Supplementary materials and methods

Targeted resequencing

RNA probes complementary in sequence to the coding exons (± 2bp) of target genes were designed using e-array (https://earray.chem.agilent.com/earray/) and manufactured as part of custom SureSelect kits (Agilent, Santa Clara, CA). A tiling frequency of 2x was selected during kit design and small target intervals were extended such that all intervals were targeted by at least 2 distinct probes. Paired end Illumina sequencing libraries were prepared for 444 cases and 311 controls using DNA extracted from venous blood (KBioscience, Herts, England). Libraries were subsequently enriched for target sequences using the custom SureSelect kits. Library preparation and target enrichment was performed as per the "SureSelect Target Enrichment System for Illumina Paired-End Sequencing Library protocol version 2.3" protocol with the following modifications. 1) Only 1 µg of DNA was used in library preparations. 2) DNA fragmentation was performed either using a Covaris system (Covaris, Woburn, MA) as per the manufacturer's instructions, enzymatically or through sonication. Where enzymatic fragmentation was performed, 15.8 µl of DNA (63.3 ng/µl) was incubated with 2 µl NEBNext dsDNA fragmentase (New England Biolabs, Ipswich, MA) in NEBNext fragmentase reaction buffer @ 30 °C for 20 min. 5 µl of 0.5 M EDTA was then used to stop the reaction. Where fragmentation was performed through sonication, 100 µl of DNA (10 ng/µl) was subjected to 60 min of sonication (30 sec on, 30 sec off) using a Bioruptor (Diagenode, Liège, Belgium) at low power. 3) Extended Illumina adapters including 6 bp indexes were used in place of standard adapters (Metabion, Martinsried, Germany). All indexes were at least an edit distance of 2 from one another. 4) Following adapter ligation, libraries were subjected to electrophoresis through a 2% Low Range Ultra agarose gel (BioRad, Hercules, CA) stained with SYBR Green 1 nucleic acid gel stain (Sigma, Arklow, Ireland) for 90 mins @ 140 V. Fragments of 300 – 400 bp were manually

excised from gel, using a Safe Imager™ 2.0 Blue Light Transilluminator (Invitrogen, Carlsbad, CA) for visualization. DNA was extracted from gel slices using QIAquick Gel Extraction Kits (Qiagen, West Sussex, UK) as per the manufacturer's instructions. 5) 5 cycles of PCR were performed instead of 4 prior to target enrichment, and Phusion (New England Biolabs) was used for all PCR reactions. 6) Libraries labelled with distinct indexes were pooled prior to target enrichment such that the expected sequence coverage of target intervals would have a mean of ~ 30x per sample. 203 case libraries were resequenced by 80 bp paired-end resequencing on an Illumina Genome Analyzer II at TrinSeq, Institute of Molecular Medicine, Trinity College Dublin, Ireland. The remainder of libraries were resequenced by 101 bp single-end sequencing on a HiSeq 2000 at GATC Biotech AG, Konstanz, Switzerland.

Analysis of sequence data

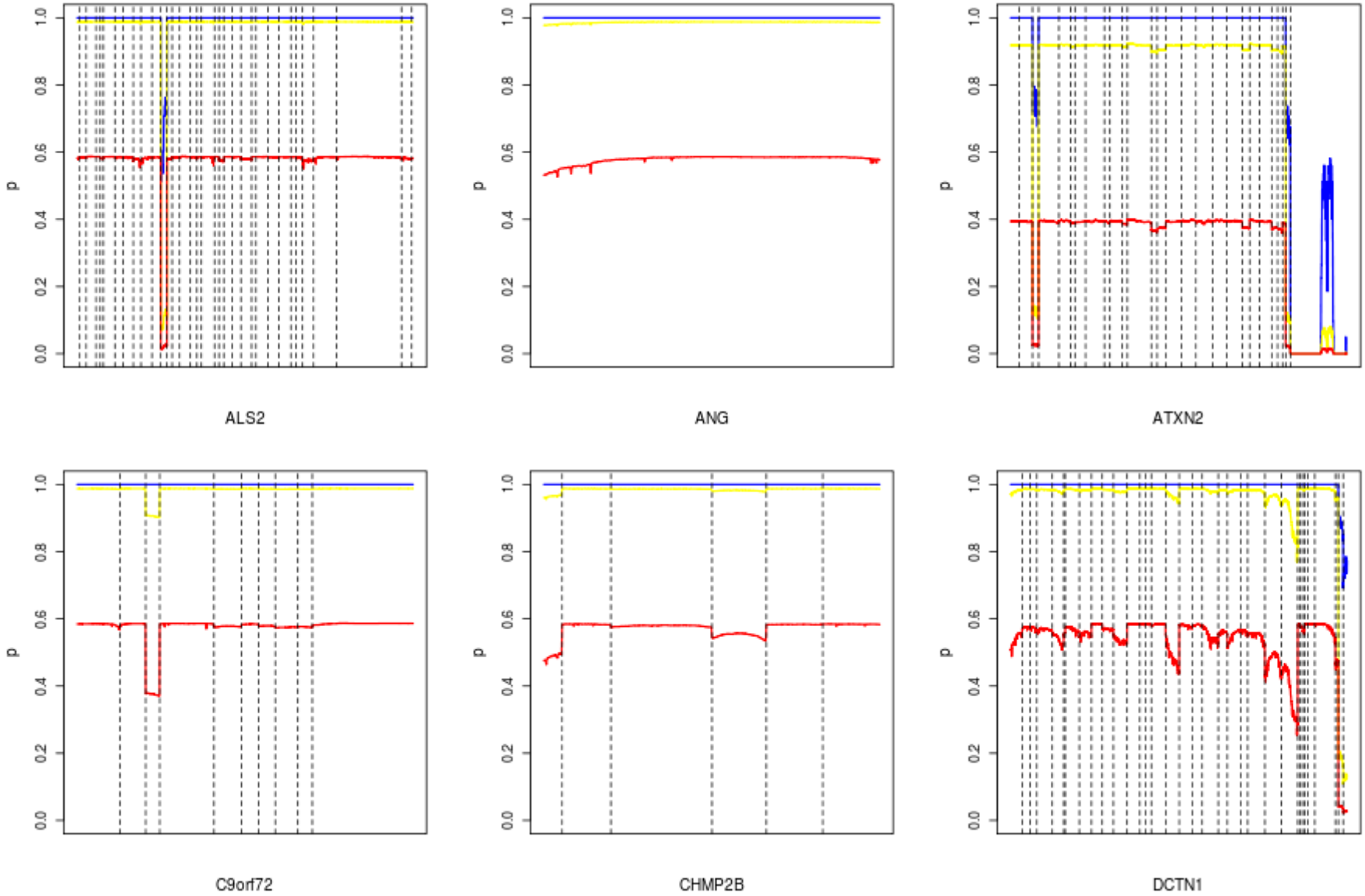
Sequencing reads were aligned using BWA to build GRCh37 of the human genome ("http://www.broadinstitute.org/gsa/wiki/index.php/GSA_FTP_Server"). Alignments were annotated with sample of origin based on detected adapter indexes. SAMtools was used to sort and index alignment files. Picard was used to merge alignment files and remove PCR duplicates. The GATK was used to perform local indel realignment, recalibrate base quality (BQ) scores, determine per sample depth of coverage across target intervals, call variants and assign sample genotypes. Base calls were only considered for depth of coverage analyses or variant calling when they were associated with BQ scores >= 20 and alignment mapping quality scores >=20.

Variant annotation and quality control

The effects of variants at the protein level were predicted using Variant Effect Predictor version 2.7 (http://www.ensembl.org/info/docs/variation/vep/index.html) and Ensembl 69 (http://www.ensembl.org/). Predictions were based only on transcripts annotated as protein coding and included PolyPhen and SIFT pathogenicity classifications. Genotype frequencies within external control cohorts were imported from the March 2012 release of the 1000 genomes project (http://www.1000genomes.org/) and the ESP6500 release of the NHLBI exome sequencing project (Exome Variant Server, NHLBI Exome Sequencing Project (ESP), Seattle, WA (URL: http://evs.gs.washington.edu/EVS/) [Accessed 2012 Jul 18]). Variants were assigned a status of either "pass" or "fail" based on whether they met a series of quality control criteria. These criteria included a minimum variant quality score of 30, a genotype call rate ≥10%, and "QD", "FS", "ReadPosRankSum" and "HaplotypeScore" values within the range observed for the subset of variants noted also to have been reported by the 1000 genomes or NHLBI Exome Sequencing projects. Additionally, novel case variants which were observed to occur only *in cis* with other novel case variants were interpreted as possible misalignments and assigned a status of "fail". Variant quality score, "QD", "FS", "ReadPosRankSum" and "HaplotypeScore" values were calculated using the GATK. Genotype calls associated with genotype quality scores <15 were reset to missing prior to variant quality control.

Estimation of variant detection power

For each target position, per sample sequence coverage was used to calculate the expected total of resequenced chromosomes. The probability of including a mutated chromosome among this total was then calculated for a range of minor allele frequencies using R version 2.14.1 (http://www.r-project.org/). Both the sampling of patients from the Irish ALS population and the sampling of chromosomes during resequencing were assumed to follow binomial distributions.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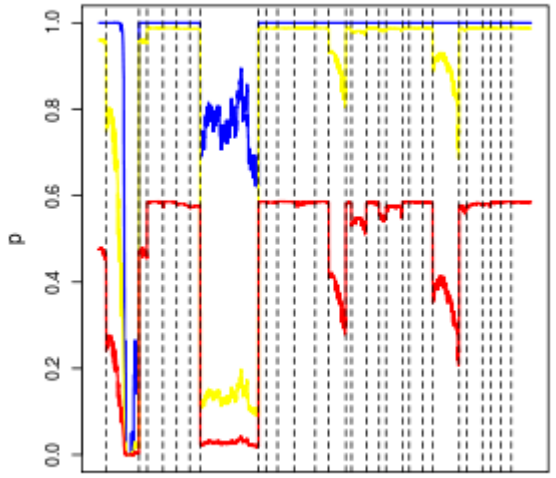
ALS2
ANG
ATXN2
C9orf72
CHMP2B
DCTN1



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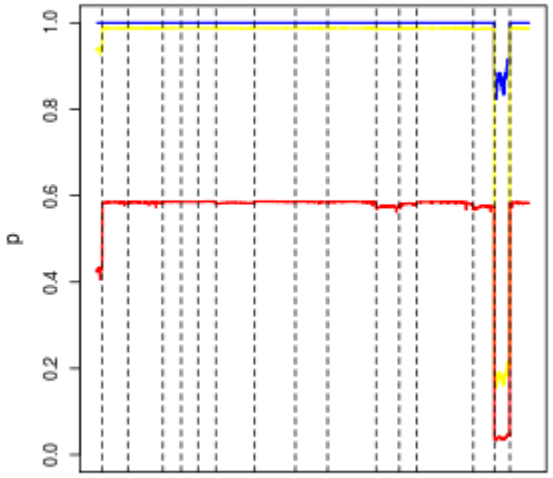
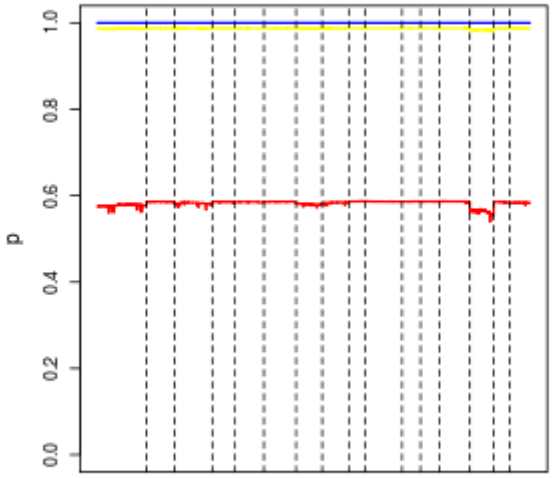


