | 56 | 0.047 |
| 437422 | 7 | 120428042 | 120428042 | C | T | SNP | <i>TSPAN12</i> | ENST00000222747 | -1 | 3_prime_untranslated_region | c.*604 | NULL | tier2 | 229 | 1 | 0.004 | 192 | 138 | 0.417 |
| 437422 | 8 | 113299258 | 113299258 | T | C | SNP | <i>CSMD3</i> | ENST00000297405 | -1 | intronic | c.9331+35 | e58+35 | tier3 | 347 | 2 | 0.006 | 524 | 228 | 0.302 |
| 437422 | 8 | 145754744 | 145754744 | T | C | SNP | <i>ARHGAP39</i> | ENST00000377307 | -1 | 3_prime_untranslated_region | c.*1062 | NULL | tier4 | 60 | 0 | 0.000 | 109 | 52 | 0.321 |
| 437422 | 9 | 114341237 | 114341237 | T | A | SNP | <i>PTGR1</i> | ENST00000309195 | -1 | splice_region | c.496-6 | e6-6 | tier3 | 48 | 0 | 0.000 | 82 | 14 | 0.146 |
| 437422 | 11 | 763314 | 763315 | - | T | INS | <i>TALDO1</i> | ENST00000319006 | 1 | intronic | c.462-30_462-29 | e5-29 | tier4 | 6 | 0 | 0.000 | 2 | 7 | 0.778 |
| 437422 | 11 | 46686904 | 46686904 | T | G | SNP | <i>ATG13</i> | ENST00000312040 | 1 | intronic | c.901-29 | e11-29 | tier3 | 77 | 0 | 0.000 | 127 | 58 | 0.312 |
| 437422 | 11 | 130067769 | 130067769 | G | A | SNP | <i>ST14</i> | ENST00000278742 | 1 | missense | c.1388 | p.R463Q | tier1 | 26 | 0 | 0.000 | 26 | 18 | 0.409 |
| 437422 | 12 | 122091150 | 122091150 | T | G | SNP | <i>MORN3</i> | ENST00000355329 | -1 | missense | c.479 | p.Y160S | tier1 | 116 | 1 | 0.009 | 106 | 42 | 0.284 |
| 437422 | 16 | 10721483 | 10721483 | C | T | SNP | <i>TEKT5</i> | ENST00000283025 | -1 | missense | c.1415 | p.R472H | tier1 | 88 | 0 | 0.000 | 95 | 46 | 0.326 |
| 437422 | 17 | 7576851 | 7576851 | A | C | SNP | <i>TP53</i> | ENST00000269305 | -1 | splice_site | c.993+2 | e8+2 | tier1 | 1059 | 8 | 0.008 | 1443 | 648 | 0.309 |
| 437422 | 17 | 7578431 | 7578431 | G | A | SNP | <i>TP53</i> | ENST00000269305 | -1 | nonsense | c.499 | p.Q167* | tier1 | 1051 | 9 | 0.009 | 1349 | 612 | 0.312 |
| 437422 | 17 | 78114602 | 78114603 | - | T | INS | <i>EIF4A3</i> | ENST00000269349 | -1 | intronic | c.372+515_372+514 | e4+515 | tier4 | 114 | 1 | 0.009 | 190 | 19 | 0.091 |
| 437422 | 18 | 33269094 | 33269094 | G | A | SNP | <i>GALNT1</i> | ENST00000269195 | 1 | intronic | c.861-43 | e6-43 | tier3 | 191 | 0 | 0.000 | 235 | 88 | 0.272 |
| 437422 | 20 | 9360833 | 9360834 | - | A | INS | <i>PLCB4</i> | ENST00000334005 | 1 | intronic | c.853+24_853+23 | e10+24 | tier3 | 113 | 1 | 0.009 | 207 | 25 | 0.108 |
| 437422 | 20 | 33068598 | 33068598 | G | A | SNP | <i>ITCH</i> | ENST00000262650 | 1 | intronic | c.2075+61 | e18+61 | tier3 | 137 | 0 | 0.000 | 202 | 40 | 0.165 |
| 437422 | X | 31139616 | 31139616 | T | A | SNP | <i>DMD</i> | ENST00000357033 | -1 | 3_prime_untranslated_region | c.*420 | NULL | tier2 | 213 | 0 | 0.000 | 416 | 19 | 0.044 |
| 437422 | X | 76812922 | 76812922 | C | T | SNP | <i>ATRX</i> | ENST00000373344 | -1 | silent | c.6699 | p.K2233 | tier1 | 292 | 2 | 0.007 | 485 | 107 | 0.180 |
| 439820 | 1 | 2077399 | 2077399 | C | G | SNP | <i>PRKCZ</i> | ENST00000378567 | 1 | intronic | c.553-67 | e7-67 | tier3 | 49 | 0 | 0.000 | 106 | 19 | 0.152 |
| 439820 | 1 | 22927251 | 22927251 | G | C | SNP | <i>EPHA8</i> | ENST00000166244 | 1 | missense | c.2486 | p.W829S | tier1 | 133 | 0 | 0.000 | 179 | 13 | 0.068 |
| 439820 | 1 | 35250948 | 35250948 | C | T | SNP | <i>GJB3</i> | ENST00000373362 | 1 | silent | c.585 | p.S195 | tier1 | 203 | 1 | 0.005 | 314 | 79 | 0.200 |
| 439820 | 1 | 91182861 | 91182861 | A | - | DEL | <i>BARHL2</i> | ENST00000370445 | -1 | 5_prime_flanking_region | c.-109 | NULL | tier2 | 38 | 0 | 0.000 | 72 | 14 | 0.163 |
| 439820 | 1 | 97981408 | 97981408 | G | A | SNP | <i>DPYD</i> | ENST00000370192 | -1 | silent | c.1614 | p.A538 | tier1 | 96 | 1 | 0.010 | 91 | 71 | 0.438 |
| 439820 | 1 | 115258745 | 115258745 | C | G | SNP | <i>NRAS</i> | ENST00000369535 | -1 | missense | c.37 | p.G13R | tier1 | 399 | 0 | 0.000 | 616 | 109 | 0.150 |
| 439820 | 1 | 115258747 | 115258747 | C | G | SNP | <i>NRAS</i> | ENST00000369535 | -1 | missense | c.35 | p.G12A | tier1 | 401 | 0 | 0.000 | 694 | 27 | 0.037 |
| 439820 | 1 | 145417057 | 145417057 | C | G | SNP | <i>HFE2</i> | ENST00000336751 | 1 | 3_prime_untranslated_region | c.*121 | NULL | tier3 | 55 | 0 | 0.000 | 70 | 69 | 0.496 |
| 439820 | 1 | 160090882 | 160090882 | C | T | SNP | <i>ATP1A2</i> | ENST00000478587 | 1 | 3_prime_untranslated_region | c.*160090882 | NULL | tier3 | 81 | 0 | 0.000 | 120 | 34 | 0.219 |
| 439820 | 1 | 186089086 | 186089086 | G | C | SNP | <i>HMCN1</i> | ENST00000271588 | 1 | intronic | c.12095-57 | e80-57 | tier3 | 317 | 0 | 0.000 | 549 | 90 | 0.141 |
| 439820 | 2 | 38295746 | 38295746 | C | T | SNP | <i>CYP1B1</i> | ENST00000260630 | -1 | 3_prime_untranslated_region | c.*2119 | NULL | tier3 | 143 | 2 | 0.014 | 170 | 130 | 0.433 |
| 439820 | 2 | 89292205 | 89292205 | C | T | SNP | <i>IGKV1-8</i> | ENST00000495489 | -1 | missense | c.68 | p.R23Q | tier1 | 265 | 0 | 0.000 | 396 | 33 | 0.077 |
| 439820 | 2 | 91927043 | 91927043 | T | C | SNP | <i>ENSG00000223703</i> | ENST00000445234 | 1 | 3_prime_untranslated_region | c.*919270 | | | | | | | | |

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|--------|----|-----------|-----------|-----|---|-----|------------------------|-----------------|----|-----------------------------|---------------|--------------------|-------|-----|----|-------|------|-----|-------|
| 439820 | 3 | 164783108 | 164783108 | G | A | SNP | <i>SI</i> | ENST00000264382 | -1 | missense | c.748 | p.R250C | tier1 | 376 | 1 | 0.003 | 620 | 99 | 0.138 |
| 439820 | 3 | 194146027 | 194146027 | A | G | SNP | <i>ATP13A3</i> | ENST00000256031 | -1 | intronic | c.3313+44 | e28+44 | tier3 | 26 | 2 | 0.071 | 50 | 48 | 0.490 |
| 439820 | 4 | 23796009 | 23796009 | G | A | SNP | <i>PPARGC1A</i> | ENST00000264867 | -1 | 3_prime_untranslated_region | c.*1436 | NULL | tier3 | 39 | 0 | 0.000 | 49 | 17 | 0.258 |
| 439820 | 4 | 159634313 | 159634313 | C | G | SNP | <i>PPID</i> | ENST00000307720 | -1 | silent | c.852 | p.L284 | tier1 | 99 | 0 | 0.000 | 156 | 13 | 0.077 |
| 439820 | 5 | 40998245 | 40998245 | C | T | SNP | <i>MROH2B</i> | ENST00000399564 | -1 | missense | c.4667 | p.R1556H | tier1 | 80 | 0 | 0.000 | 93 | 71 | 0.433 |
| 439820 | 5 | 81583110 | 81583110 | G | C | SNP | <i>ATP6AP1L</i> | ENST00000380167 | 1 | intronic | c.1+6280 | e0+6280 | tier3 | 113 | 0 | 0.000 | 183 | 15 | 0.075 |
| 439820 | 6 | 131941851 | 131941851 | C | A | SNP | <i>MED23</i> | ENST00000368058 | -1 | nonsense | c.514 | p.E172* | tier1 | 100 | 0 | 0.000 | 122 | 96 | 0.440 |
| 439820 | 6 | 146244843 | 146244843 | G | C | SNP | <i>SHPRH</i> | ENST00000367503 | -1 | missense | c.3508 | p.R1170G | tier1 | 108 | 0 | 0.000 | 127 | 109 | 0.460 |
| 439820 | 6 | 168466523 | 168466523 | G | C | SNP | <i>FRMD1</i> | ENST00000358587 | -1 | 5_prime_untranslated_region | c.-168466523 | NULL | tier3 | 55 | 0 | 0.000 | 66 | 13 | 0.165 |
| 439820 | 7 | 14217686 | 14217686 | C | T | SNP | <i>DGKB</i> | ENST00000258767 | -1 | missense | c.2216 | p.R739Q | tier1 | 64 | 1 | 0.015 | 49 | 34 | 0.410 |
| 439820 | 7 | 92888838 | 92888838 | G | A | SNP | <i>CCDC132</i> | ENST00000495039 | 1 | 3_prime_untranslated_region | c.*92888838 | NULL | tier3 | 76 | 0 | 0.000 | 127 | 102 | 0.444 |
| 439820 | 7 | 102427887 | 102427887 | T | C | SNP | <i>FAM185A</i> | ENST00000413034 | 1 | missense | c.1037 | p.V346A | tier1 | 205 | 0 | 0.000 | 296 | 15 | 0.048 |
| 439820 | 7 | 139045095 | 139045095 | T | G | SNP | <i>LUC7L2</i> | ENST00000541515 | 1 | intronic | c.259+14728 | e2+14728 | tier2 | 150 | 2 | 0.013 | 123 | 16 | 0.114 |
| 439820 | 8 | 2909983 | 2909983 | C | T | SNP | <i>CSMD1</i> | ENST00000520002 | -1 | splice_site | c.7663+1 | e51+1 | tier1 | 617 | 3 | 0.005 | 501 | 360 | 0.417 |
| 439820 | 8 | 52732287 | 52732287 | C | T | SNP | <i>PCMTD1</i> | ENST00000360540 | -1 | 3_prime_untranslated_region | c.*624 | NULL | tier3 | 120 | 1 | 0.008 | 224 | 21 | 0.085 |
| 439820 | 8 | 102555673 | 102555673 | C | A | SNP | <i>GRHL2</i> | ENST00000472106 | 1 | 3_prime_untranslated_region | c.*102555673 | NULL | tier3 | 51 | 0 | 0.000 | 61 | 42 | 0.408 |
| 439820 | 8 | 133192501 | 133192501 | C | T | SNP | <i>KCNQ3</i> | ENST00000388996 | -1 | missense | c.680 | p.R227Q | tier1 | 105 | 0 | 0.000 | 129 | 11 | 0.078 |
| 439820 | 9 | 39178241 | 39178241 | C | A | SNP | <i>CNTNAP3</i> | ENST00000297668 | -1 | nonsense | c.655 | p.G219* | tier1 | 212 | 0 | 0.000 | 454 | 130 | 0.222 |
| 439820 | 10 | 5925962 | 5925962 | G | A | SNP | <i>ANKRD16</i> | ENST00000380092 | -1 | silent | c.657 | p.D219 | tier1 | 45 | 0 | 0.000 | 53 | 35 | 0.398 |
| 439820 | 10 | 30316501 | 30316503 | CTG | - | DEL | <i>KIAA1462</i> | ENST00000375377 | -1 | in_frame_del | c.2576_2574 | p.S859in_frame_del | tier1 | 49 | 0 | 0.000 | 67 | 12 | 0.152 |
| 439820 | 10 | 46210670 | 46210670 | G | C | SNP | <i>FAM21FP</i> | ENST00000426241 | -1 | 5_prime_untranslated_region | c.-46210670 | NULL | tier2 | 220 | 0 | 0.000 | 454 | 28 | 0.058 |
| 439820 | 10 | 71627506 | 71627506 | C | T | SNP | <i>COL13A1</i> | ENST00000356340 | 1 | intronic | c.365-4426 | e3-4426 | tier2 | 49 | 1 | 0.020 | 57 | 36 | 0.387 |
| 439820 | 10 | 105073978 | 105073978 | C | - | DEL | <i>PCGF6</i> | ENST00000369847 | -1 | frame_shift_del | c.961 | p.E321fs | tier1 | 64 | 1 | 0.015 | 104 | 72 | 0.409 |
| 439820 | 10 | 114925313 | 114925313 | G | C | SNP | <i>TCF7L2</i> | ENST00000355995 | 1 | splice_site | c.1443-1 | e15-1 | tier1 | 78 | 0 | 0.000 | 96 | 18 | 0.158 |
| 439820 | 11 | 2906566 | 2906566 | C | A | SNP | <i>CDKN1C</i> | ENST00000414822 | -1 | nonsense | c.154 | p.E52* | tier1 | 87 | 0 | 0.000 | 96 | 53 | 0.353 |
| 439820 | 11 | 4360536 | 4360536 | C | G | SNP | <i>OR52B3P</i> | ENST00000314715 | 1 | 5_prime_flanking_region | c.-38976 | NULL | tier3 | 57 | 0 | 0.000 | 120 | 17 | 0.124 |
| 439820 | 11 | 57471814 | 57471814 | G | A | SNP | <i>MED19</i> | ENST00000431606 | -1 | missense | c.626 | p.P209L | tier1 | 55 | 3 | 0.052 | 88 | 61 | 0.407 |
| 439820 | 11 | 62364143 | 62364143 | T | C | SNP | <i>MTA2</i> | ENST00000278823 | -1 | missense | c.848 | p.Y283C | tier1 | 446 | 9 | 0.020 | 476 | 382 | 0.444 |
| 439820 | 11 | 124413410 | 124413410 | C | G | SNP | <i>OR8B12</i> | ENST00000306842 | -1 | silent | c.141 | p.L47 | tier1 | 654 | 0 | 0.000 | 1056 | 79 | 0.070 |
| 439820 | 11 | 124789263 | 124789263 | C | G | SNP | <i>HEPN1</i> | ENST00000408930 | 1 | 5_prime_untranslated_region | c.-384 | NULL | tier3 | 80 | 0 | 0.000 | 140 | 23 | 0.141 |
| 439820 | 12 | 6965657 | 6965657 | A | G | SNP | <i>USP5</i> | ENST00000229268 | 1 | intronic | c.584+43 | e5+43 | tier3 | 18 | 0 | 0.000 | 23 | 21 | 0.477 |
| 439820 | 12 | 47186868 | 47186868 | G | A | SNP | <i>SLC38A4</i> | ENST00000266579 | -1 | 5_prime_untranslated_region | c.-14 | NULL | tier3 | 62 | 0 | 0.000 | 93 | 75 | 0.446 |
| 439820 | 12 | 50026780 | 50026780 | C | G | SNP | <i>PRPF40B</i> | ENST00000548825 | 1 | intronic | c.349-17 | e7-17 | tier3 | 353 | 4 | 0.010 | 498 | 26 | 0.046 |
| 439820 | 12 | 51844830 | 51844830 | G | C | SNP | <i>SLC4A8</i> | ENST00000453097 | 1 | intronic | c.277+24 | e3+24 | tier3 | 163 | 0 | 0.000 | 237 | 15 | 0.060 |
| 439820 | 12 | 112888165 | 112888165 | C | G | SNP | <i>PTPN11</i> | ENST00000351677 | 1 | missense | c.181 | p.D61H | tier1 | 484 | 0 | 0.000 | 1051 | 46 | 0.042 |
| 439820 | 14 | 24518609 | 24518609 | A | G | SNP | <i>DHRS4L1</i> | ENST00000558293 | 1 | intronic | c.NULL | NULL | tier3 | 114 | 4 | 0.034 | 109 | 104 | 0.488 |
| 439820 | 14 | 24524337 | 24524337 | G | T | SNP | <i>LRRC16B</i> | ENST00000342740 | 1 | missense | c.523 | p.V175F | tier1 | 90 | 0 | 0.000 | 72 | 66 | 0.475 |
| 439820 | 14 | 100792064 | 100792064 | A | G | SNP | <i>SLC25A47</i> | ENST00000361529 | 1 | intronic | c.29-61 | e2-61 | tier3 | 22 | 0 | 0.000 | 32 | 23 | 0.418 |
| 439820 | 15 | 91438809 | 91438809 | G | C | SNP | <i>FES</i> | ENST00000328850 | 1 | 3_prime_untranslated_region | c.*21 | NULL | tier3 | 31 | 1 | 0.031 | 57 | 33 | 0.367 |
| 439820 | 16 | 2098338 | 2098338 | A | G | SNP | <i>TSC2</i> | ENST00000219476 | 1 | intronic | c.1-250 | e1-250 | tier2 | 98 | 1 | 0.010 | 123 | 88 | 0.417 |
| 439820 | 16 | 7761443 | 7761443 | G | A | SNP | <i>RBFOX1</i> | ENST00000547372 | 1 | 3_prime_untranslated_region | c.*1800 | NULL | tier3 | 101 | 0 | 0.000 | 144 | 28 | 0.159 |
| 439820 | 16 | 24582766 | 24582766 | G | A | SNP | <i>RBBP6</i> | ENST00000319715 | 1 | missense | c.4379 | p.R1460H | tier1 | 24 | 0 | 0.000 | 46 | 25 | 0.352 |
| 439820 | 16 | 29576812 | 29576812 | T | C | SNP | <i>ENSG00000205534</i> | ENST00000447140 | -1 | 5_prime_untranslated_region | c.-29576812 | NULL | tier3 | 115 | 0 | 0.000 | 143 | 121 | 0.458 |
| 439820 | 16 | 50835415 | 50835415 | A | G | SNP | <i>CYLD</i> | ENST00000427738 | 1 | 3_prime_untranslated_region | c.*4996 | NULL | tier3 | 75 | 1 | 0.013 | 72 | 68 | 0.486 |
| 439820 | 16 | 50835731 | 50835731 | C | G | SNP | <i>CYLD</i> | ENST00000427738 | 1 | 3_prime_untranslated_region | c.*5312 | NULL | tier3 | 66 | 0 | 0.000 | 83 | 52 | 0.382 |
| 439820 | 17 | 7577538 | 7577538 | C | T | SNP | <i>TP53</i> | ENST00000269305 | -1 | missense | c.743 | p.R248Q | tier1 | 606 | 5 | 0.008 | 558 | 600 | 0.518 |
| 439820 | 17 | 15942836 | 15942836 | G | C | SNP | <i>NCOR1</i> | ENST00000268712 | -1 | missense | c.6866 | p.P2289R | tier1 | 591 | 3 | 0.005 | 690 | 457 | 0.397 |
| 439820 | 18 | 7034550 | 7034550 | A | G | SNP | <i>LAMA1</i> | ENST00000389658 | -1 | missense | c.1979 | p.M660T | tier1 | 109 | 0 | 0.000 | 136 | 102 | 0.427 |
| 439820 | 18 | 14752893 | 14752893 | A | T | SNP | <i>ANKRD30B</i> | ENST00000358984 | 1 | missense | c.392 | p.D131V | tier1 | 111 | 2 | 0.018 | 161 | 104 | 0.393 |
| 439820 | 18 | 29046556 | 29046556 | G | C | SNP | <i>DSG3</i> | ENST00000257189 | 1 | missense | c.1475 | p.C492S | tier1 | 74 | 0 | 0.000 | 127 | 21 | 0.141 |
| 439820 | 18 | 34205430 | 34205430 | G | A | SNP | <i>FHOD3</i> | ENST00000257209 | 1 | intronic | c.958-44 | e10-44 | tier3 | 104 | 0 | 0.000 | 110 | 55 | 0.331 |
| 439820 | 18 | 42531907 | 42531907 | G | A | SNP | <i>SETBP1</i> | ENST00000282030 | 1 | missense | c.2602 | p.D868N | tier1 | 589 | 2 | 0.003 | 768 | 171 | 0.182 |
| 439820 | 19 | 1529504 | 1529504 | G | T | SNP | <i>PLK5</i> | ENST00000334770 | 1 | intronic | c.490+15 | e5+15 | tier3 | 107 | 0 | 0.000 | 175 | 90 | 0.338 |
| 439820 | 19 | 9007457 | 9007457 | C | G | SNP | <i>MUC16</i> | ENST00000397910 | -1 | intronic | c.39481+30 | e43+30 | tier3 | 633 | 0 | 0.000 | 1562 | 76 | 0.046 |
| 439820 | 19 | 42401048 | 42401048 | C | T | SNP | <i>ARHGEF1</i> | ENST00000337665 | 1 | intronic | c.1166+493 | e13+493 | tier4 | 180 | 2 | 0.011 | 111 | 87 | 0.437 |
| 439820 | 19 | 53873937 | 53873938 | - | A | INS | <i>ZNF525</i> | ENST00000474037 | 1 | intronic | c.1-1531_1530 | e1-1530 | tier3 | 10 | 0 | 0.000 | 24 | 24 | 0.500 |
| 439820 | 20 | 31022441 | 31022442 | - | G | INS | <i>ASXL1</i> | ENST00000375687 | 1 | frame_shift_ins | c.1926_1927 | p.G645fs | tier1 | 621 | 15 | 0.024 | 758 | 356 | 0.320 |
| 439820 | 20 | 61291782 | 61291782 | G | C | SNP | <i>SLCO4A1</i> | ENST00000217159 | 1 | missense | c.906 | p.E302D | tier1 | 70 | 0 | 0.000 | 61 | 14 | 0.187 |
| 439820 | 21 | 44514777 | 44514777 | T | G | SNP | <i>U2AF1</i> | ENST00000291552 | -1 | missense | c.470 | p.Q157P | tier1 | 312 | 1 | 0.003 | 308 | 229 | 0.425 |
| 439820 | X | 31462671 | 31462671 | G | A | SNP | <i>DMD</i> | ENST00000357033 | -1 | missense | c.9011 | p.T3004I | tier1 | 92 | 0 | 0.000 | 15 | 128 | 0.895 |
| 439820 | X | 110439784 | 110439784 | C | C | SNP | <i>PAK3</i> | ENST00000360648 | 1 | silent | c.1431 | p.P477 | tier1 | 106 | 0 | 0.000 | 176 | 20 | 0.101 |
| 439820 | X | 128781951 | 128781951 | C | G | SNP | <i>APLN</i> | ENST00000427399 | -1 | intronic | c.128-57 | e2-57 | tier3 | 48 | 0 | 0.000 | 96 | 17 | 0.150 |
| 439820 | X | 133511717 | 133511718 | AG | - | DEL | <i>PHF6</i> | ENST00000394292 | 1 | frame_shift_del | c.70_71 | p.D25fs | tier1 | 178 | 1 | 0.006 | 95 | 352 | 0.788 |
| 476807 | 1 | 26671642 | 26671642 | C | A | SNP | <i>AIM1L</i> | ENST00000374207 | -1 | intronic | c.169+204 | e2+204 | tier3 | 106 | 0 | 0.000 | 189 | 50 | 0.209 |
| 476807 | 1 | 94342696 | 94342696 | A | - | DEL | <i>DNTTIP2</i> | ENST00000436063 | -1 | frame_shift_del | c.795 | p.F265fs | tier1 | 31 | 0 | 0.000 | 89 | 39 | 0.305 |
| 476807 | 1 | 176525591 | 176525591 | T | C | SNP | <i>PAPPA2</i> | ENST00000367662 | 1 | silent | c.133 | p.L45 | tier1 | 88 | 0 | 0.000 | 126 | 32 | 0.203 |
| 476807 | 2 | 97833261 | 97833261 | T | A | SNP | <i>ANKRD36</i> | ENST00000420699 | 1 | intronic | c. | | | | | | | | |