ELP3



FGGY

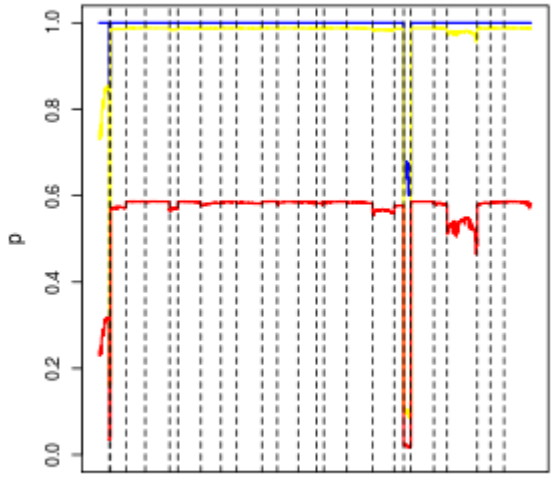
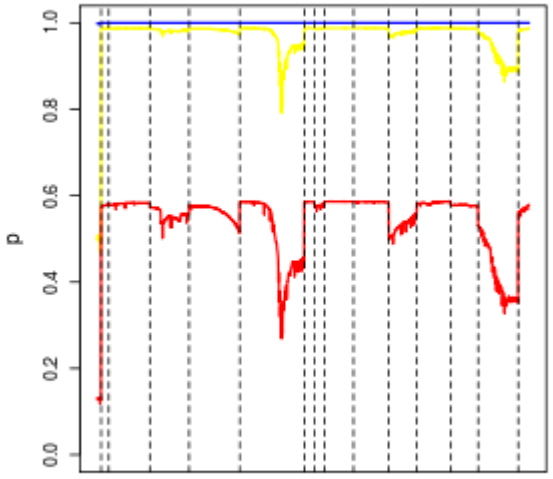
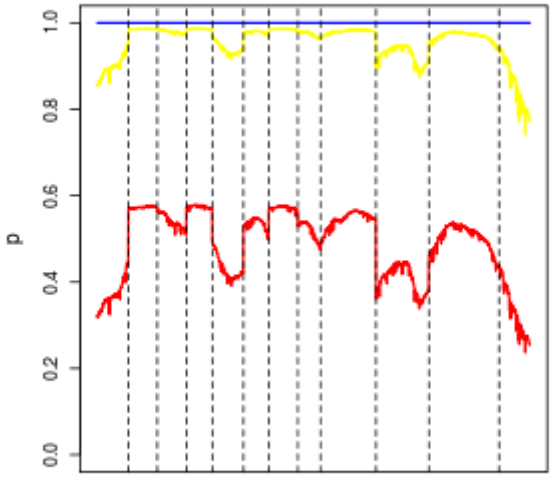


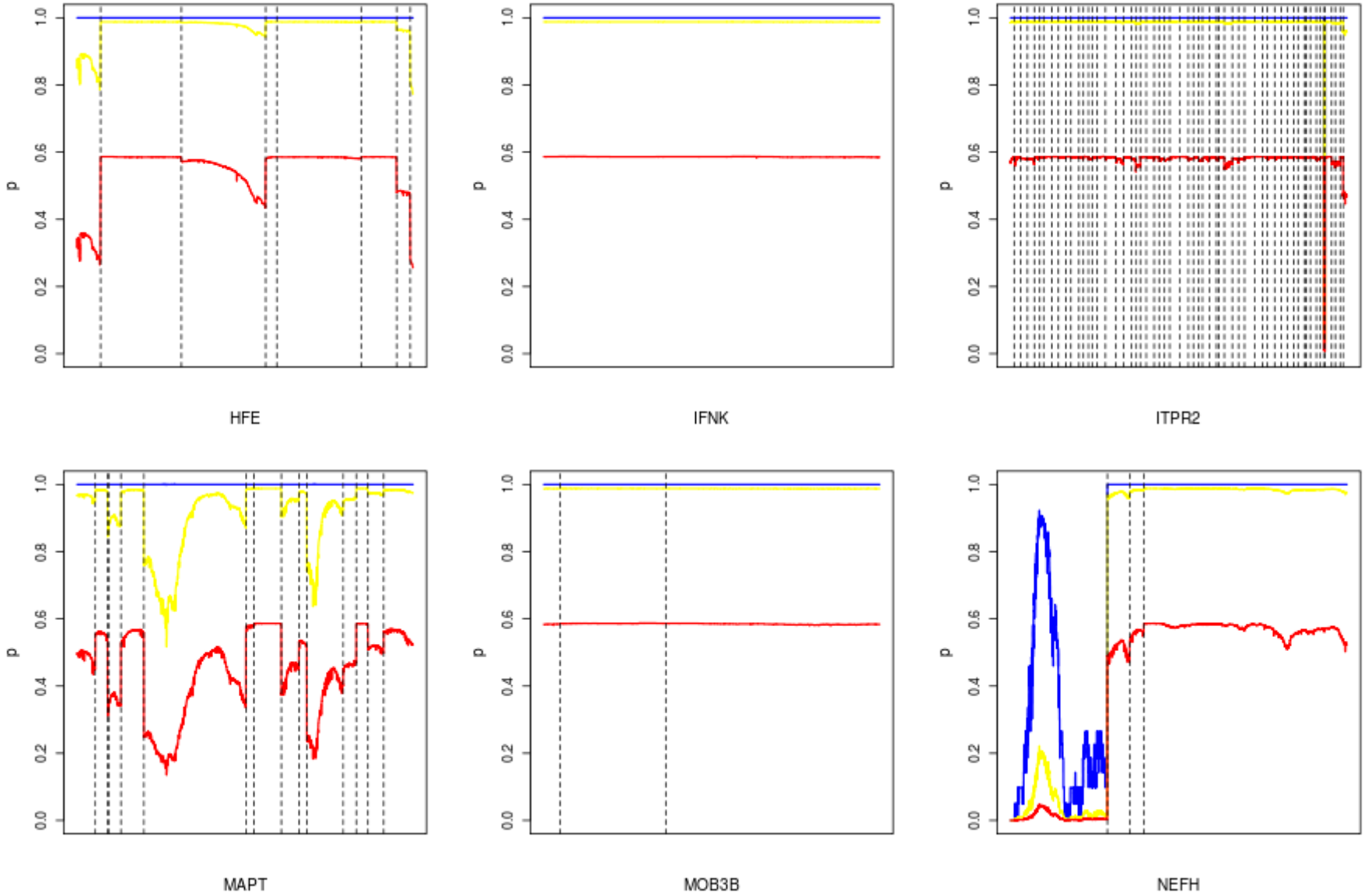
FIG4



FUS



GRN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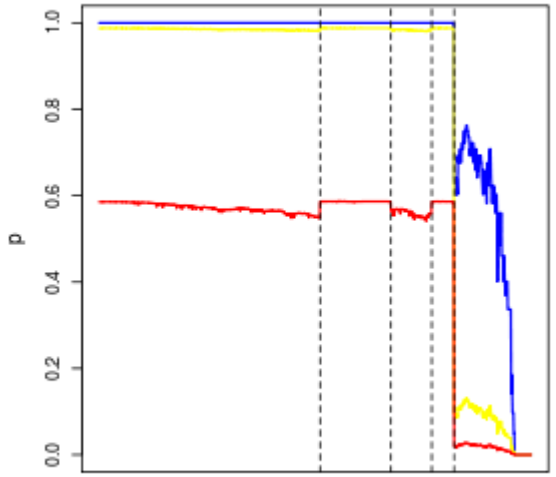
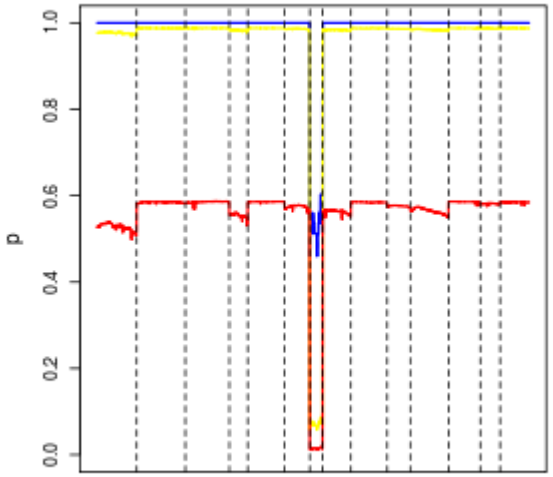


NIPA1



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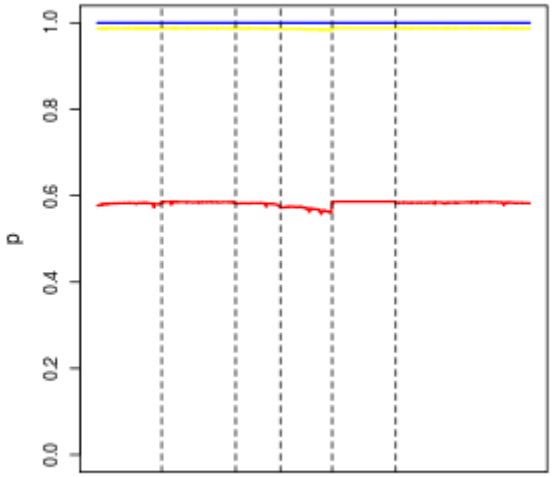
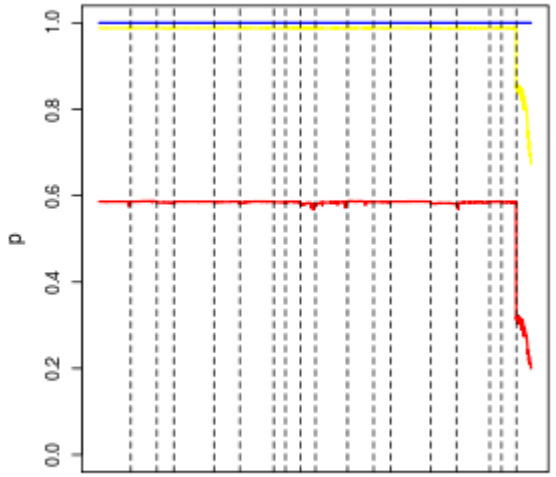
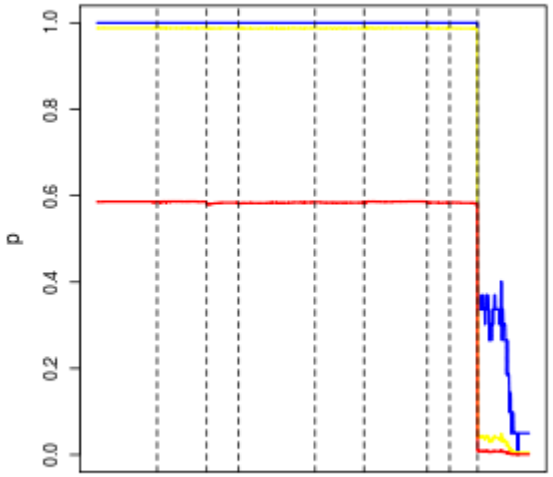