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|--------|----|-----------|-----------|----------------------------|-----------|-----|-----------------|-----------------|----|-----------------------------|--------------|---------------------|-------|------|----|-------|------|-----|-------|
| 476807 | 4 | 126389815 | 126389815 | C | G | SNP | FAT4 | ENST00000394329 | 1 | missense | c.12048 | p.D4016E | tier1 | 126 | 2 | 0.016 | 210 | 104 | 0.329 |
| 476807 | 4 | 183601392 | 183601392 | T | C | SNP | TENM3 | ENST00000406950 | 1 | splice_region | c.1538-9 | e8-9 | tier3 | 54 | 1 | 0.018 | 105 | 56 | 0.346 |
| 476807 | 5 | 137589102 | 137589102 | G | A | SNP | GFRA3 | ENST00000274721 | -1 | intronic | c.1025-38 | e7-38 | tier3 | 151 | 2 | 0.013 | 160 | 64 | 0.286 |
| 476807 | 6 | 32007827 | 32007827 | C | A | SNP | CYP21A2 | ENST00000418967 | 1 | missense | c.784 | p.L262I | tier1 | 64 | 0 | 0.000 | 100 | 52 | 0.342 |
| 476807 | 7 | 75123378 | 75123378 | C | A | SNP | POM121C | ENST00000453279 | -1 | 5_prime_flanking_region | c.-52604 | NULL | tier3 | 268 | 0 | 0.000 | 502 | 23 | 0.044 |
| 476807 | 7 | 77764563 | 77764563 | C | T | SNP | MAGI2 | ENST00000354212 | -1 | intronic | c.2846-40 | e17-40 | tier3 | 280 | 5 | 0.018 | 429 | 203 | 0.320 |
| 476807 | 7 | 92764778 | 92764778 | A | C | SNP | SAMD9L | ENST00000318238 | -1 | missense | c.507 | p.D169E | tier1 | 30 | 0 | 0.000 | 92 | 48 | 0.343 |
| 476807 | 7 | 100684392 | 100684392 | C | T | SNP | MUC17 | ENST00000306151 | 1 | missense | c.9695 | p.T3232M | tier1 | 130 | 0 | 0.000 | 237 | 39 | 0.141 |
| 476807 | 8 | 74529737 | 74529737 | T | C | SNP | STAU2 | ENST00000524300 | -1 | intronic | c.411-51 | e4-51 | tier3 | 22 | 0 | 0.000 | 76 | 36 | 0.321 |
| 476807 | 11 | 35016453 | 35016453 | C | T | SNP | PDHX | ENST00000227868 | 1 | splice_region | c.1248-8 | e11-8 | tier3 | 73 | 1 | 0.014 | 129 | 79 | 0.380 |
| 476807 | 11 | 93417105 | 93417105 | C | T | SNP | KIAA1731 | ENST00000411936 | 1 | intronic | c.950-25 | e8-25 | tier3 | 69 | 0 | 0.000 | 152 | 27 | 0.151 |
| 476807 | 12 | 53670353 | 53670353 | G | A | SNP | ESPL1 | ENST00000257934 | 1 | intronic | c.1701-51 | e7-51 | tier3 | 68 | 5 | 0.069 | 87 | 48 | 0.356 |
| 476807 | 12 | 85460596 | 85460596 | A | G | SNP | LRR1Q1 | ENST00000393217 | 1 | missense | c.2615 | p.Q872R | tier1 | 55 | 2 | 0.035 | 248 | 138 | 0.358 |
| 476807 | 12 | 114377861 | 114377861 | G | T | SNP | RBM19 | ENST00000261741 | -1 | silent | c.1842 | p.G614 | tier1 | 71 | 4 | 0.053 | 90 | 59 | 0.396 |
| 476807 | 13 | 19407870 | 19407870 | G | T | SNP | ANKRD20A9P | ENST00000457997 | -1 | 5_prime_untranslated_region | c.-19407870 | NULL | tier3 | 143 | 0 | 0.000 | 404 | 215 | 0.346 |
| 476807 | 13 | 36347975 | 36347975 | T | C | SNP | DCLK1 | ENST00000360631 | -1 | 3_prime_untranslated_region | c.*697 | NULL | tier3 | 14 | 0 | 0.000 | 22 | 19 | 0.463 |
| 476807 | 15 | 21071137 | 21071137 | G | A | SNP | POTEB2 | ENST00000454856 | -1 | intronic | c.410+64 | e1+64 | tier3 | 780 | 5 | 0.006 | 1530 | 70 | 0.044 |
| 476807 | 15 | 38228468 | 38228468 | A | G | SNP | TMCO5A | ENST00000319669 | 1 | intronic | c.1-47 | e1-47 | tier3 | 45 | 1 | 0.022 | 103 | 63 | 0.375 |
| 476807 | 15 | 87097682 | 87097682 | C | T | SNP | AGBL1 | ENST00000441037 | 1 | missense | c.2770 | p.R924W | tier1 | 61 | 1 | 0.016 | 116 | 30 | 0.206 |
| 476807 | 15 | 90631934 | 90631934 | C | T | SNP | IDH2 | ENST00000330062 | -1 | missense | c.419 | p.R140Q | tier1 | 555 | 6 | 0.011 | 597 | 311 | 0.341 |
| 476807 | 17 | 9066111 | 9066111 | T | C | SNP | NTN1 | ENST00000173229 | 1 | intronic | c.1019-19 | e2-19 | tier3 | 80 | 0 | 0.000 | 122 | 51 | 0.295 |
| 476807 | 17 | 74732959 | 74732959 | G | T | SNP | SRSF2 | ENST00000359995 | -1 | missense | c.284 | p.P95H | tier1 | 332 | 1 | 0.003 | 402 | 203 | 0.336 |
| 476807 | 18 | 14513621 | 14513621 | T | C | SNP | POTEC | ENST00000358970 | -1 | intronic | c.1533+40 | e10+40 | tier3 | 102 | 0 | 0.000 | 474 | 32 | 0.063 |
| 476807 | 19 | 8654876 | 8654876 | C | T | SNP | ADAMTS10 | ENST00000270328 | -1 | splice_site | c.1798-1 | e14-1 | tier1 | 48 | 1 | 0.020 | 60 | 31 | 0.337 |
| 476807 | 19 | 36321785 | 36321785 | C | T | SNP | NPHS1 | ENST00000378910 | -1 | silent | c.3555 | p.P1185 | tier1 | 78 | 0 | 0.000 | 101 | 34 | 0.252 |
| 476807 | 19 | 56303709 | 56303709 | G | A | SNP | NLRP11 | ENST00000443188 | -1 | missense | c.2471 | p.T824M | tier1 | 99 | 1 | 0.010 | 202 | 23 | 0.102 |
| 476807 | 20 | 31022403 | 31022425 | CACCACTGCCATAGA GAGCGGC | - | DEL | ASXL1 | ENST00000375687 | 1 | frame_shift_del | c.1888_1910 | p.H633fs | tier1 | 747 | 3 | 0.004 | 1000 | 114 | 0.102 |
| 476807 | 20 | 31531576 | 31531576 | C | T | SNP | EFCAB8 | ENST00000457731 | 1 | 5_prime_flanking_region | c.-16075 | NULL | tier3 | 94 | 0 | 0.000 | 119 | 68 | 0.362 |
| 476807 | 20 | 48914916 | 48914916 | G | T | SNP | LOC284751 | ENST00000371639 | 1 | rna | NULL | NULL | tier1 | 73 | 1 | 0.014 | 91 | 48 | 0.343 |
| 476807 | 22 | 16157940 | 16157940 | C | T | SNP | ENSG00000206195 | ENST00000413768 | -1 | intronic | c.NULL | NULL | tier2 | 28 | 0 | 0.000 | 33 | 10 | 0.233 |
| 476807 | X | 123181343 | 123181344 | - | AAGCGAAAG | INS | STAG2 | ENST00000218089 | 1 | frame_shift_ins | c.807_808 | p.L273fs | tier1 | 169 | 0 | 0.000 | 281 | 34 | 0.108 |
| 480109 | 1 | 180789156 | 180789156 | A | - | DEL | XPR1 | ENST00000367590 | 1 | intronic | c.764-4733 | e8-4733 | tier4 | 48 | 0 | 0.000 | 72 | 14 | 0.163 |
| 480109 | 2 | 55177887 | 55177887 | C | T | SNP | EML6 | ENST00000356458 | 1 | missense | c.4184 | p.A1395V | tier1 | 93 | 0 | 0.000 | 105 | 57 | 0.352 |
| 480109 | 2 | 172951837 | 172951837 | G | T | SNP | DLX1 | ENST00000409492 | 1 | 3_prime_untranslated_region | c.*172951837 | NULL | tier2 | 32 | 0 | 0.000 | 45 | 18 | 0.286 |
| 480109 | 3 | 3099496 | 3099496 | A | - | DEL | CNTN4 | ENST00000397461 | 1 | 3_prime_untranslated_region | c.*1592 | NULL | tier2 | 142 | 1 | 0.007 | 195 | 21 | 0.097 |
| 480109 | 4 | 106588612 | 106588612 | T | C | SNP | ARHGEF38 | ENST00000420470 | 1 | missense | c.1900 | p.Y634H | tier1 | 83 | 1 | 0.012 | 71 | 22 | 0.234 |
| 480109 | 4 | 153784943 | 153784943 | A | C | SNP | ARFIP1 | ENST00000353617 | 1 | intronic | c.202+77 | e2+77 | tier3 | 84 | 1 | 0.012 | 83 | 49 | 0.371 |
| 480109 | 5 | 38489182 | 38489182 | G | T | SNP | LIFR | ENST00000263409 | -1 | nonsense | c.2333 | p.S778* | tier1 | 107 | 0 | 0.000 | 95 | 34 | 0.264 |
| 480109 | 5 | 67597268 | 67597268 | G | A | SNP | PIK3R1 | ENST00000396611 | 1 | 3_prime_untranslated_region | c.*3839 | NULL | tier4 | 81 | 0 | 0.000 | 111 | 12 | 0.098 |
| 480109 | 5 | 118834979 | 118834979 | A | G | SNP | HSD17B4 | ENST00000256216 | 1 | intronic | c.973-33 | e13-33 | tier3 | 99 | 0 | 0.000 | 35 | 37 | 0.514 |
| 480109 | 6 | 87969820 | 87969822 | AAG | - | DEL | ZNF292 | ENST00000369577 | 1 | in_frame_del | c.6473_6475 | p.E2159in_frame_del | tier1 | 55 | 0 | 0.000 | 98 | 26 | 0.210 |
| 480109 | 7 | 36671732 | 36671732 | C | A | SNP | AOAH | ENST00000258749 | -1 | intronic | c.451-20 | e6-20 | tier3 | 95 | 0 | 0.000 | 157 | 13 | 0.077 |
| 480109 | 7 | 116199666 | 116199667 | - | G | INS | CAV1 | ENST00000341049 | 1 | 3_prime_untranslated_region | c.*325 | NULL | tier3 | 57 | 0 | 0.000 | 59 | 11 | 0.157 |
| 480109 | 9 | 5073770 | 5073770 | G | T | SNP | JAK2 | ENST00000381652 | 1 | missense | c.1849 | p.V617F | tier1 | 568 | 2 | 0.004 | 593 | 251 | 0.297 |
| 480109 | 9 | 125282293 | 125282293 | C | T | SNP | OR1J4 | ENST00000340750 | 1 | silent | c.874 | p.L292 | tier1 | 27 | 0 | 0.000 | 58 | 33 | 0.363 |
| 480109 | 11 | 24927450 | 24927450 | G | C | SNP | LUZP2 | ENST00000336930 | 1 | intronic | c.397-87 | e6-87 | tier3 | 73 | 0 | 0.000 | 81 | 24 | 0.229 |
| 480109 | 11 | 65655168 | 65655168 | C | T | SNP | FIBP | ENST00000338369 | -1 | missense | c.314 | p.R105Q | tier1 | 71 | 1 | 0.014 | 138 | 33 | 0.193 |
| 480109 | 11 | 112071453 | 112071453 | A | G | SNP | BCO2 | ENST00000357685 | 1 | missense | c.983 | p.Q328R | tier1 | 100 | 2 | 0.020 | 160 | 74 | 0.316 |
| 480109 | 12 | 6701122 | 6701122 | T | C | SNP | CHD4 | ENST00000309577 | -1 | missense | c.3050 | p.K1017R | tier1 | 384 | 2 | 0.005 | 494 | 174 | 0.260 |
| 480109 | 12 | 40689290 | 40689290 | G | C | SNP | LRRK2 | ENST00000298910 | 1 | missense | c.2940 | p.E980D | tier1 | 78 | 1 | 0.012 | 81 | 22 | 0.212 |
| 480109 | 12 | 122983394 | 122983394 | C | T | SNP | ZCCHC8 | ENST00000336229 | -1 | silent | c.222 | p.L74 | tier1 | 83 | 1 | 0.012 | 153 | 67 | 0.305 |
| 480109 | 14 | 21862072 | 21862072 | C | T | SNP | CHD8 | ENST00000399982 | -1 | missense | c.5882 | p.R1961H | tier1 | 28 | 0 | 0.000 | 35 | 22 | 0.386 |
| 480109 | 15 | 85742643 | 85742643 | A | G | SNP | CSPG4P12 | ENST00000559989 | 1 | intronic | c.NULL | NULL | tier3 | 19 | 1 | 0.050 | 12 | 12 | 0.500 |
| 480109 | 16 | 32956973 | 32956973 | G | T | SNP | ENSG00000260921 | ENST00000568904 | 1 | 5_prime_flanking_region | c.-49885 | NULL | tier3 | 280 | 2 | 0.007 | 469 | 84 | 0.152 |
| 480109 | 16 | 57935363 | 57935363 | G | C | SNP | CNGB1 | ENST00000251102 | -1 | intronic | c.2893-24 | e28-24 | tier3 | 48 | 0 | 0.000 | 49 | 18 | 0.265 |
| 480109 | 17 | 7577114 | 7577114 | C | T | SNP | TP53 | ENST00000269305 | -1 | missense | c.824 | p.C275Y | tier1 | 621 | 7 | 0.011 | 914 | 468 | 0.338 |
| 480109 | 17 | 16829225 | 16829225 | A | C | SNP | TBC1D27 | ENST00000261651 | -1 | rna | NULL | NULL | tier1 | 82 | 1 | 0.012 | 120 | 59 | 0.328 |
| 480109 | 19 | 18981487 | 18981487 | T | - | DEL | GDF1 | ENST00000247005 | -1 | intronic | c.1-59 | e1-59 | tier2 | 15 | 0 | 0.000 | 69 | 33 | 0.324 |
| 480109 | 20 | 43744353 | 43744353 | G | C | SNP | KCN51 | ENST00000306117 | -1 | 5_prime_flanking_region | c.-16476 | NULL | tier3 | 119 | 1 | 0.008 | 117 | 51 | 0.304 |
| 480109 | 21 | 32491425 | 32491425 | T | A | SNP | TIAM1 | ENST00000286827 | -1 | 3_prime_untranslated_region | c.*1261 | NULL | tier4 | 99 | 0 | 0.000 | 134 | 12 | 0.082 |
| 480109 | 21 | 44010250 | 44010251 | - | G | INS | ENSG00000235772 | ENST00000442605 | -1 | 5_prime_flanking_region | c.-28206 | NULL | tier2 | 19 | 0 | 0.000 | 20 | 12 | 0.375 |
| 480109 | 21 | 44010254 | 44010255 | TT | - | DEL | ENSG00000235772 | ENST00000442605 | -1 | 5_prime_flanking_region | c.-28210 | NULL | tier2 | 19 | 0 | 0.000 | 17 | 12 | 0.414 |
| 480109 | 21 | 44010258 | 44010259 | - | T | INS | ENSG00000235772 | ENST00000442605 | -1 | 5_prime_flanking_region | c.-28214 | NULL | tier2 | 26 | 0 | 0.000 | 19 | 12 | 0.387 |
| 480109 | 22 | 42046726 | 42046726 | G | C | SNP | XRCC6 | ENST00000359308 | 1 | splice_site | c.961-1 | e7-1 | tier1 | 112 | 0 | 0.000 | 168 | 91 | 0.351 |
| 480109 | X | 8538673 | 8538673 | G | T | SNP | KAL1 | ENST00000262648 | -1 | missense | c.929 | p.T310N | tier1 | 55 | 2 | 0.035 | 67 | 36 | 0.350 |
| 480109 | X | 153042601 | 153042601 | A | G | SNP | PLXNB3 | ENST00000538966 | 1 | intronic | c.4997-62 | e29-62 | tier3 | 29 | 0 | 0.000 | 52 | 17 | 0.243 |
| 481668 | 1 | 16262371 | 16262371 | C | T | SNP | SPEN | ENST00000375759 | 1 | silent | c.9636 | p.A3212 | tier1 | 726 | 8 | 0.011 | 901 | 341 | 0.273 |
| 481668 | 1 | 17256868 | 17256868 | G | A | SNP | CROCC | ENST00000375541 | 1 | intronic | c.683+26 | e6+26 | tier3 | 1326 | 12 | 0.009 | 2026 | 325 | 0.138 |
| 481668 | 1 | 467 | | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|----|---|-----|-----------------|-----------------|----|-----------------------------|-----------------|----------|-------|-----|----|-------|------|-----|-------|
| 481668 | 1 | 247094587 | 247094587 | T | C | SNP | AHCTF1 | ENST00000366508 | -1 | 5_prime_untranslated_region | c.-59 | NULL | tier2 | 34 | 0 | 0.000 | 37 | 9 | 0.196 |
| 481668 | 2 | 1457512 | 1457512 | C | G | SNP | TPO | ENST00000329066 | 1 | missense | c.529 | p.L177V | tier1 | 111 | 0 | 0.000 | 119 | 36 | 0.232 |
| 481668 | 2 | 61009939 | 61009939 | T | - | DEL | PAPOLG | ENST00000238714 | 1 | intronic | c.1112+34 | e12+34 | tier3 | 68 | 0 | 0.000 | 131 | 17 | 0.115 |
| 481668 | 2 | 98853198 | 98853198 | A | G | SNP | VWA3B | ENST00000485216 | 1 | rna | NULL | NULL | tier1 | 69 | 2 | 0.028 | 81 | 31 | 0.277 |
| 481668 | 4 | 15070967 | 15070968 | - | A | INS | CPEB2 | ENST00000538197 | 1 | 3_prime_untranslated_region | c.*2963 | NULL | tier2 | 104 | 2 | 0.019 | 179 | 57 | 0.242 |
| 481668 | 4 | 95223388 | 95223388 | A | G | SNP | HPGDS | ENST00000295256 | -1 | missense | c.344 | p.M115T | tier1 | 94 | 1 | 0.011 | 146 | 46 | 0.240 |
| 481668 | 5 | 40959580 | 40959580 | T | C | SNP | C7 | ENST00000313164 | 1 | missense | c.1519 | p.S507P | tier1 | 92 | 4 | 0.042 | 130 | 47 | 0.266 |
| 481668 | 5 | 41862863 | 41862863 | A | - | DEL | OXCT1 | ENST00000196371 | -1 | intronic | c.79-11 | e2-11 | tier4 | 93 | 0 | 0.000 | 135 | 19 | 0.123 |
| 481668 | 6 | 325150 | 325150 | T | C | SNP | DUSP22 | ENST00000419235 | 1 | intronic | c.139-9964 | e4-9964 | tier3 | 459 | 16 | 0.034 | 630 | 84 | 0.117 |
| 481668 | 6 | 31760727 | 31760727 | G | C | SNP | VARS | ENST00000495010 | -1 | 5_prime_untranslated_region | c.-31760727 | NULL | tier3 | 96 | 2 | 0.020 | 117 | 46 | 0.281 |
| 481668 | 6 | 135513482 | 135513482 | A | G | SNP | MYB | ENST00000341911 | 1 | missense | c.548 | p.N183S | tier1 | 122 | 0 | 0.000 | 131 | 47 | 0.264 |
| 481668 | 7 | 34118627 | 34118627 | C | T | SNP | BMPER | ENST00000297161 | 1 | missense | c.1237 | p.R413C | tier1 | 738 | 0 | 0.000 | 1026 | 53 | 0.049 |
| 481668 | 7 | 81593304 | 81593304 | T | G | SNP | CACNA2D1 | ENST00000356253 | -1 | intronic | c.2816+68 | e34+68 | tier3 | 51 | 0 | 0.000 | 46 | 26 | 0.361 |
| 481668 | 7 | 100635373 | 100635373 | C | A | SNP | MUC12 | ENST00000536621 | 1 | missense | c.1529 | p.A510D | tier1 | 285 | 1 | 0.004 | 519 | 68 | 0.116 |
| 481668 | 7 | 148506166 | 148506166 | T | A | SNP | EZH2 | ENST00000320356 | -1 | missense | c.2192 | p.Y731F | tier1 | 440 | 1 | 0.002 | 646 | 222 | 0.255 |
| 481668 | 9 | 117138957 | 117138957 | C | T | SNP | AKNA | ENST00000307564 | -1 | missense | c.1130 | p.R377H | tier1 | 350 | 0 | 0.000 | 729 | 24 | 0.032 |
| 481668 | 10 | 128933807 | 128933807 | T | C | SNP | FAM196A | ENST00000522781 | -1 | 3_prime_untranslated_region | c.*2294 | NULL | tier3 | 167 | 0 | 0.000 | 214 | 75 | 0.259 |
| 481668 | 10 | 133761131 | 133761131 | G | T | SNP | PPP2R2D | ENST00000455566 | 1 | missense | c.825 | p.M275I | tier1 | 148 | 1 | 0.007 | 213 | 57 | 0.211 |
| 481668 | 11 | 71903432 | 71903432 | A | G | SNP | FOLR1 | ENST00000312293 | 1 | intronic | c.168+47 | e1+47 | tier3 | 63 | 0 | 0.000 | 70 | 26 | 0.271 |
| 481668 | 12 | 21608977 | 21608977 | G | T | SNP | PYROXD1 | ENST00000240651 | 1 | missense | c.698 | p.S233I | tier1 | 91 | 2 | 0.022 | 141 | 54 | 0.277 |
| 481668 | 12 | 57929244 | 57929245 | AC | - | DEL | DCTN2 | ENST00000551142 | -1 | 5_prime_untranslated_region | c.-57929244 | NULL | tier3 | 91 | 0 | 0.000 | 137 | 17 | 0.110 |
| 481668 | 12 | 113599210 | 113599210 | C | A | SNP | DDX54 | ENST00000549271 | -1 | rna | NULL | NULL | tier1 | 109 | 0 | 0.000 | 122 | 26 | 0.176 |
| 481668 | 13 | 33223009 | 33223009 | C | T | SNP | PDS5B | ENST00000315596 | 1 | nonsense | c.100 | p.R34* | tier1 | 77 | 1 | 0.013 | 130 | 64 | 0.327 |
| 481668 | 14 | 50895475 | 50895476 | - | A | INS | MAP4K5 | ENST00000013125 | -1 | splice_region_ins | c.2234-4_2234-3 | e28-3 | tier3 | 69 | 0 | 0.000 | 144 | 16 | 0.100 |
| 481668 | 15 | 56704519 | 56704519 | G | A | SNP | TEX9 | ENST00000352903 | 1 | missense | c.848 | p.R283K | tier1 | 167 | 0 | 0.000 | 216 | 98 | 0.312 |
| 481668 | 15 | 74889854 | 74889855 | AC | - | DEL | ARID3B | ENST00000346246 | 1 | 3_prime_untranslated_region | c.*1739 | NULL | tier4 | 53 | 0 | 0.000 | 67 | 11 | 0.141 |
| 481668 | 16 | 30456662 | 30456662 | C | T | SNP | SEPHS2 | ENST00000542752 | -1 | silent | c.216 | p.G72 | tier1 | 82 | 0 | 0.000 | 84 | 23 | 0.215 |
| 481668 | 17 | 1559855 | 1559855 | T | C | SNP | PRPF8 | ENST00000304992 | -1 | missense | c.5624 | p.H1875R | tier1 | 651 | 6 | 0.009 | 997 | 410 | 0.290 |
| 481668 | 17 | 37425012 | 37425012 | A | G | SNP | FBXL20 | ENST00000264658 | -1 | intronic | c.933+68 | e12+68 | tier3 | 63 | 2 | 0.031 | 86 | 46 | 0.349 |
| 481668 | 17 | 37879957 | 37879957 | C | T | SNP | ERBB2 | ENST00000269571 | 1 | intronic | c.2208+44 | e18+44 | tier3 | 41 | 0 | 0.000 | 31 | 22 | 0.400 |
| 481668 | 19 | 9067438 | 9067438 | C | T | SNP | MUC16 | ENST00000397910 | -1 | missense | c.20008 | p.V6670I | tier1 | 906 | 9 | 0.010 | 1848 | 610 | 0.247 |
| 481668 | 19 | 15219913 | 15219913 | C | G | SNP | SYDE1 | ENST00000342784 | 1 | silent | c.135 | p.P45 | tier1 | 38 | 1 | 0.026 | 30 | 28 | 0.483 |
| 481668 | 20 | 2552803 | 2552803 | T | - | DEL | TMC2 | ENST00000358864 | 1 | intronic | c.555-22 | e5-22 | tier3 | 100 | 2 | 0.020 | 167 | 26 | 0.135 |
| 481668 | 20 | 2552806 | 2552806 | T | A | SNP | TMC2 | ENST00000358864 | 1 | intronic | c.555-19 | e5-19 | tier3 | 103 | 2 | 0.019 | 165 | 28 | 0.144 |
| 485523 | 1 | 110009655 | 110009655 | C | - | DEL | SYPL2 | ENST00000369872 | 1 | intronic | c.55-26 | e2-26 | tier2 | 10 | 0 | 0.000 | 8 | 13 | 0.619 |
| 485523 | 4 | 39474726 | 39474726 | G | A | SNP | LIAS | ENST00000261434 | 1 | missense | c.961 | p.E321K | tier1 | 148 | 0 | 0.000 | 242 | 17 | 0.066 |
| 485523 | 4 | 55976980 | 55976980 | G | A | SNP | KDR | ENST00000263923 | -1 | intronic | c.977-45 | e8-45 | tier3 | 266 | 0 | 0.000 | 310 | 23 | 0.069 |
| 485523 | 5 | 23527469 | 23527469 | C | A | SNP | PRDM9 | ENST00000296682 | 1 | missense | c.2272 | p.R758S | tier1 | 165 | 2 | 0.012 | 207 | 18 | 0.080 |
| 485523 | 5 | 54552381 | 54552381 | A | - | DEL | DHX29 | ENST00000251636 | -1 | intronic | c.4058-13 | e27-13 | tier3 | 78 | 0 | 0.000 | 79 | 10 | 0.112 |
| 485523 | 5 | 176939370 | 176939370 | C | T | SNP | DDX41 | ENST00000507955 | -1 | missense | c.1574 | p.R525H | tier1 | 524 | 0 | 0.000 | 677 | 65 | 0.088 |
| 485523 | 6 | 73904728 | 73904728 | A | T | SNP | KCNQ5 | ENST00000402622 | 1 | missense | c.2420 | p.N807I | tier1 | 82 | 0 | 0.000 | 156 | 18 | 0.103 |
| 485523 | 9 | 15472612 | 15472612 | A | - | DEL | PSIP1 | ENST00000495873 | -1 | rna | NULL | NULL | tier1 | 82 | 0 | 0.000 | 95 | 12 | 0.112 |
| 485523 | 14 | 24517447 | 24517447 | G | A | SNP | DHRS4L1 | ENST00000558293 | 1 | rna | NULL | NULL | tier1 | 152 | 0 | 0.000 | 248 | 20 | 0.074 |
| 485523 | 15 | 75706652 | 75706652 | C | T | SNP | SIN3A | ENST00000360439 | -1 | missense | c.367 | p.V123M | tier1 | 46 | 0 | 0.000 | 39 | 10 | 0.204 |
| 485523 | 18 | 47462642 | 47462642 | G | A | SNP | MYO5B | ENST00000285039 | -1 | silent | c.1983 | p.N661 | tier1 | 350 | 0 | 0.000 | 555 | 54 | 0.089 |
| 485523 | 19 | 7805616 | 7805616 | G | C | SNP | CD209 | ENST00000315599 | -1 | 3_prime_untranslated_region | c.*2309 | NULL | tier3 | 497 | 0 | 0.000 | 835 | 27 | 0.031 |
| 489196 | 1 | 1871145 | 1871145 | G | A | SNP | C1orf222 | ENST00000493964 | -1 | missense | c.614 | p.P205L | tier1 | 103 | 0 | 0.000 | 48 | 51 | 0.515 |
| 489196 | 1 | 116386058 | 116386058 | C | T | SNP | NHLH2 | ENST00000369506 | -1 | 5_prime_untranslated_region | c.-5065 | NULL | tier4 | 134 | 0 | 0.000 | 218 | 78 | 0.262 |
| 489196 | 1 | 248651829 | 248651829 | A | T | SNP | OR2G6 | ENST00000343414 | 1 | 5_prime_flanking_region | c.-33119 | NULL | tier3 | 230 | 1 | 0.004 | 253 | 52 | 0.171 |
| 489196 | 2 | 172930961 | 172930961 | A | G | SNP | METAP1D | ENST00000315796 | 1 | intronic | c.498-31 | e5-31 | tier3 | 37 | 1 | 0.026 | 62 | 34 | 0.354 |
| 489196 | 2 | 178483636 | 178483636 | C | T | SNP | TTA30A | ENST00000355689 | -1 | 5_prime_untranslated_region | c.-207 | NULL | tier2 | 102 | 0 | 0.000 | 104 | 37 | 0.262 |
| 489196 | 2 | 196863801 | 196863801 | G | T | SNP | DNAH7 | ENST00000410072 | -1 | 3_prime_untranslated_region | c.*13 | NULL | tier4 | 90 | 0 | 0.000 | 106 | 29 | 0.215 |
| 489196 | 2 | 219501054 | 219501054 | C | T | SNP | PLCD4 | ENST00000417849 | 1 | silent | c.2121 | p.D707 | tier1 | 100 | 0 | 0.000 | 95 | 36 | 0.275 |
| 489196 | 2 | 233407276 | 233407276 | C | T | SNP | CHRNA7 | ENST00000389494 | 1 | intronic | c.604+46 | e6+46 | tier3 | 32 | 0 | 0.000 | 26 | 18 | 0.409 |
| 489196 | 2 | 234591299 | 234591299 | T | C | SNP | UGT1A7 | ENST00000373426 | 1 | missense | c.716 | p.V239A | tier1 | 117 | 1 | 0.008 | 159 | 19 | 0.107 |
| 489196 | 3 | 77607305 | 77607305 | G | A | SNP | ROBO2 | ENST00000461745 | 1 | splice_region | c.1437+5 | e9+5 | tier3 | 44 | 0 | 0.000 | 57 | 27 | 0.321 |
| 489196 | 3 | 113327118 | 113327118 | G | T | SNP | SIDT1 | ENST00000393830 | 1 | intronic | c.1602+54 | e16+54 | tier3 | 67 | 0 | 0.000 | 49 | 22 | 0.310 |
| 489196 | 4 | 190876351 | 190876351 | G | C | SNP | FRG1 | ENST00000226798 | 1 | intronic | c.432+45 | e5+45 | tier3 | 169 | 2 | 0.012 | 329 | 26 | 0.073 |
| 489196 | 5 | 32135733 | 32135733 | A | G | SNP | GOLPH3 | ENST00000265070 | -1 | silent | c.417 | p.H139 | tier1 | 117 | 0 | 0.000 | 248 | 39 | 0.135 |
| 489196 | 5 | 122713937 | 122713937 | G | A | SNP | CEP120 | ENST00000306467 | -1 | intronic | c.2196+93 | e15+93 | tier4 | 93 | 0 | 0.000 | 163 | 28 | 0.147 |
| 489196 | 5 | 140768657 | 140768657 | A | T | SNP | PCDHGB4 | ENST00000519479 | 1 | missense | c.1206 | p.L402F | tier1 | 82 | 1 | 0.012 | 79 | 48 | 0.378 |
| 489196 | 6 | 2623069 | 2623069 | C | T | SNP | C6orf195 | ENST00000296847 | -1 | 3_prime_untranslated_region | c.*604 | NULL | tier3 | 142 | 0 | 0.000 | 124 | 66 | 0.347 |
| 489196 | 6 | 30954600 | 30954600 | A | C | SNP | MUC21 | ENST00000376296 | 1 | missense | c.648 | p.R216S | tier1 | 172 | 5 | 0.028 | 213 | 34 | 0.136 |
| 489196 | 6 | 35761203 | 35761203 | A | - | DEL | CLPSL1 | ENST00000428710 | 1 | intronic | c.82-62 | e2-62 | tier4 | 63 | 1 | 0.016 | 55 | 12 | 0.179 |
| 489196 | 6 | 36685089 | 36685089 | G | A | SNP | RAB44 | ENST00000229824 | 1 | missense | c.215 | p.R72K | tier1 | 124 | 1 | 0.008 | 98 | 40 | 0.290 |
| 489196 | 6 | 62611335 | 62611335 | A | - | DEL | KHDRBS2 | ENST00000281156 | -1 | intronic | c.484-59 | e5-59 | tier3 | 29 | 0 | 0.000 | 43 | 13 | 0.232 |
| 489196 | 6 | 112671088 | 112671088 | G | T | SNP | RFPL4B | ENST00000441065 | 1 | missense | c.178 | p.A60S | tier1 | 99 | 0 | 0.000 | 77 | 36 | 0.319 |
| 489196 | 6 | 143094282 | 143094282 | C | T | SNP | HIVEP2 | ENST00000012134 | -1 | missense | c.1594 | p.E532K | tier1 | 142 | 0 | 0.000 | 143 | 45 | 0.239 |
| 489196 | 7 | 23625386 | 23625386 | G | A | SNP | ENSG00000232553 | ENST00000416636 | -1 | 5_prime_untranslated_region | c.-23625386 | NULL | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|---|-----|-----|------------------------|-----------------|----|-----------------------------|--------------|----------|-------|-----|----|-------|------|-----|-------|
| 489196 | 8 | 87519260 | 87519260 | T | C | SNP | <i>RMDN1</i> | ENST00000406452 | -1 | missense | c.211 | p.I71V | tier1 | 169 | 0 | 0.000 | 275 | 99 | 0.264 |
| 489196 | 8 | 126011647 | 126011647 | T | C | SNP | <i>SQLI</i> | ENST00000265896 | 1 | missense | c.2 | p.M1T | tier1 | 79 | 0 | 0.000 | 80 | 28 | 0.259 |
| 489196 | 9 | 127300323 | 127300323 | G | C | SNP | <i>NR6A1</i> | ENST00000487099 | -1 | intronic | c.824+48 | e6+48 | tier3 | 58 | 2 | 0.033 | 38 | 23 | 0.377 |
| 489196 | 10 | 23326305 | 23326305 | G | A | SNP | <i>ARMC3</i> | ENST00000298032 | 1 | missense | c.2516 | p.G839E | tier1 | 139 | 0 | 0.000 | 98 | 49 | 0.333 |
| 489196 | 10 | 79797037 | 79797037 | A | G | SNP | <i>RPS24</i> | ENST00000440692 | 1 | missense | c.365 | p.K122R | tier1 | 68 | 0 | 0.000 | 73 | 27 | 0.270 |
| 489196 | 11 | 20676288 | 20676288 | G | A | SNP | <i>SLC6A5</i> | ENST00000525748 | 1 | silent | c.2268 | p.P756 | tier1 | 128 | 0 | 0.000 | 118 | 47 | 0.285 |
| 489196 | 11 | 46400551 | 46400551 | T | A | SNP | <i>DGKZ</i> | ENST00000454345 | 1 | missense | c.2993 | p.L998Q | tier1 | 19 | 0 | 0.000 | 24 | 18 | 0.429 |
| 489196 | 11 | 48454279 | 48454279 | G | A | SNP | <i>OR4C10P</i> | ENST00000434991 | -1 | 5_prime_untranslated_region | c.-48454279 | NULL | tier3 | 238 | 0 | 0.000 | 205 | 87 | 0.298 |
| 489196 | 11 | 56000330 | 56000330 | C | T | SNP | <i>OR5T2</i> | ENST00000313264 | -1 | missense | c.332 | p.C111Y | tier1 | 33 | 0 | 0.000 | 72 | 29 | 0.287 |
| 489196 | 12 | 9007449 | 9007449 | T | C | SNP | <i>A2ML1</i> | ENST00000299698 | 1 | intronic | c.2764+22 | e22+22 | tier3 | 176 | 0 | 0.000 | 162 | 56 | 0.257 |
| 489196 | 12 | 52946663 | 52946663 | C | T | SNP | <i>KRT71</i> | ENST00000267119 | -1 | missense | c.199 | p.G67R | tier1 | 225 | 1 | 0.004 | 202 | 82 | 0.289 |
| 489196 | 12 | 54686621 | 54686621 | C | T | SNP | <i>NFE2</i> | ENST00000312156 | -1 | missense | c.659 | p.G220E | tier1 | 76 | 0 | 0.000 | 87 | 29 | 0.250 |
| 489196 | 12 | 56110725 | 56110725 | A | G | SNP | <i>BLOC1S1</i> | ENST00000548925 | 1 | missense | c.154 | p.R52G | tier1 | 116 | 0 | 0.000 | 107 | 42 | 0.282 |
| 489196 | 12 | 76461257 | 76461257 | - | A | INS | <i>NAP1L1</i> | ENST00000551524 | -1 | 5_prime_untranslated_region | c.-24 | NULL | tier3 | 18 | 0 | 0.000 | 33 | 17 | 0.340 |
| 489196 | 13 | 28009534 | 28009534 | T | C | SNP | <i>GTF3A</i> | ENST00000419181 | 1 | intronic | c.567-36 | e6-36 | tier3 | 31 | 0 | 0.000 | 57 | 22 | 0.279 |
| 489196 | 13 | 52863939 | 52863939 | A | G | SNP | <i>LOC101930578</i> | ENST00000451298 | -1 | intronic | NULL | NULL | tier3 | 306 | 3 | 0.010 | 435 | 26 | 0.056 |
| 489196 | 14 | 59670661 | 59670661 | G | T | SNP | <i>DAAM1</i> | ENST00000351081 | 1 | intronic | c.1+15175 | e0+15175 | tier3 | 58 | 0 | 0.000 | 121 | 37 | 0.234 |
| 489196 | 14 | 106945697 | 106945697 | G | A | SNP | <i>LINC00221</i> | ENST00000334298 | 1 | intronic | c.NULL | NULL | tier3 | 62 | 1 | 0.016 | 42 | 24 | 0.364 |
| 489196 | 15 | 20457242 | 20457242 | C | T | SNP | <i>ENSG00000258628</i> | ENST00000557586 | -1 | 5_prime_untranslated_region | c.-20457242 | NULL | tier3 | 615 | 3 | 0.005 | 691 | 51 | 0.069 |
| 489196 | 15 | 66191080 | 66191080 | T | A | SNP | <i>MEGF11</i> | ENST00000409699 | -1 | missense | c.2960 | p.N987I | tier1 | 66 | 1 | 0.015 | 41 | 28 | 0.406 |
| 489196 | 16 | 57712000 | 57712000 | G | T | SNP | <i>GPR97</i> | ENST00000333493 | 1 | intronic | c.346-82 | e4-82 | tier3 | 189 | 3 | 0.016 | 139 | 55 | 0.284 |
| 489196 | 16 | 66956068 | 66956068 | G | A | SNP | <i>RRAD</i> | ENST00000299759 | -1 | missense | c.838 | p.R280C | tier1 | 66 | 1 | 0.015 | 60 | 28 | 0.318 |
| 489196 | 17 | 976071 | 976071 | C | T | SNP | <i>ABR</i> | ENST00000302538 | -1 | intronic | c.754-77 | e8-77 | tier3 | 81 | 0 | 0.000 | 71 | 27 | 0.273 |
| 489196 | 17 | 43185926 | 43185926 | G | A | SNP | <i>NMT1</i> | ENST00000258960 | 1 | 3_prime_untranslated_region | c.*2919 | NULL | tier3 | 146 | 0 | 0.000 | 131 | 53 | 0.288 |
| 489196 | 17 | 62892220 | 62892220 | G | C | SNP | <i>LRRC37A3</i> | ENST00000319651 | -1 | missense | c.1156 | p.P386A | tier1 | 374 | 0 | 0.000 | 512 | 109 | 0.175 |
| 489196 | 18 | 28971008 | 28971008 | C | T | SNP | <i>DSG4</i> | ENST00000359747 | 1 | intronic | c.685-33 | e7-33 | tier3 | 99 | 0 | 0.000 | 127 | 62 | 0.326 |
| 489196 | 19 | 40589158 | 40589158 | C | T | SNP | <i>ZNF780A</i> | ENST00000455521 | -1 | intronic | c.10-14 | e2-14 | tier3 | 208 | 1 | 0.005 | 178 | 87 | 0.327 |
| 489196 | 19 | 43920107 | 43920107 | C | T | SNP | <i>TEX101</i> | ENST00000253435 | 1 | silent | c.90 | p.L30 | tier1 | 77 | 0 | 0.000 | 40 | 22 | 0.355 |
| 489196 | 20 | 9496180 | 9496180 | G | T | SNP | <i>LAMP5</i> | ENST00000246070 | 1 | missense | c.145 | p.V49L | tier1 | 90 | 1 | 0.011 | 98 | 86 | 0.467 |
| 489196 | 20 | 29623299 | 29623299 | G | A | SNP | <i>FRG1B</i> | ENST00000278882 | 1 | intronic | c.66+45 | e1+45 | tier3 | 691 | 9 | 0.013 | 1509 | 65 | 0.041 |
| 489196 | 20 | 31073554 | 31073555 | - | GTC | INS | <i>C20orf112</i> | ENST00000201961 | -1 | 3_prime_untranslated_region | c.*9 | NULL | tier3 | 76 | 0 | 0.000 | 74 | 36 | 0.327 |
| 489196 | 21 | 45820168 | 45820168 | C | T | SNP | <i>TRPM2</i> | ENST00000300482 | 1 | silent | c.2235 | p.G745 | tier1 | 50 | 0 | 0.000 | 40 | 19 | 0.322 |
| 489196 | 22 | 46677825 | 46677825 | G | A | SNP | <i>TTC38</i> | ENST00000445282 | 1 | intronic | c.561+210 | e6+210 | tier3 | 53 | 2 | 0.036 | 54 | 21 | 0.269 |
| 489196 | X | 73041567 | 73041567 | C | T | SNP | <i>XIST</i> | ENST00000429829 | -1 | rna | NULL | NULL | tier1 | 38 | 1 | 0.026 | 57 | 33 | 0.367 |
| 489196 | X | 153049418 | 153049418 | C | G | SNP | <i>SRPK3</i> | ENST00000370101 | 1 | intronic | c.908-11 | e10-11 | tier3 | 71 | 0 | 0.000 | 56 | 21 | 0.273 |
| 489196 | X | 153799950 | 153799950 | G | A | SNP | <i>G6PD</i> | ENST00000369620 | -1 | 5_prime_flanking_region | c.-25580 | NULL | tier3 | 403 | 2 | 0.005 | 401 | 92 | 0.186 |
| 489513 | 1 | 71869848 | 71869849 | - | A | INS | <i>NEGR1</i> | ENST00000357731 | -1 | 3_prime_untranslated_region | c.*3281 | NULL | tier4 | 172 | 2 | 0.012 | 156 | 82 | 0.345 |
| 489513 | 1 | 173684860 | 173684860 | A | G | SNP | <i>KLHL20</i> | ENST00000209884 | 1 | intronic | c.1-282 | e1-282 | tier2 | 100 | 4 | 0.039 | 84 | 84 | 0.500 |
| 489513 | 1 | 240351440 | 240351440 | T | C | SNP | <i>FMN2</i> | ENST00000319653 | 1 | intronic | c.1931-67 | e4-67 | tier3 | 137 | 2 | 0.014 | 79 | 80 | 0.503 |
| 489513 | 3 | 42735190 | 42735190 | C | T | SNP | <i>HHATL</i> | ENST00000310417 | -1 | nonsense | c.1167 | p.W389* | tier1 | 92 | 2 | 0.021 | 72 | 54 | 0.429 |
| 489513 | 3 | 46792243 | 46792243 | C | T | SNP | <i>PRSS50</i> | ENST00000460241 | -1 | intronic | c.1-2102 | e1-2102 | tier2 | 30 | 0 | 0.000 | 16 | 16 | 0.500 |
| 489513 | 3 | 53844274 | 53844274 | C | T | SNP | <i>CACNA1D</i> | ENST00000288139 | 1 | silent | c.6201 | p.F2067 | tier1 | 62 | 0 | 0.000 | 40 | 43 | 0.518 |
| 489513 | 4 | 76570299 | 76570299 | T | - | DEL | <i>G3BP2</i> | ENST00000359707 | -1 | 3_prime_untranslated_region | c.*315 | NULL | tier3 | 92 | 0 | 0.000 | 155 | 16 | 0.094 |
| 489513 | 4 | 152498539 | 152498539 | G | A | SNP | <i>FAM160A1</i> | ENST00000435205 | 1 | intronic | c.106-63 | e2-63 | tier3 | 41 | 1 | 0.024 | 45 | 40 | 0.471 |
| 489513 | 5 | 20304957 | 20304957 | G | T | SNP | <i>ENSG00000214132</i> | ENST00000502634 | -1 | 5_prime_untranslated_region | c.-20304957 | NULL | tier3 | 155 | 4 | 0.025 | 159 | 117 | 0.424 |
| 489513 | 5 | 107929567 | 107929567 | C | T | SNP | <i>ENSG00000244245</i> | ENST00000466800 | 1 | 3_prime_untranslated_region | c.*107929567 | NULL | tier3 | 154 | 4 | 0.025 | 167 | 112 | 0.399 |
| 489513 | 5 | 137680472 | 137680472 | T | C | SNP | <i>FAM53C</i> | ENST00000239906 | 1 | intronic | c.137-42 | e3-42 | tier4 | 320 | 9 | 0.027 | 244 | 207 | 0.458 |
| 489513 | 5 | 140482438 | 140482438 | G | C | SNP | <i>PCDHB3</i> | ENST00000231130 | 1 | missense | c.2205 | p.Q735H | tier1 | 212 | 2 | 0.009 | 283 | 67 | 0.191 |
| 489513 | 6 | 41011404 | 41011404 | G | T | SNP | <i>TSPO2</i> | ENST00000373161 | 1 | silent | c.282 | p.L94 | tier1 | 63 | 2 | 0.031 | 42 | 32 | 0.432 |
| 489513 | 7 | 131844359 | 131844359 | G | C | SNP | <i>PLXNA4</i> | ENST00000321063 | -1 | missense | c.4533 | p.N1511K | tier1 | 120 | 3 | 0.024 | 74 | 58 | 0.439 |
| 489513 | 8 | 56367730 | 56367730 | C | T | SNP | <i>XKR4</i> | ENST00000327381 | 1 | intronic | c.1007-68110 | e3-68110 | tier2 | 397 | 13 | 0.032 | 361 | 288 | 0.442 |
| 489513 | 8 | 74495119 | 74495119 | A | - | DEL | <i>STAU2</i> | ENST00000524300 | -1 | intronic | c.1162-54 | e9-54 | tier3 | 58 | 0 | 0.000 | 65 | 13 | 0.167 |
| 489513 | 9 | 16215272 | 16215272 | C | T | SNP | <i>C9orf92</i> | ENST00000380683 | -1 | missense | c.196 | p.G66R | tier1 | 51 | 0 | 0.000 | 40 | 30 | 0.429 |
| 489513 | 9 | 32974446 | 32974446 | G | T | SNP | <i>APTX</i> | ENST00000379819 | -1 | splice_region | c.916+10 | e7+10 | tier3 | 98 | 2 | 0.020 | 103 | 73 | 0.415 |
| 489513 | 9 | 101798470 | 101798470 | C | C | SNP | <i>COL15A1</i> | ENST00000375001 | 1 | missense | c.2308 | p.G770R | tier1 | 112 | 2 | 0.018 | 119 | 80 | 0.402 |
| 489513 | 9 | 136402651 | 136402651 | G | A | SNP | <i>ADAMTSL2</i> | ENST00000393061 | 1 | missense | c.542 | p.R181Q | tier1 | 167 | 1 | 0.006 | 126 | 92 | 0.422 |
| 489513 | 10 | 48373412 | 48373412 | G | A | SNP | <i>ZNF488</i> | ENST00000395702 | 1 | 3_prime_untranslated_region | c.*1857 | NULL | tier3 | 84 | 1 | 0.012 | 63 | 41 | 0.394 |
| 489513 | 10 | 87615842 | 87615842 | G | A | SNP | <i>GRID1</i> | ENST00000327946 | -1 | missense | c.1057 | p.R353W | tier1 | 724 | 2 | 0.003 | 1088 | 48 | 0.042 |
| 489513 | 11 | 32417288 | 32417288 | G | A | SNP | <i>WT1</i> | ENST00000332351 | -1 | intronic | c.1249+515 | e7+515 | tier4 | 245 | 5 | 0.020 | 280 | 90 | 0.242 |
| 489513 | 12 | 130883190 | 130883190 | C | A | SNP | <i>RIMBP2</i> | ENST00000261655 | -1 | 3_prime_untranslated_region | c.*490 | NULL | tier3 | 88 | 2 | 0.022 | 107 | 69 | 0.390 |
| 489513 | 12 | 133050427 | 133050427 | A | G | SNP | <i>MUC8</i> | ENST00000595994 | -1 | intronic | c.148-131 | e2-131 | tier2 | 319 | 4 | 0.012 | 424 | 31 | 0.068 |
| 489513 | 15 | 97634474 | 97634474 | A | G | SNP | <i>ENSG00000215948</i> | ENST00000401129 | -1 | 3_prime_flanking_region | c.*18 | NULL | tier4 | 128 | 2 | 0.015 | 127 | 102 | 0.444 |
| 489513 | 17 | 78180746 | 78180746 | C | T | SNP | <i>SGSH</i> | ENST00000575484 | -1 | rna | NULL | NULL | tier1 | 102 | 2 | 0.019 | 94 | 61 | 0.394 |
| 489513 | 18 | 72113811 | 72113811 | C | G | SNP | <i>FAM69C</i> | ENST00000343998 | -1 | intronic | c.876+30 | e2+30 | tier3 | 62 | 0 | 0.000 | 45 | 65 | 0.591 |
| 489513 | 19 | 38161162 | 38161162 | T | - | DEL | <i>ZNF781</i> | ENST00000358582 | -1 | 5_prime_untranslated_region | c.-113 | NULL | tier3 | 106 | 1 | 0.009 | 159 | 16 | 0.091 |
| 489513 | 19 | 40834389 | 40834389 | G | T | SNP | <i>C19orf47</i> | ENST00000582783 | -1 | missense | c.481 | p.D161N | tier1 | 122 | 2 | 0.016 | 77 | 89 | 0.536 |
| 489513 | 19 | 59081697 | 59081697 | C | A | SNP | <i>MZF1</i> | ENST00000595245 | -1 | 5_prime_untranslated_region | c.-59081697 | NULL | tier3 | 79 | 5 | 0.059 | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|------|---|-----|-----------------|-----------------|----|-----------------------------|--------------|-----------|-------|------|----|-------|------|-----|-------|
| 489513 | X | 119390658 | 119390658 | G | T | SNP | ZBTB33 | ENST00000326624 | 1 | 3_prime_untranslated_region | c.*1369 | NULL | tier2 | 60 | 1 | 0.016 | 71 | 15 | 0.174 |
| 491309 | 1 | 243288182 | 243288182 | T | - | DEL | CEP170 | ENST00000468254 | -1 | rna | NULL | NULL | tier1 | 36 | 0 | 0.000 | 146 | 28 | 0.161 |
| 491309 | 5 | 179780316 | 179780316 | T | C | SNP | GFP72 | ENST00000253778 | -1 | 5_prime_untranslated_region | c.-99 | NULL | tier2 | 12 | 0 | 0.000 | 11 | 9 | 0.409 |
| 491309 | 7 | 2309174 | 2309174 | G | A | SNP | SNX8 | ENST00000222990 | -1 | intronic | c.621+20 | e5+20 | tier3 | 26 | 0 | 0.000 | 28 | 9 | 0.231 |
| 491309 | 17 | 7577580 | 7577580 | T | C | SNP | TP53 | ENST00000269305 | -1 | missense | c.701 | p.Y234C | tier1 | 711 | 3 | 0.004 | 1093 | 44 | 0.039 |
| 491309 | 19 | 16629981 | 16629981 | C | T | SNP | CHERP | ENST00000546361 | -1 | missense | c.2740 | p.E914K | tier1 | 99 | 0 | 0.000 | 156 | 16 | 0.093 |
| 496321 | 1 | 986249 | 986249 | C | T | SNP | AGRN | ENST00000379370 | 1 | intronic | c.5253+32 | e30+32 | tier2 | 27 | 0 | 0.000 | 22 | 24 | 0.522 |
| 496321 | 1 | 47276735 | 47276735 | G | A | SNP | CYP4B1 | ENST00000371923 | 1 | intronic | c.323-77 | e3-77 | tier3 | 75 | 3 | 0.039 | 39 | 29 | 0.427 |
| 496321 | 1 | 114397144 | 114397144 | C | A | SNP | PTPN22 | ENST00000359785 | -1 | missense | c.699 | p.R233S | tier1 | 29 | 2 | 0.065 | 59 | 43 | 0.422 |
| 496321 | 2 | 89102427 | 89102427 | G | A | SNP | ANKRD36BP2 | ENST00000393525 | 1 | rna | NULL | NULL | tier1 | 104 | 2 | 0.019 | 478 | 69 | 0.126 |
| 496321 | 2 | 241513196 | 241513196 | C | T | SNP | RNPEPL1 | ENST00000270357 | 1 | missense | c.133 | p.R45C | tier1 | 69 | 5 | 0.067 | 48 | 30 | 0.385 |
| 496321 | 3 | 25639916 | 25639916 | G | A | SNP | TOP2B | ENST00000264331 | -1 | missense | c.4763 | p.S1588L | tier1 | 73 | 0 | 0.000 | 53 | 31 | 0.369 |
| 496321 | 3 | 38141928 | 38141928 | G | A | SNP | DLEC1 | ENST00000346219 | 1 | intronic | c.2864+12 | e19+12 | tier3 | 92 | 6 | 0.061 | 47 | 40 | 0.460 |
| 496321 | 3 | 125643455 | 125643455 | C | T | SNP | FAM86JP | ENST00000467239 | 1 | intronic | c.NULL | NULL | tier3 | 56 | 1 | 0.018 | 29 | 31 | 0.517 |
| 496321 | 3 | 172835422 | 172835422 | A | T | SNP | SPATA16 | ENST00000351008 | -1 | missense | c.100 | p.L34I | tier1 | 62 | 2 | 0.031 | 64 | 46 | 0.418 |
| 496321 | 3 | 193125021 | 193125021 | G | A | SNP | ATP13A4 | ENST00000342695 | -1 | intronic | c.3378+81 | e29+81 | tier3 | 65 | 2 | 0.030 | 47 | 33 | 0.413 |
| 496321 | 4 | 103822448 | 103822448 | G | A | SNP | SLC9B1 | ENST00000296422 | -1 | silent | c.1374 | p.S458 | tier1 | 435 | 3 | 0.007 | 605 | 44 | 0.068 |
| 496321 | 4 | 106190881 | 106190881 | A | T | SNP | TET2 | ENST00000380013 | 1 | missense | c.4159 | p.N1387Y | tier1 | 297 | 17 | 0.054 | 195 | 224 | 0.535 |
| 496321 | 4 | 106193892 | 106193892 | C | T | SNP | TET2 | ENST00000380013 | 1 | nonsense | c.4354 | p.R1452* | tier1 | 702 | 34 | 0.046 | 568 | 426 | 0.428 |
| 496321 | 4 | 120446865 | 120446865 | G | A | SNP | PDE5A | ENST00000354960 | -1 | intronic | c.1633-15 | e12-15 | tier3 | 53 | 1 | 0.019 | 40 | 11 | 0.212 |
| 496321 | 4 | 153809253 | 153809253 | G | A | SNP | ARFIP1 | ENST00000353617 | 1 | intronic | c.792-32 | e7-32 | tier3 | 24 | 0 | 0.000 | 51 | 18 | 0.261 |
| 496321 | 5 | 140202969 | 140202969 | C | T | SNP | PCDHA5 | ENST00000529859 | 1 | missense | c.1609 | p.R537C | tier1 | 482 | 37 | 0.071 | 306 | 270 | 0.469 |
| 496321 | 6 | 152746604 | 152746604 | C | T | SNP | SYNE1 | ENST00000341594 | -1 | splice_site | c.5291-1 | e40-1 | tier1 | 55 | 1 | 0.018 | 83 | 63 | 0.429 |
| 496321 | 6 | 159660601 | 159660601 | G | A | SNP | FNDC1 | ENST00000329629 | 1 | missense | c.3919 | p.G1307R | tier1 | 160 | 13 | 0.075 | 91 | 88 | 0.492 |
| 496321 | 6 | 160199944 | 160199944 | G | A | SNP | ACAT2 | ENST00000472052 | 1 | rna | NULL | NULL | tier1 | 46 | 2 | 0.041 | 77 | 67 | 0.465 |
| 496321 | 7 | 1132922 | 1132922 | G | A | SNP | GPER1 | ENST00000297469 | 1 | 3_prime_untranslated_region | c.*430 | NULL | tier2 | 99 | 2 | 0.020 | 42 | 38 | 0.475 |
| 496321 | 7 | 100652450 | 100652450 | T | C | SNP | MUC12 | ENST00000536621 | 1 | splice_region | c.15251+10 | e5+10 | tier2 | 59 | 3 | 0.048 | 67 | 59 | 0.468 |
| 496321 | 7 | 150835438 | 150835438 | G | A | SNP | AGAP3 | ENST00000397238 | 1 | intronic | c.1666+38 | e12+38 | tier2 | 84 | 1 | 0.012 | 50 | 25 | 0.333 |
| 496321 | 12 | 48375943 | 48375943 | C | T | SNP | COL2A1 | ENST00000380518 | -1 | missense | c.2302 | p.G768S | tier1 | 106 | 5 | 0.045 | 60 | 62 | 0.508 |
| 496321 | 13 | 61141522 | 61141522 | C | T | SNP | TDRD3 | ENST00000535286 | 1 | intronic | c.2119-137 | e13-137 | tier3 | 70 | 4 | 0.053 | 107 | 84 | 0.440 |
| 496321 | 14 | 75248223 | 75248223 | C | T | SNP | YLP1M | ENST00000325680 | 1 | nonsense | c.1477 | p.Q493* | tier1 | 61 | 4 | 0.062 | 67 | 51 | 0.432 |
| 496321 | 15 | 42434172 | 42434172 | C | T | SNP | PLA2G4F | ENST00000397272 | -1 | 3_prime_untranslated_region | c.*10 | NULL | tier3 | 59 | 5 | 0.078 | 38 | 37 | 0.493 |
| 496321 | 15 | 72958364 | 72958364 | C | G | SNP | GOLGA6B | ENST00000421285 | 1 | missense | c.1849 | p.H617D | tier1 | 212 | 0 | 0.000 | 192 | 32 | 0.143 |
| 496321 | 15 | 75219024 | 75219024 | A | G | SNP | COX5A | ENST00000322347 | -1 | intronic | c.339+83 | e3+83 | tier3 | 15 | 0 | 0.000 | 29 | 38 | 0.567 |
| 496321 | 16 | 29820921 | 29820921 | C | T | SNP | MAZ | ENST00000219782 | 1 | missense | c.1340 | p.P447L | tier1 | 48 | 1 | 0.020 | 18 | 19 | 0.514 |
| 496321 | 16 | 33497567 | 33497567 | C | T | SNP | BMS1P8 | ENST00000565156 | -1 | rna | NULL | NULL | tier1 | 354 | 3 | 0.008 | 573 | 35 | 0.058 |
| 496321 | 19 | 33792431 | 33792431 | C | G | SNP | CEBPA | ENST00000498907 | -1 | missense | c.890 | p.R297P | tier1 | 1145 | 61 | 0.050 | 682 | 577 | 0.458 |
| 496321 | 19 | 36727961 | 36727961 | T | C | SNP | ZNF146 | ENST00000443387 | 1 | missense | c.619 | p.S207P | tier1 | 29 | 2 | 0.065 | 54 | 46 | 0.455 |
| 496321 | 19 | 55697038 | 55697038 | G | A | SNP | PTPRH | ENST00000376350 | -1 | splice_region | c.2902-8 | e18-8 | tier3 | 36 | 3 | 0.077 | 26 | 24 | 0.480 |
| 496321 | X | 49189242 | 49189242 | - | A | INS | GAGE13 | ENST00000381751 | 1 | frame_shift_ins | c.27_28 | p.Y9fs | tier1 | 14 | 0 | 0.000 | 3 | 4 | 0.571 |
| 496321 | Y | 22923114 | 22923114 | C | T | SNP | RPS4Y2 | ENST00000288666 | 1 | intronic | c.263-56 | e4-56 | tier3 | 36 | 3 | 0.077 | 7 | 54 | 0.885 |
| 499258 | 1 | 40033490 | 40033490 | T | C | SNP | PABPC4 | ENST00000372858 | -1 | missense | c.905 | p.D302G | tier1 | 145 | 1 | 0.007 | 205 | 51 | 0.199 |
| 499258 | 1 | 155028802 | 155028802 | C | G | SNP | ADAM15 | ENST00000525020 | 1 | 3_prime_untranslated_region | c.*155028802 | NULL | tier3 | 34 | 0 | 0.000 | 138 | 32 | 0.188 |
| 499258 | 1 | 182850554 | 182850554 | G | G | SNP | DHX9 | ENST00000367549 | 1 | missense | c.2780 | p.D927G | tier1 | 109 | 0 | 0.000 | 161 | 31 | 0.162 |
| 499258 | 1 | 240969528 | 240969528 | A | A | SNP | RGS7 | ENST00000407727 | -1 | missense | c.1181 | p.P394L | tier1 | 45 | 0 | 0.000 | 155 | 41 | 0.209 |
| 499258 | 2 | 6879873 | 6879873 | T | - | DEL | LINC00487 | ENST00000382045 | -1 | intronic | c.NULL | NULL | tier2 | 49 | 0 | 0.000 | 130 | 32 | 0.198 |
| 499258 | 2 | 43905981 | 43905981 | A | G | SNP | PLEKHH2 | ENST00000282406 | 1 | intronic | c.124-21 | e2-21 | tier2 | 232 | 3 | 0.013 | 240 | 70 | 0.225 |
| 499258 | 2 | 88825827 | 88825827 | C | T | SNP | C2orf51 | ENST00000303254 | 1 | intronic | c.28-96 | e2-96 | tier4 | 70 | 0 | 0.000 | 149 | 27 | 0.153 |
| 499258 | 2 | 197645323 | 197645323 | A | G | SNP | GTF3C3 | ENST00000263956 | -1 | missense | c.1178 | p.M393T | tier1 | 81 | 0 | 0.000 | 93 | 21 | 0.184 |
| 499258 | 4 | 100266176 | 100266176 | G | A | SNP | ADH1C | ENST00000515683 | -1 | missense | c.410 | p.P137L | tier1 | 63 | 0 | 0.000 | 237 | 75 | 0.240 |
| 499258 | 7 | 98639905 | 98639905 | T | - | DEL | SMURF1 | ENST00000361125 | -1 | intronic | c.1335-50 | e13-50 | tier4 | 38 | 0 | 0.000 | 87 | 16 | 0.155 |
| 499258 | 7 | 135376425 | 135376425 | G | A | SNP | C7orf73 | ENST00000422968 | 1 | 3_prime_untranslated_region | c.*135376425 | NULL | tier3 | 57 | 1 | 0.017 | 96 | 30 | 0.238 |
| 499258 | 7 | 144060684 | 144060684 | G | A | SNP | ARHGEF5 | ENST00000056217 | 1 | missense | c.922 | p.E308K | tier1 | 17 | 0 | 0.000 | 72 | 37 | 0.339 |
| 499258 | 9 | 87322805 | 87322805 | C | T | SNP | NTRK2 | ENST00000277120 | 1 | missense | c.406 | p.R136C | tier1 | 116 | 0 | 0.000 | 159 | 20 | 0.112 |
| 499258 | 10 | 22680773 | 22680773 | T | C | SNP | SPAG6 | ENST00000376603 | 1 | missense | c.1349 | p.V450A | tier1 | 58 | 0 | 0.000 | 141 | 32 | 0.185 |
| 499258 | 10 | 49291796 | 49291796 | C | T | SNP | ENSG00000249871 | ENST00000506185 | -1 | intronic | c.NULL | NULL | tier3 | 123 | 2 | 0.016 | 88 | 24 | 0.214 |
| 499258 | 12 | 100904941 | 100904941 | A | T | SNP | NR1H4 | ENST00000551379 | 1 | intronic | c.475+20 | e2+20 | tier3 | 43 | 0 | 0.000 | 107 | 25 | 0.189 |
| 499258 | 15 | 25224990 | 25224990 | T | C | SNP | SNURF | ENST00000551312 | 1 | intronic | c.216-104 | e4-104 | tier2 | 68 | 0 | 0.000 | 84 | 25 | 0.229 |
| 499258 | 16 | 72821316 | 72821316 | A | G | SNP | ZFH3 | ENST00000268489 | -1 | missense | c.10859 | p.V3620A | tier1 | 43 | 0 | 0.000 | 193 | 49 | 0.203 |
| 499258 | 17 | 38450574 | 38450574 | G | T | SNP | CDC6 | ENST00000209728 | 1 | intronic | c.944-42 | e6-42 | tier2 | 107 | 0 | 0.000 | 113 | 27 | 0.193 |
| 499258 | 17 | 42152474 | 42152474 | G | A | SNP | G6PC3 | ENST00000269097 | 1 | intronic | c.535+19 | e4+19 | tier2 | 30 | 0 | 0.000 | 120 | 30 | 0.200 |
| 499258 | 19 | 56423693 | 56423693 | G | A | SNP | NLRP13 | ENST00000342929 | -1 | missense | c.1490 | p.T497M | tier1 | 39 | 0 | 0.000 | 72 | 14 | 0.163 |
| 499258 | 21 | 16337046 | 16337049 | TTCT | - | DEL | NR1P1 | ENST00000318948 | -1 | frame_shift_del | c.3468_3465 | p.K1155fs | tier1 | 83 | 1 | 0.012 | 119 | 17 | 0.125 |
| 499258 | 21 | 36252865 | 36252865 | C | G | SNP | RUNX1 | ENST00000300305 | -1 | missense | c.497 | p.R166P | tier1 | 353 | 1 | 0.003 | 1294 | 196 | 0.132 |
| 499258 | 22 | 21804391 | 21804391 | A | G | SNP | HIC2 | ENST00000407464 | 1 | 3_prime_untranslated_region | c.*3359 | NULL | tier3 | 84 | 0 | 0.000 | 189 | 36 | 0.160 |
| 499258 | X | 24813697 | 24813697 | T | C | SNP | POLA1 | ENST00000379068 | 1 | intronic | c.2965-14318 | e27-14318 | tier3 | 55 | 0 | 0.000 | 61 | 55 | 0.474 |
| 502635 | 1 | 28208733 | 28208733 | C | T | SNP | THEMIS2 | ENST00000373921 | 1 | missense | c.898 | p.R300W | tier1 | 124 | 1 | 0.008 | 126 | 16 | 0.112 |
| 502635 | 1 | 52912122 | 52912122 | T | A | SNP | ZCCHC11 | ENST00000257177 | -1 | splice_region | c.3514-8 | e21-8 | tier3 | 120 | 2 | 0.016 | 78 | 14 | 0.151 |
| 502635 | 1 | 235617686 | 235617686 | A | C | SNP | B3GALNT2 | ENST00000366600 | -1 | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-------|---|-----|-----------------|-----------------|----|-----------------------------|-----------------|----------|-------|------|----|-------|-----|-----|-------|
| 502635 | 7 | 29546764 | 29546764 | T | - | DEL | CHN2 | ENST00000539406 | 1 | intronic | c.1217-80 | e12-80 | tier3 | 43 | 0 | 0.000 | 33 | 10 | 0.233 |
| 502635 | 11 | 89515227 | 89515227 | G | T | SNP | TRIM64DP | ENST00000529289 | 1 | 3_prime_untranslated_region | c.*89515227 | NULL | tier3 | 305 | 0 | 0.000 | 323 | 26 | 0.075 |
| 502635 | 12 | 5840529 | 5840529 | A | - | DEL | ANO2 | ENST00000356134 | -1 | intronic | c.1548+1157 | e16+1157 | tier3 | 60 | 0 | 0.000 | 71 | 13 | 0.155 |
| 502635 | 12 | 119583250 | 119583250 | G | A | SNP | SRRM4 | ENST00000267260 | 1 | missense | c.836 | p.R279Q | tier1 | 66 | 0 | 0.000 | 54 | 8 | 0.129 |
| 502635 | 13 | 101881935 | 101881935 | C | T | SNP | NALCN | ENST00000251127 | -1 | missense | c.1435 | p.V479I | tier1 | 487 | 0 | 0.000 | 486 | 26 | 0.051 |
| 502635 | 14 | 106573418 | 106573418 | T | C | SNP | IGHV3-11 | ENST00000390601 | -1 | missense | c.166 | p.I56V | tier1 | 166 | 7 | 0.040 | 150 | 36 | 0.194 |
| 502635 | 14 | 106573482 | 106573482 | T | C | SNP | IGHV3-11 | ENST00000390601 | -1 | silent | c.102 | p.G34 | tier1 | 195 | 5 | 0.025 | 164 | 33 | 0.168 |
| 502635 | 14 | 106573491 | 106573491 | G | T | SNP | IGHV3-11 | ENST00000390601 | -1 | silent | c.93 | p.V31 | tier1 | 200 | 5 | 0.024 | 164 | 33 | 0.168 |
| 502635 | 15 | 31086780 | 31086780 | G | T | SNP | GOLGA8UP | ENST00000527159 | 1 | 3_prime_untranslated_region | c.*31086780 | NULL | tier3 | 520 | 1 | 0.002 | 493 | 44 | 0.082 |
| 502635 | 15 | 43995984 | 43995984 | C | G | SNP | STRC | ENST00000541030 | -1 | intronic | c.1+14335 | e0+14335 | tier3 | 90 | 0 | 0.000 | 65 | 8 | 0.110 |
| 502635 | 15 | 65715171 | 65715171 | G | A | SNP | IGDCC4 | ENST00000352385 | -1 | silent | c.30 | p.R10 | tier1 | 8 | 0 | 0.000 | 0 | 5 | 1.000 |
| 502635 | 16 | 9212075 | 9212079 | TATTA | - | DEL | C16orf72 | ENST00000327827 | 1 | 3_prime_untranslated_region | c.*1306 | NULL | tier2 | 161 | 0 | 0.000 | 76 | 6 | 0.073 |
| 502635 | 17 | 1326754 | 1326754 | G | A | SNP | CRK | ENST00000572145 | -1 | rna | NULL | NULL | tier1 | 106 | 0 | 0.000 | 120 | 10 | 0.076 |
| 502635 | 17 | 7577538 | 7577538 | C | T | SNP | TP53 | ENST00000269305 | -1 | missense | c.743 | p.R248Q | tier1 | 1010 | 2 | 0.002 | 931 | 73 | 0.073 |
| 502635 | 17 | 7578551 | 7578551 | A | T | SNP | TP53 | ENST00000269305 | -1 | missense | c.379 | p.S127T | tier1 | 1112 | 5 | 0.005 | 988 | 82 | 0.077 |
| 502635 | 18 | 47455964 | 47455964 | C | A | SNP | MYO5B | ENST00000285039 | -1 | missense | c.2008 | p.D670Y | tier1 | 197 | 4 | 0.020 | 200 | 25 | 0.111 |
| 502635 | 19 | 852255 | 852255 | G | T | SNP | ELANE | ENST00000590230 | 1 | 5_prime_untranslated_region | c.-74 | NULL | tier2 | 748 | 0 | 0.000 | 579 | 41 | 0.066 |
| 502635 | 21 | 35471902 | 35471903 | - | T | INS | SLC5A3 | ENST00000381151 | 1 | 3_prime_untranslated_region | c.*2248 | NULL | tier3 | 79 | 0 | 0.000 | 66 | 8 | 0.108 |
| 565435 | 1 | 91402928 | 91402928 | A | G | SNP | ZNF644 | ENST00000337393 | -1 | intronic | c.3688+114 | e3+114 | tier3 | 9 | 0 | 0.000 | 26 | 37 | 0.587 |
| 565435 | 1 | 114641862 | 114641862 | G | A | SNP | SYT6 | ENST00000393296 | -1 | silent | c.1218 | p.L406 | tier1 | 132 | 0 | 0.000 | 114 | 50 | 0.303 |
| 565435 | 1 | 150246585 | 150246585 | A | C | SNP | C1orf54 | ENST00000369099 | 1 | intronic | c.130+12 | e2+12 | tier3 | 81 | 2 | 0.024 | 110 | 28 | 0.203 |
| 565435 | 2 | 86267544 | 86267544 | G | A | SNP | POLR1A | ENST00000263857 | -1 | silent | c.3711 | p.G1237 | tier1 | 121 | 2 | 0.016 | 71 | 32 | 0.311 |
| 565435 | 2 | 97849131 | 97849131 | A | G | SNP | ANKRD36 | ENST00000420699 | 1 | intronic | c.1838-65 | e27-65 | tier3 | 72 | 2 | 0.027 | 158 | 58 | 0.269 |
| 565435 | 2 | 130750444 | 130750444 | C | T | SNP | ENSG00000250207 | ENST00000511039 | -1 | 5_prime_untranslated_region | c.-130750444 | NULL | tier2 | 390 | 1 | 0.003 | 358 | 66 | 0.155 |
| 565435 | 2 | 219267322 | 219267322 | C | A | SNP | CTDSP1 | ENST00000273062 | 1 | intronic | c.378+194 | e4+194 | tier3 | 63 | 0 | 0.000 | 58 | 15 | 0.206 |
| 565435 | 3 | 44776777 | 44776777 | T | C | SNP | ZNF501 | ENST00000396048 | 1 | 3_prime_untranslated_region | c.*48 | NULL | tier3 | 39 | 0 | 0.000 | 76 | 22 | 0.225 |
| 565435 | 3 | 133465239 | 133465239 | A | G | SNP | TF | ENST00000402696 | 1 | 5_prime_untranslated_region | c.-46 | NULL | tier2 | 33 | 1 | 0.029 | 24 | 15 | 0.385 |
| 565435 | 3 | 155314188 | 155314188 | T | C | SNP | PLCH1 | ENST00000340059 | -1 | intronic | c.44-21 | e2-21 | tier3 | 25 | 0 | 0.000 | 13 | 6 | 0.316 |
| 565435 | 3 | 185300560 | 185300560 | G | C | SNP | SENP2 | ENST00000545472 | 1 | missense | c.26 | p.C9S | tier1 | 186 | 4 | 0.021 | 134 | 41 | 0.233 |
| 565435 | 4 | 73615881 | 73615881 | G | A | SNP | - | - | - | - | - | - | tier3 | 87 | 4 | 0.044 | 133 | 53 | 0.285 |
| 565435 | 4 | 148457005 | 148457006 | - | T | INS | EDNRA | ENST00000324300 | 1 | intronic | c.748-24_748-23 | e4-23 | tier3 | 30 | 0 | 0.000 | 27 | 9 | 0.250 |
| 565435 | 5 | 13900350 | 13900350 | G | A | SNP | DNAH5 | ENST00000265104 | -1 | nonsense | c.2224 | p.R742* | tier1 | 446 | 5 | 0.011 | 475 | 166 | 0.259 |
| 565435 | 5 | 43295963 | 43295963 | G | A | SNP | HMGCS1 | ENST00000325110 | -1 | missense | c.796 | p.P266S | tier1 | 36 | 0 | 0.000 | 98 | 25 | 0.203 |
| 565435 | 6 | 31974832 | 31974832 | G | A | SNP | CYP21A1P | ENST00000342991 | 1 | 3_prime_untranslated_region | c.*31974832 | NULL | tier3 | 136 | 2 | 0.015 | 81 | 31 | 0.274 |
| 565435 | 6 | 54208081 | 54208081 | C | T | SNP | TINAG | ENST00000259782 | 1 | missense | c.682 | p.P228S | tier1 | 40 | 0 | 0.000 | 73 | 37 | 0.336 |
| 565435 | 6 | 80774316 | 80774316 | T | G | SNP | ENSG00000218418 | ENST00000401578 | -1 | 5_prime_untranslated_region | c.-80774316 | NULL | tier3 | 135 | 1 | 0.007 | 117 | 41 | 0.260 |
| 565435 | 7 | 5434025 | 5434025 | A | T | SNP | TNRC18 | ENST00000399537 | -1 | intronic | c.343+46 | e2+46 | tier3 | 197 | 5 | 0.025 | 133 | 83 | 0.384 |
| 565435 | 7 | 122255199 | 122255199 | A | C | SNP | CADPS2 | ENST00000449022 | -1 | intronic | c.1223+36 | e6+36 | tier3 | 63 | 1 | 0.016 | 75 | 30 | 0.283 |
| 565435 | 7 | 150894251 | 150894251 | C | T | SNP | IQCA1P1 | ENST00000602757 | -1 | intronic | c.NULL | NULL | tier3 | 77 | 1 | 0.013 | 78 | 31 | 0.284 |
| 565435 | 8 | 144399939 | 144399939 | G | A | SNP | TOP1MT | ENST00000329245 | -1 | silent | c.1284 | p.Y428 | tier1 | 155 | 3 | 0.019 | 119 | 36 | 0.232 |
| 565435 | 9 | 32631391 | 32631391 | T | C | SNP | TAF1L | ENST00000242310 | -1 | missense | c.4187 | p.R1396H | tier1 | 160 | 1 | 0.006 | 144 | 36 | 0.200 |
| 565435 | 9 | 74299332 | 74299332 | C | C | SNP | TMEM2 | ENST00000377057 | -1 | 5_prime_untranslated_region | c.-74299332 | NULL | tier3 | 41 | 0 | 0.000 | 75 | 37 | 0.330 |
| 565435 | 9 | 84608191 | 84608191 | T | C | SNP | SPATA31D1 | ENST00000344803 | 1 | missense | c.2806 | p.S936P | tier1 | 128 | 0 | 0.000 | 130 | 59 | 0.312 |
| 565435 | 10 | 95849013 | 95849013 | G | A | SNP | PLCE1 | ENST00000371375 | 1 | silent | c.162 | p.Q54 | tier1 | 923 | 14 | 0.015 | 640 | 239 | 0.272 |
| 565435 | 11 | 27528351 | 27528352 | - | G | INS | LGR4 | ENST00000379214 | -1 | 5_prime_flanking_region | c.-34502 | NULL | tier2 | 128 | 3 | 0.023 | 88 | 22 | 0.200 |
| 565435 | 11 | 56738588 | 56738588 | G | A | SNP | OR5AK3P | ENST00000326876 | 1 | 3_prime_untranslated_region | c.*56738588 | NULL | tier3 | 54 | 0 | 0.000 | 120 | 18 | 0.130 |
| 565435 | 12 | 102140522 | 102140523 | - | A | INS | GNPTAB | ENST00000299314 | -1 | 3_prime_untranslated_region | c.*420 | NULL | tier3 | 59 | 0 | 0.000 | 108 | 16 | 0.129 |
| 565435 | 14 | 19685459 | 19685459 | C | T | SNP | LOC440157 | ENST00000418499 | 1 | rna | NULL | NULL | tier1 | 74 | 0 | 0.000 | 63 | 11 | 0.149 |
| 565435 | 14 | 21853909 | 21853909 | C | T | SNP | CHD8 | ENST00000399982 | -1 | missense | c.7609 | p.E2537K | tier1 | 58 | 0 | 0.000 | 43 | 21 | 0.328 |
| 565435 | 14 | 56120281 | 56120281 | C | G | SNP | KTN1 | ENST00000395309 | 1 | missense | c.2768 | p.S923C | tier1 | 37 | 0 | 0.000 | 89 | 32 | 0.265 |
| 565435 | 15 | 79065337 | 79065337 | C | T | SNP | ADAMTS7 | ENST00000388820 | -1 | intronic | c.2131+88 | e12+88 | tier3 | 85 | 2 | 0.023 | 66 | 21 | 0.241 |
| 565435 | 15 | 90631934 | 90631934 | C | T | SNP | IDH2 | ENST00000330062 | -1 | missense | c.419 | p.R140Q | tier1 | 701 | 10 | 0.014 | 588 | 212 | 0.265 |
| 565435 | 16 | 69738490 | 69738490 | C | T | SNP | ENSG00000260772 | ENST00000561622 | 1 | rna | NULL | NULL | tier1 | 30 | 0 | 0.000 | 60 | 25 | 0.294 |
| 565435 | 17 | 10402323 | 10402323 | C | T | SNP | MYH1 | ENST00000226207 | -1 | missense | c.3952 | p.E1318K | tier1 | 169 | 0 | 0.000 | 250 | 20 | 0.074 |
| 565435 | 17 | 56620367 | 56620367 | G | T | SNP | C17orf47 | ENST00000321691 | -1 | missense | c.1181 | p.P394Q | tier1 | 139 | 5 | 0.035 | 101 | 37 | 0.266 |
| 565435 | 17 | 74732959 | 74732959 | G | T | SNP | SRSF2 | ENST00000359995 | -1 | missense | c.284 | p.P95H | tier1 | 488 | 4 | 0.008 | 360 | 143 | 0.283 |
| 565435 | 19 | 11342899 | 11342899 | G | A | SNP | DOCK6 | ENST00000319867 | -1 | missense | c.697 | p.R233W | tier1 | 97 | 0 | 0.000 | 62 | 35 | 0.357 |
| 565435 | 20 | 31022441 | 31022442 | - | G | INS | ASXL1 | ENST00000375687 | 1 | frame_shift_ins | c.1926_1927 | p.G645fs | tier1 | 1055 | 26 | 0.024 | 802 | 238 | 0.229 |
| 565435 | 22 | 18027848 | 18027848 | G | T | SNP | CECR2 | ENST00000400573 | 1 | missense | c.2805 | p.L935F | tier1 | 421 | 7 | 0.016 | 531 | 50 | 0.086 |
| 565435 | X | 39916576 | 39916576 | T | C | SNP | BCOR | ENST00000378444 | -1 | splice_site | c.4429-2 | e10-2 | tier1 | 214 | 0 | 0.000 | 208 | 30 | 0.126 |
| 565435 | X | 123197716 | 123197716 | C | C | SNP | STAG2 | ENST00000218089 | 1 | nonsense | c.1840 | p.R614* | tier1 | 113 | 0 | 0.000 | 311 | 64 | 0.170 |
| 565435 | X | 123220440 | 123220440 | C | T | SNP | STAG2 | ENST00000218089 | 1 | nonsense | c.3097 | p.R1033* | tier1 | 258 | 4 | 0.015 | 279 | 101 | 0.266 |
| 596762 | 1 | 26314633 | 26314634 | - | T | INS | PAFAH2 | ENST00000374282 | -1 | intronic | c.341+88_341+87 | e3+88 | tier3 | 50 | 0 | 0.000 | 69 | 19 | 0.216 |
| 596762 | 1 | 77987426 | 77987426 | C | T | SNP | AK5 | ENST00000354567 | 1 | intronic | c.1312-86 | e12-86 | tier3 | 45 | 0 | 0.000 | 61 | 11 | 0.153 |
| 596762 | 1 | 97144881 | 97144881 | T | C | SNP | ENSG00000137970 | ENST00000468715 | -1 | 5_prime_untranslated_region | c.-97144881 | NULL | tier3 | 91 | 1 | 0.011 | 68 | 25 | 0.269 |
| 596762 | 1 | 177906320 | 177906320 | A | T | SNP | SEC16B | ENST00000308284 | -1 | intronic | c.2503+29 | e18+29 | tier3 | 37 | 0 | 0.000 | 23 | 25 | 0.521 |
| 596762 | 3 | 114214866 | 114214866 | T | - | DEL | ZBTB20 | ENST00000357258 | -1 | intronic | c.1+4333 | e0+4333 | tier3 | 83 | 1 | 0.012 | 101 | 15 | 0.129 |
| 596762 | 4 | 2160973 | 2160973 | G | A | SNP | POLN | ENST00000382865 | -1 | intronic | c.1555-35 | e12-35 | tier3 | 73 | 1 | 0.014 | 61 | 38 | 0.384 |
| 596762 | 4 | 17528425 | 17528425 | C | A | SNP | CLRN2 | ENST00000 | | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-------|---|-----|-----------------|-----------------|----|-----------------------------|---------------|-----------|-------|------|----|-------|------|-----|-------|
| 596762 | 11 | 92531510 | 92531510 | C | T | SNP | FAT3 | ENST00000298047 | 1 | silent | c.5331 | p.S1777 | tier1 | 49 | 0 | 0.000 | 43 | 16 | 0.267 |
| 596762 | 12 | 57850452 | 57850452 | G | C | SNP | INHBE | ENST00000266646 | 1 | missense | c.874 | p.A292P | tier1 | 63 | 1 | 0.015 | 59 | 21 | 0.263 |
| 596762 | 12 | 113379563 | 113379563 | G | T | SNP | OAS3 | ENST00000228928 | 1 | missense | c.366 | p.E122D | tier1 | 31 | 0 | 0.000 | 37 | 17 | 0.315 |
| 596762 | 13 | 86369756 | 86369756 | C | T | SNP | SLITRK6 | ENST00000400286 | -1 | missense | c.888 | p.M296I | tier1 | 103 | 0 | 0.000 | 90 | 43 | 0.323 |
| 596762 | 14 | 62547892 | 62547892 | G | A | SNP | SYT16 | ENST00000430451 | 1 | missense | c.1334 | p.R445Q | tier1 | 115 | 0 | 0.000 | 131 | 12 | 0.084 |
| 596762 | 15 | 85401550 | 85401550 | G | A | SNP | ALPK3 | ENST00000258888 | 1 | missense | c.4187 | p.R1396Q | tier1 | 420 | 1 | 0.002 | 364 | 201 | 0.353 |
| 596762 | 17 | 4109540 | 4109540 | A | G | SNP | ANKFY1 | ENST00000570535 | -1 | intronic | c.1024+76 | e7+76 | tier3 | 48 | 0 | 0.000 | 37 | 25 | 0.403 |
| 596762 | 17 | 38438120 | 38438120 | G | C | SNP | WIPF2 | ENST00000323571 | 1 | 3_prime_untranslated_region | c.*3643 | NULL | tier3 | 88 | 1 | 0.011 | 86 | 53 | 0.381 |
| 596762 | 17 | 56357291 | 56357291 | C | A | SNP | MPO | ENST00000340482 | -1 | silent | c.333 | p.T111 | tier1 | 62 | 2 | 0.031 | 60 | 29 | 0.326 |
| 596762 | 18 | 7774085 | 7774085 | T | A | SNP | PTPRM | ENST00000400060 | 1 | intronic | c.74-62 | e2-62 | tier3 | 76 | 0 | 0.000 | 72 | 31 | 0.301 |
| 596762 | 20 | 34430474 | 34430475 | - | T | INS | PHF20 | ENST00000374012 | 1 | intronic | c.84-21_84-20 | e2-20 | tier3 | 92 | 1 | 0.011 | 119 | 15 | 0.112 |
| 596762 | 20 | 45355585 | 45355585 | G | A | SNP | SLC2A10 | ENST00000359271 | 1 | silent | c.1371 | p.A457 | tier1 | 51 | 0 | 0.000 | 59 | 24 | 0.286 |
| 596762 | 20 | 60895795 | 60895795 | C | G | SNP | LAMA5 | ENST00000252999 | -1 | missense | c.6648 | p.Q2216H | tier1 | 72 | 0 | 0.000 | 54 | 15 | 0.217 |
| 597501 | 2 | 21228297 | 21228297 | C | T | SNP | APOB | ENST00000233242 | -1 | missense | c.11443 | p.V3815M | tier1 | 991 | 7 | 0.007 | 874 | 56 | 0.060 |
| 597501 | 2 | 61006923 | 61006923 | G | A | SNP | PAPOLG | ENST00000238714 | 1 | intronic | c.833+13 | e9+13 | tier3 | 117 | 2 | 0.017 | 65 | 30 | 0.316 |
| 597501 | 2 | 66796098 | 66796099 | - | T | INS | MEIS1 | ENST00000488550 | 1 | 3_prime_untranslated_region | c.*187 | NULL | tier3 | 87 | 0 | 0.000 | 66 | 9 | 0.120 |
| 597501 | 2 | 99936266 | 99936266 | A | - | DEL | TXNDC9 | ENST00000264255 | -1 | splice_region_del | c.564-4 | e4-4 | tier3 | 31 | 0 | 0.000 | 38 | 11 | 0.225 |
| 597501 | 2 | 121744080 | 121744080 | G | A | SNP | GLI2 | ENST00000361492 | 1 | missense | c.2183 | p.R728Q | tier1 | 60 | 1 | 0.016 | 49 | 57 | 0.533 |
| 597501 | 2 | 179363925 | 179363925 | C | T | SNP | PLEKHA3 | ENST00000234453 | 1 | intronic | c.616-13 | e6-13 | tier3 | 67 | 1 | 0.015 | 59 | 13 | 0.181 |
| 597501 | 3 | 38913145 | 38913145 | C | A | SNP | SCN11A | ENST00000302328 | -1 | missense | c.3550 | p.V1184F | tier1 | 63 | 0 | 0.000 | 42 | 25 | 0.373 |
| 597501 | 3 | 138724564 | 138724564 | C | T | SNP | PRR23A | ENST00000383163 | -1 | missense | c.547 | p.A183T | tier1 | 178 | 2 | 0.011 | 193 | 33 | 0.145 |
| 597501 | 4 | 89022286 | 89022286 | C | T | SNP | ABCG2 | ENST00000237612 | -1 | intronic | c.1367+96 | e10+96 | tier3 | 196 | 8 | 0.039 | 176 | 176 | 0.499 |
| 597501 | 4 | 106157074 | 106157074 | C | T | SNP | TET2 | ENST00000380013 | 1 | nonsense | c.1975 | p.Q659* | tier1 | 510 | 27 | 0.050 | 320 | 312 | 0.491 |
| 597501 | 4 | 106183003 | 106183003 | C | T | SNP | TET2 | ENST00000380013 | 1 | nonsense | c.4042 | p.Q1348* | tier1 | 283 | 12 | 0.041 | 104 | 112 | 0.516 |
| 597501 | 4 | 109773495 | 109773495 | C | T | SNP | COL25A1 | ENST00000399132 | -1 | intronic | c.1345-59 | e25-59 | tier3 | 68 | 2 | 0.029 | 42 | 33 | 0.440 |
| 597501 | 4 | 115544516 | 115544516 | T | C | SNP | UGT8 | ENST00000310836 | 1 | silent | c.480 | p.T160 | tier1 | 120 | 0 | 0.000 | 79 | 54 | 0.406 |
| 597501 | 5 | 31435975 | 31435975 | C | A | SNP | DROSHA | ENST00000344624 | -1 | splice_region | c.2943-4 | e22-4 | tier3 | 45 | 1 | 0.021 | 41 | 13 | 0.241 |
| 597501 | 5 | 140203381 | 140203381 | C | T | SNP | PCDHA5 | ENST00000529859 | 1 | missense | c.2021 | p.A674V | tier1 | 489 | 0 | 0.000 | 598 | 21 | 0.034 |
| 597501 | 6 | 29069942 | 29069942 | G | A | SNP | OR2B3 | ENST00000377173 | -1 | 5_prime_flanking_region | c.-14917 | NULL | tier3 | 44 | 0 | 0.000 | 12 | 17 | 0.586 |
| 597501 | 6 | 41166075 | 41166075 | C | T | SNP | TREML2 | ENST00000483722 | -1 | missense | c.148 | p.V50M | tier1 | 117 | 0 | 0.000 | 216 | 17 | 0.073 |
| 597501 | 6 | 42330640 | 42330640 | C | T | SNP | TRERF1 | ENST00000541110 | -1 | intronic | c.1+95 | e0+95 | tier3 | 57 | 3 | 0.050 | 55 | 56 | 0.500 |
| 597501 | 6 | 123702474 | 123702474 | A | - | DEL | TRDN | ENST00000398178 | -1 | intronic | c.1186+41 | e16+41 | tier3 | 48 | 0 | 0.000 | 44 | 8 | 0.154 |
| 597501 | 6 | 136429913 | 136429913 | C | T | SNP | PDE7B | ENST00000308191 | 1 | missense | c.127 | p.R43C | tier1 | 126 | 0 | 0.000 | 154 | 73 | 0.322 |
| 597501 | 6 | 139237014 | 139237014 | C | T | SNP | REPS1 | ENST00000450536 | -1 | silent | c.1701 | p.R567 | tier1 | 142 | 4 | 0.027 | 113 | 78 | 0.408 |
| 597501 | 6 | 163990584 | 163990584 | A | G | SNP | QKI | ENST00000275262 | 1 | 3_prime_untranslated_region | c.*4842 | NULL | tier3 | 64 | 1 | 0.015 | 29 | 37 | 0.561 |
| 597501 | 7 | 131122618 | 131122618 | T | G | SNP | MKLN1 | ENST00000352689 | 1 | silent | c.1035 | p.R345 | tier1 | 121 | 6 | 0.047 | 59 | 58 | 0.496 |
| 597501 | 8 | 3045393 | 3045393 | G | A | SNP | CSMD1 | ENST00000520002 | -1 | missense | c.5621 | p.S1874L | tier1 | 338 | 11 | 0.031 | 282 | 234 | 0.453 |
| 597501 | 8 | 7878120 | 7878120 | C | T | SNP | FAM90A24P | ENST00000400118 | -1 | 5_prime_untranslated_region | c.-7878120 | NULL | tier3 | 2001 | 5 | 0.003 | 3019 | 225 | 0.069 |
| 597501 | 8 | 12516668 | 12516668 | C | T | SNP | RPS3AP35 | ENST00000478386 | -1 | 5_prime_flanking_region | c.-29903 | NULL | tier2 | 48 | 2 | 0.040 | 42 | 26 | 0.382 |
| 597501 | 8 | 99443925 | 99443926 | - | A | INS | KCNS2 | ENST00000287042 | 1 | 3_prime_untranslated_region | c.*2284 | NULL | tier3 | 62 | 0 | 0.000 | 61 | 13 | 0.176 |
| 597501 | 9 | 116079186 | 116079186 | C | T | SNP | WDR31 | ENST00000374193 | -1 | missense | c.947 | p.C316Y | tier1 | 59 | 2 | 0.033 | 59 | 60 | 0.504 |
| 597501 | 9 | 130693095 | 130693095 | C | - | DEL | ST6GALNAC6 | ENST00000291839 | -1 | 5_prime_flanking_region | c.-32835 | NULL | tier2 | 17 | 0 | 0.000 | 14 | 16 | 0.533 |
| 597501 | 10 | 49447649 | 49447649 | G | A | SNP | FRMPD2 | ENST00000374201 | -1 | missense | c.787 | p.R263C | tier1 | 78 | 4 | 0.049 | 76 | 73 | 0.490 |
| 597501 | 10 | 135015401 | 135015401 | C | T | SNP | KNDC1 | ENST00000368572 | 1 | missense | c.3392 | p.P1131L | tier1 | 48 | 3 | 0.059 | 30 | 32 | 0.516 |
| 597501 | 11 | 31531364 | 31531364 | C | T | SNP | ELP4 | ENST00000395934 | 1 | silent | c.33 | p.A11 | tier1 | 53 | 0 | 0.000 | 78 | 16 | 0.170 |
| 597501 | 12 | 117715866 | 117715866 | C | A | SNP | NOS1 | ENST00000317775 | -1 | missense | c.1562 | p.R521L | tier1 | 111 | 7 | 0.059 | 119 | 87 | 0.422 |
| 597501 | 13 | 25060389 | 25060389 | G | A | SNP | PARP4 | ENST00000381989 | -1 | silent | c.1269 | p.T423 | tier1 | 109 | 4 | 0.035 | 59 | 56 | 0.487 |
| 597501 | 14 | 25100251 | 25100251 | G | A | SNP | GZMB | ENST00000382542 | -1 | 3_prime_untranslated_region | c.*26 | NULL | tier3 | 131 | 3 | 0.022 | 95 | 77 | 0.448 |
| 597501 | 14 | 86089071 | 86089071 | C | G | SNP | FLRT2 | ENST00000330753 | 1 | missense | c.1213 | p.L405V | tier1 | 876 | 37 | 0.041 | 572 | 537 | 0.484 |
| 597501 | 15 | 34651895 | 34651895 | C | T | SNP | LPCAT4 | ENST00000314891 | -1 | missense | c.1294 | p.G432S | tier1 | 96 | 2 | 0.020 | 94 | 63 | 0.401 |
| 597501 | 15 | 91574882 | 91574882 | T | A | SNP | LOC101926911 | ENST00000557804 | 1 | intronic | c.NULL | NULL | tier3 | 84 | 4 | 0.046 | 48 | 18 | 0.273 |
| 597501 | 16 | 2522630 | 2522630 | G | A | SNP | NTN3 | ENST00000293973 | 1 | missense | c.928 | p.A310T | tier1 | 17 | 0 | 0.000 | 21 | 15 | 0.417 |
| 597501 | 17 | 8397209 | 8397209 | T | - | DEL | MYH10 | ENST00000488329 | -1 | 5_prime_untranslated_region | c.-8397209 | NULL | tier3 | 79 | 0 | 0.000 | 63 | 9 | 0.125 |
| 597501 | 17 | 11532944 | 11532944 | C | T | SNP | DNAH9 | ENST00000262442 | 1 | intronic | c.1518+43 | e7+43 | tier3 | 533 | 1 | 0.002 | 873 | 42 | 0.046 |
| 597501 | 17 | 28900664 | 28900664 | C | T | SNP | LOC101927093 | ENST00000436477 | -1 | rna | NULL | NULL | tier1 | 112 | 3 | 0.026 | 71 | 81 | 0.533 |
| 597501 | 17 | 29422352 | 29422352 | T | G | SNP | NF1 | ENST00000358273 | 1 | missense | c.25 | p.W9G | tier1 | 339 | 1 | 0.003 | 323 | 63 | 0.163 |
| 597501 | 17 | 30219342 | 30219342 | C | G | SNP | UTP6 | ENST00000261708 | -1 | missense | c.361 | p.A121P | tier1 | 138 | 2 | 0.014 | 58 | 37 | 0.390 |
| 597501 | 17 | 57647955 | 57647959 | TGAAG | - | DEL | DHX40 | ENST00000251241 | 1 | frame_shift_del | c.357_361 | p.E120fs | tier1 | 103 | 0 | 0.000 | 55 | 24 | 0.304 |
| 597501 | 17 | 74732959 | 74732959 | G | T | SNP | SRSF2 | ENST00000359995 | -1 | missense | c.284 | p.P95H | tier1 | 335 | 11 | 0.032 | 292 | 273 | 0.481 |
| 597501 | 18 | 29523048 | 29523048 | C | T | SNP | ENSG00000263823 | ENST00000580420 | 1 | rna | NULL | NULL | tier1 | 170 | 6 | 0.034 | 123 | 107 | 0.465 |
| 597501 | 19 | 17440815 | 17440815 | G | A | SNP | ANO8 | ENST00000159087 | -1 | missense | c.1282 | p.R428W | tier1 | 66 | 0 | 0.000 | 89 | 17 | 0.160 |
| 597501 | 19 | 47884624 | 47884624 | G | A | SNP | DHX34 | ENST00000486327 | 1 | 3_prime_untranslated_region | c.*47884624 | NULL | tier3 | 47 | 0 | 0.000 | 42 | 27 | 0.386 |
| 597501 | 19 | 54080590 | 54080590 | T | C | SNP | ZNF331 | ENST00000253144 | 1 | missense | c.776 | p.I259T | tier1 | 115 | 0 | 0.000 | 54 | 60 | 0.526 |
| 597501 | 20 | 48098486 | 48098486 | G | A | SNP | KCNB1 | ENST00000371741 | -1 | silent | c.532 | p.L178 | tier1 | 37 | 0 | 0.000 | 40 | 45 | 0.529 |
| 597501 | X | 10084387 | 10084387 | T | C | SNP | WWC3 | ENST00000380861 | 1 | intronic | c.975-71 | e9-71 | tier3 | 143 | 1 | 0.007 | 82 | 72 | 0.468 |
| 597501 | X | 70357542 | 70357542 | C | T | SNP | MED12 | ENST00000333646 | 1 | intronic | c.5836-34 | e41-34 | tier3 | 446 | 24 | 0.051 | 379 | 379 | 0.499 |
| 621996 | 1 | 4772000 | 4772000 | C | A | SNP | AJAP1 | ENST00000378190 | 1 | missense | c.70 | p.H24N | tier1 | 15 | 0 | 0.000 | 16 | 14 | 0.467 |
| 621996 | 1 | 91404719 | 91404719 | T | C | SNP | ZNF644 | ENST00000337393 | -1 | missense | c.2192 | p.N731S | tier1 | 67 | 3 | 0.043 | 71 | 41 | 0.366 |
| 621996 | 1 | 152186038 | 152186038 | A | - | DEL | HRNR | ENST00000368801 | -1 | frame_shift_del | c.8067 | p.H2690fs | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|--------------|-----|----------|-----------------|-----------------|--------|-----------------------------|-----------------|-----------------------|-------|------|-------|-------|------|-------|-------|
| 621996 | 2 | 166223736 | 166223736 | G | A | SNP | SCN2A | ENST00000283256 | 1 | missense | c.3530 | p.R1177Q | tier1 | 156 | 1 | 0.006 | 179 | 97 | 0.351 |
| 621996 | 2 | 179604342 | 179604342 | C | T | SNP | TTN | ENST00000342175 | -1 | missense | c.13105 | p.V4369M | tier1 | 51 | 3 | 0.056 | 29 | 34 | 0.540 |
| 621996 | 2 | 197709191 | 197709191 | T | C | SNP | PGAP1 | ENST00000354764 | -1 | intronic | c.2337+57 | e24+57 | tier3 | 219 | 0 | 0.000 | 234 | 111 | 0.320 |
| 621996 | 3 | 38291377 | 38291377 | G | A | SNP | OXSR1 | ENST00000311806 | 1 | intronic | c.1323-80 | e15-80 | tier3 | 79 | 0 | 0.000 | 79 | 28 | 0.262 |
| 621996 | 3 | 50312255 | 50312255 | G | G | SNP | SEMA3B | ENST00000416295 | 1 | 3_prime_untranslated_region | c.*50312255 | NULL | tier2 | 46 | 2 | 0.042 | 58 | 30 | 0.341 |
| 621996 | 3 | 125879686 | 125879686 | C | A | SNP | ALDH1L1 | ENST00000393434 | -1 | splice_region | c.127+10 | e1+10 | tier3 | 66 | 0 | 0.000 | 81 | 36 | 0.308 |
| 621996 | 4 | 125592366 | 125592366 | T | G | SNP | ANKRD50 | ENST00000504087 | -1 | missense | c.2066 | p.H689P | tier1 | 35 | 0 | 0.000 | 24 | 16 | 0.400 |
| 621996 | 5 | 133483867 | 133483867 | T | G | SNP | TCF7 | ENST00000395023 | 1 | 3_prime_untranslated_region | c.*1873 | NULL | tier3 | 116 | 3 | 0.025 | 128 | 78 | 0.379 |
| 621996 | 5 | 154255096 | 154255096 | C | A | SNP | CNOT8 | ENST00000285896 | 1 | 3_prime_untranslated_region | c.*97 | NULL | tier3 | 66 | 1 | 0.015 | 78 | 33 | 0.297 |
| 621996 | 5 | 176012012 | 176012012 | C | A | SNP | CDHR2 | ENST00000261944 | 1 | missense | c.2730 | p.N910K | tier1 | 123 | 4 | 0.031 | 126 | 65 | 0.340 |
| 621996 | 5 | 176016191 | 176016191 | C | T | SNP | CDHR2 | ENST00000261944 | 1 | splice_region | c.3008+8 | e21+8 | tier3 | 94 | 0 | 0.000 | 89 | 69 | 0.437 |
| 621996 | 6 | 36569780 | 36569780 | A | G | SNP | SRSF3 | ENST00000373715 | 1 | 3_prime_untranslated_region | c.*14 | NULL | tier3 | 38 | 0 | 0.000 | 35 | 17 | 0.321 |
| 621996 | 7 | 48964818 | 48964818 | G | A | SNP | ENSG00000218305 | ENST00000406468 | 1 | 3_prime_untranslated_region | c.*48964818 | NULL | tier2 | 184 | 0 | 0.000 | 142 | 66 | 0.317 |
| 621996 | 7 | 100846447 | 100846447 | C | T | SNP | DGAT2L7P | ENST00000379421 | -1 | 5_prime_untranslated_region | c.-100846447 | NULL | tier2 | 57 | 0 | 0.000 | 83 | 49 | 0.371 |
| 621996 | 7 | 112724366 | 112724366 | C | T | SNP | GPR85 | ENST00000297146 | -1 | silent | c.411 | p.T137 | tier1 | 60 | 0 | 0.000 | 61 | 36 | 0.371 |
| 621996 | 8 | 3224668 | 3224668 | C | T | SNP | CSMD1 | ENST00000520002 | -1 | missense | c.3004 | p.G1002R | tier1 | 558 | 11 | 0.019 | 517 | 311 | 0.375 |
| 621996 | 9 | 141071154 | 141071154 | T | C | SNP | TUBBP5 | ENST00000290377 | 1 | rna | NULL | NULL | tier1 | 178 | 1 | 0.006 | 245 | 19 | 0.072 |
| 621996 | 11 | 6243580 | 6243580 | G | A | SNP | FAM160A2 | ENST00000265978 | -1 | intronic | c.1191+92 | e5+92 | tier3 | 59 | 2 | 0.033 | 60 | 43 | 0.418 |
| 621996 | 11 | 24750764 | 24750764 | C | T | SNP | LUZP2 | ENST00000336930 | 1 | nonsense | c.112 | p.R38* | tier1 | 107 | 4 | 0.036 | 91 | 64 | 0.413 |
| 621996 | 11 | 46404356 | 46404356 | G | A | SNP | MDK | ENST00000489525 | 1 | missense | c.464 | p.R155H | tier1 | 36 | 1 | 0.021 | 43 | 19 | 0.247 |
| 621996 | 11 | 116718092 | 116718092 | C | T | SNP | SIK3 | ENST00000375300 | -1 | intronic | c.3808+100 | e22+100 | tier3 | 35 | 0 | 0.000 | 37 | 21 | 0.362 |
| 621996 | 12 | 113405327 | 113405327 | C | T | SNP | OAS3 | ENST00000228928 | 1 | missense | c.2794 | p.R932W | tier1 | 28 | 1 | 0.035 | 37 | 24 | 0.393 |
| 621996 | 12 | 123005987 | 123005988 | - | A | INS | RSRC2 | ENST00000331738 | -1 | intronic | c.164-13_164-12 | e3-12 | tier3 | 53 | 0 | 0.000 | 57 | 23 | 0.288 |
| 621996 | 12 | 123419982 | 123419982 | G | A | SNP | ABCB9 | ENST00000442028 | -1 | splice_region | c.1744-4 | e9-4 | tier3 | 38 | 1 | 0.026 | 57 | 36 | 0.387 |
| 621996 | 14 | 24887568 | 24887568 | C | G | SNP | NYNRIN | ENST00000382554 | 1 | 3_prime_untranslated_region | c.*916 | NULL | tier3 | 94 | 1 | 0.011 | 58 | 37 | 0.390 |
| 621996 | 14 | 63269293 | 63269293 | A | G | SNP | KCNH5 | ENST00000322893 | -1 | missense | c.1576 | p.S526P | tier1 | 38 | 0 | 0.000 | 44 | 33 | 0.429 |
| 621996 | 15 | 90631934 | 90631934 | C | T | SNP | IDH2 | ENST00000330062 | -1 | missense | c.419 | p.R140Q | tier1 | 422 | 0 | 0.000 | 475 | 258 | 0.351 |
| 621996 | 15 | 102294753 | 102294753 | T | C | SNP | DNM1P47 | ENST00000561463 | 1 | rna | NULL | NULL | tier1 | 34 | 1 | 0.029 | 41 | 18 | 0.305 |
| 621996 | 16 | 22545618 | 22545629 | TCCACCCTCAGC | - | DEL | NPIP5 | ENST00000424340 | 1 | in_frame_del | c.1314_1325 | p.APPS442in_frame_del | tier1 | 17 | 0 | 0.000 | 22 | 12 | 0.353 |
| 621996 | 16 | 33585853 | 33585853 | C | T | SNP | ENSG00000271178 | ENST00000562905 | 1 | 5_prime_flanking_region | c.-43828 | NULL | tier3 | 107 | 0 | 0.000 | 204 | 16 | 0.072 |
| 621996 | 16 | 33585887 | 33585887 | T | C | SNP | ENSG00000271178 | ENST00000562905 | 1 | 5_prime_flanking_region | c.-43794 | NULL | tier3 | 134 | 0 | 0.000 | 240 | 16 | 0.063 |
| 621996 | 17 | 12921348 | 12921348 | ELAC2 | C | SNP | ELAC2 | ENST00000338034 | -1 | 5_prime_untranslated_region | c.-84 | NULL | tier2 | 9 | 0 | 0.000 | 5 | 9 | 0.643 |
| 621996 | 17 | 45249355 | 45249355 | A | G | SNP | CDC27 | ENST00000531206 | -1 | missense | c.179 | p.L60P | tier1 | 57 | 1 | 0.017 | 73 | 36 | 0.327 |
| 621996 | 17 | 74732959 | 74732959 | G | T | SNP | SRSF2 | ENST00000359995 | -1 | missense | c.284 | p.P95H | tier1 | 199 | 5 | 0.025 | 271 | 169 | 0.384 |
| 621996 | 18 | 55024279 | 55024279 | A | C | SNP | ST8SIA3 | ENST00000324000 | 1 | silent | c.438 | p.S146 | tier1 | 85 | 1 | 0.012 | 59 | 42 | 0.416 |
| 621996 | 19 | 52726744 | 52726744 | G | A | SNP | PPP2R1A | ENST00000322088 | 1 | intronic | c.1661+1250 | e13+1250 | tier4 | 41 | 2 | 0.047 | 34 | 20 | 0.370 |
| 621996 | 20 | 29623180 | 29623180 | T | C | SNP | FRG1B | ENST00000482423 | 1 | rna | NULL | NULL | tier1 | 1531 | 18 | 0.012 | 2465 | 72 | 0.028 |
| 621996 | 20 | 31022441 | 31022442 | - | G | INS | ASXL1 | ENST00000375687 | 1 | frame_shift_ins | c.1926_1927 | p.G645fs | tier1 | 693 | 14 | 0.020 | 805 | 301 | 0.272 |
| 621996 | 21 | 36259149 | 36259158 | GATGGGCAGG | - | DEL | RUNX1 | ENST00000300305 | -1 | frame_shift_del | c.342_333 | p.P113fs | tier1 | 795 | 9 | 0.011 | 772 | 132 | 0.146 |
| 621996 | 22 | 20172250 | 20172250 | G | A | SNP | LINC00896 | ENST00000423736 | 1 | 5_prime_flanking_region | c.-21850 | NULL | tier3 | 82 | 0 | 0.000 | 59 | 33 | 0.359 |
| 621996 | X | 123205158 | 123205159 | - | AT | INS | STAG2 | ENST00000218089 | 1 | frame_shift_ins | c.2518_2519 | p.N841fs | tier1 | 189 | 0 | 0.000 | 127 | 180 | 0.586 |
| 621996 | X | 151092940 | 151092940 | G | A | SNP | MAGEA4 | ENST00000276344 | 1 | silent | c.804 | p.A268 | tier1 | 99 | 2 | 0.020 | 65 | 43 | 0.398 |
| 621996 | Y | 14077927 | 14077927 | C | T | SNP | MXRA5P1 | ENST00000420610 | 1 | 3_prime_untranslated_region | c.*14077927 | NULL | tier2 | 42 | 0 | 0.000 | 31 | 46 | 0.597 |
| 624702 | 1 | 143914212 | 143914212 | T | A | SNP | SRGAP2P2 | ENST00000494534 | 1 | 3_prime_untranslated_region | c.*143914212 | NULL | tier3 | 23 | 0 | 0.000 | 117 | 83 | 0.407 |
| 624702 | 1 | 156043138 | 156043138 | A | C | SNP | LMNA | ENST00000368300 | 1 | 5_prime_flanking_region | c.-41572 | NULL | tier3 | 83 | 1 | 0.012 | 129 | 78 | 0.375 |
| 624702 | 1 | 158057760 | 158057760 | G | A | SNP | KIRREL | ENST00000368173 | 1 | intronic | c.768-36 | e7-36 | tier2 | 29 | 1 | 0.033 | 138 | 74 | 0.349 |
| 624702 | 1 | 201868536 | 201868536 | T | G | SNP | LMOD1 | ENST00000367288 | -1 | silent | c.1605 | p.P535 | tier1 | 60 | 0 | 0.000 | 159 | 21 | 0.115 |
| 624702 | 1 | 214204427 | 214204427 | C | T | SNP | PROX1 | ENST00000261454 | 1 | intronic | c.2029-4565 | e4-4565 | tier3 | 15 | 0 | 0.000 | 39 | 23 | 0.365 |
| 624702 | 2 | 133403009 | 133403009 | C | T | SNP | GPR39 | ENST00000329321 | 1 | missense | c.1192 | p.R398C | tier1 | 26 | 0 | 0.000 | 74 | 34 | 0.315 |
| 624702 | 2 | 179454700 | 179454700 | C | T | SNP | TTN | ENST00000342175 | -1 | silent | c.35133 | p.E11711 | tier1 | 149 | 4 | 0.026 | 156 | 102 | 0.394 |
| 624702 | 2 | 187627370 | 187627370 | G | T | SNP | FAM171B | ENST00000304698 | 1 | silent | c.2301 | p.G767 | tier1 | 46 | 0 | 0.000 | 81 | 39 | 0.325 |
| 624702 | 2 | 238287631 | 238287631 | C | T | SNP | COL6A3 | ENST00000295550 | -1 | silent | c.2145 | p.S715 | tier1 | 21 | 0 | 0.000 | 60 | 29 | 0.326 |
| 624702 | 3 | 9747109 | 9747109 | A | C | SNP | CPNE9 | ENST00000383832 | 1 | intronic | c.261-39 | e5-39 | tier3 | 157 | 1 | 0.006 | 515 | 43 | 0.076 |
| 624702 | 3 | 51312620 | 51312620 | G | A | SNP | DOCK3 | ENST00000266037 | 1 | missense | c.2659 | p.V887I | tier1 | 29 | 0 | 0.000 | 59 | 33 | 0.355 |
| 624702 | 4 | 2951749 | 2951749 | C | A | SNP | NOP14 | ENST00000416614 | -1 | missense | c.1194 | p.E398D | tier1 | 47 | 4 | 0.078 | 96 | 60 | 0.382 |
| 624702 | 4 | 76711225 | 76711225 | T | A | SNP | USO1 | ENST00000538159 | 1 | intronic | c.1003-78 | e11-78 | tier3 | 50 | 0 | 0.000 | 22 | 8 | 0.070 |
| 624702 | 4 | 106157711 | 106157712 | - | T | INS | TET2 | ENST00000380013 | 1 | frame_shift_ins | c.2612_2613 | p.V872fs | tier1 | 57 | 2 | 0.034 | 39 | 68 | 0.636 |
| 624702 | 5 | 80368977 | 80368977 | T | C | SNP | RASGRF2 | ENST00000265080 | 1 | intronic | c.634-41 | e5-41 | tier3 | 64 | 1 | 0.015 | 92 | 61 | 0.396 |
| 624702 | 5 | 102503818 | 102503818 | G | A | SNP | PPIP5K2 | ENST00000451606 | 1 | missense | c.2105 | p.R702H | tier1 | 306 | 9 | 0.029 | 202 | 123 | 0.379 |
| 624702 | 5 | 139505517 | 139505517 | T | G | SNP | CYSTM1 | ENST00000261811 | 1 | 5_prime_flanking_region | c.-68534 | NULL | tier3 | 102 | 7 | 0.064 | 62 | 55 | 0.470 |
| 624702 | 5 | 140564550 | 140564550 | C | T | SNP | PCDHB16 | ENST00000361016 | 1 | 3_prime_untranslated_region | c.*85 | NULL | tier2 | 64 | 2 | 0.030 | 99 | 64 | 0.390 |
| 624702 | 5 | 159901462 | 159901463 | CT | - | DEL | MIR146A | ENST00000517927 | 1 | intronic | c.NULL | NULL | tier3 | 42 | 0 | 0.000 | 100 | 25 | 0.200 |
| 624702 | 6 | 35936771 | 35936771 | C | T | SNP | SLC26A8 | ENST00000466805 | -1 | 5_prime_untranslated_region | c.-35936771 | NULL | tier3 | 43 | 0 | 0.000 | 104 | 72 | 0.409 |
| 624702 | 6 | 109830321 | 109830321 | G | A | SNP | AKD1 | ENST00000424296 | -1 | intronic | c.4481+61 | e32+61 | tier3 | 69 | 0 | 0.000 | 95 | 69 | 0.421 |
| 624702 | 7 | 6031678 | 6031678 | A | G | SNP | PMS2 | ENST00000265849 | -1 | missense | c.914 | p.L305P | tier1 | 98 | 5 | 0.049 | 153 | 92 | 0.376 |
| 624702 | 7 | 158200082 | 158200082 | G | A | SNP | PTPRN2 | ENST00000404321 | -1 | intronic | c.232+82345 | e2+82345 | tier3 | 18 | 0 | 0.000 | 135 | 54 | 0.281 |
| 624702 | 8 | 7698557 | 7698557 | A | SNP | DEFB104A | ENST00000314265 | 1 | silent | c.60 | p.V20 | tier1 | 114 | 0 | 0.000 | 228 | 47 | 0.168 | |
| 624702 | 8 | 10467587 | 10467587 | T | C | SNP | RP1L1 | ENST00000382483 | -1 | missense | c.4021 | p.T1341A | tier1 | 134 | 1 | 0.007 | 263 | 22 | 0.076 |
| 62 | | | | | | | | | | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|---------|--|-------|-----|------------------------|-----------------|----|-----------------------------|---------------------|-----------|-------|-----|----|-------|------|-----|-------|
| 624702 | 14 | 50789470 | 50789470 | G | | T | SNP | <i>ATP5S</i> | ENST00000426751 | 1 | missense | c.394 | p.A132S | tier1 | 36 | 2 | 0.053 | 58 | 38 | 0.396 |
| 624702 | 15 | 93568489 | 93568490 | - | | TGT | INS | <i>CHD2</i> | ENST00000394196 | 1 | 3_prime_untranslated_region | c.*554 | NULL | tier3 | 141 | 0 | 0.000 | 326 | 120 | 0.269 |
| 624702 | 17 | 74732959 | 74732959 | G | | T | SNP | <i>SRSF2</i> | ENST00000392485 | -1 | missense | c.284 | p.P95H | tier1 | 72 | 2 | 0.027 | 275 | 158 | 0.364 |
| 624702 | 19 | 6678085 | 6678085 | A | | G | SNP | <i>C3</i> | ENST00000245907 | -1 | intronic | c.4851-51 | e41-51 | tier3 | 16 | 0 | 0.000 | 68 | 34 | 0.330 |
| 624702 | 19 | 10800503 | 10800506 | AAAT | | - | DEL | <i>ILF3</i> | ENST00000449870 | 1 | 3_prime_untranslated_region | c.*515 | NULL | tier3 | 98 | 2 | 0.020 | 155 | 60 | 0.279 |
| 624702 | 19 | 42861028 | 42861028 | C | | T | SNP | <i>MEGF8</i> | ENST00000251268 | 1 | silent | c.4725 | p.P1575 | tier1 | 94 | 6 | 0.060 | 420 | 286 | 0.401 |
| 624702 | 20 | 1350154 | 1350154 | T | | G | SNP | <i>FKBP1A</i> | ENST00000400137 | -1 | 3_prime_untranslated_region | c.*2602 | NULL | tier3 | 121 | 2 | 0.016 | 188 | 30 | 0.136 |
| 624702 | 20 | 18744043 | 18744043 | A | | G | SNP | <i>DTD1</i> | ENST00000377452 | 1 | 3_prime_untranslated_region | c.*19147 | NULL | tier3 | 96 | 3 | 0.030 | 104 | 69 | 0.390 |
| 624702 | 20 | 44505588 | 44505588 | C | | T | SNP | <i>ZSWIM3</i> | ENST00000255152 | 1 | nonsense | c.391 | p.Q131* | tier1 | 28 | 1 | 0.035 | 85 | 61 | 0.412 |
| 624702 | 21 | 27253248 | 27253248 | T | | C | SNP | <i>APP</i> | ENST00000346798 | -1 | 3_prime_untranslated_region | c.*733 | NULL | tier3 | 152 | 1 | 0.007 | 295 | 24 | 0.075 |
| 624702 | 22 | 20715828 | 20715828 | C | | G | SNP | <i>ENSG00000188280</i> | ENST00000434783 | 1 | intronic | c.NULL | NULL | tier2 | 30 | 0 | 0.000 | 218 | 50 | 0.185 |
| 624702 | 22 | 24176559 | 24176559 | A | | C | SNP | <i>SMARCB1</i> | ENST00000263121 | 1 | 3_prime_untranslated_region | c.*192 | NULL | tier3 | 85 | 0 | 0.000 | 157 | 18 | 0.102 |
| 624702 | 22 | 38341047 | 38341047 | A | | G | SNP | <i>C22orf23</i> | ENST00000249079 | -1 | splice_site | c.481+2 | e4+2 | tier1 | 38 | 0 | 0.000 | 105 | 56 | 0.348 |
| 624702 | X | 123176411 | 123176411 | A | | G | SNP | <i>STAG2</i> | ENST00000218089 | 1 | splice_region | c.386-8 | e5-8 | tier3 | 142 | 0 | 0.000 | 117 | 253 | 0.682 |
| 624702 | X | 153214721 | 153214721 | T | | G | SNP | <i>HCFC1</i> | ENST00000310441 | -1 | 3_prime_untranslated_region | c.*77 | NULL | tier3 | 27 | 0 | 0.000 | 74 | 23 | 0.232 |
| 625338 | 17 | 21562303 | 21562303 | A | | T | SNP | <i>ENSG00000264617</i> | ENST00000584107 | -1 | 5_prime_untranslated_region | c.-21562303 | NULL | tier3 | 193 | 0 | 0.000 | 242 | 17 | 0.066 |
| 625338 | 20 | 31022824 | 31022824 | C | | G | SNP | <i>ASXL1</i> | ENST00000375687 | 1 | nonsense | c.2309 | p.S770* | tier1 | 591 | 1 | 0.002 | 1201 | 40 | 0.032 |
| 625338 | 21 | 36231782 | 36231782 | C | | T | SNP | <i>RUNX1</i> | ENST00000300305 | -1 | missense | c.602 | p.R201Q | tier1 | 786 | 2 | 0.003 | 1092 | 28 | 0.025 |
| 625338 | 21 | 36252865 | 36252865 | C | | T | SNP | <i>RUNX1</i> | ENST00000300305 | -1 | missense | c.497 | p.R166Q | tier1 | 984 | 0 | 0.000 | 1534 | 46 | 0.029 |
| 627844 | 1 | 240073885 | 240073885 | A | | - | DEL | <i>CHRM3</i> | ENST00000255380 | 1 | 3_prime_untranslated_region | c.*1361 | NULL | tier3 | 71 | 0 | 0.000 | 109 | 16 | 0.128 |
| 627844 | 2 | 10224073 | 10224073 | G | | A | SNP | <i>ENSG00000233502</i> | ENST00000425235 | 1 | splice_site | c.165+1 | e1+1 | tier1 | 120 | 0 | 0.000 | 119 | 51 | 0.298 |
| 627844 | 2 | 87754980 | 87754981 | - | | T | INS | <i>LINC00152</i> | ENST00000331944 | 1 | rna | NULL | NULL | tier1 | 42 | 0 | 0.000 | 74 | 13 | 0.149 |
| 627844 | 3 | 31674390 | 31674390 | A | | T | SNP | <i>STT3B</i> | ENST00000295770 | 1 | intronic | c.2188-37 | e15-37 | tier3 | 90 | 0 | 0.000 | 140 | 56 | 0.286 |
| 627844 | 3 | 49036909 | 49036910 | - | | C | INS | <i>P4HTM</i> | ENST00000343546 | 1 | intronic | c.437-1962_437-1961 | e3-1961 | tier3 | 76 | 2 | 0.026 | 81 | 29 | 0.264 |
| 627844 | 3 | 122432409 | 122432409 | A | | G | SNP | <i>PARP14</i> | ENST00000474629 | 1 | missense | c.3758 | p.N1253S | tier1 | 97 | 0 | 0.000 | 169 | 71 | 0.296 |
| 627844 | 4 | 20530641 | 20530641 | G | | A | SNP | <i>SLIT2</i> | ENST00000504154 | 1 | missense | c.1532 | p.R511H | tier1 | 124 | 0 | 0.000 | 173 | 15 | 0.080 |
| 627844 | 6 | 69795034 | 69795034 | C | | T | SNP | <i>BAI3</i> | ENST00000370598 | 1 | intronic | c.2480+9104 | e15+9104 | tier3 | 74 | 0 | 0.000 | 87 | 38 | 0.304 |
| 627844 | 6 | 87968388 | 87968395 | AGTTGGT | | - | DEL | <i>ZNF292</i> | ENST00000369577 | 1 | frame_shift_del | c.5041_5048 | p.S1681fs | tier1 | 78 | 0 | 0.000 | 163 | 30 | 0.155 |
| 627844 | 6 | 135515531 | 135515531 | G | | A | SNP | <i>MYB</i> | ENST00000341911 | 1 | missense | c.881 | p.R294Q | tier1 | 62 | 1 | 0.016 | 89 | 17 | 0.160 |
| 627844 | 6 | 146719968 | 146719968 | C | | T | SNP | <i>GRM1</i> | ENST00000282753 | 1 | missense | c.1793 | p.A598V | tier1 | 111 | 1 | 0.009 | 121 | 59 | 0.326 |
| 627844 | 7 | 27236580 | 27236580 | G | | A | SNP | <i>HOXA13</i> | ENST00000222753 | -1 | 3_prime_untranslated_region | c.*1237 | NULL | tier3 | 169 | 2 | 0.012 | 441 | 221 | 0.332 |
| 627844 | 7 | 80303472 | 80303472 | G | | A | SNP | <i>CD36</i> | ENST00000309881 | 1 | 3_prime_untranslated_region | c.*9 | NULL | tier2 | 89 | 0 | 0.000 | 165 | 104 | 0.387 |
| 627844 | 8 | 82373788 | 82373788 | G | | A | SNP | <i>FABP9</i> | ENST00000379071 | -1 | 5_prime_untranslated_region | c.-30 | NULL | tier3 | 50 | 0 | 0.000 | 86 | 29 | 0.252 |
| 627844 | 8 | 105503259 | 105503259 | C | | T | SNP | <i>LRP12</i> | ENST00000424843 | -1 | missense | c.2165 | p.R722H | tier1 | 107 | 1 | 0.009 | 101 | 41 | 0.289 |
| 627844 | 10 | 120896686 | 120896686 | A | | - | DEL | <i>FAM45A</i> | ENST00000489936 | 1 | rna | NULL | NULL | tier1 | 129 | 0 | 0.000 | 306 | 20 | 0.061 |
| 627844 | 11 | 49839389 | 49839389 | G | | A | SNP | <i>ENSG00000254487</i> | ENST00000532682 | 1 | 3_prime_untranslated_region | c.*49839389 | NULL | tier2 | 217 | 0 | 0.000 | 276 | 25 | 0.083 |
| 627844 | 11 | 57268743 | 57268744 | - | | GGGT | INS | <i>SLC43A1</i> | ENST00000278426 | -1 | frame_shift_ins | c.214_213 | p.Q71fs | tier1 | 69 | 0 | 0.000 | 60 | 9 | 0.130 |
| 627844 | 11 | 57268744 | 57268745 | - | | CT | INS | <i>SLC43A1</i> | ENST00000278426 | -1 | frame_shift_ins | c.213_212 | p.Q72fs | tier1 | 69 | 0 | 0.000 | 60 | 9 | 0.130 |
| 627844 | 11 | 73980724 | 73980724 | A | | G | SNP | <i>P4HA3</i> | ENST00000331597 | -1 | silent | c.1440 | p.Y480 | tier1 | 52 | 0 | 0.000 | 39 | 20 | 0.339 |
| 627844 | 12 | 8186333 | 8186333 | G | | T | SNP | <i>FOXJ2</i> | ENST00000162391 | 1 | 5_prime_untranslated_region | c.-6096 | NULL | tier2 | 81 | 1 | 0.012 | 90 | 14 | 0.135 |
| 627844 | 12 | 46231475 | 46231476 | - | | GACC | INS | <i>ARID2</i> | ENST00000334344 | 1 | frame_shift_ins | c.1315_1316 | p.V439fs | tier1 | 62 | 1 | 0.016 | 103 | 21 | 0.169 |
| 627844 | 12 | 64841839 | 64841839 | A | | G | SNP | <i>XPOT</i> | ENST00000332707 | 1 | intronic | c.2863-46 | e24-46 | tier3 | 86 | 0 | 0.000 | 167 | 15 | 0.082 |
| 627844 | 12 | 112185009 | 112185009 | C | | T | SNP | <i>ACAD10</i> | ENST00000546647 | 1 | 3_prime_untranslated_region | c.*112185009 | NULL | tier3 | 68 | 0 | 0.000 | 82 | 20 | 0.196 |
| 627844 | 12 | 112884192 | 112884192 | C | | T | SNP | <i>PTPN11</i> | ENST00000351677 | 1 | missense | c.127 | p.L43F | tier1 | 518 | 1 | 0.002 | 638 | 100 | 0.135 |
| 627844 | 12 | 133352961 | 133352961 | A | | G | SNP | <i>GOLGA3</i> | ENST00000204726 | -1 | intronic | c.3978+259 | e20+259 | tier3 | 144 | 2 | 0.014 | 144 | 18 | 0.107 |
| 627844 | 13 | 80095144 | 80095144 | T | | - | DEL | <i>NDFIP2</i> | ENST00000218652 | 1 | intronic | c.487+34 | e2+34 | tier3 | 68 | 1 | 0.015 | 115 | 19 | 0.142 |
| 627844 | 14 | 21679136 | 21679137 | - | | T | INS | <i>HNRNPC</i> | ENST00000553444 | -1 | 5_prime_untranslated_region | c.-21679136 | NULL | tier3 | 103 | 2 | 0.019 | 183 | 50 | 0.215 |
| 627844 | 14 | 86088433 | 86088433 | T | | A | SNP | <i>FLRT2</i> | ENST00000330753 | 1 | missense | c.575 | p.I192N | tier1 | 809 | 3 | 0.004 | 1009 | 202 | 0.167 |
| 627844 | 15 | 28805975 | 28805977 | AGG | | - | DEL | <i>ENSG00000227717</i> | ENST00000419645 | 1 | 3_prime_untranslated_region | c.*28805975 | NULL | tier4 | 41 | 0 | 0.000 | 70 | 15 | 0.177 |
| 627844 | 15 | 75798204 | 75798204 | G | | C | SNP | <i>PTPN9</i> | ENST00000306726 | -1 | silent | c.780 | p.P260 | tier1 | 162 | 1 | 0.006 | 217 | 23 | 0.096 |
| 627844 | 16 | 20563602 | 20563602 | G | | C | SNP | <i>ACSM2B</i> | ENST00000329697 | -1 | missense | c.758 | p.A253G | tier1 | 127 | 1 | 0.008 | 164 | 28 | 0.146 |
| 627844 | 16 | 47675018 | 47675018 | G | | A | SNP | <i>PHKB</i> | ENST00000299167 | 1 | intronic | c.1514+17 | e15+17 | tier3 | 117 | 3 | 0.025 | 164 | 66 | 0.287 |
| 627844 | 16 | 89836214 | 89836214 | G | | C | SNP | <i>FANCA</i> | ENST00000567205 | -1 | 3_prime_untranslated_region | c.*27 | NULL | tier3 | 894 | 8 | 0.009 | 862 | 422 | 0.328 |
| 627844 | 17 | 9282009 | 9282009 | G | | G | SNP | <i>STX8</i> | ENST00000306357 | -1 | intronic | c.542-39 | e7-39 | tier3 | 127 | 0 | 0.000 | 165 | 75 | 0.313 |
| 627844 | 17 | 11622752 | 11622752 | C | | A | SNP | <i>DNAH9</i> | ENST00000262442 | 1 | missense | c.5654 | p.R1885H | tier1 | 434 | 5 | 0.011 | 432 | 208 | 0.325 |
| 627844 | 17 | 26923218 | 26923218 | C | | A | SNP | <i>SPAG5</i> | ENST00000321765 | -1 | intronic | c.177+2310 | e2+2310 | tier4 | 153 | 0 | 0.000 | 217 | 16 | 0.068 |
| 627844 | 17 | 34894610 | 34894610 | T | | - | DEL | <i>PIGW</i> | ENST00000328396 | 1 | 3_prime_untranslated_region | c.*145 | NULL | tier4 | 216 | 5 | 0.023 | 343 | 36 | 0.095 |
| 627844 | 18 | 43532461 | 43532462 | - | | ACCC | INS | <i>EPG5</i> | ENST00000282041 | -1 | frame_shift_ins | c.1157_1156 | p.S386fs | tier1 | 102 | 0 | 0.000 | 100 | 20 | 0.167 |
| 627844 | 18 | 61323095 | 61323095 | G | | A | SNP | <i>SERPINB3</i> | ENST00000283752 | -1 | silent | c.969 | p.L323 | tier1 | 121 | 2 | 0.016 | 124 | 57 | 0.313 |
| 627844 | 19 | 20045145 | 20045145 | G | | A | SNP | <i>ZNF93</i> | ENST00000343769 | 1 | missense | c.1381 | p.A461T | tier1 | 76 | 0 | 0.000 | 128 | 63 | 0.328 |
| 627844 | 20 | 57464277 | 57464278 | - | | CGGCG | INS | <i>GNAS</i> | ENST00000469431 | 1 | rna | NULL | NULL | tier1 | 13 | 0 | 0.000 | 6 | 16 | 0.727 |
| 627844 | 21 | 36259256 | 36259257 | - | | CA | INS | <i>RUNX1</i> | ENST00000300305 | -1 | frame_shift_ins | c.235_234 | p.V78fs | tier1 | 983 | 10 | 0.010 | 852 | 354 | 0.294 |
| 627844 | X | 13337504 | 13337504 | C | | T | SNP | <i>ATXN3L</i> | ENST00000380622 | -1 | missense | c.550 | p.E184K | tier1 | 46 | 0 | 0.000 | 34 | 50 | 0.595 |
| 627844 | X | 119387527 | 119387527 | A | | G | SNP | <i>ZBTB33</i> | ENST00000326624 | 1 | missense | c.257 | p.Y86C | tier1 | 435 | 16 | 0.035 | 284 | 472 | 0.624 |
| 627844 | Y | 13311260 | 13311260 | A | | G | SNP | <i>ENSG00000270570</i> | ENST00000603467 | -1 | 5_prime_flanking_region | c.-47697 | NULL | tier3 | 138 | 3 | 0.021 | 184 | 23 | 0.111 |
| 635308 | 1 | 653070 | | | | | | | | | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-----|---|-----|------------------------|-----------------|----|-----------------------------|--------------|-----------|-------|-----|---|-------|------|-----|-------|
| 635308 | 4 | 68930609 | 68930609 | G | A | SNP | <i>TMPRSS11F</i> | ENST00000356291 | -1 | missense | c.809 | p.P270L | tier1 | 75 | 0 | 0.000 | 156 | 21 | 0.119 |
| 635308 | 4 | 183723682 | 183723682 | C | G | SNP | <i>TENM3</i> | ENST00000511685 | 1 | 3_prime_untranslated_region | c.*2178 | NULL | tier2 | 87 | 0 | 0.000 | 177 | 33 | 0.157 |
| 635308 | 4 | 187088020 | 187088020 | G | C | SNP | <i>FAM149A</i> | ENST00000356371 | 1 | intronic | c.2038-102 | e12-102 | tier3 | 22 | 0 | 0.000 | 59 | 27 | 0.314 |
| 635308 | 5 | 60253633 | 60253633 | A | G | SNP | <i>NDUFAB2</i> | ENST00000296597 | 1 | intronic | c.127+12424 | e1+12424 | tier3 | 143 | 0 | 0.000 | 291 | 56 | 0.161 |
| 635308 | 5 | 73178364 | 73178364 | C | A | SNP | <i>ARHGFE28</i> | ENST00000545377 | 1 | missense | c.2882 | p.A961D | tier1 | 131 | 0 | 0.000 | 234 | 47 | 0.167 |
| 635308 | 5 | 137464931 | 137464931 | C | T | SNP | <i>NME5</i> | ENST00000265191 | -1 | intronic | c.335+21 | e2+21 | tier3 | 64 | 1 | 0.015 | 152 | 27 | 0.151 |
| 635308 | 5 | 171706385 | 171706385 | A | C | SNP | <i>UBTD2</i> | ENST00000393792 | -1 | intronic | c.70+4215 | e1+4215 | tier4 | 55 | 0 | 0.000 | 141 | 25 | 0.150 |
| 635308 | 6 | 41575854 | 41575854 | G | A | SNP | <i>MDFI</i> | ENST00000230321 | 1 | 5_prime_flanking_region | c.-30634 | NULL | tier3 | 69 | 0 | 0.000 | 171 | 32 | 0.156 |
| 635308 | 6 | 75890933 | 75890933 | T | A | SNP | <i>COL12A1</i> | ENST00000322507 | -1 | splice_region | c.1892-6 | e10-6 | tier3 | 146 | 3 | 0.020 | 262 | 74 | 0.220 |
| 635308 | 6 | 161565095 | 161565095 | C | T | SNP | <i>AGPAT4</i> | ENST00000437165 | -1 | 3_prime_untranslated_region | c.*66 | NULL | tier3 | 58 | 0 | 0.000 | 131 | 18 | 0.121 |
| 635308 | 7 | 70252341 | 70252341 | G | A | SNP | <i>AUTS2</i> | ENST00000342771 | 1 | missense | c.2455 | p.A819T | tier1 | 41 | 0 | 0.000 | 89 | 16 | 0.152 |
| 635308 | 7 | 102194790 | 102194790 | T | A | SNP | <i>SPDYE2</i> | ENST00000507918 | 1 | intronic | c.161-16 | e2-16 | tier3 | 170 | 1 | 0.006 | 454 | 50 | 0.099 |
| 635308 | 7 | 123152386 | 123152386 | A | G | SNP | <i>IQUB</i> | ENST00000324698 | -1 | silent | c.9 | p.N3 | tier1 | 35 | 0 | 0.000 | 76 | 17 | 0.183 |
| 635308 | 7 | 157629910 | 157629910 | T | C | SNP | <i>PTPRN2</i> | ENST00000404321 | -1 | intronic | c.1857+61455 | e12+61455 | tier4 | 103 | 5 | 0.046 | 366 | 96 | 0.207 |
| 635308 | 8 | 28203295 | 28203295 | C | T | SNP | <i>ZNF395</i> | ENST00000344423 | -1 | 3_prime_untranslated_region | c.*2941 | NULL | tier3 | 59 | 2 | 0.033 | 104 | 30 | 0.224 |
| 635308 | 9 | 124441020 | 124441020 | C | T | SNP | <i>DAB2IP</i> | ENST00000408936 | 1 | missense | c.188 | p.P63L | tier1 | 78 | 0 | 0.000 | 178 | 42 | 0.191 |
| 635308 | 9 | 124751442 | 124751442 | G | A | SNP | <i>TTLL11</i> | ENST00000373776 | -1 | missense | c.1571 | p.P524L | tier1 | 58 | 1 | 0.017 | 100 | 19 | 0.160 |
| 635308 | 11 | 47364238 | 47364238 | C | T | SNP | <i>MYBPC3</i> | ENST00000399249 | -1 | silent | c.1515 | p.K505 | tier1 | 52 | 0 | 0.000 | 80 | 14 | 0.149 |
| 635308 | 11 | 49813519 | 49813519 | G | T | SNP | <i>LOC440040</i> | ENST00000527477 | 1 | splice_region | c.NULL | NULL | tier4 | 22 | 0 | 0.000 | 55 | 25 | 0.305 |
| 635308 | 11 | 50379335 | 50379335 | G | A | SNP | <i>ENSG00000214883</i> | ENST00000532521 | 1 | 3_prime_untranslated_region | c.*50379335 | NULL | tier2 | 40 | 0 | 0.000 | 137 | 23 | 0.144 |
| 635308 | 11 | 56019747 | 56019747 | A | T | SNP | <i>OR5T3</i> | ENST00000303059 | 1 | silent | c.72 | p.S24 | tier1 | 93 | 2 | 0.021 | 112 | 45 | 0.283 |
| 635308 | 11 | 62327453 | 62327453 | T | A | SNP | <i>MIR3654</i> | ENST00000496634 | -1 | intronic | c.1578+88 | e9+88 | tier3 | 34 | 0 | 0.000 | 66 | 14 | 0.175 |
| 635308 | 11 | 124189425 | 124189425 | A | - | DEL | <i>OR8D2</i> | ENST00000357438 | -1 | frame_shift_del | c.669 | p.S224fs | tier1 | 40 | 0 | 0.000 | 68 | 22 | 0.244 |
| 635308 | 11 | 132306614 | 132306614 | G | A | SNP | <i>OPCML</i> | ENST00000541867 | -1 | silent | c.724 | p.L242 | tier1 | 65 | 0 | 0.000 | 86 | 14 | 0.139 |
| 635308 | 12 | 7363619 | 7363619 | T | A | SNP | <i>PEX5</i> | ENST00000266564 | 1 | 3_prime_untranslated_region | c.*800 | NULL | tier3 | 56 | 0 | 0.000 | 109 | 18 | 0.142 |
| 635308 | 12 | 21791412 | 21791412 | A | T | SNP | <i>LDHB</i> | ENST00000470985 | -1 | 5_prime_untranslated_region | c.-21791412 | NULL | tier3 | 138 | 0 | 0.000 | 306 | 20 | 0.061 |
| 635308 | 12 | 32854280 | 32854280 | A | G | SNP | <i>DNM1L</i> | ENST00000553257 | 1 | intronic | c.103-69 | e2-69 | tier3 | 72 | 0 | 0.000 | 156 | 19 | 0.108 |
| 635308 | 12 | 52966303 | 52966303 | C | T | SNP | <i>KRT74</i> | ENST00000305620 | -1 | missense | c.620 | p.G207E | tier1 | 48 | 0 | 0.000 | 91 | 17 | 0.157 |
| 635308 | 13 | 29600575 | 29600575 | G | T | SNP | <i>MTUS2</i> | ENST00000431530 | 1 | missense | c.1770 | p.L590F | tier1 | 594 | 5 | 0.008 | 1002 | 221 | 0.181 |
| 635308 | 14 | 33292181 | 33292181 | G | A | SNP | <i>AKAP6</i> | ENST00000280979 | 1 | missense | c.5162 | p.R1721H | tier1 | 58 | 0 | 0.000 | 101 | 13 | 0.114 |
| 635308 | 14 | 63453786 | 63453786 | T | A | SNP | <i>KCNH5</i> | ENST00000322893 | -1 | splice_region | c.549+4 | e5+4 | tier2 | 77 | 0 | 0.000 | 178 | 51 | 0.223 |
| 635308 | 14 | 70410184 | 70410184 | G | - | DEL | <i>SMOC1</i> | ENST00000361956 | -1 | intronic | c.100-8671 | e2-8671 | tier2 | 140 | 0 | 0.000 | 215 | 31 | 0.126 |
| 635308 | 15 | 102295698 | 102295700 | GCT | - | DEL | <i>DNM1P47</i> | ENST00000561463 | 1 | rna | NULL | NULL | tier1 | 27 | 0 | 0.000 | 32 | 10 | 0.238 |
| 635308 | 16 | 848710 | 848710 | G | A | SNP | <i>GNG13</i> | ENST00000248150 | -1 | 3_prime_untranslated_region | c.*9 | NULL | tier3 | 83 | 0 | 0.000 | 165 | 39 | 0.191 |
| 635308 | 16 | 85145834 | 85145834 | C | T | SNP | <i>FAM92B</i> | ENST00000539556 | -1 | intronic | c.20+105 | e1+105 | tier3 | 99 | 0 | 0.000 | 174 | 32 | 0.155 |
| 635308 | 17 | 78440948 | 78440948 | G | T | SNP | <i>NPTX1</i> | ENST00000535681 | -1 | 5_prime_untranslated_region | c.-78440948 | NULL | tier2 | 29 | 1 | 0.032 | 45 | 20 | 0.299 |
| 635308 | 17 | 79898724 | 79898724 | G | A | SNP | <i>MYADML2</i> | ENST00000330655 | -1 | silent | c.894 | p.S298 | tier1 | 103 | 0 | 0.000 | 182 | 32 | 0.149 |
| 635308 | 19 | 9088712 | 9088712 | C | G | SNP | <i>MUC16</i> | ENST00000397910 | -1 | missense | c.3103 | p.V1035L | tier1 | 544 | 8 | 0.015 | 1277 | 269 | 0.173 |
| 635308 | 19 | 49464172 | 49464172 | G | A | SNP | <i>BAX</i> | ENST00000345358 | -1 | splice_site | c.474+1 | e5+1 | tier1 | 53 | 0 | 0.000 | 112 | 30 | 0.208 |
| 635308 | 21 | 47918578 | 47918578 | G | C | SNP | <i>DIP2A</i> | ENST00000318711 | 1 | missense | c.487 | p.V163L | tier1 | 64 | 0 | 0.000 | 117 | 18 | 0.132 |
| 635308 | X | 57620926 | 57620926 | A | T | SNP | <i>ZXDB</i> | ENST00000374888 | 1 | 3_prime_untranslated_region | c.*33 | NULL | tier3 | 72 | 0 | 0.000 | 146 | 24 | 0.141 |
| 635308 | X | 153283630 | 153283630 | G | A | SNP | <i>IRAK1</i> | ENST00000369980 | -1 | intronic | c.795-59 | e7-59 | tier3 | 82 | 0 | 0.000 | 147 | 28 | 0.160 |
| 639691 | 1 | 45296629 | 45296629 | C | T | SNP | <i>PTCH2</i> | ENST00000372192 | -1 | missense | c.704 | p.R235Q | tier1 | 41 | 0 | 0.000 | 57 | 13 | 0.181 |
| 639691 | 1 | 118644343 | 118644343 | G | A | SNP | <i>SPAG17</i> | ENST00000336338 | -1 | intronic | c.634+20 | e5+20 | tier3 | 62 | 0 | 0.000 | 95 | 25 | 0.207 |
| 639691 | 1 | 144857704 | 144857704 | C | A | SNP | <i>PDE4DIP</i> | ENST00000369356 | -1 | missense | c.6350 | p.R2117L | tier1 | 409 | 1 | 0.002 | 601 | 20 | 0.032 |
| 639691 | 1 | 152276933 | 152276933 | G | T | SNP | <i>FLG</i> | ENST00000368799 | -1 | missense | c.10429 | p.R3477S | tier1 | 716 | 1 | 0.001 | 1022 | 24 | 0.023 |
| 639691 | 1 | 152281796 | 152281796 | G | T | SNP | <i>FLG</i> | ENST00000368799 | -1 | missense | c.5566 | p.R1856S | tier1 | 790 | 0 | 0.000 | 1023 | 23 | 0.022 |
| 639691 | 1 | 160105026 | 160105026 | G | A | SNP | <i>ATP1A2</i> | ENST00000361216 | 1 | missense | c.2056 | p.V686I | tier1 | 29 | 0 | 0.000 | 26 | 14 | 0.350 |
| 639691 | 1 | 205389686 | 205389686 | T | C | SNP | <i>LEMD1</i> | ENST00000476884 | -1 | rna | NULL | NULL | tier1 | 53 | 0 | 0.000 | 98 | 15 | 0.133 |
| 639691 | 2 | 9558907 | 9558907 | T | G | SNP | <i>ITGB1BP1</i> | ENST00000355346 | -1 | intronic | c.1-46 | e1-46 | tier4 | 37 | 0 | 0.000 | 50 | 17 | 0.254 |
| 639691 | 2 | 9558909 | 9558909 | T | A | SNP | <i>ITGB1BP1</i> | ENST00000355346 | -1 | intronic | c.1-48 | e1-48 | tier4 | 37 | 0 | 0.000 | 45 | 14 | 0.237 |
| 639691 | 2 | 21238368 | 21238368 | G | T | SNP | <i>APOB</i> | ENST00000233242 | -1 | missense | c.3382 | p.R1128S | tier1 | 372 | 0 | 0.000 | 594 | 19 | 0.031 |
| 639691 | 2 | 71762298 | 71762298 | C | A | SNP | <i>DYSF</i> | ENST00000413539 | 1 | intronic | c.1446+79 | e15+79 | tier3 | 255 | 0 | 0.000 | 297 | 15 | 0.048 |
| 639691 | 2 | 89160377 | 89160377 | G | C | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 47 | 0 | 0.000 | 18 | 30 | 0.625 |
| 639691 | 2 | 89160420 | 89160420 | T | C | SNP | <i>IGKJ4</i> | ENST00000390239 | -1 | silent | c.15 | p.G5 | tier1 | 68 | 0 | 0.000 | 44 | 12 | 0.211 |
| 639691 | 2 | 89160450 | 89160450 | T | G | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 72 | 0 | 0.000 | 45 | 10 | 0.182 |
| 639691 | 2 | 89160485 | 89160485 | A | G | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 66 | 0 | 0.000 | 45 | 7 | 0.135 |
| 639691 | 2 | 89160500 | 89160500 | C | G | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 67 | 0 | 0.000 | 48 | 7 | 0.127 |
| 639691 | 2 | 89160507 | 89160507 | T | G | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 65 | 0 | 0.000 | 47 | 7 | 0.130 |
| 639691 | 2 | 89160663 | 89160663 | T | C | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 78 | 0 | 0.000 | 41 | 31 | 0.431 |
| 639691 | 2 | 89160686 | 89160686 | T | C | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 101 | 0 | 0.000 | 66 | 41 | 0.380 |
| 639691 | 2 | 89160714 | 89160714 | T | C | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 123 | 0 | 0.000 | 82 | 53 | 0.393 |
| 639691 | 2 | 89160740 | 89160740 | A | C | SNP | <i>IGKJ3</i> | ENST00000390240 | -1 | missense | c.31 | p.Y11D | tier1 | 168 | 1 | 0.006 | 119 | 73 | 0.380 |
| 639691 | 2 | 89160789 | 89160789 | C | T | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 222 | 0 | 0.000 | 161 | 62 | 0.277 |
| 639691 | 2 | 89160801 | 89160801 | C | T | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 218 | 1 | 0.005 | 164 | 60 | 0.268 |
| 639691 | 2 | 89160806 | 89160806 | A | G | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 220 | 0 | 0.000 | 159 | 59 | 0.269 |
| 639691 | 2 | 89160841 | 89160841 | G | G | SNP | <i>ENSG00000231486</i> | ENST00000418209 | -1 | intronic | c.NULL | NULL | tier3 | 222 | 0 | 0.000 | 153 | 43 | 0.217 |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|------|---|-----|------------------------|-----------------|----|-----------------------------|-----------------------|----------|-------|-----|---|-------|------|-----|-------|
| 639691 | 2 | 89185292 | 89185292 | T | C | SNP | <i>IGKV4-1</i> | ENST00000390243 | 1 | intronic | c.50-64 | e2-64 | tier3 | 75 | 0 | 0.000 | 100 | 11 | 0.099 |
| 639691 | 2 | 89185531 | 89185531 | C | G | SNP | <i>IGKV4-1</i> | ENST00000390243 | 1 | nonsense | c.225 | p.Y75* | tier1 | 180 | 0 | 0.000 | 81 | 9 | 0.100 |
| 639691 | 2 | 89185535 | 89185535 | G | A | SNP | <i>IGKV4-1</i> | ENST00000390243 | 1 | missense | c.229 | p.A77T | tier1 | 178 | 0 | 0.000 | 78 | 8 | 0.093 |
| 639691 | 2 | 89185568 | 89185568 | T | G | SNP | <i>IGKV4-1</i> | ENST00000390243 | 1 | missense | c.262 | p.F88V | tier1 | 189 | 0 | 0.000 | 68 | 9 | 0.117 |
| 639691 | 2 | 89246846 | 89246846 | A | G | SNP | <i>IGKV1-5</i> | ENST00000496168 | -1 | silent | c.324 | p.Y108 | tier1 | 160 | 0 | 0.000 | 137 | 11 | 0.074 |
| 639691 | 2 | 89246861 | 89246861 | A | T | SNP | <i>IGKV1-5</i> | ENST00000496168 | -1 | missense | c.309 | p.D103E | tier1 | 165 | 0 | 0.000 | 153 | 15 | 0.089 |
| 639691 | 2 | 89246918 | 89246918 | G | A | SNP | <i>IGKV1-5</i> | ENST00000496168 | -1 | silent | c.252 | p.F84 | tier1 | 178 | 0 | 0.000 | 176 | 25 | 0.123 |
| 639691 | 2 | 89246947 | 89246947 | T | G | SNP | <i>IGKV1-5</i> | ENST00000496168 | -1 | missense | c.223 | p.S75R | tier1 | 178 | 1 | 0.006 | 182 | 26 | 0.125 |
| 639691 | 2 | 89246969 | 89246969 | C | G | SNP | <i>IGKV1-5</i> | ENST00000496168 | -1 | missense | c.201 | p.K67N | tier1 | 171 | 0 | 0.000 | 159 | 22 | 0.121 |
| 639691 | 2 | 89246970 | 89246970 | T | C | SNP | <i>IGKV1-5</i> | ENST00000496168 | -1 | missense | c.200 | p.K67R | tier1 | 170 | 0 | 0.000 | 157 | 23 | 0.128 |
| 639691 | 2 | 89247181 | 89247181 | T | G | SNP | <i>IGKV1-5</i> | ENST00000496168 | -1 | intronic | c.55+59 | e1+59 | tier3 | 167 | 0 | 0.000 | 168 | 66 | 0.280 |
| 639691 | 2 | 89247195 | 89247195 | T | C | SNP | <i>IGKV1-5</i> | ENST00000496168 | -1 | intronic | c.55+45 | e1+45 | tier3 | 180 | 0 | 0.000 | 177 | 64 | 0.265 |
| 639691 | 2 | 89247280 | 89247280 | G | T | SNP | <i>IGKV1-5</i> | ENST00000496168 | -1 | silent | c.15 | p.V5 | tier1 | 187 | 0 | 0.000 | 172 | 66 | 0.276 |
| 639691 | 2 | 114355305 | 114355305 | C | A | SNP | <i>WASH2P</i> | ENST00000538033 | 1 | intronic | c.NULL | NULL | tier3 | 125 | 0 | 0.000 | 129 | 10 | 0.071 |
| 639691 | 2 | 141356343 | 141356343 | C | A | SNP | <i>LRP1B</i> | ENST00000389484 | -1 | missense | c.7051 | p.G2351W | tier1 | 452 | 0 | 0.000 | 954 | 26 | 0.027 |
| 639691 | 2 | 202436665 | 202436665 | T | G | SNP | <i>ALS2CR11</i> | ENST00000286195 | -1 | missense | c.832 | p.T278P | tier1 | 54 | 1 | 0.018 | 91 | 29 | 0.242 |
| 639691 | 3 | 15176119 | 15176119 | G | T | SNP | <i>ZFYVE20</i> | ENST00000253699 | -1 | 5_prime_flanking_region | c.-38492 | NULL | tier3 | 368 | 0 | 0.000 | 597 | 19 | 0.031 |
| 639691 | 3 | 38592896 | 38592896 | G | A | SNP | <i>SCN5A</i> | ENST00000333535 | -1 | missense | c.4967 | p.A1656V | tier1 | 142 | 1 | 0.007 | 241 | 62 | 0.205 |
| 639691 | 3 | 71247370 | 71247370 | G | T | SNP | <i>FOXP1</i> | ENST00000318789 | -1 | missense | c.163 | p.Q55K | tier1 | 589 | 0 | 0.000 | 774 | 22 | 0.028 |
| 639691 | 3 | 193007798 | 193007798 | G | C | SNP | <i>ATP13A5</i> | ENST00000342358 | -1 | missense | c.2899 | p.Q967E | tier1 | 66 | 0 | 0.000 | 103 | 16 | 0.135 |
| 639691 | 3 | 193721356 | 193721356 | C | A | SNP | <i>LOC647323</i> | ENST00000397645 | -1 | intronic | c.NULL | NULL | tier2 | 100 | 0 | 0.000 | 104 | 10 | 0.088 |
| 639691 | 4 | 55594015 | 55594015 | G | T | SNP | <i>KIT</i> | ENST00000288135 | 1 | missense | c.1801 | p.G601W | tier1 | 338 | 0 | 0.000 | 610 | 20 | 0.032 |
| 639691 | 4 | 100273911 | 100273911 | G | T | SNP | <i>ADH1C</i> | ENST00000515683 | -1 | 5_prime_untranslated_region | c.-79 | NULL | tier3 | 52 | 0 | 0.000 | 81 | 22 | 0.214 |
| 639691 | 4 | 106196822 | 106196822 | G | T | SNP | <i>TET2</i> | ENST00000380013 | 1 | missense | c.5155 | p.G1719W | tier1 | 496 | 1 | 0.002 | 884 | 24 | 0.026 |
| 639691 | 4 | 114278785 | 114278785 | G | T | SNP | <i>ANK2</i> | ENST00000357077 | 1 | missense | c.9011 | p.W3004L | tier1 | 457 | 0 | 0.000 | 793 | 22 | 0.027 |
| 639691 | 4 | 189013998 | 189013998 | T | C | SNP | <i>TRIML2</i> | ENST00000503141 | -1 | 3_prime_untranslated_region | c.*4297 | NULL | tier4 | 72 | 0 | 0.000 | 96 | 33 | 0.256 |
| 639691 | 5 | 38968092 | 38968092 | G | A | SNP | <i>RICTOR</i> | ENST00000296782 | -1 | missense | c.1013 | p.P338L | tier1 | 49 | 0 | 0.000 | 133 | 29 | 0.178 |
| 639691 | 5 | 96248518 | 96248512 | TAAC | - | DEL | <i>ERAP2</i> | ENST00000437043 | 1 | intronic | c.2516+16_2516+13 | e15+16 | tier3 | 49 | 0 | 0.000 | 105 | 21 | 0.167 |
| 639691 | 5 | 140604754 | 140604754 | C | A | SNP | <i>PCDHB14</i> | ENST00000239449 | 1 | silent | c.1677 | p.P559 | tier1 | 170 | 0 | 0.000 | 192 | 11 | 0.054 |
| 639691 | 5 | 149808594 | 149808594 | G | T | SNP | <i>CD74</i> | ENST00000009530 | -1 | 5_prime_flanking_region | c.-16282 | NULL | tier4 | 127 | 0 | 0.000 | 101 | 8 | 0.073 |
| 639691 | 6 | 325891 | 325891 | G | T | SNP | <i>DUSP22</i> | ENST00000604971 | 1 | 5_prime_untranslated_region | c.-22258 | NULL | tier3 | 264 | 0 | 0.000 | 245 | 13 | 0.049 |
| 639691 | 6 | 15501389 | 15501389 | C | T | SNP | <i>JARID2</i> | ENST00000341776 | 1 | missense | c.2197 | p.R733C | tier1 | 57 | 0 | 0.000 | 69 | 16 | 0.188 |
| 639691 | 6 | 18139973 | 18139974 | AT | - | DEL | <i>TPMT</i> | ENST00000309983 | -1 | intronic | c.367-26_367-25 | e4-25 | tier4 | 52 | 0 | 0.000 | 94 | 18 | 0.161 |
| 639691 | 6 | 52701179 | 52701179 | A | - | DEL | <i>GSTA5</i> | ENST00000284562 | -1 | intronic | c.140-13 | e3-13 | tier3 | 65 | 0 | 0.000 | 87 | 35 | 0.287 |
| 639691 | 6 | 76343330 | 76343330 | T | C | SNP | <i>SENP6</i> | ENST00000370014 | 1 | missense | c.241 | p.S81P | tier1 | 42 | 0 | 0.000 | 144 | 26 | 0.152 |
| 639691 | 6 | 119285878 | 119285878 | A | C | SNP | <i>FAM184A</i> | ENST00000338891 | -1 | missense | c.3092 | p.V1031G | tier1 | 38 | 0 | 0.000 | 135 | 57 | 0.297 |
| 639691 | 6 | 127912679 | 127912679 | C | - | DEL | <i>C6orf58</i> | ENST00000329722 | 1 | splice_region_del | c.914-9 | e6-9 | tier3 | 57 | 0 | 0.000 | 292 | 42 | 0.126 |
| 639691 | 7 | 2515372 | 2515372 | G | T | SNP | <i>GRIFIN</i> | ENST00000417742 | -1 | rna | NULL | NULL | tier1 | 169 | 0 | 0.000 | 171 | 10 | 0.055 |
| 639691 | 7 | 43484053 | 43484053 | C | A | SNP | <i>HECW1</i> | ENST00000395891 | 1 | missense | c.1282 | p.Q428K | tier1 | 379 | 3 | 0.008 | 392 | 157 | 0.286 |
| 639691 | 7 | 99795840 | 99795840 | G | T | SNP | <i>STAG3</i> | ENST00000317296 | 1 | intronic | c.1244+51 | e11+51 | tier3 | 268 | 0 | 0.000 | 311 | 14 | 0.043 |
| 639691 | 7 | 142482221 | 142482221 | G | T | SNP | <i>PRSS3P2</i> | ENST00000603901 | 1 | rna | NULL | NULL | tier1 | 58 | 0 | 0.000 | 24 | 6 | 0.200 |
| 639691 | 8 | 26628125 | 26628125 | G | A | SNP | <i>ADRA1A</i> | ENST00000380586 | -1 | silent | c.942 | p.L314 | tier1 | 48 | 0 | 0.000 | 88 | 29 | 0.248 |
| 639691 | 8 | 40012484 | 40012484 | A | C | SNP | <i>C8orf4</i> | ENST00000315792 | 1 | 3_prime_untranslated_region | c.*1112 | NULL | tier2 | 15 | 0 | 0.000 | 81 | 40 | 0.331 |
| 639691 | 8 | 131073071 | 131073071 | G | T | SNP | <i>ASAP1</i> | ENST00000524124 | -1 | missense | c.2407 | p.Q803K | tier1 | 94 | 0 | 0.000 | 79 | 8 | 0.092 |
| 639691 | 8 | 142506437 | 142506437 | C | A | SNP | <i>MROH5</i> | ENST00000521161 | -1 | missense | c.140 | p.G47V | tier1 | 384 | 0 | 0.000 | 464 | 15 | 0.031 |
| 639691 | 8 | 145745801 | 145745801 | A | C | SNP | <i>LRRC14</i> | ENST00000292524 | 1 | missense | c.509 | p.D170A | tier1 | 72 | 1 | 0.014 | 75 | 21 | 0.219 |
| 639691 | 9 | 13217161 | 13217161 | C | T | SNP | <i>MPDZ</i> | ENST00000319217 | -1 | intronic | c.1201+18 | e8+18 | tier3 | 42 | 0 | 0.000 | 164 | 55 | 0.251 |
| 639691 | 9 | 119730785 | 119730787 | GAG | - | DEL | <i>ASTN2</i> | ENST00000313400 | -1 | intronic | c.1889+6700_1889+6698 | e10+6700 | tier3 | 37 | 0 | 0.000 | 50 | 12 | 0.194 |
| 639691 | 9 | 140286997 | 140286997 | G | T | SNP | <i>EXD3</i> | ENST00000465160 | -1 | 3_prime_untranslated_region | c.*575 | NULL | tier3 | 110 | 0 | 0.000 | 136 | 11 | 0.075 |
| 639691 | 9 | 141013150 | 141013150 | G | T | SNP | <i>CACNA1B</i> | ENST00000371355 | 1 | missense | c.5963 | p.G1988V | tier1 | 199 | 0 | 0.000 | 219 | 12 | 0.052 |
| 639691 | 10 | 124458903 | 124458903 | C | A | SNP | <i>C10orf120</i> | ENST00000329446 | -1 | missense | c.202 | p.D68Y | tier1 | 164 | 0 | 0.000 | 177 | 69 | 0.281 |
| 639691 | 11 | 1267352 | 1267352 | C | A | SNP | <i>MUC5B</i> | ENST00000447027 | 1 | missense | c.9251 | p.P3084Q | tier1 | 480 | 0 | 0.000 | 573 | 18 | 0.030 |
| 639691 | 11 | 1271142 | 1271142 | C | A | SNP | <i>MUC5B</i> | ENST00000447027 | 1 | silent | c.13041 | p.P4347 | tier1 | 577 | 1 | 0.002 | 580 | 21 | 0.035 |
| 639691 | 11 | 1271180 | 1271180 | C | A | SNP | <i>MUC5B</i> | ENST00000447027 | 1 | missense | c.13079 | p.P4360Q | tier1 | 681 | 0 | 0.000 | 713 | 19 | 0.026 |
| 639691 | 11 | 6235593 | 6235593 | T | C | SNP | <i>FAM160A2</i> | ENST00000265978 | -1 | intronic | c.2599+48 | e10+48 | tier3 | 57 | 0 | 0.000 | 90 | 13 | 0.126 |
| 639691 | 11 | 64794980 | 64794980 | C | T | SNP | <i>SNX15</i> | ENST00000377244 | 1 | 5_prime_untranslated_region | c.-30 | NULL | tier2 | 67 | 2 | 0.029 | 60 | 26 | 0.302 |
| 639691 | 11 | 118918816 | 118918816 | A | - | DEL | <i>HYOU1</i> | ENST00000527738 | -1 | 5_prime_untranslated_region | c.-118918816 | NULL | tier3 | 41 | 0 | 0.000 | 52 | 21 | 0.288 |
| 639691 | 12 | 4920436 | 4920436 | C | A | SNP | <i>KCNA6</i> | ENST00000280684 | 1 | missense | c.1229 | p.P410Q | tier1 | 91 | 0 | 0.000 | 72 | 8 | 0.100 |
| 639691 | 12 | 70349145 | 70349145 | C | G | SNP | <i>MYRFL</i> | ENST00000299350 | 1 | missense | c.473 | p.S158C | tier1 | 62 | 0 | 0.000 | 192 | 63 | 0.247 |
| 639691 | 12 | 93477504 | 93477504 | C | A | SNP | <i>ENSG00000257322</i> | ENST00000549930 | -1 | intronic | c.NULL | NULL | tier4 | 39 | 0 | 0.000 | 63 | 23 | 0.267 |
| 639691 | 12 | 125292397 | 125292397 | G | T | SNP | <i>SCARB1</i> | ENST00000415380 | -1 | missense | c.919 | p.L307M | tier1 | 376 | 0 | 0.000 | 553 | 20 | 0.035 |
| 639691 | 13 | 31002525 | 31002525 | G | A | SNP | <i>UBE2L5P</i> | ENST00000436446 | 1 | 3_prime_untranslated_region | c.*31002525 | NULL | tier2 | 94 | 0 | 0.000 | 151 | 31 | 0.170 |
| 639691 | 13 | 114530002 | 114530002 | C | A | SNP | <i>GAS6</i> | ENST00000357389 | -1 | missense | c.1573 | p.G525W | tier1 | 279 | 0 | 0.000 | 292 | 12 | 0.039 |
| 639691 | 14 | 20264803 | 20264803 | G | A | SNP | <i>OR4N1P</i> | ENST00000554847 | 1 | 3_prime_untranslated_region | c.*20264803 | NULL | tier3 | 131 | 0 | 0.000 | 188 | 48 | 0.203 |
| 639691 | 14 | 77844469 | 77844469 | C | A | SNP | <i>SAMD15</i> | ENST00000216471 | 1 | silent | c.708 | p.T236 | tier1 | 26 | 0 | 0.000 | 24 | 8 | 0.250 |
| 639691 | 14 | 106329350 | 106329350 | C | G | SNP | <i>ENSG00000244620</i> | ENST00000414005 | 1 | 5_prime_flanking_region | c.-26592 | NULL | tier2 | 31 | 0 | 0.000 | 14 | 8 | 0.348 |
| 639691 | 14 | 106329945 | 106329945 | C | T | SNP | <i>ENSG00000244620</i> | ENST00000414005 | 1 | 5_prime_flanking_region | c.-25997 | NULL | tier3 | 84 | 1 | 0.012 | 59</ | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|---|---|-----|------------------------|-----------------|----|-----------------------------|-------------|----------|-------|------|---|-------|------|-----|-------|
| 639691 | 14 | 107034956 | 107034956 | T | C | SNP | <i>IGHV5-51</i> | ENST00000390626 | -1 | missense | c.124 | p.K42E | tier1 | 109 | 0 | 0.000 | 96 | 23 | 0.193 |
| 639691 | 14 | 107035010 | 107035010 | C | T | SNP | <i>IGHV5-51</i> | ENST00000390626 | -1 | missense | c.70 | p.V24M | tier1 | 102 | 0 | 0.000 | 102 | 16 | 0.136 |
| 639691 | 14 | 107035027 | 107035027 | C | G | SNP | <i>IGHV5-51</i> | ENST00000390626 | -1 | missense | c.53 | p.C18S | tier1 | 110 | 0 | 0.000 | 103 | 19 | 0.155 |
| 639691 | 14 | 107035049 | 107035049 | A | G | SNP | <i>IGHV5-51</i> | ENST00000390626 | -1 | intronic | c.47-16 | e2-16 | tier3 | 113 | 0 | 0.000 | 83 | 16 | 0.162 |
| 639691 | 14 | 107035075 | 107035075 | A | G | SNP | <i>IGHV5-51</i> | ENST00000390626 | -1 | intronic | c.46+42 | e1+42 | tier3 | 108 | 0 | 0.000 | 85 | 16 | 0.158 |
| 639691 | 14 | 107035146 | 107035146 | A | G | SNP | <i>IGHV5-51</i> | ENST00000390626 | -1 | missense | c.17 | p.I6T | tier1 | 74 | 0 | 0.000 | 65 | 13 | 0.167 |
| 639691 | 14 | 107178814 | 107178814 | C | G | SNP | <i>IGHV1-69</i> | ENST00000390633 | -1 | 5_prime_flanking_region | c.-8447 | NULL | tier3 | 76 | 0 | 0.000 | 68 | 15 | 0.181 |
| 639691 | 14 | 107178822 | 107178822 | T | C | SNP | <i>IGHV2-70</i> | ENST00000390634 | -1 | missense | c.355 | p.I119V | tier1 | 79 | 0 | 0.000 | 77 | 14 | 0.154 |
| 639691 | 14 | 107178861 | 107178861 | T | C | SNP | <i>IGHV2-70</i> | ENST00000390634 | -1 | missense | c.316 | p.M106V | tier1 | 102 | 0 | 0.000 | 139 | 15 | 0.097 |
| 639691 | 14 | 107178871 | 107178871 | T | C | SNP | <i>IGHV2-70</i> | ENST00000390634 | -1 | silent | c.306 | p.T102 | tier1 | 112 | 0 | 0.000 | 138 | 28 | 0.169 |
| 639691 | 14 | 107178884 | 107178884 | T | A | SNP | <i>IGHV2-70</i> | ENST00000390634 | -1 | missense | c.293 | p.Q98L | tier1 | 120 | 0 | 0.000 | 172 | 17 | 0.090 |
| 639691 | 14 | 107178935 | 107178935 | C | A | SNP | <i>IGHV2-70</i> | ENST00000390634 | -1 | missense | c.242 | p.S81I | tier1 | 141 | 0 | 0.000 | 208 | 30 | 0.126 |
| 639691 | 14 | 107179009 | 107179009 | G | A | SNP | <i>IGHV2-70</i> | ENST00000390634 | -1 | silent | c.168 | p.S56 | tier1 | 172 | 0 | 0.000 | 203 | 47 | 0.188 |
| 639691 | 14 | 107179158 | 107179158 | C | T | SNP | <i>IGHV2-70</i> | ENST00000390634 | -1 | intronic | c.47-28 | e2-28 | tier3 | 186 | 0 | 0.000 | 256 | 47 | 0.155 |
| 639691 | 15 | 33923426 | 33923426 | G | T | SNP | <i>RYR3</i> | ENST00000389232 | 1 | silent | c.2799 | p.L933 | tier1 | 382 | 0 | 0.000 | 671 | 20 | 0.029 |
| 639691 | 16 | 1261371 | 1261371 | G | A | SNP | <i>CACNA1H</i> | ENST00000569953 | 1 | 3_prime_untranslated_region | c.*1261371 | NULL | tier3 | 75 | 1 | 0.013 | 61 | 12 | 0.164 |
| 639691 | 16 | 56652567 | 56652567 | C | T | SNP | <i>MT1L</i> | ENST00000565768 | 1 | rna | NULL | NULL | tier1 | 80 | 0 | 0.000 | 79 | 17 | 0.177 |
| 639691 | 16 | 67580350 | 67580350 | C | T | SNP | <i>FAM65A</i> | ENST00000422602 | 1 | missense | c.3704 | p.A1235V | tier1 | 252 | 1 | 0.004 | 330 | 44 | 0.118 |
| 639691 | 17 | 7386234 | 7386234 | G | A | SNP | <i>SLC35G6</i> | ENST00000412468 | 1 | missense | c.931 | p.V311M | tier1 | 115 | 3 | 0.025 | 128 | 26 | 0.169 |
| 639691 | 17 | 7579349 | 7579349 | A | C | SNP | <i>TP53</i> | ENST00000269305 | -1 | missense | c.338 | p.F113C | tier1 | 784 | 1 | 0.001 | 774 | 213 | 0.216 |
| 639691 | 17 | 56277172 | 56277172 | G | A | SNP | <i>EPX</i> | ENST00000225371 | 1 | intronic | c.1537+17 | e9+17 | tier3 | 106 | 1 | 0.009 | 97 | 45 | 0.317 |
| 639691 | 17 | 61972742 | 61972742 | T | G | SNP | <i>CSH1</i> | ENST00000329882 | -1 | missense | c.547 | p.K183Q | tier1 | 116 | 5 | 0.041 | 109 | 41 | 0.273 |
| 639691 | 17 | 79206209 | 79206209 | C | A | SNP | <i>ENTHD2</i> | ENST00000374769 | -1 | silent | c.180 | p.R60 | tier1 | 181 | 0 | 0.000 | 172 | 10 | 0.054 |
| 639691 | 18 | 14810028 | 14810028 | A | G | SNP | <i>ANKRD30B</i> | ENST00000358984 | 1 | intronic | c.2313+1450 | e25+1450 | tier3 | 107 | 0 | 0.000 | 357 | 31 | 0.080 |
| 639691 | 18 | 20979505 | 20979505 | T | C | SNP | <i>TMEM241</i> | ENST00000383233 | -1 | intronic | c.279+25 | e4+25 | tier3 | 49 | 0 | 0.000 | 89 | 33 | 0.271 |
| 639691 | 19 | 11564674 | 11564674 | G | T | SNP | <i>ELAVL3</i> | ENST00000359227 | -1 | 3_prime_untranslated_region | c.*667 | NULL | tier4 | 41 | 1 | 0.023 | 57 | 22 | 0.279 |
| 639691 | 19 | 49548481 | 49548481 | C | T | SNP | <i>CGB5</i> | ENST00000301408 | 1 | missense | c.428 | p.A143V | tier1 | 69 | 1 | 0.014 | 74 | 21 | 0.221 |
| 639691 | 19 | 54742838 | 54742838 | G | A | SNP | <i>LILRA6</i> | ENST00000245621 | -1 | silent | c.1386 | p.A462 | tier1 | 187 | 0 | 0.000 | 276 | 17 | 0.058 |
| 639691 | 21 | 38885295 | 38885295 | G | T | SNP | <i>DYRK1A</i> | ENST00000339659 | 1 | 3_prime_untranslated_region | c.*461 | NULL | tier3 | 161 | 0 | 0.000 | 226 | 21 | 0.084 |
| 639691 | 22 | 23223246 | 23223246 | A | T | SNP | <i>IGLV3-1</i> | ENST00000390319 | 1 | intronic | c.47-31 | e2-31 | tier3 | 32 | 0 | 0.000 | 35 | 22 | 0.386 |
| 639691 | 22 | 23223346 | 23223346 | T | C | SNP | <i>IGLV3-1</i> | ENST00000390319 | 1 | missense | c.116 | p.I39T | tier1 | 53 | 0 | 0.000 | 49 | 8 | 0.140 |
| 639691 | 22 | 23223372 | 23223372 | G | A | SNP | <i>IGLV3-1</i> | ENST00000390319 | 1 | missense | c.142 | p.D48N | tier1 | 50 | 0 | 0.000 | 46 | 11 | 0.193 |
| 639691 | 22 | 23223397 | 23223397 | A | C | SNP | <i>IGLV3-1</i> | ENST00000390319 | 1 | missense | c.167 | p.Q56P | tier1 | 58 | 0 | 0.000 | 58 | 11 | 0.159 |
| 639691 | 22 | 23223436 | 23223436 | A | G | SNP | <i>IGLV3-1</i> | ENST00000390319 | 1 | missense | c.206 | p.D69G | tier1 | 56 | 0 | 0.000 | 74 | 14 | 0.159 |
| 639691 | 22 | 23223446 | 23223446 | G | C | SNP | <i>IGLV3-1</i> | ENST00000390319 | 1 | silent | c.216 | p.R72 | tier1 | 57 | 0 | 0.000 | 74 | 16 | 0.178 |
| 639691 | 22 | 23223480 | 23223480 | A | C | SNP | <i>IGLV3-1</i> | ENST00000390319 | 1 | missense | c.250 | p.N84H | tier1 | 53 | 0 | 0.000 | 67 | 21 | 0.236 |
| 639691 | 22 | 23660791 | 23660791 | C | T | SNP | <i>ENSG00000236611</i> | ENST00000450776 | 1 | 5_prime_flanking_region | c.-8012 | NULL | tier3 | 93 | 0 | 0.000 | 133 | 13 | 0.089 |
| 639691 | X | 15841049 | 15841049 | G | T | SNP | <i>ZRSR2</i> | ENST00000307771 | 1 | missense | c.1133 | p.R378L | tier1 | 493 | 0 | 0.000 | 758 | 22 | 0.028 |
| 639691 | X | 18323300 | 18323300 | C | A | SNP | <i>SCML2</i> | ENST00000251900 | -1 | silent | c.522 | p.V174 | tier1 | 383 | 0 | 0.000 | 732 | 23 | 0.030 |
| 639691 | X | 48163116 | 48163116 | G | T | SNP | <i>SSX9</i> | ENST00000608568 | -1 | 5_prime_untranslated_region | c.-48163116 | NULL | tier3 | 216 | 0 | 0.000 | 346 | 17 | 0.046 |
| 639691 | X | 62857793 | 62857793 | G | T | SNP | <i>ARHGEF9</i> | ENST00000253401 | -1 | 3_prime_untranslated_region | c.*115 | NULL | tier4 | 318 | 4 | 0.012 | 500 | 37 | 0.069 |
| 639691 | X | 69455923 | 69455923 | C | T | SNP | <i>AWA1</i> | ENST00000374521 | 1 | silent | c.189 | p.G63 | tier1 | 95 | 0 | 0.000 | 116 | 12 | 0.094 |
| 639691 | X | 101972409 | 101972409 | T | C | SNP | <i>GPRASP2</i> | ENST00000332262 | 1 | 3_prime_untranslated_region | c.*95 | NULL | tier3 | 72 | 1 | 0.014 | 156 | 67 | 0.300 |
| 639691 | X | 147031603 | 147031603 | T | - | DEL | <i>FMR1</i> | ENST00000370475 | 1 | 3_prime_untranslated_region | c.*1239 | NULL | tier3 | 79 | 0 | 0.000 | 294 | 30 | 0.093 |
| 639691 | X | 153134255 | 153134255 | C | G | SNP | <i>L1CAM</i> | ENST00000543994 | -1 | intronic | c.1385+41 | e11+41 | tier3 | 64 | 0 | 0.000 | 76 | 21 | 0.217 |
| 678913 | 1 | 155884156 | 155884156 | A | - | DEL | <i>KIAA0907</i> | ENST00000368320 | -1 | 3_prime_untranslated_region | c.*1458 | NULL | tier3 | 56 | 0 | 0.000 | 74 | 14 | 0.159 |
| 678913 | 2 | 96557526 | 96557526 | T | A | SNP | <i>ANKRD36C</i> | ENST00000456556 | -1 | intronic | c.2785-41 | e46-41 | tier3 | 301 | 3 | 0.010 | 592 | 33 | 0.053 |
| 678913 | 3 | 78700929 | 78700929 | C | A | SNP | <i>ROBO1</i> | ENST00000464233 | -1 | missense | c.2765 | p.R922L | tier1 | 130 | 2 | 0.015 | 160 | 82 | 0.337 |
| 678913 | 3 | 78700930 | 78700930 | G | T | SNP | <i>ROBO1</i> | ENST00000464233 | -1 | missense | c.2764 | p.R922S | tier1 | 131 | 2 | 0.015 | 160 | 84 | 0.343 |
| 678913 | 5 | 150407642 | 150407642 | T | G | SNP | <i>GPX3</i> | ENST00000388825 | 1 | missense | c.632 | p.I211S | tier1 | 85 | 1 | 0.012 | 121 | 66 | 0.353 |
| 678913 | 6 | 33261595 | 33261595 | G | A | SNP | <i>RGL2</i> | ENST00000497454 | -1 | silent | c.1485 | p.L495 | tier1 | 131 | 2 | 0.015 | 112 | 79 | 0.414 |
| 678913 | 7 | 116166678 | 116166678 | G | T | SNP | <i>CAV1</i> | ENST00000341049 | 1 | missense | c.130 | p.A44S | tier1 | 70 | 2 | 0.028 | 91 | 66 | 0.418 |
| 678913 | 7 | 141321598 | 141321598 | C | A | SNP | <i>AGK</i> | ENST00000355413 | 1 | silent | c.585 | p.I195 | tier1 | 90 | 2 | 0.022 | 132 | 75 | 0.359 |
| 678913 | 7 | 151945083 | 151945083 | C | T | SNP | <i>KMT2C</i> | ENST00000355193 | -1 | missense | c.2436 | p.M812I | tier1 | 2504 | 5 | 0.002 | 5543 | 178 | 0.031 |
| 678913 | 12 | 2912387 | 2912387 | C | T | SNP | <i>FKBP4</i> | ENST0000001008 | 1 | missense | c.1343 | p.T448M | tier1 | 57 | 0 | 0.000 | 60 | 38 | 0.388 |
| 678913 | 12 | 39761659 | 39761659 | T | C | SNP | <i>KIF21A</i> | ENST00000395670 | -1 | intronic | c.600+26 | e4+26 | tier3 | 43 | 0 | 0.000 | 66 | 44 | 0.400 |
| 678913 | 15 | 50784950 | 50784950 | C | T | SNP | <i>USP8</i> | ENST00000307179 | 1 | missense | c.2287 | p.R763W | tier1 | 67 | 0 | 0.000 | 117 | 22 | 0.158 |
| 678913 | 16 | 5040788 | 5040788 | C | T | SNP | <i>SEC14L5</i> | ENST00000251170 | 1 | silent | c.366 | p.D122 | tier1 | 118 | 0 | 0.000 | 123 | 88 | 0.415 |
| 678913 | 17 | 58740533 | 58740533 | A | - | DEL | <i>PPM1D</i> | ENST00000305921 | 1 | frame_shift_del | c.1438 | p.A481fs | tier1 | 63 | 0 | 0.000 | 82 | 62 | 0.431 |
| 678913 | 20 | 29628225 | 29628225 | A | G | SNP | <i>FRG1B</i> | ENST00000278882 | 1 | splice_site | c.229-2 | e4-2 | tier1 | 602 | 5 | 0.008 | 1566 | 51 | 0.032 |
| 678913 | 20 | 30618789 | 30618789 | C | T | SNP | <i>LOC101929636</i> | ENST00000449519 | -1 | rna | NULL | NULL | tier1 | 133 | 1 | 0.008 | 147 | 79 | 0.350 |
| 678913 | 21 | 27542825 | 27542825 | G | C | SNP | <i>APP</i> | ENST00000346798 | -1 | intronic | c.57+57 | e1+57 | tier2 | 39 | 0 | 0.000 | 52 | 20 | 0.278 |
| 715413 | 1 | 172378884 | 172378884 | - | T | INS | <i>DNM3</i> | ENST00000358155 | 1 | 3_prime_untranslated_region | c.*1903 | NULL | tier3 | 12 | 0 | 0.000 | 65 | 41 | 0.387 |
| 715413 | 1 | 201781078 | 201781078 | A | C | SNP | <i>NAV1</i> | ENST00000367296 | 1 | missense | c.5000 | p.N1667T | tier1 | 315 | 2 | 0.006 | 977 | 64 | 0.062 |
| 715413 | 6 | 325294 | 325294 | A | G | SNP | <i>DUSP22</i> | ENST00000604971 | 1 | 5_prime_untranslated_region | c.-22855 | NULL | tier3 | 212 | 3 | 0.014 | 579 | 41 | 0.066 |
| 715413 | 6 | 32548143 | 32548143 | T | C | SNP | <i>HLA-DRB1</i> | ENST00000360004 | -1 | intronic | c.764-96 | e5-96 | tier3 | 36 | 1 | 0.027 | 90 | 29 | 0.244 |
| 715413 | 6 | 32548146 | 32548146 | C | T | SNP | <i>HLA-DRB1</i> | ENST00000360004 | -1 | intronic | c.764-99 | e5-99 | tier3 | 35 | 1 | 0.028 | 82 | 29 | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|---|---|-----|------------------------|-----------------|----|-----------------------------|--------------|-----------|-------|-----|----|-------|------|-----|-------|
| 715413 | 20 | 29612347 | 29612347 | C | A | SNP | <i>FRG1B</i> | ENST00000482423 | 1 | rna | NULL | NULL | tier1 | 250 | 1 | 0.004 | 815 | 51 | 0.059 |
| 715413 | X | 34961763 | 34961763 | A | C | SNP | <i>FAM47B</i> | ENST00000329357 | 1 | missense | c.815 | p.H272P | tier1 | 422 | 2 | 0.005 | 1081 | 47 | 0.041 |
| 715413 | X | 154009603 | 154009603 | G | A | SNP | <i>MPP1</i> | ENST00000482757 | -1 | 5_prime_untranslated_region | c.-154009603 | NULL | tier3 | 62 | 0 | 0.000 | 137 | 17 | 0.110 |
| 717045 | 1 | 32381592 | 32381592 | T | A | SNP | <i>PTP4A2</i> | ENST00000344035 | -1 | splice_region | c.97-4 | e2-4 | tier4 | 122 | 1 | 0.008 | 141 | 15 | 0.096 |
| 717045 | 1 | 47548156 | 47548156 | G | A | SNP | <i>CYP4Z1</i> | ENST00000334194 | 1 | intronic | c.492+23 | e4+23 | tier3 | 89 | 2 | 0.022 | 80 | 52 | 0.394 |
| 717045 | 1 | 200878061 | 200878061 | G | A | SNP | <i>C1orf106</i> | ENST00000367342 | 1 | missense | c.1033 | p.E345K | tier1 | 51 | 1 | 0.019 | 60 | 49 | 0.450 |
| 717045 | 1 | 216678795 | 216678796 | - | T | INS | <i>ESRRG</i> | ENST00000408911 | -1 | 3_prime_untranslated_region | c.*1486 | NULL | tier3 | 66 | 0 | 0.000 | 70 | 9 | 0.114 |
| 717045 | 2 | 32666548 | 32666548 | T | G | SNP | <i>BIRC6</i> | ENST00000421745 | 1 | intronic | c.3944+18 | e17+18 | tier3 | 123 | 3 | 0.024 | 79 | 51 | 0.392 |
| 717045 | 2 | 122368248 | 122368248 | T | G | SNP | <i>CLASP1</i> | ENST00000263710 | -1 | intronic | c.1-4492 | e1-4492 | tier4 | 70 | 1 | 0.014 | 87 | 17 | 0.160 |
| 717045 | 2 | 131947387 | 131947387 | G | T | SNP | <i>PLEKHB2</i> | ENST00000303908 | 1 | intronic | c.423+56823 | e5+56823 | tier3 | 463 | 6 | 0.013 | 412 | 24 | 0.055 |
| 717045 | 2 | 132290441 | 132290441 | G | A | SNP | <i>CCDC74A</i> | ENST00000295171 | 1 | missense | c.881 | p.R294Q | tier1 | 96 | 0 | 0.000 | 171 | 16 | 0.086 |
| 717045 | 2 | 179481333 | 179481333 | A | T | SNP | <i>TTN</i> | ENST00000342992 | -1 | nonsense | c.40481 | p.L13494* | tier1 | 97 | 1 | 0.010 | 84 | 47 | 0.359 |
| 717045 | 2 | 198266834 | 198266834 | T | C | SNP | <i>SF3B1</i> | ENST00000335508 | -1 | missense | c.2098 | p.K700E | tier1 | 308 | 4 | 0.013 | 303 | 204 | 0.401 |
| 717045 | 2 | 217280235 | 217280235 | T | C | SNP | <i>SMARCAL1</i> | ENST00000357276 | 1 | missense | c.808 | p.Y270H | tier1 | 36 | 1 | 0.027 | 25 | 19 | 0.432 |
| 717045 | 3 | 27208497 | 27208497 | A | T | SNP | <i>NEK10</i> | ENST00000383771 | -1 | intronic | c.768-4268 | e8-4268 | tier3 | 43 | 0 | 0.000 | 10 | 19 | 0.655 |
| 717045 | 3 | 71355167 | 71355167 | T | C | SNP | <i>FOXP1</i> | ENST00000318789 | -1 | intronic | c.1-6136 | e1-6136 | tier2 | 31 | 0 | 0.000 | 33 | 12 | 0.267 |
| 717045 | 4 | 25407313 | 25407313 | G | A | SNP | <i>ANAPC4</i> | ENST00000510092 | 1 | intronic | c.1320+69 | e17+69 | tier3 | 36 | 0 | 0.000 | 22 | 9 | 0.290 |
| 717045 | 4 | 105393360 | 105393360 | C | T | SNP | <i>CXXC4</i> | ENST00000466963 | -1 | rna | NULL | NULL | tier1 | 55 | 0 | 0.000 | 26 | 15 | 0.357 |
| 717045 | 6 | 129475728 | 129475728 | G | A | SNP | <i>LAMA2</i> | ENST00000421865 | 1 | missense | c.1106 | p.R369H | tier1 | 204 | 0 | 0.000 | 138 | 64 | 0.317 |
| 717045 | 7 | 73973313 | 73973313 | G | A | SNP | <i>GTF2IRD1</i> | ENST00000265755 | 1 | missense | c.2275 | p.G759S | tier1 | 62 | 0 | 0.000 | 43 | 42 | 0.494 |
| 717045 | 7 | 151878771 | 151878771 | T | C | SNP | <i>KMT2C</i> | ENST00000355193 | -1 | silent | c.6174 | p.G2058 | tier1 | 694 | 10 | 0.014 | 649 | 333 | 0.338 |
| 717045 | 8 | 9639151 | 9639151 | A | G | SNP | <i>TNKS</i> | ENST00000310430 | 1 | 3_prime_untranslated_region | c.*4905 | NULL | tier3 | 87 | 1 | 0.011 | 74 | 47 | 0.388 |
| 717045 | 10 | 61666207 | 61666207 | G | A | SNP | <i>CCDC6</i> | ENST00000263102 | -1 | 5_prime_untranslated_region | c.-25 | NULL | tier2 | 15 | 0 | 0.000 | 6 | 6 | 0.500 |
| 717045 | 10 | 71620368 | 71620368 | C | T | SNP | <i>COL13A1</i> | ENST00000356340 | 1 | intronic | c.365-11564 | e3-11564 | tier3 | 138 | 4 | 0.028 | 137 | 81 | 0.370 |
| 717045 | 11 | 1770224 | 1770224 | C | G | SNP | <i>IFITM10</i> | ENST00000482459 | -1 | rna | NULL | NULL | tier1 | 8 | 0 | 0.000 | 1 | 4 | 0.800 |
| 717045 | 12 | 49724201 | 49724201 | G | A | SNP | <i>TROAP</i> | ENST00000257909 | 1 | missense | c.1573 | p.A525T | tier1 | 47 | 3 | 0.060 | 40 | 30 | 0.423 |
| 717045 | 14 | 88945999 | 88945999 | G | A | SNP | <i>PTPN21</i> | ENST00000328736 | -1 | silent | c.1776 | p.P592 | tier1 | 167 | 1 | 0.006 | 105 | 83 | 0.442 |
| 717045 | 14 | 106805481 | 106805481 | G | A | SNP | <i>IGHV4-31</i> | ENST00000438142 | -1 | missense | c.82 | p.P28S | tier1 | 210 | 3 | 0.014 | 259 | 28 | 0.097 |
| 717045 | 14 | 106805485 | 106805485 | C | G | SNP | <i>IGHV4-31</i> | ENST00000438142 | -1 | silent | c.78 | p.S26 | tier1 | 213 | 3 | 0.014 | 268 | 28 | 0.095 |
| 717045 | 14 | 106805502 | 106805502 | C | G | SNP | <i>IGHV4-31</i> | ENST00000438142 | -1 | missense | c.61 | p.V21L | tier1 | 232 | 2 | 0.009 | 274 | 27 | 0.090 |
| 717045 | 14 | 106805533 | 106805533 | A | C | SNP | <i>IGHV4-31</i> | ENST00000438142 | -1 | intronic | c.47-17 | e2-17 | tier3 | 258 | 2 | 0.008 | 319 | 24 | 0.070 |
| 717045 | 15 | 101432730 | 101432730 | G | C | SNP | <i>ALDH1A3</i> | ENST00000329841 | 1 | missense | c.361 | p.D121H | tier1 | 88 | 3 | 0.033 | 70 | 59 | 0.457 |
| 717045 | 16 | 66432084 | 66432084 | G | T | SNP | <i>CDH5</i> | ENST00000341529 | 1 | intronic | c.1485+75 | e8+75 | tier3 | 41 | 0 | 0.000 | 40 | 33 | 0.452 |
| 717045 | 17 | 40731892 | 40731892 | C | T | SNP | <i>FAM134C</i> | ENST00000309428 | -1 | 3_prime_untranslated_region | c.*1939 | NULL | tier3 | 111 | 5 | 0.043 | 82 | 38 | 0.317 |
| 717045 | 17 | 58979988 | 58979988 | G | T | SNP | <i>BCAS3</i> | ENST00000390652 | 1 | missense | c.746 | p.R249L | tier1 | 118 | 0 | 0.000 | 75 | 7 | 0.085 |
| 717045 | 19 | 1046259 | 1046259 | C | G | SNP | <i>ABCA7</i> | ENST00000263094 | 1 | silent | c.1476 | p.P492 | tier1 | 165 | 2 | 0.012 | 118 | 70 | 0.368 |
| 717045 | 20 | 39992390 | 39992390 | C | T | SNP | <i>EMILIN3</i> | ENST00000332312 | -1 | silent | c.402 | p.T134 | tier1 | 102 | 1 | 0.010 | 91 | 74 | 0.449 |
| 717045 | 20 | 61444605 | 61444605 | A | G | SNP | <i>OGFR</i> | ENST00000290291 | 1 | silent | c.1638 | p.P546 | tier1 | 27 | 0 | 0.000 | 30 | 10 | 0.250 |
| 717045 | 22 | 24400302 | 24400302 | G | A | SNP | <i>GSTTP2</i> | ENST00000389399 | -1 | splice_region | c.NULL | NULL | tier3 | 121 | 0 | 0.000 | 223 | 21 | 0.086 |
| 732217 | 1 | 1896486 | 1896486 | G | A | SNP | <i>C1orf222</i> | ENST00000604159 | -1 | silent | c.171 | p.D57 | tier1 | 34 | 0 | 0.000 | 49 | 39 | 0.443 |
| 732217 | 1 | 19549161 | 19549161 | C | T | SNP | <i>EMC1</i> | ENST00000486238 | -1 | missense | c.95 | p.R32K | tier1 | 29 | 1 | 0.033 | 42 | 39 | 0.482 |
| 732217 | 1 | 20518895 | 20518895 | C | T | SNP | <i>UBXN10</i> | ENST00000375099 | 1 | 3_prime_untranslated_region | c.*998 | NULL | tier3 | 81 | 0 | 0.000 | 97 | 63 | 0.389 |
| 732217 | 1 | 46079781 | 46079781 | A | G | SNP | <i>NASP</i> | ENST00000402363 | 1 | missense | c.1526 | p.N509S | tier1 | 82 | 3 | 0.035 | 144 | 125 | 0.465 |
| 732217 | 1 | 237777658 | 237777658 | A | - | DEL | <i>RYR2</i> | ENST00000360064 | 1 | frame_shift_del | c.5224 | p.K1743fs | tier1 | 42 | 2 | 0.046 | 55 | 28 | 0.337 |
| 732217 | 2 | 112588927 | 112588927 | C | G | SNP | <i>ANAPC1</i> | ENST00000341068 | -1 | missense | c.2561 | p.G854A | tier1 | 195 | 8 | 0.039 | 400 | 328 | 0.451 |
| 732217 | 2 | 236949476 | 236949476 | G | A | SNP | <i>AGAP1</i> | ENST00000304032 | 1 | missense | c.1882 | p.E628K | tier1 | 51 | 3 | 0.056 | 113 | 51 | 0.311 |
| 732217 | 4 | 106164914 | 106164914 | G | A | SNP | <i>TET2</i> | ENST00000380013 | 1 | missense | c.3782 | p.R1261H | tier1 | 171 | 4 | 0.023 | 30 | 571 | 0.945 |
| 732217 | 4 | 125593542 | 125593542 | A | G | SNP | <i>ANKRD50</i> | ENST00000504087 | -1 | missense | c.890 | p.I297T | tier1 | 71 | 3 | 0.041 | 7 | 145 | 0.954 |
| 732217 | 5 | 69826356 | 69826356 | G | A | SNP | <i>ENSG00000250918</i> | ENST00000509075 | 1 | 3_prime_untranslated_region | c.*69826356 | NULL | tier3 | 166 | 4 | 0.023 | 720 | 87 | 0.107 |
| 732217 | 5 | 98109985 | 98109985 | G | A | SNP | <i>RGMB</i> | ENST00000308234 | 1 | intronic | c.259+75 | e2+75 | tier2 | 7 | 0 | 0.000 | 1 | 6 | 0.857 |
| 732217 | 5 | 171723836 | 171723836 | G | A | SNP | <i>UBTD2</i> | ENST00000393792 | -1 | 5_prime_flanking_region | c.-13167 | NULL | tier2 | 73 | 0 | 0.000 | 140 | 19 | 0.120 |
| 732217 | 6 | 150064734 | 150064734 | A | - | DEL | <i>NUP43</i> | ENST00000340413 | -1 | intronic | c.321+40 | e3+40 | tier4 | 48 | 0 | 0.000 | 134 | 26 | 0.163 |
| 732217 | 7 | 31378474 | 31378474 | G | A | SNP | <i>NEUROD6</i> | ENST00000297142 | -1 | nonsense | c.409 | p.R137* | tier1 | 121 | 3 | 0.024 | 153 | 97 | 0.388 |
| 732217 | 7 | 42003680 | 42003680 | A | T | SNP | <i>GLI3</i> | ENST00000479210 | -1 | rna | NULL | NULL | tier1 | 71 | 1 | 0.014 | 251 | 51 | 0.168 |
| 732217 | 7 | 42003681 | 42003681 | A | T | SNP | <i>GLI3</i> | ENST00000479210 | -1 | rna | NULL | NULL | tier1 | 74 | 0 | 0.000 | 271 | 25 | 0.084 |
| 732217 | 7 | 48545892 | 48545892 | G | A | SNP | <i>ABCA13</i> | ENST00000435803 | 1 | intronic | c.13292-40 | e49-40 | tier3 | 32 | 1 | 0.030 | 62 | 38 | 0.380 |
| 732217 | 7 | 73475359 | 73475359 | T | C | SNP | <i>ELN</i> | ENST00000358929 | 1 | intronic | c.1952-72 | e27-72 | tier4 | 46 | 0 | 0.000 | 86 | 70 | 0.449 |
| 732217 | 7 | 142223845 | 142223845 | T | C | SNP | <i>TRBV11-1</i> | ENST00000390367 | -1 | missense | c.322 | p.M108V | tier1 | 47 | 0 | 0.000 | 96 | 15 | 0.135 |
| 732217 | 8 | 52732382 | 52732382 | C | T | SNP | <i>PCMTD1</i> | ENST00000360540 | -1 | 3_prime_untranslated_region | c.*529 | NULL | tier3 | 47 | 0 | 0.000 | 141 | 23 | 0.139 |
| 732217 | 8 | 77764819 | 77764819 | C | - | DEL | <i>ZFX4</i> | ENST00000521891 | 1 | frame_shift_del | c.5662 | p.Q1888fs | tier1 | 44 | 1 | 0.022 | 56 | 47 | 0.456 |
| 732217 | 8 | 145255402 | 145255402 | G | A | SNP | <i>MROH1</i> | ENST00000326134 | 1 | missense | c.1099 | p.V367M | tier1 | 28 | 0 | 0.000 | 33 | 34 | 0.500 |
| 732217 | 9 | 14788 | 14788 | G | A | SNP | <i>ENSG00000181404</i> | ENST00000330546 | -1 | 5_prime_untranslated_region | c.-14788 | NULL | tier3 | 397 | 8 | 0.020 | 655 | 56 | 0.078 |
| 732217 | 9 | 33386977 | 33386977 | G | T | SNP | <i>AQP7</i> | ENST00000297988 | -1 | silent | c.258 | p.G86 | tier1 | 304 | 4 | 0.013 | 802 | 125 | 0.135 |
| 732217 | 9 | 130897518 | 130897518 | A | C | SNP | <i>PTGES2</i> | ENST00000338961 | -1 | 5_prime_flanking_region | c.-7522 | NULL | tier4 | 139 | 2 | 0.014 | 267 | 115 | 0.300 |
| 732217 | 10 | 102296470 | 102296470 | T | C | SNP | <i>HIF1AN</i> | ENST00000299163 | 1 | intronic | c.428+52 | e2+52 | tier2 | 21 | 0 | 0.000 | 35 | 43 | 0.551 |
| 732217 | 10 | 119003712 | 119003712 | C | T | SNP | <i>SLC18A2</i> | ENST00000298472 | 1 | missense | c.352 | p.P118S | tier1 | 41 | 2 | 0.047 | 44 | 40 | 0.476 |
| 732217 | 11 | 14279305 | 14279305 | G | T | SNP | <i>SPON1</i> | ENST00000310358 | 1 | rna | NULL | NULL | tier1 | 55 | 2 | 0.035 | 80 | 65 | 0.445 |
| 732217 | 11 | 59244984 | 592449 | | | | | | | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-----------------------------|---|------|------------------------|-----------------|----|-----------------------------|-------------------|----------|-------|-----|----|-------|-----|-----|-------|
| 732217 | 17 | 263412 | 263412 | G | A | SNP | <i>C17orf97</i> | ENST00000360127 | 1 | missense | c.778 | p.E260K | tier1 | 322 | 6 | 0.018 | 706 | 59 | 0.077 |
| 732217 | 17 | 15984046 | 15984047 | - | A | INS | <i>NCOR1</i> | ENST00000268712 | -1 | splice_region_ins | c.3178-6_3178-5 | e23-5 | tier2 | 271 | 5 | 0.018 | 466 | 219 | 0.320 |
| 732217 | 17 | 72363579 | 72363579 | T | A | SNP | <i>GPR142</i> | ENST00000582579 | 1 | 5_prime_untranslated_region | c.-66 | NULL | tier3 | 17 | 0 | 0.000 | 39 | 28 | 0.418 |
| 732217 | 17 | 72948180 | 72948180 | C | T | SNP | <i>HID1</i> | ENST00000425042 | -1 | intronic | c.2145-22 | e18-22 | tier3 | 33 | 0 | 0.000 | 35 | 18 | 0.340 |
| 732217 | 17 | 74732959 | 74732959 | G | A | SNP | <i>SRSF2</i> | ENST00000359995 | -1 | missense | c.284 | p.P95L | tier1 | 164 | 5 | 0.030 | 254 | 232 | 0.476 |
| 732217 | 19 | 41557967 | 41557967 | C | T | SNP | <i>CYP2G2P</i> | ENST00000594920 | -1 | 5_prime_untranslated_region | c.-41557967 | NULL | tier3 | 96 | 2 | 0.020 | 110 | 115 | 0.509 |
| 732217 | 20 | 31022441 | 31022442 | - | G | INS | <i>ASXL1</i> | ENST00000375687 | 1 | frame_shift_ins | c.1926_1927 | p.G645fs | tier1 | 553 | 14 | 0.025 | 689 | 347 | 0.335 |
| 732217 | 21 | 36164760 | 36164761 | - | - | CCAT | <i>RUNX1</i> | ENST00000300305 | -1 | frame_shift_ins | c.1115_1114 | p.G372fs | tier1 | 764 | 7 | 0.009 | 691 | 414 | 0.375 |
| 732217 | 21 | 47976820 | 47976820 | T | - | DEL | <i>DIP2A</i> | ENST00000481883 | 1 | 3_prime_untranslated_region | c.*47976820 | NULL | tier3 | 56 | 0 | 0.000 | 145 | 20 | 0.121 |
| 732217 | X | 106482271 | 106482271 | T | C | SNP | <i>PIH1D3</i> | ENST00000336387 | 1 | intronic | c.515+34 | e5+34 | tier3 | 40 | 2 | 0.048 | 13 | 120 | 0.902 |
| 754150 | 1 | 10683092 | 10683092 | T | C | SNP | <i>PEX14</i> | ENST00000356607 | 1 | missense | c.401 | p.L134P | tier1 | 160 | 0 | 0.000 | 154 | 22 | 0.124 |
| 754150 | 1 | 115401238 | 115401239 | - | A | INS | <i>SYCP1</i> | ENST00000369518 | 1 | frame_shift_ins | c.362_363 | p.S122fs | tier1 | 65 | 0 | 0.000 | 98 | 12 | 0.109 |
| 754150 | 2 | 46707812 | 46707812 | G | A | SNP | <i>TMEM247</i> | ENST00000434431 | 1 | missense | c.386 | p.R129Q | tier1 | 65 | 0 | 0.000 | 57 | 8 | 0.123 |
| 754150 | 2 | 149793838 | 149793838 | G | A | SNP | <i>KIF5C</i> | ENST00000435030 | 1 | missense | c.332 | p.R111Q | tier1 | 52 | 0 | 0.000 | 55 | 10 | 0.154 |
| 754150 | 2 | 208394531 | 208394531 | G | T | SNP | <i>CREB1</i> | ENST00000430624 | 1 | 5_prime_untranslated_region | c.-25829 | NULL | tier2 | 6 | 0 | 0.000 | 1 | 5 | 0.833 |
| 754150 | 2 | 219221645 | 219221645 | C | G | SNP | <i>C2orf62</i> | ENST00000289388 | 1 | intronic | c.25+13 | e1+13 | tier3 | 171 | 0 | 0.000 | 119 | 11 | 0.085 |
| 754150 | 2 | 239973727 | 239973727 | T | A | SNP | <i>HDAC4</i> | ENST00000345617 | -1 | 3_prime_untranslated_region | c.*1066 | NULL | tier3 | 180 | 4 | 0.022 | 109 | 73 | 0.401 |
| 754150 | 3 | 52012429 | 52012429 | T | A | SNP | <i>ABHD14A</i> | ENST00000474575 | 1 | 3_prime_untranslated_region | c.*52012429 | NULL | tier3 | 137 | 1 | 0.007 | 104 | 22 | 0.175 |
| 754150 | 3 | 69915190 | 69915190 | G | A | SNP | <i>MITF</i> | ENST00000448226 | 1 | intronic | c.105-13095 | e2-13095 | tier3 | 82 | 0 | 0.000 | 81 | 17 | 0.174 |
| 754150 | 3 | 182933850 | 182933850 | G | C | SNP | <i>MCF2L2</i> | ENST00000328913 | -1 | missense | c.2403 | p.F801L | tier1 | 130 | 0 | 0.000 | 146 | 11 | 0.070 |
| 754150 | 4 | 4190576 | 4190576 | C | G | SNP | <i>OTOP1</i> | ENST00000296358 | -1 | missense | c.1793 | p.R598P | tier1 | 62 | 0 | 0.000 | 97 | 14 | 0.126 |
| 754150 | 4 | 4190577 | 4190577 | G | C | SNP | <i>OTOP1</i> | ENST00000296358 | -1 | missense | c.1792 | p.R598G | tier1 | 60 | 0 | 0.000 | 96 | 14 | 0.127 |
| 754150 | 4 | 4190595 | 4190595 | G | C | SNP | <i>OTOP1</i> | ENST00000296358 | -1 | missense | c.1774 | p.P592A | tier1 | 63 | 0 | 0.000 | 90 | 14 | 0.135 |
| 754150 | 4 | 9389009 | 9389009 | A | G | SNP | <i>ENSG00000219492</i> | ENST00000508324 | -1 | splice_region | c.13-8 | e2-8 | tier3 | 497 | 13 | 0.026 | 243 | 176 | 0.419 |
| 754150 | 4 | 20852133 | 20852133 | G | T | SNP | <i>KCNIP4</i> | ENST00000515373 | -1 | 5_prime_untranslated_region | c.-20852133 | NULL | tier3 | 110 | 4 | 0.035 | 57 | 46 | 0.447 |
| 754150 | 4 | 25262265 | 25262265 | C | T | SNP | <i>PI4K2B</i> | ENST00000264864 | 1 | intronic | c.978+52 | e6+52 | tier4 | 41 | 1 | 0.024 | 54 | 17 | 0.239 |
| 754150 | 4 | 25677760 | 25677760 | G | A | SNP | <i>SLC34A2</i> | ENST00000382051 | 1 | missense | c.1462 | p.A488T | tier1 | 54 | 0 | 0.000 | 59 | 9 | 0.132 |
| 754150 | 5 | 26906108 | 26906108 | C | G | SNP | <i>CDH9</i> | ENST00000231021 | -1 | silent | c.771 | p.T257 | tier1 | 154 | 2 | 0.013 | 158 | 37 | 0.189 |
| 754150 | 5 | 55168187 | 55168187 | C | T | SNP | <i>IL31RA</i> | ENST00000447346 | 1 | missense | c.362 | p.T121M | tier1 | 162 | 0 | 0.000 | 126 | 89 | 0.414 |
| 754150 | 5 | 115177743 | 115177743 | G | C | SNP | <i>AP3S1</i> | ENST00000316788 | 1 | missense | c.9 | p.K3N | tier1 | 22 | 0 | 0.000 | 19 | 11 | 0.367 |
| 754150 | 5 | 139941115 | 139941115 | T | C | SNP | <i>APBB3</i> | ENST00000356738 | -1 | intronic | c.762+57 | e7+57 | tier2 | 96 | 3 | 0.030 | 48 | 44 | 0.478 |
| 754150 | 5 | 140554370 | 140554370 | C | T | SNP | <i>PCDHB7</i> | ENST00000231137 | 1 | missense | c.1954 | p.R652C | tier1 | 829 | 7 | 0.008 | 707 | 79 | 0.100 |
| 754150 | 5 | 178413301 | 178413301 | C | T | SNP | <i>GRM6</i> | ENST00000231188 | -1 | missense | c.1954 | p.A652T | tier1 | 83 | 3 | 0.035 | 65 | 43 | 0.398 |
| 754150 | 6 | 52400654 | 52400654 | A | C | SNP | <i>TRAM2</i> | ENST00000182527 | -1 | splice_region | c.121-10 | e2-10 | tier2 | 87 | 2 | 0.023 | 73 | 58 | 0.443 |
| 754150 | 7 | 31127292 | 31127292 | T | C | SNP | <i>ADCYAP1R1</i> | ENST00000436116 | 1 | missense | c.85 | p.C29R | tier1 | 108 | 0 | 0.000 | 99 | 15 | 0.132 |
| 754150 | 7 | 42005086 | 42005086 | G | T | SNP | <i>GLI3</i> | ENST00000395925 | -1 | missense | c.3585 | p.N1195K | tier1 | 83 | 4 | 0.046 | 58 | 42 | 0.420 |
| 754150 | 7 | 77584160 | 77584160 | T | - | DEL | <i>PHTF2</i> | ENST00000248550 | 1 | intronic | c.2338-15 | e19-15 | tier3 | 152 | 0 | 0.000 | 226 | 23 | 0.092 |
| 754150 | 11 | 2443297 | 2443297 | G | A | SNP | <i>TRPM5</i> | ENST00000452833 | -1 | intronic | c.298+74 | e2+74 | tier3 | 25 | 0 | 0.000 | 14 | 6 | 0.300 |
| 754150 | 11 | 34192643 | 34192643 | C | T | SNP | <i>ABTB2</i> | ENST00000435224 | -1 | intronic | c.1398-25 | e5-25 | tier3 | 71 | 0 | 0.000 | 62 | 18 | 0.225 |
| 754150 | 11 | 48346968 | 48346968 | C | T | SNP | <i>OR4C3</i> | ENST00000319856 | 1 | missense | c.476 | p.T159L | tier1 | 85 | 1 | 0.012 | 98 | 23 | 0.190 |
| 754150 | 12 | 19653148 | 19653148 | A | T | SNP | <i>AEBP2</i> | ENST00000398864 | 1 | missense | c.1292 | p.H431L | tier1 | 92 | 0 | 0.000 | 75 | 50 | 0.400 |
| 754150 | 12 | 50746672 | 50746672 | A | G | SNP | <i>FAM186A</i> | ENST00000327337 | -1 | missense | c.3943 | p.F1315L | tier1 | 285 | 4 | 0.014 | 283 | 28 | 0.090 |
| 754150 | 13 | 101712169 | 101712169 | C | T | SNP | <i>NALCN</i> | ENST00000251127 | -1 | splice_site | c.4905+1 | e41+1 | tier1 | 606 | 19 | 0.030 | 313 | 251 | 0.445 |
| 754150 | 14 | 91467486 | 91467486 | C | T | SNP | <i>RPS6KA5</i> | ENST00000261991 | -1 | missense | c.121 | p.A41T | tier1 | 190 | 4 | 0.021 | 126 | 88 | 0.407 |
| 754150 | 15 | 76576183 | 76576183 | A | - | DEL | <i>ETFA</i> | ENST00000557943 | -1 | intronic | c.665-17 | e8-17 | tier3 | 30 | 0 | 0.000 | 23 | 8 | 0.258 |
| 754150 | 15 | 90631934 | 90631934 | C | T | SNP | <i>IDH2</i> | ENST00000330062 | -1 | missense | c.419 | p.R140Q | tier1 | 589 | 12 | 0.020 | 280 | 269 | 0.490 |
| 754150 | 15 | 102295647 | 102295647 | C | G | SNP | <i>DNM1P47</i> | ENST00000561463 | 1 | rna | NULL | NULL | tier1 | 37 | 1 | 0.026 | 21 | 12 | 0.364 |
| 754150 | 16 | 12297294 | 12297294 | T | G | SNP | <i>SNX29</i> | ENST00000306030 | 1 | intronic | c.523+3754 | e6+3754 | tier3 | 572 | 7 | 0.012 | 619 | 28 | 0.043 |
| 754150 | 17 | 2595220 | 2595233 | CCCCGTGCCCGCGG | - | DEL | <i>CLUH</i> | ENST00000435359 | -1 | intronic | c.3538-44_3538-31 | e23-31 | tier2 | 28 | 0 | 0.000 | 19 | 11 | 0.367 |
| 754150 | 17 | 74732959 | 74732959 | G | C | SNP | <i>SRSF2</i> | ENST00000359995 | -1 | missense | c.284 | p.P95R | tier1 | 390 | 20 | 0.049 | 228 | 138 | 0.377 |
| 754150 | 19 | 426497 | 426497 | C | T | SNP | <i>SHC2</i> | ENST00000264554 | -1 | intronic | c.1175-1266 | e10-1266 | tier4 | 59 | 0 | 0.000 | 34 | 6 | 0.143 |
| 754150 | 19 | 6906516 | 6906516 | C | T | SNP | <i>EMR1</i> | ENST00000312053 | 1 | missense | c.1022 | p.A341V | tier1 | 218 | 7 | 0.031 | 121 | 133 | 0.522 |
| 754150 | 19 | 15654862 | 15654862 | G | A | SNP | <i>CYP4F22</i> | ENST00000269703 | 1 | intronic | c.1006+14 | e7+14 | tier3 | 148 | 3 | 0.020 | 91 | 48 | 0.345 |
| 754150 | 19 | 41355820 | 41355820 | A | G | SNP | <i>CYP2A6</i> | ENST00000301141 | -1 | silent | c.246 | p.C82 | tier1 | 83 | 0 | 0.000 | 83 | 13 | 0.135 |
| 754150 | 19 | 41355828 | 41355828 | C | T | SNP | <i>CYP2A6</i> | ENST00000301141 | -1 | missense | c.238 | p.V80M | tier1 | 78 | 0 | 0.000 | 83 | 13 | 0.135 |
| 754150 | 20 | 3127469 | 3127469 | C | G | SNP | <i>FASTKD5</i> | ENST00000380266 | -1 | missense | c.2248 | p.E750Q | tier1 | 70 | 3 | 0.041 | 33 | 49 | 0.598 |
| 754150 | 20 | 29632749 | 29632749 | A | G | SNP | <i>FRG1B</i> | ENST00000278882 | 1 | intronic | c.536+28 | e6+28 | tier3 | 262 | 3 | 0.011 | 443 | 27 | 0.057 |
| 754150 | 20 | 29652147 | 29652147 | A | G | SNP | <i>MLLT10P1</i> | ENST00000418346 | -1 | 5_prime_flanking_region | c.-14087 | NULL | tier4 | 267 | 4 | 0.015 | 342 | 27 | 0.073 |
| 754150 | 20 | 31022403 | 31022425 | CACCACTGCCATAGA GAGGCGGC | - | DEL | <i>ASXL1</i> | ENST00000375687 | 1 | frame_shift_del | c.1888_1910 | p.H633fs | tier1 | 875 | 7 | 0.008 | 481 | 78 | 0.140 |
| 754150 | 20 | 31022417 | 31022417 | A | - | DEL | <i>ASXL1</i> | ENST00000375687 | 1 | frame_shift_del | c.1902 | p.E635fs | tier1 | 843 | 7 | 0.008 | 390 | 45 | 0.103 |
| 754150 | 20 | 55967777 | 55967777 | G | C | SNP | <i>RBM38</i> | ENST00000356208 | 1 | missense | c.305 | p.R102P | tier1 | 53 | 3 | 0.054 | 47 | 31 | 0.397 |
| 754150 | 22 | 24094823 | 24094828 | ATCCCC | - | DEL | <i>MMP11</i> | ENST00000215743 | 1 | 5_prime_flanking_region | c.-20235 | NULL | tier3 | 34 | 1 | 0.029 | 30 | 19 | 0.388 |
| 754150 | 22 | 50903351 | 50903351 | C | T | SNP | <i>SBF1</i> | ENST00000380817 | -1 | splice_region | c.1333-5 | e13-5 | tier2 | 252 | 10 | 0.038 | 144 | 107 | 0.423 |
| 754150 | X | 73064091 | 73064091 | A | G | SNP | <i>XIST</i> | ENST00000429829 | -1 | rna | NULL | NULL | tier1 | 65 | 1 | 0.015 | 13 | 67 | 0.838 |
| 754150 | X | 123505288 | 123505288 | T | C | SNP | <i>SH2D1A</i> | ENST00000491950 | 1 | rna | NULL | NULL | tier1 | 140 | 6 | 0.041 | 22 | 143 | 0.867 |
| 769516 | 1 | 15046 | 15046 | G | A | SNP | <i>WASH7P</i> | ENST00000423562 | -1 | splice_region | c.NULL | NULL | tier3 | 162 | 0 | 0.000 | 501 | 44 | 0.081 |
| 769516 | 1 | 116947126 | 116947126 | C | G | SNP | <i>ATP1A1</i> | ENST00000295598 | 1 | 3_prime_untranslated_region | c.*60 | NULL | tier3 | 28 | 0 | 0.0 | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|---|-----|-----|-----------------|-----------------|----|-----------------------------|-------------------|-------------------|-------|-----|---|-------|------|-----|-------|
| 769516 | 4 | 187541903 | 187541903 | C | T | SNP | FAT1 | ENST00000441802 | -1 | missense | c.5837 | p.S1946N | tier1 | 16 | 0 | 0.000 | 92 | 41 | 0.308 |
| 769516 | 5 | 15936967 | 15936967 | C | T | SNP | FBXL7 | ENST00000504595 | 1 | missense | c.1148 | p.A383V | tier1 | 13 | 0 | 0.000 | 33 | 30 | 0.476 |
| 769516 | 5 | 73168759 | 73168759 | T | A | SNP | ARHGFEF28 | ENST00000545377 | 1 | intronic | c.2567-65 | e21-65 | tier3 | 30 | 0 | 0.000 | 99 | 45 | 0.313 |
| 769516 | 6 | 56466430 | 56466430 | A | T | SNP | DST | ENST00000370754 | -1 | silent | c.11259 | p.T3753 | tier1 | 78 | 0 | 0.000 | 796 | 310 | 0.280 |
| 769516 | 9 | 100137897 | 100137897 | C | G | SNP | CCDC180 | ENST00000526038 | 1 | 3_prime_untranslated_region | c.*100137897 | NULL | tier3 | 57 | 0 | 0.000 | 405 | 171 | 0.297 |
| 769516 | 9 | 127549340 | 127549340 | C | T | SNP | OLFML2A | ENST00000373580 | 1 | silent | c.177 | p.D59 | tier1 | 22 | 0 | 0.000 | 133 | 49 | 0.269 |
| 769516 | 11 | 376244 | 376245 | - | A | INS | B4GALNT4 | ENST00000329962 | 1 | splice_region_ins | c.1197-7_1197-6 | e13-6 | tier2 | 38 | 0 | 0.000 | 233 | 78 | 0.251 |
| 769516 | 11 | 68273518 | 68273518 | T | C | SNP | PPP6R3 | ENST00000393799 | 1 | intronic | c.1-13451 | e1-13451 | tier3 | 47 | 0 | 0.000 | 216 | 37 | 0.146 |
| 769516 | 12 | 125478381 | 125478382 | - | CTG | INS | BRI3BP | ENST00000341446 | 1 | in_frame_ins | c.45_46 | p.19in_frame_insL | tier1 | 8 | 0 | 0.000 | 7 | 10 | 0.588 |
| 769516 | 13 | 32783001 | 32783001 | C | T | SNP | FRY | ENST00000380250 | 1 | nonsense | c.4030 | p.R1344* | tier1 | 28 | 0 | 0.000 | 135 | 37 | 0.214 |
| 769516 | 14 | 59113492 | 59113492 | C | T | SNP | DACT1 | ENST00000335867 | 1 | silent | c.2151 | p.Y717 | tier1 | 30 | 0 | 0.000 | 149 | 51 | 0.255 |
| 769516 | 15 | 40942716 | 40942716 | T | - | DEL | CASC5 | ENST00000346991 | 1 | intronic | c.6251-26 | e19-26 | tier3 | 32 | 0 | 0.000 | 112 | 34 | 0.233 |
| 769516 | 16 | 81208267 | 81208267 | A | C | SNP | PKD1L2 | ENST00000527937 | -1 | missense | c.604 | p.C202G | tier1 | 181 | 0 | 0.000 | 693 | 288 | 0.293 |
| 769516 | 17 | 71361450 | 71361450 | T | C | SNP | SDK2 | ENST00000392650 | -1 | missense | c.5252 | p.N1751S | tier1 | 26 | 0 | 0.000 | 128 | 43 | 0.250 |
| 769516 | 17 | 74732959 | 74732959 | G | C | SNP | SRSF2 | ENST00000359995 | -1 | missense | c.284 | p.P95R | tier1 | 114 | 0 | 0.000 | 454 | 183 | 0.285 |
| 769516 | 18 | 14361468 | 14361468 | G | A | SNP | ENSG00000265437 | ENST00000584979 | -1 | 5_prime_untranslated_region | c.-14361468 | NULL | tier3 | 18 | 0 | 0.000 | 107 | 54 | 0.335 |
| 769516 | 22 | 19130308 | 19130308 | G | A | SNP | DGCR14 | ENST00000252137 | -1 | missense | c.235 | p.R79W | tier1 | 14 | 0 | 0.000 | 56 | 31 | 0.356 |
| 783437 | 1 | 38488507 | 38488507 | T | - | DEL | UTP11L | ENST00000373014 | 1 | intronic | c.678+26 | e7+26 | tier3 | 68 | 0 | 0.000 | 114 | 14 | 0.109 |
| 783437 | 3 | 47263926 | 47263926 | T | - | DEL | KIF9-AS1 | ENST00000429315 | 1 | intronic | c.NULL | NULL | tier4 | 75 | 1 | 0.013 | 124 | 19 | 0.133 |
| 783437 | 3 | 50137714 | 50137714 | T | - | DEL | RBM5 | ENST00000469838 | 1 | 3_prime_untranslated_region | c.*207 | NULL | tier3 | 82 | 1 | 0.012 | 107 | 15 | 0.123 |
| 783437 | 6 | 31498127 | 31498127 | T | G | SNP | DDX39B | ENST00000462421 | -1 | rna | NULL | NULL | tier1 | 125 | 1 | 0.008 | 159 | 20 | 0.107 |
| 783437 | 6 | 102516350 | 102516350 | C | T | SNP | GRIK2 | ENST00000421544 | 1 | silent | c.2091 | p.N897 | tier1 | 791 | 2 | 0.003 | 1072 | 40 | 0.036 |
| 783437 | 7 | 95818865 | 95818865 | A | - | DEL | SLC25A13 | ENST00000416240 | -1 | intronic | c.848+28 | e8+28 | tier3 | 65 | 0 | 0.000 | 113 | 17 | 0.131 |
| 783437 | 8 | 143369132 | 143369132 | G | C | SNP | TSNARE1 | ENST00000307180 | -1 | intronic | c.1291-3286 | e10-3286 | tier3 | 585 | 4 | 0.007 | 869 | 28 | 0.031 |
| 783437 | 9 | 131104965 | 131104965 | T | - | DEL | TMSB4XP4 | ENST00000323496 | 1 | 3_prime_untranslated_region | c.*131104965 | NULL | tier3 | 122 | 1 | 0.008 | 123 | 12 | 0.089 |
| 783437 | 16 | 85689387 | 85689387 | A | C | SNP | GSE1 | ENST00000253458 | 1 | missense | c.853 | p.T285P | tier1 | 146 | 3 | 0.020 | 174 | 26 | 0.129 |
| 783437 | 17 | 76001145 | 76001145 | G | T | SNP | TNRC6C | ENST00000335749 | 1 | 5_prime_untranslated_region | c.-43999 | NULL | tier3 | 266 | 2 | 0.007 | 626 | 34 | 0.051 |
| 783437 | 21 | 46927371 | 46927412 | GGGAGGGCCAGGTG CTGGGGCGGGAGAG TCGGGTGCTGGCA | - | DEL | COL18A1 | ENST00000473212 | 1 | 3_prime_untranslated_region | c.*46927371 | NULL | tier3 | 58 | 1 | 0.017 | 74 | 16 | 0.178 |
| 796518 | 1 | 55611693 | 55611693 | C | T | SNP | USP24 | ENST00000407756 | -1 | silent | c.1860 | p.E620 | tier1 | 30 | 1 | 0.032 | 95 | 48 | 0.336 |
| 796518 | 2 | 98846643 | 98846643 | G | A | SNP | VWA3B | ENST00000477737 | 1 | missense | c.2281 | p.V761I | tier1 | 77 | 1 | 0.013 | 161 | 36 | 0.181 |
| 796518 | 2 | 171379066 | 171379066 | T | C | SNP | MYO3B | ENST00000334231 | 1 | intronic | c.3602+3016 | e30+3016 | tier3 | 50 | 0 | 0.000 | 101 | 48 | 0.322 |
| 796518 | 2 | 179346878 | 179346878 | G | A | SNP | PLEKHA3 | ENST00000234453 | 1 | intronic | c.40+1242 | e1+1242 | tier3 | 26 | 0 | 0.000 | 98 | 38 | 0.279 |
| 796518 | 2 | 198266834 | 198266834 | T | C | SNP | SF3B1 | ENST00000335508 | -1 | missense | c.2098 | p.K700E | tier1 | 239 | 0 | 0.000 | 664 | 39 | 0.056 |
| 796518 | 2 | 198267483 | 198267483 | C | A | SNP | SF3B1 | ENST00000335508 | -1 | missense | c.1874 | p.R625L | tier1 | 485 | 1 | 0.002 | 1322 | 59 | 0.043 |
| 796518 | 4 | 151682922 | 151682922 | T | C | SNP | LRBA | ENST00000357115 | -1 | intronic | c.5645+13 | e34+13 | tier4 | 126 | 0 | 0.000 | 649 | 54 | 0.077 |
| 796518 | 4 | 155254236 | 155254236 | C | T | SNP | DCHS2 | ENST00000357232 | -1 | missense | c.1627 | p.G543S | tier1 | 334 | 0 | 0.000 | 514 | 239 | 0.317 |
| 796518 | 5 | 140202971 | 140202971 | C | T | SNP | PCDHA5 | ENST00000529859 | 1 | silent | c.1611 | p.R537 | tier1 | 316 | 0 | 0.000 | 556 | 262 | 0.320 |
| 796518 | 6 | 170591580 | 170591580 | G | A | SNP | DLL1 | ENST00000366756 | -1 | 3_prime_untranslated_region | c.*382 | NULL | tier3 | 38 | 0 | 0.000 | 68 | 29 | 0.299 |
| 796518 | 7 | 28319235 | 28319235 | C | G | SNP | ENSG00000174495 | ENST00000445031 | -1 | 5_prime_untranslated_region | c.-28319235 | NULL | tier3 | 42 | 0 | 0.000 | 60 | 32 | 0.344 |
| 796518 | 11 | 55035844 | 55035844 | T | C | SNP | TRIM48 | ENST00000417545 | 1 | missense | c.574 | p.Y192H | tier1 | 46 | 2 | 0.042 | 0 | 15 | 1.000 |
| 796518 | 11 | 55035854 | 55035854 | A | A | SNP | TRIM48 | ENST00000417545 | 1 | splice_region | c.578+6 | e4+6 | tier3 | 46 | 2 | 0.042 | 0 | 13 | 1.000 |
| 796518 | 11 | 55035855 | 55035855 | T | G | SNP | TRIM48 | ENST00000417545 | 1 | splice_region | c.578+7 | e4+7 | tier3 | 45 | 2 | 0.042 | 0 | 12 | 1.000 |
| 796518 | 12 | 48179692 | 48179692 | G | T | SNP | ENSG00000268069 | ENST00000599515 | 1 | 3_prime_untranslated_region | c.*353 | NULL | tier3 | 30 | 0 | 0.000 | 61 | 28 | 0.315 |
| 796518 | 13 | 101733940 | 101733940 | G | A | SNP | NALCN | ENST00000251127 | -1 | nonsense | c.3823 | p.R1275* | tier1 | 299 | 1 | 0.003 | 722 | 44 | 0.057 |
| 796518 | 14 | 106788772 | 106788772 | G | A | SNP | IGHV2-26 | ENST00000390611 | -1 | 5_prime_flanking_region | c.-30680 | NULL | tier3 | 22 | 0 | 0.000 | 47 | 17 | 0.266 |
| 796518 | 14 | 106788774 | 106788774 | T | C | SNP | IGHV2-26 | ENST00000390611 | -1 | 5_prime_flanking_region | c.-30682 | NULL | tier3 | 21 | 0 | 0.000 | 46 | 16 | 0.258 |
| 796518 | 15 | 32929534 | 32929534 | C | A | SNP | ARHGAP11A | ENST00000361627 | 1 | missense | c.2560 | p.Q854K | tier1 | 92 | 0 | 0.000 | 181 | 78 | 0.301 |
| 796518 | 16 | 56919177 | 56919177 | A | G | SNP | SLC12A3 | ENST00000438926 | 1 | missense | c.1826 | p.E609G | tier1 | 370 | 0 | 0.000 | 581 | 264 | 0.312 |
| 796518 | 17 | 986858 | 986858 | G | A | SNP | ABR | ENST00000302538 | -1 | missense | c.541 | p.L181F | tier1 | 39 | 0 | 0.000 | 44 | 22 | 0.328 |
| 796518 | 17 | 51900882 | 51900882 | G | A | SNP | KIF2B | ENST00000268919 | 1 | missense | c.488 | p.R163H | tier1 | 535 | 0 | 0.000 | 794 | 370 | 0.318 |
| 796518 | 18 | 47383269 | 47383269 | A | G | SNP | MYO5B | ENST00000285039 | -1 | splice_region | c.3945-9 | e30-9 | tier3 | 243 | 0 | 0.000 | 461 | 194 | 0.295 |
| 796518 | 20 | 31877714 | 31877714 | T | C | SNP | BPIFB1 | ENST00000253354 | 1 | missense | c.281 | p.I94T | tier1 | 38 | 0 | 0.000 | 89 | 27 | 0.231 |
| 818831 | 1 | 75107026 | 75107026 | G | A | SNP | C1orf173 | ENST00000326665 | -1 | missense | c.433 | p.P145S | tier1 | 70 | 0 | 0.000 | 166 | 37 | 0.182 |
| 818831 | 1 | 181706695 | 181706695 | C | T | SNP | CACNA1E | ENST00000367573 | 1 | missense | c.3457 | p.R1153C | tier1 | 411 | 1 | 0.002 | 799 | 24 | 0.029 |
| 818831 | 2 | 92031582 | 92031582 | G | A | SNP | ABCD1P5 | ENST00000445663 | 1 | 3_prime_untranslated_region | c.*92031582 | NULL | tier3 | 80 | 1 | 0.012 | 164 | 38 | 0.186 |
| 818831 | 2 | 101666881 | 101666881 | G | A | SNP | TBC1D8 | ENST00000409318 | -1 | missense | c.854 | p.P285L | tier1 | 41 | 0 | 0.000 | 74 | 16 | 0.176 |
| 818831 | 4 | 175220362 | 175220362 | G | A | SNP | CEP44 | ENST00000426172 | 1 | splice_site | c.89+1 | e1+1 | tier1 | 41 | 0 | 0.000 | 65 | 15 | 0.188 |
| 818831 | 4 | 185612642 | 185612642 | T | A | SNP | PRIMPOL | ENST00000512658 | 1 | 3_prime_untranslated_region | c.*185612642 | NULL | tier3 | 56 | 1 | 0.018 | 190 | 46 | 0.195 |
| 818831 | 5 | 112926812 | 112926812 | C | A | SNP | YTHDC2 | ENST00000161863 | 1 | missense | c.3900 | p.N1300K | tier1 | 72 | 0 | 0.000 | 138 | 19 | 0.120 |
| 818831 | 6 | 168314069 | 168314069 | C | G | SNP | MLL7 | ENST00000366806 | 1 | intronic | c.2038-779 | e16-779 | tier2 | 55 | 1 | 0.018 | 73 | 28 | 0.277 |
| 818831 | 7 | 74212011 | 74212011 | C | T | SNP | GTF2IRD2 | ENST00000405086 | -1 | missense | c.1840 | p.V614I | tier1 | 99 | 1 | 0.010 | 195 | 29 | 0.130 |
| 818831 | 7 | 100350095 | 100350095 | T | C | SNP | ZAN | ENST00000546292 | 1 | silent | c.2367 | p.I789 | tier1 | 124 | 2 | 0.016 | 243 | 29 | 0.106 |
| 818831 | 12 | 133050449 | 133050450 | - | T | INS | MUC8 | ENST00000595994 | -1 | intronic | c.147+130_147+129 | e1+130 | tier2 | 6 | 0 | 0.000 | 1 | 5 | 0.833 |
| 818831 | 14 | 102445717 | 102445717 | C | A | SNP | DYNC1H1 | ENST00000360184 | 1 | silent | c.406 | p.R136 | tier1 | 72 | 0 | 0.000 | 175 | 23 | 0.116 |
| 818831 | 15 | 74363428 | 74363428 | C | T | SNP | GOLGA6A | ENST00000290438 | -1 | intronic | c.1954+37 | e17+37 | tier3 | 310 | 0 | 0.000 | 580 | 28 | 0.046 |
| 818831 | 15 | 89856067 | 89856067 | T | A | SNP | FANCI | ENST00000310775 | 1 | intronic | c.3652-68 | e34-68 | tier3 | 52 | 0 | 0.000 | 74 | 12 | 0.140 |
| 818831 | 15 | 90631934 | 90631934 | C | T | SNP | IDH2 | ENST00000330062 | -1 | missense | c.419 | p.R140Q | tier1 | 453 | 4 | 0.009 | 780 | 128 | 0.141 |
| 818831 | 17 | 18334137 | 18334137 | G | G | SNP | KRT17P2 | ENST00000581146 | 1 | intronic | c.NULL | NULL | tier3 | 58 | 1 | 0.0 | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-------|-----|---------|-----------------|-----------------|-----------------------------|-----------------------------|--------------|----------|-------|-----|-------|-------|------|-------|-------|
| 819544 | 2 | 236480647 | 236480647 | C | T | SNP | ENSG00000221704 | ENST00000408777 | 1 | rna | NULL | NULL | tier1 | 69 | 0 | 0.000 | 186 | 20 | 0.097 |
| 819544 | 5 | 41312207 | 41312207 | A | T | SNP | PLCXD3 | ENST00000328457 | -1 | 3_prime_untranslated_region | c.*1512 | NULL | tier3 | 100 | 1 | 0.010 | 261 | 32 | 0.109 |
| 819544 | 5 | 176940748 | 176940748 | C | A | SNP | DDX41 | ENST00000507955 | -1 | missense | c.1036 | p.A346P | tier1 | 315 | 0 | 0.000 | 593 | 28 | 0.045 |
| 819544 | 6 | 130504496 | 130504496 | G | A | SNP | SAMD3 | ENST00000524930 | -1 | 3_prime_untranslated_region | c.*31139 | NULL | tier4 | 33 | 0 | 0.000 | 95 | 26 | 0.213 |
| 819544 | 8 | 110457215 | 110457215 | A | G | SNP | PKHD1L1 | ENST00000378402 | 1 | missense | c.5117 | p.N1706S | tier1 | 500 | 1 | 0.002 | 1136 | 34 | 0.029 |
| 819544 | 12 | 50037698 | 50037698 | G | T | SNP | PRPF40B | ENST00000548825 | 1 | nonsense | c.2497 | p.E833* | tier1 | 498 | 0 | 0.000 | 1072 | 29 | 0.026 |
| 819544 | 15 | 90229787 | 90229787 | A | - | DEL | PEX11A | ENST00000300056 | -1 | splice_region_del | c.57-10 | e2-10 | tier3 | 45 | 0 | 0.000 | 69 | 12 | 0.148 |
| 819544 | 17 | 30321557 | 30321557 | T | - | DEL | SUZ12 | ENST00000578106 | 1 | 3_prime_untranslated_region | c.*30321557 | NULL | tier3 | 181 | 4 | 0.022 | 496 | 58 | 0.105 |
| 819544 | 17 | 36358189 | 36358189 | G | T | SNP | LOC101929950 | ENST00000312412 | -1 | intronic | c.947-31 | e8-31 | tier4 | 197 | 2 | 0.010 | 604 | 42 | 0.065 |
| 829970 | 1 | 151238730 | 151238730 | G | T | SNP | PSMD4 | ENST00000476855 | 1 | 3_prime_untranslated_region | c.*151238730 | NULL | tier3 | 23 | 0 | 0.000 | 145 | 116 | 0.441 |
| 829970 | 2 | 1665807 | 1665807 | T | C | SNP | PXDN | ENST00000252804 | -1 | intronic | c.1680+37 | e13+37 | tier2 | 20 | 0 | 0.000 | 172 | 124 | 0.418 |
| 829970 | 2 | 21238083 | 21238083 | G | A | SNP | APOB | ENST00000233242 | -1 | silent | c.3558 | p.T1186 | tier1 | 222 | 0 | 0.000 | 361 | 277 | 0.434 |
| 829970 | 2 | 95944840 | 95944840 | G | T | SNP | PROM2 | ENST00000317620 | 1 | nonsense | c.1222 | p.E408* | tier1 | 13 | 0 | 0.000 | 114 | 81 | 0.415 |
| 829970 | 2 | 105442572 | 105442572 | C | G | SNP | LOC100506421 | ENST00000413121 | -1 | intronic | c.NULL | NULL | tier3 | 66 | 0 | 0.000 | 132 | 126 | 0.487 |
| 829970 | 3 | 38151817 | 38151817 | G | C | SNP | DLEC1 | ENST00000346219 | 1 | intronic | c.3443+45 | e23+45 | tier3 | 38 | 0 | 0.000 | 123 | 92 | 0.426 |
| 829970 | 3 | 56766462 | 56766462 | A | G | SNP | ARHGEF3 | ENST00000338458 | -1 | splice_region | c.1138-10 | e11-10 | tier3 | 87 | 0 | 0.000 | 161 | 131 | 0.447 |
| 829970 | 3 | 101477008 | 101477008 | A | T | SNP | CEP97 | ENST00000327230 | 1 | missense | c.1558 | p.N520Y | tier1 | 60 | 0 | 0.000 | 79 | 83 | 0.512 |
| 829970 | 3 | 180320681 | 180320681 | AAGAG | - | DEL | TTC14 | ENST00000296015 | 1 | frame_shift_del | c.164_168 | p.K57fs | tier1 | 109 | 0 | 0.000 | 181 | 91 | 0.335 |
| 829970 | 3 | 182910694 | 182910694 | G | C | SNP | MCF2L2 | ENST00000488149 | -1 | 5_prime_untranslated_region | c.-182910694 | NULL | tier3 | 26 | 0 | 0.000 | 91 | 86 | 0.486 |
| 829970 | 4 | 5857824 | 5857824 | A | T | SNP | CRMP1 | ENST00000324989 | -1 | intronic | c.820+46 | e4+46 | tier3 | 42 | 0 | 0.000 | 88 | 86 | 0.494 |
| 829970 | 4 | 15839802 | 15839802 | T | A | SNP | CD38 | ENST00000226279 | 1 | intronic | c.659+14 | e5+14 | tier3 | 107 | 0 | 0.000 | 105 | 82 | 0.439 |
| 829970 | 4 | 70255405 | 70255405 | C | A | SNP | ENSG00000215110 | ENST00000511504 | 1 | 3_prime_untranslated_region | c.*70255405 | NULL | tier3 | 173 | 0 | 0.000 | 298 | 219 | 0.423 |
| 829970 | 5 | 7303612 | 7303612 | G | A | SNP | LOC442132 | ENST00000512854 | -1 | intronic | c.NULL | NULL | tier3 | 61 | 0 | 0.000 | 259 | 193 | 0.427 |
| 829970 | 5 | 149439406 | 149439406 | C | T | SNP | CSF1R | ENST00000286301 | -1 | silent | c.1989 | p.T663 | tier1 | 57 | 0 | 0.000 | 26 | 214 | 0.892 |
| 829970 | 6 | 66803321 | 66803321 | C | T | SNP | NUFIP1P | ENST00000402902 | -1 | 3_prime_flanking_region | c.*3 | NULL | tier3 | 211 | 0 | 0.000 | 346 | 65 | 0.158 |
| 829970 | 6 | 125231919 | 125231919 | T | G | SNP | STL | ENST00000439075 | -1 | 5_prime_untranslated_region | c.-125231919 | NULL | tier3 | 234 | 0 | 0.000 | 498 | 20 | 0.039 |
| 829970 | 7 | 128292569 | 128292569 | C | T | SNP | FAM71F2 | ENST00000480462 | 1 | 5_prime_flanking_region | c.-19883 | NULL | tier3 | 11 | 0 | 0.000 | 3 | 21 | 0.840 |
| 829970 | 7 | 138126219 | 138126219 | C | T | SNP | ENSG00000214815 | ENST00000399037 | 1 | 3_prime_untranslated_region | c.*138126219 | NULL | tier2 | 54 | 1 | 0.018 | 211 | 44 | 0.173 |
| 829970 | 8 | 73125567 | 73125567 | A | - | DEL | ENSG00000250979 | ENST00000503430 | -1 | 5_prime_untranslated_region | c.-73125567 | NULL | tier2 | 148 | 0 | 0.000 | 381 | 150 | 0.283 |
| 829970 | 8 | 73125568 | 73125568 | C | T | SNP | ENSG00000250979 | ENST00000503430 | -1 | 5_prime_untranslated_region | c.-73125568 | NULL | tier2 | 147 | 1 | 0.007 | 361 | 164 | 0.311 |
| 829970 | 9 | 100122246 | 100122246 | A | - | DEL | C9orf174 | ENST00000375202 | 1 | intronic | c.3496-18 | e26-18 | tier3 | 155 | 0 | 0.000 | 417 | 285 | 0.406 |
| 829970 | 10 | 82279156 | 82279156 | G | SNP | TSPAN14 | ENST00000429989 | 1 | 3_prime_untranslated_region | c.*1424 | NULL | tier3 | 72 | 0 | 0.000 | 136 | 137 | 0.502 | |
| 829970 | 10 | 127477858 | 127477858 | G | T | SNP | UROS | ENST00000465577 | -1 | 5_prime_untranslated_region | c.-127477858 | NULL | tier3 | 30 | 0 | 0.000 | 111 | 116 | 0.511 |
| 829970 | 11 | 49897871 | 49897871 | C | T | SNP | ENSG00000255190 | ENST00000529746 | 1 | 3_prime_untranslated_region | c.*49897871 | NULL | tier3 | 323 | 0 | 0.000 | 538 | 414 | 0.434 |
| 829970 | 12 | 5603366 | 5603366 | C | A | SNP | NTF3 | ENST00000423158 | 1 | missense | c.25 | p.Q9K | tier1 | 36 | 0 | 0.000 | 68 | 57 | 0.456 |
| 829970 | 12 | 18760087 | 18760087 | T | G | SNP | PIK3C2G | ENST00000538779 | 1 | intronic | c.4088-2382 | e30-2382 | tier2 | 88 | 0 | 0.000 | 191 | 33 | 0.147 |
| 829970 | 12 | 27919782 | 27919782 | C | A | SNP | MANSC4 | ENST00000381273 | -1 | intronic | c.230-50 | e2-50 | tier2 | 58 | 0 | 0.000 | 125 | 99 | 0.440 |
| 829970 | 13 | 99446895 | 99446895 | C | T | SNP | DOCK9 | ENST00000400228 | -1 | missense | c.1993 | p.V665I | tier1 | 60 | 0 | 0.000 | 104 | 119 | 0.531 |
| 829970 | 14 | 60577741 | 60577741 | A | G | SNP | C14orf135 | ENST00000391611 | 1 | 3_prime_untranslated_region | c.*2692 | NULL | tier4 | 45 | 0 | 0.000 | 82 | 81 | 0.494 |
| 829970 | 14 | 90650882 | 90650882 | A | C | SNP | KCNK13 | ENST00000282146 | 1 | missense | c.762 | p.Q254H | tier1 | 99 | 1 | 0.010 | 755 | 120 | 0.137 |
| 829970 | 15 | 24926165 | 24926165 | A | T | SNP | C15orf2 | ENST00000329468 | 1 | 3_prime_untranslated_region | c.*1680 | NULL | tier4 | 49 | 0 | 0.000 | 74 | 48 | 0.393 |
| 829970 | 15 | 58007312 | 58007312 | A | T | SNP | POLR2M | ENST00000299638 | 1 | 3_prime_untranslated_region | c.*435 | NULL | tier3 | 87 | 0 | 0.000 | 190 | 31 | 0.140 |
| 829970 | 16 | 72829450 | 72829450 | A | G | SNP | ZFH3 | ENST00000268489 | -1 | silent | c.7131 | p.P2377 | tier1 | 28 | 0 | 0.000 | 105 | 91 | 0.464 |
| 829970 | 17 | 263645 | 263645 | C | T | SNP | C17orf97 | ENST00000360127 | 1 | silent | c.1011 | p.P337 | tier1 | 129 | 2 | 0.015 | 447 | 82 | 0.154 |
| 829970 | 17 | 7577094 | 7577094 | G | A | SNP | TP53 | ENST00000269305 | -1 | missense | c.844 | p.R282W | tier1 | 117 | 1 | 0.008 | 116 | 696 | 0.857 |
| 829970 | 17 | 61995243 | 61995243 | C | T | SNP | GH1 | ENST00000323322 | -1 | silent | c.333 | p.S111 | tier1 | 39 | 0 | 0.000 | 213 | 212 | 0.499 |
| 829970 | 19 | 17763404 | 17763404 | A | T | SNP | UNC13A | ENST00000428389 | -1 | intronic | c.1703+36 | e13+36 | tier3 | 38 | 0 | 0.000 | 83 | 70 | 0.458 |
| 829970 | 19 | 18378165 | 18378165 | T | G | SNP | KIAA1683 | ENST00000392413 | -1 | missense | c.185 | p.K62T | tier1 | 106 | 0 | 0.000 | 751 | 111 | 0.129 |
| 829970 | 19 | 39214520 | 39214520 | G | C | SNP | ACTN4 | ENST00000252699 | 1 | intronic | c.1552-57 | e14-57 | tier3 | 36 | 0 | 0.000 | 174 | 127 | 0.421 |
| 829970 | 19 | 53793213 | 53793213 | A | T | SNP | BIRC8 | ENST00000426466 | -1 | missense | c.415 | p.S139T | tier1 | 64 | 0 | 0.000 | 87 | 69 | 0.442 |
| 829970 | 20 | 30785389 | 30785389 | G | C | SNP | PLAGL2 | ENST00000246229 | -1 | silent | c.357 | p.T119 | tier1 | 16 | 0 | 0.000 | 125 | 82 | 0.394 |
| 829970 | 21 | 31655211 | 31655211 | C | G | SNP | KRTAP24-1 | ENST00000340345 | -1 | missense | c.40 | p.V14L | tier1 | 32 | 0 | 0.000 | 58 | 41 | 0.414 |
| 829970 | 21 | 42694456 | 42694456 | G | A | SNP | FAM3B | ENST00000357985 | 1 | intronic | c.20-394 | e2-394 | tier4 | 57 | 0 | 0.000 | 121 | 106 | 0.465 |
| 829970 | 21 | 45877697 | 45877697 | G | A | SNP | LRRC3 | ENST00000291592 | 1 | 3_prime_untranslated_region | c.*396 | NULL | tier3 | 23 | 0 | 0.000 | 83 | 80 | 0.491 |
| 829970 | 22 | 33255649 | 33255649 | T | C | SNP | TIMP3 | ENST00000266085 | 1 | 3_prime_untranslated_region | c.*285 | NULL | tier3 | 196 | 0 | 0.000 | 220 | 112 | 0.337 |
| 839903 | 1 | 82436028 | 82436028 | C | T | SNP | LPHN2 | ENST00000370717 | 1 | missense | c.2752 | p.H918Y | tier1 | 90 | 0 | 0.000 | 85 | 67 | 0.441 |
| 839903 | 1 | 115258747 | 115258747 | C | T | SNP | NRAS | ENST00000369535 | -1 | missense | c.35 | p.G12D | tier1 | 390 | 1 | 0.003 | 588 | 83 | 0.124 |
| 839903 | 1 | 144619315 | 144619315 | G | T | SNP | ENSG00000225241 | ENST00000421407 | 1 | intronic | c.NULL | NULL | tier3 | 190 | 1 | 0.005 | 286 | 67 | 0.190 |
| 839903 | 1 | 177927426 | 177927426 | C | T | SNP | SEC16B | ENST00000527976 | -1 | missense | c.136 | p.A46T | tier1 | 72 | 0 | 0.000 | 43 | 40 | 0.482 |
| 839903 | 1 | 222705451 | 222705451 | C | A | SNP | HHIPL2 | ENST00000343410 | -1 | missense | c.1580 | p.R527L | tier1 | 57 | 1 | 0.017 | 35 | 45 | 0.563 |
| 839903 | 1 | 223568401 | 223568401 | G | A | SNP | C1orf65 | ENST00000366875 | 1 | silent | c.1584 | p.K528 | tier1 | 44 | 0 | 0.000 | 43 | 39 | 0.476 |
| 839903 | 2 | 26677567 | 26677567 | G | T | SNP | DRC1 | ENST00000288710 | 1 | missense | c.1972 | p.D658Y | tier1 | 74 | 0 | 0.000 | 42 | 48 | 0.533 |
| 839903 | 2 | 44203321 | 44203321 | G | A | SNP | LRPPRC | ENST00000260665 | -1 | missense | c.698 | p.A233V | tier1 | 44 | 0 | 0.000 | 110 | 23 | 0.172 |
| 839903 | 2 | 97784048 | 97784048 | G | A | SNP | ANKRD36 | ENST00000420699 | 1 | intronic | c.313-33 | e3-33 | tier3 | 205 | 3 | 0.014 | 322 | 25 | 0.072 |
| 839903 | 2 | 97817745 | 97817745 | G | T | SNP | ANKRD36 | ENST00000420699 | 1 | intronic | c.1162+69 | e13+69 | tier4 | 29 | 0 | 0.000 | 56 | 14 | 0.200 |
| 839903 | 2 | 187367384 | 187367384 | G | A | SNP | ZC3H15 | ENST00000337859 | 1 | intronic | c.534+70 | e5+70 | tier3 | 53 | 0 | 0.000 | 100 | 41 | 0.289 |
| 839903 | 3 | 16217151 | 16217151 | G | T | SNP | GALNT15 | ENST00000339732 | 1 | missense | c.493 | p.A165S | tier1 | 104 | 0 | 0.000 | 69 | 45 | 0.395 |
| 839903 | 3 | 57447266 | 57447266 | G | A | SNP | DNAH12 | ENST00000351747 | -1 | nonsense | c.2617 | p.Q873* | tier1 | 49 | 0 | 0.000 | 93 | 70 | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|----|-----|----------------|------------------------|-----------------|----------|-----------------------------|-----------------|-----------|-------|-----|-------|-------|-----|-------|-------|
| 839903 | 5 | 140203552 | 140203552 | G | T | SNP | <i>PCDHA5</i> | ENST00000529859 | 1 | missense | c.2192 | p.C731F | tier1 | 267 | 2 | 0.007 | 237 | 208 | 0.466 |
| 839903 | 6 | 30229312 | 30229312 | G | T | SNP | <i>HCG17</i> | ENST00000453558 | -1 | intronic | c.NULL | NULL | tier3 | 127 | 0 | 0.000 | 82 | 67 | 0.450 |
| 839903 | 7 | 151082221 | 151082221 | G | A | SNP | <i>WDR86</i> | ENST00000334493 | -1 | missense | c.815 | p.T272M | tier1 | 66 | 0 | 0.000 | 84 | 16 | 0.160 |
| 839903 | 8 | 1649371 | 1649371 | G | A | SNP | <i>DLGAP2</i> | ENST00000520901 | 1 | missense | c.2735 | p.R912H | tier1 | 182 | 0 | 0.000 | 118 | 108 | 0.478 |
| 839903 | 8 | 65496059 | 65496059 | C | T | SNP | <i>BHLHE22</i> | ENST00000321870 | 1 | 3_prime_untranslated_region | c.*1566 | NULL | tier2 | 106 | 0 | 0.000 | 156 | 35 | 0.182 |
| 839903 | 9 | 32624147 | 32624147 | T | A | SNP | <i>ENSG00000223440</i> | ENST00000430787 | 1 | 5_prime_flanking_region | c.-9305 | NULL | tier4 | 55 | 0 | 0.000 | 66 | 90 | 0.577 |
| 839903 | 10 | 5494360 | 5494360 | G | A | SNP | <i>NET1</i> | ENST00000355029 | 1 | missense | c.403 | p.A135T | tier1 | 100 | 2 | 0.020 | 73 | 54 | 0.425 |
| 839903 | 10 | 49220302 | 49220302 | G | A | SNP | <i>CTGLF12P</i> | ENST00000339771 | -1 | 5_prime_untranslated_region | c.-49220302 | NULL | tier3 | 56 | 0 | 0.000 | 99 | 53 | 0.349 |
| 839903 | 10 | 90582658 | 90582658 | G | A | SNP | <i>ANKRD22</i> | ENST00000476963 | -1 | rna | NULL | NULL | tier1 | 96 | 0 | 0.000 | 96 | 79 | 0.451 |
| 839903 | 10 | 112356174 | 112356174 | G | C | SNP | <i>SMC3</i> | ENST00000361804 | 1 | missense | c.1982 | p.R661P | tier1 | 392 | 1 | 0.003 | 687 | 160 | 0.189 |
| 839903 | 10 | 117486773 | 117486773 | C | T | SNP | <i>ATRNL1</i> | ENST00000355044 | 1 | nonsense | c.3811 | p.R1271* | tier1 | 165 | 0 | 0.000 | 177 | 12 | 0.064 |
| 839903 | 10 | 131634749 | 131634749 | C | T | SNP | <i>EBF3</i> | ENST00000368648 | -1 | 3_prime_untranslated_region | c.*1444 | NULL | tier3 | 345 | 0 | 0.000 | 314 | 86 | 0.215 |
| 839903 | 11 | 1083328 | 1083328 | C | A | SNP | <i>MUC2</i> | ENST00000441003 | 1 | splice_region | c.2219+9 | e16+9 | tier2 | 60 | 0 | 0.000 | 77 | 38 | 0.330 |
| 839903 | 11 | 46334349 | 46334349 | C | T | SNP | <i>CREB3L1</i> | ENST00000288400 | 1 | intronic | c.963-53 | e8-53 | tier3 | 52 | 0 | 0.000 | 49 | 37 | 0.430 |
| 839903 | 11 | 55322753 | 55322753 | C | T | SNP | <i>OR4C15</i> | ENST00000314644 | 1 | missense | c.971 | p.A324V | tier1 | 57 | 0 | 0.000 | 134 | 38 | 0.221 |
| 839903 | 11 | 63997883 | 63997883 | T | - | DEL | <i>DNAJC4</i> | ENST00000321685 | 1 | intronic | c.1-21 | e1-21 | tier2 | 17 | 0 | 0.000 | 21 | 11 | 0.344 |
| 839903 | 11 | 107219570 | 107219570 | T | G | SNP | <i>CWF19L2</i> | ENST00000462890 | -1 | 5_prime_untranslated_region | c.-107219570 | NULL | tier3 | 79 | 0 | 0.000 | 85 | 71 | 0.455 |
| 839903 | 12 | 43945607 | 43945607 | G | T | SNP | <i>ADAMTS20</i> | ENST00000389420 | -1 | intronic | c.91+27 | e1+27 | tier2 | 134 | 0 | 0.000 | 86 | 63 | 0.417 |
| 839903 | 12 | 48360948 | 48360948 | A | - | DEL | <i>TMEM106C</i> | ENST00000429772 | 1 | intronic | c.603-43 | e6-43 | tier3 | 57 | 0 | 0.000 | 56 | 9 | 0.139 |
| 839903 | 12 | 49725038 | 49725038 | C | T | SNP | <i>TROAP</i> | ENST00000257909 | 1 | missense | c.2140 | p.R714C | tier1 | 74 | 0 | 0.000 | 73 | 67 | 0.479 |
| 839903 | 13 | 36401953 | 36401953 | T | G | SNP | <i>DCLK1</i> | ENST00000360631 | -1 | intronic | c.1288-62 | e9-62 | tier3 | 208 | 0 | 0.000 | 201 | 164 | 0.448 |
| 839903 | 14 | 76211445 | 76211445 | G | A | SNP | <i>TTL5</i> | ENST00000298832 | 1 | missense | c.1289 | p.R430H | tier1 | 124 | 1 | 0.008 | 190 | 20 | 0.095 |
| 839903 | 14 | 106135124 | 106135124 | C | T | SNP | <i>IGHGP</i> | ENST00000390555 | -1 | 5_prime_untranslated_region | c.-106135124 | NULL | tier3 | 170 | 0 | 0.000 | 272 | 32 | 0.105 |
| 839903 | 15 | 57999148 | 57999148 | C | T | SNP | <i>POLR2M</i> | ENST00000299638 | 1 | silent | c.108 | p.R36 | tier1 | 113 | 0 | 0.000 | 134 | 21 | 0.136 |
| 839903 | 16 | 29851667 | 29851667 | C | T | SNP | <i>MVP</i> | ENST00000357402 | 1 | missense | c.1078 | p.R360C | tier1 | 31 | 1 | 0.031 | 48 | 41 | 0.461 |
| 839903 | 16 | 30722141 | 30722141 | G | T | SNP | <i>SRCAP</i> | ENST00000262518 | 1 | nonsense | c.1201 | p.E401* | tier1 | 56 | 0 | 0.000 | 39 | 36 | 0.480 |
| 839903 | 16 | 87160454 | 87160454 | A | G | SNP | <i>C16orf95</i> | ENST00000562840 | -1 | intronic | c.NULL | NULL | tier3 | 77 | 0 | 0.000 | 104 | 21 | 0.168 |
| 839903 | 17 | 7010496 | 7010496 | G | T | SNP | <i>ASGR2</i> | ENST00000355035 | -1 | intronic | c.512-26 | e6-26 | tier3 | 58 | 1 | 0.017 | 60 | 71 | 0.542 |
| 839903 | 17 | 39041424 | 39041424 | C | T | SNP | <i>KRT20</i> | ENST00000167588 | -1 | missense | c.14 | p.R5H | tier1 | 82 | 2 | 0.024 | 68 | 69 | 0.504 |
| 839903 | 18 | 21452994 | 21452994 | C | T | SNP | <i>LAMA3</i> | ENST00000269217 | 1 | 5_prime_untranslated_region | c.-15 | NULL | tier3 | 50 | 0 | 0.000 | 48 | 41 | 0.461 |
| 839903 | 19 | 13136234 | 13136234 | G | T | SNP | <i>NFIX</i> | ENST00000585575 | 1 | nonsense | c.403 | p.E135* | tier1 | 114 | 0 | 0.000 | 143 | 44 | 0.233 |
| 839903 | 19 | 42219296 | 42219296 | G | A | SNP | <i>CEACAM5</i> | ENST00000596606 | 1 | 3_prime_untranslated_region | c.*42219296 | NULL | tier3 | 42 | 0 | 0.000 | 24 | 26 | 0.520 |
| 839903 | 20 | 16349047 | 16349047 | C | G | SNP | <i>KIF16B</i> | ENST00000408042 | -1 | intronic | c.3499-576 | e23-576 | tier3 | 139 | 0 | 0.000 | 77 | 70 | 0.470 |
| 839903 | X | 48926088 | 48926088 | C | T | SNP | <i>CCDC120</i> | ENST00000603986 | 1 | missense | c.1985 | p.A662V | tier1 | 96 | 2 | 0.020 | 67 | 53 | 0.442 |
| 839903 | X | 77222318 | 77222318 | C | T | SNP | <i>ATP7A</i> | ENST00000341514 | 1 | intronic | c.1-4800 | e1-4800 | tier4 | 38 | 0 | 0.000 | 49 | 32 | 0.395 |
| 839903 | X | 153631127 | 153631127 | G | A | SNP | <i>DNASE1L1</i> | ENST0000014935 | -1 | missense | c.830 | p.A277V | tier1 | 158 | 0 | 0.000 | 316 | 17 | 0.051 |
| 848524 | 1 | 99164247 | 99164247 | T | - | DEL | <i>SNX7</i> | ENST00000306121 | 1 | intronic | c.839-15 | e6-15 | tier3 | 46 | 0 | 0.000 | 93 | 15 | 0.139 |
| 848524 | 1 | 150432709 | 150432709 | C | T | SNP | <i>RPRD2</i> | ENST00000369068 | 1 | silent | c.1327 | p.L443 | tier1 | 59 | 0 | 0.000 | 82 | 12 | 0.128 |
| 848524 | 1 | 155580877 | 155580877 | T | T | SNP | <i>MSTO1</i> | ENST00000245564 | 1 | nonsense | c.268 | p.Q90* | tier1 | 18 | 0 | 0.000 | 20 | 16 | 0.444 |
| 848524 | 1 | 168161365 | 168161365 | C | T | SNP | <i>TIPRL</i> | ENST00000367833 | 1 | intronic | c.516+627 | e4+627 | tier3 | 55 | 3 | 0.052 | 70 | 53 | 0.427 |
| 848524 | 1 | 177898245 | 177898245 | A | - | DEL | <i>ENSG00000254154</i> | ENST00000354921 | -1 | rna | NULL | NULL | tier1 | 81 | 2 | 0.024 | 76 | 43 | 0.361 |
| 848524 | 1 | 243419492 | 243419492 | A | SNP | <i>SDCCAG8</i> | ENST00000366541 | 1 | missense | c.17 | p.E6G | tier1 | 98 | 0 | 0.000 | 106 | 87 | 0.451 | |
| 848524 | 2 | 237103618 | 237103618 | C | T | SNP | <i>ASB18</i> | ENST00000409749 | -1 | missense | c.1298 | p.R433H | tier1 | 55 | 2 | 0.035 | 46 | 50 | 0.521 |
| 848524 | 3 | 50138116 | 50138116 | G | A | SNP | <i>RBM5</i> | ENST00000469838 | 1 | 3_prime_untranslated_region | c.*609 | NULL | tier3 | 52 | 0 | 0.000 | 62 | 47 | 0.431 |
| 848524 | 3 | 57611690 | 57611690 | A | G | SNP | <i>DENND6A</i> | ENST00000311128 | -1 | 3_prime_untranslated_region | c.*2251 | NULL | tier2 | 116 | 2 | 0.017 | 236 | 196 | 0.454 |
| 848524 | 4 | 1803182 | 1803182 | C | A | SNP | <i>FGFR3</i> | ENST00000340107 | 1 | silent | c.534 | p.A178 | tier1 | 19 | 0 | 0.000 | 19 | 24 | 0.558 |
| 848524 | 4 | 8234226 | 8234226 | G | A | SNP | <i>SH3TC1</i> | ENST00000502350 | 1 | 3_prime_untranslated_region | c.*8234226 | NULL | tier2 | 57 | 2 | 0.034 | 58 | 46 | 0.442 |
| 848524 | 4 | 54324769 | 54324769 | T | C | SNP | <i>FIP1L1</i> | ENST00000507206 | 1 | 3_prime_untranslated_region | c.*54324769 | NULL | tier3 | 35 | 1 | 0.028 | 57 | 46 | 0.447 |
| 848524 | 4 | 67142574 | 67142574 | T | A | SNP | <i>MIR1269A</i> | ENST00000408636 | 1 | rna | NULL | NULL | tier1 | 65 | 0 | 0.000 | 61 | 39 | 0.390 |
| 848524 | 4 | 165187407 | 165187407 | T | C | SNP | <i>01.Mär</i> | ENST00000503008 | -1 | intronic | c.1+116932 | e0+116932 | tier4 | 232 | 3 | 0.013 | 318 | 108 | 0.254 |
| 848524 | 5 | 80064748 | 80064748 | C | G | SNP | <i>MSH3</i> | ENST00000265081 | 1 | missense | c.2179 | p.R727G | tier1 | 161 | 0 | 0.000 | 280 | 29 | 0.094 |
| 848524 | 5 | 87502321 | 87502321 | G | - | DEL | <i>TMEM161B</i> | ENST00000296595 | -1 | splice_region_del | c.599-5 | e7-5 | tier3 | 168 | 2 | 0.012 | 315 | 25 | 0.074 |
| 848524 | 5 | 140221091 | 140221091 | G | T | SNP | <i>PCDHA8</i> | ENST00000531613 | 1 | missense | c.185 | p.R62L | tier1 | 548 | 12 | 0.021 | 577 | 361 | 0.384 |
| 848524 | 5 | 176016454 | 176016454 | C | A | SNP | <i>CDHR2</i> | ENST00000508085 | 1 | 3_prime_untranslated_region | c.*176016454 | NULL | tier3 | 47 | 1 | 0.021 | 42 | 43 | 0.506 |
| 848524 | 6 | 88757740 | 88757740 | C | T | SNP | <i>SPACA1</i> | ENST00000237201 | 1 | silent | c.117 | p.A39 | tier1 | 51 | 2 | 0.038 | 45 | 37 | 0.451 |
| 848524 | 7 | 27210530 | 27210530 | G | A | SNP | <i>HOXA10</i> | ENST00000283921 | -1 | 3_prime_untranslated_region | c.*988 | NULL | tier2 | 244 | 1 | 0.004 | 269 | 15 | 0.053 |
| 848524 | 7 | 53103572 | 53103572 | C | T | SNP | <i>POM121L12</i> | ENST00000408890 | 1 | missense | c.208 | p.R70C | tier1 | 11 | 0 | 0.000 | 9 | 15 | 0.625 |
| 848524 | 7 | 142353940 | 142353941 | TG | - | DEL | <i>TRBV23-1</i> | ENST00000390396 | 1 | frame_shift_del | c.323_324 | p.Y109fs | tier1 | 62 | 1 | 0.016 | 36 | 46 | 0.561 |
| 848524 | 8 | 11189142 | 11189142 | G | A | SNP | <i>SLC35G5</i> | ENST00000382435 | 1 | missense | c.527 | p.G176E | tier1 | 110 | 1 | 0.009 | 128 | 14 | 0.099 |
| 848524 | 8 | 51449330 | 51449330 | G | A | SNP | <i>SNTG1</i> | ENST00000518864 | 1 | silent | c.642 | p.S214 | tier1 | 50 | 0 | 0.000 | 34 | 40 | 0.541 |
| 848524 | 8 | 92982935 | 92982935 | G | A | SNP | <i>RUNX1T1</i> | ENST00000436581 | -1 | missense | c.1523 | p.A508V | tier1 | 437 | 13 | 0.029 | 449 | 398 | 0.469 |
| 848524 | 8 | 144800290 | 144800290 | C | G | SNP | <i>MAPK15</i> | ENST00000475376 | 1 | 3_prime_untranslated_region | c.*144800290 | NULL | tier3 | 79 | 2 | 0.025 | 58 | 48 | 0.453 |
| 848524 | 9 | 98242628 | 98242628 | T | C | SNP | <i>PTCH1</i> | ENST00000331920 | -1 | intronic | c.945+44 | e6+44 | tier3 | 409 | 6 | 0.014 | 628 | 102 | 0.140 |
| 848524 | 10 | 75560546 | 75560546 | C | - | DEL | <i>ZSWIM8</i> | ENST00000603195 | 1 | rna | NULL | NULL | tier1 | 124 | 2 | 0.016 | 182 | 20 | 0.099 |
| 848524 | 11 | 69631012 | 69631012 | C | A | SNP | <i>FGF3</i> | ENST00000334134 | -1 | intronic | c.324+76 | e2+76 | tier3 | 29 | 1 | 0.033 | 25 | 28 | 0.528 |
| 848524 | 11 | 78601098 | 78601098 | G | A | SNP | <i>TENM4</i> | ENST00000278550 | -1 | intronic | c.849-33 | e5-33 | tier3 | 49 | 0 | 0.000 | 17 | 27 | 0.614 |
| 848524 | 11 | 102395831 | 102395832 | - | T | INS | <i>MMP7</i> | ENST00000260227 | -1 | intronic | c.485-37_485-36 | e4-36 | tier3 | 79 | 0 | 0.000 | 101 | 12 | 0.106 |
| 848524 | 12 | 34 | | | | | | | | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-----------------------------|---|-----|-----------------|-----------------|----|-----------------------------|---------------------|------------------------|-------|------|----|-------|------|-----|-------|
| 848524 | 18 | 66344112 | 66344115 | AATA | - | DEL | TMX3 | ENST00000578816 | -1 | 5_prime_untranslated_region | c.-66344112 | NULL | tier3 | 66 | 3 | 0.044 | 70 | 40 | 0.364 |
| 848524 | 19 | 1043430 | 1043430 | C | G | SNP | ABCA7 | ENST00000263094 | 1 | silent | c.888 | p.L296 | tier1 | 58 | 2 | 0.033 | 66 | 55 | 0.455 |
| 848524 | 19 | 37064363 | 37064363 | A | T | SNP | LOC101927599 | ENST00000448373 | 1 | rna | NULL | NULL | tier1 | 111 | 0 | 0.000 | 145 | 122 | 0.457 |
| 848524 | 19 | 56173950 | 56173964 | AGATTAAACCAGGACA | - | DEL | U2AF2 | ENST00000308924 | 1 | in_frame_del | c.569_583 | p.INQDK191in_frame_del | tier1 | 514 | 3 | 0.006 | 497 | 89 | 0.152 |
| 848524 | 20 | 31022403 | 31022425 | CACCACTGCCATAGA GAGGCGGC | - | DEL | ASXL1 | ENST00000375687 | 1 | frame_shift_del | c.1888_1910 | p.H633fs | tier1 | 652 | 5 | 0.008 | 914 | 129 | 0.124 |
| 848524 | 20 | 31022417 | 31022417 | A | - | DEL | ASXL1 | ENST00000375687 | 1 | frame_shift_del | c.1902 | p.E635fs | tier1 | 640 | 3 | 0.005 | 740 | 91 | 0.110 |
| 848524 | 21 | 14424204 | 14424204 | C | T | SNP | ANKRD30BP2 | ENST00000471407 | 1 | rna | NULL | NULL | tier1 | 407 | 6 | 0.015 | 562 | 151 | 0.211 |
| 855934 | 1 | 228403429 | 228403429 | G | A | SNP | OBSCN | ENST00000422127 | 1 | missense | c.1994 | p.R665Q | tier1 | 100 | 7 | 0.065 | 144 | 111 | 0.432 |
| 855934 | 2 | 81421390 | 81421390 | G | A | SNP | ENSG00000234429 | ENST00000378334 | -1 | 3_prime_flanking_region | c.*71 | NULL | tier2 | 99 | 0 | 0.000 | 102 | 87 | 0.456 |
| 855934 | 2 | 166611398 | 166611398 | G | T | SNP | GALNT3 | ENST00000392701 | -1 | intronic | c.1524+44 | e7+44 | tier3 | 117 | 0 | 0.000 | 94 | 73 | 0.437 |
| 855934 | 2 | 172231602 | 172231602 | C | T | SNP | ENSG00000224553 | ENST00000428525 | -1 | 5_prime_untranslated_region | c.-172231602 | NULL | tier3 | 68 | 0 | 0.000 | 105 | 103 | 0.495 |
| 855934 | 3 | 133087476 | 133087477 | CA | - | DEL | TMEM108 | ENST00000321871 | 1 | intronic | c.41-11120_41-11119 | e2-11119 | tier3 | 55 | 0 | 0.000 | 74 | 11 | 0.129 |
| 855934 | 4 | 106157845 | 106157845 | C | T | SNP | TET2 | ENST00000380013 | 1 | nonsense | c.2746 | p.Q916* | tier1 | 50 | 3 | 0.057 | 49 | 40 | 0.449 |
| 855934 | 5 | 140783754 | 140783754 | G | A | SNP | PCDHGA9 | ENST00000573521 | 1 | missense | c.1235 | p.R412Q | tier1 | 44 | 0 | 0.000 | 56 | 60 | 0.517 |
| 855934 | 6 | 30530949 | 30530949 | C | T | SNP | PRR3 | ENST00000481741 | 1 | rna | NULL | NULL | tier1 | 95 | 0 | 0.000 | 102 | 87 | 0.460 |
| 855934 | 6 | 31637272 | 31637272 | C | T | SNP | CSNK2B | ENST00000375865 | 1 | nonsense | c.544 | p.Q182* | tier1 | 18 | 0 | 0.000 | 20 | 23 | 0.535 |
| 855934 | 6 | 118800974 | 118800974 | T | C | SNP | CEP85L | ENST00000368488 | -1 | intronic | c.1923+60 | e10+60 | tier3 | 68 | 0 | 0.000 | 35 | 28 | 0.444 |
| 855934 | 7 | 6439714 | 6439714 | T | C | SNP | RAC1 | ENST00000473564 | 1 | 3_prime_untranslated_region | c.*6439714 | NULL | tier3 | 195 | 0 | 0.000 | 164 | 134 | 0.450 |
| 855934 | 7 | 101758501 | 101758501 | C | T | SNP | CUX1 | ENST00000360264 | 1 | nonsense | c.655 | p.R219* | tier1 | 74 | 0 | 0.000 | 46 | 66 | 0.589 |
| 855934 | 8 | 139668092 | 139668092 | C | T | SNP | COL22A1 | ENST00000303045 | -1 | intronic | c.3333+48 | e44+48 | tier3 | 98 | 0 | 0.000 | 63 | 41 | 0.394 |
| 855934 | 8 | 140767744 | 140767744 | T | G | SNP | TRAPPC9 | ENST00000389328 | -1 | intronic | c.3350-23299 | e22-23299 | tier3 | 74 | 0 | 0.000 | 209 | 21 | 0.091 |
| 855934 | 8 | 145773426 | 145773426 | C | T | SNP | ARHGAP39 | ENST00000377307 | -1 | silent | c.1044 | p.S348 | tier1 | 12 | 0 | 0.000 | 18 | 15 | 0.455 |
| 855934 | 10 | 3183033 | 3183033 | A | G | SNP | PITRM1 | ENST00000380989 | -1 | intronic | c.2649-29 | e24-29 | tier3 | 80 | 0 | 0.000 | 78 | 55 | 0.410 |
| 855934 | 10 | 43651051 | 43651051 | A | G | SNP | CSGALNACT2 | ENST00000374466 | 1 | missense | c.454 | p.T152A | tier1 | 73 | 0 | 0.000 | 55 | 48 | 0.466 |
| 855934 | 10 | 71695049 | 71695049 | G | A | SNP | COL13A1 | ENST00000356340 | 1 | intronic | c.1696-57 | e30-57 | tier3 | 66 | 0 | 0.000 | 62 | 79 | 0.560 |
| 855934 | 10 | 104120904 | 104120904 | G | A | SNP | GBF1 | ENST00000476019 | 1 | rna | NULL | NULL | tier1 | 38 | 0 | 0.000 | 30 | 24 | 0.444 |
| 855934 | 10 | 115528652 | 115528652 | G | A | SNP | PLEKHS1 | ENST00000369310 | 1 | splice_region | c.417+3 | e5+3 | tier2 | 48 | 0 | 0.000 | 28 | 26 | 0.482 |
| 855934 | 10 | 135292946 | 135292946 | A | C | SNP | SPRN | ENST00000541506 | -1 | intronic | c.1-55745 | e1-55745 | tier3 | 38 | 0 | 0.000 | 46 | 11 | 0.193 |
| 855934 | 12 | 54796846 | 54796846 | G | A | SNP | ITGA5 | ENST00000551564 | -1 | 5_prime_untranslated_region | c.-54796846 | NULL | tier3 | 70 | 1 | 0.014 | 103 | 74 | 0.418 |
| 855934 | 12 | 70064230 | 70064230 | T | G | SNP | BEST3 | ENST00000476098 | -1 | missense | c.590 | p.D197A | tier1 | 47 | 0 | 0.000 | 54 | 50 | 0.481 |
| 855934 | 16 | 84888372 | 84888372 | C | T | SNP | CRISPLD2 | ENST00000262424 | 1 | missense | c.646 | p.R216W | tier1 | 77 | 0 | 0.000 | 115 | 113 | 0.496 |
| 855934 | 18 | 30253100 | 30253100 | C | T | SNP | KLHL14 | ENST00000359358 | -1 | 3_prime_untranslated_region | c.*1520 | NULL | tier2 | 121 | 1 | 0.008 | 74 | 44 | 0.373 |
| 855934 | 18 | 44555207 | 44555207 | G | A | SNP | TCEB3C | ENST00000330682 | -1 | missense | c.1007 | p.T336M | tier1 | 574 | 0 | 0.000 | 1089 | 41 | 0.036 |
| 855934 | 19 | 43679576 | 43679576 | C | T | SNP | PSG5 | ENST00000342951 | -1 | missense | c.755 | p.R252H | tier1 | 206 | 1 | 0.005 | 194 | 192 | 0.497 |
| 855934 | 19 | 57772501 | 57772501 | C | G | SNP | ZNF460 | ENST00000360338 | 1 | 5_prime_flanking_region | c.-19674 | NULL | tier3 | 77 | 1 | 0.013 | 57 | 46 | 0.442 |
| 855934 | 20 | 31022442 | 31022443 | GG | - | DEL | ASXL1 | ENST00000375687 | 1 | frame_shift_del | c.1927_1928 | p.G645fs | tier1 | 732 | 15 | 0.020 | 990 | 621 | 0.386 |
| 855934 | 20 | 49199177 | 49199177 | A | C | SNP | PTPN1 | ENST00000371621 | 1 | intronic | c.1285-52 | e10-52 | tier3 | 99 | 3 | 0.029 | 96 | 78 | 0.448 |
| 855934 | 21 | 44514777 | 44514777 | T | G | SNP | U2AF1 | ENST00000291552 | -1 | missense | c.470 | p.Q157P | tier1 | 374 | 7 | 0.018 | 449 | 328 | 0.421 |
| 855934 | X | 73071895 | 73071895 | C | A | SNP | XIST | ENST00000429829 | -1 | rna | NULL | NULL | tier1 | 74 | 4 | 0.051 | 105 | 41 | 0.281 |
| 855934 | X | 76888698 | 76888698 | G | A | SNP | ATRX | ENST00000373344 | -1 | missense | c.5131 | p.P1711S | tier1 | 25 | 0 | 0.000 | 2 | 25 | 0.926 |
| 855934 | X | 133511773 | 133511774 | - | A | INS | PHF6 | ENST00000394292 | 1 | frame_shift_ins | c.126_127 | p.H42fs | tier1 | 144 | 0 | 0.000 | 32 | 165 | 0.838 |
| 856550 | 1 | 26317279 | 26317279 | G | A | SNP | PAFAH2 | ENST00000374282 | -1 | missense | c.29 | p.P10L | tier1 | 65 | 3 | 0.044 | 61 | 41 | 0.402 |
| 856550 | 1 | 32127089 | 32127089 | C | T | SNP | COL16A1 | ENST00000271069 | -1 | intronic | c.3727-30 | e60-30 | tier3 | 80 | 1 | 0.012 | 55 | 46 | 0.455 |
| 856550 | 1 | 115258747 | 115258747 | C | T | SNP | NRAS | ENST00000369535 | -1 | missense | c.35 | p.G12D | tier1 | 470 | 3 | 0.006 | 461 | 250 | 0.351 |
| 856550 | 1 | 142699411 | 142699412 | - | T | INS | ENSG00000203849 | ENST00000369381 | -1 | rna | NULL | NULL | tier1 | 170 | 1 | 0.006 | 212 | 77 | 0.266 |
| 856550 | 1 | 204960404 | 204960404 | G | A | SNP | NFASC | ENST00000367172 | 1 | intronic | c.3092+2466 | e23+2466 | tier2 | 83 | 0 | 0.000 | 73 | 51 | 0.411 |
| 856550 | 1 | 206941873 | 206941873 | A | G | SNP | IL10 | ENST00000423557 | -1 | 3_prime_untranslated_region | c.*108 | NULL | tier3 | 156 | 0 | 0.000 | 152 | 149 | 0.493 |
| 856550 | 2 | 202514343 | 202514343 | C | T | SNP | MPP4 | ENST00000359962 | -1 | missense | c.1540 | p.V514I | tier1 | 85 | 2 | 0.023 | 75 | 40 | 0.348 |
| 856550 | 2 | 216967347 | 216967347 | T | - | DEL | TMEM169 | ENST00000295658 | 1 | 3_prime_untranslated_region | c.*2082 | NULL | tier4 | 121 | 3 | 0.024 | 169 | 25 | 0.129 |
| 856550 | 3 | 57232440 | 57232440 | A | T | SNP | HESX1 | ENST00000295934 | -1 | missense | c.438 | p.N146K | tier1 | 130 | 1 | 0.008 | 148 | 131 | 0.470 |
| 856550 | 3 | 141892042 | 141892042 | A | - | DEL | GK5 | ENST00000463349 | -1 | splice_region_del | c.29-3 | e2-3 | tier4 | 60 | 1 | 0.016 | 119 | 22 | 0.156 |
| 856550 | 3 | 178546013 | 178546013 | C | T | SNP | KCNMB2 | ENST00000358316 | 1 | missense | c.275 | p.T92M | tier1 | 98 | 3 | 0.030 | 72 | 89 | 0.553 |
| 856550 | 5 | 24487247 | 24487247 | C | A | SNP | CDH10 | ENST00000264463 | -1 | 3_prime_untranslated_region | c.*525 | NULL | tier4 | 262 | 4 | 0.015 | 282 | 219 | 0.435 |
| 856550 | 5 | 112403792 | 112403792 | C | A | SNP | MCC | ENST00000302475 | -1 | missense | c.1444 | p.V482L | tier1 | 47 | 0 | 0.000 | 36 | 39 | 0.520 |
| 856550 | 5 | 153190809 | 153190809 | G | A | SNP | GRIA1 | ENST00000448073 | 1 | 3_prime_untranslated_region | c.*24 | NULL | tier3 | 49 | 2 | 0.039 | 34 | 26 | 0.433 |
| 856550 | 6 | 66044805 | 66044805 | C | T | SNP | EYS | ENST00000342421 | -1 | 3_prime_untranslated_region | c.*49 | NULL | tier2 | 68 | 1 | 0.015 | 90 | 65 | 0.419 |
| 856550 | 6 | 131179391 | 131179391 | G | C | SNP | EPB41L2 | ENST00000526782 | -1 | 5_prime_untranslated_region | c.-131179391 | NULL | tier3 | 53 | 0 | 0.000 | 49 | 32 | 0.386 |
| 856550 | 7 | 131853016 | 131853016 | A | G | SNP | PLXNA4 | ENST00000321063 | -1 | intronic | c.4286+47 | e21+47 | tier3 | 53 | 0 | 0.000 | 32 | 28 | 0.467 |
| 856550 | 7 | 148507436 | 148507436 | T | C | SNP | EZH2 | ENST00000320356 | -1 | missense | c.2018 | p.N673S | tier1 | 368 | 3 | 0.008 | 286 | 197 | 0.407 |
| 856550 | 9 | 99233221 | 99233221 | T | C | SNP | HABP4 | ENST00000375249 | 1 | intronic | c.744-72 | e5-72 | tier3 | 78 | 1 | 0.013 | 44 | 31 | 0.408 |
| 856550 | 9 | 99233241 | 99233241 | G | C | SNP | HABP4 | ENST00000375249 | 1 | intronic | c.744-52 | e5-52 | tier3 | 105 | 1 | 0.009 | 61 | 50 | 0.446 |
| 856550 | 10 | 89118109 | 89118109 | T | C | SNP | NUTM2D | ENST00000381697 | 1 | silent | c.87 | p.L29 | tier1 | 1001 | 9 | 0.009 | 1024 | 294 | 0.223 |
| 856550 | 11 | 5537218 | 5537218 | C | T | SNP | UBQLNL | ENST00000380184 | -1 | missense | c.454 | p.V152M | tier1 | 29 | 0 | 0.000 | 20 | 13 | 0.394 |
| 856550 | 11 | 72294529 | 72294529 | G | T | SNP | PDE2A | ENST00000334456 | -1 | missense | c.1681 | p.Q561K | tier1 | 95 | 0 | 0.000 | 80 | 65 | 0.448 |
| 856550 | 11 | 83770488 | 83770488 | C | T | SNP | DLG2 | ENST00000376104 | -1 | silent | c.789 | p.V263 | tier1 | 130 | 0 | 0.000 | 74 | 51 | 0.408 |
| 856550 | 11 | 117282559 | 117282559 | G | A | SNP | CEP164 | ENST00000278935 | 1 | silent | c.4212 | p.A1404 | tier1 | 16 | 0 | 0.000 | 20 | 18 | 0.474 |
| 856550 | 12 | 25378647 | 25378647 | T | A | SNP | KRAS | ENST00000256078 | -1 | missense | c.351 | p.K117N | tier1 | 603 | 4 | 0.007 | 1082 | 59 | 0.052 |
| 856550 | 12 | 46205132 | 46205132 | T | G | SNP | ARID2 | ENST00000334344 | 1 | intronic | c.285-69 | e4-69 | tier3 | 46 | 0 | 0.000 | 69 | 35 | 0.337 |
| 856550 | 12 | 483769 | | | | | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|---|-----|----------------|------------------------|-----------------|----------|-----------------------------|---------------------|-----------|-------|------|-------|-------|------|-------|-------|
| 856550 | 15 | 78397250 | 78397250 | T | - | DEL | <i>CIB2</i> | ENST00000258930 | -1 | 3_prime_untranslated_region | c.*403 | NULL | tier3 | 111 | 2 | 0.018 | 139 | 20 | 0.126 |
| 856550 | 16 | 13297355 | 13297355 | C | G | SNP | <i>SHISA9</i> | ENST00000558583 | 1 | missense | c.919 | p.Q307E | tier1 | 78 | 1 | 0.013 | 59 | 37 | 0.381 |
| 856550 | 17 | 1028517 | 1028517 | C | A | SNP | <i>ABR</i> | ENST00000302538 | -1 | splice_site | c.246+1 | e2+1 | tier1 | 107 | 0 | 0.000 | 107 | 79 | 0.425 |
| 856550 | 17 | 11757380 | 11757380 | G | A | SNP | <i>DNAH9</i> | ENST00000262442 | 1 | missense | c.9568 | p.A3190T | tier1 | 730 | 1 | 0.001 | 965 | 31 | 0.031 |
| 856550 | 17 | 48356336 | 48356336 | G | A | SNP | <i>TMEM92</i> | ENST00000300433 | 1 | silent | c.345 | p.A115 | tier1 | 154 | 4 | 0.025 | 139 | 85 | 0.380 |
| 856550 | 17 | 59479337 | 59479337 | C | G | SNP | <i>TBX2</i> | ENST00000419047 | 1 | intronic | c.441+25 | e2+25 | tier2 | 73 | 0 | 0.000 | 48 | 25 | 0.343 |
| 856550 | 17 | 74387223 | 74387223 | G | C | SNP | <i>UBE2O</i> | ENST00000319380 | -1 | missense | c.3680 | p.S1227W | tier1 | 54 | 0 | 0.000 | 46 | 29 | 0.382 |
| 856550 | 20 | 34099166 | 34099166 | T | C | SNP | <i>CEP250</i> | ENST00000397527 | 1 | intronic | c.7066-26 | e32-26 | tier3 | 44 | 0 | 0.000 | 63 | 11 | 0.126 |
| 856550 | 20 | 45052040 | 45052040 | C | T | SNP | <i>ELMO2</i> | ENST00000445496 | -1 | intronic | c.1+9493 | e0+9493 | tier4 | 87 | 1 | 0.011 | 46 | 52 | 0.531 |
| 856550 | 20 | 57484420 | 57484420 | C | T | SNP | <i>GNAS</i> | ENST00000354359 | 1 | missense | c.604 | p.R202C | tier1 | 631 | 6 | 0.009 | 457 | 358 | 0.439 |
| 856550 | 21 | 36252998 | 36252998 | C | G | SNP | <i>RUNX1</i> | ENST00000300305 | -1 | missense | c.364 | p.G122R | tier1 | 1222 | 24 | 0.019 | 206 | 1486 | 0.878 |
| 856550 | 22 | 16414079 | 16414079 | G | A | SNP | <i>ENSG00000215268</i> | ENST00000417863 | 1 | 3_prime_untranslated_region | c.*16414079 | NULL | tier3 | 182 | 9 | 0.047 | 160 | 73 | 0.312 |
| 856550 | 22 | 17029213 | 17029213 | A | G | SNP | <i>ENSG00000233995</i> | ENST00000454360 | 1 | 5_prime_flanking_region | c.-23704 | NULL | tier3 | 483 | 7 | 0.014 | 570 | 32 | 0.053 |
| 856550 | 22 | 18628675 | 18628675 | C | T | SNP | <i>TUBA8</i> | ENST00000608634 | 1 | missense | c.745 | p.R249C | tier1 | 82 | 1 | 0.012 | 56 | 71 | 0.559 |
| 856550 | 22 | 45277142 | 45277142 | G | A | SNP | <i>PHF21B</i> | ENST00000313237 | -1 | 3_prime_untranslated_region | c.*1824 | NULL | tier2 | 156 | 1 | 0.006 | 150 | 96 | 0.390 |
| 856550 | X | 41782248 | 41782248 | G | C | SNP | <i>CASK</i> | ENST00000468986 | -1 | rna | NULL | NULL | tier1 | 96 | 0 | 0.000 | 69 | 54 | 0.439 |
| 856550 | X | 86063662 | 86063662 | A | G | SNP | <i>DACH2</i> | ENST00000373125 | 1 | intronic | c.1241-4197 | e8-4197 | tier3 | 136 | 0 | 0.000 | 146 | 84 | 0.362 |
| 856550 | X | 133527979 | 133527979 | G | T | SNP | <i>PHF6</i> | ENST00000394292 | 1 | nonsense | c.415 | p.E139* | tier1 | 561 | 3 | 0.005 | 683 | 498 | 0.422 |
| 875301 | 1 | 1717243 | 1717243 | T | - | DEL | <i>GNB1</i> | ENST00000378609 | -1 | 3_prime_untranslated_region | c.*1527 | NULL | tier2 | 39 | 0 | 0.000 | 25 | 7 | 0.219 |
| 875301 | 1 | 148887714 | 148887714 | C | T | SNP | <i>ENSG00000231448</i> | ENST00000444424 | 1 | 3_prime_untranslated_region | c.*148887714 | NULL | tier2 | 1927 | 1 | 0.001 | 2460 | 75 | 0.030 |
| 875301 | 2 | 64807082 | 64807082 | T | G | SNP | <i>AFTPH</i> | ENST00000498706 | 1 | 3_prime_untranslated_region | c.*64807082 | NULL | tier3 | 194 | 0 | 0.000 | 135 | 11 | 0.075 |
| 875301 | 2 | 179321040 | 179321040 | A | - | DEL | <i>DFNB59</i> | ENST00000375129 | 1 | intronic | c.549+162 | e3+162 | tier3 | 108 | 1 | 0.009 | 73 | 10 | 0.121 |
| 875301 | 2 | 198264776 | 198264776 | T | G | SNP | <i>SF3B1</i> | ENST00000335508 | -1 | splice_region | c.3013+3 | e20+3 | tier3 | 605 | 0 | 0.000 | 577 | 42 | 0.068 |
| 875301 | 2 | 198267359 | 198267359 | C | G | SNP | <i>SF3B1</i> | ENST00000335508 | -1 | missense | c.1998 | p.K666N | tier1 | 828 | 1 | 0.001 | 926 | 46 | 0.047 |
| 875301 | 3 | 50334761 | 50334761 | G | C | SNP | <i>NAT6</i> | ENST00000354862 | -1 | missense | c.200 | p.P67R | tier1 | 64 | 0 | 0.000 | 64 | 9 | 0.123 |
| 875301 | 4 | 183723267 | 183723267 | G | A | SNP | <i>TENM3</i> | ENST00000511685 | 1 | 3_prime_untranslated_region | c.*1763 | NULL | tier2 | 94 | 0 | 0.000 | 57 | 7 | 0.109 |
| 875301 | 7 | 48467344 | 48467345 | - | T | INS | <i>ABCA13</i> | ENST00000435803 | 1 | intronic | c.12460-19_12460-18 | e42-18 | tier3 | 130 | 0 | 0.000 | 126 | 14 | 0.100 |
| 875301 | 7 | 50450292 | 50450292 | A | G | SNP | <i>IKZF1</i> | ENST00000331340 | 1 | missense | c.476 | p.N159S | tier1 | 292 | 0 | 0.000 | 404 | 19 | 0.045 |
| 875301 | 7 | 104946931 | 104946932 | - | A | INS | <i>SRPK2</i> | ENST00000465112 | -1 | frame_shift_ins | c.159_158 | p.L53fs | tier1 | 108 | 0 | 0.000 | 75 | 9 | 0.107 |
| 875301 | 9 | 79256934 | 79256934 | G | A | SNP | <i>PRUNE2</i> | ENST00000428286 | -1 | intronic | c.7654+2721 | e5+2721 | tier4 | 20 | 0 | 0.000 | 22 | 10 | 0.313 |
| 875301 | 11 | 33361114 | 33361114 | A | - | DEL | <i>HIPK3</i> | ENST00000303296 | 1 | intronic | c.1613+34 | e5+34 | tier3 | 113 | 2 | 0.017 | 54 | 9 | 0.143 |
| 875301 | 11 | 121475955 | 121475955 | C | T | SNP | <i>SORL1</i> | ENST00000260197 | 1 | splice_region | c.4778+7 | e34+7 | tier3 | 119 | 0 | 0.000 | 128 | 12 | 0.086 |
| 875301 | 12 | 31247676 | 31247676 | T | A | SNP | <i>DDX11</i> | ENST00000407793 | 1 | intronic | c.1415-13 | e13-13 | tier3 | 505 | 1 | 0.002 | 546 | 29 | 0.050 |
| 875301 | 15 | 60745782 | 60745782 | A | - | DEL | <i>NARG2</i> | ENST00000561114 | -1 | frame_shift_del | c.1145 | p.F382fs | tier1 | 91 | 1 | 0.011 | 41 | 7 | 0.146 |
| 876752 | 1 | 153190890 | 153190890 | G | A | SNP | <i>PRR9</i> | ENST00000368744 | 1 | silent | c.270 | p.Q90 | tier1 | 73 | 0 | 0.000 | 49 | 26 | 0.347 |
| 876752 | 1 | 207881643 | 207881643 | A | G | SNP | <i>CR1L</i> | ENST00000508064 | 1 | intronic | c.1414+35 | e10+35 | tier4 | 278 | 3 | 0.011 | 337 | 63 | 0.158 |
| 876752 | 1 | 207881649 | 207881649 | C | T | SNP | <i>CR1L</i> | ENST00000508064 | 1 | intronic | c.1414+41 | e10+41 | tier4 | 267 | 2 | 0.007 | 318 | 60 | 0.157 |
| 876752 | 1 | 237947039 | 237947039 | C | T | SNP | <i>RYR2</i> | ENST00000360064 | 1 | silent | c.12045 | p.N4015 | tier1 | 150 | 1 | 0.007 | 94 | 54 | 0.365 |
| 876752 | 2 | 1099077 | 1099077 | A | T | SNP | <i>SNTG2</i> | ENST00000308624 | 1 | intronic | c.325+4981 | e4+4981 | tier4 | 34 | 0 | 0.000 | 45 | 11 | 0.196 |
| 876752 | 2 | 233899445 | 233899445 | G | A | SNP | <i>NEU2</i> | ENST00000233840 | 1 | missense | c.821 | p.G274E | tier1 | 50 | 3 | 0.057 | 35 | 20 | 0.357 |
| 876752 | 3 | 37340696 | 37340696 | A | - | DEL | <i>GOLGA4</i> | ENST00000361924 | 1 | intronic | c.1003-83 | e9-83 | tier3 | 144 | 1 | 0.007 | 144 | 98 | 0.405 |
| 876752 | 3 | 38621021 | 38621021 | C | T | SNP | <i>SCN5A</i> | ENST00000333535 | -1 | intronic | c.3229-35 | e17-35 | tier3 | 40 | 0 | 0.000 | 20 | 13 | 0.382 |
| 876752 | 4 | 82026868 | 82026868 | C | T | SNP | <i>PRKG2</i> | ENST00000264399 | -1 | intronic | c.2063+99 | e15+99 | tier3 | 106 | 1 | 0.009 | 82 | 73 | 0.468 |
| 876752 | 11 | 193204 | 193204 | G | SNP | <i>SCGB1C1</i> | ENST00000342878 | 1 | intronic | c.55+50 | e1+50 | tier3 | 169 | 4 | 0.023 | 141 | 22 | 0.133 | |
| 876752 | 11 | 71373771 | 71373771 | T | A | SNP | <i>ENPP7P8</i> | ENST00000529093 | 1 | 5_prime_flanking_region | c.-49015 | NULL | tier3 | 630 | 8 | 0.013 | 504 | 312 | 0.382 |
| 876752 | 15 | 21119477 | 21119477 | T | C | SNP | <i>POTEB2</i> | ENST00000454856 | -1 | 5_prime_flanking_region | c.-47867 | NULL | tier4 | 51 | 0 | 0.000 | 64 | 11 | 0.147 |
| 876752 | 15 | 90631934 | 90631934 | C | T | SNP | <i>IDH2</i> | ENST00000330062 | -1 | missense | c.419 | p.R140Q | tier1 | 660 | 0 | 0.000 | 790 | 29 | 0.035 |
| 876752 | 16 | 32890900 | 32890900 | C | T | SNP | <i>SLC6A10P</i> | ENST00000330048 | -1 | intronic | c.NULL | NULL | tier3 | 301 | 0 | 0.000 | 262 | 11 | 0.040 |
| 876752 | 16 | 84227714 | 84227714 | G | A | SNP | <i>ADAD2</i> | ENST00000268624 | 1 | missense | c.521 | p.R174H | tier1 | 103 | 0 | 0.000 | 62 | 54 | 0.466 |
| 876752 | 17 | 6703354 | 6703354 | C | G | SNP | <i>TEKT1</i> | ENST00000338694 | -1 | missense | c.1249 | p.V417L | tier1 | 40 | 0 | 0.000 | 19 | 23 | 0.548 |
| 876752 | 18 | 47380016 | 47380016 | G | A | SNP | <i>MYO5B</i> | ENST00000285039 | -1 | silent | c.4024 | p.L1342 | tier1 | 380 | 6 | 0.016 | 290 | 209 | 0.418 |
| 876752 | 19 | 7944301 | 7944301 | G | A | SNP | <i>FLJ22184</i> | ENST00000539422 | -1 | 5_prime_flanking_region | c.-5727 | NULL | tier3 | 73 | 0 | 0.000 | 50 | 39 | 0.438 |
| 876752 | 22 | 21153574 | 21153574 | G | A | SNP | <i>PI4KA</i> | ENST00000255882 | -1 | splice_region | c.1821-10 | e16-10 | tier3 | 73 | 1 | 0.014 | 51 | 42 | 0.452 |
| 882325 | 1 | 112525116 | 112525116 | T | G | SNP | <i>KCND3</i> | ENST00000315987 | -1 | missense | c.233 | p.D78A | tier1 | 57 | 1 | 0.017 | 84 | 46 | 0.351 |
| 882325 | 1 | 152186038 | 152186038 | A | - | DEL | <i>HRNR</i> | ENST00000368801 | -1 | frame_shift_del | c.8067 | p.H2690fs | tier1 | 14 | 0 | 0.000 | 7 | 10 | 0.588 |
| 882325 | 1 | 152186047 | 152186047 | A | - | DEL | <i>HRNR</i> | ENST00000368801 | -1 | frame_shift_del | c.8058 | p.R2687fs | tier1 | 10 | 0 | 0.000 | 5 | 10 | 0.667 |
| 882325 | 1 | 220369746 | 220369746 | A | - | DEL | <i>RAB3GAP2</i> | ENST00000358951 | -1 | splice_region_del | c.812-6 | e10-6 | tier3 | 77 | 1 | 0.013 | 178 | 24 | 0.119 |
| 882325 | 1 | 228464970 | 228464970 | G | T | SNP | <i>OBSCN</i> | ENST00000422127 | 1 | missense | c.6710 | p.C2237F | tier1 | 35 | 0 | 0.000 | 50 | 40 | 0.444 |
| 882325 | 1 | 228464971 | 228464971 | T | A | SNP | <i>OBSCN</i> | ENST00000422127 | 1 | nonsense | c.6711 | p.C2237* | tier1 | 37 | 0 | 0.000 | 51 | 40 | 0.440 |
| 882325 | 2 | 25457242 | 25457242 | C | T | SNP | <i>DNMT3A</i> | ENST00000264709 | -1 | missense | c.2645 | p.R882H | tier1 | 270 | 9 | 0.032 | 370 | 228 | 0.380 |
| 882325 | 2 | 209113112 | 209113112 | C | A | SNP | <i>IDH1</i> | ENST00000345146 | -1 | missense | c.395 | p.R132L | tier1 | 301 | 1 | 0.003 | 564 | 362 | 0.391 |
| 882325 | 2 | 211421474 | 211421474 | C | T | SNP | <i>CPS1</i> | ENST00000430249 | 1 | missense | c.35 | p.T12I | tier1 | 76 | 1 | 0.013 | 158 | 66 | 0.295 |
| 882325 | 2 | 238253632 | 238253632 | T | C | SNP | <i>COL6A3</i> | ENST00000295550 | -1 | intronic | c.7163-37 | e34-37 | tier3 | 71 | 2 | 0.027 | 103 | 45 | 0.302 |
| 882325 | 3 | 38948846 | 38948846 | T | C | SNP | <i>ENSG00000215941</i> | ENST00000401122 | 1 | rna | NULL | NULL | tier1 | 19 | 0 | 0.000 | 31 | 21 | 0.404 |
| 882325 | 3 | 184100959 | 184100959 | G | - | DEL | <i>CHRD</i> | ENST00000460627 | 1 | 3_prime_untranslated_region | c.*184100959 | NULL | tier3 | 82 | 2 | 0.024 | 114 | 22 | 0.162 |
| 882325 | 4 | 9785355 | 9785355 | A | - | DEL | <i>DRD5</i> | ENST00000304374 | 1 | 3_prime_untranslated_region | c.*268 | NULL | tier3 | 53 | 0 | 0.000 | 143 | 20 | 0.123 |
| 882325 | 4 | 170913470 | 170913470 | G | A | SNP | <i>MFAP3L</i> | ENST00000361618 | -1 | splice_region | c.299-10 | e2-10 | tier3 | 31 | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|---|---|-----|-----------------|-----------------|----|-----------------------------|-----------------|-----------|-------|-----|----|-------|------|------|-------|
| 882325 | 12 | 21051353 | 21051353 | C | A | SNP | SLCO1B3 | ENST00000553473 | 1 | intronic | c.1683-17 | e12-17 | tier2 | 110 | 1 | 0.009 | 246 | 125 | 0.336 |
| 882325 | 12 | 111655648 | 111655648 | G | A | SNP | CUX2 | ENST00000261726 | 1 | intronic | c.175-46 | e3-46 | tier3 | 70 | 0 | 0.000 | 104 | 41 | 0.279 |
| 882325 | 14 | 74196463 | 74196464 | - | G | INS | ELMSAN1 | ENST00000286523 | -1 | frame_shift_ins | c.1975_1974 | p.I658fs | tier1 | 52 | 1 | 0.019 | 43 | 25 | 0.368 |
| 882325 | 15 | 33833126 | 33833126 | G | T | SNP | RYR3 | ENST00000389232 | 1 | intronic | c.646+35 | e7+35 | tier3 | 188 | 1 | 0.005 | 357 | 273 | 0.432 |
| 882325 | 17 | 74732959 | 74732959 | G | A | SNP | SRSF2 | ENST00000359995 | -1 | missense | c.284 | p.P95L | tier1 | 320 | 2 | 0.006 | 427 | 232 | 0.352 |
| 882325 | 18 | 55996142 | 55996142 | G | A | SNP | NEDD4L | ENST00000400345 | 1 | intronic | c.681-85 | e10-85 | tier3 | 23 | 0 | 0.000 | 40 | 25 | 0.379 |
| 882325 | 19 | 2435136 | 2435136 | G | A | SNP | LMNB2 | ENST00000325327 | -1 | missense | c.718 | p.R240C | tier1 | 15 | 0 | 0.000 | 21 | 15 | 0.417 |
| 882325 | X | 50350816 | 50350816 | C | G | SNP | SHROOM4 | ENST00000289292 | -1 | missense | c.3326 | p.W1109S | tier1 | 25 | 1 | 0.039 | 9 | 25 | 0.735 |
| 882325 | X | 53441798 | 53441798 | A | G | SNP | SMO1A | ENST00000322213 | -1 | missense | c.320 | p.I107T | tier1 | 222 | 2 | 0.009 | 440 | 64 | 0.126 |
| 882325 | X | 77911875 | 77911875 | T | C | SNP | ZCCHC5 | ENST00000321110 | -1 | 3_prime_untranslated_region | c.*615 | NULL | tier2 | 32 | 1 | 0.030 | 37 | 56 | 0.602 |
| 907471 | 1 | 221875157 | 221875157 | A | - | DEL | DUSP10 | ENST00000468085 | -1 | rna | NULL | NULL | tier1 | 40 | 0 | 0.000 | 40 | 11 | 0.216 |
| 907471 | 1 | 237947546 | 237947546 | C | T | SNP | RYR2 | ENST00000360064 | 1 | silent | c.12552 | p.N4184 | tier1 | 304 | 2 | 0.007 | 454 | 23 | 0.048 |
| 907471 | 2 | 25457243 | 25457243 | G | A | SNP | DNMT3A | ENST00000264709 | -1 | missense | c.2644 | p.R882C | tier1 | 309 | 1 | 0.003 | 399 | 18 | 0.043 |
| 907471 | 5 | 82817864 | 82817864 | G | A | SNP | VCAN | ENST00000265077 | 1 | missense | c.3739 | p.D1247N | tier1 | 811 | 0 | 0.000 | 1203 | 28 | 0.023 |
| 907471 | 5 | 112227415 | 112227415 | G | A | SNP | ZRSR1 | ENST00000391338 | 1 | missense | c.79 | p.A27T | tier1 | 636 | 0 | 0.000 | 793 | 25 | 0.031 |
| 907471 | 5 | 142301123 | 142301123 | G | T | SNP | ARHGAP26 | ENST00000477867 | 1 | rna | NULL | NULL | tier1 | 225 | 0 | 0.000 | 175 | 11 | 0.059 |
| 907471 | 5 | 142301124 | 142301124 | G | T | SNP | ARHGAP26 | ENST00000477867 | 1 | rna | NULL | NULL | tier1 | 225 | 0 | 0.000 | 174 | 10 | 0.054 |
| 907471 | 7 | 126716306 | 126716306 | G | A | SNP | GRM8 | ENST00000339582 | -1 | intronic | c.727+30244 | e2+30244 | tier3 | 341 | 0 | 0.000 | 462 | 16 | 0.033 |
| 907471 | 12 | 32948971 | 32948971 | C | T | SNP | PKP2 | ENST0000070846 | -1 | intronic | c.2489+72 | e12+72 | tier3 | 72 | 0 | 0.000 | 121 | 14 | 0.104 |
| 907471 | 12 | 55820945 | 55820945 | G | A | SNP | OR6C76 | ENST00000328314 | 1 | missense | c.908 | p.R303K | tier1 | 29 | 0 | 0.000 | 23 | 8 | 0.258 |
| 907471 | 12 | 111701486 | 111701487 | - | A | INS | CUX2 | ENST00000261726 | 1 | intronic | c.223-79_223-78 | e4-78 | tier3 | 76 | 0 | 0.000 | 121 | 13 | 0.097 |
| 907471 | 14 | 36334922 | 36334922 | A | T | SNP | BRMS1L | ENST00000216807 | 1 | splice_region | c.688-6 | e8-6 | tier4 | 47 | 2 | 0.041 | 35 | 16 | 0.314 |
| 907471 | 16 | 89805075 | 89805075 | G | A | SNP | FANCA | ENST00000389301 | -1 | silent | c.4302 | p.A1434 | tier1 | 814 | 0 | 0.000 | 1086 | 27 | 0.024 |
| 907471 | 21 | 19628809 | 19628810 | - | T | INS | CHODL | ENST00000299295 | 1 | intronic | c.80-17_80-16 | e2-16 | tier3 | 12 | 0 | 0.000 | 13 | 13 | 0.500 |
| 913698 | 1 | 91973859 | 91973859 | A | G | SNP | CDC7 | ENST00000234626 | 1 | silent | c.240 | p.Q80 | tier1 | 128 | 3 | 0.023 | 194 | 102 | 0.345 |
| 913698 | 1 | 118426264 | 118426264 | G | A | SNP | GDAP2 | ENST00000369443 | -1 | intronic | c.1108-15 | e10-15 | tier3 | 56 | 0 | 0.000 | 79 | 44 | 0.358 |
| 913698 | 1 | 142826527 | 142826527 | G | A | SNP | ANKRD20A14P | ENST00000457776 | 1 | 3_prime_untranslated_region | c.*142826527 | NULL | tier3 | 22 | 1 | 0.044 | 20 | 23 | 0.535 |
| 913698 | 1 | 182791320 | 182791320 | G | A | SNP | NPL | ENST00000258317 | 1 | missense | c.724 | p.A242T | tier1 | 95 | 3 | 0.031 | 157 | 63 | 0.286 |
| 913698 | 2 | 132723935 | 132723935 | G | A | SNP | ENSG00000235615 | ENST00000419362 | -1 | rna | NULL | NULL | tier1 | 9 | 0 | 0.000 | 2 | 9 | 0.818 |
| 913698 | 2 | 196636498 | 196636498 | G | A | SNP | DNAH7 | ENST00000312428 | -1 | silent | c.11319 | p.Y3773 | tier1 | 102 | 0 | 0.000 | 205 | 40 | 0.162 |
| 913698 | 2 | 227924194 | 227924194 | C | T | SNP | COL4A4 | ENST00000396625 | -1 | silent | c.2310 | p.P770 | tier1 | 75 | 3 | 0.039 | 152 | 65 | 0.298 |
| 913698 | 3 | 47050909 | 47050909 | G | - | DEL | NBEAL2 | ENST00000450053 | -1 | 3_prime_untranslated_region | c.*99 | NULL | tier2 | 6 | 0 | 0.000 | 8 | 15 | 0.652 |
| 913698 | 3 | 112300106 | 112300106 | C | T | SNP | SLC35A5 | ENST00000492406 | 1 | missense | c.1142 | p.P381L | tier1 | 75 | 4 | 0.051 | 133 | 51 | 0.276 |
| 913698 | 4 | 1656787 | 1656787 | C | T | SNP | FAM53A | ENST00000308132 | -1 | missense | c.800 | p.G267E | tier1 | 21 | 0 | 0.000 | 23 | 23 | 0.489 |
| 913698 | 4 | 184019703 | 184019703 | C | G | SNP | WWC2-AS2 | ENST00000506413 | -1 | rna | NULL | NULL | tier1 | 11 | 0 | 0.000 | 6 | 7 | 0.539 |
| 913698 | 5 | 43277155 | 43277155 | G | A | SNP | NIM1 | ENST00000326035 | 1 | splice_region | c.293-4 | e2-4 | tier3 | 42 | 1 | 0.023 | 63 | 27 | 0.300 |
| 913698 | 6 | 26536971 | 26536971 | A | - | DEL | HMGNA4 | ENST00000377575 | 1 | 5_prime_flanking_region | c.-8464 | NULL | tier2 | 34 | 0 | 0.000 | 59 | 14 | 0.192 |
| 913698 | 7 | 100468311 | 100468311 | C | T | SNP | TRIP6 | ENST00000429658 | 1 | missense | c.266 | p.A89V | tier1 | 65 | 0 | 0.000 | 142 | 32 | 0.184 |
| 913698 | 9 | 119461034 | 119461034 | C | T | SNP | TRIM32 | ENST00000373983 | 1 | missense | c.1013 | p.A338V | tier1 | 41 | 0 | 0.000 | 52 | 32 | 0.381 |
| 913698 | 9 | 124530916 | 124530916 | G | G | SNP | DAB2IP | ENST00000408936 | 1 | splice_region | c.1899+4 | e10+4 | tier3 | 92 | 4 | 0.042 | 110 | 56 | 0.335 |
| 913698 | 11 | 5536564 | 5536564 | C | A | SNP | UBQLNL | ENST00000380184 | -1 | missense | c.1108 | p.H370Y | tier1 | 29 | 1 | 0.033 | 42 | 21 | 0.333 |
| 913698 | 11 | 6891758 | 6891758 | A | T | SNP | OR10A2 | ENST00000307322 | 1 | missense | c.773 | p.K258M | tier1 | 36 | 3 | 0.039 | 72 | 64 | 0.377 |
| 913698 | 11 | 120744937 | 120744937 | G | A | SNP | GRIK4 | ENST00000527524 | 1 | splice_region | c.1059+10 | e8+10 | tier3 | 301 | 7 | 0.023 | 515 | 191 | 0.270 |
| 913698 | 12 | 91363823 | 91363823 | G | A | SNP | EPYC | ENST00000261172 | -1 | nonsense | c.796 | p.Q266* | tier1 | 130 | 0 | 0.000 | 241 | 38 | 0.136 |
| 913698 | 15 | 41223819 | 41223819 | C | - | DEL | DLL4 | ENST00000249749 | 1 | frame_shift_del | c.513 | p.R172fs | tier1 | 49 | 2 | 0.039 | 75 | 35 | 0.318 |
| 913698 | 15 | 90631934 | 90631934 | C | T | SNP | IDH2 | ENST00000330062 | -1 | missense | c.419 | p.R140Q | tier1 | 358 | 3 | 0.008 | 838 | 203 | 0.195 |
| 913698 | 16 | 88808530 | 88808530 | G | A | SNP | ENSG00000260617 | ENST00000566114 | 1 | rna | NULL | NULL | tier1 | 229 | 3 | 0.013 | 547 | 38 | 0.065 |
| 913698 | 17 | 39122986 | 39122986 | G | A | SNP | KRT39 | ENST00000355612 | -1 | silent | c.123 | p.V41 | tier1 | 64 | 3 | 0.045 | 142 | 57 | 0.286 |
| 913698 | 17 | 74732959 | 74732959 | G | T | SNP | SRSF2 | ENST00000359995 | -1 | missense | c.284 | p.P95H | tier1 | 199 | 1 | 0.005 | 414 | 165 | 0.285 |
| 913698 | 20 | 31022441 | 31022442 | - | G | INS | ASXL1 | ENST00000375687 | 1 | frame_shift_ins | c.1926_1927 | p.G645fs | tier1 | 590 | 12 | 0.020 | 1159 | 352 | 0.233 |
| 913698 | 20 | 40701696 | 40701696 | C | T | SNP | PTPR1 | ENST00000373198 | -1 | 3_prime_untranslated_region | c.*7823 | NULL | tier2 | 62 | 0 | 0.000 | 80 | 26 | 0.245 |
| 913698 | 20 | 60773672 | 60773672 | G | A | SNP | MTG2 | ENST00000370823 | 1 | intronic | c.469-18 | e4-18 | tier3 | 52 | 3 | 0.055 | 95 | 41 | 0.302 |
| 913698 | 21 | 36252877 | 36252877 | C | T | SNP | RUNX1 | ENST00000300305 | -1 | missense | c.485 | p.R162K | tier1 | 921 | 54 | 0.055 | 761 | 1152 | 0.602 |
| 913698 | X | 52679352 | 52679352 | T | G | SNP | SSX7 | ENST00000298181 | -1 | intronic | c.330+51 | e4+51 | tier3 | 254 | 6 | 0.023 | 486 | 218 | 0.309 |
| 913698 | X | 140967238 | 140967238 | G | A | SNP | MAGEC3 | ENST00000298296 | 1 | intronic | c.515+21 | e3+21 | tier3 | 180 | 6 | 0.032 | 258 | 104 | 0.287 |
| 918051 | 1 | 94586694 | 94586694 | A | G | SNP | ABCA4 | ENST00000370225 | -1 | 5_prime_flanking_region | c.-93 | NULL | tier3 | 31 | 0 | 0.000 | 88 | 61 | 0.409 |
| 918051 | 1 | 94586695 | 94586695 | C | G | SNP | ABCA4 | ENST00000370225 | -1 | 5_prime_flanking_region | c.-94 | NULL | tier3 | 32 | 0 | 0.000 | 88 | 61 | 0.409 |
| 918051 | 1 | 150121929 | 150121929 | C | G | SNP | PLEKHO1 | ENST00000369126 | 1 | 5_prime_untranslated_region | c.-9109 | NULL | tier2 | 11 | 0 | 0.000 | 27 | 36 | 0.571 |
| 918051 | 1 | 152186047 | 152186047 | A | - | DEL | HRNR | ENST00000368801 | -1 | frame_shift_del | c.8058 | p.R2687fs | tier1 | 8 | 0 | 0.000 | 3 | 6 | 0.667 |
| 918051 | 2 | 128621736 | 128621736 | C | T | SNP | AMMECR1L | ENST00000272647 | -1 | 3_prime_untranslated_region | c.*932 | NULL | tier3 | 8 | 0 | 0.000 | 59 | 90 | 0.604 |
| 918051 | 3 | 112998095 | 112998095 | G | A | SNP | BOC | ENST00000273395 | 1 | splice_site | c.1817-1 | e10-1 | tier1 | 14 | 0 | 0.000 | 31 | 39 | 0.549 |
| 918051 | 4 | 7780663 | 7780664 | - | T | INS | AFAP1 | ENST00000505447 | -1 | 5_prime_untranslated_region | c.-7780663 | NULL | tier3 | 80 | 0 | 0.000 | 106 | 11 | 0.094 |
| 918051 | 5 | 36876937 | 36876937 | C | - | DEL | NIPBL | ENST00000282516 | 1 | 5_prime_untranslated_region | c.-76862 | NULL | tier2 | 52 | 1 | 0.019 | 137 | 51 | 0.271 |
| 918051 | 5 | 140232564 | 140232564 | C | T | SNP | PCDHA9 | ENST00000378122 | 1 | 3_prime_untranslated_region | c.*1955 | NULL | tier4 | 36 | 0 | 0.000 | 13 | 7 | 0.350 |
| 918051 | 6 | 32557378 | 32557378 | T | C | SNP | HLA-DRB1 | ENST00000360004 | -1 | intronic | c.100+42 | e1+42 | tier3 | 31 | 0 | 0.000 | 158 | 37 | 0.190 |
| 918051 | 7 | 105668989 | 105668989 | C | T | SNP | CDHR3 | ENST00000468143 | 1 | 3_prime_untranslated_region | c.*105668989 | NULL | tier3 | 45 | 0 | 0.000 | 82 | 72 | 0.468 |
| 918051 | 9 | 2907139 | 2907139 | G | A | SNP | CARM1P1 | ENST00000426329 | -1 | 5_prime_untranslated_region | c.-2907139 | NULL | tier3 | 14 | 0 | 0.000 | 82 | 53 | 0.393 |
| 918051 | 9 | 88452656 | 88452656 | C | T | SNP | ENSG00000165121 | ENST00000431724 | 1 | intronic | c.NULL | NULL | tier3 | 48 | 0 | 0.000 | 126 | 77 | 0.379 |
| 918051 | 10 | 19616672 | 19616672 | T | A | SNP | C10orf112 | ENST00000454679 | 1 | intronic | c.1575+70 | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|-----------------|------|-------|-----------------|-----------------|----|-----------------------------|-----------------------|--------------------|-------|------|----|-------|------|-----|-------|
| 918051 | 11 | 20696148 | 20696148 | G | A | SNP | NELL1 | ENST00000357134 | 1 | intronic | c.56-3330 | e2-3330 | tier4 | 9 | 0 | 0.000 | 35 | 42 | 0.546 |
| 918051 | 11 | 63342376 | 63342376 | T | C | SNP | PLA2G16 | ENST00000394613 | -1 | rna | NULL | NULL | tier1 | 23 | 0 | 0.000 | 84 | 69 | 0.451 |
| 918051 | 11 | 92533422 | 92533422 | T | C | SNP | FAT3 | ENST00000298047 | 1 | missense | c.7243 | p.C2415R | tier1 | 17 | 0 | 0.000 | 88 | 59 | 0.401 |
| 918051 | 12 | 116446681 | 116446681 | T | C | SNP | MED13L | ENST00000281928 | -1 | missense | c.1537 | p.I513V | tier1 | 22 | 0 | 0.000 | 87 | 68 | 0.439 |
| 918051 | 13 | 42118880 | 42118880 | A | G | SNP | VWA8 | ENST00000379310 | -1 | 3_prime_flanking_region | c.*23453 | NULL | tier4 | 30 | 0 | 0.000 | 51 | 13 | 0.203 |
| 918051 | 14 | 57070634 | 57070634 | A | G | SNP | TMEM260 | ENST00000261556 | 1 | missense | c.446 | p.N149S | tier1 | 134 | 0 | 0.000 | 496 | 391 | 0.440 |
| 918051 | 15 | 62359822 | 62359822 | C | A | SNP | C2CD4A | ENST00000355522 | 1 | missense | c.10 | p.L4M | tier1 | 22 | 0 | 0.000 | 63 | 50 | 0.443 |
| 918051 | 16 | 780298 | 780298 | G | A | SNP | NARFL | ENST00000562862 | -1 | rna | NULL | NULL | tier1 | 25 | 0 | 0.000 | 96 | 77 | 0.445 |
| 918051 | 19 | 1026637 | 1026637 | C | A | SNP | CNN2 | ENST00000562015 | 1 | rna | NULL | NULL | tier1 | 9 | 0 | 0.000 | 14 | 22 | 0.611 |
| 918051 | 19 | 37063218 | 37063219 | - | A | INS | ZNF529 | ENST00000591340 | -1 | intronic | c.14+16_14+15 | e1+16 | tier3 | 12 | 0 | 0.000 | 57 | 42 | 0.424 |
| 918051 | 19 | 39869235 | 39869235 | G | A | SNP | SAMD4B | ENST00000314471 | 1 | splice_region | c.1530+6 | e7+6 | tier3 | 23 | 0 | 0.000 | 35 | 21 | 0.375 |
| 918051 | 20 | 1920551 | 1920551 | T | C | SNP | LOC727993 | ENST00000446562 | 1 | 5_prime_flanking_region | c.-7341 | NULL | tier2 | 49 | 2 | 0.039 | 60 | 48 | 0.444 |
| 918051 | 20 | 34022502 | 34022502 | G | A | SNP | GDF5 | ENST00000374369 | -1 | silent | c.711 | p.A237 | tier1 | 11 | 0 | 0.000 | 30 | 28 | 0.483 |
| 918051 | 20 | 44979120 | 44979120 | G | C | SNP | SLC35C2 | ENST00000543605 | -1 | silent | c.1098 | p.P366 | tier1 | 29 | 0 | 0.000 | 73 | 75 | 0.507 |
| 918051 | 22 | 23055600 | 23055600 | C | T | SNP | IGLV3-21 | ENST00000390308 | 1 | missense | c.263 | p.T88M | tier1 | 40 | 0 | 0.000 | 74 | 63 | 0.460 |
| 918051 | X | 123197782 | 123197783 | - | A | INS | STAG2 | ENST00000218089 | 1 | frame_shift_ins | c.1906_1907 | p.Y636fs | tier1 | 37 | 0 | 0.000 | 117 | 468 | 0.800 |
| 922185 | 1 | 149400471 | 149400471 | T | C | SNP | HIST2H3PS2 | ENST00000392948 | -1 | silent | c.72 | p.K24 | tier1 | 188 | 4 | 0.021 | 197 | 57 | 0.224 |
| 922185 | 2 | 20100829 | 20100829 | T | A | SNP | TTC32 | ENST00000431392 | -1 | silent | c.54 | p.S18 | tier1 | 85 | 0 | 0.000 | 115 | 38 | 0.248 |
| 922185 | 2 | 179638759 | 179638759 | A | G | SNP | TTN | ENST00000342992 | -1 | missense | c.7136 | p.V2379A | tier1 | 77 | 0 | 0.000 | 110 | 14 | 0.113 |
| 922185 | 4 | 25930000 | 25930000 | A | - | DEL | SMIM20 | ENST00000506197 | 1 | splice_region_del | c.166+9 | e2+9 | tier3 | 87 | 0 | 0.000 | 112 | 15 | 0.118 |
| 922185 | 4 | 71463976 | 71463976 | C | T | SNP | AMB1 | ENST00000322937 | 1 | intronic | c.136-97 | e4-97 | tier3 | 74 | 1 | 0.013 | 128 | 20 | 0.135 |
| 922185 | 4 | 100544120 | 100544120 | T | A | SNP | MTTP | ENST00000265517 | 1 | 3_prime_untranslated_region | c.*115 | NULL | tier3 | 77 | 0 | 0.000 | 109 | 32 | 0.227 |
| 922185 | 5 | 82817833 | 82817833 | C | T | SNP | VCAN | ENST00000265077 | 1 | silent | c.3708 | p.I1236 | tier1 | 739 | 3 | 0.004 | 1063 | 55 | 0.049 |
| 922185 | 5 | 126753323 | 126753323 | C | T | SNP | MEGF10 | ENST00000274473 | 1 | splice_region | c.1131-7 | e9-7 | tier3 | 47 | 0 | 0.000 | 55 | 24 | 0.304 |
| 922185 | 5 | 137298188 | 137298188 | T | C | SNP | FAM13B | ENST0000033079 | -1 | intronic | c.1180-30 | e9-30 | tier3 | 49 | 1 | 0.020 | 78 | 25 | 0.240 |
| 922185 | 7 | 39990405 | 39990406 | - | CCGC | INS | CDK13 | ENST00000181839 | 1 | frame_shift_ins | c.165_166 | p.L57fs | tier1 | 27 | 0 | 0.000 | 19 | 7 | 0.269 |
| 922185 | 7 | 55241540 | 55241540 | G | T | SNP | EGFR | ENST00000275493 | 1 | intronic | c.2062-74 | e18-74 | tier3 | 187 | 1 | 0.005 | 211 | 13 | 0.058 |
| 922185 | 9 | 111954473 | 111954473 | A | T | SNP | EPB41L4B | ENST00000374566 | -1 | intronic | c.2301+85 | e22+85 | tier3 | 83 | 0 | 0.000 | 86 | 25 | 0.225 |
| 922185 | 10 | 126678193 | 126678193 | A | G | SNP | CTBP2 | ENST00000309035 | -1 | missense | c.2852 | p.I951T | tier1 | 70 | 0 | 0.000 | 81 | 15 | 0.156 |
| 922185 | 11 | 57970725 | 57970725 | A | C | SNP | OR1S2 | ENST00000302592 | -1 | missense | c.929 | p.M310R | tier1 | 105 | 3 | 0.028 | 142 | 45 | 0.241 |
| 922185 | 11 | 117340626 | 117340626 | C | T | SNP | DSCAML1 | ENST00000321322 | -1 | silent | c.3204 | p.K1068 | tier1 | 134 | 1 | 0.007 | 122 | 41 | 0.252 |
| 922185 | 11 | 134605431 | 134605431 | G | A | SNP | ENSG00000251226 | ENST00000513405 | 1 | 5_prime_flanking_region | c.-59 | NULL | tier2 | 449 | 14 | 0.030 | 333 | 121 | 0.266 |
| 922185 | 12 | 10146243 | 10146243 | A | C | SNP | CLEC1B | ENST00000298527 | -1 | intronic | c.546-357 | e6-357 | tier3 | 45 | 0 | 0.000 | 57 | 16 | 0.219 |
| 922185 | 12 | 50026453 | 50026453 | G | T | SNP | PRPF40B | ENST00000548436 | 1 | 3_prime_untranslated_region | c.*50026453 | NULL | tier3 | 710 | 1 | 0.001 | 594 | 169 | 0.221 |
| 922185 | 14 | 21968662 | 21968662 | A | G | SNP | METTL3 | ENST00000298717 | -1 | missense | c.1279 | p.F427L | tier1 | 60 | 1 | 0.016 | 80 | 26 | 0.245 |
| 922185 | 14 | 93704243 | 93704243 | T | C | SNP | BTBD7 | ENST00000334746 | -1 | 3_prime_untranslated_region | c.*4376 | NULL | tier3 | 50 | 0 | 0.000 | 67 | 22 | 0.247 |
| 922185 | 15 | 34519872 | 34519872 | A | C | SNP | EMC4 | ENST00000561246 | 1 | 3_prime_untranslated_region | c.*34519872 | NULL | tier3 | 41 | 0 | 0.000 | 45 | 11 | 0.196 |
| 922185 | 15 | 49926994 | 49926994 | A | - | DEL | DTWD1 | ENST00000251250 | 1 | splice_region_del | c.667+3 | e3+3 | tier3 | 77 | 0 | 0.000 | 145 | 16 | 0.099 |
| 922185 | 15 | 89386622 | 89386622 | C | T | SNP | ACAN | ENST00000439576 | 1 | missense | c.794 | p.T265I | tier1 | 86 | 4 | 0.044 | 71 | 27 | 0.276 |
| 922185 | 17 | 15457016 | 15457016 | C | A | SNP | TVP23C | ENST00000225576 | -1 | missense | c.223 | p.D75Y | tier1 | 108 | 1 | 0.009 | 170 | 46 | 0.213 |
| 922185 | 17 | 48668000 | 48668000 | G | A | SNP | CACNA1G | ENST00000359106 | 1 | intronic | c.2453+17 | e10+17 | tier3 | 461 | 0 | 0.000 | 578 | 45 | 0.072 |
| 922185 | 18 | 19781636 | 19781636 | A | T | SNP | GATA6 | ENST00000269216 | 1 | 3_prime_untranslated_region | c.*850 | NULL | tier2 | 144 | 0 | 0.000 | 260 | 70 | 0.212 |
| 922185 | 18 | 21140392 | 21140392 | G | A | SNP | NPC1 | ENST00000269228 | -1 | silent | c.684 | p.D228 | tier1 | 31 | 0 | 0.000 | 54 | 14 | 0.206 |
| 922185 | 22 | 38686807 | 38686808 | - | T | INS | CSNK1E | ENST00000359867 | -1 | 3_prime_untranslated_region | c.*2486 | NULL | tier2 | 78 | 0 | 0.000 | 134 | 16 | 0.107 |
| 933226 | 1 | 45798446 | 45798446 | C | T | SNP | MUTYH | ENST00000450313 | -1 | missense | c.565 | p.G189R | tier1 | 53 | 0 | 0.000 | 68 | 34 | 0.333 |
| 933226 | 2 | 179532466 | 179532466 | T | - | DEL | TTN | ENST00000342992 | -1 | intronic | c.31483+2479 | e151+2479 | tier2 | 15 | 0 | 0.000 | 76 | 43 | 0.361 |
| 933226 | 2 | 228512849 | 228512849 | G | A | SNP | ENSG00000250599 | ENST00000509872 | -1 | 5_prime_untranslated_region | c.-228512849 | NULL | tier3 | 64 | 0 | 0.000 | 119 | 53 | 0.308 |
| 933226 | 2 | 241976165 | 241976165 | G | A | SNP | SNED1 | ENST00000310397 | 1 | intronic | c.806-42 | e5-42 | tier2 | 14 | 0 | 0.000 | 7 | 8 | 0.533 |
| 933226 | 2 | 242755726 | 242755726 | G | T | SNP | NEU4 | ENST00000325935 | 1 | silent | c.84 | p.R28 | tier1 | 36 | 0 | 0.000 | 36 | 30 | 0.455 |
| 933226 | 3 | 11303055 | 11303055 | C | T | SNP | HRH1 | ENST00000397056 | 1 | 3_prime_untranslated_region | c.*868 | NULL | tier4 | 40 | 0 | 0.000 | 62 | 50 | 0.446 |
| 933226 | 3 | 142546497 | 142546497 | G | A | SNP | ENSG00000240950 | ENST00000489462 | -1 | 5_prime_untranslated_region | c.-142546497 | NULL | tier3 | 44 | 0 | 0.000 | 63 | 32 | 0.337 |
| 933226 | 3 | 195512549 | 195512549 | A | G | SNP | MUC4 | ENST00000463781 | -1 | missense | c.5902 | p.S1968P | tier1 | 209 | 0 | 0.000 | 335 | 72 | 0.176 |
| 933226 | 4 | 665776 | 665776 | G | T | SNP | PCGF3 | ENST00000362003 | 1 | 5_prime_flanking_region | c.-61694 | NULL | tier3 | 76 | 0 | 0.000 | 70 | 41 | 0.369 |
| 933226 | 4 | 7741871 | 7741871 | C | T | SNP | SORCS2 | ENST00000507866 | 1 | intronic | c.3416-56 | e27-56 | tier3 | 20 | 0 | 0.000 | 19 | 19 | 0.500 |
| 933226 | 4 | 55161723 | 55161723 | G | A | SNP | PDGFRA | ENST00000257290 | 1 | 3_prime_untranslated_region | c.*284 | NULL | tier3 | 42 | 0 | 0.000 | 125 | 70 | 0.359 |
| 933226 | 4 | 187628190 | 187628190 | T | A | SNP | FAT1 | ENST00000441802 | -1 | missense | c.2792 | p.N931I | tier1 | 37 | 0 | 0.000 | 55 | 37 | 0.402 |
| 933226 | 4 | 189068340 | 189068342 | TGG | - | DEL | TRIML1 | ENST00000332517 | 1 | in_frame_del | c.1221_1223 | p.G408in_frame_del | tier1 | 65 | 0 | 0.000 | 99 | 52 | 0.344 |
| 933226 | 5 | 1283509 | 1283509 | A | C | SNP | TERT | ENST00000310581 | -1 | intronic | c.1574-770 | e3-770 | tier3 | 904 | 15 | 0.016 | 1313 | 83 | 0.059 |
| 933226 | 5 | 13793657 | 13793657 | A | G | SNP | DNAH5 | ENST00000265104 | -1 | silent | c.8191 | p.L273I | tier1 | 390 | 0 | 0.000 | 514 | 271 | 0.344 |
| 933226 | 5 | 31511227 | 31511227 | C | T | SNP | DROSHA | ENST00000512076 | -1 | missense | c.631 | p.G211R | tier1 | 65 | 0 | 0.000 | 101 | 48 | 0.322 |
| 933226 | 5 | 34182770 | 34182770 | C | T | SNP | LOC100653061 | ENST00000514048 | -1 | intronic | c.NULL | NULL | tier3 | 1352 | 1 | 0.001 | 2412 | 159 | 0.062 |
| 933226 | 5 | 118309302 | 118309319 | GCCTCCTTGCTCGCG | - | DEL | DTWD2 | ENST00000510708 | -1 | intronic | c.218+14670_218+14653 | e1+14670 | tier2 | 64 | 2 | 0.030 | 43 | 16 | 0.271 |
| 933226 | 6 | 29911836 | 29911836 | GCA | T | SNP | HLA-A | ENST00000376806 | 1 | intronic | c.620-63 | e4-63 | tier2 | 208 | 1 | 0.005 | 312 | 19 | 0.057 |
| 933226 | 6 | 29911844 | 29911844 | T | A | SNP | HLA-A | ENST00000376806 | 1 | intronic | c.620-55 | e4-55 | tier2 | 230 | 1 | 0.004 | 341 | 20 | 0.055 |
| 933226 | 6 | 169623535 | 169623535 | C | T | SNP | THBS2 | ENST00000366787 | -1 | missense | c.2809 | p.D937N | tier1 | 36 | 0 | 0.000 | 80 | 44 | 0.355 |
| 933226 | 7 | 72455428 | 72455428 | C | G | SNP | STAG3L3 | ENST00000436857 | -1 | intronic | c.NULL | NULL | tier3 | 16 | 0 | 0.000 | 18 | 29 | 0.617 |
| 933226 | 7 | 94157586 | 94157586 | T | - | DEL | CASD1 | ENST00000297273 | 1 | intronic | c.459+24 | e5+24 | tier4 | 18 | 0 | 0.000 | 151 | 69 | 0.314 |
| 933226 | 7 | 157652472 | 157652472 | A | C | SNP | PTPRN2 | ENST00000404321 | -1 | intronic | c.1857+38893 | e12+38893 | tier3 | 433 | 7 | 0.016 | 742 | 49 | 0.062 |
| 933226 | 7 | 157897557 | 157897557 | T | C | SNP</ | | | | | | | | | | | | | |