PARK7



PON1



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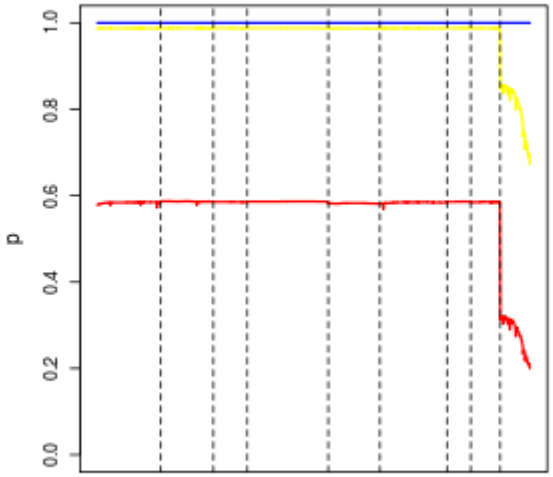
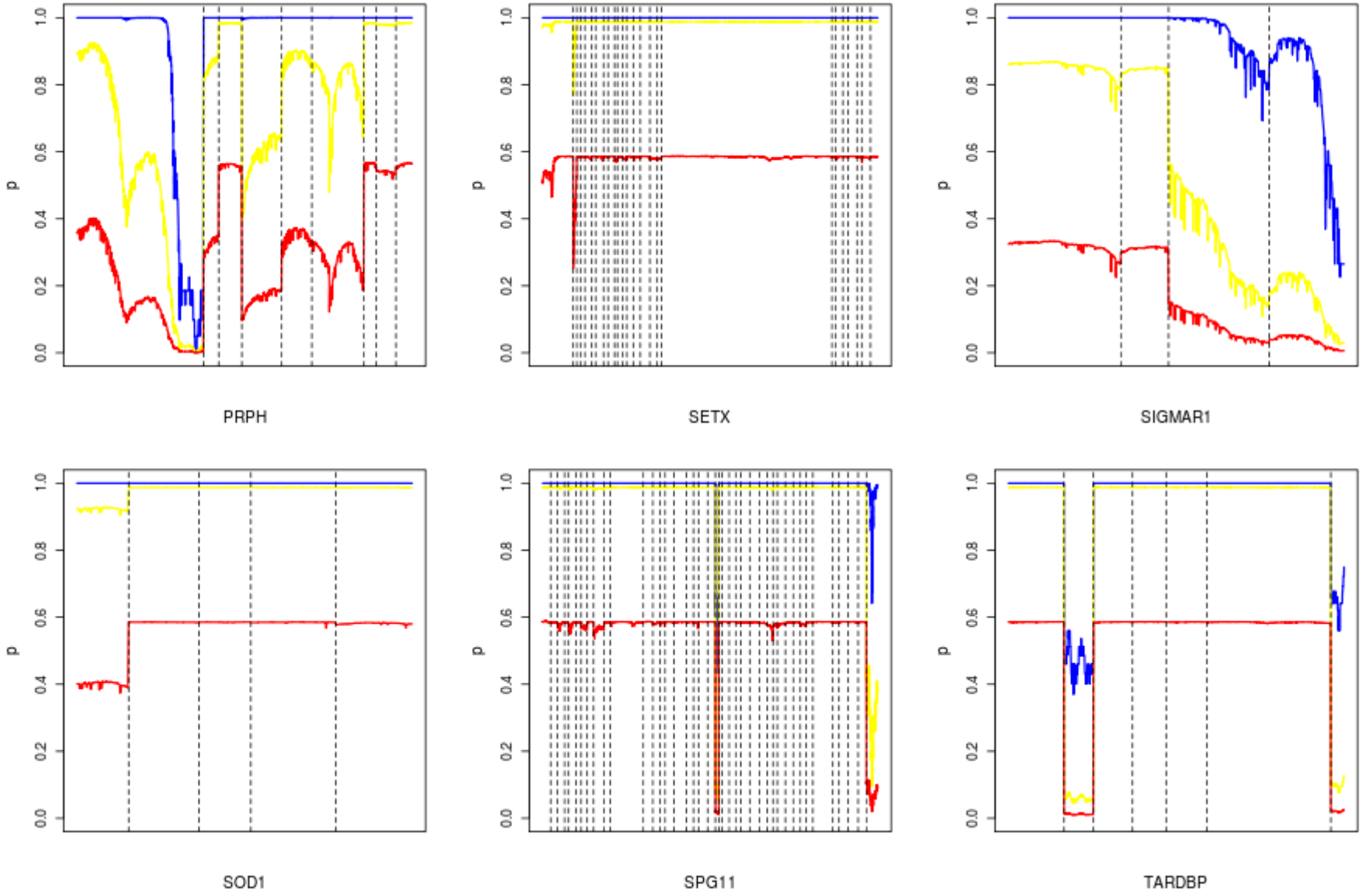


PON3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SIGMAR1
SOD1
SPG11
TARDBP

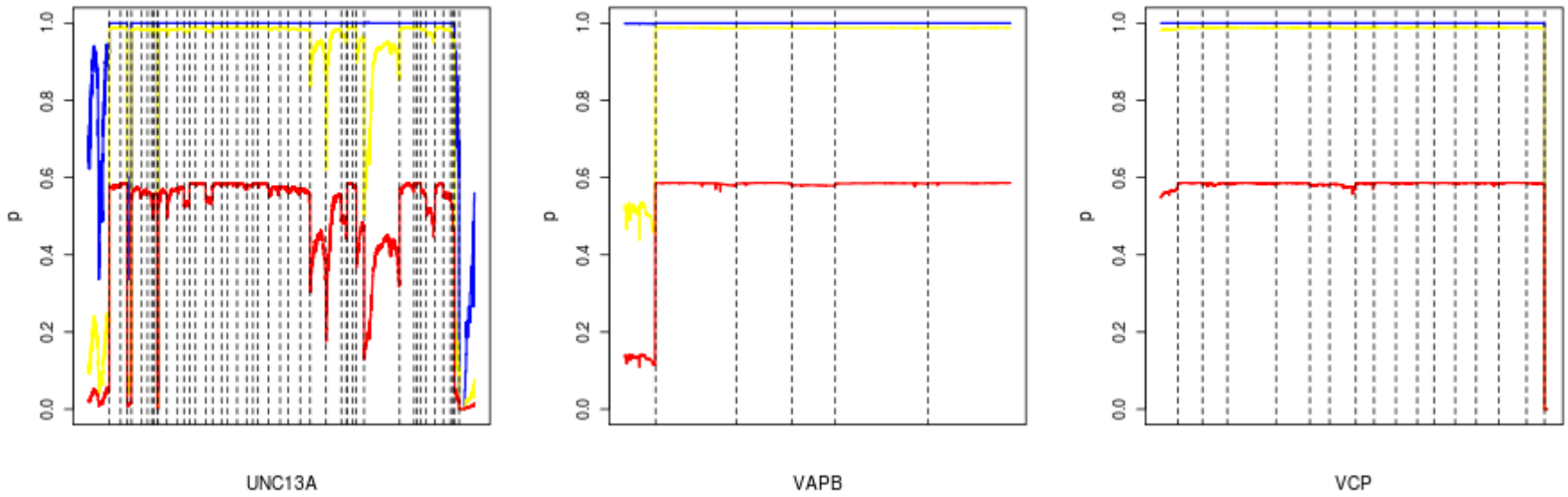

Figure S1: Variant detection power

The horizontal axes denote position within the coding sequence of each target gene. The vertical axes denote the probability that a variant with an allele frequency of 5% (blue line), 0.5% (yellow line) or 0.1% (red line) among Irish ALS patients was sampled during resequencing. The coding sequence of a target gene refers to the union of all coding transcripts reported in Ensembl 69. Exon boundaries are demarcated by vertical dotted lines.

Table S1: Target gene resequencing

Gene	Designation	% Sequenced (>=1 X coverage)	Mean Case Coverage
ALS2	Mendelian	100	35.6 + 25.4
ANG	Mendelian	100	30.4 + 22.2
ATXN2	Low penetrance/ tenative	87.1	21.4 + 26.7
C9orf72	Mendelian	100	36.7 + 27.1
CHMP2B	Low penetrance/ tenative	100	25.8 + 19.7
DCTN1	Low penetrance/ tenative	100	16.6 + 17.1
DPP6	Low penetrance/ tenative	98.8	20.6 + 22.2
ELP3	Low penetrance/ tenative	100	32.3 + 21.9
FGGY	Low penetrance/ tenative	100	30.8 + 20.1
FIG4	Low penetrance/ tenative	100	33.4 + 24.3
FUS	Mendelian	100	17.9 + 18.1
GRN	Low penetrance/ tenative	100	10.1 + 12.3
HFE	Low penetrance/ tenative	100	28.7 + 25.4
IFNK	Low penetrance/ tenative	100	38.8 + 21.6
ITPR2	Low penetrance/ tenative	100	31.6 + 21.3
MAPT	Low penetrance/ tenative	100	12.4 + 18.8
MOB3B	Low penetrance/ tenative	100	38.9 + 23.9
NEFH	Low penetrance/ tenative	97.7	11.4 + 13.5
NIPA1	Low penetrance/ tenative	96.1	22 + 20.7
OPTN	Mendelian	100	21.3 + 15.9
PARK7	Low penetrance/ tenative	100	26.7 + 18.1
PON1	Low penetrance/ tenative	100	37.1 + 24.7

PON2	Low penetrance/ tenative	99.9	34.7 + 25.3
PON3	Low penetrance/ tenative	100	32.7 + 22.2
PRPH	Low penetrance/ tenative	99.4	4.2 + 8.7
SETX	Mendelian	100	41.2 + 25.7
SIGMAR1	Low penetrance/ tenative	100	2.9 + 7.5
SOD1	Mendelian	100	29.7 + 24.6
SPG11	Low penetrance/ tenative	100	32.9 + 23.7
TARDBP	Mendelian	100	30.3 + 22.1
UNC13A	Low penetrance/ tenative	98.6	15.5 + 17.9
VAPB	Mendelian	100	31.8 + 22.3
VCP	Mendelian	99.2	34.1 + 23

Table S2: ALS gene variants

Gene	Class	Transcript	DNA	Amino.Acid	SIFT	Poly Phen	fALS	sALS	Irish	European	Global
ALS2	missense	*ENST00000264276, ENST00000467448	c.1102G>A, c.1102G>A	p.Val368Met, p.Val368Met	0, 0	0, 0	0,4,46	5,70,315	2,48,240	45,871,3894	145,1662,5633
ALS2	missense, splice region	*ENST00000264276	c.1115C>G	p.Pro372Arg	0	1	50,0,0	389,1,0	294,2,0	4742,22,0	7306,23,0
ALS2	***missense***	****ENST00000264276***	***c.1226C>G***	***p.Ala409Gly***	***0***	***0***	***48,0,0***	***358,12,0***	***252,23,0***	***252,23,0***	***252,23,0***
ALS2	missense	*ENST00000264276	c.1265T>C	p.Met422Thr	1	0	50,0,0	388,1,0	302,0,0	4454,1,0	6399,1,0
ALS2	***missense***	****ENST00000264276***	***c.1283C>A***	***p.Thr428Asn***	***0***	***0***	***50,0,0***	***388,1,0***	***302,0,0***	***302,0,0***	***302,0,0***
ALS2	missense	*ENST00000264276	c.1627G>A	p.Asp543Asn	1	2	47,0,0	290,0,0	247,4,0	4389,11,0	6357,11,0
ALS2	splice region, synonymous	*ENST00000264276	c.1641G>A	p.(=)			50,0,0	380,1,0	277,0,0	4388,4,0	6255,9,0
ALS2	synonymous	*ENST00000264276	c.2028A>G	p.(=)			50,0,0	383,0,0	267,1,0	267,1,0	267,1,0
ALS2	missense	*ENST00000264276, ENST00000457679	c.2098A>G, c.34A>G	p.Thr700Ala, p.Thr12Ala	0, 0	0, 0	50,0,0	384,2,0	279,0,0	279,0,0	279,0,0
ALS2	missense	*ENST00000264276, ENST00000457679	c.2155C>T, c.91C>T	p.Pro719Ser, p.Pro31Ser	1, 1	2, 2	50,0,0	389,0,0	282,1,0	282,1,0	282,1,0
ALS2	***missense***	****ENST00000264276, ENST00000457679***	***c.2216C>T, c.152C>T***	***p.Ala739Val, p.Ala51Val***	***0, 0***	***0, 0***	***46,2,0***	***304,52,0***	***210,54,0***	***210,54,0***	***210,54,0***
ALS2	synonymous	*ENST00000264276, ENST00000457679	c.2241C>T, c.177C>T	p.(=), p.(=)	,	,	50,0,0	388,1,0	295,1,0	4445,25,0	6403,28,0
ALS2	missense	*ENST00000264276, ENST00000457679	c.2408A>G, c.344A>G	p.Lys803Arg, p.Lys115Arg	0, 0	1, 0	50,0,0	390,1,0	289,0,0	289,0,0	289,0,0
ALS2	synonymous	*ENST00000264276, ENST00000457679	c.2466G>A, c.402G>A	p.(=), p.(=)	,	,	8,25,15	57,175,145	37,117,102	750,2244,1719	2360,2984,1894
ALS2	missense	*ENST00000264276, ENST00000457679	c.2566A>G, c.502A>G	p.Thr856Ala, p.Thr168Ala	0, 0	1, 0	47,0,0	372,1,0	253,0,0	253,0,0	253,0,0
ALS2	missense	*ENST00000264276, ENST00000457679	c.2606A>C, c.542A>C	p.Gln869Pro, p.Gln181Pro	1, 0	2, 1	50,0,0	389,1,0	290,0,0	290,0,0	290,0,0
ALS2	synonymous	*ENST00000264276, ENST00000457679	c.2796C>T, c.732C>T	p.(=), p.(=)	,	,	34,15,1	308,75,4	241,54,2	4029,745,31	6262,1120,53
ALS2	missense	*ENST00000264276, ENST00000467448,	c.280A>G, c.280A>G,	p.Ile94Val, p.Ile94Val,	0, 0, 0, 0	0, 0, 0, 0	47,3,0	371,15,1	285,14,0	4596,232,4	7069,445,7

		ENST00000409632, ENST00000410052	c.280A>G, c.280A>G	p.Ile94Val, p.Ile94Val							
ALS2	***synonymous***	****ENST00000264276, ENST00000457679***	***c.2992C>A, c.928C>A***	***p.(=), p.(=)***	***,***	***,***	***48,0,0***	***352,1,0***	***268,2,0***	***268,2,0***	***268,2,0***
ALS2	missense	*ENST00000264276, ENST00000457679	c.3046C>G, c.982C>G	p.Pro1016Ala, p.Pro328Ala	0, 0	0, 0	50,0,0	386,1,0	291,0,0	4445,1,0	6365,17,0
ALS2	missense	*ENST00000264276, ENST00000457679	c.3094C>T, c.1030C>T	p.Arg1032Cys, p.Arg344Cys	1, 1	2, 2	49,0,0	374,1,0	274,0,0	274,0,0	274,0,0
ALS2	synonymous	*ENST00000264276, ENST00000457679	c.3129C>G, c.1065C>G	p.(=), p.(=)	,	,	50,0,0	387,1,0	294,0,0	294,0,0	294,0,0
ALS2	***splice donor***	****ENST00000264276, ENST00000457679***	***c.3182+2T>G, c.1118+2T>G***	***,***	***,***	***,***	***46,0,0***	***361,3,0***	***254,6,0***	***254,6,0***	***254,6,0***
ALS2	***missense, splice region***	****ENST00000264276, ENST00000457679***	***c.3250T>G, c.1186T>G***	***p.Tyr1084Asp, p.Tyr396Asp***	***1, 1***	***1, 1***	***40,3,0***	***280,20,0***	***187,18,0***	***187,18,0***	***187,18,0***
ALS2	missense	*ENST00000264276, ENST00000457679	c.3307C>A, c.1243C>A	p.His1103Asn, p.His415Asn	0, 1	0, 0	49,0,0	384,0,0	277,1,0	277,1,0	277,1,0
ALS2	synonymous	*ENST00000264276	c.3741T>G	p.(=)			50,0,0	385,1,0	282,0,0	4728,20,0	7267,20,0
ALS2	missense	*ENST00000264276	c.3863C>T	p.Pro1288Leu	0	0	50,0,0	375,1,0	302,0,0	4403,3,0	6299,3,0
ALS2	synonymous	*ENST00000264276	c.3885G>A	p.(=)			45,3,0	349,14,0	266,15,1	4404,366,7	6998,399,7
ALS2	synonymous	*ENST00000264276	c.4015C>T	p.(=)			0,3,44	1,57,288	0,25,218	25,682,3997	83,1354,5829
ALS2	synonymous	*ENST00000264276	c.4107G>A	p.(=)			46,0,0	352,1,0	245,0,0	245,0,0	245,0,0
ALS2	missense	*ENST00000264276	c.4119A>G	p.Ile1373Met	1	0	46,0,0	342,2,0	243,0,0	4664,43,0	7189,47,0
ALS2	missense	*ENST00000264276	c.4135C>A	p.Pro1379Thr	1	1	50,0,0	386,0,0	296,2,0	296,2,0	296,2,0
ALS2	missense	*ENST00000264276	c.4382G>A	p.Arg1461Gln	0	0	50,0,0	384,1,0	282,1,0	282,1,0	282,1,0
ALS2	synonymous	*ENST00000264276	c.4641G>A	p.(=)			49,0,0	387,1,0	277,2,0	277,2,0	277,2,0
ALS2	missense	*ENST00000264276	c.4957C>T	p.Arg1653Cys	1	1	50,0,0	356,0,0	239,1,0	4337,2,0	6197,3,0
ANG	missense	*ENST00000336811, ENST00000397990	c.122A>T, c.122A>T	p.Lys41Ile, p.Lys41Ile	1, 1	0, 0	50,0,0	361,5,0	283,5,0	4938,28,0	7850,32,0
ANG	missense	*ENST00000336811, ENST00000397990	c.208A>G, c.208A>G	p.Ile70Val, p.Ile70Val	0, 0	0, 0	50,0,0	387,1,0	301,2,0	4596,7,0	6797,9,0
ANG	synonymous	*ENST00000336811, ENST00000397990	c.330T>G, c.330T>G	p.(=), p.(=)	,	,	32,15,3	304,76,7	220,76,10	3725,1169,90	5982,1771,147
ANG	synonymous	*ENST00000336811,	c.363A>T,	p.(=), p.(=)	,	,	50,0,0	388,0,0	305,1,0	4978,6,0	7669,223,8

		ENST00000397990	c.363A>T								
ATXN2	missense	*ENST00000377617, ENST00000535949, ENST00000550104, ENST00000542287, ENST00000471866, ENST00000389153, ENST00000548492	c.1123G>A, c.256G>A, c.1123G>A, c.328G>A, c.151G>A, c.328G>A, c.352G>A	p.Asp375Asn, p.Asp86Asn, p.Asp375Asn, p.Asp110Asn, p.Asp51Asn, p.Asp110Asn, p.Asp118Asn	1, 1, 1, 1, 1, 1, 1	0, 0, 0, 0, 0, 0, 0	37,0,0	210,0,0	158,1,0	158,1,0	158,1,0
ATXN2	missense	*ENST00000377617, ENST00000535949, ENST00000550104, ENST00000542287, ENST00000389153	c.1472A>G, c.605A>G, c.1472A>G, c.677A>G, c.677A>G	p.Asn491Ser, p.Asn202Ser, p.Asn491Ser, p.Asn226Ser, p.Asn226Ser	0, 0, 0, 0, 0	0, 0, 0, 0, 0	36,1,0	206,4,0	163,0,0	4821,20,0	7733,24,0
ATXN2	missense, splice region, synonymous	*ENST00000377617, ENST00000542287, ENST00000550104, ENST00000389153, ENST00000389154, ENST00000535949, ENST00000482777	c.3000A>G, c.2205A>G, c.2939A>G, c.2211A>G, c.165A>G, c.2133A>G, c.57A>G	p.(=), p.(=), p.Tyr980Cys, p.(=), p.(=), p.(=), p.(=)	, , 0, , , ,	, , 0, , , ,	37,0,0	208,2,0	164,1,0	4803,40,0	7712,47,0
ATXN2	***missense***	****ENST00000377617, ENST00000550104***	***c.319C>G, c.319C>G***	***p.Leu107Val, p.Leu107Val***	***0, 0***	***,***	***0,0,0***	***0,0,0***	***0,0,0***	***19,122,237***	***416,351,324***
ATXN2	missense	*ENST00000377617, ENST00000550844, ENST00000389154, ENST00000542287, ENST00000389153	c.3322C>T, c.97C>T, c.487C>T, c.2527C>T, c.2533C>T	p.Pro1108Ser, p.Pro33Ser, p.Pro163Ser, p.Pro843Ser, p.Pro845Ser	0, 0, 0, 0, 0	2, 2, 1, 2, 2	37,0,0	200,1,0	147,3,0	4811,17,0	7724,20,0
ATXN2	missense, synonymous	*ENST00000377617, ENST00000535949, ENST00000550844, ENST00000542287, ENST00000389154, ENST00000389153, ENST00000482777, ENST00000550889	c.3411C>T, c.2490C>T, c.186C>T, c.2616C>T, c.576C>T, c.2622C>T, c.414C>T, c.65C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.Thr22Met	, , , , , , ,	, , , , , , ,	37,0,0	209,1,0	165,0,0	165,0,0	165,0,0

ATXN2	missense	*ENST00000377617, ENST00000550844, ENST00000389154, ENST00000482777, ENST00000550889, ENST00000389153, ENST00000535949, ENST00000542287	c.3491G>A, c.266G>A, c.656G>A, c.494G>A, c.145G>A, c.2702G>A, c.2570G>A, c.2696G>A	p.Ser1164Asn , p.Ser89Asn, p.Ser219Asn, p.Ser165Asn, p.Val49Ile, p.Ser901Asn, p.Ser857Asn, p.Ser899Asn	0, 0, 0, 0, , 0, 0, 0	0, 0, 0, 0, , 0, 0, 0	36,1,0	210,0,0	165,0,0	4464,1,0	6667,1,0
ATXN2	synonymous	*ENST00000377617, ENST00000389154, ENST00000550844, ENST00000542287, ENST00000389153, ENST00000535949, ENST00000482777	c.3708G>A, c.873G>A, c.483G>A, c.2913G>A, c.2919G>A, c.2787G>A, c.711G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , ,	, , , , , ,	37,0,0	207,3,0	165,0,0	4805,38,0	7714,45,0
ATXN2	missense	*ENST00000377617, ENST00000550104	c.743G>A, c.743G>A	p.Ser248Asn, p.Ser248Asn	0, 0	1, 0	36,0,0	197,3,0	145,4,0	4790,37,0	6858,776,108
C9orf72	synonymous	*ENST00000380003	c.1275G>A	p.(=)			48,0,0	362,0,0	252,1,0	4552,1,0	6752,2,0
C9orf72	synonymous	*ENST00000380003	c.1404C>T	p.(=)			49,1,0	383,3,0	270,6,0	4898,56,0	7809,61,0
C9orf72	missense	*ENST00000380003, ENST00000379995, ENST00000379997	c.620A>G, c.620A>G, c.620A>G	p.Asn207Ser, p.Asn207Ser, p.Asn207Ser	0, 0, 0	0, 0, 0	38,9,1	301,69,6	194,47,5	3788,1048,87	6523,1227,89
C9orf72	synonymous	*ENST00000380003	c.870C>T	p.(=)			35,12,3	275,95,16	193,84,11	3232,1534,200	5090,2459,333
CHMP2B	missense	*ENST00000263780, ENST00000494980	c.118A>G, c.118A>G	p.Lys40Glu, p.Lys40Glu	1, 1	1, 0	50,0,0	372,1,0	261,0,0	261,0,0	261,0,0
CHMP2B	missense	*ENST00000263780, ENST00000494980	c.123G>T, c.123G>T	p.Gln41His, p.Gln41His	1, 1	1, 2	50,0,0	372,1,0	263,0,0	263,0,0	263,0,0
CHMP2B	synonymous	*ENST00000263780, ENST00000494980	c.27C>T, c.27C>T	p.(=), p.(=)	,	,	24,4,0	135,42,3	104,26,1	4016,752,41	6298,1343,84
CHMP2B	synonymous	*ENST00000263780, ENST00000471660	c.312T>C, c.189T>C	p.(=), p.(=)	,	,	0,3,43	1,23,336	0,26,214	16,506,4396	467,1777,5590
CHMP2B	synonymous	*ENST00000263780, ENST00000471660, ENST00000494980	c.372A>C, c.249A>C, c.282A>C	p.(=), p.(=), p.(=)	, ,	, ,	33,8,1	248,53,4	176,36,0	4084,760,38	6382,1338,78
DCTN1	synonymous	*ENST00000361874,	c.1059C>T,	p.(=), p.(=),	, , , ,	, , , ,	46,0,0	274,1,0	211,1,0	211,1,0	211,1,0