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|--------|----|-----------|-----------|---|----|-----|------------------------|-----------------|----|-----------------------------|-------------------|-----------|-------|------|---|-------|------|------|-------|
| 933226 | 9 | 68728891 | 68728891 | C | T | SNP | <i>ENSG00000229789</i> | ENST00000427066 | 1 | 3_prime_untranslated_region | c.*68728891 | NULL | tier3 | 72 | 1 | 0.014 | 477 | 62 | 0.115 |
| 933226 | 9 | 93976302 | 93976302 | A | G | SNP | <i>AUH</i> | ENST00000473695 | -1 | rna | NULL | NULL | tier1 | 22 | 0 | 0.000 | 152 | 92 | 0.377 |
| 933226 | 10 | 81609037 | 81609037 | G | A | SNP | <i>NUFM2E</i> | ENST00000429984 | 1 | intronc | c.1735-90 | e6-90 | tier3 | 69 | 0 | 0.000 | 131 | 20 | 0.132 |
| 933226 | 11 | 62037739 | 62037739 | C | T | SNP | <i>SCGB2A2</i> | ENST00000227918 | 1 | silent | c.51 | p.Y17 | tier1 | 39 | 1 | 0.025 | 37 | 28 | 0.431 |
| 933226 | 11 | 83497858 | 83497858 | A | T | SNP | <i>DLG2</i> | ENST00000376104 | -1 | intronc | c.1723-23 | e16-23 | tier3 | 53 | 0 | 0.000 | 49 | 33 | 0.402 |
| 933226 | 12 | 59307704 | 59307704 | G | A | SNP | <i>LRIG3</i> | ENST00000548968 | -1 | 5_prime_untranslated_region | c.-59307704 | NULL | tier3 | 37 | 0 | 0.000 | 175 | 91 | 0.342 |
| 933226 | 13 | 37270408 | 37270408 | A | G | SNP | <i>SERTM1</i> | ENST00000315190 | 1 | 3_prime_untranslated_region | c.*869 | NULL | tier3 | 13 | 0 | 0.000 | 96 | 51 | 0.347 |
| 933226 | 13 | 108518148 | 108518148 | T | A | SNP | <i>FAM155A</i> | ENST00000375915 | -1 | missense | c.797 | p.E266V | tier1 | 21 | 0 | 0.000 | 40 | 27 | 0.403 |
| 933226 | 14 | 75151331 | 75151331 | A | G | SNP | <i>AREL1</i> | ENST00000356357 | -1 | silent | c.69 | NULL | tier1 | 26 | 1 | 0.037 | 53 | 35 | 0.398 |
| 933226 | 14 | 103059220 | 103059220 | G | A | SNP | <i>RCOR1</i> | ENST00000262241 | 1 | 5_prime_untranslated_region | c.-4 | NULL | tier2 | 8 | 0 | 0.000 | 4 | 7 | 0.583 |
| 933226 | 15 | 48056944 | 48056944 | G | A | SNP | <i>SEMA6D</i> | ENST00000316364 | 1 | missense | c.1207 | p.V403I | tier1 | 70 | 0 | 0.000 | 97 | 35 | 0.265 |
| 933226 | 16 | 12297268 | 12297268 | A | G | SNP | <i>SNX29</i> | ENST00000306030 | 1 | intronc | c.523+3728 | e6+3728 | tier3 | 407 | 7 | 0.017 | 740 | 55 | 0.069 |
| 933226 | 16 | 12297294 | 12297294 | T | G | SNP | <i>SNX29</i> | ENST00000306030 | 1 | intronc | c.523+3754 | e6+3754 | tier3 | 389 | 4 | 0.010 | 709 | 34 | 0.046 |
| 933226 | 16 | 72094682 | 72094682 | A | G | SNP | <i>HP</i> | ENST00000355906 | 1 | missense | c.1114 | p.T372A | tier1 | 120 | 0 | 0.000 | 190 | 25 | 0.116 |
| 933226 | 17 | 7578406 | 7578406 | C | T | SNP | <i>TP53</i> | ENST00000269305 | -1 | missense | c.524 | p.R175H | tier1 | 1041 | 3 | 0.003 | 407 | 1037 | 0.716 |
| 933226 | 17 | 18638428 | 18638428 | C | A | SNP | <i>TRIM16L</i> | ENST00000395902 | 1 | missense | c.864 | p.D288E | tier1 | 35 | 0 | 0.000 | 61 | 37 | 0.378 |
| 933226 | 17 | 19643645 | 19643645 | G | A | SNP | <i>ALDH3A1</i> | ENST00000487650 | -1 | 5_prime_untranslated_region | c.-19643645 | NULL | tier3 | 40 | 0 | 0.000 | 61 | 30 | 0.330 |
| 933226 | 18 | 42531913 | 42531913 | G | A | SNP | <i>SETBP1</i> | ENST00000282030 | -1 | missense | c.2608 | p.G870S | tier1 | 532 | 2 | 0.004 | 514 | 310 | 0.376 |
| 933226 | 19 | 1011071 | 1011071 | C | T | SNP | <i>TMEM259</i> | ENST00000591128 | -1 | 5_prime_untranslated_region | c.-1011071 | NULL | tier2 | 72 | 0 | 0.000 | 81 | 31 | 0.277 |
| 933226 | 19 | 1754673 | 1754673 | A | T | SNP | <i>ONECUT3</i> | ENST00000382349 | 1 | missense | c.1012 | p.S338C | tier1 | 55 | 0 | 0.000 | 42 | 25 | 0.373 |
| 933226 | 19 | 49316512 | 49316512 | C | T | SNP | <i>HSD17B14</i> | ENST00000263278 | -1 | missense | c.733 | p.V245M | tier1 | 51 | 0 | 0.000 | 38 | 25 | 0.397 |
| 933226 | 19 | 51380151 | 51380151 | T | A | SNP | <i>KLK2</i> | ENST00000325321 | 1 | missense | c.517 | p.C173S | tier1 | 77 | 0 | 0.000 | 97 | 59 | 0.378 |
| 933226 | 20 | 13269233 | 13269233 | C | T | SNP | <i>ISM1</i> | ENST00000262487 | 1 | silent | c.690 | p.S230 | tier1 | 68 | 0 | 0.000 | 74 | 40 | 0.351 |
| 933226 | 20 | 60499501 | 60499501 | G | A | SNP | <i>CDH4</i> | ENST00000360469 | 1 | missense | c.1738 | p.V580I | tier1 | 54 | 0 | 0.000 | 25 | 24 | 0.490 |
| 933226 | 20 | 62715089 | 62715089 | G | A | SNP | <i>OPRL1</i> | ENST00000336866 | 1 | intronc | c.1+3384 | e0+3384 | tier2 | 6 | 0 | 0.000 | 1 | 6 | 0.857 |
| 933226 | 21 | 11098586 | 11098586 | G | A | SNP | <i>BAGE2</i> | ENST00000470054 | -1 | intronc | c.NULL | NULL | tier4 | 38 | 0 | 0.000 | 61 | 22 | 0.265 |
| 933226 | 21 | 34923934 | 34923934 | T | C | SNP | <i>SON</i> | ENST00000356577 | 1 | silent | c.2397 | p.S799 | tier1 | 175 | 4 | 0.022 | 264 | 39 | 0.128 |
| 933226 | 22 | 16414219 | 16414219 | G | A | SNP | <i>ENSG00000215268</i> | ENST00000417863 | 1 | 3_prime_untranslated_region | c.*16414219 | NULL | tier3 | 147 | 0 | 0.000 | 221 | 75 | 0.253 |
| 933226 | 22 | 42381976 | 42381976 | G | - | DEL | <i>O3.Sep</i> | ENST00000460267 | 1 | 3_prime_untranslated_region | c.*42381976 | NULL | tier3 | 24 | 0 | 0.000 | 43 | 24 | 0.358 |
| 933226 | X | 5821695 | 5821695 | G | A | SNP | <i>NLGN4X</i> | ENST00000381093 | -1 | missense | c.1084 | p.P362S | tier1 | 48 | 0 | 0.000 | 14 | 72 | 0.837 |
| 933226 | X | 43626915 | 43626915 | C | G | SNP | <i>MAOB</i> | ENST00000378069 | -1 | intronc | c.1411-50 | e15-50 | tier4 | 38 | 0 | 0.000 | 34 | 78 | 0.696 |
| 933226 | X | 101624781 | 101624781 | C | T | SNP | <i>NXF2B</i> | ENST00000372752 | -1 | 5_prime_untranslated_region | c.-155 | NULL | tier3 | 132 | 0 | 0.000 | 239 | 147 | 0.380 |
| 933226 | X | 130423302 | 130423302 | C | A | SNP | <i>IGSF1</i> | ENST00000370903 | -1 | 5_prime_untranslated_region | c.-2654 | NULL | tier3 | 21 | 0 | 0.000 | 13 | 54 | 0.806 |
| 944541 | 1 | 50552429 | 50552429 | G | A | SNP | <i>ELAVL4</i> | ENST00000448907 | -1 | intronc | c.18+38575 | e1+38575 | tier3 | 50 | 1 | 0.020 | 128 | 61 | 0.321 |
| 944541 | 2 | 43453288 | 43453288 | G | A | SNP | <i>THADA</i> | ENST00000330266 | -1 | intronc | c.3189-59383 | e20-59383 | tier2 | 60 | 1 | 0.016 | 170 | 108 | 0.386 |
| 944541 | 2 | 97483175 | 97483175 | C | G | SNP | <i>CNNM3</i> | ENST00000305510 | 1 | missense | c.1161 | p.N387K | tier1 | 22 | 0 | 0.000 | 29 | 30 | 0.509 |
| 944541 | 3 | 130145131 | 130145131 | G | T | SNP | <i>COL6A5</i> | ENST00000265379 | 1 | intronc | c.5199+38 | e29+38 | tier3 | 30 | 0 | 0.000 | 62 | 40 | 0.392 |
| 944541 | 3 | 195690360 | 195690360 | G | A | SNP | <i>SDHAP1</i> | ENST00000354559 | -1 | intronc | c.NULL | NULL | tier3 | 32 | 0 | 0.000 | 167 | 46 | 0.214 |
| 944541 | 4 | 44638032 | 44638032 | C | T | SNP | <i>YIPF7</i> | ENST00000332990 | -1 | missense | c.259 | p.A87T | tier1 | 35 | 0 | 0.000 | 69 | 38 | 0.355 |
| 944541 | 5 | 140482151 | 140482151 | C | T | SNP | <i>PCDHB3</i> | ENST00000231130 | 1 | silent | c.1918 | p.L640 | tier1 | 223 | 2 | 0.009 | 467 | 308 | 0.397 |
| 944541 | 6 | 33144473 | 33144473 | G | C | SNP | <i>COL11A2</i> | ENST00000341947 | -1 | intronc | c.2115+25 | e26+25 | tier3 | 49 | 0 | 0.000 | 113 | 66 | 0.369 |
| 944541 | 6 | 43127615 | 43127615 | A | G | SNP | <i>PTK7</i> | ENST00000230419 | 1 | missense | c.2963 | p.D988G | tier1 | 41 | 0 | 0.000 | 79 | 40 | 0.336 |
| 944541 | 7 | 142242469 | 142242469 | G | T | SNP | <i>TRBV5-3</i> | ENST00000390362 | -1 | missense | c.155 | p.S52Y | tier1 | 90 | 1 | 0.011 | 189 | 24 | 0.113 |
| 944541 | 8 | 3253958 | 3253958 | C | T | SNP | <i>CSMD1</i> | ENST00000520002 | -1 | intronc | c.2409-55 | e18-55 | tier3 | 142 | 3 | 0.021 | 356 | 163 | 0.314 |
| 944541 | 8 | 21824261 | 21824261 | A | G | SNP | <i>XPO7</i> | ENST00000434536 | 1 | intronc | c.19-107 | e2-107 | tier3 | 30 | 0 | 0.000 | 132 | 94 | 0.416 |
| 944541 | 8 | 102504720 | 102504720 | C | G | SNP | <i>LOC101927172</i> | ENST00000520268 | -1 | rna | NULL | NULL | tier1 | 83 | 0 | 0.000 | 169 | 108 | 0.390 |
| 944541 | 9 | 27268422 | 27268422 | G | C | SNP | <i>LINC00032</i> | ENST00000425633 | -1 | rna | NULL | NULL | tier1 | 13 | 0 | 0.000 | 31 | 23 | 0.426 |
| 944541 | 9 | 139903352 | 139903352 | - | C | INS | <i>ABCA2</i> | ENST00000265662 | -1 | intronc | c.6931-33_6931-32 | e47-32 | tier3 | 19 | 1 | 0.050 | 45 | 32 | 0.416 |
| 944541 | 11 | 32414262 | 32414262 | C | G | SNP | <i>WT1</i> | ENST00000332351 | -1 | missense | c.1289 | p.R430P | tier1 | 215 | 0 | 0.000 | 743 | 71 | 0.087 |
| 944541 | 11 | 32417909 | 32417910 | - | GA | INS | <i>WT1</i> | ENST00000332351 | -1 | frame_shift_ins | c.1143_1142 | p.A382fs | tier1 | 330 | 0 | 0.000 | 751 | 305 | 0.289 |
| 944541 | 11 | 32417914 | 32417915 | - | T | INS | <i>WT1</i> | ENST00000332351 | -1 | frame_shift_ins | c.1138_1137 | p.R379fs | tier1 | 322 | 0 | 0.000 | 925 | 106 | 0.103 |
| 944541 | 12 | 6697549 | 6697549 | C | T | SNP | <i>CHD4</i> | ENST00000309577 | -1 | missense | c.3380 | p.R1127Q | tier1 | 358 | 0 | 0.000 | 1112 | 95 | 0.079 |
| 944541 | 12 | 96330312 | 96330312 | A | - | DEL | <i>CCDC38</i> | ENST00000344280 | -1 | intronc | c.1-11 | e1-11 | tier3 | 7 | 0 | 0.000 | 21 | 29 | 0.580 |
| 944541 | 12 | 112888189 | 112888189 | G | A | SNP | <i>PTPN11</i> | ENST00000351677 | 1 | missense | c.205 | p.E69K | tier1 | 204 | 3 | 0.015 | 535 | 381 | 0.416 |
| 944541 | 13 | 33017732 | 33017732 | C | T | SNP | <i>N4BP2L2</i> | ENST00000399396 | -1 | silent | c.942 | p.A314 | tier1 | 17 | 0 | 0.000 | 51 | 28 | 0.354 |
| 944541 | 13 | 50115190 | 50115190 | C | T | SNP | <i>RCBTB1</i> | ENST00000258646 | -1 | intronc | c.1325-48 | e10-48 | tier3 | 13 | 0 | 0.000 | 29 | 31 | 0.517 |
| 944541 | 15 | 52427942 | 52427942 | C | A | SNP | <i>GNB5</i> | ENST00000261837 | -1 | silent | c.639 | p.A213 | tier1 | 9 | 0 | 0.000 | 15 | 16 | 0.516 |
| 944541 | 15 | 89386618 | 89386618 | T | A | SNP | <i>ACAN</i> | ENST00000439576 | 1 | missense | c.790 | p.F264I | tier1 | 33 | 0 | 0.000 | 79 | 44 | 0.358 |
| 944541 | 16 | 11541929 | 11541929 | G | A | SNP | <i>LOC101929274</i> | ENST00000344649 | -1 | missense | c.1222 | p.P408S | tier1 | 61 | 0 | 0.000 | 85 | 85 | 0.500 |
| 944541 | 17 | 40322106 | 40322106 | G | A | SNP | <i>KCNH4</i> | ENST00000264661 | -1 | intronc | c.1390+19 | e8+19 | tier3 | 31 | 0 | 0.000 | 79 | 42 | 0.342 |
| 944541 | 17 | 42635299 | 42635299 | G | A | SNP | <i>FZD2</i> | ENST00000315323 | 1 | silent | c.243 | p.K81 | tier1 | 34 | 0 | 0.000 | 51 | 27 | 0.346 |
| 944541 | 18 | 43684347 | 43684347 | A | C | SNP | <i>HAUS1</i> | ENST00000282058 | 1 | 5_prime_untranslated_region | c.-31 | NULL | tier2 | 208 | 0 | 0.000 | 533 | 369 | 0.409 |
| 944541 | 19 | 7964800 | 7964800 | C | T | SNP | <i>LRRC8E</i> | ENST00000306708 | 1 | missense | c.1393 | p.L465F | tier1 | 30 | 0 | 0.000 | 64 | 33 | 0.340 |
| 944541 | 19 | 10812636 | 10812636 | T | C | SNP | <i>QTRT1</i> | ENST00000250237 | 1 | missense | c.254 | p.L85P | tier1 | 45 | 0 | 0.000 | 77 | 36 | 0.316 |
| 944541 | 20 | 56847816 | 56847816 | T | C | SNP | <i>PPP4R1L</i> | ENST00000334187 | -1 | splice_region | c.292+9 | e4+9 | tier3 | 8 | 0 | 0.000 | 36 | 53 | 0.596 |
| 944541 | 22 | 20710847 | 20710847 | C | T | SNP | <i>FAM230A</i> | ENST00000434783 | 1 | missense | c.2579 | p.A860V | tier1 | 142 | 0 | 0.000 | 497 | 29 | 0.055 |
| 944541 | 22 | 22842205 | 22842205 | G | C | SNP | <i>ZNF280B</i> | ENST00000360412 | -1 | missense | c.1519 | p.L507V | tier1 | 45 | 0 | 0.000 | 126 | | |