		ENST00000394003, ENST00000409438, ENST00000409567, ENST00000409868, ENST00000407639, ENST00000409240	c.1038C>T, c.657C>T, c.999C>T, c.1008C>T, c.657C>T, c.948C>T	p.(=), p.(=), p.(=), p.(=), p.(=)	, ,	, ,					
DCTN1	missense	*ENST00000361874, ENST00000409567, ENST00000407639, ENST00000409438, ENST00000409240, ENST00000394003, ENST00000409868	c.1060G>A, c.1000G>A, c.658G>A, c.658G>A, c.949G>A, c.1039G>A, c.1009G>A	p.Ala354Thr, p.Ala334Thr, p.Ala220Thr, p.Ala220Thr, p.Ala317Thr, p.Ala347Thr, p.Ala337Thr	1, 1, 1, 1, 1, 1, 1	2, 2, 2, 2, 2, 2, 2	45,1,0	272,1,0	194,1,0	194,1,0	194,1,0
DCTN1	***missense***	****ENST00000361874, ENST00000409438, ENST00000394003, ENST00000407639, ENST00000409567, ENST00000409240, ENST00000409868***	***c.1231C>A, c.829C>A, c.1210C>A, c.829C>A, c.1171C>A, c.1120C>A, c.1180C>A***	***p.Arg411Ser, p.Arg277Ser, p.Arg404Ser, p.Arg277Ser, p.Arg391Ser, p.Arg374Ser, p.Arg394Ser***	***0, 0, 0, 0, 0, 0, 0***	***0, 0, 0, 0, 0, 0, 0***	***41,0,0***	***246,2,0***	***167,0,0***	***167,0,0***	***167,0,0***
DCTN1	***missense***	****ENST00000361874, ENST00000409868, ENST00000409567, ENST00000409240, ENST00000394003, ENST00000409438, ENST00000407639***	***c.1480G>A, c.1429G>A, c.1420G>A, c.1369G>A, c.1459G>A, c.1078G>A, c.1078G>A***	***p.Ala494Thr, p.Ala477Thr, p.Ala474Thr, p.Ala457Thr, p.Ala487Thr, p.Ala360Thr, p.Ala360Thr***	***0, 0, 0, 0, 0, 0, 0***	***2, 2, 0, 1, 0, 0, 0***	***37,0,0***	***203,1,0***	***178,1,0***	***4478,1,0***	***6676,6,0***
DCTN1	missense	*ENST00000361874, ENST00000409868,	c.1484G>A, c.1433G>A,	p.Arg495Gln, p.Arg478Gln,	0, 0, 0, 0,	0, 0, 0, 0,	34,7,0	202,9,1	183,5,0	4687,178,1	7578,203,1

		ENST00000409567, ENST00000394003, ENST00000409240, ENST00000407639, ENST00000409438	c.1424G>A, c.1463G>A, c.1373G>A, c.1082G>A, c.1082G>A	p.Arg475Gln, p.Arg488Gln, p.Arg458Gln, p.Arg361Gln, p.Arg361Gln	0, 0, 0	0, 0, 0					
DCTN1	synonymous	*ENST00000361874, ENST00000394003, ENST00000409868, ENST00000409567, ENST00000407639, ENST00000409240, ENST00000409438	c.2019C>T, c.1998C>T, c.1968C>T, c.1959C>T, c.1617C>T, c.1908C>T, c.1617C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , ,	, , , , , ,	49,0,0	380,0,0	300,1,0	300,1,0	300,1,0
DCTN1	***missense***	****ENST00000361874, ENST00000409567, ENST00000409240, ENST00000407639, ENST00000409438, ENST00000409868, ENST00000394003***	***c.2209G>C, c.2149G>C, c.2098G>C, c.1807G>C, c.1807G>C, c.2158G>C, c.2188G>C***	***p.Glu737Gln, p.Glu717Gln, p.Glu700Gln, p.Glu603Gln, p.Glu603Gln, p.Glu720Gln, p.Glu730Gln***	***1, 1, 1, 1, 1, 1, 1***	***1, 0, 1, 0, 0, 1, 0***	***49,0,0***	***360,1,0***	***289,1,0***	***289,1,0***	***289,1,0***
DCTN1	missense	*ENST00000361874, ENST00000407639, ENST00000409438, ENST00000409868, ENST00000409567, ENST00000394003, ENST00000409240	c.2339T>C, c.1937T>C, c.1937T>C, c.2288T>C, c.2279T>C, c.2318T>C, c.2228T>C	p.Ile780Thr, p.Ile646Thr, p.Ile646Thr, p.Ile763Thr, p.Ile760Thr, p.Ile773Thr, p.Ile743Thr	1, 1, 1, 1, 1, 1, 1	0, 0, 0, 0, 0, 0, 0	50,0,0	384,1,0	304,0,0	4603,1,0	6806,1,0
DCTN1	missense	*ENST00000361874, ENST00000409438, ENST00000407639, ENST00000394003, ENST00000409240, ENST00000409868, ENST00000409567	c.2353C>T, c.1951C>T, c.1951C>T, c.2332C>T, c.2242C>T, c.2302C>T, c.2293C>T	p.Arg785Trp, p.Arg651Trp, p.Arg651Trp, p.Arg778Trp, p.Arg748Trp, p.Arg768Trp, p.Arg765Trp	1, 1, 1, 1, 1, 1, 1	2, 2, 2, 2, 2, 2, 2	50,0,0	377,1,0	301,0,0	4594,7,0	6795,9,0
DCTN1	synonymous	*ENST00000361874, ENST00000409438,	c.2448A>G, c.2046A>G,	p.(=), p.(=), p.(=), p.(=),	, , , , , ,	, , , , , ,	48,0,0	369,4,0	301,2,0	4907,74,0	7613,281,3

		ENST00000409868, ENST00000407639, ENST00000409240, ENST00000394003, ENST00000409567	c.2397A>G, c.2046A>G, c.2337A>G, c.2427A>G, c.2388A>G	p.(=), p.(=), p.(=)							
DCTN1	missense	*ENST00000361874, ENST00000409438, ENST00000407639, ENST00000409240, ENST00000394003, ENST00000409868, ENST00000409567	c.2551C>G, c.2149C>G, c.2149C>G, c.2440C>G, c.2530C>G, c.2500C>G, c.2491C>G	p.Leu851Val, p.Leu717Val, p.Leu717Val, p.Leu814Val, p.Leu844Val, p.Leu834Val, p.Leu831Val	0, 0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0, 0	27,0,0	212,0,0	222,1,0	600,1,0	1312,2,0
DCTN1	synonymous	*ENST00000361874, ENST00000409240, ENST00000407639, ENST00000409567, ENST00000409868, ENST00000394003, ENST00000409438	c.2559C>T, c.2448C>T, c.2157C>T, c.2499C>T, c.2508C>T, c.2538C>T, c.2157C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , ,	, , , , , ,	30,0,0	222,1,0	216,1,0	216,1,0	216,1,0
DCTN1	splice acceptor	*ENST00000361874, ENST00000409868, ENST00000409567, ENST00000394003, ENST00000409438, ENST00000407639, ENST00000409240	c.2887-2A>G, c.2836-2A>G, c.2827-2A>G, c.2866-2A>G, c.2485-2A>G, c.2485-2A>G, c.2776-2A>G	, , , , , ,	, , , , , ,	, , , , , ,	39,0,0	237,2,0	246,0,0	246,0,0	246,0,0
DCTN1	synonymous	*ENST00000361874, ENST00000409567,	c.2952C>G, c.2892C>G,	p.(=), p.(=), p.(=), p.(=),	, , , , , ,	, , , , , ,	46,0,0	287,0,0	280,1,0	4579,2,0	6782,2,0

		ENST00000409868, ENST00000409240, ENST00000409438, ENST00000407639, ENST00000394003	c.2901C>G, c.2841C>G, c.2550C>G, c.2550C>G, c.2931C>G	p.(=), p.(=), p.(=)							
DCTN1	missense	*ENST00000361874, ENST00000409438, ENST00000394003, ENST00000409240, ENST00000407639, ENST00000409868, ENST00000409567	c.3146G>A, c.2744G>A, c.3125G>A, c.3035G>A, c.2744G>A, c.3095G>A, c.3086G>A	p.Arg1049Gln, p.Arg915Gln, p.Arg1042Gln, p.Arg1012Gln, p.Arg915Gln, p.Arg1032Gln, p.Arg1029Gln	0, 0, 0, 0, 0, 0, 0	2, 2, 2, 2, 2, 2, 2	47,0,0	313,4,0	273,2,0	4930,23,0	7845,24,0
DCTN1	synonymous	*ENST00000361874, ENST00000409868, ENST00000409567, ENST00000409438, ENST00000407639, ENST00000394003, ENST00000409240	c.3501C>T, c.3435C>T, c.3426C>T, c.3084C>T, c.3099C>T, c.3480C>T, c.3375C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , ,	, , , , , ,	45,0,0	299,2,0	224,3,0	224,3,0	224,3,0
DCTN1	missense	*ENST00000361874, ENST00000407639, ENST00000409240, ENST00000394003, ENST00000409438, ENST00000409868, ENST00000409567	c.3746C>T, c.3344C>T, c.3620C>T, c.3725C>T, c.3329C>T, c.3680C>T, c.3671C>T	p.Thr1249Ile, p.Thr1115Ile, p.Thr1207Ile, p.Thr1242Ile, p.Thr1110Ile, p.Thr1227Ile, p.Thr1224Ile	1, 1, 1, 1, 1, 1, 1	0, 0, 0, 0, 0, 0, 0	41,0,0	272,3,0	266,4,0	4905,43,0	7816,48,0
DCTN1	***missense***	****ENST00000361874, ENST00000409438, ENST00000407639, ENST00000409868, ENST00000394003,***	***c.3788T>G, c.3371T>G, c.3386T>G, c.3722T>G, c.3767T>G,***	***p.Val1263Gly, p.Val1124Gly, p.Val1129Gly***	***1, 1, 1, 1, 1, 1, 1***	***2, 1, 1, 2, 1, 1, 2***	***32,3,0***	***135,52,0***	***114,52,0***	***114,52,0***	***114,52,0***

		ENST00000409567, ENST00000409240	***c.3713T>G, c.3662T>G***	***, p.Val1241Gly , p.Val1256Gly , p.Val1238Gly , p.Val1221Gly***							
DCTN1	synonymous	*ENST00000361874, ENST00000394003, ENST00000409567, ENST00000458655, ENST00000417090	c.42C>T, c.42C>T, c.42C>T, c.63C>T, c.54C>T	p.(=), p.(=), p.(=), p.(=), p.(=)	, , , ,	, , , ,	37,0,0	275,1,0	221,1,0	221,1,0	221,1,0
DCTN1	missense	*ENST00000361874, ENST00000407639, ENST00000409567, ENST00000409240, ENST00000409438, ENST00000394003, ENST00000409868	c.586A>G, c.184A>G, c.526A>G, c.475A>G, c.184A>G, c.565A>G, c.535A>G	p.Ile196Val, p.Ile62Val, p.Ile176Val, p.Ile159Val, p.Ile62Val, p.Ile189Val, p.Ile179Val	0, 0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0, 0	18,0,0	120,3,0	104,3,0	4711,69,2	7617,76,2
DCTN1	synonymous	*ENST00000361874, ENST00000409868, ENST00000409567, ENST00000409438, ENST00000409240, ENST00000394003, ENST00000407639	c.597G>A, c.546G>A, c.537G>A, c.195G>A, c.486G>A, c.576G>A, c.195G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , ,	, , , , , ,	18,0,0	121,1,0	97,0,0	4393,0,0	6583,7,0
DCTN1	***missense***	****ENST00000361874, ENST00000394003, ENST00000409868, ENST00000409438, ENST00000409240, ENST00000409567, ENST00000407639***	***c.713G>T, c.692G>T, c.662G>T, c.311G>T, c.602G>T, c.653G>T, c.311G>T***	***p.Arg238Leu, p.Arg231Leu, p.Arg221Leu, p.Arg104Leu, p.Arg201Leu, p.Arg218Leu, p.Arg104Leu***	***1, 1, 1, 1, 1, 1, 1***	***2, 2, 2, 2, 2, 2, 2***	***26,0,0***	***164,0,0***	***146,1,0***	***146,1,0***	***146,1,0***
DCTN1	***synonymous***	****ENST00000361874,***	***c.801G>A,***	***p.(=), p.(=),***	***, , , ,***	***, , , ,***	***16,0,0***	***128,1,0***	***143,0,0***	***143,0,0***	***143,0,0***

		ENST00000407639, ENST00000409567, ENST00000409438, ENST00000394003, ENST00000409868, ENST00000409240	***c.399G>A, c.741G>A, c.399G>A, c.780G>A, c.750G>A, c.690G>A***	***p.(=), p.(=), p.(=), p.(=), p.(=)***	***, ,***	***, ,***					
DCTN1	missense	*ENST00000361874, ENST00000458655, ENST00000417090, ENST00000409868, ENST00000413111, ENST00000437375, ENST00000394003, ENST00000421392, ENST00000440727, ENST00000454119, ENST00000409567, ENST00000409240, ENST00000449655	c.82C>T, c.103C>T, c.94C>T, c.31C>T, c.31C>T, c.31C>T, c.82C>T, c.31C>T, c.31C>T, c.31C>T, c.82C>T, c.31C>T, c.31C>T	p.Arg28Trp, p.Arg35Trp, p.Arg32Trp, p.Arg11Trp, p.Arg11Trp, p.Arg11Trp, p.Arg28Trp, p.Arg11Trp, p.Arg11Trp, p.Arg11Trp, p.Arg28Trp, p.Arg11Trp, p.Arg11Trp	1, 1, 1, 1, 1, 1, 1, 1, 1, 1, 1, 1, 1	1, 1, 2, 1, 1, 1, 1, 1, 1, 1, 1, 1, 1	45,0,0	308,0,0	235,2,0	235,2,0	235,2,0
DPP6	***synonymous***	****ENST00000377770, ENST00000406326***	***c.114C>T, c.114C>T***	***p.(=), p.(=)***	***,***	***,***	***0,1,0***	***0,1,0***	***0,0,0***	***0,0,0***	***0,0,0***
DPP6	synonymous	*ENST00000377770, ENST00000332007, ENST00000404039, ENST00000427557	c.1458C>T, c.1272C>T, c.1266C>T, c.1137C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	46,1,0	364,2,0	293,1,0	4469,8,0	6544,11,0
DPP6	missense	*ENST00000377770, ENST00000332007, ENST00000427557, ENST00000404039	c.1459G>A, c.1273G>A, c.1138G>A, c.1267G>A	p.Val487Met, p.Val425Met, p.Val380Met, p.Val423Met	1, 1, 1, 1	2, 2, 2, 2	49,0,0	364,0,0	298,1,0	298,1,0	298,1,0
DPP6	***missense***	****ENST00000377770, ENST00000427557, ENST00000332007, ENST00000404039***	***c.1616T>C, c.1295T>C, c.1430T>C, c.1424T>C***	***p.Phe539Ser, p.Phe432Ser, p.Phe477Ser, p.Phe475Ser***	***1, 1, 1, 1***	***2, 1, 1, 2***	***50,0,0***	***351,0,0***	***296,1,0***	***296,1,0***	***296,1,0***
DPP6	missense	*ENST00000377770, ENST00000404039,	c.1711A>C, c.1519A>C,	p.Lys571Gln, p.Lys507Gln,	0, 0, 0, 0	0, 0, 0, 0	50,0,0	389,0,0	294,1,0	4770,8,0	7337,25,0

		ENST00000427557, ENST00000332007	c.1390A>C, c.1525A>C	p.Lys464Gln, p.Lys509Gln							
DPP6	synonymous	*ENST00000377770, ENST00000427557, ENST00000332007, ENST00000404039	c.1896A>G, c.1575A>G, c.1710A>G, c.1704A>G	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	2,9,0	18,56,10	29,49,2	1087,2329,1226	1343,3452,2626
DPP6	synonymous	*ENST00000377770, ENST00000332007, ENST00000427557, ENST00000404039	c.1911G>A, c.1725G>A, c.1590G>A, c.1719G>A	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	5,10,1	33,72,14	35,58,4	1971,2118,580	3968,2838,652
DPP6	synonymous	*ENST00000377770, ENST00000332007, ENST00000404039, ENST00000427557	c.2127T>C, c.1941T>C, c.1935T>C, c.1806T>C	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	28,16,2	197,118,14	170,90,17	3275,1447,165	4371,2723,627
DPP6	synonymous	*ENST00000377770, ENST00000427557, ENST00000404039, ENST00000332007	c.2205C>T, c.1884C>T, c.2013C>T, c.2019C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	49,0,0	344,4,0	261,7,0	4756,43,0	7352,114,7
DPP6	***synonymous***	****ENST00000377770, ENST00000404039, ENST00000427557, ENST00000332007***	***c.2295A>G , c.2103A>G , c.1974A>G , c.2109A>G***	***p.(=), p.(=), p.(=), p.(=)***	***, , ,***	***, , ,***	***49,0,0***	***356,2,0***	***296,0,0***	***296,0,0***	***296,0,0***
DPP6	synonymous	*ENST00000377770, ENST00000404039, ENST00000427557, ENST00000332007	c.2331C>T, c.2139C>T, c.2010C>T, c.2145C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	48,0,0	372,0,0	286,0,0	4841,6,0	7577,13,0
DPP6	synonymous	*ENST00000377770, ENST00000404039, ENST00000332007, ENST00000427557	c.2373C>T, c.2181C>T, c.2187C>T, c.2052C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	36,13,0	313,58,3	236,43,4	3799,975,60	6182,1258,109
DPP6	***splice acceptor***	****ENST00000377770, ENST00000332007,***	***c.2378-1G>A,***	***, , ,***	***, , ,***	***, , ,***	***48,0,0***	***358,6,0***	***254,2,0***	***254,2,0***	***254,2,0***

		ENST00000427557, ENST00000404039	***c.2192-1G>A, c.2057-1G>A, c.2186-1G>A***								
DPP6	synonymous	*ENST00000377770, ENST00000427557, ENST00000332007, ENST00000404039	c.2427G>A, c.2106G>A, c.2241G>A, c.2235G>A	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	34,14,2	299,86,6	226,62,5	3578,1116,101	4732,2251,422
DPP6	missense	*ENST00000377770, ENST00000332007, ENST00000404039, ENST00000427557	c.2561T>C, c.2375T>C, c.2369T>C, c.2240T>C	p.Leu854Pro, p.Leu792Pro, p.Leu790Pro, p.Leu747Pro	1, 0, 0, 0	0, 0, 0, 0	23,21,6	211,144,29	154,122,24	2637,1875,406	3479,3231,1056
DPP6	synonymous	*ENST00000377770, ENST00000404039, ENST00000332007	c.657C>T, c.465C>T, c.471C>T	p.(=), p.(=), p.(=)	, ,	, ,	4,28,17	39,167,182	31,127,124	419,1939,2397	910,3103,3299
DPP6	synonymous	*ENST00000377770, ENST00000404039, ENST00000332007	c.666C>T, c.474C>T, c.480C>T	p.(=), p.(=), p.(=)	, ,	, ,	48,2,0	382,9,0	282,3,0	4707,46,0	7261,52,0
DPP6	synonymous	*ENST00000377770, ENST00000332007, ENST00000404039	c.723A>G, c.537A>G, c.531A>G	p.(=), p.(=), p.(=)	, ,	, ,	0,8,41	4,52,324	3,49,245	51,812,3908	297,1822,5229
DPP6	***missense***	****, ENST00000406326***	***c.785C>T***	***p.Ser262Leu***			***0,0,0***	***0,0,0***	***0,1,0***	***1946,400,23***	***3326,597,34***
DPP6	synonymous	*ENST00000377770, ENST00000404039, ENST00000332007, ENST00000427557	c.879T>C, c.687T>C, c.693T>C, c.558T>C	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	388,1,0	296,0,0	296,0,0	296,0,0
DPP6	missense, splice region	*ENST00000377770, ENST00000332007, ENST00000427557, ENST00000404039	c.883G>A, c.697G>A, c.562G>A, c.691G>A	p.Glu295Lys, p.Glu233Lys, p.Glu188Lys, p.Glu231Lys	1, 1, 1, 1	2, 2, 2, 2	50,0,0	389,1,0	296,0,0	296,0,0	296,0,0
DPP6	synonymous	*ENST00000377770, ENST00000332007, ENST00000427557,	c.945C>T, c.759C>T, c.624C>T,	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	45,3,0	325,44,2	248,49,1	4080,736,32	6300,1226,61