| | | | | | | | | | | | | | | | | | | | | |
|--------|----|-----------|-----------|----|----|-----|-----------------|-----------------|-----------------|-----------------------------|------------------------|-----------------------|---------|-------|-----|-------|-------|-----|-------|-------|
| 955252 | 5 | 140563408 | 140563408 | A | G | SNP | PCDHB16 | ENST00000361016 | 1 | missense | c.1274 | p.D425G | tier1 | 235 | 1 | 0.004 | 485 | 24 | 0.047 | |
| 955252 | 7 | 157652095 | 157652096 | - | CA | INS | PTPRN2 | ENST00000404321 | -1 | intronic | c.1857+39269-1857+3926 | e12+39269 | tier3 | 133 | 2 | 0.015 | 148 | 17 | 0.103 | |
| 955252 | 9 | 139342941 | 139342941 | T | G | SNP | SEC16A | ENST00000313084 | -1 | missense | c.91 | p.T31P | tier1 | 69 | 0 | 0.000 | 133 | 18 | 0.115 | |
| 955252 | 11 | 67432799 | 67432799 | G | A | SNP | ALDH3B2 | ENST00000349015 | -1 | silent | c.663 | p.R221 | tier1 | 240 | 0 | 0.000 | 370 | 16 | 0.041 | |
| 955252 | 12 | 30790136 | 30790137 | - | A | INS | IPO8 | ENST00000256079 | -1 | intronic | c.2490-16_2490-15 | e22-15 | tier3 | 18 | 0 | 0.000 | 49 | 25 | 0.338 | |
| 955252 | 20 | 29628205 | 29628205 | T | A | SNP | FRG1B | ENST00000278882 | 1 | intronic | c.229-22 | e4-22 | tier3 | 469 | 7 | 0.015 | 1948 | 99 | 0.048 | |
| 955252 | 20 | 29628229 | 29628229 | G | T | SNP | FRG1B | ENST00000479318 | 1 | nonsense | c.115 | p.E39* | tier1 | 534 | 8 | 0.015 | 2248 | 106 | 0.045 | |
| 955252 | 22 | 20402385 | 20402386 | GC | - | DEL | PI4KAP1 | ENST00000416922 | -1 | rna | NULL | NULL | tier1 | 17 | 0 | 0.000 | 12 | 9 | 0.429 | |
| 955252 | 22 | 34019122 | 34019122 | T | - | DEL | LARGE | ENST00000354992 | -1 | intronic | c.491+3106 | e3+3106 | tier3 | 108 | 1 | 0.009 | 284 | 32 | 0.101 | |
| 988638 | 1 | 46505876 | 46505876 | G | A | SNP | PIK3R3 | ENST00000262741 | -1 | 3_prime_untranslated_region | c.*3469 | NULL | tier3 | 182 | 0 | 0.000 | 193 | 34 | 0.150 | |
| 988638 | 1 | 161519606 | 161519606 | A | G | SNP | FCGR3A | ENST00000367969 | -1 | missense | c.29 | p.F10S | tier1 | 128 | 0 | 0.000 | 123 | 26 | 0.173 | |
| 988638 | 1 | 231956089 | 231956090 | - | - | T | INS | DISC1 | ENST00000439617 | 1 | intronic | c.1981+1826_1981+1825 | e9+1826 | tier3 | 122 | 0 | 0.000 | 186 | 20 | 0.097 |
| 988638 | 1 | 233091444 | 233091444 | G | A | SNP | NTPCR | ENST00000366628 | 1 | missense | c.176 | p.R59Q | tier1 | 124 | 0 | 0.000 | 163 | 41 | 0.200 | |
| 988638 | 2 | 102484410 | 102484410 | C | T | SNP | MAP4K4 | ENST00000498066 | 1 | rna | NULL | NULL | tier1 | 104 | 0 | 0.000 | 86 | 10 | 0.104 | |
| 988638 | 2 | 128387436 | 128387436 | A | G | SNP | MYO7B | ENST00000389524 | 1 | intronic | c.4728+35 | e33+35 | tier3 | 108 | 0 | 0.000 | 108 | 15 | 0.122 | |
| 988638 | 2 | 135957920 | 135957920 | T | C | SNP | ZRANB3 | ENST00000264159 | -1 | missense | c.3232 | p.K1078E | tier1 | 94 | 0 | 0.000 | 116 | 34 | 0.227 | |
| 988638 | 2 | 175689250 | 175689250 | A | - | DEL | CHN1 | ENST00000409900 | -1 | splice_region_del | c.628-4 | e8-4 | tier3 | 44 | 0 | 0.000 | 58 | 11 | 0.159 | |
| 988638 | 2 | 192257750 | 192257750 | G | C | SNP | MYO1B | ENST00000304164 | 1 | intronic | c.2077-49 | e19-49 | tier3 | 84 | 0 | 0.000 | 112 | 15 | 0.118 | |
| 988638 | 2 | 202467998 | 202467998 | C | A | SNP | ALS2CR11 | ENST00000286195 | -1 | missense | c.319 | p.V107F | tier1 | 76 | 0 | 0.000 | 128 | 39 | 0.234 | |
| 988638 | 3 | 32997048 | 32997048 | T | A | SNP | CCR4 | ENST00000330953 | 1 | 3_prime_untranslated_region | c.*1051 | NULL | tier3 | 146 | 3 | 0.020 | 242 | 29 | 0.107 | |
| 988638 | 3 | 55061774 | 55061774 | C | T | SNP | CACNA2D3 | ENST00000288197 | 1 | intronic | c.2987+9430 | e35+9430 | tier2 | 91 | 0 | 0.000 | 101 | 21 | 0.172 | |
| 988638 | 6 | 55990914 | 55990914 | G | T | SNP | COL21A1 | ENST00000244728 | -1 | missense | c.1576 | p.H526N | tier1 | 96 | 0 | 0.000 | 154 | 56 | 0.265 | |
| 988638 | 7 | 20826548 | 20826548 | G | T | SNP | SP8 | ENST00000418710 | -1 | 5_prime_flanking_region | c.-131 | NULL | tier2 | 110 | 0 | 0.000 | 91 | 17 | 0.157 | |
| 988638 | 7 | 74225436 | 74225436 | T | C | SNP | GTF2IRD2 | ENST00000405086 | -1 | missense | c.799 | p.M267V | tier1 | 142 | 2 | 0.014 | 177 | 118 | 0.400 | |
| 988638 | 8 | 94937549 | 94937550 | - | T | INS | PDP1 | ENST00000396200 | 1 | 3_prime_untranslated_region | c.*1648 | NULL | tier4 | 27 | 0 | 0.000 | 55 | 17 | 0.236 | |
| 988638 | 9 | 44874097 | 44874097 | G | A | SNP | ENSG00000231454 | ENST00000450729 | 1 | 3_prime_untranslated_region | c.*44874097 | NULL | tier3 | 190 | 1 | 0.005 | 224 | 27 | 0.108 | |
| 988638 | 9 | 128083745 | 128083745 | G | T | SNP | GAPVD1 | ENST00000394105 | 1 | nonsense | c.1636 | p.G546* | tier1 | 76 | 0 | 0.000 | 162 | 34 | 0.174 | |
| 988638 | 10 | 51829388 | 51829388 | C | T | SNP | FAM21A | ENST00000282633 | 1 | missense | c.208 | p.R70W | tier1 | 196 | 0 | 0.000 | 234 | 46 | 0.164 | |
| 988638 | 11 | 5842368 | 5842368 | G | C | SNP | OR52N2 | ENST00000317037 | 1 | missense | c.803 | p.G268A | tier1 | 84 | 0 | 0.000 | 130 | 26 | 0.166 | |
| 988638 | 11 | 89531801 | 89531801 | C | A | SNP | TRIM49 | ENST00000329758 | -1 | splice_region | c.860-4 | e6-4 | tier3 | 57 | 0 | 0.000 | 140 | 29 | 0.171 | |
| 988638 | 12 | 40815079 | 40815079 | G | A | SNP | MUC19 | ENST00000471230 | 1 | rna | NULL | NULL | tier1 | 115 | 0 | 0.000 | 296 | 55 | 0.157 | |
| 988638 | 16 | 89650436 | 89650436 | G | A | SNP | CPNE7 | ENST00000268720 | 1 | missense | c.658 | p.V220M | tier1 | 96 | 1 | 0.010 | 95 | 21 | 0.181 | |
| 988638 | 17 | 44074056 | 44074056 | G | T | SNP | MAPT | ENST00000344290 | 1 | intronic | c.1827+26 | e10+26 | tier3 | 70 | 0 | 0.000 | 41 | 18 | 0.305 | |
| 988638 | 17 | 74732959 | 74732959 | G | T | SNP | SRSF2 | ENST00000359995 | -1 | missense | c.284 | p.P95H | tier1 | 392 | 1 | 0.003 | 397 | 70 | 0.150 | |
| 988638 | 18 | 42531907 | 42531907 | G | A | SNP | SETBP1 | ENST00000282030 | 1 | missense | c.2602 | p.D868N | tier1 | 576 | 0 | 0.000 | 629 | 106 | 0.144 | |
| 988638 | 19 | 7675544 | 7675544 | G | C | SNP | CAMSAP3 | ENST00000446248 | 1 | intronic | c.982-42 | e9-42 | tier3 | 102 | 2 | 0.019 | 88 | 20 | 0.184 | |
| 988638 | 19 | 10561262 | 10561262 | T | - | DEL | PDE4A | ENST00000352831 | 1 | intronic | c.621-17 | e5-17 | tier3 | 73 | 0 | 0.000 | 86 | 13 | 0.131 | |
| 988638 | 19 | 35265460 | 35265460 | C | T | SNP | ZNF599 | ENST00000329285 | -1 | 5_prime_flanking_region | c.-1714 | NULL | tier4 | 47 | 0 | 0.000 | 53 | 10 | 0.159 | |
| 988638 | 20 | 31022441 | 31022442 | - | G | INS | ASXL1 | ENST00000375687 | 1 | frame_shift_ins | c.1926_1927 | p.G645fs | tier1 | 805 | 6 | 0.007 | 781 | 132 | 0.145 | |
| 988638 | 20 | 44815531 | 44815531 | G | A | SNP | CDH22 | ENST00000372262 | -1 | silent | c.1479 | p.N493 | tier1 | 88 | 0 | 0.000 | 92 | 18 | 0.164 | |
| 988638 | X | 14338161 | 14338161 | C | T | SNP | - | - | - | - | - | - | tier3 | 42 | 0 | 0.000 | 48 | 25 | 0.343 | |
| 988638 | X | 73071846 | 73071846 | A | - | DEL | XIST | ENST00000429829 | -1 | rna | NULL | NULL | tier1 | 80 | 1 | 0.012 | 97 | 16 | 0.142 | |
| 991140 | 1 | 142653596 | 142653596 | C | T | SNP | ENSG00000266657 | ENST00000580249 | 1 | rna | NULL | NULL | tier1 | 211 | 0 | 0.000 | 423 | 50 | 0.106 | |
| 991140 | 1 | 152279155 | 152279155 | C | - | DEL | FLG | ENST00000368799 | -1 | frame_shift_del | c.8207 | p.G2736fs | tier1 | 374 | 0 | 0.000 | 691 | 94 | 0.120 | |
| 991140 | 1 | 181702771 | 181702771 | C | T | SNP | CACNA1E | ENST00000367573 | 1 | silent | c.3147 | p.S1049 | tier1 | 268 | 0 | 0.000 | 491 | 126 | 0.204 | |
| 991140 | 1 | 223938499 | 223938499 | C | T | SNP | CAPN2 | ENST00000295006 | 1 | intronic | c.814-93 | e7-93 | tier3 | 40 | 0 | 0.000 | 92 | 22 | 0.193 | |
| 991140 | 1 | 235926068 | 235926068 | C | T | SNP | LYST | ENST00000389793 | -1 | missense | c.6205 | p.G2069R | tier1 | 51 | 0 | 0.000 | 132 | 64 | 0.327 | |
| 991140 | 2 | 97833261 | 97833261 | T | A | SNP | ANKRD36 | ENST00000420699 | 1 | intronic | c.1532-53 | e1-53 | tier3 | 186 | 4 | 0.021 | 666 | 63 | 0.086 | |
| 991140 | 2 | 166187904 | 166187904 | T | C | SNP | SCN2A | ENST00000283256 | 1 | silent | c.2214 | p.I738 | tier1 | 217 | 1 | 0.005 | 503 | 146 | 0.224 | |
| 991140 | 2 | 189997806 | 189997806 | G | A | SNP | MIR3129 | ENST00000581095 | -1 | rna | NULL | NULL | tier1 | 55 | 1 | 0.018 | 106 | 37 | 0.259 | |
| 991140 | 3 | 116163747 | 116163747 | C | T | SNP | LSAMP | ENST00000490035 | -1 | silent | c.132 | p.R44 | tier1 | 67 | 0 | 0.000 | 89 | 43 | 0.326 | |
| 991140 | 3 | 132201248 | 132201248 | C | T | SNP | DNAJC13 | ENST00000260818 | 1 | intronic | c.2979+74 | e26+74 | tier3 | 62 | 2 | 0.031 | 105 | 44 | 0.295 | |
| 991140 | 4 | 16503736 | 16503737 | - | A | INS | LDB2 | ENST00000509803 | -1 | 5_prime_untranslated_region | c.-16503736 | NULL | tier3 | 66 | 0 | 0.000 | 121 | 18 | 0.130 | |
| 991140 | 4 | 36093672 | 36093672 | T | A | SNP | ARAP2 | ENST00000303965 | -1 | intronic | c.4286-30 | e27-30 | tier3 | 170 | 1 | 0.006 | 474 | 120 | 0.202 | |
| 991140 | 6 | 147006987 | 147006987 | C | G | SNP | ADGB | ENST00000397944 | 1 | intronic | c.1311+23 | e10+23 | tier3 | 39 | 0 | 0.000 | 158 | 30 | 0.159 | |
| 991140 | 7 | 34889221 | 34889221 | C | A | SNP | NPSR1 | ENST00000381539 | 1 | missense | c.1170 | p.N390K | tier1 | 47 | 0 | 0.000 | 49 | 30 | 0.380 | |
| 991140 | 8 | 97506480 | 97506480 | G | A | SNP | SDC2 | ENST00000520233 | 1 | rna | NULL | NULL | tier1 | 142 | 0 | 0.000 | 367 | 132 | 0.265 | |
| 991140 | 8 | 142166004 | 142166004 | G | T | SNP | DENND3 | ENST00000262585 | 1 | missense | c.891 | p.K297N | tier1 | 131 | 0 | 0.000 | 216 | 73 | 0.253 | |
| 991140 | 8 | 142517302 | 142517302 | G | C | SNP | MROH5 | ENST00000521053 | -1 | 5_prime_untranslated_region | c.-53 | NULL | tier3 | 273 | 1 | 0.004 | 459 | 155 | 0.251 | |
| 991140 | 9 | 133254693 | 133254693 | G | A | SNP | HMCN2 | ENST00000480829 | 1 | 5_prime_flanking_region | c.-19336 | NULL | tier3 | 56 | 0 | 0.000 | 109 | 31 | 0.220 | |
| 991140 | 9 | 135521201 | 135521201 | C | A | SNP | DDX31 | ENST00000372159 | -1 | intronic | c.1695+81 | e13+81 | tier3 | 42 | 0 | 0.000 | 61 | 26 | 0.299 | |
| 991140 | 11 | 19735518 | 19735518 | G | A | SNP | NAV2 | ENST00000396087 | 1 | splice_region | c.267+10 | e1+10 | tier2 | 35 | 0 | 0.000 | 53 | 13 | 0.197 | |
| 991140 | 11 | 30034426 | 30034426 | A | G | SNP | KCNA4 | ENST00000328224 | -1 | 5_prime_untranslated_region | c.-201 | NULL | tier3 | 123 | 0 | 0.000 | 195 | 71 | 0.266 | |
| 991140 | 12 | 15749242 | 15749242 | C | T | SNP | PTPRO | ENST00000544706 | 1 | rna | NULL | NULL | tier1 | 138 | 0 | 0.000 | 149 | 71 | 0.323 | |
| 991140 | 12 | 31297989 | 31297989 | C | T | SNP | LOC100509445 | ENST00000398963 | -1 | splice_site | c.NULL | NULL | tier1 | 294 | 0 | 0.000 | 743 | 27 | 0.035 | |
| 991140 | 12 | 48378352 | 48378352 | G | A | SNP | COL2A1 | ENST00000380518 | -1 | silent | c.1864 | p.L622 | tier1 | 38 | 0 | 0.000 | 63 | 22 | 0.259 | |
| 991140 | 15 | 45059668 | 45059668 | C | T | SNP | TRIM69 | ENST00000329464 | 1 | missense | c.1201 | p.R401W | tier1 | 72 | 0 | 0.000 | 126 | 49 | 0.280 | |
| 991140 | 16 | 21974018 | 21974018 | G | A | SNP | UQCRC2 | ENST00000563711 | 1 | 3_prime_untranslated_region | c.*21974018 | NULL | tier3 | 75 | 0 | 0.000 | 170 | 64 | 0.274 | |
| 991140 | 16 | 70923652 | 70923652 | G | A | SNP | HYDIN | ENST00000393 | | | | | | | | | | | | |

| | | | | | | | | | | | | | | | | | | | |
|--------|----|----------|----------|---|---|-----|--------|-----------------|----|----------|----------|---------|-------|-----|---|-------|-----|-----|-------|
| 991140 | 19 | 44680481 | 44680481 | G | T | SNP | ZNF226 | ENST00000337433 | 1 | missense | c.1066 | p.V356F | tier1 | 76 | 2 | 0.026 | 153 | 46 | 0.231 |
| 991140 | 19 | 54395724 | 54395724 | A | G | SNP | PRKCG | ENST00000540413 | 1 | intronic | c.687-39 | e7-39 | tier3 | 102 | 1 | 0.010 | 213 | 62 | 0.224 |
| 991140 | 20 | 57464246 | 57464246 | C | A | SNP | GNAS | ENST00000464788 | 1 | rna | NULL | NULL | tier1 | 20 | 0 | 0.000 | 31 | 12 | 0.279 |
| 991140 | 21 | 41550865 | 41550865 | G | A | SNP | DSCAM | ENST00000400454 | -1 | missense | c.2936 | p.A979V | tier1 | 469 | 0 | 0.000 | 876 | 283 | 0.244 |

All genome coordinates use NCBI Build 37.

UPN, unique patient number.

Chr, chromosome; Var, variant; Ref, reference; VAF, variant allele frequency, listed as a fraction.

The VAF calculation includes non-supporting reads.

Tier, defined in Walter et. al., NEJM. 2012 Mar 22;366(12):1090-8.