		ENST00000404039	c.753C>T								
DPP6	synonymous	*ENST00000377770, ENST00000404039, ENST00000427557, ENST00000332007	c.948C>T, c.756C>T, c.627C>T, c.762C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	47,0,0	371,1,0	292,0,0	4460,0,0	6472,3,0
DPP6	***missense***	****, ENST00000406326***	***c.982T>G***	***p.Cys328Gly***			***0,0,0***	***0,0,0***	***0,0,0***	***0,0,0***	***0,0,0***
ELP3	synonymous	*ENST00000256398, ENST00000524103, ENST00000380353, ENST00000537665, ENST00000521015, ENST00000542181	c.1068G>A, c.852G>A, c.792G>A, c.711G>A, c.1026G>A, c.681G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	50,0,0	386,1,0	298,1,0	298,1,0	298,1,0
ELP3	missense	*ENST00000256398, ENST00000517975, ENST00000523357, ENST00000380353, ENST00000524103, ENST00000521015, ENST00000537665, ENST00000542181	c.1318G>A, c.97G>A, c.115G>A, c.1042G>A, c.1102G>A, c.1276G>A, c.961G>A, c.931G>A	p.Glu440Lys, p.Glu33Lys, p.Glu39Lys, p.Glu348Lys, p.Glu368Lys, p.Glu426Lys, p.Glu321Lys, p.Glu311Lys	1, 1, 1, 1, 1, 1, 1, 1	1, 2, 2, 1, 1, 1, 1, 1	49,0,0	363,1,0	259,2,0	4936,3,0	7852,3,0
ELP3	missense	*ENST00000256398, ENST00000521015, ENST00000521570, ENST00000524024, ENST00000521099, ENST00000520288, ENST00000520270	c.139G>A, c.97G>A, c.97G>A, c.139G>A, c.97G>A, c.97G>A, c.97G>A	p.Ala47Thr, p.Ala33Thr, p.Ala33Thr, p.Ala47Thr, p.Ala33Thr, p.Ala33Thr, p.Ala33Thr	1, 1, 1, 1, 1, 1, 1	0, 0, 0, 0, 0, 0, 0	50,0,0	380,3,0	302,0,0	4600,2,0	6803,2,0
ELP3	missense	*ENST00000256398, ENST00000380353, ENST00000521015, ENST00000542181, ENST00000524103, ENST00000523357, ENST00000537665	c.1459C>T, c.1183C>T, c.1417C>T, c.1072C>T, c.1243C>T, c.256C>T, c.1102C>T	p.Arg487Trp, p.Arg395Trp, p.Arg473Trp, p.Arg358Trp, p.Arg415Trp, p.Arg86Trp, p.Arg368Trp	1, 0, 1, 0, 0, 0, 0	2, 2, 2, 2, 2, 2, 2	46,0,0	330,1,0	199,1,0	4870,8,0	7786,8,0
ELP3	missense	*ENST00000256398,	c.206G>T,	p.Arg69Leu,	1, 1,	0, 0,	50,0,0	380,1,0	294,0,0	294,0,0	294,0,0

		ENST00000520288, ENST00000521570, ENST00000521015, ENST00000521099, ENST00000524024, ENST00000520270	c.164G>T, c.164G>T, c.164G>T, c.164G>T, c.206G>T, c.164G>T	p.Arg55Leu, p.Arg55Leu, p.Arg55Leu, p.Arg55Leu, p.Arg69Leu, p.Arg55Leu	1, 1, 1, 1, 1	0, 0, 0, 1, 0					
ELP3	initiator codon, missense	*ENST00000256398, ENST00000521099, ENST00000524103, ENST00000380353, ENST00000520288, ENST00000542181, ENST00000521570, ENST00000524024, ENST00000521015, ENST00000520270, ENST00000537665	c.326G>A, c.284G>A, c.110G>A, c.50G>A, c.284G>A, c.3G>A, c.284G>A, c.326G>A, c.284G>A, c.284G>A, c.33G>A	p.Cys109Tyr, p.Cys95Tyr, p.Cys37Tyr, p.Cys17Tyr, p.Cys95Tyr, p.Met1?, p.Cys95Tyr, p.Cys109Tyr, p.Cys95Tyr, p.Cys95Tyr, p.Met11Ile	1, 1, 1, 1, 1, , 1, 1, 1, 1, 1	2, 2, 2, 2, 2, 0, 2, 2, 2, 2, 0	49,1,0	389,1,0	278,0,0	4577,1,0	6780,1,0
ELP3	***splice region, synonymous***	****, ENST00000523357***	***c.366G>A***	***p.(=)***			***0,0,0***	***0,0,0***	***0,0,0***	***287,82,9***	***833,223,35***
FGGY	synonymous	*ENST00000371218, ENST00000303721, ENST00000371210, ENST00000371212	c.1053A>G, c.1053A>G, c.156A>G, c.789A>G	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	389,1,0	281,2,0	4580,3,0	6783,3,0
FGGY	splice donor	*ENST00000371218, ENST00000303721, ENST00000371212, ENST00000371210	c.1221+2T>C, c.1221+2T>C, c.957+2T>C, c.324+2T>C	, , ,	, , ,	, , ,	49,1,0	384,6,0	287,6,0	4924,47,0	7835,52,0
FGGY	missense	*ENST00000371218, ENST00000582567, ENST00000413489,	c.129T>G, c.129T>G, c.129T>G,	p.Asn43Lys, p.Asn43Lys, p.Asn43Lys,	0, 0, 0, 0, 0	0, 0, 0, 0, 0	17,24,8	143,187,31	117,139,46	1848,1892,521	2391,3053,1098

		ENST00000303721, ENST00000371212	c.129T>G, c.129T>G	p.Asn43Lys, p.Asn43Lys							
FGGY	synonymous	*ENST00000371218, ENST00000303721, ENST00000371212, ENST00000371210	c.1560C>T, c.1488C>T, c.1224C>T, c.591C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	46,0,0	310,0,0	285,1,0	285,1,0	285,1,0
FGGY	missense	*ENST00000371218, ENST00000371210, ENST00000303721, ENST00000371212	c.1716G>A, c.747G>A, c.1644G>A, c.1380G>A	p.Met572Ile, p.Met249Ile, p.Met548Ile, p.Met460Ile	0, 0, 0, 0	1, 1, 1, 1	48,0,0	375,1,0	285,0,0	285,0,0	285,0,0
FGGY	missense	*ENST00000371218, ENST00000413489, ENST00000303721, ENST00000371212	c.188G>C, c.188G>C, c.188G>C, c.188G>C	p.Cys63Ser, p.Cys63Ser, p.Cys63Ser, p.Cys63Ser	1, 1, 1, 1	1, 1, 1, 0	49,0,0	351,1,0	293,0,0	4251,2,0	6529,5,0
FGGY	***feature elongation, frameshift***	****ENST00000371218, ENST00000303721, ENST00000413489***	***c.411_412insG, c.411_412insG, c.411_412insG***	***p.Val140Glyfs X25, p.Val140Glyfs X25, p.Val140Glyfs X25***	***, ,***	***, ,***	***50,0,0***	***365,1,0***	***299,0,0***	***299,0,0***	***299,0,0***
FGGY	missense	*ENST00000371218, ENST00000303721, ENST00000371212, ENST00000582567, ENST00000413489	c.49G>A, c.49G>A, c.49G>A, c.49G>A, c.49G>A	p.Val17Ile, p.Val17Ile, p.Val17Ile, p.Val17Ile, p.Val17Ile	1, 1, 1, 1, 1	2, 2, 2, 2, 2	47,1,0	308,2,0	225,1,0	3800,8,0	5367,9,0
FGGY	synonymous	*ENST00000371218, ENST00000371212, ENST00000413489, ENST00000303721	c.516G>A, c.252G>A, c.516G>A, c.516G>A	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	17,24,9	137,199,49	91,151,44	1899,2327,737	2388,3679,1812
FGGY	synonymous	*ENST00000371218, ENST00000413489, ENST00000371212, ENST00000303721	c.516G>C, c.516G>C, c.252G>C, c.516G>C	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	384,1,0	286,0,0	286,0,0	286,0,0
FGGY	missense	*ENST00000371218, ENST00000303721,	c.614G>A, c.614G>A,	p.Ser205Asn, p.Ser205Asn,	0, 0, 0	0, 0, 0	50,0,0	388,1,0	284,0,0	4582,2,0	6785,2,0

		ENST00000371212	c.350G>A	p.Ser117Asn							
FGGY	***missense***	****ENST00000371218, ENST00000303721, ENST00000371212***	***c.622A>G, c.622A>G, c.358A>G***	***p.Lys208Glu, p.Lys208Glu, p.Lys120Glu***	***0, 0, 0***	***0, 0, 0***	***50,0,0***	***389,0,0***	***280,1,0***	***280,1,0***	***280,1,0***
FGGY	synonymous	*ENST00000371218, ENST00000371212, ENST00000303721	c.630T>A, c.366T>A, c.630T>A	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	387,1,0	281,0,0	281,0,0	281,0,0
FGGY	missense, splice region	*ENST00000371218, ENST00000371212, ENST00000303721	c.800G>T, c.536G>T, c.800G>T	p.Gly267Val, p.Gly179Val, p.Gly267Val	1, 1, 1	2, 2, 2	49,0,0	359,1,0	294,4,0	4958,18,0	7872,20,0
FGGY	missense	*ENST00000371218, ENST00000303721, ENST00000371212	c.857C>T, c.857C>T, c.593C>T	p.Thr286Met, p.Thr286Met, p.Thr198Met	0, 0, 1	1, 2, 1	39,5,2	292,55,3	238,46,8	4364,579,27	7223,635,28
FGGY	splice region, synonymous	*ENST00000371218, ENST00000371212, ENST00000303721, ENST00000371210	c.903G>T, c.639G>T, c.903G>T, c.6G>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	357,1,0	302,1,0	4600,3,0	6803,3,0
FGGY	missense	*ENST00000371218, ENST00000303721, ENST00000371212, ENST00000371210	c.979A>C, c.979A>C, c.715A>C, c.82A>C	p.Asn327His, p.Asn327His, p.Asn239His, p.Asn28His	1, 1, 1, 1	0, 2, 2, 2	50,0,0	383,1,0	295,0,0	4586,9,0	6789,9,0
FIG4	missense	*ENST00000230124, ENST00000441478	c.1090A>T, c.259A>T	p.Met364Leu, p.Met87Leu	0, 0	0, 0	47,3,0	363,24,1	271,24,0	4611,355,7	7272,592,25
FIG4	missense	*ENST00000230124, ENST00000454215	c.122T>C, c.59T>C	p.Ile41Thr, p.Ile20Thr	1, 1	2, 2	46,0,0	360,1,0	238,2,0	4899,18,0	7814,19,0
FIG4	synonymous	*ENST00000230124, ENST00000441478	c.1305G>C, c.474G>C	p.(=), p.(=)	,	,	50,0,0	388,1,0	281,0,0	4579,2,0	6782,2,0
FIG4	missense	*ENST00000230124, ENST00000441478	c.1426C>T, c.595C>T	p.Arg476Cys, p.Arg199Cys	1, 1	0, 0	47,0,0	354,1,0	244,0,0	622,0,0	1334,1,0
FIG4	synonymous	*ENST00000230124, ENST00000441478	c.1525C>T, c.694C>T	p.(=), p.(=)	,	,	49,0,0	387,1,0	287,0,0	4586,1,0	6789,1,0
FIG4	synonymous	*ENST00000230124, ENST00000441478	c.1527G>A, c.696G>A	p.(=), p.(=)	,	,	50,0,0	387,0,0	287,1,0	287,1,0	287,1,0
FIG4	missense	*ENST00000230124, ENST00000441478,	c.1793T>A, c.962T>A,	p.Phe598Tyr, p.Phe321Tyr,	0, 0, 0	0, 0, 0	38,0,0	336,0,0	219,1,0	219,1,0	219,1,0