Table S4. Validated Somatic Mutations by Ultra-Deep Error-Corrected Sequencing

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|---------------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 611 | 1 | 53547545 | 53547545 | C | T | <i>PODN</i> | intronic | c.1806-108 | e10-108 | tier3 | 8982 | 895 | 887 | 5 | 0.006 | 23761 | 3272 | 2325 | 923 | 0.284 | 21120 | 2119 | 1473 | 622 | 0.297 | 35709 | 4411 | 4405 | 4 | 0.000 | 25601 | 3509 | 3504 | 0 | 0.000 |
| 611 | 2 | 25464481 | 25464481 | G | A | <i>DNMT3A</i> | nonsense | c.2032 | p.Q678* | tier1 | 2028 | 226 | 225 | 1 | 0.004 | 4329 | 790 | 747 | 41 | 0.052 | 4130 | 517 | 491 | 24 | 0.047 | 3049 | 490 | 487 | 3 | 0.006 | 3039 | 452 | 451 | 0 | 0.000 |
| 611 | 3 | 31495819 | 31495819 | G | A | <i>THRAP3P1</i> | 3_prime_untranslated_region | c.*31495819 | NULL | tier3 | 3035 | 365 | 365 | 0 | 0.000 | 3102 | 654 | 500 | 152 | 0.233 | 1732 | 188 | 138 | 50 | 0.266 | 4527 | 844 | 842 | 1 | 0.000 | 4125 | 0 | 0 | 0 | NA |
| 611 | 3 | 64644370 | 64644370 | G | A | <i>ADAMTS9</i> | silent | c.777 | p.S259 | tier1 | 3123 | 364 | 363 | 0 | 0.000 | 3968 | 649 | 567 | 77 | 0.120 | 6484 | 898 | 790 | 107 | 0.119 | 8990 | 1366 | 1364 | 0 | 0.000 | 7793 | 1287 | 1285 | 1 | 0.000 |
| 611 | 3 | 130720537 | 130720537 | T | A | <i>ATP2C1</i> | 3_prime_untranslated_region | c.*343 | NULL | tier4 | 288 | 17 | 17 | 0 | 0.000 | 425 | 69 | 45 | 24 | 0.348 | 254 | 17 | 10 | 7 | 0.412 | 756 | 94 | 93 | 0 | 0.000 | 530 | 78 | 77 | 0 | 0.000 |
| 611 | 4 | 187540539 | 187540539 | C | T | <i>FAT1</i> | missense | c.7201 | p.E2401K | tier1 | 21282 | 2384 | 2364 | 9 | 0.004 | 32719 | 4299 | 2970 | 1289 | 0.303 | 24310 | 2850 | 2007 | 820 | 0.290 | 52691 | 6679 | 6675 | 1 | 0.000 | 33041 | 4166 | 4160 | 1 | 0.000 |
| 611 | 6 | 33037020 | 33037020 | G | A | <i>HLA-DPA1</i> | missense | c.404 | p.T135I | tier1 | 2634 | 286 | 282 | 1 | 0.004 | 18565 | 2550 | 1805 | 734 | 0.289 | 13574 | 1591 | 1098 | 483 | 0.306 | 31106 | 3993 | 3991 | 0 | 0.000 | 21027 | 2758 | 2753 | 0 | 0.000 |
| 611 | 6 | 43206066 | 43206066 | C | G | <i>TBKI</i> | 5_prime_flanking_region | c.-8333 | NULL | tier4 | 5847 | 541 | 536 | 3 | 0.006 | 15507 | 2392 | 1662 | 697 | 0.295 | 16426 | 1814 | 1285 | 520 | 0.288 | 19550 | 2486 | 2476 | 0 | 0.000 | 13342 | 1837 | 1821 | 0 | 0.000 |
| 611 | 7 | 45753309 | 45753309 | C | T | <i>ADCY1</i> | silent | c.3075 | p.H102S | tier1 | 21752 | 2553 | 2548 | 0 | 0.000 | 9088 | 1296 | 1201 | 91 | 0.070 | 7784 | 909 | 870 | 38 | 0.042 | 31696 | 4296 | 4293 | 0 | 0.000 | 19415 | 2938 | 2929 | 0 | 0.000 |
| 611 | 7 | 100549883 | 100549883 | G | A | <i>LOC100131514</i> | missense | c.464 | p.S155N | tier1 | 12274 | 1185 | 1181 | 2 | 0.002 | 14429 | 1813 | 1189 | 602 | 0.336 | 14811 | 1654 | 1141 | 505 | 0.307 | 28000 | 3067 | 3062 | 0 | 0.000 | 20095 | 2213 | 2207 | 0 | 0.000 |
| 611 | 8 | 17944578 | 17944578 | C | T | <i>LOC101929066</i> | rna | NULL | NULL | tier1 | 2684 | 191 | 190 | 1 | 0.005 | 4051 | 574 | 409 | 161 | 0.282 | 5753 | 548 | 386 | 158 | 0.290 | 6560 | 884 | 877 | 1 | 0.000 | 4619 | 670 | 665 | 0 | 0.000 |
| 611 | 8 | 72127533 | 72127533 | C | T | <i>EYA1</i> | intronic | c.1597+89 | e14+89 | tier3 | 1549 | 105 | 104 | 0 | 0.000 | 1463 | 177 | 121 | 52 | 0.301 | 2375 | 229 | 148 | 75 | 0.336 | 4084 | 459 | 455 | 0 | 0.000 | 2230 | 279 | 275 | 0 | 0.000 |
| 611 | 8 | 116424437 | 116424437 | G | C | <i>TRPS1</i> | 3_prime_untranslated_region | c.*1814 | NULL | tier3 | 445 | 34 | 34 | 0 | 0.000 | 738 | 133 | 99 | 33 | 0.250 | 812 | 69 | 49 | 20 | 0.290 | 993 | 143 | 142 | 0 | 0.000 | 837 | 0 | 0 | 0 | NA |
| 611 | 9 | 103338826 | 103338826 | C | T | <i>TMEFF1</i> | nonsense | c.1087 | p.R363* | tier1 | 9353 | 999 | 983 | 10 | 0.010 | 123 | 2 | 1 | 1 | 0.500 | 14226 | 1630 | 1016 | 601 | 0.372 | 27451 | 3387 | 3382 | 4 | 0.000 | 19021 | 2398 | 2388 | 4 | 0.000 |
| 611 | 9 | 131502394 | 131502394 | G | A | <i>ZER1</i> | silent | c.1858 | p.L620 | tier1 | 13177 | 1286 | 1276 | 7 | 0.005 | 14496 | 1927 | 1487 | 430 | 0.224 | 11271 | 1139 | 883 | 255 | 0.224 | 38977 | 4802 | 4799 | 1 | 0.000 | 22185 | 2704 | 2702 | 0 | 0.000 |
| 611 | 11 | 6648113 | 6648113 | C | T | <i>DCHS1</i> | missense | c.6157 | p.V2053M | tier1 | 13448 | 1544 | 1529 | 9 | 0.006 | 18292 | 2787 | 2385 | 394 | 0.142 | 13078 | 1624 | 1304 | 316 | 0.195 | 27951 | 3995 | 3992 | 1 | 0.000 | 21789 | 3272 | 3265 | 0 | 0.000 |
| 611 | 11 | 25004690 | 25004690 | A | G | <i>LUZP2</i> | missense | c.616 | p.M206V | tier1 | 42802 | 4243 | 4231 | 2 | 0.000 | 38780 | 4819 | 4449 | 348 | 0.073 | 46612 | 4847 | 4425 | 399 | 0.083 | 85559 | 10251 | 10249 | 0 | 0.000 | 58070 | 7108 | 7089 | 0 | 0.000 |
| 611 | 11 | 125298875 | 125298875 | C | T | <i>PKNOX2</i> | intronic | c.937-33 | e8-33 | tier2 | 23043 | 2669 | 2659 | 4 | 0.002 | 21617 | 2936 | 2431 | 492 | 0.168 | 27834 | 3446 | 2883 | 532 | 0.156 | 55863 | 7646 | 7643 | 0 | 0.000 | 36314 | 5239 | 5229 | 0 | 0.000 |
| 611 | 12 | 53553483 | 53553483 | T | C | <i>CSAD</i> | missense | c.1313 | p.N438S | tier1 | 7170 | 932 | 929 | 3 | 0.003 | 20403 | 2365 | 2345 | 879 | 0.267 | 19174 | 2587 | 1818 | 748 | 0.262 | 26492 | 3963 | 3962 | 0 | 0.000 | 18518 | 2758 | 2744 | 0 | 0.000 |
| 611 | 14 | 23072764 | 23072764 | T | A | <i>ABHD4</i> | 3_prime_untranslated_region | c.*23072764 | NULL | tier3 | 15259 | 1488 | 1480 | 8 | 0.005 | 21294 | 3178 | 2359 | 777 | 0.248 | 11665 | 1264 | 956 | 303 | 0.241 | 25949 | 3426 | 3424 | 0 | 0.000 | 20752 | 2877 | 2866 | 0 | 0.000 |
| 611 | 14 | 104216205 | 104216205 | G | A | <i>PPP1R13B</i> | missense | c.895 | p.R299C | tier1 | 5783 | 516 | 515 | 0 | 0.000 | 7995 | 1079 | 757 | 317 | 0.295 | 7698 | 833 | 589 | 241 | 0.290 | 10755 | 1235 | 1231 | 2 | 0.000 | 7363 | 927 | 925 | 0 | 0.000 |
| 611 | 15 | 66008017 | 66008017 | G | A | <i>DENND4A</i> | intronic | c.1808-56 | e12-56 | tier4 | 832 | 102 | 102 | 0 | 0.000 | 1718 | 327 | 236 | 89 | 0.274 | 2125 | 293 | 237 | 56 | 0.191 | 2938 | 509 | 508 | 0 | 0.000 | 2159 | 356 | 356 | 0 | 0.000 |
| 611 | 16 | 3071729 | 3071729 | G | A | <i>TNFRSF12A</i> | intronic | c.334+39 | e3+39 | tier2 | 15049 | 1546 | 1536 | 6 | 0.004 | 26664 | 3781 | 2611 | 1133 | 0.303 | 23166 | 2648 | 1856 | 765 | 0.292 | 34221 | 4344 | 4340 | 0 | 0.000 | 23753 | 3174 | 3167 | 0 | 0.000 |
| 611 | 17 | 3661144 | 3661144 | G | A | <i>ITGA6</i> | silent | c.876 | p.I292 | tier1 | 7346 | 765 | 764 | 0 | 0.000 | 13405 | 2318 | 1752 | 544 | 0.249 | 6899 | 821 | 629 | 183 | 0.225 | 19367 | 2754 | 2752 | 1 | 0.000 | 14580 | 2149 | 2149 | 0 | 0.000 |
| 611 | 17 | 26087697 | 26087697 | G | T | <i>NOS2</i> | missense | c.2962 | p.P988T | tier1 | 40614 | 4444 | 4420 | 16 | 0.004 | 49671 | 6438 | 4380 | 1988 | 0.312 | 36749 | 3875 | 2631 | 1191 | 0.312 | 92973 | 11598 | 11592 | 1 | 0.000 | 56334 | 6900 | 6884 | 0 | 0.000 |
| 611 | 18 | 42531913 | 42531913 | G | A | <i>SETBP1</i> | missense | c.2608 | p.G870S | tier1 | 29437 | 3285 | 3261 | 14 | 0.004 | 36346 | 4819 | 3395 | 1393 | 0.291 | 33568 | 3669 | 2475 | 1139 | 0.315 | 61869 | 7748 | 7737 | 5 | 0.000 | 43134 | 5370 | 5361 | 1 | 0.000 |
| 611 | 19 | 39787541 | 39787541 | C | T | <i>IFNL1</i> | intronic | c.249+19 | e2+19 | tier3 | 12877 | 1445 | 1436 | 5 | 0.003 | 14639 | 2041 | 1449 | 576 | 0.284 | 14489 | 1615 | 1077 | 532 | 0.331 | 24841 | 3192 | 3190 | 0 | 0.000 | 17614 | 2334 | 2329 | 0 | 0.000 |
| 611 | 21 | 33684165 | 33684165 | A | G | <i>MRAP</i> | missense | c.377 | p.E126G | tier1 | 21623 | 2546 | 2534 | 10 | 0.004 | 24384 | 3554 | 2469 | 1057 | 0.300 | 23550 | 2879 | 1997 | 839 | 0.296 | 46032 | 6446 | 6441 | 1 | 0.000 | 29013 | 4082 | 4073 | 0 | 0.000 |
| 611 | 22 | 23922123 | 23922123 | G | A | <i>IGLL1</i> | intronic | c.206+49 | e1+49 | tier2 | 5101 | 490 | 463 | 24 | 0.049 | 12689 | 1827 | 1364 | 433 | 0.241 | 14202 | 1363 | 917 | 435 | 0.322 | 27798 | 3368 | 3350 | 8 | 0.002 | 17682 | 2407 | 2385 | 3 | 0.001 |
| 611 | X | 9757836 | 9757836 | C | T | <i>SHROOM2</i> | intronic | c.165+3086 | e1+3086 | tier3 | 3004 | 273 | 273 | 0 | 0.000 | 6031 | 821 | 665 | 149 | 0.183 | 7411 | 693 | 556 | 135 | 0.195 | 21649 | 2484 | 2483 | 0 | 0.000 | 14127 | 1764 | 1757 | 0 | 0.000 |
| 130968 | 1 | 6101941 | 6101941 | T | A | <i>KCNAB2</i> | splice_region | c.119+9 | e2+9 | tier2 | 9117 | 1310 | 1252 | 54 | 0.041 | 5823 | 791 | 466 | 313 | 0.402 | 6722 | 729 | 426 | 297 | 0.411 | 7561 | 1246 | 1000 | 237 | 0.192 | 11513 | 1814 | 1225 | 575 | 0.319 |
| 130968 | 1 | 115258747 | 115258747 | C | T | <i>NRAS</i> | missense | c.35 | p.G12D | tier1 | 22176 | 2785 | 2772 | 8 | 0.003 | 16437 | 1825 | 1544 | 239 | 0.134 | 19940 | 2099 | 1435 | 552 | 0.278 | 14718 | 1972 | 1785 | 120 | 0.063 | 29726 | 4004 | 4000 | 1 | 0.000 |
| 130968 | 1 | 154960473 | 154960473 | C | A | <i>FLAD1</i> | rna | NULL | NULL | tier1 | 26444 | 3393 | 3384 | 5 | 0.001 | 20965 | 2676 | 2324 | 340 | 0.128 | 18949 | 2200 | 1638 | 550 | 0.251 | 14463 | 2251 | 2202 | 48 | 0.021 | 36546 | 5275 | 5274 | 0 | 0.000 |
| 130968 | 1 | 160144523 | 160144523 | C | T | <i>ATP1A4</i> | missense | c.2297 | p.T766M | tier1 | 2849 | 369 | 362 | 7 | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|------------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 145094 | 21 | 44324363 | 44324363 | G | A | <i>NDUVF3</i> | missense | c.1241 | p.G414E | tier1 | 64410 | 7969 | 7908 | 54 | 0.007 | 24417 | 3048 | 1615 | 1419 | 0.468 | 40570 | 5090 | 3905 | 1167 | 0.230 | 694415 | 26990 | 26965 | 18 | 0.001 | 218769 | 18522 | 18445 | 61 | 0.003 |
| 145094 | X | 18845595 | 18845595 | A | T | <i>PPEF1</i> | missense | c.1952 | p.N651I | tier1 | 23300 | 3453 | 3410 | 41 | 0.012 | 12714 | 1690 | 93 | 1596 | 0.945 | 22525 | 3192 | 1768 | 1418 | 0.445 | 403663 | 20214 | 20160 | 36 | 0.002 | 140265 | 14019 | 13687 | 306 | 0.022 |
| 147444 | 1 | 34164429 | 34164429 | G | T | <i>CSMD2</i> | missense | c.3849 | p.S1283R | tier1 | 10290 | 1129 | 1128 | 0 | 0.000 | 16483 | 2413 | 1916 | 485 | 0.202 | 6187 | 946 | 590 | 354 | 0.375 | 15888 | 2354 | 2354 | 0 | 0.000 | 21486 | 0 | 0 | 0 | NA |
| 147444 | 1 | 38262323 | 38262323 | C | A | <i>MANEAL</i> | intronic | c.661-93 | e3-93 | tier3 | 2165 | 275 | 275 | 0 | 0.000 | 7229 | 1513 | 1130 | 379 | 0.251 | 462 | 97 | 63 | 34 | 0.351 | 4264 | 891 | 890 | 0 | 0.000 | 3351 | 714 | 714 | 0 | 0.000 |
| 147444 | 1 | 89480937 | 89480937 | G | A | <i>GBP3</i> | intronic | c.318+33 | e2+33 | tier3 | 7300 | 649 | 647 | 2 | 0.003 | 11614 | 1649 | 1280 | 359 | 0.219 | 9529 | 1155 | 783 | 363 | 0.317 | 13756 | 1811 | 1803 | 0 | 0.000 | 16354 | 1990 | 1986 | 0 | 0.000 |
| 147444 | 1 | 163306613 | 163306613 | C | T | <i>NUF2</i> | missense | c.410 | p.T137M | tier1 | 3443 | 301 | 301 | 0 | 0.000 | 4347 | 572 | 427 | 145 | 0.254 | 5278 | 633 | 411 | 221 | 0.350 | 5388 | 705 | 701 | 2 | 0.000 | 6979 | 866 | 864 | 1 | 0.000 |
| 147444 | 1 | 201016781 | 201016781 | C | T | <i>CACNA1S</i> | intronic | c.4442-27 | e37-27 | tier3 | 16351 | 1886 | 1885 | 0 | 0.000 | 25629 | 3574 | 2693 | 870 | 0.244 | 14418 | 1908 | 1146 | 754 | 0.397 | 25369 | 3725 | 3717 | 1 | 0.000 | 36926 | 5116 | 5106 | 2 | 0.000 |
| 147444 | 1 | 226061960 | 226061960 | G | A | <i>TMEM63A</i> | intronic | c.266+28 | e2+28 | tier3 | 16399 | 2317 | 2312 | 5 | 0.002 | 29734 | 3693 | 2795 | 881 | 0.240 | 15664 | 2217 | 1412 | 799 | 0.361 | 30394 | 4252 | 4244 | 3 | 0.001 | 41701 | 5988 | 5975 | 2 | 0.000 |
| 147444 | 2 | 42119915 | 42119915 | C | A | <i>LOC388942</i> | rna | NULL | NULL | tier1 | 15872 | 1949 | 1949 | 0 | 0.000 | 33223 | 4789 | 3666 | 1062 | 0.225 | 13828 | 1925 | 1192 | 728 | 0.379 | 29304 | 4375 | 4363 | 1 | 0.000 | 43219 | 6097 | 6090 | 1 | 0.000 |
| 147444 | 2 | 42936013 | 42936013 | G | T | <i>MTA3</i> | splice_site | c.1303-1 | e14-1 | tier1 | 5984 | 0 | 0 | 0 | 0.000 | 1321 | 196 | 141 | 54 | 0.277 | 2977 | 393 | 226 | 166 | 0.423 | 7442 | 921 | 920 | 0 | 0.000 | 2843 | 354 | 353 | 0 | 0.000 |
| 147444 | 2 | 128262356 | 128262356 | T | C | <i>IWS1</i> | missense | c.1123 | p.M375V | tier1 | 1566 | 0 | 0 | 0 | 0.000 | 5088 | 1002 | 775 | 218 | 0.220 | 1101 | 192 | 125 | 63 | 0.335 | 3646 | 695 | 694 | 0 | 0.000 | 2270 | 377 | 377 | 0 | 0.000 |
| 147444 | 2 | 216972458 | 216972458 | C | G | <i>XRC5</i> | intronic | c.1+37 | e0+37 | tier2 | 13512 | 1330 | 1329 | 0 | 0.000 | 22640 | 3716 | 2783 | 915 | 0.247 | 7246 | 1084 | 652 | 424 | 0.394 | 18493 | 2929 | 2923 | 1 | 0.000 | 24099 | 3335 | 3330 | 0 | 0.000 |
| 147444 | 3 | 158306755 | 158306755 | A | T | <i>MLF1</i> | intronic | c.96-3468 | e2-3468 | tier4 | 10373 | 943 | 942 | 0 | 0.000 | 14220 | 2013 | 1495 | 498 | 0.250 | 6438 | 865 | 527 | 337 | 0.390 | 15470 | 2192 | 2188 | 0 | 0.000 | 16084 | 2054 | 2052 | 0 | 0.000 |
| 147444 | 4 | 100761543 | 100761543 | C | A | <i>DAPP1</i> | missense | c.322 | p.H108N | tier1 | 16889 | 1615 | 1614 | 0 | 0.000 | 16531 | 2137 | 1767 | 362 | 0.170 | 15332 | 1944 | 1456 | 479 | 0.248 | 23704 | 3119 | 3117 | 0 | 0.000 | 39234 | 5007 | 5006 | 0 | 0.000 |
| 147444 | 4 | 149358015 | 149358015 | C | T | <i>NR3C2</i> | splice_site | c.1-1 | e1-1 | tier1 | 8884 | 737 | 737 | 0 | 0.000 | 18409 | 2745 | 2187 | 536 | 0.197 | 7838 | 1045 | 720 | 318 | 0.306 | 17182 | 2480 | 2478 | 0 | 0.000 | 17484 | 2372 | 2370 | 0 | 0.000 |
| 147444 | 5 | 1331845 | 1331845 | C | T | <i>CLPTM1L</i> | 5_prime_untranslated_region | c.-1331845 | NULL | tier3 | 7188 | 709 | 709 | 0 | 0.000 | 18907 | 2183 | 1651 | 528 | 0.242 | 8869 | 1189 | 735 | 452 | 0.381 | 18715 | 2550 | 2548 | 0 | 0.000 | 27835 | 3489 | 3488 | 1 | 0.000 |
| 147444 | 5 | 38464253 | 38464253 | C | T | <i>EGFLAM</i> | 3_prime_untranslated_region | c.*165 | NULL | tier3 | 25273 | 2674 | 2673 | 1 | 0.000 | 37488 | 4629 | 3523 | 1081 | 0.235 | 21658 | 2897 | 1888 | 994 | 0.345 | 39935 | 5325 | 5322 | 0 | 0.000 | 65825 | 8669 | 8666 | 1 | 0.000 |
| 147444 | 5 | 39331903 | 39331903 | C | T | <i>C9</i> | missense | c.490 | p.G164R | tier1 | 3614 | 396 | 392 | 3 | 0.008 | 7230 | 386 | 578 | 266 | 0.315 | 5114 | 681 | 419 | 257 | 0.380 | 7180 | 929 | 928 | 0 | 0.000 | 10463 | 1327 | 1323 | 2 | 0.000 |
| 147444 | 6 | 39841127 | 39841127 | G | A | <i>DAAM2</i> | missense | c.1158 | p.M386I | tier1 | 22892 | 2709 | 2708 | 1 | 0.000 | 36571 | 4423 | 3222 | 1184 | 0.269 | 23555 | 3154 | 1965 | 1172 | 0.374 | 27379 | 3620 | 3616 | 1 | 0.000 | 55044 | 7235 | 7228 | 2 | 0.000 |
| 147444 | 6 | 72086766 | 72086766 | A | G | <i>LINC00472</i> | intronic | c.NULL | NULL | tier3 | 5832 | 722 | 719 | 1 | 0.001 | 8420 | 1484 | 1127 | 352 | 0.238 | 5386 | 913 | 569 | 341 | 0.375 | 8670 | 1526 | 1524 | 1 | 0.000 | 12365 | 2034 | 2032 | 1 | 0.000 |
| 147444 | 6 | 167571516 | 167571516 | G | A | <i>GPR31</i> | 5_prime_untranslated_region | c.-197 | NULL | tier3 | 8744 | 1006 | 1006 | 0 | 0.000 | 14622 | 2606 | 1969 | 630 | 0.242 | 3344 | 584 | 391 | 192 | 0.329 | 16196 | 2761 | 2759 | 0 | 0.000 | 14664 | 2330 | 2329 | 0 | 0.000 |
| 147444 | 7 | 57522854 | 57522854 | A | T | <i>ZNF716</i> | missense | c.242 | p.E81V | tier1 | 2895 | 288 | 287 | 1 | 0.003 | 3974 | 838 | 584 | 249 | 0.299 | 1176 | 177 | 70 | 107 | 0.605 | 4731 | 890 | 888 | 0 | 0.000 | 4150 | 625 | 624 | 0 | 0.000 |
| 147444 | 7 | 100190385 | 100190385 | C | T | <i>FBXO24</i> | intronic | c.673-21 | e5-21 | tier3 | 4295 | 457 | 457 | 0 | 0.000 | 7017 | 1252 | 863 | 378 | 0.305 | 1137 | 187 | 56 | 130 | 0.699 | 6729 | 0 | 0 | 0 | NA | 10618 | 1514 | 1510 | 3 | 0.000 |
| 147444 | 9 | 43628454 | 43628454 | G | A | <i>SPATA31A6</i> | intronic | c.309-76 | e4-76 | tier3 | 3697 | 578 | 578 | 0 | 0.000 | 8414 | 1541 | 1206 | 330 | 0.215 | 2425 | 526 | 356 | 170 | 0.323 | 6629 | 1354 | 1352 | 0 | 0.000 | 8862 | 1746 | 1746 | 0 | 0.000 |
| 147444 | 10 | 24762962 | 24762962 | C | T | <i>KIAA1217</i> | missense | c.1652 | p.A551V | tier1 | 24207 | 2668 | 2665 | 1 | 0.000 | 34026 | 4225 | 3132 | 1062 | 0.253 | 14479 | 1937 | 1231 | 705 | 0.364 | 37714 | 4858 | 4845 | 1 | 0.000 | 45456 | 5957 | 5948 | 2 | 0.000 |
| 147444 | 10 | 46148449 | 46148449 | A | T | <i>ZFAND4</i> | nonsense | c.243 | p.Y81* | tier1 | 15979 | 0 | 0 | 0 | 0.000 | 15247 | 2183 | 1575 | 601 | 0.276 | 20851 | 2692 | 1644 | 1028 | 0.385 | 24361 | 3340 | 3337 | 0 | 0.000 | 32088 | 4172 | 4172 | 0 | 0.000 |
| 147444 | 11 | 3687330 | 3687330 | C | T | <i>CHRNA10</i> | 3_prime_untranslated_region | c.*7 | NULL | tier3 | 11245 | 1190 | 1189 | 0 | 0.000 | 10857 | 1639 | 1316 | 319 | 0.195 | 4935 | 619 | 394 | 225 | 0.363 | 13933 | 1998 | 1996 | 0 | 0.000 | 19592 | 2631 | 2631 | 0 | 0.000 |
| 147444 | 11 | 64821688 | 64821688 | C | A | <i>NAALADL1</i> | intronic | c.992+34 | e6+34 | tier3 | 4423 | 555 | 554 | 0 | 0.000 | 12071 | 2706 | 2112 | 581 | 0.216 | 782 | 165 | 95 | 70 | 0.424 | 8350 | 1819 | 1817 | 0 | 0.000 | 4226 | 885 | 884 | 0 | 0.000 |
| 147444 | 12 | 57579580 | 57579580 | C | T | <i>LRP1</i> | missense | c.6730 | p.R2244W | tier1 | 6181 | 696 | 696 | 0 | 0.000 | 26763 | 3903 | 2873 | 1013 | 0.261 | 6627 | 914 | 554 | 360 | 0.394 | 17975 | 2775 | 2772 | 0 | 0.000 | 33845 | 4700 | 4693 | 4 | 0.000 |
| 147444 | 12 | 57627594 | 57627594 | G | T | <i>SHMT2</i> | missense | c.1186 | p.A396S | tier1 | 13274 | 1571 | 1571 | 0 | 0.000 | 22545 | 3253 | 2583 | 886 | 0.274 | 7162 | 1122 | 636 | 483 | 0.432 | 18905 | 3067 | 3061 | 2 | 0.000 | 25936 | 3859 | 3858 | 1 | 0.000 |
| 147444 | 12 | 113444369 | 113444369 | G | A | <i>OAS2</i> | silent | c.1620 | p.K540 | tier1 | 15418 | 1653 | 1651 | 1 | 0.001 | 21520 | 2598 | 2053 | 539 | 0.208 | 17323 | 2268 | 1479 | 773 | 0.343 | 31905 | 4057 | 4054 | 1 | 0.000 | 39100 | 4943 | 4940 | 2 | 0.000 |
| 147444 | 14 | 50612016 | 50612016 | C | G | <i>SOS2</i> | missense | c.2539 | p.V847L | tier1 | 10465 | 965 | 965 | 0 | 0.000 | 18080 | 2343 | 1784 | 553 | 0.237 | 18691 | 2434 | 1589 | 838 | 0.345 | 21243 | 2773 | 2770 | 2 | 0.000 | 31737 | 3941 | 3935 | 0 | 0.000 |
| 147444 | 16 | 4384836 | 4384836 | C | T | <i>GLIS2</i> | missense | c.380 | p.P127L | tier1 | 7854 | 786 | 786 | 0 | 0.000 | 15154 | 2076 | 1685 | 388 | 0.187 | 3524 | 497 | 356 | 141 | 0.284 | 13459 | 1988 | 1988 | 0 | 0.000 | 29256 | 3913 | 3913 | 0 | 0.000 |
| 147444 | 16 | 68721437 | 68721437 | G | A | <i>CDH3</i> | silent | c.1593 | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 147457 | 2 | 26177162 | 26177162 | G | A | KIF3C | silent | c.1863 | p.N62I | tier1 | 27245 | 3864 | 3823 | 35 | 0.009 | 34757 | 3636 | 2585 | 1031 | 0.285 | NA | NA | NA | NA | NA | 55934 | 352 | 344 | 7 | 0.020 | NA | NA | NA | NA | NA |
| 147457 | 2 | 26644243 | 26644243 | G | A | CCDC164 | missense | c.331 | p.E111K | tier1 | 52526 | 6231 | 6157 | 71 | 0.011 | 49967 | 4340 | 3163 | 1169 | 0.270 | NA | NA | NA | NA | NA | 419492 | 1765 | 1757 | 8 | 0.005 | NA | NA | NA | NA | NA |
| 147457 | 2 | 26800492 | 26800492 | G | A | C2orf70 | missense | c.457 | p.V153I | tier1 | 17275 | 2496 | 2483 | 12 | 0.005 | 31085 | 3339 | 2489 | 836 | 0.251 | NA | NA | NA | NA | NA | 37946 | 275 | 274 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 27261955 | 27261955 | C | T | TMEM214 | missense | c.1448 | p.T483M | tier1 | 25164 | 3393 | 3357 | 31 | 0.009 | 28439 | 2917 | 2030 | 875 | 0.301 | NA | NA | NA | NA | NA | 196050 | 815 | 813 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 27310227 | 27310227 | C | T | KHK | splice_region | c.92+8 | e1+8 | tier2 | 12150 | 1593 | 1573 | 17 | 0.011 | 19205 | 2114 | 1308 | 798 | 0.379 | NA | NA | NA | NA | NA | 98513 | 389 | 384 | 5 | 0.013 | NA | NA | NA | NA | NA |
| 147457 | 2 | 27592008 | 27592008 | G | A | EIF2B4 | missense | c.283 | p.R95W | tier1 | 48723 | 5745 | 5696 | 44 | 0.008 | 31799 | 2663 | 1991 | 660 | 0.249 | NA | NA | NA | NA | NA | 340406 | 1359 | 1357 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 27801023 | 27801023 | G | A | C2orf16 | silent | c.1584 | p.T528 | tier1 | 49704 | 5839 | 5767 | 68 | 0.012 | 62611 | 4740 | 3400 | 1317 | 0.279 | NA | NA | NA | NA | NA | 398132 | 1510 | 1504 | 4 | 0.003 | NA | NA | NA | NA | NA |
| 147457 | 2 | 56403145 | 56403145 | C | T | LOC100129434 | 5_prime_untranslated_region | c.-56403145 | NULL | tier2 | 30072 | 3662 | 3615 | 35 | 0.010 | 39079 | 3050 | 2166 | 878 | 0.288 | NA | NA | NA | NA | NA | 123501 | 631 | 627 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 58459235 | 58459235 | G | A | FANCL | missense | c.109 | p.H37Y | tier1 | 13637 | 1776 | 1763 | 13 | 0.007 | 17012 | 1659 | 1163 | 491 | 0.297 | NA | NA | NA | NA | NA | 57782 | 276 | 273 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 61331050 | 61331050 | G | A | KIAA1841 | silent | c.1428 | p.P476 | tier1 | 18861 | 2518 | 2491 | 24 | 0.010 | 26024 | 2812 | 1988 | 814 | 0.291 | NA | NA | NA | NA | NA | 58194 | 266 | 265 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 70504464 | 70504464 | C | T | PCYOX1 | silent | c.1458 | p.N486 | tier1 | 20184 | 2781 | 2738 | 41 | 0.015 | 42952 | 4283 | 2982 | 1283 | 0.301 | NA | NA | NA | NA | NA | 229202 | 948 | 945 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 73829499 | 73829499 | C | T | ALMS1 | splice_region | c.12295+4 | e20+4 | tier2 | 36808 | 4521 | 4479 | 37 | 0.008 | 33371 | 3007 | 2047 | 950 | 0.317 | NA | NA | NA | NA | NA | 184306 | 816 | 802 | 8 | 0.010 | NA | NA | NA | NA | NA |
| 147457 | 2 | 74173887 | 74173887 | G | A | DGUOK | missense | c.297 | p.M99I | tier1 | 20765 | 3307 | 3272 | 34 | 0.010 | 28136 | 3815 | 2704 | 1098 | 0.289 | NA | NA | NA | NA | NA | 91628 | 539 | 518 | 18 | 0.034 | NA | NA | NA | NA | NA |
| 147457 | 2 | 74867224 | 74867224 | C | T | C2orf65 | missense | c.179 | p.R60H | tier1 | 55784 | 6862 | 6761 | 84 | 0.012 | 52644 | 5052 | 3606 | 1419 | 0.282 | NA | NA | NA | NA | NA | 475210 | 2051 | 2017 | 28 | 0.014 | NA | NA | NA | NA | NA |
| 147457 | 2 | 75158001 | 75158001 | C | T | ENSG00000204792 | rna | NULL | NULL | tier1 | 34294 | 4184 | 4139 | 43 | 0.010 | 47513 | 4285 | 3048 | 1224 | 0.287 | NA | NA | NA | NA | NA | 190008 | 830 | 821 | 7 | 0.008 | NA | NA | NA | NA | NA |
| 147457 | 2 | 91800800 | 91800800 | G | A | ENSG00000233991 | 3_prime_untranslated_region | c.*91800800 | NULL | tier2 | 133344 | 17358 | 17292 | 41 | 0.002 | 147828 | 13612 | 12816 | 745 | 0.055 | NA | NA | NA | NA | NA | 424083 | 2440 | 2412 | 8 | 0.003 | NA | NA | NA | NA | NA |
| 147457 | 2 | 131704217 | 131704217 | C | T | ARHGEF4 | missense | c.3004 | p.R1002C | tier1 | 18730 | 2500 | 2478 | 22 | 0.009 | 24811 | 2622 | 1910 | 699 | 0.268 | NA | NA | NA | NA | NA | 74943 | 362 | 350 | 7 | 0.020 | NA | NA | NA | NA | NA |
| 147457 | 2 | 141459402 | 141459402 | G | A | LRP1B | silent | c.6315 | p.N2105 | tier1 | 59363 | 6861 | 6824 | 30 | 0.004 | 54726 | 4076 | 3676 | 1009 | 0.215 | NA | NA | NA | NA | NA | 582577 | 2229 | 2229 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 160983105 | 160983105 | G | A | ITGB6 | silent | c.1668 | p.G556 | tier1 | 8514 | 1059 | 1053 | 6 | 0.006 | 8706 | 948 | 719 | 228 | 0.241 | NA | NA | NA | NA | NA | 69591 | 277 | 277 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 170134318 | 170134318 | C | T | LRP2 | missense | c.1709 | p.R570H | tier1 | 22666 | 2925 | 2893 | 31 | 0.011 | 20311 | 2603 | 1812 | 786 | 0.303 | NA | NA | NA | NA | NA | 241150 | 975 | 970 | 5 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 179613743 | 179613743 | C | T | TTN | missense | c.13384 | p.E4462K | tier1 | 2413 | 310 | 307 | 3 | 0.010 | 6760 | 742 | 540 | 202 | 0.272 | NA | NA | NA | NA | NA | 13490 | 65 | 65 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 198274685 | 198274685 | C | T | SF3B1 | missense | c.713 | p.R238H | tier1 | 15354 | 1483 | 1470 | 11 | 0.007 | 20271 | 1773 | 1310 | 458 | 0.259 | NA | NA | NA | NA | NA | 73244 | 222 | 222 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 201619547 | 201619547 | C | T | AOX2P | 3_prime_untranslated_region | c.*201619547 | NULL | tier3 | 36167 | 4367 | 4329 | 33 | 0.008 | 36897 | 3282 | 2461 | 803 | 0.246 | NA | NA | NA | NA | NA | 392259 | 1448 | 1432 | 12 | 0.008 | NA | NA | NA | NA | NA |
| 147457 | 2 | 203948117 | 203948117 | G | A | NBEAL1 | missense | c.860 | p.R287H | tier1 | 12825 | 2428 | 2401 | 26 | 0.011 | 19494 | 2420 | 1709 | 709 | 0.293 | NA | NA | NA | NA | NA | 65429 | 799 | 799 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 213886807 | 213886807 | G | A | IKZF2 | missense | c.622 | p.R208C | tier1 | 12114 | 1633 | 1621 | 12 | 0.007 | 21353 | 2293 | 1778 | 513 | 0.224 | NA | NA | NA | NA | NA | 84581 | 416 | 414 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 215798769 | 215798769 | C | T | ABCA12 | intronic | c.7680+33 | e52+33 | tier3 | 17922 | 2244 | 2233 | 10 | 0.004 | 12457 | 1227 | 1003 | 221 | 0.181 | NA | NA | NA | NA | NA | 123380 | 531 | 528 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 218938006 | 218938006 | C | T | RUFY4 | missense | c.427 | p.R143C | tier1 | 44597 | 5378 | 5337 | 35 | 0.007 | 49790 | 4124 | 2927 | 1187 | 0.289 | NA | NA | NA | NA | NA | 315806 | 1307 | 1293 | 7 | 0.005 | NA | NA | NA | NA | NA |
| 147457 | 2 | 219538351 | 219538351 | G | A | STK36 | missense | c.88 | p.V30M | tier1 | 26783 | 3613 | 3585 | 27 | 0.007 | 29394 | 2461 | 1875 | 1009 | 0.289 | NA | NA | NA | NA | NA | 193441 | 843 | 830 | 12 | 0.014 | NA | NA | NA | NA | NA |
| 147457 | 2 | 219544699 | 219544699 | C | T | STK36 | silent | c.1032 | p.L344 | tier1 | 13816 | 2376 | 2359 | 17 | 0.007 | 21860 | 2554 | 1869 | 680 | 0.267 | NA | NA | NA | NA | NA | 43770 | 343 | 343 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 220312930 | 220312930 | C | T | SPEG | silent | c.1050 | p.T350 | tier1 | 51172 | 6462 | 6396 | 54 | 0.008 | 39196 | 3226 | 2368 | 846 | 0.263 | NA | NA | NA | NA | NA | 265917 | 1402 | 1396 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 226447163 | 226447163 | G | A | NYAP2 | missense | c.1030 | p.V344M | tier1 | 7589 | 1459 | 1362 | 16 | 0.012 | 9152 | 1699 | 1258 | 390 | 0.237 | NA | NA | NA | NA | NA | 57406 | 316 | 311 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 226447731 | 226447731 | G | A | NYAP2 | missense | c.1598 | p.R533H | tier1 | 26535 | 3480 | 3444 | 35 | 0.010 | 27168 | 2294 | 1629 | 659 | 0.288 | NA | NA | NA | NA | NA | 68068 | 384 | 383 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 227872842 | 227872842 | C | T | COL4A4 | silent | c.4701 | p.A1567 | tier1 | 43657 | 4699 | 4678 | 17 | 0.004 | 31418 | 2319 | 1979 | 338 | 0.146 | NA | NA | NA | NA | NA | 221277 | 930 | 920 | 5 | 0.005 | NA | NA | NA | NA | NA |
| 147457 | 2 | 227915822 | 227915822 | G | A | COL4A4 | silent | c.3021 | p.Y1007 | tier1 | 8324 | 1031 | 1022 | 9 | 0.009 | 12382 | 1111 | 782 | 327 | 0.295 | NA | NA | NA | NA | NA | 16473 | 103 | 103 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 238245159 | 238245159 | C | T | COL6A3 | missense | c.8584 | p.V2862I | tier1 | 4878 | 1195 | 1178 | 16 | 0.013 | 6734 | 1357 | 968 | 385 | 0.285 | NA | NA | NA | NA | NA | 14056 | 127 | 122 | 4 | 0.032 | NA | NA | NA | NA | NA |
| 147457 | 2 | 238461018 | 238461018 | C | T | MLPH | nonsense | c.1714 | p.R572* | tier1 | 17479 | 2156 | 2135 | 19 | 0.009 | 19667 | 1753 | 1365 | 384 | 0.220 | NA | NA | NA | NA | NA | 63549 | 352 | 352 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 2 | 241517127 | 241517127 | C | T | RNPEL1 | missense | c.1303 | p.R435C | tier1 | 53678 | 6279 | 6228 | 45 | 0.007 | 44715 | 3667 | 2675 | 980 | 0.268 | NA | NA | NA | NA | NA | 297421 | 1153 | 1141 | 8 | 0.007 | NA | NA | NA | NA | NA |
| 147457 | 3 | 9768381 | 9768381 | C | T | CPNE9 | missense | c.572 | p.S191L | tier1 | 10305 | 1355 | 1341 | 12 | 0.009 | 11500 | 1246 | 879 | 367 | 0.295 | NA | NA | NA | NA | NA | 69500 | 326 | 324 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 3 | 11468460 | 11468460 | C | T | ATG7 | intronic | c.2079+60 | e17+60 | tier3 | 2640 | 574 | 564 | 9 | 0.016 | 8229 | 1675 | 1158 | 512 | 0.307 | NA | NA | NA | NA | NA | 5342 | 69 | 69 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 3 | 14552987 | 14552987 | G | A | GRIP2 | 5_prime_untranslated_region | c.-14552987 | NULL | tier3 | 2635 | 638 | 632 | 5 | 0.008 | 4623 | 983 | 678 | 303 | 0.309 | NA | NA | NA | NA | NA | 3152 | 44 | 37 | 7 | 0.159 | NA | NA | NA | NA | NA |
| 147457 | 3 | 14862667 | 14862667 | C | T | FGD5 | missense | c.2089 | p.R697W | tier1 | 55830 | 6689 | 6614 | 71 | 0.011 | 48724 | 4014 | 2853 | 1146 | 0.287 | NA | NA | NA | NA</ | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 147457 | 5 | 1338034 | 1338034 | G | A | CLPTM1L | silent | c.663 | p.R221 | tier1 | 87830 | 9964 | 9853 | 77 | 0.008 | 70248 | 5076 | 3463 | 1585 | 0.314 | NA | NA | NA | NA | NA | 638897 | 2695 | 2660 | 23 | 0.009 | NA | NA | NA | NA | NA |
| 147457 | 5 | 1483662 | 1483662 | G | A | LPCAT1 | intronic | c.668-61 | e6-61 | tier3 | 14481 | 2628 | 2599 | 26 | 0.010 | 24837 | 3664 | 2555 | 1093 | 0.300 | NA | NA | NA | NA | NA | 24241 | 220 | 210 | 9 | 0.041 | NA | NA | NA | NA | NA |
| 147457 | 5 | 6602599 | 6602599 | C | T | NSUN2 | missense | c.1972 | p.V658M | tier1 | 23099 | 3676 | 3627 | 44 | 0.012 | 34628 | 4399 | 3049 | 1337 | 0.305 | NA | NA | NA | NA | NA | 44723 | 352 | 352 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 13762777 | 13762777 | C | T | DNAH5 | intronic | c.10281+54 | e60+54 | tier3 | 8630 | 2100 | 2088 | 11 | 0.005 | 13119 | 2377 | 1745 | 623 | 0.263 | NA | NA | NA | NA | NA | 24603 | 236 | 236 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 31995780 | 31995780 | C | T | PDZD2 | silent | c.1077 | p.H359 | tier1 | 28942 | 3461 | 3416 | 37 | 0.011 | 63181 | 4432 | 3112 | 1307 | 0.296 | NA | NA | NA | NA | NA | 191301 | 881 | 872 | 5 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 34190535 | 34190535 | C | T | ENSG00000215156 | 3_prime_untranslated_region | c.*34190535 | NULL | tier3 | 65126 | 8005 | 7980 | 11 | 0.001 | 88107 | 7460 | 7089 | 361 | 0.048 | NA | NA | NA | NA | NA | 92824 | 652 | 645 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 35910093 | 35910093 | C | T | CAPSL | missense | c.400 | p.E134K | tier1 | 35003 | 4451 | 4416 | 32 | 0.007 | 29265 | 3263 | 2422 | 839 | 0.257 | NA | NA | NA | NA | NA | 232307 | 923 | 917 | 5 | 0.005 | NA | NA | NA | NA | NA |
| 147457 | 5 | 41149449 | 41149449 | G | A | C6 | silent | c.2517 | p.D839 | tier1 | 4240 | 1041 | 1032 | 9 | 0.009 | 6757 | 1406 | 973 | 432 | 0.307 | NA | NA | NA | NA | NA | 7136 | 88 | 88 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 129521471 | 129521471 | G | A | CHSY3 | missense | c.2636 | p.R879Q | tier1 | 20992 | 3037 | 3009 | 28 | 0.009 | 29467 | 3080 | 2128 | 946 | 0.308 | NA | NA | NA | NA | NA | 74738 | 397 | 396 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 140308059 | 140308059 | C | T | PCDHAC1 | missense | c.1582 | p.R528W | tier1 | 6970 | 635 | 624 | 10 | 0.016 | 15984 | 2042 | 1421 | 617 | 0.303 | NA | NA | NA | NA | NA | 25924 | 136 | 136 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 140751057 | 140751057 | G | A | PCDHGB3 | missense | c.1096 | p.V366I | tier1 | 8638 | 2079 | 2062 | 15 | 0.007 | 14673 | 2882 | 2012 | 864 | 0.300 | NA | NA | NA | NA | NA | 11893 | 154 | 154 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 145610359 | 145610359 | C | T | RBM27 | silent | c.729 | p.I243 | tier1 | 64532 | 7397 | 7324 | 63 | 0.009 | 54129 | 4069 | 3128 | 921 | 0.227 | NA | NA | NA | NA | NA | 293020 | 1215 | 1207 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 146236055 | 146236055 | C | T | PPP2R2B | splice_region | c.37+7 | e1+7 | tier3 | 14142 | 1911 | 1880 | 26 | 0.014 | 12947 | 1599 | 1138 | 456 | 0.286 | NA | NA | NA | NA | NA | 84759 | 354 | 350 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 147929769 | 147929769 | G | A | HTRA | missense | c.83 | p.T28M | tier1 | 26378 | 4464 | 4435 | 27 | 0.006 | 28473 | 3328 | 2600 | 719 | 0.217 | NA | NA | NA | NA | NA | 93684 | 524 | 520 | 4 | 0.008 | NA | NA | NA | NA | NA |
| 147457 | 5 | 149584221 | 149584221 | C | T | SLC6A7 | intronic | c.1432+27 | e11+27 | tier3 | 48554 | 5469 | 5434 | 33 | 0.006 | 27050 | 2421 | 1948 | 470 | 0.194 | NA | NA | NA | NA | NA | 244160 | 1102 | 1102 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 153434852 | 153434852 | C | T | MFAP3 | 3_prime_untranslated_region | c.*1579 | NULL | tier3 | 7924 | 1862 | 1841 | 16 | 0.009 | 14135 | 2844 | 2136 | 699 | 0.247 | NA | NA | NA | NA | NA | 15556 | 242 | 242 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 159665575 | 159665575 | G | A | FABP6 | intronic | c.481-38 | e6-38 | tier3 | 3160 | 438 | 433 | 4 | 0.009 | 5099 | 602 | 433 | 169 | 0.281 | NA | NA | NA | NA | NA | 11801 | 57 | 57 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 173534383 | 173534383 | C | T | HMP19 | missense | c.391 | p.R131C | tier1 | 41742 | 5492 | 5454 | 28 | 0.005 | 45667 | 4730 | 3448 | 1268 | 0.269 | NA | NA | NA | NA | NA | 292563 | 1662 | 1656 | 5 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 5 | 176308809 | 176308809 | G | A | HK3 | silent | c.2277 | p.I759 | tier1 | 37127 | 4454 | 4404 | 44 | 0.010 | 29955 | 2621 | 1856 | 753 | 0.289 | NA | NA | NA | NA | NA | 224006 | 899 | 890 | 4 | 0.004 | NA | NA | NA | NA | NA |
| 147457 | 6 | 3127540 | 3127540 | C | T | BPHL | silent | c.276 | p.V92 | tier1 | 13044 | 1802 | 1772 | 26 | 0.014 | 19683 | 2361 | 1583 | 770 | 0.227 | NA | NA | NA | NA | NA | 25878 | 400 | 397 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 25727357 | 25727357 | G | A | HIST1H2BA | missense | c.221 | p.R74H | tier1 | 14792 | 1833 | 1813 | 17 | 0.009 | 19814 | 1607 | 1139 | 466 | 0.290 | NA | NA | NA | NA | NA | 56511 | 318 | 318 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 27217008 | 27217008 | C | T | PRSS16 | missense | c.467 | p.A156V | tier1 | 23019 | 2712 | 2696 | 15 | 0.006 | 23150 | 2008 | 1579 | 421 | 0.211 | NA | NA | NA | NA | NA | 153372 | 626 | 626 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 31913016 | 31913016 | G | A | C2 | missense | c.2141 | p.R714H | tier1 | 19525 | 2823 | 2787 | 34 | 0.012 | 29159 | 3121 | 2181 | 932 | 0.299 | NA | NA | NA | NA | NA | 100033 | 489 | 480 | 5 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 33554497 | 33554497 | C | T | GGNBP1 | missense | c.148 | p.R50C | tier1 | 29872 | 3713 | 3672 | 35 | 0.009 | 37994 | 3349 | 2304 | 1027 | 0.308 | NA | NA | NA | NA | NA | 122612 | 513 | 512 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 33990389 | 33990389 | C | T | GRM4 | 3_prime_untranslated_region | c.*209 | NULL | tier3 | 27642 | 3366 | 3335 | 26 | 0.008 | 19411 | 1722 | 1180 | 539 | 0.314 | NA | NA | NA | NA | NA | 205524 | 762 | 751 | 5 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 42985429 | 42985429 | C | T | KLHDC3 | missense | c.41 | p.T14M | tier1 | 43747 | 5968 | 5909 | 54 | 0.009 | 51510 | 5286 | 3654 | 1608 | 0.306 | NA | NA | NA | NA | NA | 163688 | 764 | 761 | 3 | 0.004 | NA | NA | NA | NA | NA |
| 147457 | 6 | 52522264 | 52522264 | C | T | TMEM14A | 5_prime_flanking_region | c.-19637 | NULL | tier3 | 15182 | 1890 | 1872 | 12 | 0.006 | 30946 | 2509 | 2109 | 392 | 0.157 | NA | NA | NA | NA | NA | 94944 | 690 | 677 | 7 | 0.010 | NA | NA | NA | NA | NA |
| 147457 | 6 | 55270421 | 55270421 | G | A | GFRAL | 3_prime_flanking_region | c.*3770 | NULL | tier3 | 27260 | 3329 | 3300 | 26 | 0.008 | 27849 | 2668 | 1698 | 565 | 0.250 | NA | NA | NA | NA | NA | 135270 | 550 | 550 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 105726111 | 105726111 | C | T | PRPF | missense | c.2041 | p.G681R | tier1 | 22255 | 2815 | 2780 | 33 | 0.012 | 34450 | 3182 | 2236 | 933 | 0.294 | NA | NA | NA | NA | NA | 50811 | 292 | 292 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 108985689 | 108985689 | G | A | FOXO3 | silent | c.1653 | p.S551 | tier1 | 18776 | 2205 | 2194 | 11 | 0.005 | 19185 | 1635 | 1344 | 289 | 0.177 | NA | NA | NA | NA | NA | 53710 | 394 | 394 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 136709445 | 136709445 | C | T | MAP7 | intronic | c.592+86 | e5+86 | tier3 | 15212 | 2356 | 2341 | 7 | 0.003 | 16160 | 2179 | 1732 | 435 | 0.201 | NA | NA | NA | NA | NA | 111582 | 514 | 509 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 136944051 | 136944051 | G | A | MAP3K5 | silent | c.2085 | p.Y695 | tier1 | 9715 | 1825 | 1813 | 10 | 0.005 | 14977 | 2452 | 1768 | 677 | 0.277 | NA | NA | NA | NA | NA | 23103 | 210 | 210 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 151672062 | 151672062 | G | A | AKAP12 | missense | c.2536 | p.V846M | tier1 | 9376 | 1208 | 1197 | 9 | 0.007 | 16711 | 1668 | 1268 | 394 | 0.237 | NA | NA | NA | NA | NA | 32831 | 197 | 197 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 157527582 | 157527582 | C | T | ARID1B | silent | c.5427 | p.D1809 | tier1 | 6651 | 981 | 966 | 13 | 0.013 | 10590 | 1189 | 855 | 333 | 0.280 | NA | NA | NA | NA | NA | 30684 | 153 | 153 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 157529705 | 157529705 | G | A | ARID18 | 3_prime_untranslated_region | c.*680 | NULL | tier3 | 44344 | 5882 | 5858 | 19 | 0.003 | 29280 | 3098 | 2477 | 614 | 0.199 | NA | NA | NA | NA | NA | 293392 | 1040 | 1040 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 6 | 160481709 | 160481709 | G | A | IGF2R | silent | c.3222 | p.A107A | tier1 | 57923 | 7512 | 7427 | 82 | 0.011 | 53917 | 5179 | 3685 | 1478 | 0.286 | NA | NA | NA | NA | NA | 333617 | 1392 | 1388 | 4 | 0.003 | NA | NA | NA | NA | NA |
| 147457 | 6 | 170594817 | 170594817 | C | T | D | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 147457 | 9 | 116050499 | 116050499 | C | T | PRPF4 | missense | c.980 | p.A327V | tier1 | 41232 | 4913 | 4862 | 42 | 0.009 | 28328 | 2358 | 1785 | 569 | 0.242 | NA | NA | NA | NA | NA | 289464 | 1202 | 1198 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 9 | 117087344 | 117087344 | C | T | ORM1 | missense | c.452 | p.T151M | tier1 | 43442 | 5148 | 5102 | 40 | 0.008 | 45158 | 3364 | 2337 | 1019 | 0.304 | NA | NA | NA | NA | NA | 321610 | 1637 | 1632 | 4 | 0.002 | NA | NA | NA | NA | NA |
| 147457 | 9 | 117143546 | 117143546 | C | T | AKNA | missense | c.68 | p.R23H | tier1 | 25536 | 3062 | 3042 | 20 | 0.007 | 21248 | 1684 | 1201 | 479 | 0.285 | NA | NA | NA | NA | NA | 100152 | 464 | 462 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 9 | 123370046 | 123370046 | C | T | MEGF9 | missense | c.1441 | p.D481N | tier1 | 48167 | 6040 | 5974 | 60 | 0.010 | 56049 | 4463 | 3161 | 1287 | 0.289 | NA | NA | NA | NA | NA | 458612 | 2039 | 2009 | 26 | 0.013 | NA | NA | NA | NA | NA |
| 147457 | 9 | 131394806 | 131394806 | G | A | SPTAN1 | intronic | c.7013+43 | e52+43 | tier3 | 21776 | 3028 | 2987 | 36 | 0.012 | 25400 | 2621 | 1827 | 783 | 0.300 | NA | NA | NA | NA | NA | 109225 | 451 | 449 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 9 | 131893899 | 131893899 | G | A | PPP2R4 | silent | c.546 | p.T182 | tier1 | 36364 | 4427 | 4364 | 57 | 0.013 | 43101 | 3550 | 2453 | 1079 | 0.305 | NA | NA | NA | NA | NA | 181531 | 921 | 914 | 6 | 0.007 | NA | NA | NA | NA | NA |
| 147457 | 9 | 139616703 | 139616703 | C | T | FAM69B | missense | c.433 | p.R145W | tier1 | 46754 | 5817 | 5759 | 54 | 0.009 | 56331 | 4669 | 3392 | 1261 | 0.271 | NA | NA | NA | NA | NA | 276498 | 1161 | 1146 | 10 | 0.009 | NA | NA | NA | NA | NA |
| 147457 | 10 | 21827795 | 21827795 | C | T | MLL10 | missense | c.194 | p.P65L | tier1 | 7289 | 1124 | 1110 | 14 | 0.012 | 9731 | 1542 | 1083 | 459 | 0.298 | NA | NA | NA | NA | NA | 42645 | 199 | 199 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 25890983 | 25890983 | G | A | GPR158 | 3_prime_untranslated_region | c.*2780 | NULL | tier2 | 13525 | 1967 | 1954 | 9 | 0.005 | 19086 | 2231 | 1525 | 704 | 0.316 | NA | NA | NA | NA | NA | 49958 | 313 | 313 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 27048066 | 27048066 | G | A | ABI1 | nonsense | c.1006 | p.R336* | tier1 | 30980 | 4260 | 4208 | 51 | 0.012 | 39115 | 3912 | 2803 | 1098 | 0.281 | NA | NA | NA | NA | NA | 144662 | 759 | 758 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 28023604 | 28023604 | G | A | MXK | missense | c.619 | p.R207W | tier1 | 25388 | 3737 | 3698 | 39 | 0.010 | 23584 | 2608 | 1889 | 709 | 0.273 | NA | NA | NA | NA | NA | 230541 | 986 | 976 | 9 | 0.009 | NA | NA | NA | NA | NA |
| 147457 | 10 | 29813480 | 29813480 | C | T | SVIL | missense | c.2507 | p.R836Q | tier1 | 28768 | 3511 | 3456 | 50 | 0.014 | 28761 | 3325 | 2225 | 1090 | 0.329 | NA | NA | NA | NA | NA | 142275 | 604 | 604 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 30654823 | 30654823 | C | T | ENSG00000241621 | 5_prime_untranslated_region | c.-30654823 | NULL | tier3 | 64504 | 7125 | 7065 | 53 | 0.007 | 51786 | 3709 | 2698 | 1002 | 0.271 | NA | NA | NA | NA | NA | 700337 | 2714 | 2703 | 6 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 34671634 | 34671634 | C | T | PARD3 | silent | c.1233 | p.P411 | tier1 | 11500 | 2162 | 2143 | 17 | 0.008 | 10021 | 1482 | 1071 | 407 | 0.275 | NA | NA | NA | NA | NA | 22089 | 180 | 177 | 3 | 0.017 | NA | NA | NA | NA | NA |
| 147457 | 10 | 46210567 | 46210567 | G | A | ENSG00000237840 | intronic | c.NULL | NULL | tier3 | 11019 | 1471 | 1449 | 19 | 0.013 | 17159 | 1585 | 1144 | 436 | 0.276 | NA | NA | NA | NA | NA | 128881 | 549 | 544 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 47747160 | 47747160 | G | A | ENSG00000244599 | 3_prime_untranslated_region | c.*47747160 | NULL | tier3 | 3978 | 970 | 963 | 6 | 0.006 | 8468 | 1471 | 1038 | 427 | 0.291 | NA | NA | NA | NA | NA | 5260 | 56 | 56 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 64967543 | 64967543 | C | T | JMJD1C | missense | c.3886 | p.G1296R | tier1 | 14831 | 2382 | 2356 | 23 | 0.010 | 22850 | 3087 | 2166 | 912 | 0.296 | NA | NA | NA | NA | NA | 42188 | 322 | 320 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 86004953 | 86004953 | C | T | RGR | intronic | c.79+28 | e1+28 | tier3 | 45596 | 5198 | 5142 | 51 | 0.010 | 44955 | 3371 | 2361 | 1004 | 0.298 | NA | NA | NA | NA | NA | 185736 | 888 | 878 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 87379729 | 87379729 | G | A | GRID1 | missense | c.2255 | p.T752M | tier1 | 19872 | 2337 | 2307 | 30 | 0.013 | 29761 | 2116 | 1519 | 590 | 0.280 | NA | NA | NA | NA | NA | 51666 | 289 | 282 | 6 | 0.021 | NA | NA | NA | NA | NA |
| 147457 | 10 | 100022513 | 100022513 | G | A | LOXL4 | silent | c.264 | p.Y88 | tier1 | 37675 | 4141 | 4091 | 43 | 0.010 | 49311 | 3360 | 2608 | 744 | 0.222 | NA | NA | NA | NA | NA | 366339 | 1433 | 1433 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 103870964 | 103870964 | G | A | LDB1 | intronic | c.173+49 | e3+49 | tier3 | 1757 | 446 | 438 | 6 | 0.014 | 3090 | 494 | 338 | 155 | 0.314 | NA | NA | NA | NA | NA | 3584 | 48 | 48 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 104245343 | 104245343 | C | T | ACTR1A | intronic | c.440+23 | e5+23 | tier3 | 51377 | 6085 | 6025 | 53 | 0.009 | 52683 | 4547 | 3342 | 1183 | 0.261 | NA | NA | NA | NA | NA | 326326 | 1340 | 1329 | 11 | 0.008 | NA | NA | NA | NA | NA |
| 147457 | 10 | 104628872 | 104628872 | C | T | C10orf32 | intronic | c.321-690 | e6-690 | tier4 | 17829 | 2892 | 2865 | 24 | 0.008 | 20664 | 2389 | 1688 | 694 | 0.291 | NA | NA | NA | NA | NA | 84668 | 455 | 454 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 106160477 | 106160477 | C | T | CCDC147 | missense | c.1855 | p.R619W | tier1 | 22373 | 3031 | 2997 | 30 | 0.010 | 33327 | 3504 | 2500 | 995 | 0.285 | NA | NA | NA | NA | NA | 98505 | 490 | 490 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 116008560 | 116008560 | C | T | VWAZ | intronic | c.52+36 | e1+36 | tier3 | 6753 | 1120 | 1109 | 10 | 0.009 | 12508 | 1762 | 1266 | 490 | 0.279 | NA | NA | NA | NA | NA | 17474 | 125 | 125 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 118969094 | 118969094 | G | A | KCNK18 | missense | c.439 | p.V147I | tier1 | 2666 | 631 | 627 | 4 | 0.006 | 5543 | 1106 | 792 | 311 | 0.282 | NA | NA | NA | NA | NA | 3206 | 54 | 54 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 123976222 | 123976222 | G | A | TACC2 | silent | c.7425 | p.A247S | tier1 | 78841 | 7991 | 7897 | 87 | 0.011 | 63935 | 3972 | 2763 | 1196 | 0.302 | NA | NA | NA | NA | NA | 624264 | 2450 | 2405 | 36 | 0.015 | NA | NA | NA | NA | NA |
| 147457 | 10 | 124402685 | 124402685 | G | A | DMBT1 | missense | c.7400 | p.R2467H | tier1 | 34281 | 4393 | 4356 | 34 | 0.008 | 44382 | 4010 | 2919 | 1079 | 0.270 | NA | NA | NA | NA | NA | 204867 | 908 | 907 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 10 | 135011989 | 135011989 | C | T | KNDC1 | silent | c.2055 | p.N685 | tier1 | 27057 | 3060 | 3036 | 24 | 0.008 | 28776 | 2078 | 1584 | 492 | 0.237 | NA | NA | NA | NA | NA | 156800 | 712 | 704 | 6 | 0.008 | NA | NA | NA | NA | NA |
| 147457 | 11 | 1316941 | 1316941 | C | T | TOLLIP | silent | c.117 | p.A39 | tier1 | 50306 | 6544 | 6491 | 49 | 0.007 | 39252 | 3768 | 2776 | 983 | 0.262 | NA | NA | NA | NA | NA | 220882 | 846 | 841 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 11 | 6048700 | 6048700 | C | T | OR56A1 | missense | c.235 | p.V79M | tier1 | 29802 | 3684 | 3637 | 43 | 0.012 | 34623 | 2788 | 1947 | 830 | 0.299 | NA | NA | NA | NA | NA | 232288 | 963 | 949 | 14 | 0.015 | NA | NA | NA | NA | NA |
| 147457 | 11 | 6232207 | 6232207 | G | A | C11orf42 | missense | c.937 | p.D313N | tier1 | 22205 | 3081 | 3046 | 31 | 0.010 | 33765 | 3860 | 2727 | 1117 | 0.291 | NA | NA | NA | NA | NA | 153786 | 771 | 770 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 11 | 10500213 | 10500213 | C | T | AMPD3 | missense | c.362 | p.T121M | tier1 | 24495 | 3140 | 3106 | 33 | 0.011 | 28673 | 2613 | 1799 | 805 | 0.309 | NA | NA | NA | NA | NA | 68968 | 442 | 438 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 11 | 16823374 | 16823374 | G | A | PLEKHA7 | splice_region | c.2158-10 | e16-10 | tier3 | 54269 | 6564 | 6457 | 75 | 0.011 | 83499 | 6625 | 4752 | 1842 | 0.279 | NA | NA | NA | NA | NA | 518776 | 2490 | 2478 | 8 | 0.003 | NA | NA | NA | NA | NA |
| 147457 | 11 | 17659970 | 17659970 | C | T | OTOA | intronic | c.7832-28 | e47-28 | tier3 | 24964 | 3796 | 3751 | 42 | 0.011 | 39494 | 4142 | 2869 | 1259 | 0.305 | NA | NA | NA | NA | NA | 85212 | 505 | 505 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 11 | 17801160 | 17801160 | C | T | KCNK1 | silent | c.1662 | p.Y554 | tier1 | 45883 | 5356 | 5290 | 61 | 0.011 | 28596 | 2556 | 1800 | 747 | 0.293 | NA | NA | NA | NA | NA | 249496 | 1005 | 1000 | 2 | 0.000 | NA | NA | NA | NA | NA</ |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 147457 | 12 | 123201344 | 123201344 | G | A | HCAR3 | 5_prime_untranslated_region | c.-60 | NULL | tier3 | 602 | 115 | 115 | 0 | 0.000 | 991 | 231 | 168 | 63 | 0.273 | NA | NA | NA | NA | NA | 1932 | 27 | 27 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 12 | 124175149 | 124175149 | G | A | TCTN2 | missense | c.961 | p.V321I | tier1 | 20603 | 2953 | 2938 | 13 | 0.004 | 21561 | 2690 | 2187 | 493 | 0.184 | NA | NA | NA | NA | NA | 116220 | 518 | 512 | 5 | 0.010 | NA | NA | NA | NA | NA |
| 147457 | 12 | 124422048 | 124422048 | C | T | CDC92 | missense | c.553 | p.V185M | tier1 | 37792 | 4600 | 4553 | 44 | 0.010 | 37393 | 3037 | 2219 | 811 | 0.268 | NA | NA | NA | NA | NA | 236682 | 1101 | 1092 | 7 | 0.006 | NA | NA | NA | NA | NA |
| 147457 | 12 | 131498748 | 131498748 | G | A | GPR133 | missense | c.1336 | p.A446T | tier1 | 28429 | 3454 | 3433 | 21 | 0.006 | 33326 | 3258 | 2562 | 688 | 0.212 | NA | NA | NA | NA | NA | 204438 | 786 | 773 | 8 | 0.010 | NA | NA | NA | NA | NA |
| 147457 | 13 | 21562125 | 21562125 | C | T | LATS2 | silent | c.1794 | p.S598 | tier1 | 42168 | 6122 | 6054 | 62 | 0.010 | 55136 | 5267 | 3828 | 1417 | 0.270 | NA | NA | NA | NA | NA | 236772 | 1173 | 1172 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 13 | 23928871 | 23928871 | G | A | SACS | missense | c.1880 | p.T627M | tier1 | 28547 | 3238 | 3203 | 35 | 0.011 | 55441 | 3717 | 2559 | 1150 | 0.310 | NA | NA | NA | NA | NA | 356541 | 1497 | 1494 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 13 | 24410420 | 24410420 | G | A | MIPEP | nonsense | c.1612 | p.R538* | tier1 | 17591 | 2092 | 2070 | 16 | 0.008 | 13194 | 1411 | 1088 | 319 | 0.227 | NA | NA | NA | NA | NA | 128482 | 562 | 553 | 7 | 0.013 | NA | NA | NA | NA | NA |
| 147457 | 13 | 39450219 | 39450219 | G | A | FREM2 | missense | c.8342 | p.R2781H | tier1 | 56370 | 6018 | 5976 | 38 | 0.006 | 53571 | 3379 | 2385 | 982 | 0.292 | NA | NA | NA | NA | NA | 341187 | 1499 | 1473 | 20 | 0.013 | NA | NA | NA | NA | NA |
| 147457 | 13 | 45857726 | 45857726 | C | T | GT2F2 | 3_prime_untranslated_region | c.*30 | NULL | tier3 | 10482 | 1435 | 1417 | 16 | 0.011 | 18585 | 2325 | 1580 | 738 | 0.318 | NA | NA | NA | NA | NA | 37285 | 287 | 287 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 13 | 95118904 | 95118904 | G | A | DC7 | missense | c.604 | p.R202C | tier1 | 24558 | 3082 | 3055 | 26 | 0.008 | 19234 | 1790 | 1347 | 438 | 0.245 | NA | NA | NA | NA | NA | 267773 | 950 | 933 | 11 | 0.012 | NA | NA | NA | NA | NA |
| 147457 | 13 | 111329450 | 111329450 | G | A | CARS2 | missense | c.656 | p.A219V | tier1 | 23587 | 3509 | 3489 | 19 | 0.005 | 39503 | 3377 | 2638 | 733 | 0.217 | NA | NA | NA | NA | NA | 271081 | 1204 | 1199 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 13 | 113838700 | 113838700 | G | A | PCID2 | silent | c.807 | p.Y269I | tier1 | 18347 | 2497 | 2466 | 28 | 0.011 | 22041 | 2449 | 1688 | 751 | 0.308 | NA | NA | NA | NA | NA | 88239 | 440 | 431 | 7 | 0.016 | NA | NA | NA | NA | NA |
| 147457 | 14 | 21552150 | 21552150 | C | T | ARHGGEF40 | missense | c.3730 | p.R1244W | tier1 | 53336 | 6562 | 6516 | 44 | 0.007 | 33253 | 2855 | 2223 | 624 | 0.219 | NA | NA | NA | NA | NA | 537389 | 2063 | 2040 | 11 | 0.005 | NA | NA | NA | NA | NA |
| 147457 | 14 | 21861926 | 21861926 | C | T | CHD8 | missense | c.6028 | p.E2010K | tier1 | 51044 | 5879 | 5834 | 42 | 0.007 | 52833 | 3891 | 3029 | 852 | 0.220 | NA | NA | NA | NA | NA | 369809 | 1535 | 1515 | 12 | 0.008 | NA | NA | NA | NA | NA |
| 147457 | 14 | 38261600 | 38261600 | G | A | TTC6 | missense | c.3689 | p.R1230Q | tier1 | 13147 | 1634 | 1613 | 20 | 0.012 | 11439 | 1085 | 757 | 324 | 0.300 | NA | NA | NA | NA | NA | 86742 | 327 | 323 | 4 | 0.012 | NA | NA | NA | NA | NA |
| 147457 | 14 | 45664649 | 45664649 | G | A | FANCM | intronic | c.5341-726 | e21-726 | tier3 | 6493 | 1025 | 1013 | 5 | 0.005 | 7469 | 1293 | 955 | 334 | 0.259 | NA | NA | NA | NA | NA | 41637 | 242 | 235 | 5 | 0.021 | NA | NA | NA | NA | NA |
| 147457 | 14 | 52471285 | 52471285 | C | T | C14orf166 | 3_prime_untranslated_region | c.*51 | NULL | tier3 | 31774 | 3449 | 3414 | 33 | 0.010 | 41248 | 2746 | 1943 | 794 | 0.290 | NA | NA | NA | NA | NA | 191435 | 811 | 808 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 14 | 57713531 | 57713531 | C | T | EXOC5 | missense | c.168 | p.M56I | tier1 | 12407 | 1685 | 1663 | 19 | 0.011 | 16095 | 1998 | 1443 | 551 | 0.276 | NA | NA | NA | NA | NA | 60344 | 299 | 299 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 14 | 68047847 | 68047847 | G | A | PLEKHH1 | intronic | c.3284+92 | e22+92 | tier3 | 55976 | 6306 | 6230 | 71 | 0.011 | 61545 | 4371 | 3135 | 1225 | 0.281 | NA | NA | NA | NA | NA | 433248 | 1975 | 1959 | 15 | 0.008 | NA | NA | NA | NA | NA |
| 147457 | 14 | 76259462 | 76259462 | G | A | TTL5 | intronic | c.3171+19 | e26+19 | tier3 | 42262 | 4989 | 4948 | 37 | 0.007 | 41206 | 3264 | 2439 | 814 | 0.250 | NA | NA | NA | NA | NA | 321279 | 1389 | 1380 | 6 | 0.004 | NA | NA | NA | NA | NA |
| 147457 | 14 | 91211188 | 91211188 | C | T | TTC7B | missense | c.524 | p.C175Y | tier1 | 4775 | 584 | 574 | 8 | 0.014 | 7167 | 818 | 581 | 234 | 0.287 | NA | NA | NA | NA | NA | 12924 | 76 | 72 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 14 | 91700696 | 91700696 | G | A | GPR68 | silent | c.729 | p.T243 | tier1 | 16166 | 2906 | 2875 | 27 | 0.009 | 27269 | 3480 | 2500 | 969 | 0.279 | NA | NA | NA | NA | NA | 31381 | 206 | 206 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 14 | 93706769 | 93706769 | G | A | BTBD7 | 3_prime_untranslated_region | c.*1850 | NULL | tier2 | 20889 | 3634 | 3629 | 4 | 0.001 | 30925 | 4724 | 4371 | 344 | 0.073 | NA | NA | NA | NA | NA | 77610 | 543 | 543 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 14 | 102801015 | 102801015 | G | A | ZNF839 | intronic | c.1509+32 | e4+32 | tier3 | 3908 | 447 | 441 | 6 | 0.013 | 5131 | 700 | 484 | 211 | 0.304 | NA | NA | NA | NA | NA | 31345 | 165 | 164 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 14 | 105419206 | 105419206 | G | A | AHNAK2 | missense | c.2582 | p.P861L | tier1 | 95151 | 10551 | 10435 | 84 | 0.008 | 67942 | 4996 | 3922 | 1058 | 0.212 | NA | NA | NA | NA | NA | 649421 | 2498 | 2456 | 30 | 0.012 | NA | NA | NA | NA | NA |
| 147457 | 15 | 23313159 | 23313159 | G | A | HERC2P2 | 5_prime_untranslated_region | c.-23313159 | NULL | tier2 | 10419 | 1202 | 1188 | 12 | 0.010 | 9851 | 1031 | 739 | 291 | 0.283 | NA | NA | NA | NA | NA | 59430 | 252 | 251 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 15 | 25349836 | 25349836 | C | T | SNORD116-28 | rna | NULL | NULL | tier1 | 14452 | 2046 | 2022 | 23 | 0.011 | 26056 | 2703 | 1904 | 791 | 0.294 | NA | NA | NA | NA | NA | 90007 | 468 | 457 | 9 | 0.019 | NA | NA | NA | NA | NA |
| 147457 | 15 | 28517461 | 28517461 | C | T | HERC2 | missense | c.983 | p.R328H | tier1 | 34159 | 3813 | 3791 | 18 | 0.005 | 49992 | 3031 | 2665 | 360 | 0.119 | NA | NA | NA | NA | NA | 117607 | 580 | 570 | 6 | 0.010 | NA | NA | NA | NA | NA |
| 147457 | 15 | 33936693 | 33936693 | C | T | RYR3 | silent | c.3738 | p.N1246 | tier1 | 16350 | 2252 | 2241 | 11 | 0.005 | 25093 | 2819 | 1995 | 810 | 0.289 | NA | NA | NA | NA | NA | 52243 | 342 | 342 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 15 | 40099429 | 40099429 | C | T | GPR176 | missense | c.203 | p.R68H | tier1 | 5650 | 698 | 697 | 1 | 0.001 | 2937 | 311 | 260 | 51 | 0.164 | NA | NA | NA | NA | NA | 36078 | 141 | 140 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 15 | 40532849 | 40532849 | C | T | ENSG00000259288 | missense | c.131 | p.T44M | tier1 | 12911 | 1449 | 1446 | 3 | 0.002 | 15680 | 1124 | 1064 | 60 | 0.053 | NA | NA | NA | NA | NA | 142267 | 730 | 729 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 15 | 40583762 | 40583762 | C | T | PLCB2 | 3_prime_untranslated_region | c.*205 | NULL | tier2 | 31095 | 4688 | 4604 | 81 | 0.017 | 41254 | 4079 | 2789 | 1268 | 0.313 | NA | NA | NA | NA | NA | 238420 | 1788 | 1780 | 6 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 15 | 44673070 | 44673070 | G | A | CASC4 | missense | c.968 | p.R323H | tier1 | 38594 | 4678 | 4650 | 28 | 0.006 | 25298 | 2118 | 1740 | 374 | 0.177 | NA | NA | NA | NA | NA | 371329 | 1453 | 1448 | 4 | 0.003 | NA | NA | NA | NA | NA |
| 147457 | 15 | 52622645 | 52622645 | C | T | MYO5A | missense | c.4460 | p.R1487Q | tier1 | 46289 | 5046 | 5014 | 32 | 0.006 | 33849 | 2611 | 1936 | 671 | 0.257 | NA | NA | NA | NA | NA | 352307 | 1397 | 1384 | 13 | 0.009 | NA | NA | NA | NA | NA |
| 147457 | 15 | 63964722 | 63964722 | G | A | HERC1 | missense | c.8018 | p.P2673L | tier1 | 25385 | 3203 | 3190 | 12 | 0.004 | 44121 | 3995 | 2987 | 996 | 0.250 | NA | NA | NA | NA | NA | 224348 | 1001 | 1001 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 15 | 66181422 | 66181422 | C | T | RAB11A | 3_prime_untranslated_region | c.*1244 | NULL | tier3 | 20600 | 3162 | 3142 | 16 | 0.005 | 17204 | 2414 | 1938 | 474 | 0.197 | NA | NA | NA | NA | NA | 239065 | 910 | 910 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 15 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|----------|----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 147457 | 17 | 38146349 | 38146349 | C | T | PSMD3 | nonsense | c.880 | p.R294* | tier1 | 15821 | 1944 | 1926 | 16 | 0.008 | 19025 | 1675 | 1172 | 494 | 0.297 | NA | NA | NA | NA | NA | 60870 | 367 | 365 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 39740595 | 39740595 | C | T | KRT14 | missense | c.679 | p.E227K | tier1 | 12535 | 1278 | 1268 | 8 | 0.006 | 31582 | 1908 | 1363 | 538 | 0.283 | NA | NA | NA | NA | NA | 436058 | 1697 | 1666 | 12 | 0.007 | NA | NA | NA | NA | NA |
| 147457 | 17 | 40063711 | 40063711 | C | T | ACLY | missense | c.893 | p.R298Q | tier1 | 23162 | 2794 | 2766 | 26 | 0.009 | 26911 | 2118 | 1442 | 672 | 0.318 | NA | NA | NA | NA | NA | 124013 | 539 | 539 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 41026210 | 41026210 | C | T | ENSG00000260105 | 3_prime_untranslated_region | c.*41026210 | NULL | tier2 | 1422 | 359 | 356 | 3 | 0.008 | 4299 | 654 | 457 | 196 | 0.300 | NA | NA | NA | NA | NA | 5356 | 61 | 61 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 44144025 | 44144025 | G | A | KANSL1 | nonsense | c.1726 | p.R576* | tier1 | 35110 | 3766 | 3729 | 37 | 0.010 | 42552 | 2795 | 1981 | 808 | 0.290 | NA | NA | NA | NA | NA | 375355 | 1418 | 1413 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 48434597 | 48434597 | G | A | XYLT2 | missense | c.1925 | p.R642Q | tier1 | 19353 | 2525 | 2506 | 17 | 0.007 | 46569 | 3984 | 3073 | 905 | 0.228 | NA | NA | NA | NA | NA | 178105 | 817 | 809 | 7 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 52993020 | 52993020 | C | T | TOM1L1 | intronic | c.604-87 | e7-87 | tier2 | 21550 | 2773 | 2745 | 26 | 0.009 | 20172 | 1948 | 1404 | 536 | 0.276 | NA | NA | NA | NA | NA | 137545 | 687 | 682 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 56606542 | 56606542 | C | T | 43347 | 5_prime_untranslated_region | c.-7 | NULL | tier2 | 17969 | 2205 | 2189 | 14 | 0.006 | 18362 | 1795 | 1240 | 549 | 0.307 | NA | NA | NA | NA | NA | 171324 | 724 | 724 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 59668192 | 59668192 | G | A | NACA2 | missense | c.350 | p.S117L | tier1 | 14123 | 1670 | 1661 | 8 | 0.005 | 14304 | 1283 | 1006 | 274 | 0.214 | NA | NA | NA | NA | NA | 96326 | 548 | 544 | 3 | 0.005 | NA | NA | NA | NA | NA |
| 147457 | 17 | 60754754 | 60754754 | G | A | MRC2 | silent | c.1959 | p.T653 | tier1 | 30319 | 3746 | 3700 | 42 | 0.011 | 27678 | 2592 | 1859 | 723 | 0.280 | NA | NA | NA | NA | NA | 381542 | 1304 | 1285 | 10 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 61611215 | 61611215 | C | T | KCNH6 | intronic | c.676-32 | e5-32 | tier2 | 18072 | 3168 | 3132 | 36 | 0.011 | 30425 | 3932 | 2680 | 1240 | 0.316 | NA | NA | NA | NA | NA | 17372 | 175 | 175 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 64873540 | 64873540 | C | T | CACNG5 | silent | c.90 | p.T30 | tier1 | 37343 | 4442 | 4400 | 39 | 0.009 | 41951 | 3026 | 2150 | 867 | 0.287 | NA | NA | NA | NA | NA | 138308 | 761 | 745 | 16 | 0.021 | NA | NA | NA | NA | NA |
| 147457 | 17 | 74900341 | 74900341 | G | A | MGAT5B | missense | c.560 | p.R187H | tier1 | 40682 | 5669 | 5644 | 21 | 0.004 | 33268 | 3488 | 2977 | 709 | 0.192 | NA | NA | NA | NA | NA | 320545 | 1296 | 1296 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 76167847 | 76167847 | C | T | SYNGR2 | missense | c.505 | p.R169C | tier1 | 32825 | 3983 | 3945 | 35 | 0.009 | 34027 | 2940 | 2163 | 769 | 0.262 | NA | NA | NA | NA | NA | 216621 | 937 | 919 | 16 | 0.017 | NA | NA | NA | NA | NA |
| 147457 | 17 | 76803689 | 76803689 | C | T | USP36 | silent | c.1437 | p.P479 | tier1 | 10289 | 1675 | 1653 | 20 | 0.012 | 14710 | 1628 | 1139 | 489 | 0.300 | NA | NA | NA | NA | NA | 34702 | 197 | 197 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 77081692 | 77081692 | C | T | ENGASE | splice_region | c.1701-10 | e13-10 | tier3 | 35488 | 4166 | 4123 | 38 | 0.009 | 30387 | 2481 | 1844 | 631 | 0.255 | NA | NA | NA | NA | NA | 195147 | 883 | 881 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 17 | 77083117 | 77083117 | G | A | ENGASE | 3_prime_untranslated_region | c.*686 | NULL | tier3 | 37228 | 4808 | 4758 | 47 | 0.010 | 38239 | 3488 | 2378 | 1096 | 0.315 | NA | NA | NA | NA | NA | 185511 | 842 | 824 | 17 | 0.020 | NA | NA | NA | NA | NA |
| 147457 | 17 | 78247071 | 78247071 | G | A | RNF213 | silent | c.129 | p.S43 | tier1 | 19809 | 2110 | 2092 | 17 | 0.008 | 15825 | 1260 | 971 | 284 | 0.226 | NA | NA | NA | NA | NA | 95659 | 421 | 417 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 18 | 5397381 | 5397381 | G | A | EPB41L3 | silent | c.2517 | p.P839 | tier1 | 30093 | 3685 | 3664 | 21 | 0.006 | 27923 | 2749 | 2147 | 597 | 0.218 | NA | NA | NA | NA | NA | 321328 | 1314 | 1313 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 18 | 6837314 | 6837314 | C | T | ARHGAP28 | silent | c.444 | p.A148 | tier1 | 29197 | 3249 | 3249 | 17 | 0.005 | 20827 | 1396 | 1152 | 241 | 0.173 | NA | NA | NA | NA | NA | 169370 | 663 | 658 | 5 | 0.008 | NA | NA | NA | NA | NA |
| 147457 | 18 | 13072806 | 13072806 | C | T | CEP192 | nonsense | c.5401 | p.R1801* | tier1 | 44687 | 5534 | 5494 | 37 | 0.007 | 36812 | 3414 | 2561 | 841 | 0.247 | NA | NA | NA | NA | NA | 264181 | 1119 | 1119 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 18 | 19024347 | 19024347 | C | T | GREB1L | missense | c.1370 | p.T457M | tier1 | 12675 | 1867 | 1849 | 18 | 0.010 | 20959 | 2736 | 2049 | 680 | 0.249 | NA | NA | NA | NA | NA | 104212 | 442 | 437 | 5 | 0.011 | NA | NA | NA | NA | NA |
| 147457 | 18 | 19780629 | 19780629 | C | T | GATA6 | missense | c.1370 | p.P544L | tier1 | 18601 | 2243 | 2220 | 23 | 0.010 | 17809 | 1436 | 1015 | 417 | 0.291 | NA | NA | NA | NA | NA | 92247 | 478 | 478 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 18 | 24126926 | 24126926 | G | A | KCTD1 | silent | c.1575 | p.D525 | tier1 | 23659 | 3227 | 3201 | 24 | 0.007 | 16203 | 1472 | 1164 | 302 | 0.206 | NA | NA | NA | NA | NA | 200356 | 716 | 709 | 5 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 18 | 34340699 | 34340699 | C | T | FHOD3 | silent | c.A029 | p.D1343 | tier1 | 26245 | 2927 | 2899 | 27 | 0.009 | 23979 | 2017 | 1324 | 687 | 0.342 | NA | NA | NA | NA | NA | 202310 | 811 | 800 | 10 | 0.012 | NA | NA | NA | NA | NA |
| 147457 | 18 | 47527556 | 47527556 | C | T | MYO5B | intronic | c.612+69 | e5+69 | tier2 | 40300 | 4764 | 4690 | 65 | 0.014 | 50113 | 4494 | 3058 | 1412 | 0.316 | NA | NA | NA | NA | NA | 291438 | 1179 | 1153 | 14 | 0.012 | NA | NA | NA | NA | NA |
| 147457 | 19 | 648900 | 648900 | C | T | RNF126 | missense | c.652 | p.V218I | tier1 | 31606 | 3928 | 3893 | 31 | 0.008 | 28990 | 2506 | 1763 | 738 | 0.295 | NA | NA | NA | NA | NA | 158706 | 694 | 680 | 10 | 0.014 | NA | NA | NA | NA | NA |
| 147457 | 19 | 806592 | 806592 | C | T | PTBP1 | intronic | c.1119+36 | e10+36 | tier2 | 21508 | 2823 | 2798 | 21 | 0.007 | 21674 | 2125 | 1472 | 651 | 0.307 | NA | NA | NA | NA | NA | 62240 | 341 | 341 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 19 | 10462666 | 10462666 | G | A | ABCA7 | missense | c.1483 | p.D495N | tier1 | 33339 | 3688 | 3643 | 38 | 0.010 | 25302 | 1769 | 1199 | 568 | 0.321 | NA | NA | NA | NA | NA | 167520 | 673 | 666 | 7 | 0.010 | NA | NA | NA | NA | NA |
| 147457 | 19 | 3976654 | 3976654 | G | A | EEF2 | silent | c.2475 | p.F825 | tier1 | 33995 | 4940 | 4882 | 52 | 0.011 | 41540 | 4404 | 3006 | 1377 | 0.314 | NA | NA | NA | NA | NA | 110150 | 630 | 626 | 4 | 0.006 | NA | NA | NA | NA | NA |
| 147457 | 19 | 4511513 | 4511513 | G | A | PLIN4 | missense | c.2417 | p.T806M | tier1 | 10301 | 1067 | 1066 | 1 | 0.001 | 25303 | 1776 | 1715 | 60 | 0.034 | NA | NA | NA | NA | NA | 530049 | 1684 | 1678 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 19 | 4930798 | 4930798 | C | T | UHRF1 | missense | c.518 | p.T173M | tier1 | 31456 | 3133 | 3095 | 35 | 0.011 | 50025 | 3203 | 2266 | 931 | 0.291 | NA | NA | NA | NA | NA | 390756 | 1523 | 1517 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 19 | 7267653 | 7267653 | C | T | INSR | missense | c.355 | p.A119T | tier1 | 49792 | 5721 | 5642 | 76 | 0.013 | 40105 | 3725 | 2635 | 1083 | 0.291 | NA | NA | NA | NA | NA | 276016 | 1029 | 1020 | 9 | 0.009 | NA | NA | NA | NA | NA |
| 147457 | 19 | 7620282 | 7620282 | G | A | PNPLA6 | intronic | c.2966+29 | e24+29 | tier2 | 3735 | 398 | 386 | 12 | 0.030 | 7418 | 967 | 510 | 457 | 0.473 | NA | NA | NA | NA | NA | 15629 | 104 | 102 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 19 | 8027941 | 8027941 | C | T | ELAVL1 | 3_prime_untranslated_region | c.*426 | NULL | tier3 | 20071 | 2492 | 2462 | 27 | 0.011 | 33174 | 2997 | 2189 | 799 | 0.267 | NA | NA | NA | NA | NA | 103429 | 541 | 541 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 19 | 8617070 | 8617070 | C | T | MYO1F | intronic | c.505-22 | e7-22 | tier3 | 31249 | 3658 | 3627 | 27 | 0.007 | 30018 | 2426 | 1757 | 664 | 0.274 | NA | NA | NA | NA | NA | 154879 | 643 | 643 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 147457 | 19 | 10099957 | 10099957 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF | |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|----|
| 147457 | 21 | 43984766 | 43984766 | G | A | SLC37A1 | intronic | c.1136-52 | e13-52 | tier3 | 78694 | 8566 | 8479 | 72 | 0.008 | 64098 | 4715 | 3720 | 981 | 0.209 | NA | NA | NA | NA | NA | 707573 | 2923 | 2911 | 4 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | 21 | 46363698 | 46363698 | G | A | FAM207A | missense | c.229 | p.V771 | tier1 | 16968 | 2090 | 2071 | 16 | 0.008 | 20962 | 1807 | 1262 | 540 | 0.300 | NA | NA | NA | NA | NA | 41648 | 241 | 236 | 4 | 0.017 | NA | NA | NA | NA | NA | |
| 147457 | 22 | 21349265 | 21349265 | G | A | LZTR1 | missense | c.1892 | p.R631Q | tier1 | 13742 | 1710 | 1691 | 16 | 0.009 | 18524 | 1659 | 1164 | 493 | 0.298 | NA | NA | NA | NA | NA | 42995 | 251 | 251 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | 22 | 23077532 | 23077532 | C | T | IGLV2-18 | silent | c.309 | p.D103 | tier1 | 37981 | 5120 | 5091 | 28 | 0.005 | 38538 | 3976 | 2949 | 1016 | 0.256 | NA | NA | NA | NA | NA | 246785 | 1093 | 1087 | 6 | 0.005 | NA | NA | NA | NA | NA | |
| 147457 | 22 | 25598652 | 25598652 | C | T | CRYBB3 | silent | c.87 | p.Y29 | tier1 | 26233 | 3344 | 3300 | 39 | 0.012 | 35699 | 3034 | 2170 | 851 | 0.282 | NA | NA | NA | NA | NA | 253231 | 1082 | 1067 | 7 | 0.007 | NA | NA | NA | NA | NA | |
| 147457 | 22 | 37319436 | 37319436 | C | T | CSF2RB | intronic | c.200+27 | e2+27 | tier3 | 29488 | 3041 | 3023 | 14 | 0.005 | 25804 | 1542 | 1248 | 290 | 0.189 | NA | NA | NA | NA | NA | 130760 | 569 | 569 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | 22 | 37892522 | 37892522 | C | T | CARD10 | missense | c.1993 | p.E665K | tier1 | 7072 | 1147 | 1135 | 10 | 0.009 | 12107 | 1715 | 1279 | 433 | 0.253 | NA | NA | NA | NA | NA | 18615 | 146 | 146 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | 22 | 38062929 | 38062929 | G | A | PDXP | 3_prime_untranslated_region | c.*1051 | NULL | tier2 | 32675 | 3739 | 3698 | 34 | 0.009 | 27225 | 1997 | 1519 | 474 | 0.238 | NA | NA | NA | NA | NA | 275521 | 1042 | 1040 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 2406917 | 2406917 | C | T | ZBED1 | missense | c.1844 | p.R615H | tier1 | 43312 | 6503 | 6426 | 69 | 0.011 | 44160 | 4762 | 3374 | 1379 | 0.290 | NA | NA | NA | NA | NA | 120964 | 708 | 707 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 20070889 | 20070889 | C | T | MAP7D2 | intronic | c.595+107 | e5+107 | tier3 | 53914 | 6343 | 6289 | 47 | 0.007 | 45177 | 3601 | 2758 | 838 | 0.233 | NA | NA | NA | NA | NA | 188162 | 742 | 730 | 9 | 0.012 | NA | NA | NA | NA | NA | |
| 147457 | X | 29973688 | 29973688 | G | A | ILIRAPL1 | silent | c.1842 | p.T614 | tier1 | 45477 | 5734 | 5671 | 58 | 0.010 | 49124 | 4546 | 3206 | 1318 | 0.291 | NA | NA | NA | NA | NA | 120988 | 512 | 498 | 12 | 0.024 | NA | NA | NA | NA | NA | |
| 147457 | X | 29974566 | 29974566 | G | A | ILIRAPL1 | 3_prime_untranslated_region | c.*629 | NULL | tier3 | 11625 | 1711 | 1697 | 12 | 0.007 | 15316 | 2088 | 1494 | 586 | 0.282 | NA | NA | NA | NA | NA | 34432 | 365 | 363 | 1 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 41530801 | 41530801 | C | T | CASK | intronic | c.430-18 | e6-18 | tier4 | 6687 | 1019 | 1003 | 16 | 0.016 | 9343 | 1184 | 822 | 360 | 0.305 | NA | NA | NA | NA | NA | 21317 | 139 | 139 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 49032048 | 49032048 | G | A | PRICKLE3 | nonsense | c.1822 | p.R608* | tier1 | 30094 | 3327 | 3286 | 39 | 0.012 | 24630 | 1705 | 1302 | 401 | 0.235 | NA | NA | NA | NA | NA | 144633 | 581 | 570 | 6 | 0.010 | NA | NA | NA | NA | NA | |
| 147457 | X | 69595890 | 69595890 | C | T | KIF4A | intronic | c.1924-60 | e17-60 | tier3 | 20196 | 2557 | 2542 | 13 | 0.005 | 16532 | 1619 | 1383 | 234 | 0.145 | NA | NA | NA | NA | NA | 94147 | 348 | 345 | 3 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 70836264 | 70836264 | C | T | CXCR3 | missense | c.1199 | p.R400Q | tier1 | 41171 | 5462 | 5428 | 31 | 0.006 | 42969 | 4051 | 2958 | 1079 | 0.267 | NA | NA | NA | NA | NA | 78679 | 373 | 351 | 22 | 0.059 | NA | NA | NA | NA | NA | |
| 147457 | X | 71427464 | 71427464 | C | A | ERCC6L | missense | c.1153 | p.V385L | tier1 | 12556 | 1953 | 1940 | 13 | 0.007 | 15638 | 2553 | 1965 | 584 | 0.229 | NA | NA | NA | NA | NA | 52191 | 273 | 270 | 3 | 0.011 | NA | NA | NA | NA | NA | |
| 147457 | X | 79281277 | 79281277 | G | A | TBX22 | splice_site | c.633+1 | e4+1 | tier1 | 29891 | 3894 | 3872 | 21 | 0.005 | 48498 | 4549 | 3408 | 1128 | 0.249 | NA | NA | NA | NA | NA | 203552 | 841 | 841 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 84525966 | 84525966 | C | T | ZNF711 | missense | c.1442 | p.P481L | tier1 | 52528 | 6434 | 6365 | 62 | 0.010 | 60448 | 5486 | 4068 | 1386 | 0.254 | NA | NA | NA | NA | NA | 210960 | 859 | 842 | 17 | 0.020 | NA | NA | NA | NA | NA | |
| 147457 | X | 103316458 | 103316458 | C | T | H2BFW2 | 5_prime_flanking_region | c.-48226 | NULL | tier3 | 61088 | 7275 | 7196 | 68 | 0.009 | 52134 | 3525 | 3233 | 1101 | 0.254 | NA | NA | NA | NA | NA | 375428 | 1507 | 1481 | 18 | 0.012 | NA | NA | NA | NA | NA | |
| 147457 | X | 105144665 | 105144665 | C | T | NRK | missense | c.766 | p.P256S | tier1 | 7692 | 1223 | 1215 | 6 | 0.005 | 16562 | 2319 | 1838 | 466 | 0.202 | NA | NA | NA | NA | NA | 27407 | 128 | 128 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 120181966 | 120181966 | G | A | GLUD2 | missense | c.428 | p.R143H | tier1 | 19731 | 3613 | 3563 | 42 | 0.012 | 29697 | 3525 | 2480 | 1024 | 0.292 | NA | NA | NA | NA | NA | 42373 | 740 | 738 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 123215311 | 123215311 | C | T | STAG2 | nonsense | c.2857 | p.R953* | tier1 | 38027 | 4879 | 4852 | 21 | 0.004 | 33850 | 3454 | 2661 | 781 | 0.227 | NA | NA | NA | NA | NA | 171103 | 691 | 687 | 3 | 0.004 | NA | NA | NA | NA | NA | |
| 147457 | X | 128929233 | 128929233 | G | A | ENSG00000240143 | intronic | c.NULL | NULL | tier3 | 20269 | 3119 | 3079 | 37 | 0.012 | 26671 | 3093 | 2179 | 902 | 0.293 | NA | NA | NA | NA | NA | 87583 | 382 | 378 | 4 | 0.010 | NA | NA | NA | NA | NA | |
| 147457 | X | 129190113 | 129190113 | C | T | BCORL1 | 3_prime_untranslated_region | c.*2 | NULL | tier3 | 40006 | 4594 | 4545 | 37 | 0.008 | 31277 | 2243 | 1814 | 421 | 0.188 | NA | NA | NA | NA | NA | 132083 | 501 | 499 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 129318354 | 129318354 | C | T | RAB33A | silent | c.354 | p.D118 | tier1 | 33357 | 4217 | 4176 | 40 | 0.009 | 47449 | 3781 | 2746 | 1021 | 0.271 | NA | NA | NA | NA | NA | 55018 | 250 | 250 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 129658059 | 129658059 | G | A | ENSG00000229702 | 5_prime_untranslated_region | c.-129658059 | NULL | tier2 | 8763 | 1510 | 1498 | 7 | 0.005 | 13164 | 1850 | 1397 | 445 | 0.242 | NA | NA | NA | NA | NA | 35695 | 149 | 147 | 2 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 138678815 | 138678815 | G | A | MCF2 | nonsense | c.2605 | p.R869* | tier1 | 8524 | 1103 | 1091 | 11 | 0.010 | 13628 | 1460 | 1022 | 455 | 0.312 | NA | NA | NA | NA | NA | 21660 | 117 | 117 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 153660704 | 153660704 | C | T | ATPGAP1 | silent | c.456 | p.L152 | tier1 | 38293 | 4612 | 4576 | 35 | 0.008 | 33550 | 2766 | 2138 | 625 | 0.226 | NA | NA | NA | NA | NA | 146124 | 512 | 509 | 2 | 0.000 | NA | NA | NA | NA | NA | |
| 147457 | X | 153696165 | 153696165 | G | A | PLXNA3 | missense | c.3641 | p.R1214Q | tier1 | 67610 | 7812 | 7711 | 89 | 0.011 | 72459 | 5126 | 3485 | 1625 | 0.318 | NA | NA | NA | NA | NA | 268586 | 1007 | 976 | 15 | 0.015 | NA | NA | NA | NA | NA | |
| 169510 | 1 | 115258745 | 115258745 | C | G | NRAS | missense | c.37 | p.G13R | tier1 | 22977 | 2771 | 2677 | 86 | 0.031 | 18987 | 2368 | 1379 | 967 | 0.412 | 4019 | 24 | 24 | 0 | 0.000 | 15410 | 508 | 507 | 1 | 0.000 | NA | NA | NA | NA | NA | |
| 169510 | 2 | 39053173 | 39053173 | C | T | DHX57 | missense | c.2830 | p.G944R | tier1 | 8622 | 1010 | 939 | 68 | 0.068 | 7536 | 923 | 502 | 414 | 0.452 | 1116 | 7 | 3 | 4 | NA | 7696 | 322 | 322 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 169510 | 2 | 73316541 | 73316541 | G | A | RAB11FIP5 | rna | NULL | NULL | tier1 | 21839 | 2804 | 2637 | 155 | 0.056 | 14239 | 1684 | 888 | 775 | 0.466 | 1793 | 12 | 8 | 4 | 0.333 | 13260 | 417 | 417 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 169510 | 3 | 184922434 | 184922434 | C | G | EHHADH | missense | c.680 | p.C227S | tier1 | 40109 | 5263 | 5092 | 156 | 0.030 | 25056 | 3318 | 2023 | 1238 | 0.380 | 13593 | 41 | 41 | 0 | 0.000 | 26178 | 718 | 717 | 1 | 0.000 | NA | NA | NA | NA | NA | |
| 169510 | 4 | 96044976 | 96044976 | C | G | BMPRI1B | missense | c.455 | p.P152R | tier1 | 3573 | 342 | 334 | 6 | 0.018 | 3154 | 340 | 232 | 104 | 0.310 | 1108 | 0 | 0 | 0 | 0 | NA | 5083 | 298 | 298 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 169510 | 5 | 141236840 | 141236840 | C | T | PCDH1 | missense | c.3296 | p.G1099E | tier1 | 20878 | 2560 | 2394 | 153 | 0.060 | 22627 | 2768 | 1582 | 1152 | 0.421 | 7304 | 32 | 31 | 1 | 0.000 | 15208 | 502 | 502 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 169 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 185295 | 12 | 65272304 | 65272304 | C | T | TBC1D30 | 3_prime_untranslated_region | c.*2736 | NULL | tier3 | 30023 | 4019 | 4010 | 4 | 0.001 | 99196 | 12907 | 9722 | 3053 | 0.239 | 37985 | 2579 | 1622 | 951 | 0.370 | 80434 | 10102 | 10088 | 4 | 0.000 | 71793 | 6371 | 6363 | 4 | 0.000 |
| 185295 | 12 | 124427322 | 124427322 | A | C | CCDC92 | nonsense | c.192 | p.Y64* | tier1 | 8592 | 1257 | 1245 | 11 | 0.009 | 25738 | 3997 | 2472 | 1482 | 0.375 | 4710 | 568 | 317 | 251 | 0.442 | 7199 | 1065 | 1065 | 0 | 0.000 | 12984 | 1924 | 1922 | 0 | 0.000 |
| 185295 | 13 | 48547335 | 48547335 | G | A | SUCLA2 | intronc | c.663+64 | e5+64 | tier3 | 1632 | 210 | 210 | 0 | 0.000 | 3506 | 497 | 366 | 127 | 0.258 | 2675 | 283 | 166 | 117 | 0.413 | 3706 | 530 | 529 | 1 | 0.000 | 2230 | 0 | 0 | 0 | NA |
| 185295 | 13 | 52684711 | 52684711 | G | T | NEK5 | silent | c.315 | p.I105 | tier1 | 1982 | 320 | 318 | 2 | 0.006 | 19764 | 2677 | 1922 | 745 | 0.279 | 6913 | 564 | 327 | 235 | 0.418 | 18570 | 2471 | 2466 | 4 | 0.000 | 13850 | 1560 | 1560 | 0 | 0.000 |
| 185295 | 14 | 96552991 | 96552991 | C | T | C14orf132 | rna | NULL | NULL | tier1 | 10922 | 1708 | 1703 | 3 | 0.002 | 37248 | 5111 | 3487 | 1592 | 0.313 | 7280 | 638 | 335 | 302 | 0.474 | 15816 | 2168 | 2165 | 2 | 0.000 | 17449 | 2002 | 1997 | 1 | 0.000 |
| 185295 | 15 | 85343270 | 85343270 | C | G | ZNF592 | intronc | c.3273+62 | e7+62 | tier3 | 11581 | 1946 | 1935 | 6 | 0.003 | 52415 | 8134 | 5754 | 2324 | 0.288 | 18696 | 1449 | 947 | 498 | 0.345 | 32984 | 4648 | 4644 | 2 | 0.000 | 39476 | 4479 | 4474 | 4 | 0.000 |
| 185295 | 17 | 10545798 | 10545798 | C | T | MYH3 | silent | c.1824 | p.G608 | tier1 | 24528 | 3432 | 3417 | 2 | 0.001 | 67363 | 9567 | 7743 | 1748 | 0.184 | 13879 | 1099 | 836 | 260 | 0.237 | 45257 | 5804 | 5793 | 2 | 0.000 | 46955 | 4732 | 4720 | 3 | 0.000 |
| 185295 | 17 | 43008929 | 43008929 | C | T | KIF18B | intronc | c.1553+41 | e10+41 | tier3 | 34219 | 4512 | 4494 | 10 | 0.002 | 110631 | 14002 | 9579 | 4259 | 0.308 | 25946 | 1668 | 996 | 669 | 0.402 | 67163 | 8199 | 8194 | 1 | 0.000 | 79687 | 6980 | 6972 | 2 | 0.000 |
| 185295 | 21 | 44524456 | 44524456 | G | A | U2AF1 | missense | c.101 | p.S34F | tier1 | 7843 | 1166 | 1157 | 8 | 0.007 | 15332 | 2695 | 1999 | 687 | 0.256 | 7658 | 909 | 734 | 172 | 0.190 | 13525 | 1904 | 1903 | 0 | 0.000 | 14386 | 2266 | 2263 | 0 | 0.000 |
| 185295 | 21 | 47847509 | 47847509 | C | T | PCNT | intronc | c.7321-27 | e34-27 | tier3 | 27599 | 3605 | 3580 | 25 | 0.007 | 79444 | 10216 | 6138 | 3984 | 0.394 | 29939 | 1712 | 1051 | 656 | 0.384 | 47606 | 5922 | 5915 | 6 | 0.000 | 64194 | 5201 | 5195 | 4 | 0.000 |
| 185295 | 22 | 39887386 | 39887386 | G | A | MGAT3 | 3_prime_untranslated_region | c.*2432 | NULL | tier3 | 4360 | 592 | 592 | 0 | 0.000 | 22192 | 2926 | 2019 | 891 | 0.306 | 7993 | 574 | 311 | 262 | 0.457 | 11291 | 1466 | 1464 | 0 | 0.000 | 11878 | 1150 | 1148 | 0 | 0.000 |
| 185295 | X | 39921391 | 39921391 | C | T | BCOR | splice_site | c.4428+1 | e9+1 | tier1 | 8025 | 986 | 984 | 1 | 0.001 | 21687 | 2820 | 1323 | 1482 | 0.158 | 5162 | 371 | 64 | 306 | 0.827 | 35766 | 4512 | 4505 | 2 | 0.000 | 19031 | 1882 | 1881 | 1 | 0.000 |
| 187374 | 1 | 44467034 | 44467034 | C | G | SLC6A9 | intronc | c.1419+28 | e9+28 | tier3 | 18382 | 2475 | 2458 | 15 | 0.006 | 12992 | 1796 | 1550 | 241 | 0.125 | NA | NA | NA | NA | NA | 31219 | 4582 | 4570 | 11 | 0.002 | 19177 | 2711 | 2662 | 48 | 0.018 |
| 187374 | 1 | 47512254 | 47512254 | G | A | CYP4X1 | missense | c.1189 | p.G397R | tier1 | 23710 | 2775 | 2766 | 9 | 0.003 | 29898 | 3978 | 2796 | 1157 | 0.293 | NA | NA | NA | NA | NA | 56781 | 7735 | 7681 | 48 | 0.006 | 33334 | 4529 | 4318 | 197 | 0.044 |
| 187374 | 1 | 176564033 | 176564033 | C | T | PAPPA2 | silent | c.1293 | p.T431 | tier1 | 10718 | 1674 | 1666 | 8 | 0.005 | 14829 | 2077 | 1311 | 734 | 0.355 | NA | NA | NA | NA | NA | 26831 | 3972 | 3936 | 31 | 0.008 | 17316 | 2573 | 2388 | 181 | 0.070 |
| 187374 | 1 | 179526224 | 179526224 | G | A | NPHS2 | silent | c.676 | p.L226 | tier1 | 35689 | 4191 | 4166 | 22 | 0.005 | 33298 | 4346 | 3044 | 1267 | 0.294 | NA | NA | NA | NA | NA | 80138 | 10706 | 10634 | 66 | 0.006 | 42424 | 5718 | 5487 | 226 | 0.040 |
| 187374 | 2 | 25463295 | 25463295 | T | A | DNMT3A | missense | c.2198 | p.E733V | tier1 | 3678 | 430 | 429 | 1 | 0.002 | 6584 | 873 | 565 | 303 | 0.349 | NA | NA | NA | NA | NA | 12858 | 2072 | 2053 | 15 | 0.007 | 8016 | 1237 | 1142 | 91 | 0.074 |
| 187374 | 2 | 37458814 | 37458814 | C | A | CEBPZ | 5_prime_untranslated_region | c.-104 | NULL | tier2 | 19902 | 2151 | 2134 | 14 | 0.007 | 19554 | 2449 | 1636 | 805 | 0.330 | NA | NA | NA | NA | NA | 36543 | 4866 | 4828 | 38 | 0.008 | 20569 | 2735 | 2574 | 150 | 0.055 |
| 187374 | 2 | 42280542 | 42280542 | G | A | PKDCC | 3_prime_untranslated_region | c.*42280542 | NULL | tier3 | 13283 | 2086 | 2076 | 8 | 0.004 | 13586 | 2073 | 1352 | 706 | 0.343 | NA | NA | NA | NA | NA | 26366 | 4754 | 4713 | 36 | 0.008 | 17954 | 3152 | 2969 | 178 | 0.057 |
| 187374 | 2 | 196673523 | 196673523 | C | A | DNAH7 | missense | c.9966 | p.W3322C | tier1 | 18655 | 2110 | 2094 | 9 | 0.004 | 19135 | 2563 | 1980 | 570 | 0.224 | NA | NA | NA | NA | NA | 43137 | 6001 | 5962 | 29 | 0.005 | 24544 | 3162 | 3057 | 97 | 0.031 |
| 187374 | 2 | 207570559 | 207570559 | G | A | DYTN | missense | c.335 | p.A112V | tier1 | 2160 | 370 | 367 | 2 | 0.005 | 1755 | 327 | 206 | 121 | 0.370 | NA | NA | NA | NA | NA | 4203 | 944 | 935 | 8 | 0.008 | 1952 | 421 | 397 | 24 | 0.057 |
| 187374 | 3 | 133667500 | 133667500 | C | T | SLCO2A1 | missense | c.985 | p.V329I | tier1 | 33511 | 3691 | 3666 | 19 | 0.005 | 28340 | 3526 | 2315 | 1167 | 0.335 | NA | NA | NA | NA | NA | 57124 | 7710 | 7637 | 62 | 0.008 | 35246 | 4692 | 4467 | 216 | 0.046 |
| 187374 | 6 | 49676860 | 49676860 | A | G | CRISP2 | missense | c.50 | p.L175 | tier1 | 12559 | 1320 | 1316 | 4 | 0.003 | 13955 | 1701 | 1102 | 579 | 0.344 | NA | NA | NA | NA | NA | 26392 | 3746 | 3702 | 40 | 0.011 | 15048 | 2009 | 1882 | 120 | 0.060 |
| 187374 | 7 | 139083345 | 139083345 | A | T | LUC7L2 | nonsense | c.355 | p.R119* | tier1 | 8170 | 870 | 868 | 1 | 0.001 | 6425 | 801 | 482 | 317 | 0.397 | NA | NA | NA | NA | NA | 19092 | 2513 | 2489 | 20 | 0.008 | 8096 | 1026 | 954 | 72 | 0.070 |
| 187374 | 9 | 101751401 | 101751401 | C | T | COL15A1 | intronc | c.724-59 | e5-59 | tier3 | 23453 | 3030 | 3001 | 23 | 0.008 | 20110 | 2820 | 1909 | 889 | 0.318 | NA | NA | NA | NA | NA | 36101 | 5615 | 5567 | 44 | 0.008 | 25441 | 3820 | 3645 | 169 | 0.044 |
| 187374 | 9 | 131708388 | 131708388 | G | A | DOLK | nonsense | c.1195 | p.R399* | tier1 | 4590 | 743 | 737 | 6 | 0.008 | 4222 | 670 | 456 | 212 | 0.317 | NA | NA | NA | NA | NA | 8862 | 1935 | 1918 | 16 | 0.008 | 4382 | 960 | 902 | 56 | 0.058 |
| 187374 | 9 | 132980111 | 132980111 | G | T | NCS1 | missense | c.90 | p.W30C | tier1 | 11268 | 1362 | 1357 | 2 | 0.001 | 9849 | 1446 | 934 | 499 | 0.348 | NA | NA | NA | NA | NA | 18614 | 3366 | 3330 | 32 | 0.010 | 11011 | 1855 | 1740 | 112 | 0.060 |
| 187374 | 10 | 25465090 | 25465090 | C | T | GPR158 | silent | c.741 | p.G247 | tier1 | 5731 | 724 | 718 | 5 | 0.007 | 4535 | 581 | 410 | 167 | 0.289 | NA | NA | NA | NA | NA | 10977 | 1648 | 1637 | 8 | 0.005 | 5191 | 767 | 726 | 34 | 0.045 |
| 187374 | 11 | 36601073 | 36601073 | T | C | RAG1 | 3_prime_untranslated_region | c.*3087 | NULL | tier2 | 557 | 42 | 42 | 0 | 0.000 | 834 | 98 | 64 | 34 | 0.347 | NA | NA | NA | NA | NA | 1373 | 197 | 196 | 1 | 0.000 | 560 | 73 | 69 | 4 | 0.055 |
| 187374 | 11 | 70779207 | 70779207 | C | T | SHANK2 | intronc | c.1072+71 | e14+71 | tier4 | 2108 | 378 | 377 | 1 | 0.003 | 1520 | 247 | 180 | 64 | 0.262 | NA | NA | NA | NA | NA | 3063 | 703 | 696 | 7 | 0.010 | 1696 | 347 | 331 | 16 | 0.046 |
| 187374 | 11 | 93439939 | 93439939 | C | A | KIAA1731 | missense | c.5625 | p.D1875E | tier1 | 7573 | 678 | 671 | 3 | 0.004 | 8150 | 846 | 542 | 300 | 0.356 | NA | NA | NA | NA | NA | 15939 | 2029 | 2012 | 17 | 0.008 | 8653 | 1087 | 1004 | 79 | 0.073 |
| 187374 | 11 | 93826848 | 93826848 | T | C | HEPHL1 | intronc | c.2434+42 | e13+42 | tier2 | 22662 | 2638 | 2623 | 12 | 0.005 | 22411 | 2829 | 1821 | 989 | 0.352 | NA | NA | NA | NA | NA | 39413 | 4962 | 4927 | 30 | 0.006 | 28176 | 3611 | 3387 | 218 | 0.060 |
| 187374 | 12 | 53699798 | 53699798 | T | G | C12orf10 | missense | c.107 | p.V36G | tier1 | 18490 | 2188 | 2182 | 4 | 0.002 | 28643 | 3793 | 3053 | 706 | 0.188 | NA | NA | NA | NA | NA | 48631 | 6846 | 6813 | 28 | 0.004 | 36841 | 5083 | 4947 | 127 | 0.025 |
| 187374 | 12 | 56811467 | 56811467 | G | A | TIMELESS | 5_prime_untranslated_region | c.-56811467 | NULL | tier3 | 10249 | 1138 | 1128 | 9 | 0.008 | 12404 | 1528 | 980 | 539 | 0.355 | NA | NA | NA | NA | NA | 24965 | 3707 | 3664 | 32 | 0.009 | 17083 | 2466 | 2319 | 141 | 0.057 |
| 187374 | 13 | 73334857 | 73334857 | C | T | DIS3 | intronc | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF | |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|-------|
| 199019 | 2 | 212288901 | 212288901 | C | T | ERBB4 | missense | c.2845 | p.V949I | tier1 | 10803 | 1014 | 1012 | 2 | 0.002 | 7060 | 740 | 589 | 146 | 0.199 | NA | NA | NA | NA | NA | 12503 | 1694 | 1692 | 1 | 0.000 | 14646 | 273 | 272 | 0 | 0.000 | |
| 199019 | 2 | 219267899 | 219267899 | C | T | CTDSP1 | intronic | c.471+49 | e+49 | tier3 | 14694 | 1478 | 1439 | 38 | 0.026 | 13162 | 1294 | 887 | 397 | 0.309 | NA | NA | NA | NA | NA | 39467 | 5308 | 5263 | 39 | 0.007 | 31541 | 500 | 500 | 0 | 0.000 | |
| 199019 | 4 | 106156453 | 106156453 | G | T | TET2 | nonsense | c.1354 | p.E452* | tier1 | 10834 | 996 | 955 | 37 | 0.037 | 8474 | 834 | 638 | 186 | 0.226 | NA | NA | NA | NA | NA | 16977 | 2319 | 2300 | 15 | 0.006 | 17092 | 286 | 286 | 0 | 0.000 | |
| 199019 | 5 | 7302322 | 7302322 | G | A | ENSG00000205976 | intronic | c.NULL | NULL | tier3 | 11932 | 1270 | 1246 | 24 | 0.019 | 7810 | 833 | 585 | 242 | 0.293 | NA | NA | NA | NA | NA | 18945 | 2643 | 2629 | 8 | 0.003 | 18343 | 245 | 245 | 0 | 0.000 | |
| 199019 | 7 | 57529430 | 57529430 | A | C | ZNF716 | silent | p.S421 | tier1 | 9585 | 1016 | 1007 | 9 | 0.009 | 5410 | 548 | 447 | 101 | 0.184 | NA | NA | NA | NA | NA | NA | 5713 | 743 | 743 | 0 | 0.000 | 18964 | 228 | 228 | 0 | 0.000 | |
| 199019 | 8 | 38835514 | 38835514 | C | T | HTRA4 | missense | c.814 | p.R272W | tier1 | 4084 | 299 | 285 | 14 | 0.047 | 4666 | 387 | 261 | 123 | 0.320 | NA | NA | NA | NA | NA | 4648 | 605 | 596 | 5 | 0.008 | 4974 | 69 | 68 | 0 | 0.000 | |
| 199019 | 11 | 46747390 | 46747390 | C | T | F2 | intronic | c.560-19 | e-19 | tier4 | 9824 | 959 | 944 | 14 | 0.015 | 5954 | 609 | 415 | 189 | 0.313 | NA | NA | NA | NA | NA | 11707 | 1655 | 1647 | 3 | 0.002 | 13687 | 191 | 191 | 0 | 0.000 | |
| 199019 | 11 | 65062076 | 65062076 | G | A | POLA2 | silent | p.L471 | tier1 | 16562 | 1974 | 1964 | 9 | 0.005 | 9872 | 1140 | 924 | 206 | 0.182 | NA | NA | NA | NA | NA | NA | 15439 | 2237 | 2226 | 9 | 0.000 | 17132 | 300 | 299 | 1 | 0.000 | |
| 199019 | 11 | 71949104 | 71949104 | G | A | INPP1 | missense | c.3571 | p.G1191R | tier1 | 15200 | 1686 | 1686 | 0 | 0.000 | 9280 | 1082 | 1033 | 47 | 0.044 | NA | NA | NA | NA | NA | 12253 | 1852 | 1850 | 1 | 0.000 | 15189 | 0 | 0 | 0 | NA | |
| 199019 | 11 | 89018012 | 89018012 | G | T | TYR | missense | c.1256 | p.G419V | tier1 | 8959 | 836 | 831 | 3 | 0.004 | 5386 | 546 | 423 | 122 | 0.224 | NA | NA | NA | NA | NA | 11470 | 1595 | 1590 | 4 | 0.003 | 11318 | 215 | 215 | 0 | 0.000 | |
| 199019 | 12 | 124241407 | 124241407 | G | A | ATP6V0A2 | missense | c.2339 | p.R780H | tier1 | 20664 | 2603 | 2550 | 48 | 0.018 | 14513 | 1805 | 1201 | 589 | 0.329 | NA | NA | NA | NA | NA | 24533 | 3871 | 3847 | 18 | 0.005 | 33089 | 498 | 498 | 0 | 0.000 | |
| 199019 | 14 | 106357572 | 106357572 | A | G | IGHD6-19 | silent | c.6 | p.Y2 | tier1 | 12705 | 1521 | 1508 | 12 | 0.008 | 7058 | 805 | 592 | 207 | 0.259 | NA | NA | NA | NA | NA | 8271 | 1276 | 1274 | 0 | 0.000 | 11138 | 240 | 240 | 0 | 0.000 | |
| 199019 | 15 | 50897271 | 50897271 | C | T | TRPM7 | missense | c.2780 | p.S927N | tier1 | 7473 | 768 | 761 | 6 | 0.008 | 4586 | 531 | 368 | 159 | 0.302 | NA | NA | NA | NA | NA | 8393 | 1257 | 1252 | 2 | 0.000 | 10511 | 242 | 242 | 0 | 0.000 | |
| 199019 | 16 | 4736332 | 4736332 | G | A | MGRN1 | 3_prime_untranslated_region | c.*32 | NULL | tier2 | 8269 | 796 | 776 | 19 | 0.024 | 4986 | 598 | 418 | 175 | 0.295 | NA | NA | NA | NA | NA | 7903 | 1257 | 1248 | 9 | 0.007 | 11765 | 242 | 242 | 0 | 0.000 | |
| 199019 | 17 | 7578445 | 7578445 | A | T | TP53 | missense | c.485 | p.I162N | tier1 | 20913 | 2310 | 2229 | 78 | 0.034 | 11259 | 1203 | 489 | 705 | 0.590 | NA | NA | NA | NA | NA | NA | 30370 | 3906 | 3879 | 25 | 0.006 | 36911 | 427 | 426 | 0 | 0.000 |
| 199019 | 17 | 61898424 | 61898424 | A | G | FTSJ3 | silent | c.1938 | p.D646 | tier1 | 32049 | 3302 | 3159 | 125 | 0.038 | 21842 | 2214 | 1447 | 734 | 0.337 | NA | NA | NA | NA | NA | 41729 | 5776 | 5724 | 49 | 0.008 | 49762 | 746 | 746 | 0 | 0.000 | |
| 199019 | 17 | 72589154 | 72589154 | T | C | C17orf77 | 3_prime_untranslated_region | c.*237 | NULL | tier3 | 2098 | 168 | 163 | 4 | 0.024 | 1336 | 130 | 70 | 60 | 0.462 | NA | NA | NA | NA | NA | 1207 | 181 | 178 | 3 | 0.017 | 4356 | 99 | 99 | 0 | 0.000 | |
| 199019 | 17 | 75209492 | 75209492 | G | A | SEC14L1 | missense | p.D654N | tier1 | 22753 | 2266 | 2264 | 1 | 0.000 | 14132 | 1349 | 1198 | 138 | 0.103 | NA | NA | NA | NA | NA | NA | 24385 | 3201 | 3193 | 3 | 0.000 | 32465 | 489 | 488 | 0 | 0.000 | |
| 199019 | X | 46513121 | 46513121 | A | T | SLC9A7 | missense | c.1067 | p.F356Y | tier1 | 5001 | 733 | 726 | 7 | 0.010 | 3527 | 529 | 379 | 147 | 0.279 | NA | NA | NA | NA | NA | 2380 | 579 | 575 | 3 | 0.005 | 3081 | 129 | 129 | 0 | 0.000 | |
| 199019 | X | 132437274 | 132437274 | C | T | GPC4 | missense | c.1388 | p.R463H | tier1 | 8105 | 808 | 797 | 11 | 0.014 | 5560 | 537 | 388 | 148 | 0.276 | NA | NA | NA | NA | NA | NA | 6138 | 825 | 814 | 10 | 0.012 | 5139 | 87 | 87 | 0 | 0.000 |
| 221833 | 1 | 3566445 | 3566445 | A | C | WRAP73 | intronic | c.69+50 | e1+50 | tier2 | 12673 | 1394 | 1370 | 22 | 0.016 | 14612 | 1766 | 1007 | 741 | 0.424 | 9654 | 742 | 714 | 28 | 0.038 | 24767 | 3365 | 3358 | 2 | 0.000 | 40647 | 5815 | 5810 | 1 | 0.000 | |
| 221833 | 2 | 59482372 | 59482372 | G | A | LOC101927285 | rna | NULL | NULL | tier1 | 30083 | 3504 | 3476 | 21 | 0.006 | 27872 | 3237 | 2355 | 839 | 0.263 | 15680 | 1369 | 1337 | 29 | 0.021 | 37259 | 4749 | 4742 | 3 | 0.001 | 64870 | 8520 | 8504 | 1 | 0.000 | |
| 221833 | 2 | 74742861 | 74742861 | C | A | TLX2 | missense | c.502 | p.Q168K | tier1 | 140 | 0 | 0 | 0 | 0.000 | 789 | 66 | 21 | 45 | 0.682 | 251 | 11 | 11 | 0 | 0.000 | 881 | 98 | 98 | 0 | 0.000 | 1602 | 193 | 192 | 0 | 0.000 | |
| 221833 | 2 | 86923062 | 86923062 | T | A | CHMP3 | intronic | c.1-7614 | e1-7614 | tier4 | 20740 | 2330 | 2253 | 74 | 0.032 | 26026 | 2919 | 1097 | 1774 | 0.618 | 14545 | 1214 | 1107 | 98 | 0.081 | 30356 | 3916 | 3894 | 13 | 0.003 | 55953 | 7036 | 7020 | 0 | 0.000 | |
| 221833 | 2 | 198638852 | 198638852 | G | A | ENSG00000222017 | 3_prime_untranslated_region | c.*71 | NULL | tier3 | 8677 | 1095 | 1084 | 8 | 0.007 | 9797 | 1314 | 989 | 313 | 0.240 | 5521 | 577 | 566 | 10 | 0.017 | 12615 | 1904 | 1902 | 1 | 0.000 | 18821 | 3192 | 3190 | 0 | 0.000 | |
| 221833 | 3 | 38766591 | 38766591 | C | A | SCN10A | intronic | c.3228+74 | e17+74 | tier3 | 8106 | 1584 | 1569 | 13 | 0.008 | 7231 | 1323 | 757 | 554 | 0.423 | 6156 | 871 | 831 | 39 | 0.045 | 12094 | 2566 | 2561 | 4 | 0.002 | 22037 | 5407 | 5403 | 1 | 0.000 | |
| 221833 | 3 | 57616622 | 57616622 | G | A | DENND6A | intronic | c.1407+30 | e16+30 | tier3 | 989 | 138 | 137 | 1 | 0.007 | 2530 | 330 | 195 | 133 | 0.405 | 2802 | 242 | 231 | 11 | 0.045 | 4159 | 606 | 604 | 0 | 0.000 | 6817 | 1005 | 1005 | 0 | 0.000 | |
| 221833 | 3 | 97806175 | 97806175 | C | T | OR54C | silent | c.159 | p.N53 | tier1 | 36366 | 4540 | 4475 | 61 | 0.013 | 42497 | 5143 | 2863 | 2207 | 0.435 | 32028 | 3014 | 2868 | 134 | 0.045 | 70471 | 9583 | 9575 | 5 | 0.000 | 138668 | 17965 | 17934 | 3 | 0.000 | |
| 221833 | 4 | 115898324 | 115898324 | A | G | NDST4 | intronic | c.1066+19 | e2+19 | tier3 | 9917 | 1076 | 1070 | 6 | 0.006 | 11131 | 1177 | 766 | 401 | 0.344 | 11510 | 905 | 841 | 62 | 0.069 | 21535 | 2633 | 2625 | 1 | 0.000 | 36675 | 4698 | 4688 | 0 | 0.000 | |
| 221833 | 5 | 13901373 | 13901373 | C | T | DNAH5 | silent | c.2040 | p.A680 | tier1 | 25893 | 3110 | 3059 | 51 | 0.016 | 23822 | 2809 | 1645 | 1130 | 0.407 | 19890 | 1649 | 1563 | 71 | 0.043 | 38164 | 4915 | 4908 | 3 | 0.000 | 78953 | 10064 | 10050 | 4 | 0.000 | |
| 221833 | 6 | 33381539 | 33381539 | G | A | PHF1 | missense | c.614 | p.R205Q | tier1 | 15906 | 1875 | 1769 | 102 | 0.055 | 23175 | 2738 | 1414 | 1303 | 0.480 | 14525 | 1154 | 734 | 411 | 0.359 | 27727 | 3409 | 3401 | 7 | 0.002 | 55834 | 6834 | 6801 | 9 | 0.001 | |
| 221833 | 7 | 36396735 | 36396735 | G | A | KIAA0895 | missense | c.643 | p.P215S | tier1 | 1273 | 188 | 184 | 3 | 0.016 | 1215 | 172 | 124 | 48 | 0.279 | 877 | 82 | 79 | 3 | 0.037 | 3222 | 463 | 462 | 1 | 0.000 | 7875 | 1523 | 1522 | 0 | 0.000 | |
| 221833 | 7 | 63032656 | 63032656 | G | A | TNRC18P2 | intronic | c.NULL | NULL | tier2 | 1219 | 221 | 220 | 1 | 0.005 | 1165 | 236 | 180 | 56 | 0.237 | 329 | 33 | 31 | 2 | 0.000 | 1729 | 325 | 324 | 0 | 0.000 | 3386 | 772 | 771 | 0 | 0.000 | |
| 221833 | 7 | 74482588 | 74482588 | C | T | WBSCR16 | missense | c.472 | p.V158M | tier1 | 14272 | 1754 | 1704 | 48 | 0.027 | 14578 | 1890 | 1105 | 765 | 0.409 | 11661 | 1069 | 945 | 122 | 0.114 | 23290 | 3271 | 3260 | 5 | 0.002 | 46754 | 6677 | 6671 | 3 | 0.000 | |
| 221833 | 9 | 138642911 | 138642911 | G | A | KCN11 | intronic | c.434+24 | e4+24 | tier3 | 18945 | 2191 | 2124 | 64 | 0.029 | 14896 | 1690 | 905 | 770 | 0.460 | 11753 | 950 | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 243777 | 1 | 3428641 | 3428641 | T | C | MEGF6 | missense | c.905 | p.N302S | tier1 | 47798 | 5255 | 5247 | 6 | 0.001 | 34014 | 4096 | 3761 | 316 | 0.078 | NA | NA | NA | NA | NA | 66578 | 9124 | 9116 | 0 | 0.000 | 116508 | 16030 | 16007 | 0 | 0.000 |
| 243777 | 1 | 24383863 | 24383863 | C | T | MYOM3 | silent | c.4314 | p.K1438 | tier1 | 19547 | 2264 | 2197 | 63 | 0.028 | 10682 | 1405 | 1027 | 370 | 0.265 | NA | NA | NA | NA | NA | 23625 | 3618 | 3615 | 1 | 0.000 | 50047 | 7861 | 7851 | 0 | 0.000 |
| 243777 | 1 | 230921753 | 230921753 | A | G | CAPN9 | missense | c.1508 | p.D503G | tier1 | 22696 | 2680 | 2678 | 0 | 0.000 | 16707 | 2266 | 2171 | 87 | 0.039 | NA | NA | NA | NA | NA | 32519 | 5023 | 5012 | 1 | 0.000 | 42813 | 7340 | 7335 | 0 | 0.000 |
| 243777 | 2 | 103124587 | 103124587 | C | T | SLC9A4 | silent | c.1248 | p.F416 | tier1 | 22627 | 2438 | 2425 | 8 | 0.003 | 16664 | 1902 | 1700 | 193 | 0.102 | NA | NA | NA | NA | NA | 34960 | 4647 | 4642 | 1 | 0.000 | 53972 | 7229 | 7218 | 0 | 0.000 |
| 243777 | 2 | 152534686 | 152534686 | C | G | NEB | missense | c.3271 | p.D1091H | tier1 | 8375 | 762 | 735 | 23 | 0.030 | 6759 | 698 | 538 | 158 | 0.227 | NA | NA | NA | NA | NA | 13477 | 1743 | 1741 | 1 | 0.000 | 22802 | 3175 | 3172 | 0 | 0.000 |
| 243777 | 3 | 58870344 | 58870344 | C | T | C3orf67 | silent | c.267 | p.Q89 | tier1 | 3330 | 452 | 436 | 16 | 0.035 | 3569 | 601 | 457 | 144 | 0.240 | NA | NA | NA | NA | NA | 6408 | 1316 | 1315 | 0 | 0.000 | 8656 | 1950 | 1948 | 2 | 0.000 |
| 243777 | 4 | 2932778 | 2932778 | G | A | MFSD10 | missense | c.1262 | p.A421V | tier1 | 7096 | 673 | 669 | 2 | 0.003 | 11229 | 1548 | 1227 | 312 | 0.203 | NA | NA | NA | NA | NA | 22441 | 3640 | 3636 | 1 | 0.000 | 37714 | 6377 | 6374 | 2 | 0.000 |
| 243777 | 5 | 82833952 | 82833952 | C | T | VCAN | silent | c.5130 | p.D1710 | tier1 | 2852 | 293 | 291 | 1 | 0.003 | 4791 | 783 | 766 | 14 | 0.018 | NA | NA | NA | NA | NA | 7568 | 1416 | 1414 | 0 | 0.000 | 13561 | 2847 | 2842 | 1 | 0.000 |
| 243777 | 5 | 140201927 | 140201927 | C | A | PCDH5A | missense | c.567 | p.N189K | tier1 | 15891 | 1566 | 1546 | 16 | 0.010 | 14937 | 1745 | 1372 | 364 | 0.210 | NA | NA | NA | NA | NA | 31992 | 4432 | 4426 | 5 | 0.000 | 52355 | 7894 | 7882 | 0 | 0.000 |
| 243777 | 6 | 7829666 | 7829666 | G | A | BMP6 | intronic | c.665-15707 | e2-15707 | tier4 | 4290 | 487 | 477 | 9 | 0.019 | 4377 | 567 | 403 | 162 | 0.287 | NA | NA | NA | NA | NA | 8651 | 1296 | 1295 | 0 | 0.000 | 8860 | 1588 | 1587 | 0 | 0.000 |
| 243777 | 6 | 42985369 | 42985369 | C | A | KLHDC3 | silent | c.267 | p.V89 | tier1 | 9228 | 1036 | 1030 | 2 | 0.002 | 16814 | 2347 | 2065 | 267 | 0.114 | NA | NA | NA | NA | NA | 29057 | 4779 | 4778 | 0 | 0.000 | 50504 | 8987 | 8981 | 0 | 0.000 |
| 243777 | 6 | 42985377 | 42985377 | G | A | KLHDC3 | nonsense | c.275 | p.W92* | tier1 | 9020 | 1024 | 1013 | 3 | 0.003 | 16185 | 2304 | 2003 | 274 | 0.120 | NA | NA | NA | NA | NA | 28330 | 4709 | 4699 | 0 | 0.000 | 49613 | 8924 | 8916 | 1 | 0.000 |
| 243777 | 6 | 64290808 | 64290808 | G | A | PTPA41 | 3_prime_untranslated_region | c.*729 | NULL | tier3 | 4544 | 490 | 485 | 4 | 0.008 | 3755 | 395 | 334 | 61 | 0.154 | NA | NA | NA | NA | NA | 6555 | 800 | 800 | 0 | 0.000 | 14693 | 1912 | 1908 | 0 | 0.000 |
| 243777 | 7 | 135263622 | 135263622 | C | T | NUP205 | missense | c.1001 | p.A334V | tier1 | 11944 | 1309 | 1276 | 29 | 0.022 | 15884 | 1819 | 1347 | 461 | 0.255 | NA | NA | NA | NA | NA | 27824 | 3567 | 3563 | 3 | 0.000 | 56597 | 7291 | 7263 | 2 | 0.000 |
| 243777 | 8 | 117859879 | 117859879 | G | A | RAD21 | nonsense | c.1756 | p.R586* | tier1 | 7435 | 729 | 724 | 2 | 0.003 | 9929 | 1059 | 943 | 112 | 0.106 | NA | NA | NA | NA | NA | 18835 | 2368 | 2364 | 2 | 0.000 | 31132 | 4057 | 4057 | 0 | 0.000 |
| 243777 | 9 | 108125237 | 108125237 | G | A | SLC44A1 | missense | c.1036 | p.V346I | tier1 | 4098 | 611 | 598 | 11 | 0.018 | 3651 | 620 | 502 | 116 | 0.188 | NA | NA | NA | NA | NA | 6819 | 1356 | 1354 | 0 | 0.000 | 11488 | 2588 | 2585 | 0 | 0.000 |
| 243777 | 10 | 54076396 | 54076396 | T | C | DKK1 | silent | c.630 | p.C210 | tier1 | 11704 | 1458 | 1417 | 41 | 0.028 | 6320 | 772 | 543 | 228 | 0.296 | NA | NA | NA | NA | NA | 10253 | 1441 | 1433 | 4 | 0.003 | 26513 | 4029 | 4012 | 1 | 0.000 |
| 243777 | 10 | 135350563 | 135350563 | G | T | CYP2E1 | splice_region | c.968-4 | e7-4 | tier3 | 15475 | 1702 | 1677 | 22 | 0.013 | 11458 | 1294 | 1022 | 268 | 0.208 | NA | NA | NA | NA | NA | 21275 | 2954 | 2953 | 0 | 0.000 | 34953 | 5018 | 5016 | 1 | 0.000 |
| 243777 | 11 | 67552796 | 67552796 | G | T | ALG1L8P | 3_prime_untranslated_region | c.*67552796 | NULL | tier3 | 7882 | 748 | 741 | 6 | 0.008 | 6391 | 684 | 520 | 164 | 0.240 | NA | NA | NA | NA | NA | 14249 | 1756 | 1756 | 0 | 0.000 | 24092 | 3075 | 3075 | 0 | 0.000 |
| 243777 | 11 | 71175151 | 71175151 | C | A | NADSYN1 | missense | c.370 | p.R124S | tier1 | 20939 | 2924 | 2886 | 28 | 0.010 | 34569 | 4519 | 3606 | 873 | 0.195 | NA | NA | NA | NA | NA | 62386 | 9159 | 9151 | 2 | 0.000 | 104320 | 14866 | 14851 | 4 | 0.000 |
| 243777 | 11 | 132081872 | 132081872 | T | A | NTM | intronic | c.401-44 | e4-44 | tier3 | 12716 | 1184 | 1158 | 25 | 0.021 | 10366 | 1094 | 855 | 233 | 0.214 | NA | NA | NA | NA | NA | 18915 | 2518 | 2515 | 2 | 0.000 | 35985 | 4874 | 4867 | 0 | 0.000 |
| 243777 | 12 | 11992117 | 11992117 | G | A | ETV6 | nonsense | c.207 | p.W69* | tier1 | 33041 | 3548 | 3526 | 13 | 0.004 | 25437 | 2831 | 2496 | 318 | 0.113 | NA | NA | NA | NA | NA | 48212 | 6185 | 6184 | 0 | 0.000 | 91635 | 11949 | 11938 | 0 | 0.000 |
| 243777 | 16 | 51175096 | 51175096 | G | A | SALL1 | missense | c.1532 | p.A346V | tier1 | 15532 | 1553 | 1509 | 42 | 0.027 | 16802 | 1918 | 1412 | 492 | 0.258 | NA | NA | NA | NA | NA | 29105 | 3770 | 3765 | 3 | 0.000 | 49464 | 6730 | 6722 | 3 | 0.000 |
| 243777 | 17 | 10451010 | 10451010 | G | A | MYH2 | intronic | c.204+24 | e1+24 | tier3 | 26858 | 2722 | 2685 | 23 | 0.008 | 23033 | 2555 | 2039 | 494 | 0.195 | NA | NA | NA | NA | NA | 39719 | 5095 | 5089 | 2 | 0.000 | 72769 | 9624 | 9612 | 1 | 0.000 |
| 243777 | 17 | 78313118 | 78313118 | C | G | RNF213 | missense | c.4951 | p.L1651V | tier1 | 12490 | 1398 | 1365 | 28 | 0.020 | 25412 | 2949 | 2179 | 743 | 0.254 | NA | NA | NA | NA | NA | 34291 | 4477 | 4474 | 0 | 0.000 | 55311 | 7290 | 7285 | 0 | 0.000 |
| 243777 | 18 | 42531917 | 42531917 | T | C | SETBP1 | missense | c.2612 | p.I871T | tier1 | 29045 | 3318 | 3307 | 10 | 0.003 | 28940 | 3401 | 3292 | 96 | 0.028 | NA | NA | NA | NA | NA | 48772 | 6435 | 6431 | 0 | 0.000 | 78461 | 10289 | 10276 | 0 | 0.000 |
| 243777 | 18 | 77896475 | 77896475 | A | T | ADNP2 | missense | c.3179 | p.D1060V | tier1 | 2959 | 248 | 244 | 4 | 0.016 | 3053 | 287 | 221 | 64 | 0.225 | NA | NA | NA | NA | NA | 6457 | 762 | 761 | 0 | 0.000 | 4593 | 560 | 560 | 0 | 0.000 |
| 243777 | 19 | 1525633 | 1525633 | C | T | PLK5 | 5_prime_untranslated_region | c.-1363 | NULL | tier2 | 38807 | 4443 | 4372 | 59 | 0.013 | 31055 | 3789 | 3055 | 701 | 0.187 | NA | NA | NA | NA | NA | 64060 | 8600 | 8586 | 6 | 0.000 | 88350 | 12391 | 12363 | 0 | 0.000 |
| 243777 | 19 | 51358189 | 51358189 | T | A | KLK3 | 5_prime_untranslated_region | c.-23 | NULL | tier3 | 6819 | 572 | 555 | 15 | 0.026 | 5847 | 628 | 449 | 176 | 0.282 | NA | NA | NA | NA | NA | 11601 | 1447 | 1446 | 0 | 0.000 | 18744 | 2464 | 2460 | 2 | 0.000 |
| 243777 | 19 | 57722941 | 57722941 | A | G | ZNF264 | missense | c.476 | p.D159G | tier1 | 2742 | 330 | 330 | 0 | 0.000 | 3665 | 574 | 513 | 60 | 0.105 | NA | NA | NA | NA | NA | 7563 | 1361 | 1359 | 0 | 0.000 | 8522 | 1895 | 1894 | 0 | 0.000 |
| 243777 | 20 | 3214593 | 3214593 | C | T | SLC4A11 | silent | c.708 | p.R236 | tier1 | 29045 | 3167 | 3081 | 78 | 0.025 | 30713 | 3700 | 2683 | 983 | 0.268 | NA | NA | NA | NA | NA | 62507 | 8355 | 8341 | 6 | 0.001 | 95373 | 12885 | 12860 | 2 | 0.000 |
| 243777 | 21 | 44514777 | 44514777 | T | G | U2AF1 | missense | c.470 | p.Q157P | tier1 | 38655 | 4203 | 4150 | 44 | 0.010 | 31107 | 3449 | 2647 | 775 | 0.226 | NA | NA | NA | NA | NA | 59638 | 7201 | 7198 | 1 | 0.000 | 120187 | 14390 | 14365 | 0 | 0.000 |
| 266995 | 1 | 24417462 | 24417462 | C | A | MYOM3 | missense | c.1260 | p.W420C | tier1 | 43336 | 5494 | 5485 | 4 | 0.001 | 33505 | 3643 | 3483 | 155 | 0.043 | 20125 | 2128 | 2114 | 12 | 0.006 | 11871 | 1038 | 1036 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 266995 | 1 | 181706618 | 181706618 | G | A | CACNA1E | intronic | c.3423-43 | e23-43 | tier3 | 70463 | 9735 | 9678 | 46 | 0.005 | 40500 | 4603 | 4293 | 289 | 0.063 | 18267 | 2079 | 1097 | 970 | 0.469 | 17477 | 1623 | 1618 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 266995 | 3 | 39230412 | 39230412 | C | T | XIRP1 | silent | c.525 | p.L175 | tier1 | 4494 | 539 | 533 | 3 | 0.006 | 7456 | 722 | 623 | 44 | 0.061 | 7423 | 798 | 460 | 334 | 0.421 | 2735 | 192 | 192 | 0 | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF | |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|----|
| 280837 | 1 | 1669668 | 1669668 | G | C | SLC35E2 | intronic | c.586+92 | e3+92 | tier2 | 9681 | 1209 | 1190 | 19 | 0.016 | 10714 | 998 | 534 | 462 | 0.464 | 11692 | 1479 | 826 | 640 | 0.437 | 60277 | 2293 | 2262 | 31 | 0.014 | NA | NA | NA | NA | NA | |
| 280837 | 1 | 28605680 | 28605680 | G | A | SES2N | silent | c.1284 | p.V428 | tier1 | 13319 | 1360 | 1352 | 7 | 0.005 | 14899 | 2048 | 1169 | 873 | 0.428 | 19279 | 2400 | 1402 | 972 | 0.409 | 102550 | 7789 | 7714 | 67 | 0.009 | NA | NA | NA | NA | NA | |
| 280837 | 2 | 25467497 | 25467497 | G | A | DNMT3A | nonsense | c.1579 | p.Q527* | tier1 | 70333 | 10023 | 9820 | 176 | 0.018 | 51291 | 6893 | 3714 | 3137 | 0.458 | 64265 | 9581 | 5279 | 4051 | 0.434 | 278961 | 16532 | 16337 | 173 | 0.010 | NA | NA | NA | NA | NA | |
| 280837 | 2 | 131453866 | 131453866 | T | C | ENSG00000240253 | intronic | c.NULL | NULL | tier3 | 6258 | 584 | 582 | 2 | 0.003 | 5510 | 740 | 410 | 330 | 0.446 | 6138 | 628 | 348 | 278 | 0.444 | 27099 | 2343 | 2320 | 22 | 0.009 | NA | NA | NA | NA | NA | |
| 280837 | 2 | 234184342 | 234184342 | G | A | ATG16L1 | intronic | c.954+918 | e9+918 | tier4 | 31935 | 4714 | 4707 | 7 | 0.001 | 24213 | 3828 | 3112 | 707 | 0.185 | 31204 | 4812 | 4478 | 317 | 0.066 | 124297 | 8955 | 8938 | 11 | 0.001 | NA | NA | NA | NA | NA | |
| 280837 | 3 | 118913221 | 118913221 | C | T | UPK1B | silent | c.624 | p.G208 | tier1 | 39622 | 5047 | 5017 | 29 | 0.006 | 20108 | 2658 | 1488 | 1162 | 0.438 | 26153 | 3649 | 2269 | 1355 | 0.374 | 127966 | 7136 | 7057 | 75 | 0.011 | NA | NA | NA | NA | NA | |
| 280837 | 4 | 1949812 | 1949812 | A | C | WHSC1 | 3_prime_untranslated_region | c.*5684 | NULL | tier3 | 6331 | 1296 | 1289 | 7 | 0.005 | 7076 | 1601 | 902 | 696 | 0.436 | 7007 | 1524 | 850 | 665 | 0.439 | 37456 | 4426 | 4379 | 47 | 0.011 | NA | NA | NA | NA | NA | |
| 280837 | 4 | 71888174 | 71888174 | G | T | DKC | missense | c.298 | p.A100S | tier1 | 5890 | 646 | 642 | 4 | 0.006 | 6429 | 1102 | 621 | 480 | 0.436 | 5748 | 744 | 477 | 262 | 0.355 | 29463 | 3599 | 3565 | 30 | 0.008 | NA | NA | NA | NA | NA | |
| 280837 | 4 | 76817552 | 76817552 | G | C | PPEF2 | 5_prime_untranslated_region | c.-74 | NULL | tier2 | 42602 | 5574 | 5560 | 13 | 0.002 | 28801 | 4099 | 2468 | 1613 | 0.395 | 30910 | 4269 | 2964 | 1262 | 0.299 | 199372 | 11731 | 11635 | 84 | 0.007 | NA | NA | NA | NA | NA | |
| 280837 | 4 | 109769882 | 109769882 | C | T | COL25A1 | intronic | c.1434+33 | e26+33 | tier3 | 2621 | 967 | 958 | 9 | 0.009 | 2906 | 724 | 414 | 310 | 0.428 | 3662 | 1188 | 659 | 529 | 0.445 | 18032 | 2053 | 2039 | 14 | 0.007 | NA | NA | NA | NA | NA | |
| 280837 | 5 | 74712651 | 74712651 | C | A | COL4A3BP | intronic | c.1221+50 | e8+50 | tier3 | 8707 | 875 | 872 | 2 | 0.002 | 6129 | 801 | 466 | 334 | 0.418 | 8869 | 1013 | 696 | 311 | 0.309 | 76736 | 6352 | 6305 | 46 | 0.007 | NA | NA | NA | NA | NA | |
| 280837 | 6 | 33136506 | 33136506 | C | T | COL11A2 | missense | c.3883 | p.E1295K | tier1 | 57055 | 6946 | 6901 | 42 | 0.006 | 25285 | 2410 | 1348 | 1055 | 0.439 | 34912 | 4264 | 2439 | 1786 | 0.423 | 134392 | 5779 | 5727 | 52 | 0.009 | NA | NA | NA | NA | NA | |
| 280837 | 7 | 150714308 | 150714308 | C | T | ATG9B | 5_prime_untranslated_region | c.-150714308 | NULL | tier2 | 40833 | 5544 | 5536 | 4 | 0.001 | 15091 | 1946 | 1318 | 613 | 0.317 | 19717 | 2684 | 2446 | 230 | 0.086 | 197641 | 12873 | 12844 | 17 | 0.001 | NA | NA | NA | NA | NA | |
| 280837 | 8 | 131129068 | 131129068 | C | T | ASAP1 | intronic | c.2005+58 | e21+58 | tier3 | 3325 | 612 | 605 | 6 | 0.010 | 4520 | 1144 | 642 | 501 | 0.438 | 4563 | 974 | 561 | 411 | 0.423 | 21880 | 2898 | 2864 | 33 | 0.011 | NA | NA | NA | NA | NA | |
| 280837 | 9 | 139903977 | 139903977 | G | A | ABCA2 | intronic | c.6824+16 | e44+16 | tier2 | 966 | 63 | 62 | 1 | 0.016 | 6142 | 788 | 92 | 696 | 0.883 | 4670 | 633 | 77 | 554 | 0.878 | 15744 | 2436 | 2339 | 96 | 0.039 | NA | NA | NA | NA | NA | |
| 280837 | 10 | 38651191 | 38651191 | G | A | HSD17B7P2 | 3_prime_untranslated_region | c.*38651191 | NULL | tier3 | 18221 | 2023 | 2015 | 8 | 0.004 | 16976 | 2382 | 1369 | 1010 | 0.425 | 19355 | 2319 | 1407 | 897 | 0.389 | 111636 | 8539 | 8418 | 109 | 0.013 | NA | NA | NA | NA | NA | |
| 280837 | 10 | 68139043 | 68139043 | T | C | CTNNA3 | silent | c.1599 | p.L533 | tier1 | 44029 | 6712 | 6687 | 23 | 0.003 | 26718 | 3792 | 2344 | 1435 | 0.380 | 33542 | 5048 | 3133 | 1859 | 0.372 | 161892 | 9926 | 9843 | 77 | 0.008 | NA | NA | NA | NA | NA | |
| 280837 | 10 | 119044294 | 119044294 | T | C | PDZD8 | silent | c.1950 | p.Q650 | tier1 | 56391 | 7083 | 7072 | 10 | 0.001 | 27321 | 3874 | 2478 | 1294 | 0.343 | 28831 | 3811 | 2909 | 870 | 0.230 | 248111 | 15317 | 15248 | 68 | 0.004 | NA | NA | NA | NA | NA | |
| 280837 | 11 | 4107876 | 4107876 | G | T | STIM1 | intronic | c.1541+103 | e11+103 | tier3 | 10940 | 1341 | 1332 | 9 | 0.007 | 5943 | 599 | 495 | 100 | 0.168 | 7722 | 1005 | 881 | 117 | 0.117 | 5489 | 831 | 820 | 11 | 0.013 | NA | NA | NA | NA | NA | |
| 280837 | 11 | 12225810 | 12225810 | G | T | MICAL2 | missense | c.278 | p.G93A | tier1 | 59097 | 7374 | 7316 | 55 | 0.007 | 25404 | 4645 | 1379 | 1260 | 0.477 | 31790 | 4467 | 2317 | 2104 | 0.476 | 148293 | 4929 | 4866 | 63 | 0.013 | NA | NA | NA | NA | NA | |
| 280837 | 12 | 14689595 | 14689595 | A | G | PLBD1 | missense | c.608 | p.L203S | tier1 | 7917 | 901 | 891 | 9 | 0.010 | 7044 | 832 | 480 | 351 | 0.422 | 8647 | 1018 | 613 | 397 | 0.393 | 54095 | 2606 | 2576 | 29 | 0.011 | NA | NA | NA | NA | NA | |
| 280837 | 17 | 1563288 | 1563288 | T | A | PRPF8 | missense | c.4793 | p.D1598V | tier1 | 53750 | 6375 | 6329 | 36 | 0.006 | 40103 | 5019 | 2849 | 2148 | 0.430 | 52592 | 6484 | 3701 | 2660 | 0.418 | 273050 | 15221 | 15037 | 164 | 0.011 | NA | NA | NA | NA | NA | |
| 280837 | 17 | 27210235 | 27210235 | C | T | FLOT2 | silent | c.402 | p.T134 | tier1 | 16063 | 2496 | 2410 | 81 | 0.033 | 21267 | 3874 | 2150 | 1711 | 0.443 | 26403 | 3875 | 2134 | 1690 | 0.442 | 139781 | 13386 | 13242 | 130 | 0.010 | NA | NA | NA | NA | NA | |
| 280837 | 17 | 73910078 | 73910078 | C | T | FBF1 | silent | c.3003 | p.Q100I | tier1 | 40681 | 4966 | 4946 | 19 | 0.004 | 21923 | 2287 | 1507 | 775 | 0.340 | 28674 | 3794 | 2479 | 1285 | 0.341 | 172441 | 7316 | 7263 | 51 | 0.007 | NA | NA | NA | NA | NA | |
| 280837 | 18 | 5891961 | 5891961 | C | G | TMEM200C | missense | c.102 | p.R34S | tier1 | 72630 | 8874 | 8814 | 54 | 0.006 | 45356 | 4284 | 2653 | 2147 | 0.447 | 48621 | 5914 | 3328 | 2469 | 0.426 | 317261 | 13074 | 12898 | 169 | 0.013 | NA | NA | NA | NA | NA | |
| 280837 | 19 | 9001863 | 9001863 | G | A | MUC16 | nonsense | c.904 | p.R302* | tier1 | 16873 | 2093 | 2081 | 12 | 0.006 | 8565 | 884 | 521 | 361 | 0.409 | 12012 | 1511 | 890 | 613 | 0.408 | 59170 | 2293 | 2275 | 18 | 0.008 | NA | NA | NA | NA | NA | |
| 280837 | 20 | 30409206 | 30409206 | C | T | MYLK2 | intronic | c.474-36 | e3-36 | tier3 | 24229 | 2778 | 2770 | 8 | 0.003 | 35463 | 3643 | 2316 | 1314 | 0.362 | 47807 | 5947 | 3804 | 2069 | 0.352 | 333071 | 13372 | 13277 | 80 | 0.006 | NA | NA | NA | NA | NA | |
| 280837 | X | 129147939 | 129147939 | G | A | BCORL1 | missense | c.1191 | p.M397I | tier1 | 29995 | 4025 | 3955 | 55 | 0.014 | 17931 | 1969 | 234 | 1730 | 0.881 | 24280 | 3188 | 469 | 2707 | 0.852 | 99901 | 6449 | 6284 | 142 | 0.022 | NA | NA | NA | NA | NA | |
| 283773 | 9 | 20923717 | 20923717 | G | A | FOCAD | missense | c.2911 | p.V971I | tier1 | 19746 | 2206 | 2203 | 3 | 0.001 | 15008 | 1546 | 1249 | 290 | 0.188 | 19650 | 2393 | 1998 | 381 | 0.160 | 11988 | 1046 | 1043 | 0 | 0.000 | 34366 | 4442 | 4433 | 1 | 0.000 | |
| 283773 | 19 | 2121672 | 2121672 | C | T | AP3D1 | intronic | c.1101+61 | e12+61 | tier3 | 20522 | 2418 | 2411 | 0 | 0.000 | 12541 | 1319 | 1096 | 218 | 0.166 | 18871 | 2400 | 2149 | 242 | 0.101 | 11961 | 1035 | 1035 | 0 | 0.000 | 29951 | 3921 | 3913 | 0 | 0.000 | |
| 283773 | 20 | 31023112 | 31023112 | T | G | ASXL1 | nonsense | c.2597 | p.L866* | tier1 | 21481 | 2590 | 2585 | 2 | 0.001 | 17274 | 1785 | 1528 | 243 | 0.137 | 23913 | 2958 | 2620 | 335 | 0.113 | 17173 | 1473 | 1470 | 0 | 0.000 | 41409 | 5450 | 5440 | 0 | 0.000 | |
| 283773 | X | 70339865 | 70339865 | T | A | MED12 | missense | c.398 | p.V133D | tier1 | 23909 | 2632 | 2624 | 5 | 0.002 | 24201 | 2615 | 2505 | 96 | 0.037 | 29792 | 3513 | 3462 | 49 | 0.014 | 9035 | 687 | 685 | 0 | 0.000 | 25021 | 0 | 0 | 0 | 0 | NA |
| 283773 | X | 106788960 | 106788960 | T | C | FRMPD3-AS1 | rna | NULL | NULL | tier1 | 24504 | 2819 | 2789 | 24 | 0.009 | 18288 | 1879 | 1688 | 180 | 0.096 | 19702 | 2363 | 2120 | 237 | 0.101 | 6765 | 531 | 530 | 0 | 0.000 | 16237 | 0 | 0 | 0 | 0 | NA |
| 288033 | 2 | 33614218 | 33614218 | G | T | LTPB1 | intronic | c.4715-33 | e32-33 | tier3 | 21884 | 2522 | 2455 | 66 | 0.026 | 21049 | 2058 | 1174 | 851 | 0.420 | 44295 | 6019 | 5863 | 148 | 0.025 | 21500 | 2622 | 2614 | 6 | 0.002 | 69944 | 6949 | 6933 | 10 | 0.001 | |
| 288033 | 3 | 126219009 | 12 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 307995 | 2 | 61510631 | 61510631 | T | C | USP34 | intronic | c.4833-47 | e36-47 | tier3 | 3787 | 0 | 0 | 0 | 0.000 | 1349 | 111 | 94 | 17 | 0.153 | 2077 | 0 | 0 | 0 | NA | 4450 | 429 | 427 | 0 | 0.000 | 2281 | 0 | 0 | 0 | NA |
| 307995 | 2 | 138738953 | 138738953 | C | T | HNMT | silent | c.358 | p.L120 | tier1 | 22240 | 2688 | 2684 | 0 | 0.000 | 16074 | 1610 | 1361 | 234 | 0.147 | 21010 | 2141 | 2133 | 0 | 0.000 | 35264 | 4055 | 4048 | 0 | 0.000 | 49152 | 6483 | 6477 | 0 | 0.000 |
| 307995 | 2 | 207310122 | 207310122 | T | C | ADAM23 | silent | c.306 | p.S102 | tier1 | 16606 | 2066 | 2063 | 0 | 0.000 | 7808 | 803 | 660 | 137 | 0.172 | 14513 | 1574 | 1573 | 0 | 0.000 | 22705 | 2581 | 2575 | 0 | 0.000 | 25337 | 3372 | 3366 | 0 | 0.000 |
| 307995 | 3 | 9517257 | 9517257 | G | T | SETD5 | nonsense | c.3811 | p.G1271* | tier1 | 16932 | 2046 | 2042 | 1 | 0.000 | 12666 | 1192 | 986 | 197 | 0.167 | 16625 | 1771 | 1756 | 1 | 0.000 | 30581 | 3499 | 3493 | 0 | 0.000 | 40390 | 5238 | 5230 | 0 | 0.000 |
| 307995 | 3 | 30702951 | 30702951 | G | A | TGFBR2 | intronic | c.530-10179 | e5-10179 | tier4 | 21932 | 3088 | 3087 | 1 | 0.000 | 12719 | 1402 | 1136 | 255 | 0.183 | 19415 | 2486 | 2478 | 0 | 0.000 | 35761 | 4596 | 4593 | 1 | 0.000 | 44285 | 6232 | 6230 | 1 | 0.000 |
| 307995 | 3 | 126180517 | 126180517 | G | A | ZKDC | missense | c.1988 | p.P663L | tier1 | 26121 | 3489 | 3479 | 3 | 0.001 | 17143 | 1897 | 1582 | 303 | 0.161 | 19681 | 2031 | 2023 | 0 | 0.000 | 28774 | 2952 | 2950 | 1 | 0.000 | 46891 | 6709 | 6704 | 1 | 0.000 |
| 307995 | 4 | 107154095 | 107154095 | C | A | TBCK | missense | c.1639 | p.G547C | tier1 | 22774 | 2500 | 2497 | 2 | 0.001 | 12234 | 1077 | 886 | 186 | 0.174 | 17884 | 1895 | 1891 | 0 | 0.000 | 36704 | 3940 | 3938 | 0 | 0.000 | 38889 | 4788 | 4787 | 1 | 0.000 |
| 307995 | 5 | 5235176 | 5235176 | C | T | ADAMTS16 | missense | c.1900 | p.L634F | tier1 | 41498 | 5993 | 5982 | 1 | 0.000 | 24175 | 2732 | 2248 | 461 | 0.170 | 28275 | 3291 | 3286 | 0 | 0.000 | 52906 | 6393 | 6393 | 0 | 0.000 | 64254 | 9267 | 9257 | 0 | 0.000 |
| 307995 | 5 | 14364017 | 14364017 | C | A | TRIO | silent | c.2568 | p.V856 | tier1 | 11759 | 2486 | 2485 | 0 | 0.000 | 8545 | 1420 | 1183 | 229 | 0.162 | 7732 | 1311 | 1309 | 0 | 0.000 | 14980 | 2672 | 2669 | 1 | 0.000 | 20910 | 4722 | 4719 | 1 | 0.000 |
| 307995 | 5 | 140174742 | 140174742 | C | T | PCDHA2 | missense | c.193 | p.R65W | tier1 | 10300 | 1723 | 1722 | 1 | 0.001 | 5235 | 1925 | 1496 | 96 | 0.162 | 5095 | 646 | 645 | 0 | 0.000 | 12490 | 1585 | 1583 | 0 | 0.000 | 14042 | 2508 | 2506 | 0 | 0.000 |
| 307995 | 6 | 38743672 | 38743672 | G | A | DNAH8 | missense | c.1256 | p.R419K | tier1 | 12876 | 1538 | 1531 | 0 | 0.000 | 7777 | 745 | 677 | 64 | 0.086 | 11981 | 1279 | 1277 | 0 | 0.000 | 20259 | 2218 | 2218 | 0 | 0.000 | 25829 | 3434 | 3432 | 0 | 0.000 |
| 307995 | 7 | 29689532 | 29689532 | T | A | ZNRF2P2 | intronic | c.NULL | NULL | tier2 | 9555 | 1238 | 1236 | 0 | 0.000 | 3907 | 344 | 326 | 14 | 0.041 | 8708 | 1054 | 1053 | 0 | 0.000 | 15734 | 1964 | 1960 | 0 | 0.000 | 22140 | 2998 | 2995 | 0 | 0.000 |
| 307995 | 9 | 38414100 | 38414100 | C | T | IGFBP1 | silent | c.561 | p.T187 | tier1 | 850 | 0 | 0 | 0 | 0.000 | 1029 | 101 | 84 | 17 | 0.168 | 601 | 52 | 52 | 0 | 0.000 | 1054 | 73 | 73 | 0 | 0.000 | 2312 | 0 | 0 | 0 | NA |
| 307995 | 9 | 124737164 | 124737164 | T | C | TTL11 | missense | c.1634 | p.E545G | tier1 | 4530 | 842 | 841 | 0 | 0.000 | 3253 | 448 | 363 | 82 | 0.184 | 2211 | 0 | 0 | 0 | NA | 4800 | 565 | 561 | 0 | 0.000 | 6841 | 0 | 0 | 0 | NA |
| 307995 | 11 | 67049830 | 67049830 | C | G | ADRBK1 | missense | c.1046 | p.A349G | tier1 | 17607 | 2373 | 2372 | 1 | 0.000 | 9871 | 930 | 780 | 146 | 0.158 | 13509 | 1415 | 1412 | 1 | 0.000 | 24268 | 2672 | 2668 | 0 | 0.000 | 32239 | 4290 | 4286 | 0 | 0.000 |
| 307995 | 11 | 90016247 | 90016247 | G | A | ENSG00000214391 | 3_prime_untranslated_region | c.*90016247 | NULL | tier2 | 9251 | 1416 | 1410 | 1 | 0.001 | 5881 | 615 | 505 | 109 | 0.178 | 8316 | 1001 | 997 | 0 | 0.000 | 14878 | 1962 | 1962 | 0 | 0.000 | 19995 | 3123 | 3120 | 0 | 0.000 |
| 307995 | 12 | 9095076 | 9095076 | C | G | M6PR | missense | c.647 | p.G216A | tier1 | 18052 | 2269 | 2268 | 0 | 0.000 | 14745 | 1560 | 1265 | 284 | 0.183 | 14814 | 1463 | 1459 | 0 | 0.000 | 26480 | 2763 | 2759 | 0 | 0.000 | 36033 | 4801 | 4793 | 1 | 0.000 |
| 307995 | 12 | 50027356 | 50027356 | G | A | PRPF40B | intronic | c.580+26 | e8+26 | tier3 | 20184 | 2279 | 2278 | 0 | 0.000 | 17274 | 1679 | 1356 | 308 | 0.185 | 19082 | 1865 | 1862 | 0 | 0.000 | 38807 | 4116 | 4112 | 0 | 0.000 | 44347 | 5501 | 5499 | 0 | 0.000 |
| 307995 | 12 | 52715016 | 52715016 | G | T | KRT83 | missense | c.104 | p.A35D | tier1 | 17872 | 2232 | 2228 | 2 | 0.001 | 11270 | 1091 | 906 | 174 | 0.161 | 14742 | 1522 | 1520 | 0 | 0.000 | 24864 | 2656 | 2656 | 0 | 0.000 | 30211 | 4075 | 4072 | 1 | 0.000 |
| 307995 | 12 | 52891238 | 52891238 | T | A | KRT6C | 5_prime_flanking_region | c.-23717 | NULL | tier3 | 37827 | 4684 | 4677 | 4 | 0.001 | 17563 | 1842 | 1493 | 338 | 0.185 | 13579 | 4197 | 4197 | 0 | 0.000 | 59543 | 7510 | 7503 | 0 | 0.000 | 74155 | 9567 | 9558 | 0 | 0.000 |
| 307995 | 12 | 109519720 | 109519720 | C | T | USP30 | intronic | c.781-18 | e9-18 | tier3 | 18185 | 2137 | 2136 | 1 | 0.000 | 14507 | 1438 | 1220 | 204 | 0.143 | 21551 | 2264 | 2261 | 0 | 0.000 | 37214 | 4211 | 4209 | 0 | 0.000 | 51875 | 6669 | 6664 | 1 | 0.000 |
| 307995 | 14 | 94945375 | 94945375 | G | A | SERPINA9 | intronic | c.NULL | NULL | tier3 | 14412 | 2018 | 2013 | 0 | 0.000 | 8542 | 927 | 754 | 164 | 0.179 | 9099 | 884 | 881 | 0 | 0.000 | 12602 | 1228 | 1222 | 0 | 0.000 | 18950 | 2683 | 2678 | 0 | 0.000 |
| 307995 | 17 | 61986092 | 61986092 | G | C | GH2 | 5_prime_flanking_region | c.-26931 | NULL | tier3 | 7436 | 993 | 993 | 0 | 0.000 | 5504 | 643 | 532 | 107 | 0.167 | 4489 | 420 | 418 | 1 | 0.000 | 8630 | 980 | 978 | 0 | 0.000 | 11989 | 1814 | 1811 | 1 | 0.000 |
| 307995 | 18 | 50683690 | 50683690 | C | T | DCC | intronic | c.1262-36 | e8-36 | tier3 | 7199 | 1369 | 1368 | 1 | 0.001 | 7187 | 937 | 779 | 156 | 0.167 | 6715 | 805 | 802 | 1 | 0.000 | 10471 | 1351 | 1349 | 1 | 0.000 | 15612 | 2820 | 2817 | 1 | 0.000 |
| 307995 | 20 | 47247205 | 47247205 | C | T | PREX1 | intronic | c.4593+61 | e36+61 | tier3 | 53682 | 6200 | 6199 | 0 | 0.000 | 29856 | 2963 | 2421 | 515 | 0.175 | 41606 | 4321 | 4313 | 1 | 0.000 | 80057 | 9181 | 9177 | 0 | 0.000 | 100875 | 12640 | 12635 | 2 | 0.000 |
| 307995 | 20 | 56082765 | 56082765 | C | T | CTCFL | silent | c.1326 | p.P442 | tier1 | 46865 | 5735 | 5729 | 3 | 0.001 | 18320 | 1737 | 1393 | 333 | 0.193 | 36410 | 4191 | 4180 | 2 | 0.000 | 72397 | 9117 | 9111 | 0 | 0.000 | 87757 | 11408 | 11399 | 0 | 0.000 |
| 307995 | 20 | 58349240 | 58349240 | G | A | PHACTR3 | intronic | c.927-58 | e7-58 | tier3 | 9829 | 1423 | 1419 | 1 | 0.001 | 6694 | 832 | 688 | 143 | 0.172 | 7166 | 850 | 848 | 0 | 0.000 | 11139 | 1465 | 1464 | 0 | 0.000 | 15974 | 2886 | 2880 | 1 | 0.000 |
| 307995 | 22 | 20171118 | 20171118 | G | T | LINC00896 | 5_prime_flanking_region | c.-22982 | NULL | tier3 | 41685 | 5144 | 5137 | 2 | 0.000 | 27941 | 2833 | 2333 | 468 | 0.167 | 35577 | 3647 | 3638 | 0 | 0.000 | 64807 | 7402 | 7398 | 0 | 0.000 | 82450 | 10682 | 10662 | 1 | 0.000 |
| 307995 | X | 39911641 | 39911641 | C | T | BCOR | nonsense | c.4989 | p.W1663* | tier1 | 2771 | 0 | 0 | 0 | 0.000 | 1661 | 163 | 120 | 42 | 0.259 | 2252 | 209 | 209 | 0 | 0.000 | 3184 | 355 | 354 | 0 | 0.000 | 4228 | 0 | 0 | 0 | NA |
| 307995 | X | 53574723 | 53574723 | G | T | HUWE1 | missense | c.10547 | p.A3516D | tier1 | 4030 | 855 | 853 | 1 | 0.001 | 3570 | 485 | 322 | 160 | 0.332 | 2956 | 479 | 477 | 0 | 0.000 | 6543 | 1022 | 1021 | 0 | 0.000 | 8509 | 1948 | 1947 | 1 | 0.000 |
| 307995 | X | 151336800 | 151336800 | C | T | GABRA3 | missense | c.1379 | p.R460H | tier1 | 20311 | 2431 | 2425 | 4 | 0.002 | 16974 | 1748 | 1243 | 482 | 0.279 | 22921 | 2528 | 2526 | 0 | 0.000 | 43526 | 5186 | 5172 | 1 | 0.000 | 59634 | 7616 | 7603 | 1 | 0.000 |
| 311426 | 1 | 15987264 | 15987264 | T | C | RSC1A1 | silent | c.901 | p.L301 | tier1 | 11236 | 1410 | 1362 | 48 | 0.034 | 13278 | 1685 | 958 | 720 | 0.429 | NA | NA | NA | NA | NA | 17268 | 2601 | 2577 | 12 | 0.005 | 10555 | 751 | 693 | 54 | 0.072 |
| 311426 | 1 | 23660085 | 23660085 | T | C | HNRNP1 | missense | c.424 | p.T142A | tier1 | 34856 | 4681 | 4522 | 155 | 0.033 | 31282 | 4074 | 2318 | 1713 | 0.245 | NA | NA | NA | NA | NA | 40078 | 5587 | 5558 | 18 | 0.003 | 35251 | 2705 | 2676 | 23 | 0.009 |
| 311426 | 1 | 26795865 | 26795865 | A | G | DHDDS | 3_prime_untranslated_region | c.*243 | NULL | tier3 | 18900 | 2359 | 2222 | 133 | 0.056 | 37140 | 4742 | 2588 | 2089 | 0.447 | NA | NA | NA | NA | NA | 56774 | 7787 | 7730 | 37 | 0.005 | 42915 | 3349 | 2143 | 11 | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-------------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 346313 | 1 | 115258744 | 115258744 | C | T | <i>NRAS</i> | missense | c.38 | p.G13D | tier1 | 15135 | 2345 | 2345 | 0 | 0.000 | 22148 | 2769 | 1917 | 817 | 0.299 | 9508 | 75 | 44 | 31 | 0.413 | 12656 | 1666 | 1659 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 1 | 185815276 | 185815276 | G | A | <i>HMCN1</i> | intronic | c.339+48 | e2+48 | tier3 | 6596 | 904 | 899 | 5 | 0.006 | 9266 | 1023 | 626 | 388 | 0.383 | 6413 | 50 | 30 | 20 | 0.400 | 5756 | 719 | 712 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 1 | 237494338 | 237494338 | G | A | <i>RYR2</i> | intronic | c.270+52 | e4+52 | tier3 | 6976 | 0 | 0 | 0 | 0.000 | 10457 | 1484 | 1109 | 367 | 0.249 | 4945 | 59 | 39 | 20 | 0.339 | 9559 | 1403 | 1398 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 2 | 79914651 | 79914651 | C | T | <i>CTNNA2</i> | intronic | c.204+35867 | e2+35867 | tier3 | 8130 | 1132 | 1127 | 4 | 0.004 | 7887 | 997 | 599 | 392 | 0.396 | 9373 | 58 | 31 | 27 | 0.466 | 10541 | 1507 | 1504 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 2 | 95976762 | 95976762 | G | A | <i>KCNIP3</i> | 3_prime_untranslated_region | c.*95976762 | NULL | tier3 | 14797 | 1887 | 1879 | 8 | 0.004 | 12604 | 1599 | 937 | 647 | 0.408 | 8658 | 46 | 21 | 24 | 0.533 | 13523 | 1768 | 1765 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 2 | 97493997 | 97493997 | C | T | <i>CNNM3</i> | 3_prime_untranslated_region | c.*97493997 | NULL | tier3 | 17734 | 2331 | 2323 | 7 | 0.003 | 16375 | 1841 | 1134 | 695 | 0.380 | 16681 | 97 | 58 | 39 | 0.402 | 17673 | 2370 | 2365 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 2 | 103148926 | 103148926 | G | A | <i>SLC9A4</i> | missense | c.2176 | p.G726S | tier1 | 15907 | 2288 | 2279 | 9 | 0.004 | 18575 | 2517 | 1566 | 930 | 0.373 | 9661 | 60 | 38 | 22 | 0.367 | 17004 | 2467 | 2463 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 2 | 198267359 | 198267359 | C | A | <i>SF3B1</i> | missense | c.1998 | p.K666N | tier1 | 1971 | 484 | 484 | 0 | 0.000 | 3428 | 660 | 449 | 211 | 0.320 | 1140 | 27 | 20 | 7 | 0.259 | 1344 | 284 | 283 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 3 | 124180873 | 124180873 | A | G | <i>KALRN</i> | intronic | c.3915+70 | e24+70 | tier3 | 6616 | 1154 | 1153 | 0 | 0.000 | 8408 | 1184 | 818 | 352 | 0.301 | 3526 | 41 | 29 | 12 | 0.293 | 4038 | 543 | 541 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 3 | 157363624 | 157363624 | A | T | <i>C3orf55</i> | nonsense | c.151 | p.K51* | tier1 | 12912 | 2008 | 2005 | 3 | 0.001 | 12504 | 1663 | 998 | 656 | 0.397 | 11554 | 80 | 52 | 28 | 0.350 | 17018 | 2350 | 2344 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 3 | 185362401 | 185362401 | C | T | <i>IGF2BP2</i> | 3_prime_untranslated_region | c.*918 | NULL | tier3 | 3106 | 473 | 470 | 2 | 0.004 | 3119 | 384 | 193 | 188 | 0.493 | 2875 | 20 | 11 | 9 | 0.450 | 4110 | 620 | 615 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 4 | 47888007 | 47888007 | T | C | <i>NFXL1</i> | missense | c.1553 | p.Y518C | tier1 | 7936 | 1262 | 1261 | 1 | 0.001 | 10623 | 1323 | 793 | 524 | 0.398 | 9853 | 74 | 41 | 33 | 0.446 | 12932 | 1797 | 1792 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 5 | 180659955 | 180659955 | C | T | <i>TRIM41</i> | 3_prime_untranslated_region | c.*180659955 | NULL | tier3 | 6908 | 1005 | 1001 | 3 | 0.003 | 8436 | 948 | 681 | 263 | 0.279 | 2747 | 24 | 19 | 5 | 0.208 | 6031 | 819 | 818 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 7 | 141794384 | 141794384 | G | A | <i>MGAM</i> | intronic | c.4619-36 | e38-36 | tier3 | 3954 | 754 | 745 | 9 | 0.012 | 9432 | 1324 | 227 | 1086 | 0.827 | 8804 | 44 | 12 | 32 | 0.727 | 19245 | 2505 | 2495 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 8 | 39111964 | 39111964 | C | T | <i>ADAM32</i> | missense | c.1934 | p.S645L | tier1 | 2218 | 306 | 305 | 1 | 0.003 | 2951 | 311 | 201 | 108 | 0.350 | 1323 | 11 | 6 | 5 | 0.455 | 2929 | 348 | 345 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 8 | 56362027 | 56362027 | G | A | <i>XKR4</i> | intronic | c.1007-73813 | e3-73813 | tier2 | 10991 | 1801 | 1799 | 2 | 0.001 | 19862 | 2655 | 1622 | 1018 | 0.386 | 6570 | 45 | 32 | 13 | 0.289 | 10399 | 1572 | 1571 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 9 | 2058252 | 2058252 | A | T | <i>SMARCA2</i> | intronic | c.1348-39 | e7-39 | tier3 | 6687 | 1167 | 1166 | 1 | 0.001 | 12790 | 1563 | 924 | 626 | 0.404 | 3677 | 28 | 20 | 8 | 0.286 | 7690 | 1002 | 997 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 9 | 116770653 | 116770653 | C | T | <i>ZNF618</i> | missense | c.670 | p.R224W | tier1 | 23849 | 3026 | 3015 | 11 | 0.004 | 27911 | 3504 | 1840 | 1188 | 0.392 | 21495 | 115 | 70 | 45 | 0.391 | 29547 | 3875 | 3864 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 9 | 139848263 | 139848263 | G | C | <i>LCN12</i> | rna | NULL | NULL | tier1 | 4511 | 1130 | 1128 | 2 | 0.002 | 5237 | 1046 | 627 | 416 | 0.399 | 1844 | 25 | 20 | 5 | 0.200 | 1463 | 327 | 325 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 10 | 43612131 | 43612131 | C | T | <i>RET</i> | silent | c.2236 | p.L746 | tier1 | 11710 | 1924 | 1921 | 3 | 0.002 | 12939 | 1785 | 1093 | 678 | 0.383 | 7954 | 54 | 30 | 24 | 0.444 | 8090 | 1127 | 1127 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 10 | 71631922 | 71631922 | C | T | <i>COL13A1</i> | splice_region | c.365-10 | e3-10 | tier3 | 6454 | 0 | 0 | 0 | 0.000 | 5986 | 868 | 573 | 287 | 0.334 | 4679 | 34 | 23 | 11 | 0.324 | 2959 | 550 | 547 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 11 | 32413565 | 32413565 | C | T | <i>WT1</i> | missense | c.1385 | p.R462Q | tier1 | 1659 | 0 | 0 | 0 | 0.000 | 1890 | 218 | 131 | 86 | 0.396 | 1176 | 13 | 7 | 6 | 0.462 | 729 | 82 | 80 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 11 | 66330311 | 66330311 | C | T | <i>ACTN3</i> | silent | c.2562 | p.N854 | tier1 | 25840 | 3654 | 3646 | 4 | 0.001 | 26088 | 3426 | 2080 | 1320 | 0.388 | 15836 | 115 | 75 | 40 | 0.446 | 22332 | 3070 | 3063 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 11 | 74880778 | 74880778 | C | T | <i>SLCO2B1</i> | silent | c.750 | p.R250 | tier1 | 12979 | 2156 | 2152 | 4 | 0.002 | 17961 | 2483 | 1507 | 942 | 0.385 | 6260 | 44 | 25 | 19 | 0.432 | 9284 | 1391 | 1384 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 12 | 22606832 | 22606832 | G | C | <i>CZCD5</i> | missense | c.2869 | p.Q957E | tier1 | 10580 | 1420 | 1415 | 4 | 0.003 | 11566 | 1283 | 914 | 362 | 0.284 | 6766 | 56 | 43 | 13 | 0.232 | 17261 | 2289 | 2288 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 12 | 112926910 | 112926910 | G | C | <i>PTPN11</i> | missense | c.1530 | p.Q510H | tier1 | 18281 | 2384 | 2384 | 0 | 0.000 | 17722 | 2050 | 1967 | 67 | 0.033 | 8551 | 50 | 50 | 0 | 0.000 | 15428 | 2108 | 2098 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 12 | 133384599 | 133384599 | C | T | <i>GOLGA3</i> | silent | c.1056 | p.A352 | tier1 | 20252 | 2717 | 2714 | 3 | 0.001 | 18422 | 2142 | 1324 | 795 | 0.375 | 8874 | 61 | 39 | 22 | 0.361 | 14650 | 1946 | 1944 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 13 | 95363388 | 95363388 | G | A | <i>SOX21</i> | 3_prime_untranslated_region | c.*85 | NULL | tier2 | 10957 | 1679 | 1673 | 3 | 0.002 | 9798 | 1204 | 751 | 447 | 0.373 | 8922 | 64 | 35 | 29 | 0.453 | 11181 | 1486 | 1484 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 13 | 99030086 | 99030086 | C | T | <i>FARP1</i> | missense | c.410 | p.A137V | tier1 | 10284 | 1799 | 1794 | 5 | 0.003 | 13249 | 1897 | 1169 | 718 | 0.381 | 5654 | 44 | 28 | 16 | 0.364 | 13410 | 1963 | 1958 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 14 | 38274022 | 38274022 | A | T | <i>TTCC6</i> | splice_region | c.357+3 | e3+3 | tier3 | 11618 | 1556 | 1555 | 0 | 0.000 | 14057 | 1605 | 1126 | 467 | 0.293 | 10576 | 81 | 56 | 25 | 0.309 | 15067 | 1903 | 1899 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 14 | 70442537 | 70442537 | G | A | <i>SMOC1</i> | splice_region | c.478+6 | e4+6 | tier2 | 9708 | 1305 | 1303 | 1 | 0.001 | 11480 | 1164 | 833 | 324 | 0.280 | 10514 | 77 | 51 | 26 | 0.338 | 11093 | 1380 | 1375 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 14 | 101429460 | 101429460 | A | T | <i>SNORD114-7</i> | rna | NULL | NULL | tier1 | 1456 | 395 | 394 | 1 | 0.003 | 858 | 143 | 82 | 61 | 0.427 | 453 | 9 | 4 | 5 | NA | 714 | 139 | 138 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 16 | 790001 | 790001 | G | A | <i>NARFL</i> | 5_prime_untranslated_region | c.-3603 | NULL | tier3 | 23157 | 3246 | 3238 | 3 | 0.001 | 15854 | 1949 | 1462 | 460 | 0.239 | 4948 | 30 | 28 | 2 | 0.000 | 16881 | 2183 | 2175 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 19 | 35991219 | 35991219 | T | C | <i>DMKN</i> | 5_prime_untranslated_region | c.-35991219 | NULL | tier3 | 7531 | 1700 | 1694 | 3 | 0.002 | 7260 | 1150 | 677 | 463 | 0.406 | 2283 | 31 | 22 | 9 | 0.290 | 1591 | 251 | 248 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 346313 | 19 | 38782452 | 38782452 | C | T | <i>SPINT2</i> | 3_prime_untranslated_region | c.*38782452 | NULL | tier3 | 11953 | 1849 | 1845 | 4 | 0.002 | 16199 | 2163 | 1360 | 788 | 0.367 | 8989 | 44 | 31 | 13 | 0.295 | 12445 | 1704 | 1698 | 2 | 0.000 | NA | NA | NA | NA | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 377158 | 7 | 44268486 | 44268486 | C | T | CAMK2B | silent | c.1377 | p.R459 | tier1 | 31191 | 3768 | 3745 | 19 | 0.005 | 19872 | 2875 | 1857 | 996 | 0.349 | 22934 | 2598 | 1604 | 971 | 0.377 | 24679 | 2025 | 2016 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 8 | 16166004 | 16166004 | C | T | DLGAP2 | missense | c.1730 | p.T577M | tier1 | 15778 | 2105 | 2028 | 76 | 0.036 | 9926 | 1438 | 870 | 567 | 0.395 | 12449 | 1643 | 894 | 744 | 0.454 | 10886 | 1208 | 1207 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 8 | 93029456 | 93029456 | G | A | RUNX1T1 | missense | c.257 | p.T86M | tier1 | 27247 | 3518 | 3500 | 16 | 0.005 | 13337 | 1840 | 1085 | 749 | 0.408 | 16786 | 2071 | 1165 | 889 | 0.433 | 15475 | 1318 | 1317 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 10 | 125521468 | 125521468 | C | A | CPXM2 | missense | c.1697 | p.R566L | tier1 | 33234 | 4104 | 4081 | 17 | 0.004 | 16084 | 2218 | 1289 | 921 | 0.417 | 19750 | 2413 | 1228 | 1167 | 0.487 | 17617 | 1314 | 1313 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 11 | 132177495 | 132177495 | A | G | NTM | intronic | c.527-88 | e5-88 | tier3 | 28441 | 4036 | 4028 | 6 | 0.001 | 11035 | 1798 | 1189 | 599 | 0.335 | 12341 | 1711 | 1086 | 615 | 0.362 | 15135 | 1756 | 1754 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 12 | 53439709 | 53439709 | C | T | LOC283335 | intronic | c.NULL | NULL | tier3 | 7825 | 1595 | 1585 | 8 | 0.005 | 6157 | 1216 | 747 | 462 | 0.382 | 5787 | 1148 | 636 | 510 | 0.445 | 6550 | 1008 | 1004 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 14 | 20216351 | 20216351 | C | T | ORAQ3 | silent | c.765 | p.C255 | tier1 | 62824 | 8606 | 8581 | 21 | 0.002 | 35001 | 5096 | 3330 | 1744 | 0.344 | 42145 | 5873 | 3768 | 2041 | 0.351 | 37687 | 2135 | 2134 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 14 | 38296537 | 38296537 | G | A | TTC6 | missense | c.1457 | p.G486E | tier1 | 11863 | 1418 | 1416 | 2 | 0.001 | 5038 | 661 | 514 | 146 | 0.221 | 7275 | 826 | 635 | 190 | 0.230 | 9058 | 682 | 678 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 14 | 94750258 | 94750258 | A | T | SERPINA10 | 3_prime_untranslated_region | c.*44 | NULL | tier3 | 8996 | 1348 | 1344 | 4 | 0.003 | 4692 | 846 | 620 | 225 | 0.266 | 5827 | 917 | 616 | 298 | 0.326 | 7137 | 775 | 770 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 16 | 67242195 | 67242195 | C | T | LRRC29 | intronic | c.285+20 | e2+20 | tier3 | 8952 | 1373 | 1365 | 7 | 0.005 | 16228 | 2461 | 1502 | 941 | 0.385 | 17279 | 2332 | 1324 | 1389 | 0.427 | 13870 | 1272 | 1270 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 17 | 37901877 | 37901877 | C | A | GRB7 | intronic | c.1278+86 | e11+86 | tier3 | 16607 | 2191 | 2180 | 8 | 0.004 | 9627 | 1320 | 807 | 509 | 0.387 | 10492 | 1311 | 763 | 534 | 0.412 | 8051 | 460 | 460 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 17 | 48653205 | 48653205 | G | A | CACNA1G | missense | c.1442 | p.S481N | tier1 | 9770 | 1740 | 1735 | 5 | 0.003 | 11808 | 1725 | 1037 | 683 | 0.397 | 12564 | 1917 | 1026 | 876 | 0.461 | 9331 | 842 | 842 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 19 | 12221114 | 12221114 | T | C | ENSG00000188474 | splice_region | c.1-6 | e1-6 | tier3 | 7396 | 1332 | 1325 | 5 | 0.004 | 9178 | 1894 | 1189 | 699 | 0.370 | 9686 | 1822 | 1117 | 694 | 0.383 | 8348 | 1197 | 1195 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 19 | 16008165 | 16008165 | G | A | CYP4F2 | intronic | c.198+59 | e1+59 | tier3 | 3096 | 291 | 279 | 12 | 0.041 | 2033 | 290 | 229 | 57 | 0.199 | 2418 | 217 | 192 | 24 | 0.111 | 3932 | 493 | 490 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 19 | 33792822 | 33792822 | C | A | CEBPA | nonsense | c.499 | p.E167* | tier1 | 13866 | 1815 | 1811 | 4 | 0.002 | 7305 | 1070 | 998 | 71 | 0.066 | 8461 | 1146 | 1041 | 103 | 0.090 | 9772 | 666 | 665 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 20 | 33573852 | 33573852 | T | C | MYH7B | intronic | c.932-51 | e12-51 | tier3 | 21988 | 3958 | 3933 | 22 | 0.006 | 21653 | 3676 | 2260 | 1395 | 0.382 | 25678 | 3746 | 2188 | 1507 | 0.408 | 19145 | 1491 | 1489 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 21 | 36231773 | 36231773 | C | T | RUNX1 | missense | c.611 | p.R204Q | tier1 | 40567 | 5888 | 5865 | 20 | 0.003 | 21930 | 3339 | 2312 | 1004 | 0.303 | 24949 | 3385 | 2261 | 1093 | 0.326 | 25246 | 2469 | 2464 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 21 | 39672197 | 39672197 | T | C | KCNK15 | silent | c.1014 | p.D338 | tier1 | 11992 | 1306 | 1297 | 6 | 0.005 | 8572 | 1316 | 797 | 513 | 0.392 | 9267 | 1048 | 623 | 419 | 0.402 | 11157 | 882 | 881 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 21 | 44514777 | 44514777 | T | G | UZAF1 | missense | c.470 | p.Q157P | tier1 | 82039 | 9989 | 9944 | 40 | 0.004 | 49155 | 6122 | 3547 | 2538 | 0.417 | 55897 | 6555 | 3561 | 2916 | 0.450 | 43879 | 2129 | 2126 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 21 | 44838927 | 44838927 | G | A | SIK1 | missense | c.1436 | p.T479I | tier1 | 26814 | 3642 | 3635 | 6 | 0.002 | 19946 | 3328 | 1988 | 1334 | 0.402 | 22098 | 3416 | 1998 | 1389 | 0.410 | 21113 | 1921 | 1919 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | 22 | 40425476 | 40425476 | G | A | FAM83F | intronic | c.1454-44 | e5-44 | tier3 | 34812 | 4648 | 4507 | 131 | 0.028 | 26490 | 3367 | 1999 | 1351 | 0.403 | 30682 | 4106 | 2159 | 1897 | 0.468 | 26129 | 1479 | 1476 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | X | 54948751 | 54948751 | G | A | TRO | 5_prime_untranslated_region | c.-347 | NULL | tier3 | 37994 | 5332 | 5310 | 13 | 0.002 | 19061 | 2629 | 1655 | 954 | 0.366 | 21036 | 2797 | 1648 | 1114 | 0.403 | 9334 | 684 | 681 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | X | 62974526 | 62974526 | G | A | ARHGEF9 | rna | NULL | NULL | tier1 | 43937 | 6008 | 5985 | 20 | 0.003 | 21090 | 2911 | 1755 | 1143 | 0.394 | 25009 | 3235 | 1844 | 1370 | 0.426 | 10902 | 688 | 688 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 377158 | X | 152826317 | 152826317 | C | A | ATP2B3 | missense | c.3023 | p.T1008N | tier1 | 92654 | 12135 | 12117 | 9 | 0.001 | 46732 | 5759 | 4027 | 1702 | 0.297 | 50740 | 6400 | 4392 | 1959 | 0.308 | 23593 | 942 | 942 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 378511 | 1 | 38185727 | 38185727 | C | T | EPHA10 | missense | c.2416 | p.G806S | tier1 | 21546 | 2925 | 2920 | 1 | 0.000 | 18962 | 1928 | 1164 | 758 | 0.394 | NA | NA | NA | NA | NA | 30926 | 3276 | 3272 | 0 | 0.000 | 23367 | 3297 | 3288 | 3 | 0.000 |
| 378511 | 1 | 103471831 | 103471831 | T | A | COL11A1 | missense | c.1760 | p.K587I | tier1 | 1429 | 0 | 0 | 0 | 0.000 | 1611 | 288 | 222 | 66 | 0.229 | NA | NA | NA | NA | NA | 2658 | 183 | 183 | 0 | 0.000 | 1680 | 217 | 216 | 0 | 0.000 |
| 378511 | 1 | 159505705 | 159505705 | G | C | OR10J5 | missense | c.93 | p.F31L | tier1 | 3661 | 659 | 658 | 0 | 0.000 | 13904 | 1999 | 1476 | 511 | 0.257 | NA | NA | NA | NA | NA | 15527 | 1741 | 1739 | 0 | 0.000 | 15301 | 2267 | 2260 | 1 | 0.000 |
| 378511 | 1 | 215990362 | 215990362 | T | C | USH2A | missense | c.9547 | p.I3183V | tier1 | 14189 | 1685 | 1684 | 1 | 0.001 | 16726 | 1755 | 889 | 862 | 0.492 | NA | NA | NA | NA | NA | 25758 | 2729 | 2728 | 0 | 0.000 | 22257 | 2928 | 2913 | 1 | 0.000 |
| 378511 | 1 | 218458652 | 218458652 | C | T | RRP15 | 5_prime_untranslated_region | c.-7 | NULL | tier2 | 12418 | 1616 | 1615 | 0 | 0.000 | 13735 | 1749 | 1315 | 420 | 0.242 | NA | NA | NA | NA | NA | 16938 | 1787 | 1783 | 1 | 0.000 | 16115 | 2104 | 2103 | 1 | 0.000 |
| 378511 | 2 | 10797867 | 10797867 | C | T | NOL10 | splice_site | c.906+1 | e11+1 | tier1 | 806 | 0 | 0 | 0 | 0.000 | 1567 | 294 | 199 | 94 | 0.321 | NA | NA | NA | NA | NA | 960 | 117 | 116 | 1 | 0.000 | 837 | 0 | 0 | 0 | 0.000 |
| 378511 | 2 | 89246763 | 89246763 | C | A | IGKV7-3 | 5_prime_flanking_region | c.-31869 | NULL | tier3 | 850 | 178 | 177 | 0 | 0.000 | 1668 | 263 | 165 | 98 | 0.373 | NA | NA | NA | NA | NA | 1504 | 199 | 199 | 0 | 0.000 | 1151 | 237 | 237 | 0 | 0.000 |
| 378511 | 2 | 127805801 | 127805801 | G | A | BIN1 | 5_prime_untranslated_region | c.-127805801 | NULL | tier3 | 10081 | 1171 | 1169 | 0 | 0.000 | 6982 | 753 | 456 | 295 | 0.393 | NA | NA | NA | NA | NA | 14152 | 1420 | 1420 | 0 | 0.000 | 11943 | 1540 | 1538 | 1 | 0.000 |
| 378511 | 2 | 191366422 | 191366422 | T | A | MFSD6 | 3_prime_untranslated_region | c.*1478 | NULL | tier3 | 11321 | 1652 | 1650 | 1 | 0.001 | 10329 | 1372 | 935 | 430 | 0.315 | NA | NA | NA | NA | NA | 17494 | 1967 | 1967 | 0 | 0.000 | 19206 | 2853 | 2838 | 0 | 0.000 |
| 378511 | 4 | 70826715 | 70826715 | G | T | CSN2 | 5_prime_untranslated_region | c.-2 | NULL | tier3 | 7585 | 1007 | 1004 | 0 | 0.000 | 4801 | 516 | 339 | 176 | 0.342 | NA | NA | NA | NA | NA | 9615 | 1042 | 1042 | 0 | 0.000 | 7253 | 931 | 926 | 3 | 0.000 |
| 378511 | 4 | 85562062 | 85562062 | C | T | CDS1 | silent | c.951 | p.F317 | tier1 | 41450 | 542 | 540 | 1 | 0.002 | 5099 | 804 | 499 | 303 | 0.378 | NA | NA | NA | NA | NA | 5142 | 466 | 464 | 0 | 0.000 | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 395344 | 8 | 124383531 | 124383531 | C | T | ATAD2 | missense | c.584 | p.R195H | tier1 | 14629 | 1830 | 1818 | 11 | 0.006 | 14848 | 1841 | 1287 | 550 | 0.299 | NA | NA | NA | NA | NA | 20090 | 1662 | 1632 | 24 | 0.014 | 11134 | 1240 | 1225 | 10 | 0.008 |
| 395344 | 11 | 3041385 | 3041385 | C | T | CARS | intronic | c.1266+65 | e11+65 | tier3 | 7112 | 1194 | 1183 | 11 | 0.009 | 9548 | 1111 | 665 | 436 | 0.396 | NA | NA | NA | NA | NA | 10051 | 908 | 902 | 5 | 0.006 | 6338 | 725 | 720 | 4 | 0.006 |
| 395344 | 11 | 103043939 | 103043939 | C | A | DYNC2H1 | missense | c.5463 | p.D1821E | tier1 | 30737 | 3704 | 3676 | 23 | 0.006 | 27370 | 3236 | 1869 | 1333 | 0.416 | NA | NA | NA | NA | NA | 50846 | 4421 | 4370 | 50 | 0.011 | 25479 | 3026 | 3002 | 19 | 0.006 |
| 395344 | 12 | 42866304 | 42866304 | C | T | PRICKLE1 | missense | c.15 | p.M5I | tier1 | 42612 | 5402 | 5337 | 61 | 0.011 | 27001 | 3358 | 2428 | 902 | 0.271 | NA | NA | NA | NA | NA | 55646 | 4485 | 4462 | 17 | 0.004 | 29418 | 3245 | 3236 | 9 | 0.003 |
| 395344 | 13 | 95095724 | 95095724 | G | A | DC7 | silent | c.1446 | p.D482 | tier1 | 37506 | 5536 | 5386 | 143 | 0.026 | 20964 | 3033 | 1890 | 1115 | 0.371 | NA | NA | NA | NA | NA | 27709 | 2945 | 2915 | 22 | 0.007 | 17198 | 2402 | 2383 | 14 | 0.006 |
| 395344 | 16 | 32914951 | 32914951 | C | A | IGHV3OR16-15 | 3_prime_untranslated_region | c.*32914951 | NULL | tier3 | 16195 | 1983 | 1979 | 0 | 0.000 | 11281 | 1319 | 967 | 343 | 0.262 | NA | NA | NA | NA | NA | 25985 | 2350 | 2340 | 6 | 0.003 | 12523 | 1498 | 1488 | 1 | 0.000 |
| 395344 | 19 | 36216047 | 36216047 | T | C | KMT2B | intronic | c.3528+59 | e10+59 | tier3 | 27609 | 3427 | 3388 | 36 | 0.011 | 18206 | 1968 | 1122 | 817 | 0.421 | NA | NA | NA | NA | NA | 21799 | 2020 | 2002 | 15 | 0.007 | 14284 | 1689 | 1669 | 16 | 0.009 |
| 395344 | 21 | 36252865 | 36252865 | C | T | RUNX1 | missense | c.497 | p.R166Q | tier1 | 26896 | 3450 | 3425 | 23 | 0.007 | 17808 | 2122 | 1297 | 806 | 0.383 | NA | NA | NA | NA | NA | 19449 | 2241 | 2214 | 23 | 0.010 | 14055 | 1978 | 1949 | 20 | 0.010 |
| 395344 | 21 | 44524456 | 44524456 | G | A | U2AF1 | missense | c.101 | p.S34F | tier1 | 12143 | 1825 | 1800 | 25 | 0.014 | 7376 | 961 | 651 | 308 | 0.321 | NA | NA | NA | NA | NA | 17805 | 2119 | 2109 | 10 | 0.005 | 9216 | 1398 | 1390 | 7 | 0.005 |
| 395344 | 22 | 19452777 | 19452777 | T | A | UFD1L | missense | c.443 | p.N148I | tier1 | 50171 | 6290 | 6212 | 63 | 0.010 | 39442 | 5176 | 3069 | 2030 | 0.398 | NA | NA | NA | NA | NA | 58929 | 4568 | 4513 | 39 | 0.009 | 31360 | 3453 | 3388 | 29 | 0.008 |
| 429745 | 1 | 1078202 | 1078202 | G | A | LOC254099 | rna | NULL | NULL | tier1 | 5931 | 911 | 907 | 4 | 0.004 | 4908 | 690 | 421 | 263 | 0.385 | 2088 | 234 | 151 | 81 | 0.349 | 7726 | 722 | 622 | 93 | 0.130 | 13784 | 2100 | 1849 | 240 | 0.115 |
| 429745 | 1 | 78189035 | 78189035 | T | C | USP33 | missense | c.1463 | p.Q488R | tier1 | 2431 | 445 | 441 | 4 | 0.009 | 2906 | 416 | 253 | 163 | 0.392 | 795 | 123 | 92 | 31 | 0.252 | 2851 | 277 | 248 | 29 | 0.105 | 5122 | 835 | 734 | 99 | 0.119 |
| 429745 | 1 | 103400670 | 103400670 | C | T | COL11A1 | splice_site | c.3475-1 | e45-1 | tier1 | 154 | 0 | 0 | 0 | 0.000 | 334 | 49 | 43 | 6 | 0.122 | 79 | 6 | 4 | 2 | NA | 345 | 0 | 0 | NA | 643 | 106 | 106 | 0 | 0.000 | |
| 429745 | 1 | 193018974 | 193018974 | G | T | UCHL5 | missense | c.148 | p.H50N | tier1 | 635 | 102 | 102 | 0 | 0.000 | 697 | 103 | 52 | 51 | 0.495 | 265 | 24 | 17 | 7 | 0.292 | 758 | 55 | 48 | 7 | 0.127 | 1191 | 186 | 161 | 25 | 0.134 |
| 429745 | 1 | 197021696 | 197021696 | T | A | F13B | intronic | c.1555+68 | e9+68 | tier3 | 2218 | 180 | 180 | 0 | 0.000 | 1632 | 136 | 76 | 60 | 0.441 | 1399 | 89 | 68 | 20 | 0.227 | 3010 | 151 | 137 | 13 | 0.087 | 4203 | 326 | 291 | 35 | 0.107 |
| 429745 | 1 | 240071866 | 240071866 | C | T | CHRM3 | missense | c.1115 | p.P372L | tier1 | 18828 | 3031 | 3005 | 21 | 0.007 | 12958 | 1506 | 905 | 580 | 0.391 | 11480 | 1361 | 966 | 389 | 0.287 | 30654 | 3547 | 3130 | 382 | 0.109 | 48285 | 6995 | 6216 | 749 | 0.108 |
| 429745 | 2 | 234231616 | 234231616 | G | A | SAG | missense | c.400 | p.V134M | tier1 | 366 | 64 | 63 | 1 | 0.016 | 775 | 112 | 74 | 38 | 0.309 | 253 | 32 | 18 | 14 | 0.438 | 914 | 71 | 68 | 3 | 0.042 | 1357 | 190 | 171 | 19 | 0.100 |
| 429745 | 3 | 38010850 | 38010850 | G | T | CTDSPL | intronic | c.426+1477 | e5+1477 | tier3 | 18584 | 2730 | 2701 | 28 | 0.010 | 14632 | 1646 | 959 | 660 | 0.438 | 8380 | 807 | 522 | 282 | 0.351 | 23175 | 2227 | 1951 | 249 | 0.014 | 38870 | 5267 | 4584 | 644 | 0.123 |
| 429745 | 3 | 197807179 | 197807179 | C | T | ANKRD18BP | splice_region | c.NULL | NULL | tier2 | 5395 | 618 | 617 | 0 | 0.000 | 2321 | 200 | 143 | 54 | 0.274 | 2081 | 182 | 158 | 24 | 0.132 | 6352 | 550 | 520 | 27 | 0.049 | 12351 | 1431 | 1356 | 73 | 0.051 |
| 429745 | 5 | 126667153 | 126667153 | G | A | MEGF10 | intronic | c.116+37 | e1+37 | tier3 | 1043 | 100 | 99 | 1 | 0.010 | 1329 | 126 | 61 | 63 | 0.508 | 475 | 37 | 26 | 11 | 0.297 | 1280 | 82 | 67 | 15 | 0.083 | 2260 | 222 | 189 | 33 | 0.149 |
| 429745 | 5 | 134705108 | 134705108 | G | C | H2AFY | silent | c.465 | p.A155 | tier1 | 5079 | 644 | 639 | 5 | 0.008 | 5428 | 589 | 360 | 226 | 0.386 | 2986 | 271 | 160 | 110 | 0.407 | 8240 | 703 | 598 | 99 | 0.142 | 12226 | 1587 | 1360 | 223 | 0.141 |
| 429745 | 5 | 139222426 | 139222426 | G | A | PSD2 | 3_prime_untranslated_region | c.*367 | NULL | tier3 | 4475 | 724 | 717 | 5 | 0.007 | 3764 | 458 | 266 | 187 | 0.413 | 1861 | 187 | 122 | 64 | 0.344 | 5627 | 555 | 487 | 67 | 0.121 | 9745 | 1340 | 1177 | 155 | 0.116 |
| 429745 | 5 | 176008570 | 176008570 | C | T | CDHR2 | missense | c.2045 | p.T682I | tier1 | 33969 | 4456 | 4449 | 6 | 0.001 | 20616 | 2221 | 1786 | 411 | 0.187 | 19249 | 2116 | 1955 | 152 | 0.072 | 48165 | 5253 | 5006 | 221 | 0.042 | 74751 | 9832 | 9357 | 447 | 0.046 |
| 429745 | 5 | 176709402 | 176709402 | A | G | NSD1 | intronic | c.5893-64 | e18-64 | tier3 | 91 | 0 | 0 | 0 | 0.000 | 262 | 39 | 25 | 13 | 0.342 | 44 | 4 | 3 | 1 | NA | 257 | 18 | 15 | 3 | 0.167 | 648 | 94 | 89 | 5 | 0.053 |
| 429745 | 6 | 27278373 | 27278373 | T | C | POM121L2 | missense | c.1577 | p.D526G | tier1 | 13531 | 2189 | 2166 | 20 | 0.009 | 10136 | 1360 | 783 | 565 | 0.419 | 5544 | 677 | 493 | 176 | 0.263 | 15079 | 1771 | 1562 | 191 | 0.109 | 28268 | 4382 | 3844 | 516 | 0.118 |
| 429745 | 7 | 53854870 | 53854870 | C | T | ENSG00000232418 | 3_prime_untranslated_region | c.*53854870 | NULL | tier2 | 28223 | 3696 | 3663 | 32 | 0.009 | 16551 | 1547 | 869 | 669 | 0.435 | 21358 | 2326 | 1589 | 720 | 0.312 | 32748 | 3504 | 3109 | 367 | 0.106 | 53630 | 6754 | 5954 | 768 | 0.114 |
| 429745 | 10 | 11376350 | 11376350 | T | G | CELF2 | 3_prime_untranslated_region | c.*5334 | NULL | tier3 | 7924 | 1006 | 997 | 8 | 0.008 | 9395 | 951 | 590 | 350 | 0.372 | 4952 | 443 | 266 | 174 | 0.395 | 11674 | 1004 | 875 | 124 | 0.124 | 22812 | 2631 | 2291 | 331 | 0.126 |
| 429745 | 10 | 35300792 | 35300792 | T | C | CUL2 | missense | c.12147 | p.N716S | tier1 | 12515 | 1709 | 1701 | 8 | 0.005 | 7221 | 506 | 360 | 416 | 0.578 | 882 | 612 | 266 | 303 | 0.354 | 15443 | 1835 | 1662 | 164 | 0.090 | 22763 | 3133 | 2791 | 325 | 0.104 |
| 429745 | 11 | 62302518 | 62302518 | C | T | AHNAK | nonsense | c.299 | p.W100* | tier1 | 40270 | 5304 | 5267 | 33 | 0.006 | 32662 | 3139 | 1825 | 1256 | 0.408 | 27065 | 2795 | 1838 | 928 | 0.336 | 63305 | 6506 | 5702 | 719 | 0.112 | 103605 | 12896 | 11222 | 1538 | 0.121 |
| 429745 | 11 | 67020214 | 67020214 | G | A | KDM2A | missense | c.2818 | p.A940T | tier1 | 4708 | 612 | 604 | 6 | 0.010 | 4879 | 579 | 347 | 223 | 0.391 | 2010 | 203 | 123 | 78 | 0.388 | 5587 | 502 | 445 | 54 | 0.108 | 10662 | 1403 | 1212 | 181 | 0.130 |
| 429745 | 11 | 67565182 | 67565182 | C | T | FAM86C2P | 5_prime_untranslated_region | c.-67565182 | NULL | tier3 | 5336 | 901 | 891 | 10 | 0.011 | 4344 | 544 | 333 | 209 | 0.386 | 3552 | 451 | 355 | 95 | 0.211 | 11610 | 1395 | 1295 | 97 | 0.070 | 20829 | 3034 | 2825 | 201 | 0.066 |
| 429745 | 12 | 102074329 | 102074329 | G | A | MYBPC1 | intronic | c.3433+22 | e29+22 | tier3 | 8975 | 1137 | 1127 | 10 | 0.009 | 5589 | 529 | 322 | 204 | 0.388 | 6302 | 560 | 413 | 145 | 0.260 | 10666 | 939 | 834 | 102 | 0.109 | 17843 | 2176 | 1926 | 245 | 0.113 |
| 429745 | 12 | 109569189 | 109569189 | G | T | ACACB | 5_prime_untranslated_region | c.-8097 | NULL | tier3 | 30163 | 4131 | 4094 | 36 | 0.009 | 17652 | 1755 | 1153 | 585 | 0.337 | 20162 | 2093 | 1634 | 444 | 0.214 | 41852 | 4379 | 4009 | 331 | 0.076 | 65401 | 8618 | 7781 | 798 | 0.093 |
| 429745 | 14 | 31405745 | 31405745 | C | A | STRN3 | missense | c.802 | p.G268C | tier1 | 5682 | 743 | 736 | 7 | 0.009 | 3901 | 377 | 200 | 176 | 0.468 | 3361 | 300 | 192 | 107 | 0.358 | 6926 | 594 | 518 | 72 | 0.122 | 11384 | 1425 | 1212 | 211 | 0.148 |
| 429745 | 14 | 64921502 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 435866 | 7 | 151882645 | 151882645 | C | A | MLL3 | missense | c.5080 | p.V1694L | tier1 | 18341 | 3179 | 3137 | 41 | 0.013 | 8337 | 1660 | 1049 | 606 | 0.366 | 19306 | 2029 | 1995 | 34 | 0.017 | 105404 | 6095 | 6092 | 0 | 0.000 | 100372 | 6151 | 6136 | 11 | 0.002 |
| 435866 | 8 | 68024182 | 68024182 | G | A | CSPPI1 | intronic | c.1226-25 | e12-25 | tier3 | 28617 | 2605 | 2573 | 32 | 0.012 | 14377 | 1643 | 1008 | 631 | 0.385 | 24083 | 1401 | 1369 | 31 | 0.022 | 163255 | 4606 | 4606 | 0 | 0.000 | 128108 | 3781 | 3773 | 8 | 0.002 |
| 435866 | 8 | 139727925 | 139727925 | C | A | COL22A1 | missense | c.2517 | p.K839N | tier1 | 14770 | 1481 | 1456 | 25 | 0.017 | 6594 | 812 | 465 | 344 | 0.425 | 13358 | 871 | 843 | 28 | 0.032 | 74830 | 2249 | 2249 | 0 | 0.000 | 47347 | 2114 | 2110 | 4 | 0.002 |
| 435866 | 10 | 81921715 | 81921715 | G | T | ANXA11 | silent | c.1156 | p.R386 | tier1 | 28956 | 2256 | 2244 | 12 | 0.005 | 27498 | 2104 | 1088 | 1007 | 0.481 | 32356 | 1496 | 1486 | 10 | 0.007 | 230558 | 5119 | 5117 | 1 | 0.000 | 262348 | 5468 | 5448 | 18 | 0.003 |
| 435866 | 10 | 120810716 | 120810716 | A | G | EIF3A | missense | c.2314 | p.Y772H | tier1 | 15834 | 2265 | 2253 | 10 | 0.004 | 8732 | 1502 | 893 | 604 | 0.403 | 16716 | 1341 | 1328 | 13 | 0.010 | 89665 | 3829 | 3829 | 0 | 0.000 | 82069 | 3736 | 3729 | 6 | 0.002 |
| 435866 | 15 | 28420657 | 28420657 | C | T | HERC2 | splice_site | c.9831+1 | e63+1 | tier1 | 9040 | 828 | 812 | 16 | 0.019 | 5494 | 551 | 305 | 245 | 0.445 | 8678 | 438 | 423 | 15 | 0.034 | 32185 | 1144 | 1144 | 0 | 0.000 | 52632 | 1429 | 1425 | 4 | 0.003 |
| 435866 | 16 | 3341019 | 3341019 | C | T | ZNF263 | 3_prime_untranslated_region | c.*461 | NULL | tier2 | 41409 | 2681 | 2649 | 32 | 0.012 | 27111 | 1794 | 1274 | 516 | 0.288 | 37079 | 1501 | 1476 | 25 | 0.017 | 310483 | 6336 | 6332 | 4 | 0.000 | 258758 | 5137 | 5126 | 10 | 0.002 |
| 435866 | 16 | 87870242 | 87870242 | C | T | SLC7A5 | silent | c.1152 | p.T384 | tier1 | 51148 | 3783 | 3763 | 17 | 0.004 | 34652 | 3116 | 1778 | 1330 | 0.428 | 44181 | 1996 | 1972 | 22 | 0.011 | 277199 | 6340 | 6339 | 1 | 0.000 | 345963 | 7896 | 7869 | 24 | 0.003 |
| 435866 | 17 | 40049260 | 40049260 | A | G | ACLY | intronic | c.1763+26 | e15+26 | tier2 | 19218 | 1932 | 1908 | 23 | 0.012 | 12423 | 1333 | 925 | 404 | 0.304 | 19972 | 1270 | 1245 | 23 | 0.018 | 129367 | 3722 | 3722 | 0 | 0.000 | 150179 | 3843 | 3842 | 1 | 0.000 |
| 435866 | 17 | 74732959 | 74732959 | G | T | SRSF2 | missense | c.284 | p.P95H | tier1 | 17829 | 1296 | 1268 | 26 | 0.020 | 18244 | 1580 | 873 | 702 | 0.446 | 23322 | 1205 | 1184 | 21 | 0.017 | 114677 | 3195 | 3193 | 2 | 0.000 | 168820 | 4035 | 4026 | 9 | 0.002 |
| 435866 | 18 | 61226995 | 61226995 | C | G | SERPINF12 | intronic | c.384+44 | e3+44 | tier2 | 18158 | 2791 | 2763 | 27 | 0.010 | 8346 | 1647 | 998 | 647 | 0.393 | 17288 | 1453 | 1426 | 27 | 0.019 | 76142 | 3486 | 3485 | 1 | 0.000 | 88700 | 4323 | 4312 | 9 | 0.002 |
| 435866 | 20 | 14830634 | 14830634 | A | G | MACROD2 | 3_prime_untranslated_region | c.*14830634 | NULL | tier3 | 4060 | 789 | 786 | 3 | 0.004 | 1600 | 356 | 194 | 160 | 0.452 | 3377 | 364 | 362 | 2 | 0.000 | 16068 | 924 | 922 | 0 | 0.000 | 12242 | 801 | 799 | 2 | 0.000 |
| 435866 | 22 | 38884063 | 38884063 | T | C | DDX17 | missense | c.1505 | p.Y502C | tier1 | 47465 | 4127 | 4050 | 74 | 0.018 | 31334 | 2786 | 1724 | 1053 | 0.379 | 48316 | 2487 | 2423 | 64 | 0.026 | 330616 | 8032 | 8031 | 0 | 0.000 | 314458 | 7601 | 7588 | 13 | 0.002 |
| 435866 | G | 79971691 | 79971691 | C | G | BRWD3 | missense | c.2290 | p.E764Q | tier1 | 15971 | 1677 | 1649 | 28 | 0.017 | 7953 | 825 | 474 | 347 | 0.423 | 11898 | 765 | 748 | 17 | 0.022 | 48921 | 1557 | 1556 | 1 | 0.000 | 33426 | 1092 | 1087 | 4 | 0.004 |
| 435866 | X | 105450024 | 105450024 | C | T | MUM1L1 | missense | c.599 | p.S200L | tier1 | 20377 | 2063 | 2022 | 41 | 0.020 | 9473 | 1402 | 770 | 625 | 0.448 | 16794 | 1075 | 1037 | 38 | 0.035 | 57612 | 1879 | 1879 | 0 | 0.000 | 47350 | 1819 | 1809 | 10 | 0.005 |
| 437422 | 1 | 22186507 | 22186507 | G | T | HSPG2 | intronic | c.5015-12 | e41-12 | tier3 | 6210 | 757 | 751 | 6 | 0.008 | 18971 | 2526 | 1695 | 806 | 0.322 | 7469 | 1114 | 676 | 435 | 0.392 | 20271 | 2076 | 2075 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 1 | 27736195 | 27736195 | T | C | WASF2 | missense | c.1330 | p.I444V | tier1 | 39553 | 5203 | 5176 | 24 | 0.005 | 45505 | 6561 | 4508 | 1091 | 0.195 | 22789 | 3091 | 2535 | 547 | 0.177 | 38378 | 4209 | 4208 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 1 | 167802237 | 167802237 | C | A | ADCY10 | missense | p.S1194L | tier1 | 48246 | 6354 | 6310 | 38 | 0.006 | 54754 | 6118 | 4536 | 1892 | 0.294 | 36721 | 4905 | 3194 | 1683 | 0.345 | 53584 | 5840 | 5839 | 0 | 0.000 | NA | NA | NA | NA | NA | |
| 437422 | 1 | 190067860 | 190067860 | C | T | BRINP3 | missense | c.1589 | p.R530H | tier1 | 5912 | 779 | 772 | 7 | 0.009 | 10489 | 1421 | 937 | 476 | 0.337 | 3869 | 611 | 378 | 232 | 0.380 | 6350 | 563 | 563 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 1 | 208061976 | 208061976 | C | T | CD3A | intronic | c.972+51 | e7+51 | tier3 | 18308 | 2319 | 2298 | 17 | 0.007 | 27880 | 3295 | 2198 | 1014 | 0.316 | 13762 | 1929 | 1131 | 744 | 0.397 | 14095 | 1205 | 1205 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 2 | 75105821 | 75105821 | G | A | HK2 | silent | c.1038 | p.K346 | tier1 | 44629 | 5762 | 5722 | 36 | 0.006 | 50097 | 6406 | 4761 | 1563 | 0.247 | 24618 | 3335 | 2544 | 777 | 0.234 | 48945 | 5265 | 5265 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 3 | 167006710 | 167006710 | C | G | ZBBX | missense | c.1941 | p.L647F | tier1 | 9013 | 1149 | 1145 | 3 | 0.003 | 17092 | 2055 | 1482 | 553 | 0.272 | 6258 | 844 | 515 | 327 | 0.388 | 10012 | 996 | 996 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 4 | 166416855 | 166416855 | C | T | CPE | intronic | c.1332+26 | e8+26 | tier3 | 6557 | 608 | 599 | 6 | 0.010 | 8555 | 928 | 684 | 240 | 0.260 | 2666 | 365 | 298 | 65 | 0.179 | 5769 | 485 | 484 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 5 | 133898014 | 133898014 | G | A | PHF15 | missense | c.809 | p.G270E | tier1 | 50979 | 6709 | 6650 | 53 | 0.008 | 73568 | 8933 | 6541 | 2249 | 0.256 | 35058 | 4418 | 2614 | 1778 | 0.405 | 66875 | 7560 | 7556 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 7 | 6193554 | 6193554 | C | T | USP42 | missense | c.2369 | p.A790V | tier1 | 3404 | 344 | 341 | 3 | 0.009 | 11954 | 1499 | 952 | 538 | 0.361 | 2454 | 303 | 149 | 154 | 0.508 | 13019 | 1462 | 1462 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 7 | 77885784 | 77885784 | C | T | MAGI2 | missense | c.1523 | p.R508H | tier1 | 30588 | 3946 | 3942 | 2 | 0.001 | 34134 | 4080 | 3868 | 189 | 0.047 | 17554 | 2389 | 2364 | 19 | 0.008 | 30241 | 3233 | 3231 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 7 | 120428042 | 120428042 | C | T | TSPAN12 | 3_prime_untranslated_region | c.*604 | NULL | tier2 | 1472 | 133 | 132 | 1 | 0.008 | 1502 | 165 | 97 | 66 | 0.405 | 1501 | 200 | 78 | 122 | 0.610 | 1946 | 142 | 142 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 8 | 113299258 | 113299258 | T | C | CSMD3 | intronic | c.9331+35 | e58+35 | tier3 | 17383 | 1991 | 1982 | 7 | 0.004 | 18396 | 2131 | 1693 | 419 | 0.198 | 9338 | 1227 | 981 | 237 | 0.195 | 14994 | 1457 | 1457 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 11 | 46686904 | 46686904 | T | G | ATG13 | intronic | c.901-29 | e11-29 | tier3 | 14943 | 2657 | 2636 | 20 | 0.008 | 23533 | 3805 | 2787 | 978 | 0.260 | 6979 | 1265 | 771 | 489 | 0.388 | 13833 | 1866 | 1863 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 11 | 130067769 | 130067769 | G | A | ST14 | missense | p.R463Q | tier1 | 42157 | 5856 | 5813 | 38 | 0.006 | 65488 | 8559 | 5739 | 2666 | 0.317 | 23607 | 3278 | 2152 | 1116 | 0.341 | 54687 | 6245 | 6239 | 1 | 0.000 | NA | NA | NA | NA | NA | |
| 437422 | 12 | 122091150 | 122091150 | T | G | MORN3 | missense | c.479 | p.Y160S | tier1 | 16248 | 1872 | 1852 | 17 | 0.009 | 24384 | 2836 | 2111 | 692 | 0.247 | 8373 | 1173 | 890 | 279 | 0.239 | 22977 | 2190 | 2189 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 16 | 10721483 | 10721483 | C | T | TEKTS | missense | c.1415 | p.R472H | tier1 | 55554 | 7362 | 7296 | 62 | 0.008 | 70491 | 8711 | 5995 | 2588 | 0.302 | 39020 | 4875 | 3188 | 1662 | 0.343 | 73592 | 8724 | 8714 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 17 | 7576851 | 7576851 | A | C | TP53 | splice_site | c.993+2 | e8+2 | tier1 | 44688 | 5679 | 5636 | 42 | 0.007 | 52860 | 6575 | 4754 | 1704 | 0.264 | 31257 | 4164 | 2524 | 1613 | 0.390 | 47704 | 5128 | 5127 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 437422 | 17 | 7578431 | 7578431 | G | A | TP53 | nonsense | c.499 | p.Q167* | tier1 | 52545 | 6923 | 6900 | 20 | 0.003 | 78898 | 9845 | 7659 | 2086 | 0.214 | 28499 | 3380 | 2758 | 613 | 0.182 | 80392 | 9700 | 9697 | 2 | 0.000 | NA | NA | NA | NA | NA |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 439820 | 12 | 112888165 | 112888165 | G | C | PTPN11 | missense | c.181 | p.D61H | tier1 | 17019 | 2381 | 2381 | 0 | 0.000 | 27691 | 3692 | 3552 | 127 | 0.035 | NA | NA | NA | NA | NA | 8182 | 600 | 599 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 439820 | 14 | 24518609 | 24518609 | A | G | DHRS4L1 | intronic | c.NULL | NULL | tier3 | 11208 | 1382 | 1346 | 35 | 0.025 | 8552 | 1062 | 569 | 488 | 0.462 | NA | NA | NA | NA | NA | 8219 | 520 | 285 | 229 | 0.446 | NA | NA | NA | NA | NA |
| 439820 | 14 | 24524337 | 24524337 | G | T | LRRCL16B | missense | c.523 | p.V175F | tier1 | 42204 | 5515 | 5451 | 59 | 0.011 | 64239 | 8489 | 5435 | 2861 | 0.345 | NA | NA | NA | NA | NA | 27810 | 2145 | 1365 | 730 | 0.348 | NA | NA | NA | NA | NA |
| 439820 | 14 | 100792064 | 100792064 | A | G | SLC25A47 | intronic | c.29-61 | e2-61 | tier3 | 33480 | 4481 | 4466 | 13 | 0.003 | 48595 | 6186 | 3338 | 2733 | 0.450 | NA | NA | NA | NA | NA | 33562 | 2482 | 1356 | 1052 | 0.437 | NA | NA | NA | NA | NA |
| 439820 | 15 | 91438809 | 91438809 | G | C | FES | 3_prime_untranslated_region | c.*21 | NULL | tier3 | 65267 | 8280 | 8038 | 226 | 0.027 | 71347 | 8751 | 4374 | 4236 | 0.492 | NA | NA | NA | NA | NA | 71441 | 5563 | 2723 | 2601 | 0.489 | NA | NA | NA | NA | NA |
| 439820 | 16 | 2098338 | 2098338 | A | G | TSC2 | intronic | c.1-250 | e1-250 | tier2 | 18307 | 2518 | 2446 | 70 | 0.028 | 21592 | 2809 | 1461 | 1303 | 0.471 | NA | NA | NA | NA | NA | 22564 | 1514 | 841 | 630 | 0.428 | NA | NA | NA | NA | NA |
| 439820 | 16 | 7761443 | 7761443 | G | A | RBFOX1 | 3_prime_untranslated_region | c.*1800 | NULL | tier3 | 2496 | 298 | 298 | 0 | 0.000 | 2507 | 381 | 315 | 64 | 0.169 | NA | NA | NA | NA | NA | 1075 | 65 | 61 | 4 | 0.062 | NA | NA | NA | NA | NA |
| 439820 | 16 | 24582766 | 24582766 | G | A | RBBP6 | missense | c.4379 | p.R1460H | tier1 | 17959 | 2428 | 2420 | 7 | 0.003 | 17409 | 2195 | 1405 | 773 | 0.355 | NA | NA | NA | NA | NA | 12458 | 864 | 832 | 31 | 0.036 | NA | NA | NA | NA | NA |
| 439820 | 16 | 29576812 | 29576812 | T | C | ENSG00000205534 | 5_prime_untranslated_region | c.-29576812 | NULL | tier3 | 1391 | 306 | 304 | 1 | 0.003 | 467 | 90 | 70 | 20 | 0.222 | NA | NA | NA | NA | NA | 872 | 87 | 69 | 18 | 0.207 | NA | NA | NA | NA | NA |
| 439820 | 16 | 50835415 | 50835415 | A | G | CYLD | 3_prime_untranslated_region | c.*4996 | NULL | tier3 | 4653 | 771 | 766 | 5 | 0.006 | 4221 | 757 | 475 | 281 | 0.272 | NA | NA | NA | NA | NA | 1830 | 149 | 51 | 98 | 0.658 | NA | NA | NA | NA | NA |
| 439820 | 16 | 50835731 | 50835731 | C | G | CYLD | 3_prime_untranslated_region | c.*5312 | NULL | tier3 | 8889 | 1109 | 1102 | 7 | 0.006 | 7198 | 877 | 494 | 379 | 0.434 | NA | NA | NA | NA | NA | 4248 | 288 | 275 | 13 | 0.045 | NA | NA | NA | NA | NA |
| 439820 | 17 | 7577538 | 7577538 | C | T | TP53 | missense | c.743 | p.R248Q | tier1 | 52868 | 6841 | 6796 | 41 | 0.006 | 48827 | 6763 | 3251 | 3434 | 0.514 | NA | NA | NA | NA | NA | 54609 | 5015 | 583 | 4355 | 0.882 | NA | NA | NA | NA | NA |
| 439820 | 17 | 15942836 | 15942836 | G | C | NCOR1 | missense | c.6866 | p.P2289R | tier1 | 10804 | 1457 | 1450 | 7 | 0.005 | 27645 | 3562 | 1980 | 1546 | 0.438 | NA | NA | NA | NA | NA | 15923 | 1078 | 1056 | 22 | 0.020 | NA | NA | NA | NA | NA |
| 439820 | 18 | 7034550 | 7034550 | A | G | LAMA1 | missense | c.1979 | p.M660T | tier1 | 23162 | 3026 | 3013 | 12 | 0.004 | 25313 | 3185 | 1683 | 1473 | 0.467 | NA | NA | NA | NA | NA | 16596 | 1119 | 611 | 492 | 0.446 | NA | NA | NA | NA | NA |
| 439820 | 18 | 14752893 | 14752893 | A | T | ANKRD30B | missense | c.392 | p.D131V | tier1 | 20437 | 2533 | 2516 | 15 | 0.006 | 23599 | 2708 | 1563 | 1114 | 0.416 | NA | NA | NA | NA | NA | 14513 | 973 | 552 | 408 | 0.425 | NA | NA | NA | NA | NA |
| 439820 | 18 | 29046556 | 29046556 | G | C | DSG3 | missense | c.1475 | p.C492S | tier1 | 10536 | 1356 | 1354 | 1 | 0.001 | 10670 | 1545 | 1355 | 181 | 0.118 | NA | NA | NA | NA | NA | 7036 | 455 | 455 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 439820 | 18 | 34205430 | 34205430 | G | A | FHOD3 | intronic | c.958-44 | e10-44 | tier3 | 41360 | 5457 | 5440 | 14 | 0.003 | 72467 | 9398 | 5602 | 3588 | 0.390 | NA | NA | NA | NA | NA | 37662 | 2681 | 2623 | 54 | 0.020 | NA | NA | NA | NA | NA |
| 439820 | 18 | 42531907 | 42531907 | G | A | SETBP1 | missense | c.2602 | p.D868N | tier1 | 51353 | 6527 | 6524 | 2 | 0.000 | 57353 | 7166 | 5804 | 1291 | 0.182 | NA | NA | NA | NA | NA | 43185 | 3202 | 3197 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 439820 | 19 | 1529504 | 1529504 | G | T | PLK5 | intronic | c.490+15 | e5+15 | tier3 | 31908 | 4066 | 4053 | 8 | 0.002 | 6117 | 8129 | 6082 | 1914 | 0.239 | NA | NA | NA | NA | NA | 48578 | 3616 | 2823 | 674 | 0.193 | NA | NA | NA | NA | NA |
| 439820 | 19 | 9007457 | 9007457 | C | G | MUC16 | intronic | c.39481+30 | e43+30 | tier3 | 10321 | 1911 | 1909 | 0 | 0.000 | 17429 | 2955 | 2833 | 114 | 0.039 | NA | NA | NA | NA | NA | 11975 | 901 | 897 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 439820 | 20 | 61291782 | 61291782 | G | C | SLCO4A1 | missense | c.906 | p.E302D | tier1 | 28170 | 0 | 0 | 0 | 0.000 | 35961 | 4117 | 3851 | 241 | 0.059 | NA | NA | NA | NA | NA | 34128 | 2211 | 2203 | 4 | 0.002 | NA | NA | NA | NA | NA |
| 439820 | 21 | 44514777 | 44514777 | T | G | U2AF1 | missense | c.470 | p.Q157P | tier1 | 82663 | 9996 | 9964 | 29 | 0.003 | 72468 | 8904 | 4774 | 3977 | 0.454 | NA | NA | NA | NA | NA | 71838 | 5386 | 2886 | 2289 | 0.442 | NA | NA | NA | NA | NA |
| 439820 | X | 31462671 | 31462671 | G | A | DMD | missense | c.9011 | p.T3004I | tier1 | 1252 | 252 | 250 | 2 | 0.008 | 1950 | 343 | 305 | 308 | 0.898 | NA | NA | NA | NA | NA | 1066 | 82 | 11 | 71 | 0.866 | NA | NA | NA | NA | NA |
| 439820 | X | 110439784 | 110439784 | G | C | PAK3 | silent | c.1431 | p.P477 | tier1 | 15220 | 1855 | 1852 | 1 | 0.001 | 18680 | 2359 | 2083 | 270 | 0.115 | NA | NA | NA | NA | NA | 10952 | 717 | 715 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 439820 | X | 128781951 | 128781951 | C | G | APLN | intronic | c.128-57 | e2-57 | tier3 | 15355 | 1915 | 1910 | 1 | 0.001 | 27274 | 3302 | 2875 | 403 | 0.123 | NA | NA | NA | NA | NA | 19751 | 1355 | 1354 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 1 | 26671642 | 26671642 | C | A | AIM1L | intronic | c.169+204 | e2+204 | tier3 | 26825 | 3130 | 3079 | 47 | 0.015 | 18889 | 2234 | 1466 | 735 | 0.334 | 10329 | 930 | 926 | 0 | 0.000 | 19161 | 1789 | 1786 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 1 | 176525591 | 176525591 | T | C | PAPPA2 | silent | c.133 | p.L45 | tier1 | 27459 | 4743 | 4734 | 6 | 0.001 | 33177 | 5047 | 4199 | 797 | 0.160 | 23438 | 2273 | 2272 | 1 | 0.000 | 38444 | 5788 | 5765 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 2 | 177065811 | 177065811 | C | T | ENSG00000218175 | 5_prime_untranslated_region | c.-177065811 | NULL | tier2 | 12225 | 2393 | 2352 | 36 | 0.015 | 13526 | 2481 | 1643 | 821 | 0.333 | 8020 | 1176 | 1175 | 1 | 0.000 | 22146 | 3628 | 3620 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 2 | 231906196 | 231906196 | G | T | Corf72 | intronic | c.748+72 | e2+72 | tier3 | 43393 | 6048 | 5956 | 89 | 0.015 | 39518 | 5641 | 3617 | 1951 | 0.350 | 25848 | 2421 | 2167 | 251 | 0.104 | 41531 | 5734 | 5722 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 3 | 41290971 | 41290971 | C | T | ULK4 | splice_region | c.3764+9 | e35+9 | tier3 | 12559 | 1618 | 1563 | 53 | 0.033 | 15126 | 2777 | 1729 | 1016 | 0.370 | 6906 | 1192 | 712 | 426 | 0.401 | 14701 | 2140 | 2130 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 4 | 15937803 | 15937803 | G | A | FGFBP1 | silent | c.453 | p.V151 | tier1 | 20767 | 2413 | 2405 | 7 | 0.003 | 25432 | 4080 | 3441 | 598 | 0.148 | 22016 | 2491 | 2489 | 1 | 0.000 | 30540 | 4620 | 4601 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 4 | 66388703 | 66388703 | G | A | EPH5 | intronic | c.911-27442 | e4-27442 | tier3 | 24146 | 2659 | 2619 | 38 | 0.014 | 18130 | 2190 | 1474 | 707 | 0.324 | 13564 | 1493 | 1492 | 1 | 0.000 | 22748 | 2567 | 2565 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 4 | 126389815 | 126389815 | C | G | FAT4 | missense | c.12048 | p.D4016E | tier1 | 25267 | 2770 | 2721 | 45 | 0.016 | 16861 | 2011 | 1288 | 713 | 0.356 | 20450 | 2061 | 2056 | 3 | 0.001 | 23099 | 2502 | 2498 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 4 | 183601392 | 183601392 | T | C | TENM3 | splice_region | c.1538-9 | e8-9 | tier3 | 33948 | 3900 | 3791 | 104 | 0.027 | 33966 | 4245 | 2462 | 1729 | 0.413 | 27668 | 2943 | 1787 | 1154 | 0.392 | 36682 | 4303 | 4291 | 0 | 0.000 | 2 | 0 | 0 | 0 | 0 |
| 476807 | 5 | 137589102 | 137589102 | G | A | GFR3 | intronic | c.1025-38 | e7-38 | tier3 | 20660 | 2558 | 2533 | 22 | 0.009 | 36371 | 5100 | 3300 | 1706 | 0.339 | 27372 | 2641 | 2634 | 3 | 0.000 | 37247 | 4754 | 4744 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 476807 | 6 | 32007827 | 32007827 | C | A | CYP21A2 | missense | c.784 | p.L262I | tier1 | 56872 | 7169 | 7050 | 100 | 0.014 | 57645 | 7513 | 4682 | 2714 | 0.367 | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 481668 | 1 | 46724289 | 46724289 | A | G | RAD54L | intronic | c.211-69 | e4-69 | tier3 | 3728 | 352 | 348 | 3 | 0.009 | 4127 | 467 | 333 | 129 | 0.279 | 4663 | 442 | 341 | 100 | 0.227 | 6618 | 663 | 660 | 0 | 0.000 | 7398 | 958 | 953 | 0 | 0.000 |
| 481668 | 1 | 46724300 | 46724300 | A | C | RAD54L | intronic | c.211-58 | e4-58 | tier3 | 3721 | 348 | 345 | 3 | 0.009 | 3860 | 409 | 290 | 114 | 0.282 | 4346 | 408 | 317 | 91 | 0.223 | 6286 | 634 | 629 | 0 | 0.000 | 6955 | 874 | 871 | 0 | 0.000 |
| 481668 | 1 | 152382172 | 152382172 | G | T | CRNN | silent | p.T462 | tier1 | 3656 | 699 | 673 | 26 | 0.037 | 4598 | 845 | 495 | 347 | 0.412 | 3837 | 682 | 429 | 251 | 0.369 | 7407 | 1549 | 1542 | 0 | 0.000 | 6010 | 1393 | 1393 | 0 | 0.000 | |
| 481668 | 1 | 182827980 | 182827980 | A | G | DHX9 | missense | c.1013 | p.N338S | tier1 | 13613 | 1470 | 1424 | 38 | 0.026 | 11084 | 1197 | 635 | 554 | 0.466 | 12071 | 1209 | 753 | 441 | 0.369 | 24757 | 3098 | 3090 | 0 | 0.000 | 21291 | 2749 | 2744 | 0 | 0.000 |
| 481668 | 1 | 247094587 | 247094587 | T | C | AHCTF1 | 5_prime_untranslated_region | c.-59 | NULL | tier2 | 8842 | 960 | 946 | 10 | 0.010 | 9923 | 1202 | 946 | 250 | 0.209 | 11692 | 1277 | 1010 | 252 | 0.200 | 11538 | 1532 | 1526 | 0 | 0.000 | 16834 | 2591 | 2576 | 0 | 0.000 |
| 481668 | 2 | 1457512 | 1457512 | C | G | TPO | missense | c.529 | p.L177V | tier1 | 5765 | 702 | 697 | 4 | 0.006 | 6019 | 713 | 552 | 157 | 0.221 | 6186 | 692 | 562 | 128 | 0.186 | 9818 | 1206 | 1204 | 0 | 0.000 | 12114 | 2003 | 2003 | 0 | 0.000 |
| 481668 | 2 | 98853198 | 98853198 | A | G | VWA3B | rna | NULL | NULL | tier1 | 5984 | 1092 | 1074 | 16 | 0.015 | 6090 | 1039 | 745 | 290 | 0.280 | 5740 | 941 | 734 | 207 | 0.220 | 9855 | 1695 | 1694 | 0 | 0.000 | 10850 | 0 | 0 | 0 | NA |
| 481668 | 4 | 95223388 | 95223388 | A | G | HPGDS | missense | c.344 | p.M115T | tier1 | 12213 | 1137 | 1122 | 15 | 0.013 | 11031 | 1005 | 745 | 252 | 0.253 | 12423 | 1090 | 887 | 199 | 0.183 | 26792 | 2858 | 2851 | 0 | 0.000 | 22357 | 2518 | 2516 | 0 | 0.000 |
| 481668 | 5 | 40959580 | 40959580 | T | C | C7 | missense | c.1519 | p.S507P | tier1 | 7747 | 819 | 809 | 10 | 0.012 | 7204 | 885 | 667 | 210 | 0.239 | 8286 | 866 | 706 | 159 | 0.184 | 18259 | 2312 | 2310 | 0 | 0.000 | 16683 | 2352 | 2349 | 0 | 0.000 |
| 481668 | 6 | 31760727 | 31760727 | G | C | VAR5 | 5_prime_untranslated_region | c.-31760727 | NULL | tier3 | 24956 | 2944 | 2901 | 35 | 0.012 | 20786 | 2318 | 1680 | 611 | 0.267 | 23466 | 2570 | 1914 | 629 | 0.247 | 43507 | 5492 | 5491 | 0 | 0.000 | 48077 | 6563 | 6556 | 0 | 0.000 |
| 481668 | 6 | 135513482 | 135513482 | A | G | MYB | missense | c.548 | p.N183S | tier1 | 35587 | 3818 | 3791 | 18 | 0.005 | 26391 | 2634 | 2017 | 591 | 0.227 | 34159 | 3561 | 2856 | 685 | 0.193 | 76034 | 9071 | 9057 | 0 | 0.000 | 57160 | 7122 | 7118 | 0 | 0.000 |
| 481668 | 7 | 34118627 | 34118627 | C | T | BMPER | missense | c.1237 | p.R413C | tier1 | 14570 | 1549 | 1549 | 0 | 0.000 | 11823 | 1351 | 1303 | 44 | 0.033 | 12466 | 1281 | 1227 | 48 | 0.038 | 21312 | 2525 | 2522 | 0 | 0.000 | 28573 | 3860 | 3857 | 2 | 0.000 |
| 481668 | 7 | 81593304 | 81593304 | T | G | CACNA2D1 | intronic | c.2816+68 | e34+68 | tier3 | 1705 | 109 | 109 | 0 | 0.000 | 2205 | 196 | 141 | 53 | 0.273 | 12172 | 170 | 128 | 42 | 0.247 | 3870 | 330 | 329 | 0 | 0.000 | 3117 | 0 | 0 | 0 | NA |
| 481668 | 7 | 100635373 | 100635373 | C | A | MUC12 | missense | c.1529 | p.A510D | tier1 | 2463 | 396 | 392 | 4 | 0.010 | 4026 | 708 | 608 | 96 | 0.136 | 3048 | 458 | 401 | 56 | 0.123 | 3012 | 478 | 478 | 0 | 0.000 | 3124 | 688 | 688 | 0 | 0.000 |
| 481668 | 7 | 148506166 | 148506166 | T | A | EZH2 | missense | c.2192 | p.Y731F | tier1 | 8451 | 912 | 903 | 6 | 0.007 | 8077 | 890 | 667 | 213 | 0.242 | 9517 | 923 | 727 | 191 | 0.208 | 18595 | 2258 | 2248 | 0 | 0.000 | 18191 | 2380 | 2378 | 0 | 0.000 |
| 481668 | 9 | 117138957 | 117138957 | C | T | AKNA | missense | c.1130 | p.R377H | tier1 | 22983 | 2341 | 2339 | 0 | 0.000 | 28470 | 2718 | 2683 | 31 | 0.011 | 33291 | 3187 | 3146 | 36 | 0.011 | 61527 | 6990 | 6987 | 1 | 0.000 | 51395 | 6372 | 6365 | 1 | 0.000 |
| 481668 | 10 | 128933807 | 128933807 | T | C | FAM196A | 3_prime_untranslated_region | c.*2294 | NULL | tier3 | 21752 | 2267 | 2260 | 4 | 0.002 | 14782 | 1446 | 1037 | 402 | 0.279 | 17718 | 1730 | 1335 | 374 | 0.219 | 41494 | 4873 | 4871 | 0 | 0.000 | 35598 | 4377 | 4365 | 0 | 0.000 |
| 481668 | 10 | 133761131 | 133761131 | G | T | PPP2R2D | missense | c.825 | p.M275I | tier1 | 10710 | 988 | 970 | 17 | 0.017 | 10104 | 1020 | 715 | 289 | 0.288 | 11101 | 961 | 745 | 202 | 0.243 | 16775 | 1728 | 1717 | 0 | 0.000 | 18919 | 2252 | 2246 | 0 | 0.000 |
| 481668 | 11 | 71903432 | 71903432 | A | G | FOLR1 | intronic | c.168+47 | e1+47 | tier3 | 14145 | 1616 | 1592 | 21 | 0.013 | 11382 | 1245 | 938 | 302 | 0.244 | 13164 | 1428 | 1148 | 272 | 0.192 | 31410 | 3880 | 3875 | 0 | 0.000 | 26207 | 3615 | 3609 | 0 | 0.000 |
| 481668 | 12 | 21608977 | 21608977 | G | T | PYROXD1 | missense | c.698 | p.S233I | tier1 | 5224 | 530 | 528 | 2 | 0.004 | 4079 | 411 | 310 | 101 | 0.264 | 4963 | 511 | 393 | 114 | 0.223 | 11108 | 1256 | 1255 | 0 | 0.000 | 7277 | 0 | 0 | 0 | NA |
| 481668 | 12 | 113599210 | 113599210 | C | A | DDX54 | rna | NULL | NULL | tier1 | 20029 | 2240 | 2213 | 24 | 0.011 | 13203 | 1300 | 974 | 316 | 0.245 | 15970 | 1689 | 1421 | 260 | 0.155 | 43807 | 5340 | 5338 | 0 | 0.000 | 33543 | 4263 | 4263 | 0 | 0.000 |
| 481668 | 13 | 33223009 | 33223009 | C | T | PDS5B | nonsense | c.100 | p.R34* | tier1 | 16363 | 1781 | 1779 | 2 | 0.001 | 12157 | 1188 | 927 | 251 | 0.213 | 14519 | 1510 | 1322 | 180 | 0.120 | 34218 | 4018 | 4016 | 0 | 0.000 | 25116 | 3035 | 3032 | 0 | 0.000 |
| 481668 | 15 | 56704519 | 56704519 | G | A | TEX9 | missense | c.848 | p.R283K | tier1 | 2211 | 293 | 280 | 9 | 0.031 | 1985 | 289 | 210 | 76 | 0.266 | 2392 | 295 | 233 | 58 | 0.199 | 3371 | 475 | 474 | 0 | 0.000 | 3742 | 651 | 640 | 0 | 0.000 |
| 481668 | 16 | 30456662 | 30456662 | C | T | SEPHS2 | silent | c.216 | p.G72 | tier1 | 12494 | 1355 | 1353 | 2 | 0.001 | 13959 | 1454 | 1178 | 269 | 0.186 | 16910 | 1703 | 1432 | 263 | 0.155 | 43178 | 5513 | 5510 | 0 | 0.000 | 38962 | 5225 | 5222 | 0 | 0.000 |
| 481668 | 17 | 1559855 | 1559855 | T | C | PRPF8 | missense | c.5624 | p.H1875R | tier1 | 15946 | 1834 | 1797 | 34 | 0.019 | 16758 | 1956 | 1420 | 510 | 0.264 | 19459 | 2136 | 1660 | 453 | 0.213 | 31923 | 4209 | 4197 | 0 | 0.000 | 34447 | 4901 | 4898 | 0 | 0.000 |
| 481668 | 17 | 37425012 | 37425012 | A | G | FBXL20 | intronic | c.933+68 | e12+68 | tier3 | 10846 | 1172 | 1165 | 7 | 0.006 | 10269 | 1198 | 982 | 211 | 0.177 | 10353 | 1111 | 989 | 120 | 0.108 | 28442 | 3479 | 3474 | 0 | 0.000 | 15377 | 2154 | 2153 | 0 | 0.000 |
| 481668 | 17 | 37879957 | 37879957 | C | T | ERBB2 | intronic | c.2208+44 | e18+44 | tier3 | 5212 | 521 | 520 | 0 | 0.000 | 6237 | 677 | 512 | 159 | 0.237 | 6555 | 617 | 498 | 115 | 0.188 | 13991 | 1512 | 1504 | 0 | 0.000 | 14415 | 1838 | 1837 | 1 | 0.000 |
| 481668 | 19 | 9067438 | 9067438 | C | T | MUC16 | missense | c.20008 | p.V6670I | tier1 | 19427 | 2262 | 2245 | 15 | 0.007 | 19226 | 2052 | 1714 | 324 | 0.159 | 21888 | 2334 | 1986 | 340 | 0.146 | 30956 | 3817 | 3813 | 0 | 0.000 | 34532 | 4457 | 4456 | 0 | 0.000 |
| 481668 | 19 | 15219913 | 15219913 | C | G | SYDE1 | silent | c.135 | p.P45 | tier1 | 4176 | 377 | 368 | 8 | 0.021 | 5572 | 568 | 342 | 223 | 0.395 | 5464 | 497 | 328 | 166 | 0.336 | 7978 | 724 | 721 | 1 | 0.000 | 9781 | 1307 | 1306 | 0 | 0.000 |
| 485523 | 4 | 39474726 | 39474726 | G | A | LIAS | missense | c.961 | p.E321K | tier1 | 3132 | 0 | 0 | 0 | 0.000 | 2170 | 258 | 246 | 12 | 0.047 | 1363 | 116 | 115 | 1 | 0.000 | 1319 | 141 | 141 | 0 | 0.000 | 3714 | 578 | 576 | 2 | 0.000 |
| 485523 | 4 | 55976980 | 55976980 | G | A | KDR | intronic | c.977-45 | e8-45 | tier3 | 10736 | 1448 | 1448 | 0 | 0.000 | 6523 | 861 | 808 | 48 | 0.056 | 5751 | 648 | 645 | 2 | 0.000 | 4205 | 454 | 454 | 0 | 0.000 | 13940 | 2116 | 2116 | 0 | 0.000 |
| 485523 | 5 | 176939370 | 176939370 | C | T | DDX41 | missense | c.1574 | p.R525H | tier1 | 15143 | 1773 | 1771 | 2 | 0.001 | 9232 | 1070 | 965 | 101 | 0.095 | 6583 | 566 | 558 | 3 | 0.000 | 5809 | 622 | 621 | 0 | 0.000 | 17630 | 2449 | 2439 | 5 | 0.000 |
| 485523 | 6 | 73904728 | 73904728 | A | T | KCNQ5 | missense | c.2420 | p.N807I | tier1 | 4057 | 669 | 668 | 0 | 0.000 | 3091 | 499 | 451 | 46 | 0.093 | 517 | 55 | 54 | 0 | 0.000 | 536 | 52 | 52 | 0 | 0.000 | 2353 | 568 | 567 | 0 | 0.000 |
| 485523 | 14 | 24517447 | 24517447 | G | A | DHRS4L1 | rna | NULL | NULL | tier1 | 16574 | 1653 | 1652 | 0 | 0.000 | 12727 | 1385 | 1281 | 99 | 0.072 | 8756 | 776 | 773 | 2 | 0.000 | 7359 | 814 | 813 | 0 | 0.000 | 23426 | 3123 | 3117 | 2 | 0. |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 489196 | 21 | 45820168 | 45820168 | C | T | TRPM2 | silent | c.2235 | p.G745 | tier1 | 34108 | 4082 | 4056 | 22 | 0.005 | 34778 | 4093 | 2798 | 1222 | 0.304 | NA | NA | NA | NA | NA | 75462 | 10316 | 10286 | 12 | 0.001 | 54985 | 7242 | 7236 | 0 | 0.000 |
| 489196 | 22 | 46677825 | 46677825 | G | A | TTC38 | intronic | c.561+210 | e6+210 | tier3 | 34260 | 4122 | 4102 | 16 | 0.004 | 26194 | 2932 | 2248 | 660 | 0.227 | NA | NA | NA | NA | NA | 73321 | 9718 | 9703 | 6 | 0.001 | 46987 | 5789 | 5785 | 0 | 0.000 |
| 489196 | X | 73041567 | 73041567 | C | T | XIST | rna | NULL | NULL | tier1 | 11895 | 1242 | 1230 | 10 | 0.008 | 9624 | 973 | 706 | 265 | 0.273 | NA | NA | NA | NA | NA | 13631 | 1661 | 1654 | 6 | 0.004 | 8642 | 995 | 994 | 1 | 0.000 |
| 489196 | X | 153049418 | 153049418 | C | G | SRPK3 | intronic | c.908-11 | e10-11 | tier3 | 5196 | 437 | 435 | 2 | 0.005 | 4626 | 478 | 332 | 143 | 0.301 | NA | NA | NA | NA | NA | 3137 | 347 | 345 | 2 | 0.000 | 3545 | 393 | 393 | 0 | 0.000 |
| 489196 | X | 153799950 | 153799950 | G | A | G6PD | 5_prime_flanking_region | c.-25580 | NULL | tier3 | 11537 | 1214 | 1213 | 0 | 0.000 | 9553 | 1134 | 1084 | 35 | 0.031 | NA | NA | NA | NA | NA | 10962 | 1440 | 1424 | 0 | 0.000 | 11745 | 1444 | 1439 | 0 | 0.000 |
| 489513 | 1 | 173684860 | 173684860 | A | G | KLHL20 | intronic | c.1-282 | e1-282 | tier2 | 7687 | 1062 | 1032 | 29 | 0.027 | 3661 | 447 | 219 | 227 | 0.509 | NA | NA | NA | NA | NA | 3526 | 670 | 666 | 4 | 0.006 | NA | NA | NA | NA | NA |
| 489513 | 1 | 240351440 | 240351440 | T | C | FMN2 | intronic | c.1931-67 | e4-67 | tier3 | 1690 | 178 | 176 | 2 | 0.011 | 942 | 82 | 36 | 46 | 0.561 | NA | NA | NA | NA | NA | 2941 | 393 | 384 | 9 | 0.023 | NA | NA | NA | NA | NA |
| 489513 | 3 | 42735190 | 42735190 | C | T | HHATL | nonsense | c.1167 | p.W389* | tier1 | 42803 | 6640 | 6483 | 147 | 0.022 | 15733 | 2200 | 1175 | 1001 | 0.460 | NA | NA | NA | NA | NA | 39384 | 5362 | 5286 | 68 | 0.013 | NA | NA | NA | NA | NA |
| 489513 | 3 | 46792243 | 46792243 | C | T | PRSS50 | intronic | c.1-2102 | e1-2102 | tier2 | 2747 | 488 | 473 | 15 | 0.031 | 1107 | 163 | 100 | 62 | 0.383 | NA | NA | NA | NA | NA | 1506 | 306 | 303 | 3 | 0.010 | NA | NA | NA | NA | NA |
| 489513 | 3 | 53844274 | 53844274 | C | T | CACNA1D | silent | c.6201 | p.F2067 | tier1 | 41736 | 5111 | 4999 | 105 | 0.021 | 17998 | 1858 | 1010 | 823 | 0.449 | NA | NA | NA | NA | NA | 42641 | 5681 | 5590 | 72 | 0.013 | NA | NA | NA | NA | NA |
| 489513 | 4 | 152498539 | 152498539 | G | A | FAM160A1 | intronic | c.106-63 | e2-63 | tier3 | 18260 | 2039 | 1984 | 52 | 0.026 | 7899 | 824 | 443 | 374 | 0.458 | NA | NA | NA | NA | NA | 10697 | 1601 | 1578 | 19 | 0.012 | NA | NA | NA | NA | NA |
| 489513 | 5 | 20304957 | 20304957 | G | T | ENSG00000214132 | 5_prime_untranslated_region | c.-20304957 | NULL | tier3 | 3140 | 609 | 601 | 8 | 0.013 | 2086 | 339 | 191 | 146 | 0.433 | NA | NA | NA | NA | NA | 3159 | 738 | 730 | 7 | 0.009 | NA | NA | NA | NA | NA |
| 489513 | 5 | 137680472 | 137680472 | T | C | FAM53C | intronic | c.137-42 | e3-42 | tier4 | 5474 | 558 | 547 | 10 | 0.018 | 2653 | 220 | 124 | 95 | 0.434 | NA | NA | NA | NA | NA | 2757 | 365 | 362 | 3 | 0.008 | NA | NA | NA | NA | NA |
| 489513 | 6 | 41011404 | 41011404 | G | T | TSPO2 | silent | c.282 | p.L94 | tier1 | 11098 | 2070 | 2003 | 64 | 0.031 | 6397 | 997 | 553 | 441 | 0.444 | NA | NA | NA | NA | NA | 8035 | 1795 | 1781 | 13 | 0.007 | NA | NA | NA | NA | NA |
| 489513 | 7 | 131844359 | 131844359 | G | C | PLXNA4 | missense | c.4533 | p.N1511K | tier1 | 24100 | 2980 | 2902 | 76 | 0.026 | 11137 | 1173 | 659 | 502 | 0.432 | NA | NA | NA | NA | NA | 24642 | 3512 | 3457 | 47 | 0.013 | NA | NA | NA | NA | NA |
| 489513 | 8 | 56367730 | 56367730 | C | T | XKR4 | intronic | c.1007-68110 | e3-68110 | tier2 | 14043 | 1674 | 1627 | 47 | 0.028 | 5091 | 474 | 268 | 205 | 0.433 | NA | NA | NA | NA | NA | 17876 | 2343 | 2320 | 17 | 0.007 | NA | NA | NA | NA | NA |
| 489513 | 9 | 16215272 | 16215272 | C | T | C9orf92 | missense | c.196 | p.G66R | tier1 | 41132 | 5051 | 4945 | 100 | 0.020 | 12803 | 1316 | 734 | 549 | 0.421 | NA | NA | NA | NA | NA | 27768 | 3830 | 3790 | 32 | 0.008 | NA | NA | NA | NA | NA |
| 489513 | 9 | 32974446 | 32974446 | C | T | APT7 | splice_region | c.916+10 | e7+10 | tier3 | 5780 | 577 | 569 | 8 | 0.014 | 4635 | 423 | 234 | 187 | 0.444 | NA | NA | NA | NA | NA | 12127 | 1671 | 1649 | 17 | 0.010 | NA | NA | NA | NA | NA |
| 489513 | 9 | 101798470 | 101798470 | G | C | COL15A1 | missense | c.2308 | p.G770R | tier1 | 13774 | 1387 | 1357 | 27 | 0.020 | 5729 | 537 | 284 | 248 | 0.466 | NA | NA | NA | NA | NA | 5617 | 778 | 762 | 14 | 0.018 | NA | NA | NA | NA | NA |
| 489513 | 9 | 136402651 | 136402651 | G | A | ADAMTSL2 | missense | c.542 | p.R181Q | tier1 | 54189 | 7103 | 6931 | 157 | 0.022 | 18674 | 2142 | 1309 | 805 | 0.361 | NA | NA | NA | NA | NA | 44383 | 6365 | 6318 | 41 | 0.006 | NA | NA | NA | NA | NA |
| 489513 | 10 | 48373412 | 48373412 | G | A | ZNF488 | 3_prime_untranslated_region | c.*1857 | NULL | tier3 | 3512 | 641 | 618 | 22 | 0.034 | 1407 | 214 | 118 | 96 | 0.449 | NA | NA | NA | NA | NA | 1268 | 264 | 262 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 489513 | 10 | 87615842 | 87615842 | G | A | GRID1 | missense | c.1057 | p.R353W | tier1 | 23954 | 3097 | 3094 | 2 | 0.001 | 10954 | 1260 | 1204 | 49 | 0.039 | NA | NA | NA | NA | NA | 20858 | 3054 | 3041 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 489513 | 12 | 130883190 | 130883190 | C | A | RIMBP2 | 3_prime_untranslated_region | c.*490 | NULL | tier3 | 4458 | 545 | 535 | 10 | 0.018 | 6367 | 710 | 404 | 297 | 0.424 | NA | NA | NA | NA | NA | 9049 | 1279 | 1263 | 14 | 0.011 | NA | NA | NA | NA | NA |
| 489513 | 15 | 97634474 | 97634474 | A | G | ENSG00000215948 | 3_prime_flanking_region | c.*18 | NULL | tier4 | 18333 | 2225 | 2172 | 51 | 0.023 | 6681 | 641 | 372 | 265 | 0.416 | NA | NA | NA | NA | NA | 17743 | 2529 | 2500 | 24 | 0.010 | NA | NA | NA | NA | NA |
| 489513 | 17 | 78180746 | 78180746 | C | T | SGSH | rna | NULL | NULL | tier1 | 56063 | 7291 | 7156 | 123 | 0.017 | 16485 | 1730 | 951 | 754 | 0.442 | NA | NA | NA | NA | NA | 57883 | 7469 | 7371 | 85 | 0.011 | NA | NA | NA | NA | NA |
| 489513 | 18 | 72113811 | 72113811 | C | G | FAM69C | intronic | c.876+30 | e2+30 | tier3 | 22786 | 3096 | 3027 | 65 | 0.021 | 9806 | 1085 | 602 | 469 | 0.438 | NA | NA | NA | NA | NA | 20954 | 3102 | 3053 | 42 | 0.014 | NA | NA | NA | NA | NA |
| 489513 | 19 | 40834389 | 40834389 | C | T | C19orf47 | missense | c.481 | p.D161N | tier1 | 58706 | 7545 | 7359 | 172 | 0.023 | 20482 | 2078 | 1112 | 930 | 0.455 | NA | NA | NA | NA | NA | 51235 | 6581 | 6471 | 96 | 0.015 | NA | NA | NA | NA | NA |
| 489513 | 19 | 59081697 | 59081697 | G | A | MZF1 | 5_prime_untranslated_region | c.-59081697 | NULL | tier3 | 8951 | 1997 | 1910 | 83 | 0.042 | 3245 | 570 | 47 | 522 | 0.917 | NA | NA | NA | NA | NA | 2154 | 532 | 520 | 11 | 0.021 | NA | NA | NA | NA | NA |
| 489513 | 20 | 18772588 | 18772588 | G | A | LINC00652 | rna | NULL | NULL | tier1 | 23487 | 3150 | 3069 | 76 | 0.024 | 13692 | 1603 | 864 | 721 | 0.455 | NA | NA | NA | NA | NA | 27964 | 4395 | 4325 | 60 | 0.014 | NA | NA | NA | NA | NA |
| 489513 | 21 | 31964779 | 31964779 | C | T | KRTAP6-3 | 5_prime_untranslated_region | c.-7 | NULL | tier3 | 20233 | 2641 | 2574 | 67 | 0.025 | 15439 | 1508 | 837 | 653 | 0.438 | NA | NA | NA | NA | NA | 40721 | 5091 | 5022 | 60 | 0.012 | NA | NA | NA | NA | NA |
| 489513 | 21 | 44524456 | 44524456 | G | T | UZAF1 | missense | c.101 | p.S34Y | tier1 | 12826 | 1791 | 1765 | 24 | 0.013 | 4167 | 535 | 318 | 215 | 0.403 | NA | NA | NA | NA | NA | 15729 | 2277 | 2264 | 13 | 0.006 | NA | NA | NA | NA | NA |
| 489513 | X | 118215369 | 118215369 | G | A | KIAA1210 | missense | c.5053 | p.P1685S | tier1 | 4732 | 565 | 538 | 26 | 0.046 | 2442 | 275 | 17 | 256 | 0.938 | NA | NA | NA | NA | NA | 4732 | 673 | 657 | 16 | 0.024 | NA | NA | NA | NA | NA |
| 491309 | 17 | 7577580 | 7577580 | T | C | TP53 | missense | c.701 | p.Y234C | tier1 | 24823 | 3109 | 3103 | 2 | 0.001 | 48885 | 5769 | 5602 | 138 | 0.024 | 65674 | 8304 | 7619 | 641 | 0.078 | 109990 | 14706 | 14602 | 80 | 0.005 | NA | NA | NA | NA | NA |
| 491309 | 19 | 16629981 | 16629981 | C | T | CHERP | missense | c.2740 | p.E914K | tier1 | 9280 | 1125 | 1121 | 4 | 0.004 | 16539 | 2098 | 1986 | 105 | 0.050 | 18651 | 2178 | 2121 | 54 | 0.025 | 23755 | 2963 | 2952 | 7 | 0.002 | NA | NA | NA | NA | NA |
| 496321 | 1 | 986249 | 986249 | C | T | AGRN | intronic | c.5253+32 | e30+32 | tier2 | 6395 | 552 | 517 | 35 | 0.063 | 8718 | 816 | 416 | 392 | 0.485 | 772 | 719 | 2 | 0.000 | 14977 | 1510 | 1509 | 0 | 0.000 | 15007 | 0 | 0 | 0 | 0 | NA |
| 496321 | 1 | 47276735 | 47276735 | G | A | CYP4B1 | intronic | c.323-77 | e3-77 | tier3 | 4197 | 345 | 326 | 18 | 0.052 | 11848 | 1385 | 733 | 633 | 0.463 | 11871 | 1413 | 1410 | 0 | 0.000 | 17351 | 1849 | 1843 | 0 | 0.000 | 20371 | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 502635 | 4 | 119644046 | 119644046 | T | C | SEC24D | rna | NULL | NULL | tier1 | 1320 | 161 | 160 | 0 | 0.000 | 1910 | 212 | 202 | 9 | 0.043 | 3521 | 406 | 402 | 4 | 0.010 | 746 | 68 | 59 | 9 | 0.132 | NA | NA | NA | NA | NA |
| 502635 | 4 | 129192341 | 129192341 | A | G | PGRMC2 | 3_prime_untranslated_region | c.*130 | NULL | tier3 | 6482 | 696 | 694 | 1 | 0.001 | 7016 | 734 | 664 | 69 | 0.094 | 11664 | 1107 | 1088 | 18 | 0.016 | 3708 | 265 | 226 | 38 | 0.144 | NA | NA | NA | NA | NA |
| 502635 | 11 | 89515227 | 89515227 | G | T | TRIM64DP | 3_prime_untranslated_region | c.*89515227 | NULL | tier3 | 2097 | 0 | 0 | 0 | 0.000 | 3679 | 346 | 327 | 18 | 0.052 | 6779 | 607 | 593 | 14 | 0.023 | 1074 | 64 | 55 | 9 | 0.141 | NA | NA | NA | NA | NA |
| 502635 | 12 | 119583250 | 119583250 | G | A | SRRM4 | missense | c.836 | p.R279Q | tier1 | 29334 | 3284 | 3278 | 2 | 0.001 | 19115 | 2254 | 2051 | 190 | 0.085 | 24188 | 2596 | 2546 | 43 | 0.017 | 10772 | 836 | 713 | 116 | 0.140 | NA | NA | NA | NA | NA |
| 502635 | 13 | 101881935 | 101881935 | C | T | NALCN | missense | c.1435 | p.V479I | tier1 | 4923 | 554 | 554 | 0 | 0.000 | 6221 | 718 | 666 | 50 | 0.070 | 9292 | 966 | 953 | 13 | 0.013 | 2741 | 225 | 201 | 23 | 0.103 | NA | NA | NA | NA | NA |
| 502635 | 15 | 31086780 | 31086780 | G | T | GOLGA8UP | 3_prime_untranslated_region | c.*31086780 | NULL | tier3 | 10486 | 1232 | 1230 | 1 | 0.001 | 13369 | 1508 | 1435 | 71 | 0.047 | 10850 | 1174 | 1154 | 19 | 0.016 | 7523 | 517 | 489 | 24 | 0.047 | NA | NA | NA | NA | NA |
| 502635 | 15 | 43995984 | 43995984 | C | G | STRC | intronic | c.1+14335 | e0+14335 | tier3 | 767 | 138 | 137 | 0 | 0.000 | 662 | 102 | 99 | 3 | 0.029 | 846 | 107 | 104 | 3 | 0.028 | 515 | 56 | 48 | 8 | 0.143 | NA | NA | NA | NA | NA |
| 502635 | 17 | 7577538 | 7577538 | C | T | TP53 | missense | c.743 | p.R248Q | tier1 | 38153 | 4433 | 4425 | 6 | 0.001 | 32945 | 3829 | 3532 | 283 | 0.074 | 28494 | 3174 | 3108 | 63 | 0.020 | 15180 | 1077 | 928 | 139 | 0.130 | NA | NA | NA | NA | NA |
| 502635 | 17 | 7578551 | 7578551 | A | T | TP53 | missense | c.379 | p.S127T | tier1 | 13818 | 1658 | 1655 | 1 | 0.001 | 16613 | 1916 | 1722 | 184 | 0.097 | 12532 | 1356 | 1324 | 30 | 0.022 | 8583 | 676 | 560 | 110 | 0.164 | NA | NA | NA | NA | NA |
| 502635 | 19 | 852255 | 852255 | G | T | ELANE | 5_prime_untranslated_region | c.-74 | NULL | tier2 | 4171 | 489 | 487 | 2 | 0.004 | 3406 | 404 | 371 | 32 | 0.079 | 3728 | 337 | 330 | 5 | 0.015 | 1862 | 171 | 148 | 21 | 0.124 | NA | NA | NA | NA | NA |
| 565435 | 1 | 114641862 | 114641862 | G | A | SYT6 | silent | c.1218 | p.L406 | tier1 | 13332 | 1460 | 1436 | 21 | 0.014 | 12358 | 1356 | 1011 | 336 | 0.249 | 13950 | 2053 | 1500 | 545 | 0.267 | 30410 | 4732 | 4706 | 23 | 0.005 | 21075 | 2074 | 2071 | 0 | 0.000 |
| 565435 | 2 | 86267544 | 86267544 | G | A | POLR1A | silent | c.3711 | p.G1237 | tier1 | 15427 | 1412 | 1395 | 16 | 0.011 | 16396 | 1788 | 1789 | 479 | 0.270 | 13673 | 1612 | 1140 | 468 | 0.291 | 34567 | 3791 | 3764 | 23 | 0.006 | 20206 | 1117 | 1113 | 2 | 0.000 |
| 565435 | 2 | 97849131 | 97849131 | A | G | ANKRD36 | intronic | c.1838-65 | e27-65 | tier3 | 528 | 46 | 45 | 1 | 0.022 | 1085 | 113 | 85 | 28 | 0.248 | 765 | 93 | 70 | 22 | 0.239 | 1638 | 370 | 368 | 2 | 0.000 | 3649 | 0 | 0 | 0 | NA |
| 565435 | 2 | 130750444 | 130750444 | C | T | ENSG00000250207 | 5_prime_untranslated_region | c.-130750444 | NULL | tier2 | 351 | 26 | 25 | 0 | 0.000 | 636 | 61 | 48 | 12 | 0.200 | 355 | 41 | 24 | 17 | 0.415 | 563 | 120 | 119 | 1 | 0.000 | 1926 | 339 | 338 | 0 | 0.000 |
| 565435 | 2 | 219267322 | 219267322 | C | A | CTDSP1 | intronic | c.378+194 | e4+194 | tier3 | 8388 | 714 | 711 | 2 | 0.003 | 7748 | 826 | 714 | 105 | 0.128 | 4564 | 597 | 463 | 137 | 0.222 | 19292 | 2606 | 2603 | 3 | 0.001 | 14893 | 1211 | 1210 | 0 | 0.000 |
| 565435 | 3 | 44776777 | 44776777 | T | C | ZNF501 | 3_prime_untranslated_region | c.*48 | NULL | tier3 | 7090 | 579 | 573 | 5 | 0.009 | 7945 | 811 | 649 | 160 | 0.198 | 11616 | 1406 | 1190 | 216 | 0.154 | 31913 | 4940 | 4926 | 14 | 0.003 | 17342 | 2067 | 2063 | 0 | 0.000 |
| 565435 | 3 | 133465239 | 133465239 | A | G | TF | 5_prime_untranslated_region | c.-46 | NULL | tier2 | 3473 | 238 | 235 | 3 | 0.013 | 3364 | 269 | 192 | 76 | 0.284 | 1841 | 165 | 123 | 42 | 0.255 | 8407 | 1147 | 1140 | 7 | 0.006 | 5576 | 0 | 0 | 0 | NA |
| 565435 | 3 | 155314188 | 155314188 | T | C | PLCH1 | intronic | c.44-21 | e2-21 | tier3 | 9276 | 793 | 785 | 7 | 0.009 | 8745 | 953 | 753 | 191 | 0.203 | 14495 | 1629 | 1288 | 339 | 0.208 | 27101 | 3325 | 3311 | 11 | 0.003 | 15555 | 1226 | 1221 | 0 | 0.000 |
| 565435 | 3 | 185300560 | 185300560 | G | C | SENP2 | missense | c.26 | p.C9S | tier1 | 9745 | 864 | 851 | 12 | 0.014 | 14733 | 1605 | 1208 | 385 | 0.242 | 13183 | 1730 | 1266 | 452 | 0.263 | 32185 | 4173 | 4144 | 23 | 0.006 | 24573 | 1659 | 1655 | 0 | 0.000 |
| 565435 | 4 | 73615881 | 73615881 | G | A | - | - | - | - | tier3 | 26937 | 2477 | 2430 | 43 | 0.017 | 26692 | 2859 | 2248 | 589 | 0.208 | 26582 | 3244 | 2500 | 771 | 0.235 | 76644 | 8771 | 8732 | 32 | 0.004 | 36637 | 2514 | 2513 | 0 | 0.000 |
| 565435 | 5 | 13900350 | 13900350 | G | A | DNAH5 | nonsense | c.2224 | p.R742* | tier1 | 5591 | 438 | 434 | 4 | 0.009 | 4799 | 460 | 353 | 96 | 0.214 | 7223 | 868 | 628 | 234 | 0.271 | 13127 | 2282 | 2253 | 22 | 0.010 | 11748 | 1543 | 1541 | 0 | 0.000 |
| 565435 | 5 | 43295963 | 43295963 | G | A | HMGCS1 | missense | c.796 | p.P266S | tier1 | 4562 | 289 | 279 | 7 | 0.024 | 4865 | 382 | 280 | 99 | 0.261 | 8802 | 864 | 619 | 240 | 0.279 | 15512 | 2493 | 2475 | 12 | 0.005 | 11081 | 1346 | 1344 | 0 | 0.000 |
| 565435 | 6 | 31974832 | 31974832 | G | A | CYP21A1P | 3_prime_untranslated_region | c.*31974832 | NULL | tier3 | 19729 | 1949 | 1903 | 31 | 0.016 | 30673 | 3473 | 2601 | 848 | 0.246 | 21705 | 2676 | 2013 | 650 | 0.244 | 65202 | 6589 | 6567 | 18 | 0.003 | 30819 | 1590 | 1588 | 0 | 0.000 |
| 565435 | 6 | 54208081 | 54208081 | C | T | TINAG | missense | c.682 | p.P228S | tier1 | 12078 | 986 | 968 | 17 | 0.017 | 10417 | 1004 | 729 | 273 | 0.272 | 13417 | 1445 | 1043 | 387 | 0.271 | 32114 | 4012 | 3989 | 22 | 0.005 | 14691 | 1140 | 1138 | 0 | 0.000 |
| 565435 | 6 | 80774316 | 80774316 | T | G | ENSG00000218418 | 5_prime_untranslated_region | c.-80774316 | NULL | tier3 | 634 | 69 | 69 | 0 | 0.000 | 1258 | 155 | 116 | 37 | 0.242 | 732 | 108 | 73 | 35 | 0.324 | 2184 | 503 | 501 | 2 | 0.000 | 3436 | 475 | 475 | 0 | 0.000 |
| 565435 | 7 | 5434025 | 5434025 | A | T | TNRC18 | intronic | c.343+46 | e2+46 | tier3 | 28683 | 2819 | 2744 | 69 | 0.025 | 38038 | 4331 | 3092 | 1173 | 0.275 | 21117 | 2581 | 1840 | 726 | 0.283 | 81084 | 8697 | 8607 | 72 | 0.008 | 39889 | 2285 | 2281 | 0 | 0.000 |
| 565435 | 7 | 122255199 | 122255199 | A | C | CADPS2 | intronic | c.1223+36 | e6+36 | tier3 | 16095 | 1339 | 1324 | 14 | 0.010 | 13338 | 1225 | 870 | 348 | 0.286 | 20287 | 2269 | 1663 | 594 | 0.263 | 37377 | 4585 | 4559 | 22 | 0.005 | 20122 | 1472 | 1469 | 0 | 0.000 |
| 565435 | 7 | 150894251 | 150894251 | C | T | IQCA1P1 | intronic | c.NULL | NULL | tier3 | 18487 | 2029 | 2007 | 19 | 0.009 | 17305 | 2041 | 1617 | 413 | 0.203 | 13181 | 1698 | 1319 | 378 | 0.223 | 46816 | 4639 | 4622 | 13 | 0.003 | 25609 | 1611 | 1611 | 0 | 0.000 |
| 565435 | 8 | 144399939 | 144399939 | G | A | TOP1MT | silent | c.1284 | p.Y428 | tier1 | 24944 | 2355 | 2315 | 28 | 0.012 | 28184 | 3061 | 2293 | 740 | 0.244 | 23661 | 2765 | 1988 | 765 | 0.278 | 64552 | 7314 | 7266 | 40 | 0.005 | 26804 | 1782 | 1779 | 0 | 0.000 |
| 565435 | 9 | 32631391 | 32631391 | C | T | TAFL1 | missense | c.4187 | p.R1396H | tier1 | 7746 | 844 | 824 | 14 | 0.017 | 6167 | 767 | 598 | 207 | 0.271 | 8987 | 1462 | 1057 | 394 | 0.272 | 17753 | 3052 | 3028 | 16 | 0.005 | 13268 | 1185 | 1181 | 1 | 0.000 |
| 565435 | 9 | 74299332 | 74299332 | T | C | TMEM2 | 5_prime_untranslated_region | c.-74299332 | NULL | tier3 | 8801 | 690 | 684 | 5 | 0.007 | 7996 | 721 | 597 | 121 | 0.169 | 8110 | 889 | 699 | 189 | 0.213 | 27911 | 3462 | 3448 | 13 | 0.004 | 13679 | 1229 | 1229 | 0 | 0.000 |
| 565435 | 9 | 84608191 | 84608191 | T | C | SPATA31D1 | missense | c.2806 | p.S936P | tier1 | 13459 | 1305 | 1287 | 17 | 0.013 | 13293 | 1507 | 1079 | 414 | 0.277 | 12511 | 1524 | 1118 | 399 | 0.263 | 30199 | 3568 | 3552 | 11 | 0.003 | 17830 | 1385 | 1385 | 0 | 0.000 |
| 565435 | 10 | 95849013 | 95849013 | G | A | PLCE1 | silent | c.162 | p.Q54 | tier1 | 40409 | 3967 | 3916 | 47 | 0.012 | 41148 | 4573 | 3346 | 1164 | 0.258 | 32204 | 3872 | 2697 | 1136 | 0.296 | 113707 | 11487 | 11398 | 72 | 0.006 | 43245 | 2411 | 2411 | 0 | 0.000 |
| 565435 | 11 | 56738588 | 56738588 | G | A | OR5AK3P | 3_prime_untranslated_region | c.*56738588 | NULL | tier3 | 974 | 104 | 104 | 0 | 0.000 | 2719 | 366 | 332 | 32 | 0.088 | 2050 | 355 | 347 | 8 | 0.023 | 5834 | 1395 | 1389 | 4 | 0.003 | 6721 | 0 | 0 | 0 | NA |
| 565435 | 14 | 21853909 | 21853909 | C | T | CHD8 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|------------------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 597501 | 10 | 135015401 | 135015401 | C | T | <i>KNDC1</i> | missense | c.3392 | p.P1131L | tier1 | 1958 | 243 | 234 | 9 | 0.037 | 4299 | 445 | 258 | 186 | 0.419 | NA | NA | NA | NA | NA | 5562 | 473 | 466 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 597501 | 11 | 31531364 | 31531364 | C | T | <i>ELP4</i> | silent | c.33 | p.A11 | tier1 | 27473 | 3999 | 3971 | 24 | 0.006 | 19624 | 2414 | 2007 | 382 | 0.160 | NA | NA | NA | NA | NA | 20172 | 1795 | 1782 | 9 | 0.005 | NA | NA | NA | NA | NA |
| 597501 | 12 | 117715866 | 117715866 | C | A | <i>NOS1</i> | missense | c.1562 | p.R521L | tier1 | 3155 | 537 | 516 | 20 | 0.037 | 3394 | 508 | 259 | 248 | 0.489 | NA | NA | NA | NA | NA | 4660 | 918 | 907 | 7 | 0.008 | NA | NA | NA | NA | NA |
| 597501 | 13 | 25060389 | 25060389 | G | A | <i>PARP4</i> | silent | c.1269 | p.T423 | tier1 | 1065 | 174 | 170 | 4 | 0.023 | 1410 | 234 | 124 | 110 | 0.470 | NA | NA | NA | NA | NA | 1212 | 278 | 277 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 597501 | 14 | 25100251 | 25100251 | G | A | <i>GZMB</i> | 3_prime_untranslated_region | c.*26 | NULL | tier3 | 22328 | 2840 | 2769 | 70 | 0.025 | 15565 | 1542 | 810 | 722 | 0.471 | NA | NA | NA | NA | NA | 30947 | 2864 | 2816 | 32 | 0.011 | NA | NA | NA | NA | NA |
| 597501 | 14 | 86089071 | 86089071 | C | G | <i>FLRT2</i> | missense | c.1213 | p.L405V | tier1 | 11507 | 1841 | 1788 | 53 | 0.029 | 8447 | 1090 | 621 | 460 | 0.426 | NA | NA | NA | NA | NA | 26350 | 2636 | 2613 | 19 | 0.007 | NA | NA | NA | NA | NA |
| 597501 | 15 | 34651895 | 34651895 | C | T | <i>LPCAT4</i> | missense | c.1294 | p.G432S | tier1 | 20898 | 2652 | 2572 | 73 | 0.028 | 17216 | 1747 | 1007 | 707 | 0.412 | NA | NA | NA | NA | NA | 47201 | 3973 | 3940 | 24 | 0.006 | NA | NA | NA | NA | NA |
| 597501 | 15 | 91574882 | 91574882 | T | A | <i>LOC101926911</i> | intronic | c.NULL | NULL | tier3 | 134 | 20 | 19 | 1 | 0.050 | 187 | 24 | 19 | 5 | 0.208 | NA | NA | NA | NA | NA | 122 | 31 | 31 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 597501 | 16 | 2522630 | 2522630 | G | A | <i>NTN3</i> | missense | c.928 | p.A310T | tier1 | 10326 | 1406 | 1373 | 33 | 0.023 | 6192 | 761 | 444 | 315 | 0.415 | NA | NA | NA | NA | NA | 3675 | 500 | 492 | 5 | 0.010 | NA | NA | NA | NA | NA |
| 597501 | 17 | 11532944 | 11532944 | C | T | <i>DNAH9</i> | intronic | c.1518+43 | e7+43 | tier3 | 33290 | 4249 | 4236 | 12 | 0.003 | 19803 | 2073 | 1984 | 76 | 0.037 | NA | NA | NA | NA | NA | 28820 | 2610 | 2604 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 597501 | 17 | 28900664 | 28900664 | C | T | <i>LOC101927093</i> | rna | c.NULL | NULL | tier1 | 3062 | 551 | 534 | 17 | 0.031 | 3737 | 478 | 243 | 231 | 0.487 | NA | NA | NA | NA | NA | 4772 | 684 | 675 | 6 | 0.009 | NA | NA | NA | NA | NA |
| 597501 | 17 | 29422352 | 29422352 | T | G | <i>NF1</i> | missense | c.25 | p.W9G | tier1 | 1964 | 236 | 235 | 1 | 0.004 | 3233 | 309 | 266 | 40 | 0.131 | NA | NA | NA | NA | NA | 1351 | 108 | 108 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 597501 | 17 | 30219342 | 30219342 | C | G | <i>UTP6</i> | missense | c.361 | p.A121P | tier1 | 11134 | 1686 | 1655 | 29 | 0.017 | 6298 | 726 | 402 | 319 | 0.442 | NA | NA | NA | NA | NA | 20253 | 2333 | 2313 | 16 | 0.007 | NA | NA | NA | NA | NA |
| 597501 | 17 | 74732959 | 74732959 | G | T | <i>SRSF2</i> | missense | c.284 | p.P95H | tier1 | 10293 | 1262 | 1212 | 49 | 0.039 | 8221 | 963 | 489 | 448 | 0.478 | NA | NA | NA | NA | NA | 14711 | 1525 | 1507 | 17 | 0.011 | NA | NA | NA | NA | NA |
| 597501 | 18 | 29523048 | 29523048 | C | T | <i>ENSG00000263823</i> | rna | c.NULL | NULL | tier1 | 9944 | 1283 | 1238 | 44 | 0.034 | 8373 | 963 | 495 | 459 | 0.481 | NA | NA | NA | NA | NA | 10009 | 890 | 878 | 11 | 0.012 | NA | NA | NA | NA | NA |
| 597501 | 19 | 17440815 | 17440815 | G | A | <i>ANO8</i> | missense | c.1282 | p.R428W | tier1 | 18125 | 2340 | 2297 | 39 | 0.017 | 16282 | 1820 | 1289 | 504 | 0.281 | NA | NA | NA | NA | NA | 33601 | 3412 | 3379 | 21 | 0.006 | NA | NA | NA | NA | NA |
| 597501 | 19 | 47884624 | 47884624 | G | A | <i>DHX34</i> | 3_prime_untranslated_region | c.*47884624 | NULL | tier3 | 41418 | 5490 | 5361 | 121 | 0.022 | 18930 | 2084 | 1128 | 933 | 0.453 | NA | NA | NA | NA | NA | 80784 | 5946 | 5873 | 58 | 0.010 | NA | NA | NA | NA | NA |
| 597501 | 19 | 54080590 | 54080590 | T | C | <i>ZNF331</i> | missense | c.776 | p.I259T | tier1 | 15559 | 1995 | 1949 | 45 | 0.023 | 10394 | 1170 | 652 | 505 | 0.364 | NA | NA | NA | NA | NA | 28227 | 3026 | 2971 | 41 | 0.014 | NA | NA | NA | NA | NA |
| 597501 | 20 | 48098486 | 48098486 | G | A | <i>KCNB1</i> | silent | c.532 | p.L178 | tier1 | 16072 | 2068 | 1992 | 74 | 0.036 | 9762 | 1145 | 590 | 542 | 0.479 | NA | NA | NA | NA | NA | 27257 | 2577 | 2544 | 32 | 0.012 | NA | NA | NA | NA | NA |
| 597501 | X | 10084387 | 10084387 | T | C | <i>WWC3</i> | intronic | c.975-71 | e9-71 | tier3 | 32016 | 4112 | 4011 | 99 | 0.024 | 16975 | 1721 | 885 | 814 | 0.479 | NA | NA | NA | NA | NA | 63246 | 5639 | 5578 | 56 | 0.010 | NA | NA | NA | NA | NA |
| 597501 | X | 70357542 | 70357542 | C | T | <i>MED12</i> | intronic | c.5836-34 | e41-34 | tier3 | 18298 | 2446 | 2377 | 67 | 0.027 | 8295 | 924 | 465 | 456 | 0.495 | NA | NA | NA | NA | NA | 33528 | 2365 | 2337 | 25 | 0.011 | NA | NA | NA | NA | NA |
| 621996 | 1 | 4772000 | 4772000 | C | A | <i>AJAP1</i> | missense | c.70 | p.H24N | tier1 | 21542 | 2074 | 2032 | 36 | 0.017 | 34011 | 3568 | 2250 | 1270 | 0.361 | 28983 | 2915 | 1603 | 1275 | 0.443 | 33573 | 4104 | 4095 | 6 | 0.000 | 50278 | 6422 | 6415 | 1 | 0.000 |
| 621996 | 1 | 91404719 | 91404719 | T | C | <i>ZNF644</i> | missense | c.2192 | p.N731S | tier1 | 11123 | 990 | 976 | 14 | 0.014 | 14226 | 1396 | 851 | 528 | 0.383 | 15909 | 1537 | 861 | 657 | 0.433 | 20461 | 2305 | 2297 | 5 | 0.000 | 37025 | 5065 | 5060 | 4 | 0.000 |
| 621996 | 1 | 154841865 | 154841865 | G | A | <i>KCNM3</i> | silent | c.576 | p.S192 | tier1 | 7417 | 724 | 716 | 8 | 0.011 | 12115 | 1249 | 834 | 398 | 0.323 | 12983 | 1394 | 808 | 569 | 0.413 | 11537 | 1270 | 1269 | 0 | 0.000 | 18414 | 2897 | 2896 | 0 | 0.000 |
| 621996 | 1 | 176769238 | 176769238 | C | T | <i>PAPPA2</i> | silent | c.5172 | p.P1724 | tier1 | 9531 | 990 | 970 | 20 | 0.020 | 10865 | 1175 | 783 | 388 | 0.331 | 12110 | 1297 | 726 | 564 | 0.437 | 11017 | 1305 | 1305 | 0 | 0.000 | 22967 | 3292 | 3289 | 1 | 0.000 |
| 621996 | 2 | 98354060 | 98354060 | C | T | <i>ZAP70</i> | missense | c.1414 | p.R472W | tier1 | 12198 | 1552 | 1550 | 0 | 0.000 | 25854 | 2879 | 2392 | 465 | 0.163 | 26318 | 3171 | 2781 | 362 | 0.115 | 24275 | 2923 | 2916 | 1 | 0.000 | 49497 | 7446 | 7430 | 3 | 0.000 |
| 621996 | 2 | 166223736 | 166223736 | G | A | <i>SCN2A</i> | missense | c.3530 | p.R1177Q | tier1 | 98 | 9 | 9 | 0 | 0.000 | 127 | 19 | 12 | 7 | 0.368 | 213 | 28 | 13 | 15 | 0.536 | 179 | 0 | 0 | 0 | 0.000 | 488 | 84 | 84 | 0 | 0.000 |
| 621996 | 2 | 179604342 | 179604342 | C | T | <i>TTN</i> | missense | c.13105 | p.V4369M | tier1 | 6466 | 601 | 596 | 4 | 0.007 | 6700 | 691 | 437 | 250 | 0.364 | 8076 | 78 | 449 | 334 | 0.427 | 8898 | 1067 | 1065 | 1 | 0.000 | 12585 | 1891 | 1891 | 0 | 0.000 |
| 621996 | 2 | 197709191 | 197709191 | T | C | <i>PGAP1</i> | intronic | c.2337+57 | e24+57 | tier3 | 1048 | 86 | 83 | 3 | 0.035 | 1292 | 107 | 56 | 51 | 0.477 | 1690 | 128 | 72 | 56 | 0.438 | 1301 | 127 | 126 | 1 | 0.000 | 3502 | 443 | 442 | 0 | 0.000 |
| 621996 | 3 | 38291377 | 38291377 | G | A | <i>OXSRI</i> | intronic | c.1323-80 | e15-80 | tier3 | 3291 | 323 | 321 | 2 | 0.006 | 3035 | 327 | 226 | 100 | 0.307 | 3164 | 352 | 210 | 142 | 0.403 | 3956 | 441 | 441 | 0 | 0.000 | 6631 | 1199 | 1191 | 1 | 0.000 |
| 621996 | 3 | 50312255 | 50312255 | C | G | <i>SEMA3B</i> | 3_prime_untranslated_region | c.*50312255 | NULL | tier2 | 17760 | 2173 | 2145 | 25 | 0.012 | 27636 | 3136 | 2023 | 1079 | 0.348 | 27358 | 3188 | 1796 | 1328 | 0.425 | 28112 | 3621 | 3615 | 3 | 0.000 | 44541 | 7415 | 7398 | 3 | 0.000 |
| 621996 | 3 | 125879686 | 125879686 | G | A | <i>ALDH1L1</i> | splice_region | c.127+10 | e1+10 | tier3 | 57489 | 6208 | 6190 | 4 | 0.001 | 50722 | 5652 | 4444 | 1109 | 0.200 | 45533 | 4909 | 3578 | 1253 | 0.259 | 63552 | 8069 | 8058 | 0 | 0.000 | 91484 | 11615 | 11579 | 0 | 0.000 |
| 621996 | 4 | 125592366 | 125592366 | T | G | <i>ANKRD50</i> | missense | c.2066 | p.H689P | tier1 | 17618 | 2018 | 1975 | 39 | 0.019 | 32256 | 3727 | 2433 | 1239 | 0.337 | 41529 | 4967 | 2768 | 2091 | 0.430 | 35605 | 4752 | 4739 | 4 | 0.000 | 54131 | 8759 | 8755 | 2 | 0.000 |
| 621996 | 5 | 133483867 | 133483867 | T | G | <i>TCF7</i> | 3_prime_untranslated_region | c.*1873 | NULL | tier3 | 4571 | 424 | 418 | 6 | 0.014 | 5018 | 472 | 336 | 135 | 0.287 | 5461 | 504 | 317 | 184 | 0.367 | 5853 | 575 | 575 | 0 | 0.000 | 14536 | 0 | 0 | 0 | 0.000 |
| 621996 | 5 | 154255096 | 154255096 | C | A | <i>CNOT8</i> | 3_prime_untranslated_region | c.*97 | NULL | tier3 | 15241 | 1862 | 1844 | 17 | 0.009 | 18256 | 2288 | 1498 | 768 | 0.339 | 17860 | 2200 | 1256 | 920 | 0.423 | 25501 | 3479 | 3477 | 1 | 0.000 | 49890 | 7806 | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 625338 | 21 | 36231782 | 36231782 | C | T | RUNX1 | missense | c.602 | p.R201Q | tier1 | 27269 | 3823 | 3818 | 2 | 0.001 | 15956 | 2090 | 2041 | 42 | 0.020 | 13997 | 1319 | 1310 | 7 | 0.005 | 34911 | 5289 | 5285 | 3 | 0.000 | 93815 | 10945 | 10941 | 0 | 0.000 |
| 625338 | 21 | 36252865 | 36252865 | C | T | RUNX1 | missense | c.497 | p.R166Q | tier1 | 27077 | 3350 | 3349 | 1 | 0.000 | 11292 | 1186 | 1152 | 30 | 0.025 | 12298 | 1138 | 1128 | 10 | 0.009 | 37453 | 5311 | 5307 | 0 | 0.000 | 42670 | 4903 | 4900 | 1 | 0.000 |
| 627844 | 2 | 10224073 | 10224073 | G | A | ENSG00000233502 | splice_site | c.165+1 | e1+1 | tier1 | 22088 | 2486 | 2466 | 18 | 0.007 | 26382 | 3471 | 2707 | 750 | 0.217 | 33344 | 4412 | 4396 | 16 | 0.004 | 56061 | 7553 | 7544 | 5 | 0.000 | 51131 | 6045 | 6040 | 0 | 0.000 |
| 627844 | 3 | 31674390 | 31674390 | A | T | STT3B | intronic | c.2188-37 | e15-37 | tier3 | 5173 | 513 | 509 | 3 | 0.006 | 7302 | 1335 | 928 | 397 | 0.300 | 11962 | 1724 | 1720 | 4 | 0.002 | 15477 | 2362 | 2358 | 0 | 0.000 | 16990 | 2293 | 2291 | 0 | 0.000 |
| 627844 | 3 | 122432409 | 122432409 | A | G | PARP14 | missense | c.3758 | p.N1253S | tier1 | 9170 | 972 | 957 | 15 | 0.015 | 16600 | 2288 | 1487 | 787 | 0.346 | 15323 | 1939 | 1929 | 10 | 0.005 | 23362 | 3068 | 3066 | 0 | 0.000 | 25468 | 3056 | 3054 | 0 | 0.000 |
| 627844 | 4 | 20530641 | 20530641 | G | A | SLIT2 | missense | c.1532 | p.R511H | tier1 | 8277 | 901 | 899 | 2 | 0.002 | 12327 | 1644 | 1505 | 137 | 0.083 | 11470 | 1579 | 1579 | 0 | 0.000 | 21940 | 3005 | 3002 | 1 | 0.000 | 19508 | 2342 | 2337 | 2 | 0.000 |
| 627844 | 6 | 69795034 | 69795034 | C | T | BAI3 | intronic | c.2480+9104 | e15+9104 | tier3 | 6593 | 621 | 611 | 8 | 0.013 | 25424 | 3258 | 2092 | 1142 | 0.353 | 17212 | 1969 | 1953 | 14 | 0.007 | 22372 | 2603 | 2600 | 0 | 0.000 | 22841 | 2344 | 2339 | 0 | 0.000 |
| 627844 | 6 | 135515531 | 135515531 | G | A | MYB | missense | c.881 | p.R294Q | tier1 | 4308 | 415 | 412 | 3 | 0.007 | 15388 | 1791 | 1462 | 321 | 0.180 | 18041 | 2118 | 2100 | 16 | 0.008 | 30939 | 3838 | 3835 | 2 | 0.000 | 20305 | 1946 | 1941 | 1 | 0.000 |
| 627844 | 6 | 146719968 | 146719968 | C | T | GRM1 | missense | c.1793 | p.A598V | tier1 | 5415 | 502 | 499 | 3 | 0.006 | 14772 | 1879 | 1298 | 566 | 0.304 | 12703 | 1475 | 1463 | 8 | 0.005 | 23733 | 2877 | 2876 | 0 | 0.000 | 23184 | 2557 | 2552 | 0 | 0.000 |
| 627844 | 7 | 27236580 | 27236580 | G | A | HOXA13 | 3_prime_untranslated_region | c.*1237 | NULL | tier3 | 2953 | 190 | 189 | 1 | 0.005 | 2439 | 250 | 184 | 62 | 0.252 | 5482 | 560 | 557 | 0 | 0.000 | 7055 | 721 | 720 | 0 | 0.000 | 7531 | 554 | 553 | 0 | 0.000 |
| 627844 | 7 | 80303472 | 80303472 | G | A | CD36 | 3_prime_untranslated_region | c.*9 | NULL | tier2 | 4025 | 247 | 245 | 1 | 0.004 | 5162 | 620 | 430 | 183 | 0.299 | 7630 | 735 | 726 | 8 | 0.011 | 10410 | 1042 | 1041 | 0 | 0.000 | 11879 | 1070 | 1068 | 0 | 0.000 |
| 627844 | 8 | 82373788 | 82373788 | G | A | FABP9 | 5_prime_untranslated_region | c.-30 | NULL | tier3 | 22416 | 2311 | 2288 | 19 | 0.008 | 39215 | 4843 | 3315 | 1483 | 0.309 | 42733 | 5081 | 5062 | 15 | 0.003 | 71337 | 8935 | 8928 | 3 | 0.000 | 55364 | 5922 | 5917 | 1 | 0.000 |
| 627844 | 8 | 105503259 | 105503259 | C | T | LRP12 | missense | c.11375 | p.R722H | tier1 | 11375 | 1292 | 1280 | 10 | 0.008 | 16557 | 2232 | 1585 | 624 | 0.282 | 17302 | 2239 | 2233 | 5 | 0.002 | 30729 | 4178 | 4175 | 1 | 0.000 | 23471 | 2850 | 2848 | 1 | 0.000 |
| 627844 | 11 | 73980724 | 73980724 | A | G | PAHA3 | silent | c.1440 | p.Y480 | tier1 | 25154 | 2444 | 2418 | 25 | 0.010 | 39632 | 4778 | 3157 | 1575 | 0.333 | 28032 | 3297 | 3273 | 19 | 0.006 | 59221 | 7211 | 7205 | 0 | 0.000 | 57035 | 5848 | 5845 | 0 | 0.000 |
| 627844 | 12 | 8186333 | 8186333 | G | T | FOXJ2 | 5_prime_untranslated_region | c.-6096 | NULL | tier2 | 829 | 127 | 126 | 1 | 0.008 | 5559 | 1185 | 1094 | 89 | 0.075 | 5694 | 0 | 0 | 0 | NA | 7809 | 1517 | 1515 | 0 | 0.000 | 9569 | 1672 | 1672 | 0 | 0.000 |
| 627844 | 12 | 64841839 | 64841839 | A | G | XPOT | intronic | c.2863-46 | e24-46 | tier3 | 6777 | 610 | 606 | 3 | 0.005 | 20947 | 2609 | 2349 | 240 | 0.093 | 17522 | 1979 | 1977 | 0 | 0.000 | 39917 | 4834 | 4832 | 0 | 0.000 | 32445 | 3249 | 3242 | 0 | 0.000 |
| 627844 | 12 | 112185009 | 112185009 | C | T | ACAD10 | 3_prime_untranslated_region | c.*112185009 | NULL | tier3 | 5275 | 471 | 470 | 1 | 0.002 | 8750 | 1314 | 1085 | 220 | 0.169 | 9514 | 1207 | 1201 | 4 | 0.003 | 14527 | 1924 | 1922 | 0 | 0.000 | 18831 | 2384 | 2379 | 0 | 0.000 |
| 627844 | 12 | 112884192 | 112884192 | C | T | PTPN11 | missense | c.127 | p.L43F | tier1 | 25136 | 2456 | 2452 | 4 | 0.002 | 39423 | 5037 | 4467 | 535 | 0.107 | 43769 | 5135 | 5133 | 0 | 0.000 | 65353 | 8100 | 8096 | 0 | 0.000 | 61427 | 6649 | 6646 | 0 | 0.000 |
| 627844 | 14 | 86088433 | 86088433 | T | A | FLRT2 | missense | c.575 | p.I192N | tier1 | 25520 | 2429 | 2421 | 7 | 0.003 | 38670 | 4755 | 3908 | 796 | 0.169 | 29889 | 3437 | 3428 | 9 | 0.003 | 67453 | 8379 | 8369 | 0 | 0.000 | 53919 | 5524 | 5523 | 0 | 0.000 |
| 627844 | 15 | 75798204 | 75798204 | G | C | PTPN9 | silent | c.780 | p.P260 | tier1 | 15640 | 1769 | 1760 | 9 | 0.005 | 20641 | 2706 | 2528 | 169 | 0.063 | 18404 | 2319 | 2317 | 0 | 0.000 | 36426 | 4783 | 4780 | 0 | 0.000 | 31203 | 3567 | 3565 | 0 | 0.000 |
| 627844 | 16 | 47675018 | 47675018 | G | A | PHKB | intronic | c.1514+17 | e15+17 | tier3 | 7847 | 741 | 737 | 4 | 0.005 | 9527 | 1226 | 973 | 247 | 0.202 | 14238 | 1601 | 1598 | 1 | 0.000 | 19727 | 2361 | 2360 | 0 | 0.000 | 17331 | 1744 | 1740 | 0 | 0.000 |
| 627844 | 16 | 89836214 | 89836214 | G | C | FANCA | 3_prime_untranslated_region | c.*27 | NULL | tier3 | 15798 | 1453 | 1431 | 21 | 0.014 | 23007 | 3037 | 2001 | 1011 | 0.336 | 26819 | 3434 | 3406 | 26 | 0.008 | 41923 | 5467 | 5465 | 0 | 0.000 | 37877 | 4168 | 4167 | 0 | 0.000 |
| 627844 | 17 | 9282009 | 9282009 | C | G | STX8 | intronic | c.542-39 | e7-39 | tier3 | 7514 | 853 | 845 | 8 | 0.009 | 9564 | 1517 | 1010 | 497 | 0.330 | 11850 | 1693 | 1682 | 8 | 0.005 | 18722 | 2862 | 2859 | 0 | 0.000 | 16798 | 2181 | 2177 | 1 | 0.000 |
| 627844 | 17 | 11622752 | 11622752 | G | A | DNAH9 | missense | c.5654 | p.R1885H | tier1 | 9774 | 1127 | 1115 | 11 | 0.010 | 16539 | 2497 | 1641 | 840 | 0.339 | 8450 | 1159 | 1154 | 4 | 0.003 | 22633 | 3261 | 3259 | 1 | 0.000 | 25002 | 3404 | 3400 | 1 | 0.000 |
| 627844 | 18 | 61323095 | 61323095 | G | A | SERPINB3 | silent | c.969 | p.L323 | tier1 | 13174 | 1288 | 1266 | 22 | 0.017 | 24056 | 3005 | 2017 | 953 | 0.321 | 13982 | 1689 | 1677 | 11 | 0.007 | 34058 | 4209 | 4204 | 2 | 0.000 | 32700 | 3484 | 3481 | 3 | 0.000 |
| 627844 | 19 | 20045145 | 20045145 | G | A | ZNF93 | missense | c.1381 | p.A461T | tier1 | 5354 | 481 | 480 | 0 | 0.000 | 5729 | 724 | 588 | 135 | 0.187 | 11047 | 1333 | 1327 | 6 | 0.005 | 15713 | 1998 | 1998 | 0 | 0.000 | 12317 | 1297 | 1295 | 1 | 0.000 |
| 627844 | X | 13337504 | 13337504 | C | T | ATXN3 | missense | c.550 | p.E184K | tier1 | 8492 | 885 | 873 | 12 | 0.014 | 8287 | 1095 | 501 | 588 | 0.540 | 12108 | 1497 | 1482 | 15 | 0.010 | 20652 | 2716 | 2710 | 2 | 0.000 | 17110 | 1885 | 1883 | 1 | 0.000 |
| 627844 | X | 119387527 | 119387527 | A | G | ZBTB33 | missense | c.257 | p.Y86C | tier1 | 4911 | 365 | 358 | 7 | 0.019 | 9814 | 1168 | 371 | 794 | 0.682 | 10059 | 1056 | 1037 | 19 | 0.018 | 17729 | 1941 | 1939 | 0 | 0.000 | 17129 | 1615 | 1613 | 0 | 0.000 |
| 635308 | 1 | 65307066 | 65307066 | C | G | JAK1 | intronic | c.2555-44 | e18-44 | tier3 | 7983 | 863 | 861 | 0 | 0.000 | 4637 | 574 | 515 | 57 | 0.100 | 2413 | 0 | 0 | 0 | NA | 1833 | 351 | 351 | 0 | 0.000 | 21 | 0 | 0 | 0 | NA |
| 635308 | 1 | 84394863 | 84394863 | T | A | TLL7 | silent | c.1098 | p.V366 | tier1 | 15865 | 1769 | 1760 | 6 | 0.003 | 13655 | 1503 | 1341 | 153 | 0.102 | 12523 | 1522 | 1520 | 0 | 0.000 | 48958 | 5596 | 5582 | 1 | 0.000 | 48044 | 5917 | 5914 | 0 | 0.000 |
| 635308 | 1 | 86591118 | 86591118 | C | T | COL24A1 | missense | c.901 | p.V301M | tier1 | 346 | 60 | 60 | 0 | 0.000 | 504 | 81 | 68 | 13 | 0.160 | 1111 | 191 | 183 | 7 | 0.037 | 348 | 88 | 86 | 2 | 0.000 | 277 | 0 | 0 | 0 | NA |
| 635308 | 1 | 222802593 | 222802593 | C | T | MIA3 | missense | c.778 | p.L260F | tier1 | 1972 | 340 | 337 | 3 | 0.009 | 4488 | 873 | 730 | 137 | 0.158 | 4624 | 840 | 838 | 1 | 0.000 | 2106 | 476 | 471 | 3 | 0.006 | 436 | 39 | 39 | 0 | 0.000 |
| 635308 | 3 | 112328837 | 112328837 | C | T | CCDC80 | missense | c.2413 | p.A805T | tier1 | 13554 | 2190 | 2177 | 12 | 0.005 | 20404 | 2878 | 2524 | 343 | 0.120 | 16904 | 2791 | 2780 | 10 | 0.004 | 50090 | 6621 | 6605 | 7 | 0.000 | 56541 | 8126 | 8118 | 1 | 0.000 |
| 635308 | 3 | 131217024 | 131217024 | C | G | MRPL3 | missense | c.467 | p.R156P | tier1 | 9976 | 1161 | 1149 | 11 | 0.009 | 5937 | 698 | 603 | 94 | 0.135 | 6987 | 938 | 934 | 1 | 0.000 | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 639691 | 2 | 89161003 | 89161003 | C | G | ENSG00000231486 | intronic | c.NULL | NULL | tier3 | 8446 | 830 | 830 | 0 | 0.000 | 1049 | 105 | 100 | 5 | 0.048 | 5509 | 528 | 522 | 3 | 0.006 | 11048 | 1600 | 1588 | 8 | 0.005 | NA | NA | NA | NA | NA |
| 639691 | 2 | 89185292 | 89185292 | T | C | IGKV4-1 | intronic | c.50-64 | e2-64 | tier3 | 8501 | 799 | 798 | 0 | 0.000 | 2038 | 245 | 211 | 34 | 0.139 | 8231 | 787 | 704 | 80 | 0.102 | 12597 | 2121 | 2090 | 20 | 0.009 | NA | NA | NA | NA | NA |
| 639691 | 2 | 89246918 | 89246918 | G | A | IGKV1-5 | silent | c.252 | p.F84 | tier1 | 2318 | 284 | 284 | 0 | 0.000 | 1207 | 194 | 190 | 4 | 0.021 | 3708 | 464 | 460 | 4 | 0.009 | 5344 | 1224 | 1220 | 4 | 0.003 | NA | NA | NA | NA | NA |
| 639691 | 2 | 89246947 | 89246947 | T | G | IGKV1-5 | missense | c.223 | p.S75R | tier1 | 2294 | 284 | 284 | 0 | 0.000 | 1194 | 193 | 190 | 3 | 0.016 | 3680 | 457 | 452 | 4 | 0.009 | 5349 | 1215 | 1211 | 4 | 0.003 | NA | NA | NA | NA | NA |
| 639691 | 2 | 89246969 | 89246969 | C | G | IGKV1-5 | missense | c.201 | p.K67N | tier1 | 2338 | 290 | 290 | 0 | 0.000 | 1207 | 197 | 193 | 4 | 0.020 | 3690 | 450 | 446 | 4 | 0.009 | 5462 | 1236 | 1232 | 4 | 0.003 | NA | NA | NA | NA | NA |
| 639691 | 2 | 89246970 | 89246970 | T | C | IGKV1-5 | missense | c.200 | p.K67R | tier1 | 2316 | 283 | 283 | 0 | 0.000 | 1191 | 194 | 189 | 4 | 0.021 | 3698 | 463 | 459 | 4 | 0.009 | 5377 | 1209 | 1205 | 3 | 0.002 | NA | NA | NA | NA | NA |
| 639691 | 2 | 89247181 | 89247181 | T | G | IGKV1-5 | intronic | c.55+59 | e1+59 | tier3 | 6057 | 494 | 492 | 1 | 0.002 | 1359 | 163 | 125 | 38 | 0.233 | 4642 | 326 | 265 | 60 | 0.185 | 1645 | 206 | 199 | 5 | 0.025 | NA | NA | NA | NA | NA |
| 639691 | 2 | 89247195 | 89247195 | T | C | IGKV1-5 | intronic | c.55+45 | e1+45 | tier3 | 5051 | 467 | 466 | 1 | 0.002 | 1363 | 161 | 122 | 38 | 0.238 | 4210 | 304 | 251 | 50 | 0.166 | 2805 | 214 | 209 | 5 | 0.023 | NA | NA | NA | NA | NA |
| 639691 | 2 | 89247280 | 89247280 | G | T | IGKV1-5 | silent | c.15 | p.V5 | tier1 | 4549 | 674 | 673 | 1 | 0.001 | 1513 | 279 | 194 | 85 | 0.305 | 3748 | 482 | 376 | 100 | 0.210 | 4527 | 1031 | 1009 | 21 | 0.020 | NA | NA | NA | NA | NA |
| 639691 | 2 | 202436665 | 202436665 | T | G | ALS2CR11 | missense | c.832 | p.T278P | tier1 | 32477 | 3714 | 3705 | 8 | 0.002 | 11831 | 1222 | 874 | 330 | 0.274 | 37336 | 4115 | 3335 | 746 | 0.183 | 50275 | 7051 | 6874 | 139 | 0.020 | NA | NA | NA | NA | NA |
| 639691 | 3 | 38592896 | 38592896 | G | A | SCN5A | missense | c.4967 | p.A1656V | tier1 | 33890 | 3887 | 3884 | 2 | 0.001 | 8817 | 826 | 689 | 135 | 0.164 | 26746 | 2936 | 2472 | 453 | 0.155 | 30002 | 3942 | 2142 | 1781 | 0.454 | NA | NA | NA | NA | NA |
| 639691 | 3 | 193007798 | 193007798 | G | C | ATP13A5 | missense | c.2899 | p.Q967E | tier1 | 6327 | 514 | 511 | 3 | 0.006 | 2429 | 282 | 258 | 22 | 0.079 | 8015 | 657 | 609 | 41 | 0.063 | 8252 | 1270 | 1251 | 8 | 0.006 | NA | NA | NA | NA | NA |
| 639691 | 4 | 100273911 | 100273911 | G | T | ADH1C | 5_prime_untranslated_region | c.-79 | NULL | tier3 | 6054 | 655 | 650 | 1 | 0.002 | 1925 | 187 | 150 | 37 | 0.198 | 6784 | 805 | 724 | 74 | 0.093 | 6972 | 1015 | 1005 | 8 | 0.008 | NA | NA | NA | NA | NA |
| 639691 | 4 | 189013998 | 189013998 | T | C | TRIML2 | 3_prime_untranslated_region | c.*4297 | NULL | tier4 | 21908 | 3323 | 3315 | 7 | 0.002 | 8212 | 1467 | 1077 | 386 | 0.264 | 28026 | 4842 | 3885 | 926 | 0.192 | 34567 | 6431 | 6267 | 148 | 0.023 | NA | NA | NA | NA | NA |
| 639691 | 5 | 38968092 | 38968092 | G | A | RICTOR | missense | c.1013 | p.P338L | tier1 | 3780 | 382 | 382 | 0 | 0.000 | 4111 | 433 | 380 | 53 | 0.122 | 9735 | 1018 | 918 | 98 | 0.096 | 11334 | 1773 | 1309 | 461 | 0.260 | NA | NA | NA | NA | NA |
| 639691 | 6 | 15501389 | 15501389 | C | T | JARID2 | missense | c.2197 | p.R733C | tier1 | 22037 | 2217 | 2204 | 10 | 0.005 | 6584 | 650 | 515 | 131 | 0.203 | 18952 | 1713 | 1468 | 232 | 0.136 | 28697 | 3919 | 3864 | 44 | 0.011 | NA | NA | NA | NA | NA |
| 639691 | 6 | 76343330 | 76343330 | T | C | SENP6 | missense | c.241 | p.S81P | tier1 | 17217 | 1940 | 1936 | 3 | 0.002 | 4979 | 449 | 371 | 78 | 0.174 | 17536 | 2009 | 1704 | 295 | 0.148 | 18203 | 2527 | 1381 | 1125 | 0.449 | NA | NA | NA | NA | NA |
| 639691 | 6 | 119285878 | 119285878 | A | C | FAM184A | missense | c.3092 | p.V1031G | tier1 | 12696 | 1711 | 1706 | 5 | 0.003 | 4883 | 643 | 430 | 211 | 0.329 | 15829 | 2182 | 1756 | 420 | 0.193 | 19208 | 2880 | 2799 | 69 | 0.024 | NA | NA | NA | NA | NA |
| 639691 | 7 | 43484053 | 43484053 | C | A | HECW1 | missense | c.1282 | p.Q428K | tier1 | 10580 | 1259 | 1256 | 3 | 0.002 | 4591 | 606 | 447 | 156 | 0.259 | 11798 | 1306 | 1015 | 274 | 0.213 | 17024 | 2995 | 2914 | 71 | 0.024 | NA | NA | NA | NA | NA |
| 639691 | 8 | 26628125 | 26628125 | G | A | ADRA1A | silent | c.942 | p.L314 | tier1 | 17986 | 1926 | 1923 | 1 | 0.001 | 4704 | 519 | 415 | 103 | 0.199 | 14964 | 1610 | 1385 | 209 | 0.131 | 19764 | 3136 | 3080 | 50 | 0.016 | NA | NA | NA | NA | NA |
| 639691 | 8 | 40012484 | 40012484 | A | C | C8orf4 | 3_prime_untranslated_region | c.*1112 | NULL | tier2 | 8452 | 738 | 734 | 4 | 0.005 | 3364 | 306 | 233 | 72 | 0.236 | 8709 | 710 | 563 | 142 | 0.201 | 7132 | 901 | 882 | 19 | 0.021 | NA | NA | NA | NA | NA |
| 639691 | 8 | 145745801 | 145745801 | A | C | LRRC14 | missense | c.509 | p.D170A | tier1 | 11508 | 1199 | 1198 | 0 | 0.000 | 9150 | 1010 | 694 | 303 | 0.304 | 22730 | 2195 | 1756 | 418 | 0.192 | 34910 | 5021 | 4844 | 126 | 0.025 | NA | NA | NA | NA | NA |
| 639691 | 9 | 13217161 | 13217161 | C | T | MPD3 | intronic | c.1201+18 | e8+18 | tier3 | 886 | 64 | 63 | 1 | 0.016 | 1466 | 138 | 102 | 35 | 0.255 | 3896 | 246 | 190 | 55 | 0.224 | 3612 | 459 | 445 | 10 | 0.022 | NA | NA | NA | NA | NA |
| 639691 | 10 | 124458903 | 124458903 | C | A | C10orf120 | missense | c.202 | p.D68Y | tier1 | 5489 | 713 | 712 | 1 | 0.001 | 2839 | 306 | 232 | 73 | 0.239 | 9234 | 875 | 707 | 162 | 0.186 | 12332 | 1871 | 1824 | 37 | 0.020 | NA | NA | NA | NA | NA |
| 639691 | 11 | 6235593 | 6235593 | T | C | FAM160A2 | intronic | c.2599+48 | e10+48 | tier3 | 9267 | 1190 | 1187 | 3 | 0.003 | 6103 | 689 | 561 | 125 | 0.182 | 11311 | 2560 | 2215 | 335 | 0.131 | 22491 | 3358 | 1809 | 1522 | 0.457 | NA | NA | NA | NA | NA |
| 639691 | 11 | 64794980 | 64794980 | C | T | SNX15 | 5_prime_untranslated_region | c.-30 | NULL | tier2 | 2830 | 244 | 243 | 1 | 0.004 | 1687 | 163 | 131 | 32 | 0.196 | 4498 | 431 | 353 | 75 | 0.175 | 6750 | 943 | 512 | 427 | 0.455 | NA | NA | NA | NA | NA |
| 639691 | 12 | 70349145 | 70349145 | C | G | MYRFL | missense | c.473 | p.S158C | tier1 | 9660 | 807 | 805 | 2 | 0.002 | 3563 | 294 | 217 | 76 | 0.259 | 11360 | 926 | 733 | 184 | 0.201 | 9256 | 1098 | 1063 | 34 | 0.031 | NA | NA | NA | NA | NA |
| 639691 | 12 | 93477504 | 93477504 | C | A | ENSG00000257322 | intronic | c.NULL | NULL | tier4 | 5468 | 462 | 461 | 1 | 0.002 | 2282 | 274 | 227 | 45 | 0.165 | 7489 | 642 | 551 | 85 | 0.134 | 9608 | 1541 | 910 | 617 | 0.404 | NA | NA | NA | NA | NA |
| 639691 | 13 | 31002525 | 31002525 | G | A | UBE2L5P | 3_prime_untranslated_region | c.*31002525 | NULL | tier2 | 201 | 0 | 0 | 0 | 0.000 | 208 | 38 | 29 | 9 | 0.237 | 574 | 34 | 28 | 6 | 0.176 | 809 | 132 | 86 | 46 | 0.348 | NA | NA | NA | NA | NA |
| 639691 | 14 | 20264803 | 20264803 | G | A | ORAN1P | 3_prime_untranslated_region | c.*20264803 | NULL | tier3 | 9350 | 883 | 877 | 5 | 0.006 | 2503 | 276 | 215 | 61 | 0.221 | 8833 | 796 | 689 | 104 | 0.131 | 10978 | 1506 | 1474 | 28 | 0.019 | NA | NA | NA | NA | NA |
| 639691 | 14 | 77844469 | 77844469 | C | A | SAMD15 | silent | c.708 | p.T236 | tier1 | 5020 | 647 | 645 | 1 | 0.002 | 5474 | 651 | 530 | 118 | 0.182 | 16249 | 1728 | 1451 | 262 | 0.153 | 19603 | 3242 | 1742 | 1479 | 0.459 | NA | NA | NA | NA | NA |
| 639691 | 14 | 106329350 | 106329350 | C | G | ENSG00000244620 | 5_prime_flanking_region | c.-26592 | NULL | tier2 | 25170 | 2749 | 2742 | 7 | 0.003 | 6776 | 587 | 415 | 170 | 0.291 | 21423 | 2265 | 1790 | 455 | 0.203 | 27279 | 3490 | 3384 | 103 | 0.030 | NA | NA | NA | NA | NA |
| 639691 | 14 | 106329945 | 106329945 | C | T | ENSG00000244620 | 5_prime_flanking_region | c.-25997 | NULL | tier3 | 7418 | 582 | 580 | 1 | 0.002 | 1641 | 167 | 92 | 75 | 0.449 | 4990 | 365 | 278 | 84 | 0.232 | 9883 | 1275 | 1230 | 34 | 0.027 | NA | NA | NA | NA | NA |
| 639691 | 14 | 106329978 | 106329978 | C | G | ENSG00000244620 | 5_prime_flanking_region | c.-25964 | NULL | tier3 | 7435 | 580 | 579 | 1 | 0.002 | 1712 | 167 | 89 | 78 | 0.467 | 4999 | 363 | 276 | 84 | 0.233 | 10057 | 1265 | 1230 | 32 | 0.025 | NA | NA | NA | NA | NA |
| 639691 | 14 | 106330001 | 106330001 | G | C | ENSG00000244620 | 5_prime_flanking_region | c.-25941 | NULL | tier3 | 7354 | 584 | 582 | 1 | 0.002 | 1678 | 168 | 90 | 78 | 0.464 | 4936 | 366 | 277 | 87 | 0.239 | 9901 | 1278 | 1242 | 33 | 0.026 | NA | NA | NA | NA | NA |
| 639691 | 14 | 107034793 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|--------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 678913 | 16 | 5040788 | 5040788 | C | T | SEC14L5 | silent | c.366 | p.D122 | tier1 | 16442 | 1859 | 1829 | 29 | 0.016 | 19631 | 2677 | 1469 | 1174 | 0.444 | NA | NA | NA | NA | NA | 34069 | 5025 | 5010 | 11 | 0.002 | 45945 | 7001 | 6985 | 10 | 0.001 |
| 678913 | 20 | 30618789 | 30618789 | C | T | LOC101929636 | rna | NULL | NULL | tier1 | 17783 | 2184 | 2152 | 29 | 0.013 | 13197 | 1712 | 1032 | 664 | 0.392 | NA | NA | NA | NA | NA | 32513 | 3978 | 3968 | 6 | 0.002 | 36871 | 5220 | 5211 | 5 | 0.000 |
| 678913 | 21 | 27542825 | 27542825 | G | C | APP | intronic | c.57+57 | e1+57 | tier2 | 13118 | 1435 | 1410 | 24 | 0.017 | 9791 | 1054 | 590 | 454 | 0.435 | NA | NA | NA | NA | NA | 7400 | 923 | 918 | 3 | 0.003 | 21146 | 2738 | 2731 | 4 | 0.001 |
| 715413 | 1 | 201781078 | 201781078 | A | C | NAV1 | missense | c.5000 | p.N1667T | tier1 | 4255 | 598 | 597 | 0 | 0.000 | 7661 | 1306 | 1248 | 54 | 0.041 | NA | NA | NA | NA | NA | 24275 | 3980 | 3971 | 0 | 0.000 | 12702 | 701 | 698 | 0 | 0.000 |
| 715413 | 6 | 75851871 | 75851871 | C | A | COL12A1 | missense | c.4834 | p.V1612L | tier1 | 6174 | 808 | 805 | 2 | 0.002 | 6843 | 883 | 834 | 47 | 0.053 | NA | NA | NA | NA | NA | 22345 | 3156 | 3154 | 0 | 0.000 | 13555 | 735 | 734 | 1 | 0.000 |
| 715413 | 20 | 29612347 | 29612347 | C | A | FRG1B | rna | NULL | NULL | tier1 | 30952 | 3895 | 3542 | 339 | 0.087 | 29846 | 3923 | 3599 | 317 | 0.081 | NA | NA | NA | NA | NA | 106519 | 11051 | 11043 | 6 | 0.000 | 17614 | 523 | 491 | 31 | 0.000 |
| 717045 | 1 | 47548156 | 47548156 | G | A | CYP4Z1 | intronic | c.492+23 | e4+23 | tier3 | 6888 | 1164 | 1151 | 10 | 0.009 | 2447 | 334 | 198 | 135 | 0.405 | NA | NA | NA | NA | NA | 3749 | 37 | 37 | 0 | 0.000 | 2526 | 475 | 472 | 2 | 0.000 |
| 717045 | 1 | 200878061 | 200878061 | G | A | C1orf106 | missense | c.1033 | p.E345K | tier1 | 9569 | 1547 | 1527 | 15 | 0.010 | 4371 | 450 | 302 | 144 | 0.323 | NA | NA | NA | NA | NA | 13009 | 77 | 75 | 1 | 0.000 | 5912 | 879 | 872 | 5 | 0.006 |
| 717045 | 2 | 32666548 | 32666548 | T | G | BIRC6 | intronic | c.3944+18 | e17+18 | tier3 | 2230 | 340 | 333 | 7 | 0.021 | 922 | 124 | 80 | 44 | 0.355 | NA | NA | NA | NA | NA | 1367 | 14 | 14 | 0 | 0.000 | 1245 | 199 | 198 | 1 | 0.000 |
| 717045 | 2 | 179481333 | 179481333 | A | T | TTN | nonsense | c.40481 | p.L13494* | tier1 | 30653 | 4147 | 4095 | 46 | 0.011 | 8277 | 788 | 490 | 292 | 0.373 | NA | NA | NA | NA | NA | 29213 | 144 | 143 | 0 | 0.000 | 17902 | 2359 | 2342 | 9 | 0.004 |
| 717045 | 2 | 198266834 | 198266834 | T | C | SF3B1 | missense | c.2098 | p.K700E | tier1 | 25635 | 3423 | 3343 | 71 | 0.021 | 5493 | 515 | 309 | 199 | 0.392 | NA | NA | NA | NA | NA | 35694 | 201 | 201 | 0 | 0.000 | 15687 | 1931 | 1919 | 11 | 0.006 |
| 717045 | 2 | 217280235 | 217280235 | T | C | SMARCA1 | missense | c.808 | p.Y270H | tier1 | 3481 | 490 | 479 | 11 | 0.022 | 2272 | 190 | 150 | 40 | 0.211 | NA | NA | NA | NA | NA | 6845 | 0 | 0 | 0 | 0.000 | 7963 | 1112 | 1108 | 2 | 0.000 |
| 717045 | 3 | 71355167 | 71355167 | T | C | FOXO1 | intronic | c.1-6136 | e1-6136 | tier2 | 9490 | 1403 | 1390 | 3 | 0.002 | 2813 | 374 | 275 | 98 | 0.263 | NA | NA | NA | NA | NA | 11332 | 58 | 58 | 0 | 0.000 | 6491 | 872 | 869 | 2 | 0.000 |
| 717045 | 4 | 25407313 | 25407313 | G | A | ANAPC4 | intronic | c.1320+69 | e17+69 | tier3 | 3267 | 547 | 539 | 7 | 0.013 | 1934 | 290 | 174 | 114 | 0.396 | NA | NA | NA | NA | NA | 3579 | 0 | 0 | 0 | 0.000 | 1149 | 230 | 228 | 2 | 0.000 |
| 717045 | 4 | 105393360 | 105393360 | C | T | CXXC4 | rna | NULL | NULL | tier1 | 1241 | 95 | 95 | 0 | 0.000 | 675 | 49 | 31 | 18 | 0.367 | NA | NA | NA | NA | NA | 1811 | 49 | 49 | 0 | 0.000 | 923 | 116 | 116 | 0 | 0.000 |
| 717045 | 6 | 129475728 | 129475728 | G | A | LAMA2 | missense | c.1106 | p.R369H | tier1 | 19868 | 2525 | 2480 | 42 | 0.017 | 4971 | 418 | 277 | 139 | 0.334 | NA | NA | NA | NA | NA | 20889 | 117 | 115 | 1 | 0.000 | 11026 | 1492 | 1486 | 4 | 0.003 |
| 717045 | 7 | 73973313 | 73973313 | G | A | GTF2IRD1 | missense | c.2275 | p.G759S | tier1 | 3212 | 534 | 524 | 9 | 0.017 | 2398 | 309 | 216 | 90 | 0.294 | NA | NA | NA | NA | NA | 2015 | 15 | 15 | 0 | 0.000 | 1155 | 199 | 197 | 1 | 0.000 |
| 717045 | 7 | 151878771 | 151878771 | T | C | KMT2C | silent | c.6174 | p.G2058 | tier1 | 14778 | 2969 | 2922 | 46 | 0.015 | 4446 | 667 | 416 | 249 | 0.374 | NA | NA | NA | NA | NA | 15495 | 133 | 133 | 0 | 0.000 | 3741 | 821 | 817 | 4 | 0.005 |
| 717045 | 8 | 9639151 | 9639151 | A | G | TNKS | 3_prime_untranslated_region | c.*4905 | NULL | tier3 | 1352 | 195 | 193 | 2 | 0.010 | 1136 | 134 | 79 | 54 | 0.406 | NA | NA | NA | NA | NA | 1294 | 0 | 0 | 0 | 0.000 | 878 | 130 | 129 | 1 | 0.000 |
| 717045 | 10 | 61666207 | 61666207 | G | A | CCDC6 | 5_prime_untranslated_region | c.-25 | NULL | tier2 | 12004 | 1530 | 1512 | 18 | 0.012 | 3647 | 342 | 220 | 119 | 0.351 | NA | NA | NA | NA | NA | 10798 | 73 | 73 | 0 | 0.000 | 6044 | 846 | 843 | 3 | 0.004 |
| 717045 | 10 | 71620368 | 71620368 | C | T | COL13A1 | intronic | c.365-11564 | e3-11564 | tier3 | 21700 | 2851 | 2794 | 54 | 0.019 | 6809 | 636 | 400 | 234 | 0.369 | NA | NA | NA | NA | NA | 29706 | 154 | 154 | 0 | 0.000 | 10151 | 1246 | 1240 | 5 | 0.004 |
| 717045 | 12 | 49724201 | 49724201 | G | A | TROAP | missense | c.1573 | p.A525T | tier1 | 11571 | 1517 | 1495 | 17 | 0.011 | 3004 | 289 | 205 | 83 | 0.288 | NA | NA | NA | NA | NA | 10363 | 0 | 0 | 0 | 0.000 | 6945 | 886 | 879 | 4 | 0.005 |
| 717045 | 14 | 88945999 | 88945999 | G | A | PTPN21 | silent | c.1776 | p.P592 | tier1 | 9582 | 1363 | 1342 | 16 | 0.012 | 3009 | 377 | 241 | 126 | 0.343 | NA | NA | NA | NA | NA | 6962 | 79 | 79 | 0 | 0.000 | 2622 | 451 | 449 | 2 | 0.000 |
| 717045 | 15 | 101432730 | 101432730 | G | C | ALDH1A3 | missense | c.361 | p.D121H | tier1 | 18641 | 2775 | 2740 | 30 | 0.011 | 4893 | 555 | 413 | 134 | 0.245 | NA | NA | NA | NA | NA | 19272 | 102 | 102 | 0 | 0.000 | 7718 | 1098 | 1096 | 2 | 0.000 |
| 717045 | 16 | 66432084 | 66432084 | G | T | CDH5 | intronic | c.1485+75 | e8+75 | tier3 | 18545 | 2425 | 2406 | 17 | 0.007 | 4073 | 376 | 275 | 98 | 0.263 | NA | NA | NA | NA | NA | 23912 | 112 | 112 | 0 | 0.000 | 10567 | 1319 | 1315 | 4 | 0.003 |
| 717045 | 17 | 40731892 | 40731892 | C | T | FAM134C | 3_prime_untranslated_region | c.*1939 | NULL | tier3 | 5159 | 1170 | 1152 | 18 | 0.015 | 2301 | 331 | 199 | 131 | 0.397 | NA | NA | NA | NA | NA | 2789 | 36 | 36 | 0 | 0.000 | 1440 | 356 | 352 | 3 | 0.008 |
| 717045 | 19 | 1046259 | 1046259 | C | G | ABCA7 | silent | c.1476 | p.P492 | tier1 | 18745 | 2445 | 2423 | 20 | 0.008 | 4267 | 409 | 264 | 142 | 0.350 | NA | NA | NA | NA | NA | 21192 | 99 | 99 | 0 | 0.000 | 8651 | 1076 | 1075 | 1 | 0.000 |
| 717045 | 20 | 39992390 | 39992390 | C | T | EMILIN3 | silent | c.402 | p.T134 | tier1 | 17366 | 3269 | 3215 | 47 | 0.014 | 3282 | 810 | 510 | 300 | 0.370 | NA | NA | NA | NA | NA | 13665 | 0 | 0 | 0 | 0.000 | 3847 | 744 | 741 | 1 | 0.000 |
| 732217 | 1 | 1896486 | 1896486 | G | A | C1orf222 | silent | c.171 | p.D57 | tier1 | 9674 | 1101 | 1083 | 18 | 0.016 | 26093 | 3330 | 1866 | 1416 | 0.431 | 28888 | 3230 | 1802 | 1367 | 0.431 | 23324 | 2863 | 2855 | 3 | 0.001 | NA | NA | NA | NA | NA |
| 732217 | 1 | 19549161 | 19549161 | C | T | EMC1 | missense | c.95 | p.R32K | tier1 | 8174 | 922 | 907 | 13 | 0.014 | 14888 | 1744 | 908 | 819 | 0.474 | 16817 | 1911 | 958 | 1367 | 0.431 | 12097 | 1311 | 1308 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 732217 | 1 | 20518895 | 20518895 | C | T | UBXN10 | 3_prime_untranslated_region | c.*998 | NULL | tier3 | 1142 | 137 | 135 | 2 | 0.015 | 5710 | 700 | 392 | 307 | 0.439 | 6232 | 785 | 412 | 368 | 0.472 | 5373 | 633 | 633 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 732217 | 1 | 46079781 | 46079781 | A | G | NASP | missense | c.1526 | p.N509S | tier1 | 4624 | 421 | 415 | 6 | 0.014 | 7233 | 644 | 316 | 323 | 0.505 | 7093 | 528 | 242 | 280 | 0.536 | 7638 | 680 | 678 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 732217 | 2 | 112588927 | 112588927 | C | G | ANAPC1 | missense | c.2561 | p.G854A | tier1 | 604 | 50 | 46 | 3 | 0.061 | 984 | 146 | 87 | 56 | 0.392 | 1153 | 102 | 54 | 48 | 0.471 | 635 | 78 | 78 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 732217 | 2 | 236949476 | 236949476 | G | A | AGAP1 | missense | c.1882 | p.E628K | tier1 | 18761 | 2182 | 2164 | 13 | 0.006 | 18854 | 2432 | 1563 | 840 | 0.350 | 18896 | 2091 | 1336 | 718 | 0.350 | 15653 | 1820 | 1819 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 732217 | 4 | 106164914 | 106164914 | G | A | TET2 | missense | c.3782 | p.R1261H | tier1 | 5578 | 518 | 493 | 24 | 0.046 | 5690 | 711 | 73 | 626 | 0.896 | 6924 | 565 | 34 | 527 | 0.939 | 8545 | 826 | 822 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 732217 | 4 | 125593542 | 125593542 | A | G | ANKRD50 | missense | c.890 | p.I297T | tier1 | 961 | 92 | 85 | 7 | 0.076 | 1571 | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 754150 | 20 | 3127469 | 3127469 | C | G | FASTKD5 | missense | c.2248 | p.E750Q | tier1 | 8547 | 944 | 912 | 31 | 0.033 | 17344 | 2205 | 1194 | 998 | 0.455 | 10908 | 1081 | 645 | 436 | 0.403 | 30850 | 3383 | 3354 | 27 | 0.008 | 16604 | 1874 | 1515 | 355 | 0.190 |
| 754150 | 20 | 29652147 | 29652147 | A | G | MLLT10P1 | 5_prime_flanking_region | c.14087 | NULL | tier4 | 41376 | 7127 | 6538 | 527 | 0.075 | 18439 | 2659 | 2479 | 170 | 0.064 | 20947 | 4228 | 3918 | 305 | 0.000 | 33089 | 4590 | 3963 | 585 | 0.129 | 2644 | 209 | 191 | 18 | 0.000 |
| 754150 | 20 | 55967777 | 55967777 | G | C | RBM38 | missense | c.305 | p.R102P | tier1 | 15263 | 2080 | 2043 | 32 | 0.015 | 30050 | 3990 | 2450 | 1496 | 0.379 | 12918 | 1305 | 897 | 403 | 0.310 | 30621 | 3631 | 3624 | 3 | 0.001 | 25388 | 2858 | 2188 | 647 | 0.228 |
| 754150 | 22 | 50903351 | 50903351 | C | T | SBF1 | splice_region | c.1333-5 | e13-5 | tier2 | 14749 | 2452 | 2404 | 45 | 0.018 | 13321 | 2725 | 1614 | 1079 | 0.401 | 4423 | 658 | 459 | 195 | 0.298 | 12482 | 1977 | 1961 | 15 | 0.008 | 6304 | 771 | 643 | 123 | 0.161 |
| 754150 | X | 73064091 | 73064091 | A | G | XIST | rna | NULL | NULL | tier1 | 8126 | 1021 | 960 | 61 | 0.060 | 9415 | 1375 | 133 | 1241 | 0.903 | 11277 | 1103 | 323 | 777 | 0.706 | 61253 | 7703 | 7657 | 44 | 0.006 | 21689 | 2666 | 2065 | 586 | 0.221 |
| 754150 | X | 123505288 | 123505288 | T | C | SH2D1A | rna | NULL | NULL | tier1 | 2972 | 378 | 356 | 22 | 0.058 | 2096 | 333 | 32 | 300 | 0.904 | 2654 | 494 | 138 | 355 | 0.720 | 4714 | 610 | 604 | 4 | 0.007 | 8943 | 1195 | 956 | 233 | 0.196 |
| 769516 | 1 | 116947126 | 116947126 | C | G | ATP1A1 | 3_prime_untranslated_region | c.*60 | NULL | tier3 | 4379 | 903 | 898 | 5 | 0.006 | 21673 | 3270 | 2398 | 847 | 0.261 | NA | NA | NA | NA | NA | 9447 | 1255 | 1247 | 5 | 0.004 | 3764 | 301 | 147,147; | 7 | 0.023 |
| 769516 | 1 | 170927719 | 170927719 | G | T | MROH9 | intronic | c.152+38 | e3+38 | tier3 | 6124 | 916 | 916 | 0 | 0.000 | 15862 | 2328 | 2038 | 287 | 0.123 | NA | NA | NA | NA | NA | 11442 | 1559 | 1555 | 2 | 0.000 | 10996 | 1518 | 1499,8; | 8 | 0.005 |
| 769516 | 1 | 200943970 | 200943970 | G | A | KIF21B | silent | c.4686 | p.S1562 | tier1 | 30045 | 4414 | 4382 | 24 | 0.005 | 58382 | 8718 | 6087 | 2524 | 0.293 | NA | NA | NA | NA | NA | 27640 | 4100 | 4055 | 42 | 0.010 | 20505 | 2778 | 332,1934 | 208 | 0.075 |
| 769516 | 3 | 186288815 | 186288815 | A | G | DNAJB11 | intronic | c.68+61 | e1+61 | tier2 | 9068 | 1299 | 86 | 9 | 0.095 | 18749 | 2517 | 340 | 34 | 0.091 | NA | NA | NA | NA | NA | 6400 | 879 | 197 | 3 | 0.000 | 1573 | 168 | 36,29; | 0 | 0.000 |
| 769516 | 4 | 74361009 | 74361009 | C | T | AFM | 3_prime_untranslated_region | c.*74361009 | NULL | tier3 | 20688 | 2713 | 2695 | 5 | 0.002 | 31126 | 3996 | 3348 | 623 | 0.157 | NA | NA | NA | NA | NA | 24465 | 3130 | 3113 | 16 | 0.005 | 21395 | 2757 | 325,139; | 38 | 0.014 |
| 769516 | 4 | 187541903 | 187541903 | C | T | FAT1 | missense | c.5837 | p.S1946N | tier1 | 18839 | 2348 | 2344 | 4 | 0.002 | 31206 | 3969 | 2813 | 1128 | 0.286 | NA | NA | NA | NA | NA | 18065 | 2235 | 2228 | 5 | 0.002 | 12830 | 1496 | 0,1451; | 40 | 0.027 |
| 769516 | 5 | 15936967 | 15936967 | C | T | FBX17 | missense | c.1148 | p.A383V | tier1 | 45492 | 5016 | 5001 | 14 | 0.003 | 111272 | 13453 | 9607 | 3678 | 0.277 | NA | NA | NA | NA | NA | 60333 | 7882 | 7841 | 34 | 0.004 | 40486 | 5549 | 429,201; | 101 | 0.018 |
| 769516 | 5 | 73168759 | 73168759 | T | A | ARHGFE28 | intronic | c.2567-65 | e21-65 | tier3 | 3926 | 763 | 760 | 2 | 0.003 | 12210 | 1939 | 1383 | 544 | 0.282 | NA | NA | NA | NA | NA | 6441 | 913 | 911 | 2 | 0.000 | 3970 | 344 | 72,262; | 10 | 0.029 |
| 769516 | 9 | 100137897 | 100137897 | C | G | CCDC180 | 3_prime_untranslated_region | c.*100137897 | NULL | tier3 | 26103 | 4043 | 4026 | 7 | 0.002 | 69606 | 10464 | 7499 | 2842 | 0.275 | NA | NA | NA | NA | NA | 6046 | 6009 | 30 | 0.000 | 18833 | 2454 | 079,133; | 42 | 0.017 | |
| 769516 | 9 | 127549340 | 127549340 | C | T | OLFML2A | silent | c.177 | p.D59 | tier1 | 11993 | 1888 | 1881 | 6 | 0.003 | 26202 | 3916 | 2880 | 1008 | 0.259 | NA | NA | NA | NA | NA | 14504 | 2032 | 2027 | 4 | 0.002 | 8607 | 1160 | 481,645; | 32 | 0.028 |
| 769516 | 13 | 32783001 | 32783001 | C | T | FRY | nonsense | c.4030 | p.R1344* | tier1 | 29902 | 3840 | 3832 | 7 | 0.002 | 71199 | 9365 | 6966 | 2291 | 0.247 | NA | NA | NA | NA | NA | 31719 | 4091 | 4081 | 8 | 0.002 | 18021 | 2029 | 367,1618 | 41 | 0.020 |
| 769516 | 14 | 59113492 | 59113492 | C | T | DACT1 | silent | p.Y717 | tier1 | 27387 | 3265 | 3250 | 12 | 0.004 | 60077 | 7305 | 5202 | 2043 | 0.282 | NA | NA | NA | NA | NA | 36538 | 4619 | 4600 | 16 | 0.003 | 12268 | 1544 | 1420,86; | 37 | 0.024 | |
| 769516 | 16 | 81208267 | 81208267 | A | C | PKD1L2 | missense | c.604 | p.C202G | tier1 | 626 | 121 | 119 | 2 | 0.017 | 5527 | 830 | 583 | 243 | 0.294 | NA | NA | NA | NA | NA | 1417 | 199 | 198 | 1 | 0.000 | 2 | 0 | 0; | 0 | NA |
| 769516 | 17 | 71361450 | 71361450 | T | C | SDK2 | missense | c.5252 | p.N1751S | tier1 | 13306 | 2032 | 2026 | 5 | 0.002 | 25360 | 3981 | 2888 | 1069 | 0.270 | NA | NA | NA | NA | NA | 13517 | 2099 | 2089 | 7 | 0.003 | 6345 | 767 | 760,0; | 7 | 0.009 |
| 769516 | 17 | 74732959 | 74732959 | G | C | SRSF2 | missense | c.284 | p.P95R | tier1 | 13419 | 2024 | 2016 | 5 | 0.002 | 31580 | 3381 | 2412 | 946 | 0.282 | NA | NA | NA | NA | NA | 17820 | 2431 | 2420 | 10 | 0.004 | 326 | 0 | 0; | 0 | NA |
| 769516 | 18 | 14361468 | 14361468 | G | A | ENSG00000265437 | 5_prime_untranslated_region | c.-14361468 | NULL | tier3 | 21053 | 3364 | 3348 | 9 | 0.003 | 25555 | 4492 | 3223 | 1233 | 0.277 | NA | NA | NA | NA | NA | 9823 | 2009 | 2001 | 5 | 0.002 | 2823 | 282 | 49,225; | 8 | 0.028 |
| 769516 | 22 | 19130308 | 19130308 | G | A | DGCR14 | missense | c.235 | p.R79W | tier1 | 17674 | 2331 | 2318 | 12 | 0.005 | 49652 | 6802 | 4723 | 2011 | 0.299 | NA | NA | NA | NA | NA | 15778 | 2169 | 2139 | 24 | 0.011 | 5436 | 687 | 218,408; | 61 | 0.089 |
| 783437 | 6 | 102516350 | 102516350 | C | T | GRIK2 | silent | c.2691 | p.N897 | tier1 | 23399 | 3242 | 3235 | 1 | 0.000 | 13388 | 1363 | 1337 | 22 | 0.016 | 30048 | 3106 | 3065 | 34 | 0.011 | 29579 | 3249 | 3237 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 783437 | 8 | 143369132 | 143369132 | G | C | TSNARE1 | intronic | c.1291-3286 | e10-3286 | tier3 | 17065 | 2199 | 2194 | 3 | 0.001 | 7403 | 755 | 735 | 20 | 0.026 | 5800 | 6062 | 671 | 11 | 0.016 | 5172 | 631 | 631 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 796518 | 1 | 55611693 | 55611693 | C | T | USP24 | silent | c.1860 | p.E620 | tier1 | 2735 | 306 | 305 | 0 | 0.000 | 2750 | 289 | 208 | 78 | 0.273 | 4172 | 363 | 266 | 95 | 0.263 | 4630 | 544 | 544 | 0 | 0.000 | 4025 | 379 | 378 | 0 | 0.000 |
| 796518 | 2 | 98846643 | 98846643 | G | A | VWA3B | missense | c.2281 | p.V761I | tier1 | 3446 | 0 | 0 | 0 | 0.000 | 3850 | 409 | 336 | 72 | 0.176 | 3596 | 288 | 228 | 59 | 0.206 | 6741 | 798 | 796 | 0 | 0.000 | 6897 | 637 | 636 | 0 | 0.000 |
| 796518 | 2 | 171379066 | 171379066 | T | C | MYO3B | intronic | c.3602+3016 | e30+3016 | tier3 | 11063 | 0 | 0 | 0 | 0.000 | 12509 | 1466 | 1031 | 421 | 0.290 | 14926 | 1522 | 1051 | 463 | 0.306 | 20224 | 2722 | 2718 | 1 | 0.000 | 20329 | 2141 | 2138 | 2 | 0.000 |
| 796518 | 2 | 179346878 | 179346878 | G | A | PLEKHA3 | intronic | c.40+1242 | e1+1242 | tier3 | 3836 | 642 | 642 | 0 | 0.000 | 3467 | 500 | 389 | 107 | 0.216 | 4079 | 512 | 423 | 88 | 0.172 | 6296 | 1174 | 1173 | 0 | 0.000 | 45262 | 659 | 659 | 0 | 0.000 |
| 796518 | 2 | 198266834 | 198266834 | T | C | SF3B1 | missense | c.2098 | p.K700E | tier1 | 18394 | 2435 | 2433 | 0 | 0.000 | 20454 | 2571 | 2410 | 148 | 0.058 | 19730 | 2099 | 1959 | 136 | 0.065 | 30517 | 4012 | 4009 | 0 | 0.000 | 32569 | 4442 | 4432 | 0 | 0.000 |
| 796518 | 2 | 198267483 | 198267483 | C | A | SF3B1 | missense | c.1874 | p.R625L | tier1 | 3722 | 520 | 520 | 0 | 0.000 | 3949 | 538 | 521 | 16 | 0.030 | 4192 | 362 | 359 | 2 | 0.000 | 7985 | 1218 | 1217 | 1 | 0.000 | 5274 | 635 | 635 | 0 | 0.000 |
| 796518 | 4 | 151682922 | 151682922 | T | C | LRBA | intronic | c.5645+13 | e34+13 | tier4 | 674 | 0 | 0 | 0 | 0.000 | 1457 | 161 | 151 | 10 | 0.062 | 889 | 67 | 55 | 12 | 0.179 | 1728 | 185 | 185 | 0 | 0.000 | 1419 | 130 | 130 | 0 | 0.000 |
| 796518 | 4 | 155254236 | 155254236 | C | T | DCHS2 | missense | c.11627 | p.G543S | tier1 | 11394 | 1883 | 1880 | 3 | 0.002 | 13827 | 2246 | 1646 | 579 | 0.260 | 10147 | 1138 | 827 | 296 | 0.264 | 20024 | 3577 | 3572 | 4 | 0.001 | 21124 | 3282 | 3280 | 0 | 0.000 |
| 796518 | 5 | 140202971 | 140202971 | C | T | PCDH45 | silent | c.1611 | p.R537 | tier1 | 1653 | 0 | 0 | 0 | 0.000 | 655 | 122 | 53 | 69 | 0.566 | 601 | 70 | 35 | 35 | 0.500 | 1960 | 305 | 304 | 0 | 0.000 | 2503 | 394 | 394 | 0 | 0.000 |
| 796518 | 6 | 170591580 | 170591580 | G | A | DLL1 | 3_prime_untranslated_region | c.*382 | NULL | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 829970 | 15 | 58007312 | 58007312 | A | T | POLR2M | 3_prime_untranslated_region | c.*435 | NULL | tier3 | 17889 | 2733 | 2733 | 0 | 0.000 | 24527 | 2393 | 2007 | 376 | 0.158 | 10531 | 1419 | 1268 | 148 | 0.105 | 23746 | 2949 | 2946 | 0 | 0.000 | 37343 | 4007 | 4001 | 0 | 0.000 |
| 829970 | 16 | 72829450 | 72829450 | A | G | ZFXH3 | silent | c.7131 | p.P2377 | tier1 | 65842 | 8099 | 8093 | 4 | 0.000 | 220053 | 6568 | 3723 | 2801 | 0.429 | 39904 | 5153 | 2947 | 2181 | 0.425 | 274693 | 12266 | 12261 | 1 | 0.000 | 303829 | 12310 | 12307 | 0 | 0.000 |
| 829970 | 17 | 7577094 | 7577094 | G | A | TP53 | missense | c.844 | p.R282W | tier1 | 42359 | 5489 | 5479 | 7 | 0.001 | 99618 | 3652 | 409 | 3224 | 0.887 | 19432 | 2511 | 601 | 1904 | 0.760 | 208625 | 11018 | 11002 | 4 | 0.000 | 243764 | 11574 | 11565 | 2 | 0.000 |
| 829970 | 19 | 17763404 | 17763404 | A | T | UNC13A | intronic | c.1703+36 | e13+36 | tier3 | 24156 | 3848 | 3844 | 3 | 0.001 | 143446 | 6046 | 3217 | 2799 | 0.465 | 24442 | 3752 | 2115 | 1607 | 0.432 | 144133 | 9806 | 9796 | 2 | 0.000 | 158867 | 10029 | 10021 | 1 | 0.000 |
| 829970 | 19 | 18378165 | 18378165 | T | G | KIAA1683 | missense | c.185 | p.K62T | tier1 | 60614 | 7436 | 7432 | 1 | 0.000 | 351109 | 10301 | 9767 | 504 | 0.049 | 24292 | 6159 | 5891 | 256 | 0.042 | 449565 | 17831 | 17829 | 0 | 0.000 | 430796 | 16176 | 16171 | 0 | 0.000 |
| 829970 | 19 | 39214520 | 39214520 | G | C | ACTN4 | intronic | c.1552-57 | e14-57 | tier3 | 18493 | 3153 | 3143 | 5 | 0.002 | 47830 | 3698 | 2035 | 1649 | 0.448 | 9229 | 1181 | 750 | 424 | 0.361 | 33152 | 4838 | 4813 | 2 | 0.000 | 63944 | 5988 | 5983 | 0 | 0.000 |
| 829970 | 19 | 53793213 | 53793213 | A | T | BIRC8 | missense | c.415 | p.S139T | tier1 | 8821 | 1628 | 1624 | 2 | 0.001 | 45234 | 5091 | 2678 | 2380 | 0.471 | 6860 | 947 | 557 | 387 | 0.410 | 35152 | 5449 | 5446 | 0 | 0.000 | 35809 | 5161 | 5159 | 0 | 0.000 |
| 829970 | 20 | 30785389 | 30785389 | G | C | PLAGL2 | silent | c.357 | p.T119 | tier1 | 9332 | 1375 | 1375 | 0 | 0.000 | 29576 | 1963 | 1330 | 621 | 0.318 | 5178 | 699 | 508 | 190 | 0.272 | 41021 | 3266 | 3262 | 1 | 0.000 | 45205 | 3275 | 3273 | 0 | 0.000 |
| 829970 | 21 | 31655211 | 31655211 | C | G | KRTAP24-1 | missense | c.40 | p.V14L | tier1 | 33433 | 4364 | 4356 | 4 | 0.001 | 119572 | 6297 | 3361 | 2884 | 0.462 | 25184 | 3378 | 2022 | 1344 | 0.399 | 147714 | 9761 | 9751 | 2 | 0.000 | 149189 | 9355 | 9351 | 2 | 0.000 |
| 829970 | 21 | 42694456 | 42694456 | G | A | FAM3B | intronic | c.20-394 | e2-394 | tier4 | 14752 | 2300 | 2298 | 2 | 0.001 | 61745 | 4709 | 2938 | 1752 | 0.374 | 10902 | 1470 | 907 | 558 | 0.381 | 64284 | 7089 | 7086 | 1 | 0.000 | 67722 | 6909 | 6908 | 1 | 0.000 |
| 829970 | 21 | 45877697 | 45877697 | G | A | LRRC3 | 3_prime_untranslated_region | c.*396 | NULL | tier3 | 63833 | 8705 | 8704 | 1 | 0.000 | 232119 | 9581 | 5287 | 4170 | 0.441 | 45194 | 6079 | 3512 | 2501 | 0.416 | 305112 | 15863 | 15856 | 0 | 0.000 | 368306 | 16212 | 16208 | 1 | 0.000 |
| 829970 | 22 | 33255649 | 33255649 | T | C | TIMP3 | 3_prime_untranslated_region | c.*285 | NULL | tier3 | 8345 | 1083 | 1082 | 1 | 0.001 | 15331 | 923 | 561 | 360 | 0.391 | 6165 | 701 | 429 | 270 | 0.386 | 12831 | 0 | 0 | 0 | 0.000 | 21898 | 1865 | 1865 | 0 | 0.000 |
| 839903 | 1 | 82436028 | 82436028 | C | T | LPHN2 | missense | c.2752 | p.H918Y | tier1 | 16021 | 1977 | 1975 | 2 | 0.001 | 10125 | 1338 | 1077 | 254 | 0.191 | 14759 | 1912 | 1860 | 48 | 0.025 | 29834 | 2968 | 2967 | 0 | 0.000 | 35199 | 4572 | 4572 | 0 | 0.000 |
| 839903 | 1 | 115258747 | 115258747 | C | T | NRAS | missense | c.35 | p.G12D | tier1 | 13256 | 1591 | 1589 | 1 | 0.001 | 21386 | 2683 | 2358 | 310 | 0.116 | 19040 | 2396 | 2363 | 24 | 0.010 | 40180 | 4913 | 4910 | 1 | 0.000 | 56196 | 7343 | 7334 | 0 | 0.000 |
| 839903 | 1 | 177927426 | 177927426 | C | T | SEC16B | missense | c.136 | p.A46T | tier1 | 18951 | 2560 | 2556 | 3 | 0.001 | 24301 | 3416 | 1977 | 1587 | 0.349 | 24429 | 3372 | 3108 | 253 | 0.075 | 44549 | 5407 | 5404 | 0 | 0.000 | 44986 | 5858 | 5855 | 0 | 0.000 |
| 839903 | 1 | 222705451 | 222705451 | C | A | HHLPL2 | missense | c.1580 | p.R527L | tier1 | 3662 | 401 | 400 | 0 | 0.000 | 3425 | 403 | 347 | 55 | 0.137 | 5921 | 715 | 702 | 13 | 0.018 | 10791 | 1308 | 1298 | 0 | 0.000 | 16444 | 2203 | 2203 | 0 | 0.000 |
| 839903 | 1 | 223568401 | 223568401 | G | A | C1orf65 | silent | c.1584 | p.K528 | tier1 | 14942 | 1917 | 1912 | 4 | 0.002 | 23633 | 3360 | 1928 | 1405 | 0.422 | 22610 | 3139 | 2936 | 197 | 0.063 | 42509 | 5373 | 5367 | 1 | 0.000 | 44194 | 5949 | 5946 | 0 | 0.000 |
| 839903 | 2 | 26677567 | 26677567 | G | T | DRC1 | missense | c.1972 | p.D658Y | tier1 | 13984 | 1671 | 1664 | 3 | 0.002 | 18947 | 2523 | 1712 | 800 | 0.318 | 23621 | 3115 | 2989 | 124 | 0.040 | 43144 | 4466 | 4464 | 0 | 0.000 | 55025 | 7187 | 7186 | 0 | 0.000 |
| 839903 | 2 | 44203321 | 44203321 | G | A | LRPPRC | missense | c.698 | p.A233V | tier1 | 7918 | 841 | 840 | 0 | 0.000 | 17347 | 2207 | 1995 | 208 | 0.094 | 19424 | 2497 | 2470 | 25 | 0.010 | 35835 | 3621 | 3620 | 0 | 0.000 | 67422 | 8974 | 8969 | 0 | 0.000 |
| 839903 | 2 | 187367384 | 187367384 | G | A | ZC3H4 | intronic | c.534+70 | e5+70 | tier3 | 11850 | 1262 | 1259 | 1 | 0.001 | 8032 | 932 | 834 | 93 | 0.100 | 11241 | 1447 | 1343 | 2 | 0.000 | 17248 | 2132 | 2132 | 0 | 0.000 | 39208 | 5088 | 5087 | 0 | 0.000 |
| 839903 | 3 | 16217151 | 16217151 | G | T | GALNT15 | missense | c.493 | p.A165S | tier1 | 12286 | 1808 | 1798 | 5 | 0.003 | 16378 | 2137 | 1187 | 929 | 0.439 | 20824 | 3448 | 3248 | 190 | 0.055 | 31150 | 3693 | 3690 | 0 | 0.000 | 34060 | 5234 | 5226 | 0 | 0.000 |
| 839903 | 3 | 57447266 | 57447266 | G | A | DNAH12 | nonsense | c.2617 | p.Q873* | tier1 | 3954 | 451 | 449 | 0 | 0.000 | 1921 | 222 | 123 | 97 | 0.441 | 4228 | 548 | 508 | 35 | 0.064 | 3886 | 0 | 0 | 0 | 0.000 | 20142 | 2610 | 2608 | 0 | 0.000 |
| 839903 | 3 | 78680237 | 78680237 | A | G | ROBO1 | missense | c.3626+74 | e24+74 | tier3 | 27223 | 3276 | 3274 | 0 | 0.000 | 19918 | 2479 | 2184 | 284 | 0.115 | 22538 | 2981 | 2942 | 36 | 0.012 | 41854 | 4224 | 4219 | 0 | 0.000 | 85366 | 10709 | 10701 | 0 | 0.000 |
| 839903 | 3 | 130317212 | 130317212 | G | C | COL6A6 | missense | c.4537 | p.A1513P | tier1 | 321 | 45 | 45 | 0 | 0.000 | 1549 | 237 | 134 | 100 | 0.427 | 538 | 59 | 54 | 5 | 0.085 | 2842 | 697 | 697 | 0 | 0.000 | 2 | 0 | 0 | 0 | 0.000 |
| 839903 | 4 | 145050305 | 145050305 | T | A | GYP A | intronic | c.38-8564 | e2-8564 | tier3 | 533 | 0 | 0 | 0 | 0.000 | 617 | 90 | 63 | 27 | 0.300 | 255 | 25 | 25 | 0 | 0.000 | 1781 | 411 | 411 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 839903 | 5 | 101726657 | 101726657 | G | A | SLCO6A1 | intronic | c.1879+28 | e11+28 | tier3 | 1176 | 0 | 0 | 0 | 0.000 | 4934 | 496 | 286 | 207 | 0.420 | 4057 | 366 | 351 | 13 | 0.036 | 10569 | 1566 | 1560 | 0 | 0.000 | 7285 | 800 | 798 | 0 | 0.000 |
| 839903 | 5 | 131326603 | 131326603 | A | G | ACSL6 | missense | c.328 | p.Y110H | tier1 | 6567 | 821 | 814 | 0 | 0.000 | 16117 | 2125 | 1190 | 912 | 0.434 | 22355 | 2975 | 2799 | 170 | 0.057 | 38415 | 4468 | 4456 | 0 | 0.000 | 32138 | 4528 | 4519 | 0 | 0.000 |
| 839903 | 5 | 140203552 | 140203552 | G | T | PCDH45 | missense | c.2192 | p.C731F | tier1 | 8679 | 1086 | 1080 | 1 | 0.001 | 9480 | 1388 | 940 | 442 | 0.320 | 11864 | 1541 | 1484 | 55 | 0.036 | 21627 | 2771 | 2768 | 1 | 0.000 | 29412 | 3784 | 3782 | 0 | 0.000 |
| 839903 | 6 | 30229312 | 30229312 | G | T | HCG17 | intronic | c.NULL | NULL | tier3 | 831 | 125 | 125 | 0 | 0.000 | 1417 | 249 | 155 | 90 | 0.367 | 565 | 72 | 66 | 6 | 0.083 | 2257 | 521 | 521 | 0 | 0.000 | 3 | 0 | 0 | 0 | 0.000 |
| 839903 | 7 | 151082221 | 151082221 | G | A | WDR86 | missense | c.815 | p.T272M | tier1 | 5581 | 1014 | 1012 | 2 | 0.002 | 10832 | 2225 | 1875 | 340 | 0.154 | 7063 | 1288 | 1249 | 38 | 0.030 | 18522 | 3784 | 3781 | 0 | 0.000 | 3403 | 0 | 0 | 0 | 0.000 |
| 839903 | 8 | 1649371 | 1649371 | G | A | DLGAP2 | missense | c.2735 | p.R912H | tier1 | 14210 | 1645 | 1641 | 4 | 0.002 | 21248 | 2891 | 1546 | 1322 | 0.461 | 20485 | 2632 | 2432 | 193 | 0.074 | 38133 | 5051 | 5047 | 1 | 0.000 | 23433 | 3051 | 3048 | 1 | 0.000 |
| 839903 | 8 | 65496059 | 65496059 | C | T | BHLHE22 | 3_prime_untranslated_region | c.*1566 | NULL | tier2 | 7201 | 772 | 770 | 0 | 0.000 | 7932 | 936 | 819 | 110 | 0.118 | 8762 | 1004 | 993 | 8 | 0.008 | 16563 | 2506 | 2500 | 2 | 0.000 | 30797 | 3990 | 3983 | 0 | 0.000 |
| 839903 | 9 | 32624147 | 32624147 | T | A | ENSG0000023440 | 5_prime_flanking_region | c.-9305 | NULL | tier4 | 10999 | 1258 | 1253 | 0 | 0.000 | 6781 | 900 | 747 | 150 | 0.167 | 12571 | 1608 | 1601 | 7 | 0.004 | 23472 | 2945 | 2944 | 0 | 0.000 | 47045 | 6174 | 6173 | 0 | 0.000 |
| 839903 | 10 | 5494360 | 5494360 | G | A | NET1 | missense | c.403 | p.A135T | tier1 | 6161 | 947 | 943 | 2 | 0.002 | 7354 | 1317 | 705 | 602</ | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 848524 | 17 | 40666295 | 40666295 | C | T | MIRS010 | rna | NULL | NULL | tier1 | 15202 | 2089 | 2024 | 64 | 0.031 | 8197 | 876 | 487 | 387 | 0.443 | NA | NA | NA | NA | NA | 53662 | 7044 | 7038 | 0 | 0.000 | 96862 | 11601 | 11587 | 0 | 0.000 |
| 848524 | 17 | 48594960 | 48594960 | C | T | MYCBPAP | missense | c.422 | p.P141L | tier1 | 1871 | 388 | 386 | 2 | 0.005 | 1201 | 248 | 165 | 81 | 0.329 | NA | NA | NA | NA | NA | 3750 | 730 | 730 | 0 | 0.000 | 9 | 0 | 0 | 0 | NA |
| 848524 | 18 | 42531907 | 42531907 | G | A | SETBP1 | missense | c.2602 | p.D868N | tier1 | 33806 | 4405 | 4361 | 43 | 0.010 | 10538 | 1209 | 878 | 327 | 0.271 | NA | NA | NA | NA | NA | 60890 | 8051 | 8045 | 2 | 0.000 | 90832 | 9836 | 9825 | 2 | 0.000 |
| 848524 | 19 | 1043430 | 1043430 | C | G | ABCA7 | silent | c.888 | p.L296 | tier1 | 12979 | 1709 | 1680 | 27 | 0.016 | 3119 | 331 | 174 | 157 | 0.474 | NA | NA | NA | NA | NA | 17315 | 2188 | 2187 | 0 | 0.000 | 12310 | 0 | 0 | 0 | NA |
| 848524 | 19 | 37064363 | 37064363 | A | T | LOC101927599 | rna | NULL | NULL | tier1 | 4280 | 554 | 538 | 16 | 0.029 | 2810 | 339 | 194 | 143 | 0.424 | NA | NA | NA | NA | NA | 13995 | 1821 | 1819 | 0 | 0.000 | 39 | 0 | 0 | 0 | NA |
| 855934 | 1 | 228403429 | 228403429 | G | A | OBSO | missense | c.1994 | p.R665Q | tier1 | 22300 | 2989 | 2893 | 94 | 0.031 | 24643 | 2799 | 1434 | 1323 | 0.480 | 19684 | 2930 | 1552 | 1365 | 0.468 | 57126 | 5792 | 5784 | 2 | 0.000 | 82543 | 5668 | 5646 | 0 | 0.000 |
| 855934 | 2 | 81421390 | 81421390 | G | A | ENSG00000234429 | 3_prime_flanking_region | c.*71 | NULL | tier2 | 9501 | 1286 | 1266 | 20 | 0.016 | 8989 | 1121 | 583 | 530 | 0.476 | 5443 | 997 | 502 | 494 | 0.496 | 15179 | 2898 | 2896 | 1 | 0.000 | 21579 | 2130 | 2112 | 0 | 0.000 |
| 855934 | 2 | 166611398 | 166611398 | G | T | GALNT3 | intronic | c.1524+44 | e7+44 | tier3 | 3312 | 365 | 363 | 0 | 0.000 | 1413 | 122 | 100 | 22 | 0.180 | 2969 | 386 | 314 | 70 | 0.182 | 3645 | 0 | 0 | 0 | NA | 5340 | 544 | 539 | 0 | 0.000 |
| 855934 | 2 | 172231602 | 172231602 | C | T | ENSG00000224553 | 5_prime_untranslated_region | c.-172231602 | NULL | tier3 | 7597 | 1034 | 1034 | 0 | 0.000 | 8960 | 1103 | 626 | 472 | 0.430 | 6232 | 947 | 517 | 430 | 0.454 | 20792 | 2574 | 2573 | 0 | 0.000 | 20542 | 1608 | 1589 | 0 | 0.000 |
| 855934 | 4 | 106157845 | 106157845 | C | T | TET2 | nonsense | c.2746 | p.Q916* | tier1 | 5968 | 737 | 736 | 1 | 0.001 | 8791 | 959 | 682 | 274 | 0.287 | 4649 | 614 | 415 | 197 | 0.322 | 11492 | 1839 | 1839 | 0 | 0.000 | 11340 | 1010 | 1009 | 0 | 0.000 |
| 855934 | 5 | 140783754 | 140783754 | G | A | PCDHGA9 | missense | c.1235 | p.R412Q | tier1 | 23881 | 2830 | 2801 | 23 | 0.008 | 17004 | 1485 | 873 | 598 | 0.407 | 22431 | 3092 | 1773 | 1307 | 0.424 | 41261 | 4457 | 4452 | 1 | 0.000 | 54408 | 4705 | 4655 | 0 | 0.000 |
| 855934 | 6 | 30530949 | 30530949 | C | T | PRR3 | rna | NULL | NULL | tier1 | 2715 | 383 | 378 | 2 | 0.005 | 5694 | 610 | 318 | 289 | 0.476 | 2764 | 446 | 246 | 200 | 0.448 | 10805 | 1511 | 1507 | 0 | 0.000 | 10715 | 964 | 960 | 0 | 0.000 |
| 855934 | 6 | 31637272 | 31637272 | C | T | CSNK2B | nonsense | c.544 | p.Q182* | tier1 | 28782 | 3963 | 3955 | 3 | 0.001 | 32051 | 3923 | 2086 | 1757 | 0.457 | 33542 | 4653 | 2378 | 2245 | 0.486 | 56317 | 6721 | 6709 | 0 | 0.000 | 96178 | 6943 | 6885 | 0 | 0.000 |
| 855934 | 7 | 6439714 | 6439714 | T | C | RAC1 | 3_prime_untranslated_region | c.*6439714 | NULL | tier3 | 21570 | 2662 | 2660 | 2 | 0.001 | 16783 | 1701 | 1139 | 544 | 0.323 | 15399 | 2032 | 1576 | 448 | 0.221 | 50068 | 4599 | 4595 | 0 | 0.000 | 66552 | 4153 | 4147 | 0 | 0.000 |
| 855934 | 7 | 101758501 | 101758501 | C | T | CUX1 | nonsense | c.655 | p.R219* | tier1 | 11794 | 1325 | 1323 | 2 | 0.002 | 8454 | 716 | 383 | 329 | 0.462 | 13207 | 1757 | 908 | 845 | 0.482 | 18061 | 2086 | 2085 | 1 | 0.000 | 30000 | 2631 | 2612 | 0 | 0.000 |
| 855934 | 8 | 139668092 | 139668092 | C | T | COL22A1 | intronic | c.3333+48 | e44+48 | tier3 | 4597 | 599 | 594 | 4 | 0.007 | 5273 | 552 | 299 | 250 | 0.455 | 2443 | 334 | 170 | 162 | 0.488 | 7959 | 1219 | 1217 | 1 | 0.000 | 11226 | 1202 | 1183 | 0 | 0.000 |
| 855934 | 8 | 145773426 | 145773426 | C | T | ARHGAP39 | silent | c.1044 | p.S348 | tier1 | 10243 | 1257 | 1247 | 8 | 0.006 | 16542 | 1656 | 772 | 866 | 0.529 | 12829 | 1766 | 917 | 844 | 0.479 | 35100 | 3956 | 3953 | 0 | 0.000 | 52895 | 4242 | 4215 | 0 | 0.000 |
| 855934 | 10 | 3183033 | 3183033 | A | G | PITRM1 | intronic | c.2649-29 | e24-29 | tier3 | 18710 | 2268 | 2266 | 0 | 0.000 | 13126 | 1132 | 741 | 422 | 0.363 | 20424 | 2819 | 1718 | 1094 | 0.389 | 35437 | 3868 | 3866 | 0 | 0.000 | 40465 | 3273 | 3270 | 0 | 0.000 |
| 855934 | 10 | 43651051 | 43651051 | A | G | CSGALNACT2 | missense | c.454 | p.T152A | tier1 | 12655 | 1579 | 1574 | 5 | 0.003 | 8911 | 947 | 547 | 395 | 0.419 | 19417 | 2578 | 1530 | 1043 | 0.405 | 22682 | 2376 | 2372 | 0 | 0.000 | 29049 | 2253 | 2246 | 0 | 0.000 |
| 855934 | 10 | 71695049 | 71695049 | G | A | COL13A1 | intronic | c.1696-57 | e30-57 | tier3 | 24459 | 463 | 460 | 2 | 0.004 | 5289 | 873 | 489 | 377 | 0.435 | 1146 | 293 | 164 | 129 | 0.440 | 6555 | 1646 | 1643 | 0 | 0.000 | 5556 | 851 | 815 | 0 | 0.000 |
| 855934 | 10 | 104120904 | 104120904 | G | A | GBF1 | rna | NULL | NULL | tier1 | 17610 | 2163 | 2106 | 2 | 0.001 | 17647 | 1664 | 887 | 731 | 0.452 | 13624 | 1935 | 1031 | 888 | 0.463 | 32774 | 3721 | 3689 | 0 | 0.000 | 47533 | 3347 | 3160 | 0 | 0.000 |
| 855934 | 10 | 115528652 | 115528652 | G | A | PLEKHS1 | splice_region | c.417+3 | e5+3 | tier2 | 10392 | 1388 | 1377 | 11 | 0.008 | 10020 | 1043 | 590 | 440 | 0.427 | 8026 | 1167 | 671 | 493 | 0.424 | 21668 | 2847 | 2844 | 0 | 0.000 | 20478 | 1772 | 1763 | 0 | 0.000 |
| 855934 | 12 | 54796846 | 54796846 | G | A | ITGA5 | 5_prime_untranslated_region | c.-54796846 | NULL | tier3 | 13237 | 1766 | 1753 | 10 | 0.006 | 21520 | 1436 | 1280 | 1134 | 0.470 | 25246 | 3351 | 1777 | 1563 | 0.468 | 49193 | 4781 | 4777 | 1 | 0.000 | 87747 | 5500 | 5474 | 0 | 0.000 |
| 855934 | 12 | 70064230 | 70064230 | T | G | BEST3 | missense | c.590 | p.D197A | tier1 | 5724 | 810 | 802 | 8 | 0.010 | 7819 | 895 | 479 | 412 | 0.462 | 5483 | 792 | 430 | 360 | 0.456 | 16909 | 2383 | 2379 | 0 | 0.000 | 28269 | 2139 | 2113 | 0 | 0.000 |
| 855934 | 16 | 84888372 | 84888372 | C | T | CRISPLD2 | missense | c.646 | p.R216W | tier1 | 23506 | 3083 | 3052 | 28 | 0.009 | 28205 | 3122 | 1659 | 1411 | 0.460 | 31179 | 4347 | 2386 | 1941 | 0.449 | 64346 | 7136 | 7129 | 1 | 0.000 | 129438 | 10114 | 10073 | 0 | 0.000 |
| 855934 | 18 | 30253100 | 30253100 | C | T | KLHL14 | 3_prime_untranslated_region | c.*1520 | NULL | tier2 | 2685 | 230 | 226 | 3 | 0.013 | 2275 | 152 | 82 | 70 | 0.461 | 2913 | 320 | 170 | 149 | 0.467 | 3991 | 582 | 580 | 0 | 0.000 | 3466 | 448 | 426 | 0 | 0.000 |
| 855934 | 19 | 43679576 | 43679576 | C | T | PSG5 | missense | c.755 | p.R252H | tier1 | 1478 | 249 | 246 | 2 | 0.008 | 4143 | 697 | 389 | 303 | 0.438 | 669 | 168 | 83 | 84 | 0.503 | 5239 | 1286 | 1282 | 1 | 0.000 | 2985 | 492 | 482 | 0 | 0.000 |
| 855934 | 19 | 57772501 | 57772501 | C | G | ZNF460 | 5_prime_flanking_region | c.-19674 | NULL | tier3 | 13320 | 1487 | 1414 | 69 | 0.047 | 15188 | 1320 | 683 | 620 | 0.476 | 10949 | 1540 | 727 | 799 | 0.524 | 28212 | 3401 | 3399 | 0 | 0.000 | 22066 | 2146 | 2127 | 0 | 0.000 |
| 855934 | 20 | 49199177 | 49199177 | A | C | PTPN1 | intronic | c.1285-52 | e10-52 | tier3 | 7893 | 1359 | 1320 | 35 | 0.026 | 7056 | 978 | 491 | 479 | 0.494 | 4892 | 963 | 513 | 448 | 0.466 | 14097 | 2432 | 2430 | 0 | 0.000 | 15762 | 1949 | 1944 | 0 | 0.000 |
| 855934 | 21 | 44514777 | 44514777 | T | G | U2AF1 | missense | c.470 | p.Q157P | tier1 | 43629 | 4878 | 4825 | 47 | 0.010 | 37753 | 3682 | 1883 | 1742 | 0.481 | 40446 | 5307 | 2738 | 2535 | 0.481 | 92104 | 7022 | 7020 | 0 | 0.000 | 136549 | 7411 | 7406 | 0 | 0.000 |
| 855934 | X | 76888698 | 76888698 | G | A | ATRX | missense | c.5131 | p.P1711S | tier1 | 4150 | 513 | 512 | 0 | 0.000 | 4019 | 394 | 36 | 357 | 0.908 | 3741 | 542 | 29 | 513 | 0.946 | 8281 | 882 | 882 | 0 | 0.000 | 23645 | 1720 | 1700 | 0 | 0.000 |
| 856550 | 1 | 26317279 | 26317279 | G | A | PAFAH2 | missense | c.29 | p.P10L | tier1 | 16035 | 281 | 280 | 0 | 0.000 | 14052 | 1791 | 1006 | 772 | 0.434 | NA | NA | NA | NA | NA | 18225 | 2226 | 2205 | 20 | 0.009 | 26779 | 3401 | 3394 | 4 | 0.001 |
| 856550 | 1 | 32127089 | 32127089 | C | A | COL16A1 | intronic | c.3727-30 | e60-30 | tier3 | 39309 | 589 | 585 | 1 | 0.000 | 27117 | 3296 | 2237 | 1026 | 0.314 | NA | NA | NA | NA | NA | 47593 | 6147 | 6098 | 39 | 0.006 | 69107 | 9132 | 9112 | 11 | 0.001 |
| 856550 | 1 | 115258747 | 115258747 | C | T | NRAS | missense | c.35 | p.G12D | tier1 | 19330 | 438 | 438 | 0 | 0.000 | 23250 | 2588 | 1574 | 983 | 0.384 | NA | NA | NA | NA | NA | 30695 | 3931 | 3910 | 15 | 0.004 | 37678 | 5089 | 5074 | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 876752 | 1 | 207881649 | 207881649 | C | T | CR1L | intronic | c.1414+41 | e10+41 | tier4 | 12376 | 1303 | 1293 | 10 | 0.008 | 10290 | 1023 | 859 | 163 | 0.159 | 20065 | 2617 | 2138 | 472 | 0.181 | 40890 | 4862 | 4856 | 4 | 0.001 | NA | NA | NA | NA | NA |
| 876752 | 1 | 237947039 | 237947039 | C | T | RYR2 | silent | c.12045 | p.N4015 | tier1 | 14136 | 1517 | 1492 | 23 | 0.015 | 10916 | 1151 | 707 | 435 | 0.381 | 21593 | 2863 | 1626 | 1220 | 0.429 | 38543 | 4556 | 4554 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 876752 | 2 | 233899445 | 233899445 | G | A | NEU2 | missense | c.821 | p.G274E | tier1 | 9112 | 875 | 857 | 17 | 0.019 | 12898 | 1291 | 718 | 556 | 0.436 | 21521 | 2810 | 1451 | 1320 | 0.476 | 28854 | 3233 | 3230 | 3 | 0.001 | NA | NA | NA | NA | NA |
| 876752 | 3 | 38621021 | 38621021 | C | T | SCN5A | intronic | c.3229-35 | e17-35 | tier3 | 12076 | 1090 | 1072 | 17 | 0.016 | 20091 | 1904 | 1203 | 673 | 0.359 | 40819 | 4978 | 2975 | 1960 | 0.397 | 67806 | 7261 | 7255 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 876752 | 4 | 82026868 | 82026868 | C | T | PRKG2 | intronic | c.2063+99 | e15+99 | tier3 | 3984 | 382 | 377 | 5 | 0.013 | 3323 | 1203 | 190 | 90 | 0.321 | 7152 | 889 | 499 | 386 | 0.436 | 11741 | 1292 | 1288 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 876752 | 11 | 71373771 | 71373771 | G | A | ENPP7P8 | 5_prime_flanking_region | c.-49015 | NULL | tier3 | 2648 | 257 | 254 | 3 | 0.012 | 2703 | 246 | 142 | 103 | 0.420 | 3880 | 466 | 281 | 182 | 0.393 | 6877 | 829 | 829 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 876752 | 15 | 90631934 | 90631934 | C | T | IDH2 | missense | c.419 | p.R140Q | tier1 | 15949 | 1632 | 1623 | 7 | 0.004 | 19357 | 1889 | 1838 | 47 | 0.025 | 32590 | 4102 | 3874 | 210 | 0.051 | 58015 | 6552 | 6545 | 5 | 0.001 | NA | NA | NA | NA | NA |
| 876752 | 17 | 6703354 | 6703354 | C | G | TEKT1 | missense | c.1249 | p.V417L | tier1 | 20602 | 2173 | 2143 | 29 | 0.013 | 22234 | 2373 | 1464 | 880 | 0.375 | 43341 | 5841 | 3302 | 2473 | 0.428 | 77372 | 9235 | 9226 | 2 | 0.000 | NA | NA | NA | NA | NA |
| 876752 | 18 | 47380016 | 47380016 | G | A | MYO5B | silent | c.4024 | p.L1342 | tier1 | 18885 | 2141 | 2113 | 26 | 0.012 | 27633 | 3101 | 1940 | 1123 | 0.367 | 44704 | 6356 | 3691 | 2602 | 0.413 | 84493 | 10512 | 10501 | 7 | 0.001 | NA | NA | NA | NA | NA |
| 876752 | 19 | 7944301 | 7944301 | G | A | FLJ22184 | 5_prime_flanking_region | c.-5727 | NULL | tier3 | 10051 | 991 | 974 | 16 | 0.016 | 13434 | 1233 | 656 | 569 | 0.464 | 23116 | 2961 | 1442 | 1494 | 0.509 | 33369 | 3719 | 3716 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 876752 | 22 | 21153574 | 21153574 | G | A | PI4KA | splice_region | c.1821-10 | e16-10 | tier3 | 20389 | 2190 | 2152 | 36 | 0.016 | 35600 | 3730 | 2237 | 1429 | 0.390 | 55400 | 7319 | 4134 | 3109 | 0.429 | 95222 | 11027 | 11022 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 1 | 112525116 | 112525116 | T | G | KCND3 | missense | c.233 | p.D78A | tier1 | 15808 | 1730 | 1720 | 10 | 0.006 | 13690 | 1496 | 1073 | 408 | 0.275 | 10699 | 1537 | 1451 | 83 | 0.004 | 22914 | 3036 | 3030 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 1 | 228464970 | 228464970 | G | T | OBSCN | missense | c.6710 | p.C2237F | tier1 | 28759 | 3856 | 3821 | 25 | 0.007 | 34249 | 4299 | 2788 | 1435 | 0.340 | 23115 | 3128 | 3095 | 29 | 0.009 | 60989 | 8328 | 8321 | 6 | 0.001 | NA | NA | NA | NA | NA |
| 882325 | 1 | 228464971 | 228464971 | T | A | OBSCN | nonsense | c.6711 | p.C2237* | tier1 | 28078 | 3859 | 3830 | 26 | 0.007 | 33335 | 4255 | 2712 | 1465 | 0.351 | 22736 | 3116 | 3082 | 27 | 0.009 | 59209 | 8291 | 8280 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 2 | 25457242 | 25457242 | C | T | DNMT3A | missense | c.2645 | p.R882H | tier1 | 14740 | 1599 | 1564 | 30 | 0.019 | 15434 | 1252 | 1058 | 510 | 0.325 | 10194 | 1368 | 1039 | 326 | 0.239 | 43274 | 5239 | 5220 | 10 | 0.002 | NA | NA | NA | NA | NA |
| 882325 | 2 | 209113112 | 209113112 | C | A | IDH1 | 0 | c.395 | p.R132L | tier1 | 14745 | 1776 | 1760 | 7 | 0.004 | 13079 | 1345 | 881 | 451 | 0.339 | 6426 | 918 | 918 | 0 | 0.000 | 32412 | 4049 | 4039 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 2 | 211421474 | 211421474 | C | T | CP51 | missense | c.35 | p.I121 | tier1 | 5578 | 453 | 444 | 8 | 0.018 | 5055 | 420 | 278 | 141 | 0.337 | 3282 | 445 | 417 | 24 | 0.054 | 11043 | 1140 | 1133 | 4 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 2 | 238253632 | 238253632 | T | C | COL6A3 | intronic | c.7163-37 | e34-37 | tier3 | 7767 | 770 | 767 | 2 | 0.003 | 9814 | 852 | 508 | 332 | 0.395 | 5982 | 824 | 762 | 60 | 0.073 | 11970 | 1487 | 1484 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 4 | 170913470 | 170913470 | G | A | MFAP3L | splice_region | c.299-10 | e2-10 | tier3 | 12788 | 2049 | 2037 | 9 | 0.004 | 28959 | 3383 | 2143 | 1198 | 0.359 | 21112 | 3168 | 3119 | 45 | 0.014 | 51199 | 7071 | 7062 | 8 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 5 | 150923971 | 150923971 | A | G | FAT2 | silent | c.6717 | p.H2239 | tier1 | 27691 | 3126 | 3092 | 27 | 0.009 | 23836 | 2373 | 1519 | 910 | 0.375 | 19691 | 2653 | 2649 | 0 | 0.000 | 37293 | 4773 | 4766 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 8 | 54871008 | 54871008 | T | C | RG520 | missense | c.1157 | p.I386T | tier1 | 733 | 63 | 63 | 0 | 0.000 | 2216 | 177 | 113 | 63 | 0.358 | 1124 | 126 | 125 | 0 | 0.000 | 3504 | 279 | 279 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 9 | 139910760 | 139910760 | C | T | ABCA2 | missense | c.1136 | p.R379Q | tier1 | 18561 | 2153 | 2125 | 23 | 0.011 | 33358 | 3630 | 2244 | 1330 | 0.372 | 23310 | 3091 | 2947 | 138 | 0.045 | 73115 | 9243 | 9222 | 8 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 11 | 56237381 | 56237381 | A | G | OR5M3 | missense | c.593 | p.I198T | tier1 | 15520 | 1706 | 1693 | 11 | 0.006 | 12600 | 1299 | 818 | 470 | 0.365 | 13631 | 1794 | 1783 | 10 | 0.006 | 34812 | 4280 | 4278 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 11 | 64973991 | 64973991 | C | G | CAPN1 | missense | c.1411 | p.R471G | tier1 | 11183 | 1922 | 1900 | 19 | 0.010 | 19232 | 2256 | 1386 | 846 | 0.379 | 9760 | 1801 | 1716 | 81 | 0.045 | 35681 | 5415 | 5401 | 10 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 11 | 89405200 | 89405200 | C | A | FOLH1B | intronic | NULL | NULL | tier3 | 628 | 70 | 69 | 0 | 0.000 | 3671 | 315 | 261 | 52 | 0.166 | 3550 | 473 | 473 | 0 | 0.000 | 11020 | 1296 | 1294 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 12 | 21051353 | 21051353 | C | A | SLCO1B3 | intronic | c.1683-17 | e12-17 | tier2 | 5598 | 552 | 549 | 1 | 0.002 | 4182 | 404 | 268 | 135 | 0.335 | 5034 | 660 | 658 | 1 | 0.000 | 7046 | 829 | 828 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 12 | 11165648 | 11165648 | G | A | CUX2 | intronic | c.175-46 | e3-46 | tier3 | 17172 | 2192 | 2189 | 1 | 0.000 | 14704 | 1479 | 1078 | 391 | 0.266 | 11151 | 1778 | 1769 | 3 | 0.000 | 37860 | 5161 | 5152 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 15 | 33833126 | 33833126 | G | T | RYR3 | intronic | c.646+35 | e7+35 | tier3 | 10029 | 1150 | 1144 | 3 | 0.003 | 17877 | 2029 | 1565 | 446 | 0.222 | 17532 | 2516 | 2513 | 3 | 0.000 | 42068 | 5482 | 5468 | 3 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 17 | 74732959 | 74732959 | G | A | SRSF2 | missense | c.284 | p.P95L | tier1 | 4807 | 550 | 548 | 2 | 0.004 | 12515 | 1282 | 949 | 327 | 0.256 | 5526 | 932 | 931 | 1 | 0.000 | 27616 | 3520 | 3520 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 18 | 55996142 | 55996142 | G | A | NEDD4L | intronic | c.681-85 | e10-85 | tier3 | 27109 | 3159 | 3142 | 12 | 0.004 | 21948 | 2458 | 1635 | 785 | 0.324 | 22237 | 3121 | 3115 | 1 | 0.000 | 51693 | 6787 | 6775 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | 19 | 2435136 | 2435136 | G | A | LMNB2 | missense | c.718 | p.R240C | tier1 | 4710 | 463 | 457 | 6 | 0.013 | 10972 | 1161 | 671 | 474 | 0.414 | 5908 | 819 | 763 | 51 | 0.063 | 14607 | 1951 | 1945 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | X | 50350816 | 50350816 | C | G | SHROOM4 | missense | c.3326 | p.W1109S | tier1 | 63 | 10 | 9 | 1 | 0.100 | 600 | 86 | 21 | 65 | 0.756 | 301 | 74 | 27 | 47 | 0.635 | 623 | 108 | 107 | 1 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | X | 53441798 | 53441798 | A | G | SMC1A | missense | c.320 | p.I107T | tier1 | 14554 | 1637 | 1636 | 1 | 0.001 | 16035 | 1652 | 1478 | 166 | 0.101 | 14576 | 2151 | 2149 | 0 | 0.000 | 41344 | 5491 | 5490 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 882325 | X | 77911875 | 77911875 | T | C | ZCCHC5 | 3_prime_untranslated_region | c.*615 | NULL | tier2 | 8704 | 1085 | 1068 | 16 | 0.015 | 6620 | 749 | 213 | 535 | 0.715 | 7899 | 1036 | 1033 | 2 | 0.000 | 16660 | 2142 | 2141 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 907471 | 1 | 237947546 | 237947546 | C | T | RYR2 | silent | c.12552 | p.N4184 | tier1 | 6362 | 834 | 832 | 0 | 0.000 | 12673 | 1284 | 1225 | 48 | 0.038 | NA | NA | NA | NA | NA | 21648 | 2744 | 2739 | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 918051 | 19 | 39869235 | 39869235 | G | A | SAMD4B | splice_region | c.1530+6 | e7+6 | tier3 | 6187 | 876 | 875 | 0 | 0.000 | 4900 | 620 | 409 | 205 | 0.334 | 2161 | 309 | 231 | 77 | 0.250 | 3344 | 456 | 455 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 918051 | 20 | 1920551 | 1920551 | T | C | LOC727993 | 5_prime_flanking_region | c.-7341 | NULL | tier2 | 5027 | 531 | 531 | 0 | 0.000 | 3216 | 295 | 177 | 117 | 0.398 | 2798 | 245 | 173 | 71 | 0.291 | 5984 | 471 | 470 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 918051 | 20 | 34022502 | 34022502 | G | A | GDF5 | silent | c.711 | p.A237 | tier1 | 1807 | 321 | 321 | 0 | 0.000 | 1676 | 223 | 172 | 51 | 0.229 | 996 | 131 | 107 | 23 | 0.177 | 1868 | 227 | 226 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 918051 | 20 | 44979120 | 44979120 | G | C | SLC35C2 | silent | c.1098 | p.P366 | tier1 | 15851 | 2109 | 2102 | 3 | 0.001 | 16967 | 1857 | 1053 | 780 | 0.426 | 19518 | 2020 | 1321 | 676 | 0.339 | 22750 | 2141 | 2140 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 922185 | 2 | 20100829 | 20100829 | T | A | TTC32 | silent | c.54 | p.S18 | tier1 | 11579 | 1137 | 1128 | 9 | 0.008 | 9937 | 1117 | 898 | 213 | 0.192 | 19306 | 2189 | 1918 | 261 | 0.120 | 28612 | 3370 | 3364 | 0 | 0.000 | 55171 | 6624 | 6615 | 0 | 0.000 |
| 922185 | 2 | 179638759 | 179638759 | A | G | TTN | missense | c.7136 | p.V2379A | tier1 | 6889 | 676 | 663 | 9 | 0.013 | 8345 | 942 | 877 | 61 | 0.065 | 11713 | 1237 | 1150 | 82 | 0.067 | 17793 | 1942 | 1932 | 0 | 0.000 | 21615 | 2958 | 2956 | 0 | 0.000 |
| 922185 | 4 | 71463976 | 71463976 | C | T | AMBN | intronic | c.136-97 | e4-97 | tier3 | 2122 | 199 | 194 | 5 | 0.025 | 2936 | 400 | 339 | 57 | 0.144 | 3839 | 399 | 355 | 44 | 0.110 | 6701 | 928 | 928 | 0 | 0.000 | 4196 | 0 | 0 | 0 | 0.000 |
| 922185 | 4 | 100544120 | 100544120 | T | A | MTTP | 3_prime_untranslated_region | c.*115 | NULL | tier3 | 14402 | 1241 | 1225 | 14 | 0.011 | 12855 | 1228 | 1031 | 192 | 0.157 | 20965 | 2042 | 1803 | 226 | 0.111 | 31808 | 3270 | 3269 | 0 | 0.000 | 56375 | 7374 | 7357 | 0 | 0.000 |
| 922185 | 5 | 82817833 | 82817833 | C | T | VCAN | silent | c.3708 | p.I1236 | tier1 | 1830 | 234 | 231 | 3 | 0.013 | 3563 | 662 | 641 | 21 | 0.032 | 3284 | 587 | 562 | 23 | 0.039 | 6046 | 1038 | 1038 | 0 | 0.000 | 832 | 0 | 0 | 0 | 0.000 |
| 922185 | 5 | 126753323 | 126753323 | C | T | MEGF10 | splice_region | c.1131-7 | e9-7 | tier3 | 16710 | 1905 | 1877 | 24 | 0.013 | 21308 | 2854 | 2236 | 593 | 0.210 | 35477 | 4834 | 3994 | 801 | 0.167 | 46324 | 6261 | 6257 | 0 | 0.000 | 52952 | 7682 | 7674 | 0 | 0.000 |
| 922185 | 5 | 137298188 | 137298188 | T | C | FAM13B | intronic | c.1180-30 | e9-30 | tier3 | 18640 | 1982 | 1954 | 25 | 0.013 | 20216 | 2558 | 1974 | 564 | 0.222 | 32330 | 4085 | 3430 | 622 | 0.154 | 48280 | 6335 | 6330 | 0 | 0.000 | 60373 | 7832 | 7832 | 0 | 0.000 |
| 922185 | 7 | 55241540 | 55241540 | G | T | EGFR | intronic | c.2062-74 | e18-74 | tier3 | 14771 | 1690 | 1668 | 13 | 0.008 | 17884 | 2294 | 2183 | 109 | 0.048 | 26533 | 3419 | 3199 | 204 | 0.060 | 40671 | 5321 | 5314 | 1 | 0.000 | 39964 | 4525 | 4507 | 0 | 0.000 |
| 922185 | 9 | 111954473 | 111954473 | A | T | EPB41L4B | intronic | c.2301+85 | e22+85 | tier3 | 1765 | 168 | 165 | 3 | 0.018 | 4244 | 648 | 491 | 157 | 0.242 | 3941 | 521 | 396 | 124 | 0.238 | 6496 | 927 | 926 | 0 | 0.000 | 45 | 0 | 0 | 0 | 0.000 |
| 922185 | 10 | 126678193 | 126678193 | A | G | CTBP2 | missense | c.2852 | p.I951T | tier1 | 1417 | 158 | 157 | 1 | 0.006 | 2593 | 399 | 310 | 88 | 0.221 | 2973 | 423 | 341 | 82 | 0.194 | 3829 | 571 | 571 | 0 | 0.000 | 189 | 0 | 0 | 0 | 0.000 |
| 922185 | 11 | 57970725 | 57970725 | A | C | OR1S2 | missense | c.929 | p.M310R | tier1 | 8713 | 826 | 819 | 6 | 0.007 | 11552 | 1357 | 1091 | 264 | 0.195 | 18503 | 2051 | 1714 | 334 | 0.163 | 26136 | 3122 | 3120 | 0 | 0.000 | 38939 | 5766 | 5763 | 0 | 0.000 |
| 922185 | 11 | 117340626 | 117340626 | C | T | DSCAML1 | silent | c.3204 | p.K1068 | tier1 | 37155 | 4197 | 4166 | 18 | 0.004 | 46284 | 5756 | 4595 | 1093 | 0.192 | 76183 | 9897 | 8462 | 1339 | 0.137 | 118577 | 15253 | 15249 | 0 | 0.000 | 125825 | 14400 | 14392 | 2 | 0.000 |
| 922185 | 12 | 50026453 | 50026453 | G | T | PRPF40B | 3_prime_untranslated_region | c.*50026453 | NULL | tier3 | 6993 | 654 | 645 | 7 | 0.011 | 9349 | 1135 | 851 | 271 | 0.242 | 15098 | 1631 | 1344 | 275 | 0.170 | 18410 | 2035 | 2032 | 0 | 0.000 | 8904 | 1201 | 1200 | 0 | 0.000 |
| 922185 | 14 | 21968662 | 21968662 | A | G | METTL3 | missense | c.13709 | p.F427L | tier1 | 13707 | 1347 | 1329 | 15 | 0.011 | 13491 | 1612 | 1209 | 384 | 0.241 | 20059 | 2359 | 1828 | 513 | 0.219 | 28170 | 3281 | 3278 | 0 | 0.000 | 33906 | 4179 | 4169 | 0 | 0.000 |
| 922185 | 14 | 93704243 | 93704243 | T | C | BTBD7 | 3_prime_untranslated_region | c.*4376 | NULL | tier3 | 7080 | 722 | 718 | 4 | 0.006 | 7552 | 818 | 612 | 202 | 0.248 | 11852 | 1332 | 1115 | 211 | 0.159 | 16197 | 1803 | 1800 | 0 | 0.000 | 25317 | 3263 | 3261 | 0 | 0.000 |
| 922185 | 15 | 34519872 | 34519872 | A | C | EMC4 | 3_prime_untranslated_region | c.*34519872 | NULL | tier3 | 11316 | 1104 | 1095 | 8 | 0.007 | 15074 | 1648 | 1295 | 346 | 0.210 | 35477 | 4834 | 3994 | 801 | 0.167 | 46324 | 6261 | 6257 | 0 | 0.000 | 40071 | 5114 | 5103 | 0 | 0.000 |
| 922185 | 15 | 89386622 | 89386622 | C | T | ACAN | missense | c.794 | p.T265I | tier1 | 16654 | 1641 | 1620 | 19 | 0.012 | 19660 | 2404 | 1912 | 472 | 0.198 | 25186 | 2823 | 2371 | 423 | 0.151 | 40121 | 4902 | 4900 | 0 | 0.000 | 24487 | 2915 | 2914 | 0 | 0.000 |
| 922185 | 17 | 15457016 | 15457016 | C | A | TVP23C | missense | c.223 | p.D75Y | tier1 | 2110 | 109 | 108 | 1 | 0.009 | 2842 | 282 | 210 | 72 | 0.255 | 4169 | 290 | 243 | 41 | 0.144 | 6808 | 646 | 645 | 0 | 0.000 | 52 | 0 | 0 | 0 | 0.000 |
| 922185 | 17 | 48668000 | 48668000 | G | A | CACNA1G | intronic | c.2453+17 | e10+17 | tier3 | 14549 | 1571 | 1565 | 3 | 0.002 | 14559 | 1861 | 1800 | 51 | 0.028 | 25997 | 3267 | 3138 | 123 | 0.038 | 35281 | 4438 | 4436 | 1 | 0.000 | 41143 | 5542 | 5526 | 0 | 0.000 |
| 922185 | 18 | 19781636 | 19781636 | A | T | GATA6 | 3_prime_untranslated_region | c.*850 | NULL | tier2 | 2117 | 193 | 193 | 0 | 0.000 | 3671 | 420 | 336 | 84 | 0.200 | 5949 | 643 | 551 | 92 | 0.143 | 8277 | 1001 | 1000 | 0 | 0.000 | 16122 | 2031 | 2029 | 0 | 0.000 |
| 922185 | 18 | 21140392 | 21140392 | G | A | NPCL1 | silent | p.D228 | tier1 | 9686 | 1881 | 1866 | 15 | 0.008 | 12398 | 2384 | 2222 | 158 | 0.066 | 16282 | 3122 | 2822 | 292 | 0.094 | 26447 | 5240 | 5238 | 1 | 0.000 | 38363 | 6161 | 6157 | 0 | 0.000 | |
| 933226 | 1 | 45798446 | 45798446 | C | T | MUTYH | missense | c.565 | p.G189R | tier1 | 20382 | 2781 | 2778 | 1 | 0.000 | NA | NA | NA | NA | NA | 21975 | 583 | 419 | 164 | 0.281 | 7614 | 768 | 768 | 0 | 0.000 | 32898 | 4250 | 4249 | 1 | 0.000 |
| 933226 | 2 | 228512849 | 228512849 | G | A | ENSG00000250599 | 5_prime_untranslated_region | c.-228512849 | NULL | tier3 | 21594 | 3322 | 3316 | 3 | 0.001 | NA | NA | NA | NA | NA | 16075 | 434 | 302 | 131 | 0.303 | 5776 | 653 | 651 | 1 | 0.000 | 24762 | 3460 | 3451 | 6 | 0.002 |
| 933226 | 2 | 241976165 | 241976165 | G | A | SNED1 | intronic | c.806-42 | e5-42 | tier2 | 15789 | 2591 | 2590 | 1 | 0.000 | NA | NA | NA | NA | NA | 7177 | 202 | 110 | 92 | 0.455 | 3663 | 434 | 432 | 1 | 0.000 | 14457 | 1932 | 1928 | 1 | 0.000 |
| 933226 | 2 | 242755726 | 242755726 | G | T | NEU4 | silent | c.84 | p.R28 | tier1 | 17300 | 2427 | 2423 | 3 | 0.001 | NA | NA | NA | NA | NA | 11078 | 306 | 222 | 84 | 0.275 | 4801 | 445 | 443 | 2 | 0.000 | 22155 | 2880 | 2872 | 5 | 0.002 |
| 933226 | 3 | 11303055 | 11303055 | C | T | HRH1 | 3_prime_untranslated_region | c.*868 | NULL | tier4 | 12616 | 2006 | 2003 | 2 | 0.001 | NA | NA | NA | NA | NA | 6500 | 194 | 124 | 69 | 0.358 | 3807 | 432 | 432 | 0 | 0.000 | 19516 | 2753 | 2747 | 4 | 0.001 |
| 933226 | 3 | 142546497 | 142546497 | G | A | ENSG00000240950 | 5_prime_untranslated_region | c.-142546497 | NULL | tier3 | 6013 | 923 | 922 | 1 | 0.001 | NA | NA | NA | NA | NA | 1665 | 57 | 32 | 25 | 0.439 | 1235 | 141 | 140 | 1 | 0.000 | 5351 | 832 | 829 | 1 | 0.000 |
| 933226 | 4 | 665776 | 665776 | G | T | PCGF3 | 5_prime_flanking_region | c.-61694 | NULL | tier3 | 15308 | 2598 | 2594 | 1 | 0.000 | NA | NA | NA | NA | NA | 8845 | 300 | 184 | 116 | 0.387 | 3924 | 0 | 0 | 0 | 0.000 | 14810 | 2200 | 2199 | 1 | 0.001 |
| 933226 | 4 | 7741871 | 7741871 | C | T | SORCS2 | intronic | c.3416-56 | e27-56 | tier3 | 23134 | 2962 | 2960 | 1 | 0.000 | NA | NA | NA | NA | NA | 18441 | 425 | 252 | 172 | 0.406 | 7996 | 939 | 935 | 3 | 0.003 | 28710 | 3476 | 3469 | 5 | 0.001 |
| 933226 | 4 | 55161723 | 55161723 | G | A | PDGFRA | 3_prime_untranslated_region | c.*284 | NULL | tier3 | 7931 | 1253 | 1253 | 0 | 0.000 | NA | NA | NA | NA | NA | 2852 | 103 | 82 | 21 | 0.204 | 1529 | 169 | 168 | 1 | 0.000 | 8389 | 1228 | 1225 | 1 | 0.000 |
| 933226 | 4 | 187628190 | 187628190 | T | A | FAT1 | missense | c.2792 | p.N | | | | | | | | | | | | | | | | | | | | | | | | | | |

| UPN | Chr | Start | Stop | Reference | Variant | Gene Symbol | Mutation Type | Coding Position | Amino Acid Change | Tier | Normal Total Count | Normal Unique Count* | Normal Ref Count | Normal Var Count | Normal VAF | Initial Total Count | Initial Unique Count* | Initial Ref Count | Initial Var Count | Initial VAF | Pre Total Count | Pre Unique Count* | Pre Ref Count | Pre Var Count | Pre VAF | D30 Total Count | D30 Unique Count* | D30 Ref Count | D30 Var Count | D30 VAF | D100 Total Count | D100 Unique Count* | D100 Ref Count | D100 Var Count | D100 VAF |
|--------|-----|-----------|-----------|-----------|---------|-----------------|-----------------------------|-----------------|-------------------|-------|--------------------|----------------------|------------------|------------------|------------|---------------------|-----------------------|-------------------|-------------------|-------------|-----------------|-------------------|---------------|---------------|---------|-----------------|-------------------|---------------|---------------|---------|------------------|--------------------|----------------|----------------|----------|
| 944541 | 18 | 43684347 | 43684347 | A | C | HAUS1 | 5_prime_untranslated_region | c.-31 | NULL | tier2 | 11027 | 1328 | 1312 | 12 | 0.009 | 13788 | 1600 | 957 | 628 | 0.396 | NA | NA | NA | NA | NA | 33591 | 3482 | 3479 | 0 | 0.000 | 14203 | 1323 | 1322 | 0 | 0.000 |
| 944541 | 19 | 7964800 | 7964800 | C | T | LRRCE8 | missense | c.1393 | p.L465F | tier1 | 5974 | 938 | 931 | 5 | 0.005 | 7353 | 986 | 639 | 337 | 0.345 | NA | NA | NA | NA | NA | 20434 | 2639 | 2637 | 0 | 0.000 | 8027 | 502 | 502 | 0 | 0.000 |
| 944541 | 19 | 10812636 | 10812636 | T | C | QTRT1 | missense | c.254 | p.L85P | tier1 | 23240 | 2829 | 2816 | 11 | 0.004 | 20901 | 2251 | 1505 | 713 | 0.321 | NA | NA | NA | NA | NA | 6392 | 6388 | 0 | 0.000 | 30292 | 2159 | 2159 | 0 | 0.000 | |
| 944541 | 22 | 22842205 | 22842205 | G | C | ZNF280B | missense | c.1519 | p.L507V | tier1 | 8650 | 1098 | 1086 | 9 | 0.008 | 8992 | 942 | 602 | 333 | 0.356 | NA | NA | NA | NA | NA | 23496 | 2451 | 2441 | 1 | 0.000 | 11831 | 980 | 978 | 0 | 0.000 |
| 955252 | 5 | 140563408 | 140563408 | A | G | PCDHB16 | missense | c.1274 | p.D425G | tier1 | 19016 | 1999 | 1995 | 1 | 0.001 | 30781 | 3400 | 3308 | 86 | 0.025 | 20176 | 2048 | 1993 | 49 | 0.024 | 53519 | 6240 | 6236 | 0 | 0.000 | 69767 | 8440 | 8439 | 0 | 0.000 |
| 988638 | 1 | 46505876 | 46505876 | G | A | PIK3R3 | 3_prime_untranslated_region | c.*3469 | NULL | tier3 | 33975 | 4464 | 4459 | 3 | 0.001 | 16212 | 1664 | 1440 | 219 | 0.132 | 19080 | 2465 | 1453 | 1004 | 0.409 | 47391 | 6016 | 6009 | 5 | 0.001 | 74351 | 6783 | 6772 | 7 | 0.001 |
| 988638 | 1 | 161519606 | 161519606 | A | G | FCGR3A | missense | c.29 | p.F10S | tier1 | 9443 | 1261 | 1248 | 13 | 0.010 | 4771 | 514 | 392 | 118 | 0.231 | 11339 | 1462 | 834 | 624 | 0.428 | 13636 | 1782 | 1780 | 2 | 0.000 | 13208 | 1274 | 1273 | 0 | 0.000 |
| 988638 | 1 | 233091444 | 233091444 | G | A | NTPCR | missense | c.176 | p.R59Q | tier1 | 217 | 0 | 0 | 0 | 0.000 | 301 | 26 | 24 | 2 | 0.077 | 194 | 29 | 17 | 12 | 0.414 | 1262 | 114 | 114 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 988638 | 2 | 102484410 | 102484410 | C | T | MAP4K4 | rna | NULL | NULL | tier1 | 23756 | 3338 | 3337 | 1 | 0.000 | 12674 | 1483 | 1297 | 181 | 0.122 | 19108 | 2545 | 1527 | 1011 | 0.398 | 36793 | 5089 | 5084 | 4 | 0.001 | 27712 | 2853 | 2848 | 5 | 0.002 |
| 988638 | 2 | 128387436 | 128387436 | A | G | MYO7B | intronic | c.4728+35 | e33+35 | tier3 | 45360 | 6579 | 6575 | 2 | 0.000 | 29056 | 3277 | 2815 | 439 | 0.135 | 49505 | 6799 | 4037 | 2721 | 0.403 | 81115 | 11019 | 11011 | 4 | 0.000 | 89255 | 8408 | 8388 | 11 | 0.001 |
| 988638 | 2 | 135957920 | 135957920 | T | C | ZRANB3 | missense | c.3232 | p.K1078E | tier1 | 9963 | 1257 | 1240 | 15 | 0.012 | 6748 | 744 | 590 | 150 | 0.203 | 7880 | 1143 | 676 | 467 | 0.409 | 21453 | 2846 | 2841 | 3 | 0.001 | 22076 | 2352 | 2344 | 2 | 0.000 |
| 988638 | 2 | 192257750 | 192257750 | G | C | MYO1B | intronic | c.2077-49 | e19-49 | tier3 | 10356 | 1367 | 1363 | 0 | 0.000 | 4502 | 481 | 456 | 25 | 0.052 | 4538 | 710 | 505 | 204 | 0.288 | 15276 | 2111 | 2110 | 0 | 0.000 | 26840 | 2439 | 2437 | 1 | 0.000 |
| 988638 | 2 | 202467998 | 202467998 | C | A | ALS2CR11 | missense | c.319 | p.V107F | tier1 | 26814 | 3488 | 3453 | 34 | 0.010 | 17074 | 1750 | 1346 | 395 | 0.227 | 21636 | 2900 | 1679 | 1206 | 0.411 | 53474 | 6809 | 6800 | 3 | 0.000 | 90453 | 8383 | 8370 | 9 | 0.001 |
| 988638 | 3 | 55061774 | 55061774 | C | T | CACNA2D3 | intronic | c.2987+9430 | e35+9430 | tier2 | 8406 | 1406 | 1405 | 1 | 0.001 | 11215 | 1343 | 1154 | 181 | 0.136 | 17279 | 2872 | 1659 | 1198 | 0.419 | 36744 | 5437 | 5430 | 3 | 0.000 | 19032 | 1837 | 1833 | 4 | 0.002 |
| 988638 | 3 | 20826548 | 20826548 | G | T | SP8 | 5_prime_flanking_region | c.-131 | NULL | tier2 | 1843 | 367 | 367 | 0 | 0.000 | 2386 | 310 | 272 | 34 | 0.111 | 3879 | 656 | 376 | 278 | 0.425 | 8289 | 1154 | 1150 | 1 | 0.000 | 4661 | 685 | 684 | 0 | 0.000 |
| 988638 | 9 | 44874097 | 44874097 | G | A | ENSG00000231454 | 3_prime_untranslated_region | c.*44874097 | NULL | tier3 | 1723 | 0 | 0 | 0 | 0.000 | 2207 | 237 | 194 | 43 | 0.181 | 2353 | 305 | 179 | 124 | 0.409 | 5943 | 772 | 772 | 0 | 0.000 | 4939 | 0 | 0 | 0 | NA |
| 988638 | 9 | 128083745 | 128083745 | G | T | GAPVD1 | nonsense | c.1636 | p.G546* | tier1 | 22704 | 3267 | 3230 | 31 | 0.010 | 21985 | 2513 | 2059 | 438 | 0.175 | 25659 | 3664 | 2598 | 1059 | 0.290 | 63694 | 8641 | 8633 | 3 | 0.000 | 73099 | 7490 | 7478 | 4 | 0.000 |
| 988638 | 10 | 51829388 | 51829388 | C | T | FAM21A | missense | c.208 | p.R70W | tier1 | 8444 | 1026 | 1023 | 1 | 0.001 | 4290 | 461 | 381 | 79 | 0.148 | 2797 | 435 | 256 | 179 | 0.411 | 14768 | 1895 | 1894 | 1 | 0.000 | 4954 | 665 | 663 | 1 | 0.000 |
| 988638 | 11 | 5842368 | 5842368 | G | C | ORS2N2 | missense | c.803 | p.G268A | tier1 | 5574 | 1335 | 1332 | 1 | 0.001 | 4387 | 763 | 660 | 99 | 0.130 | 4901 | 1257 | 722 | 535 | 0.426 | 12603 | 2795 | 2793 | 0 | 0.000 | 1033 | 158 | 158 | 0 | 0.000 |
| 988638 | 12 | 40815079 | 40815079 | G | A | MUC19 | rna | NULL | NULL | tier1 | 811 | 117 | 117 | 0 | 0.000 | 595 | 73 | 66 | 7 | 0.096 | 575 | 106 | 91 | 15 | 0.142 | 1687 | 252 | 252 | 0 | 0.000 | NA | NA | NA | NA | NA |
| 988638 | 16 | 89650436 | 89650436 | G | A | CPNE7 | missense | c.658 | p.V220M | tier1 | 21007 | 3057 | 3049 | 4 | 0.001 | 10223 | 1196 | 1027 | 159 | 0.134 | 16016 | 2561 | 1463 | 1091 | 0.427 | 26149 | 3780 | 3771 | 4 | 0.001 | 10693 | 1371 | 1368 | 1 | 0.000 |
| 988638 | 17 | 44074056 | 44074056 | G | T | MAPT | intronic | c.1827+26 | e10+26 | tier3 | 13914 | 1839 | 1819 | 17 | 0.009 | 10418 | 1118 | 888 | 202 | 0.185 | 15722 | 2272 | 1334 | 904 | 0.404 | 28154 | 3628 | 3620 | 5 | 0.001 | 5472 | 696 | 695 | 1 | 0.000 |
| 988638 | 17 | 74732959 | 74732959 | G | T | SRSF2 | missense | c.284 | p.P95H | tier1 | 10311 | 1436 | 1435 | 1 | 0.001 | 10008 | 1210 | 1024 | 178 | 0.148 | 20068 | 3093 | 1795 | 1272 | 0.418 | 28552 | 3910 | 3907 | 2 | 0.000 | 482 | 0 | 0 | 0 | NA |
| 988638 | 18 | 42531907 | 42531907 | G | A | SETBP1 | missense | c.2602 | p.D868N | tier1 | 26792 | 3478 | 3477 | 0 | 0.000 | 26771 | 3040 | 2581 | 444 | 0.147 | 47339 | 6331 | 3697 | 2593 | 0.412 | 78462 | 10064 | 10049 | 5 | 0.000 | 77196 | 6858 | 6837 | 13 | 0.002 |
| 988638 | 17 | 7675544 | 7675544 | G | C | CAMSAP3 | intronic | c.982-42 | e9-42 | tier3 | 13605 | 1826 | 1799 | 26 | 0.014 | 8017 | 873 | 656 | 207 | 0.240 | 12692 | 1912 | 1082 | 815 | 0.430 | 22406 | 2906 | 2896 | 1 | 0.000 | 6492 | 676 | 675 | 1 | 0.000 |
| 988638 | 19 | 35265460 | 35265460 | C | T | ZNF599 | 5_prime_flanking_region | c.-1714 | NULL | tier4 | 2054 | 377 | 371 | 5 | 0.013 | 1485 | 241 | 196 | 45 | 0.187 | 2262 | 548 | 311 | 236 | 0.431 | 4699 | 938 | 937 | 1 | 0.000 | 1645 | 193 | 193 | 0 | 0.000 |
| 988638 | 20 | 44815531 | 44815531 | G | A | CDH22 | silent | c.1479 | p.N493 | tier1 | 24890 | 3995 | 3945 | 48 | 0.012 | 32926 | 3919 | 3029 | 837 | 0.217 | 69113 | 9955 | 5755 | 4119 | 0.417 | 101385 | 14575 | 14551 | 12 | 0.001 | 101380 | 10080 | 10049 | 20 | 0.002 |
| 988638 | X | 14338161 | 14338161 | C | T | - | - | - | - | tier3 | 18629 | 2604 | 2598 | 5 | 0.002 | 9550 | 1070 | 765 | 300 | 0.282 | 12520 | 1786 | 338 | 1443 | 0.410 | 29780 | 4105 | 4095 | 7 | 0.002 | 39185 | 3787 | 3779 | 7 | 0.002 |
| 991140 | 1 | 181702771 | 181702771 | C | T | CACNA1E | silent | c.3147 | p.S1049 | tier1 | 25606 | 3230 | 3224 | 5 | 0.002 | 31176 | 3713 | 2869 | 815 | 0.221 | 21422 | 2645 | 2291 | 343 | 0.130 | 43362 | 5206 | 5175 | 29 | 0.006 | 43964 | 4789 | 4781 | 0 | 0.000 |
| 991140 | 1 | 223938499 | 223938499 | C | T | CAPN2 | intronic | c.814-93 | e7-93 | tier3 | 23839 | 2789 | 2777 | 10 | 0.004 | 17359 | 1943 | 1509 | 419 | 0.217 | 22663 | 2695 | 2320 | 361 | 0.135 | 38645 | 4335 | 4307 | 23 | 0.005 | 81316 | 0 | 0 | 0 | NA |
| 991140 | 1 | 235926068 | 235926068 | C | T | LYST | missense | c.6205 | p.G2069R | tier1 | 2734 | 284 | 284 | 0 | 0.000 | 5008 | 510 | 387 | 117 | 0.232 | 2316 | 237 | 200 | 35 | 0.149 | 7062 | 713 | 707 | 4 | 0.006 | 17007 | 2081 | 2080 | 0 | 0.000 |
| 991140 | 2 | 166187904 | 166187904 | T | C | SCN2A | silent | c.2214 | p.I738 | tier1 | 12601 | 1364 | 1358 | 5 | 0.004 | 7677 | 842 | 651 | 188 | 0.224 | 8846 | 974 | 888 | 83 | 0.085 | 16189 | 1647 | 1632 | 11 | 0.007 | 43391 | 4861 | 4856 | 0 | 0.000 |
| 991140 | 3 | 116163747 | 116163747 | C | T | LSAMP | silent | c.132 | p.R44 | tier1 | 11740 | 1553 | 1550 | 3 | 0.002 | 32947 | 4167 | 3195 | 939 | 0.227 | 37323 | 4927 | 4262 | 642 | 0.131 | 61809 | 6830 | 6787 | 40 | 0.006 | 50384 | 5382 | 5378 | 0 | 0.000 |
| 991140 | 3 | 132201248 | 132201248 | C | T | DNAIC13 | intronic | c.2979+74 | e26+74 | tier3 | 11745 | 1555 | 1549 | 5 | 0.003 | 4943 | 693 | 497 | 192 | 0.279 | 10516 | 1420 | 1282 | 135 | 0.095 | 16281 | 2010 | 1994 | 14 | 0.007 | 29503 | | | | |

Table S5. International Prognostic Scoring System-Revised Classification System

| Score | Cytogenetics | Blast % | Hemoglobin | Platelets | Absolute Neutrophil Count |
|-------|--------------|---------|------------|-----------|---------------------------|
| 0 | Very Good | ≤2 | ≥10 | ≥100 | ≥0.8 |
| 0.5 | | | | 50-<100 | <0.8 |
| 1.0 | Good | >2-<5 | 8-<10 | <50 | |
| 1.5 | | | <8 | | |
| 2.0 | Intermediate | 5-10 | | | |
| 3.0 | Poor | >10 | | | |
| 4.0 | Very Poor | | | | |

| Prognostic Subgroups | Cytogenetic abnormalities |
|----------------------|---|
| Very Good | -Y, del(11q) |
| Good | Normal, del(5q), del(12p), del(20q), double including del(5q) |
| Intermediate | del(7q), +8, +19, i(17q), any other single or double independent clones |
| Poor | -7, inv(3)/t(3q)/del(3q), double including -7/del(7q), complex: 3 abnormalities |
| Very Poor | Complex:>3 abnormalities |

| International Prognostic Scoring System-Revised Score | Risk Category | Median Overall Survival (years) | Median time to 25% leukemia evolution (years) |
|---|---------------|---------------------------------|---|
| ≤1.5 | Very Low | 8.8 | Not reached |
| >1.5-3.0 | Low | 5.3 | 10.8 |
| >3-4.5 | Intermediate | 3.0 | 3.2 |
| >4.5-6 | High | 1.6 | 1.4 |
| >6 | Very High | 0.8 | 0.73 |

From: Greenberg PL, Tuechler H, Schanz J, Sanz G, Garcia-Manero G, Sole F, et al. Revised international prognostic scoring system for myelodysplastic syndromes. *Blood*. 2012;120(12):2454-65.20(12):2

Table S6. 40 Gene Panel List

| Gene symbol | Category | Target(s) |
|--------------------|---------------------------|-------------------|
| <i>BRAF</i> | Activated signaling | V600E |
| <i>FLT3</i> | Activated signaling | TKD and ITD |
| <i>JAK2</i> | Activated signaling | V617, exon 12 |
| <i>KIT</i> | Activated signaling | exons 2, 8-13, 17 |
| <i>KRAS</i> | Activated signaling | G12, G13, Q61 |
| <i>MPL</i> | Activated signaling | exon 10 |
| <i>NF1</i> | Activated signaling | whole gene |
| <i>NRAS</i> | Activated signaling | G12, G13, Q61 |
| <i>PTPN11</i> | Activated signaling | exons 3, 13, 14 |
| <i>ASXL1</i> | Chromatin modifiers | whole gene |
| <i>EZH2</i> | Chromatin modifiers | whole gene |
| <i>SUZ12</i> | Chromatin modifiers | whole gene |
| <i>CSF3R</i> | Chromatin modifiers | whole gene |
| <i>RAD21</i> | Cohesin | whole gene |
| <i>SMC1A</i> | Cohesin | whole gene |
| <i>SMC3</i> | Cohesin | whole gene |
| <i>STAG2</i> | Cohesin | whole gene |
| <i>DNMT3A</i> | DNA methylation | whole gene |
| <i>IDH1</i> | DNA methylation | R132 |
| <i>IDH2</i> | DNA methylation | R140, R172 |
| <i>TET2</i> | DNA methylation | whole gene |
| <i>CALR</i> | Other genes | exon 9 |
| <i>CBL</i> | Other genes | exons 8, 9 |
| <i>NPM1</i> | Other genes | exon 11 |
| <i>PIGA</i> | Other genes | whole gene |
| <i>PPM1D</i> | Other genes | exon 6 |
| <i>CUX1</i> | Other genes | whole gene |
| <i>SF3B1</i> | Spliceosome | whole gene |
| <i>SRSF2</i> | Spliceosome | exon 1 |
| <i>U2AF1</i> | Spliceosome | exons 2, 6 |
| <i>ZRSR2</i> | Spliceosome | whole gene |
| <i>BCOR</i> | Transcriptional regulator | whole gene |
| <i>BCORL1</i> | Transcriptional regulator | whole gene |
| <i>CEBPA</i> | Transcriptional regulator | whole gene |
| <i>ETV6</i> | Transcriptional regulator | whole gene |
| <i>GATA2</i> | Transcriptional regulator | whole gene |
| <i>RUNX1</i> | Transcriptional regulator | whole gene |
| <i>PHF6</i> | Tumor suppressors | whole gene |
| <i>TP53</i> | Tumor suppressors | whole gene |
| <i>WT1</i> | Tumor suppressors | whole gene |