		ENST00000415980	c.210T>A	p.Phe71Tyr							
FIG4	missense	*ENST00000230124, ENST00000441478	c.1961T>C, c.1130T>C	p.Val654Ala, p.Val377Ala	0, 0	0, 0	37,11,2	287,96,7	184,77,9	3552,1263,132	4030,2504,1329
FIG4	***missense***	****ENST00000230124, ENST00000419951, ENST00000441478***	***c.2200G>A , c.121G>A, c.1369G>A***	***p.Glu734Lys, p.Glu41Lys, p.Glu457Lys***	***0, , 0***	***0, 0, 1***	***41,0,0***	***216,1,0***	***216,1,0***	***4514,3,0***	***6717,3,0***
FIG4	synonymous	*ENST00000230124, ENST00000441478	c.2559G>A, c.1641G>A	p.(=), p.(=)	,	,	13,22,15	107,190,79	92,139,49	1738,2355,865	3648,3195,1031
FIG4	***splice acceptor***	****ENST00000230124, ENST00000454215, ENST00000441478, ENST00000368941***	***c.447-2A>T, c.384-2A>T, c.9-2A>T, c.216-2A>T***	***, , ,***	***, , ,***	***, , ,***	***43,1,0***	***320,6,0***	***220,1,0***	***220,1,0***	***220,1,0***
FUS	synonymous	*ENST00000254108, ENST00000380244, ENST00000568685	c.1080C>T, c.1077C>T, c.1083C>T	p.(=), p.(=), p.(=)	, ,	, ,	38,0,0	213,2,0	166,2,0	4841,5,0	7750,5,0
FUS	***inframe deletion***	****ENST00000254108, ENST00000568685, ENST00000380244***	***c.1204_1206delAGT, c.1207_1209delAGT, c.1201_1203delAGT***	***p.Ser402del, p.Ser403del, p.Ser401del***	***, ,***	***, ,***	***50,0,0***	***364,1,0***	***296,0,0***	***296,0,0***	***296,0,0***
FUS	synonymous	*ENST00000254108, ENST00000380244, ENST00000568685	c.132C>T, c.132C>T, c.132C>T	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	376,0,0	303,1,0	4601,3,0	6798,3,0
FUS	synonymous	*ENST00000254108, ENST00000380244, ENST00000568685	c.147C>A, c.147C>A, c.147C>A	p.(=), p.(=), p.(=)	, ,	, ,	29,13,6	173,158,44	133,137,33	2475,2086,420	4866,2551,474
FUS	synonymous	*ENST00000254108, ENST00000568685, ENST00000380244	c.153C>T, c.153C>T, c.153C>T	p.(=), p.(=), p.(=)	, ,	, ,	49,0,0	376,0,0	303,3,0	4947,37,0	7383,480,31
FUS	***synonymous***	****ENST00000254108, ENST00000380244, ENST00000568685***	***c.1545T>G, c.1542T>G, c.1548T>G***	***p.(=), p.(=), p.(=)***	***, ,***	***, ,***	***38,0,0***	***259,6,0***	***227,6,0***	***227,6,0***	***227,6,0***

FUS	synonymous	*ENST00000254108, ENST00000568685, ENST00000380244	c.1566G>A, c.1569G>A, c.1563G>A	p.(=), p.(=), p.(=)	, ,	, ,	47,0,0	337,1,0	280,0,0	4940,18,0	7845,23,0
FUS	missense	*ENST00000254108, ENST00000568685, ENST00000380244	c.1574C>T, c.1577C>T, c.1571C>T	p.Pro525Leu, p.Pro526Leu, p.Pro524Leu	1, 1, 1	1, 2, 2	43,0,0	317,2,0	250,0,0	250,0,0	250,0,0
FUS	***inframe deletion***	****ENST00000254108, ENST00000568685, ENST00000380244***	***c.165_167 delTTC, c.165_167 delTTC, c.165_167 delTTC***	***p.Ser56del, p.Ser56del, p.Ser56del***	***, ,***	***, ,***	***48,0,0***	***372,0,0***	***304,1,0***	***304,1,0***	***304,1,0***
FUS	***synonymous***	****ENST00000254108, ENST00000380244, ENST00000568685***	***c.231G>C, c.228G>C, c.231G>C***	***p.(=), p.(=), p.(=)***	***, ,***	***, ,***	***22,10,0***	***95,122,0***	***77,140,0***	***77,140,0***	***77,140,0***
FUS	***missense***	****ENST00000254108, ENST00000568685, ENST00000380244***	***c.232A>C, c.232A>C, c.229A>C***	***p.Thr78Pro, p.Thr78Pro, p.Thr77Pro***	***0, 0, 0***	***, ,***	***31,2,0***	***141,36,0***	***151,36,0***	***151,36,0***	***151,36,0***
FUS	***synonymous***	****ENST00000254108, ENST00000568685, ENST00000380244***	***c.234T>C, c.234T>C, c.231T>C***	***p.(=), p.(=), p.(=)***	***, ,***	***, ,***	***33,0,0***	***173,10,0***	***184,9,0***	***184,9,0***	***184,9,0***
FUS	synonymous	*ENST00000254108, ENST00000568685, ENST00000380244	c.291C>T, c.291C>T, c.288C>T	p.(=), p.(=), p.(=)	, ,	, ,	8,19,15	79,139,72	60,141,71	1000,2421,1529	2398,3518,1944
FUS	synonymous	*ENST00000254108, ENST00000380244, ENST00000568685	c.339C>T, c.336C>T, c.339C>T	p.(=), p.(=), p.(=)	, ,	, ,	49,0,0	318,0,0	277,2,0	277,2,0	277,2,0
FUS	***missense***	****ENST00000254108, ENST00000380244, ENST00000568685***	***c.422A>C, c.419A>C, c.422A>C***	***p.Gln141Pro, p.Gln140Pro, p.Gln141Pro***	***0, 0, 0***	***, ,***	***47,0,0***	***332,1,0***	***285,0,0***	***285,0,0***	***285,0,0***
FUS	***missense***	****ENST00000254108, ENST00000380244, ENST00000568685***	***c.423A>T, c.420A>T, c.423A>T***	***p.Gln141His, p.Gln140His, p.Gln141His***	***0, 0, 0***	***, ,***	***47,0,0***	***332,1,0***	***286,0,0***	***286,0,0***	***286,0,0***
FUS	missense	*ENST00000254108, ENST00000380244,	c.74A>G, c.74A>G,	p.Tyr25Cys, p.Tyr25Cys,	1, 0, 1	, ,	49,0,0	368,0,0	286,1,0	286,1,0	286,1,0

		ENST00000568685	c.74A>G	p.Tyr25Cys							
GRN	missense	*ENST00000053867, ENST00000586443, ENST00000589265	c.1297C>T, c.738C>T, c.826C>T	p.Arg433Trp, p.Arg247Trp, p.Arg276Trp	1, 1, 1	1, 1, 1	26,0,0	111,2,0	85,3,0	4734,32,0	7644,38,0
GRN	***synonymous***	****ENST00000053867, ENST00000589265***	***c.1695C>T, c.1224C>T***	***p.(=), p.(=)***	***,***	***,***	***5,0,0***	***32,2,0***	***24,3,0***	***24,3,0***	***24,3,0***
GRN	***missense***	****ENST00000053867, ENST00000589265***	***c.1762G>T, c.1291G>T***	***p.Ala588Ser, p.Ala431Ser***	***1, 1***	***1, 1***	***1,0,0***	***2,1,0***	***5,3,0***	***5,3,0***	***5,3,0***
GRN	synonymous	*ENST00000053867, ENST00000592783, ENST00000591740, ENST00000587518, ENST00000593167, ENST00000588143, ENST00000589265, ENST00000587387	c.384T>C, c.384T>C, c.384T>C, c.384T>C, c.384T>C, c.384T>C, c.384T>C, c.426T>C	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , , ,	, , , , , , ,	49,1,0	337,16,0	290,13,1	4708,269,5	7455,419,24
GRN	***missense***	****ENST00000053867, ENST00000586443, ENST00000588237***	***c.693C>G, c.134C>G, c.495C>G***	***p.Cys231Trp, p.Cys45Trp, p.Cys165Trp***	***1, 1, 1***	***2, 2, 2***	***40,0,0***	***229,3,0***	***207,5,0***	***207,5,0***	***207,5,0***
GRN	synonymous	*ENST00000053867, ENST00000586443, ENST00000588237	c.705C>A, c.146C>A, c.507C>A	p.(=), p.(=), p.(=)	, ,	, ,	37,0,0	232,0,0	206,1,0	4506,1,0	6707,3,0
GRN	missense	*ENST00000053867, ENST00000589265, ENST00000586443	c.970G>A, c.499G>A, c.411G>A	p.Ala324Thr, p.Ala167Thr, p.Ala138Thr	0, 0, 0	0, 0, 0	37,0,0	212,1,0	169,2,0	4457,14,0	6654,20,0
GRN	synonymous	*ENST00000053867, ENST00000585512, ENST00000589265, ENST00000593167, ENST00000589536, ENST00000587518, ENST00000587109, ENST00000591740, ENST00000588143, ENST00000588237,	c.99C>T, c.99C>T, c.99C>T, c.99C>T, c.99C>T, c.99C>T, c.99C>T, c.99C>T, c.99C>T, c.99C>T,	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , , , , , ,	, , , , , , , , , ,	6,0,0	50,4,0	59,2,0	4698,41,0	7612,43,0

		ENST00000587387, ENST00000592783	c.99C>T, c.99C>T								
HFE	synonymous	*ENST00000357618, ENST00000317896, ENST00000461397, ENST00000309234, ENST00000470149, ENST00000336625	c.138G>A, c.138G>A, c.138G>A, c.138G>A, c.138G>A, c.138G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	50,0,0	388,0,0	305,1,0	305,1,0	305,1,0
HFE	missense	*ENST00000357618, ENST00000309234, ENST00000336625, ENST00000317896, ENST00000461397, ENST00000470149, ENST00000397022	c.157G>A, c.157G>A, c.157G>A, c.157G>A, c.157G>A, c.157G>A, c.88G>A	p.Val53Met, p.Val53Met, p.Val53Met, p.Val53Met, p.Val53Met, p.Val53Met, p.Val30Met	1, 1, 1, 1, 1, 1, 1	1, 1, 0, 0, 1, 1, 2	50,0,0	387,0,0	301,1,0	4979,1,0	7892,4,0
HFE	missense	*ENST00000357618, ENST00000397022, ENST00000309234, ENST00000461397, ENST00000470149, ENST00000317896, ENST00000336625	c.187C>G, c.118C>G, c.187C>G, c.187C>G, c.187C>G, c.187C>G, c.187C>G	p.His63Asp, p.His40Asp, p.His63Asp, p.His63Asp, p.His63Asp, p.His63Asp, p.His63Asp	0, 0, 0, 0, 0, 0, 1	0, 0, 0, 0, 0, 0, 0	33,16,1	255,124,9	201,97,6	3558,1323,101	6275,1515,108
HFE	***missense***	****ENST00000357618, ENST00000353147, ENST00000309234, ENST00000488199, ENST00000349999, ENST00000352392, ENST00000461397, ENST00000470149, ENST00000336625, ENST00000317896, ENST00000397022***	***c.18G>C, c.18G>C, c.18G>C, c.18G>C, c.18G>C, c.18G>C, c.18G>C, c.18G>C, c.18G>C, c.18G>C, c.18G>C***	***p.Arg6Ser, p.Arg6Ser, p.Arg6Ser, p.Arg6Ser, p.Arg6Ser, p.Arg6Ser, p.Arg6Ser, p.Arg6Ser, p.Arg6Ser, p.Arg6Ser, p.Arg6Ser***	***0, 1, 0, 1, 1, 1, 0, 0, 1, 1, 1***	***0, 1, 0, 1, 1, , 0, 0, 1, 1, 0***	***12,0,0***	***61,2,0***	***43,0,0***	***4336,6,0***	***6538,7,0***
HFE	missense	*ENST00000357618, ENST00000317896,	c.193A>T, c.193A>T,	p.Ser65Cys, p.Ser65Cys,	1, 1, 1, 1,	2, 0, 2, 0,	48,2,0	371,16,0	291,13,0	4829,150,3	7728,167,3

		ENST00000470149, ENST00000336625, ENST00000397022, ENST00000309234, ENST00000461397	c.193A>T, c.193A>T, c.124A>T, c.193A>T, c.193A>T	p.Ser65Cys, p.Ser65Cys, p.Ser42Cys, p.Ser65Cys, p.Ser65Cys	1, 1, 1	1, 2, 2					
HFE	missense	*ENST00000357618, ENST00000488199, ENST00000349999, ENST00000309234, ENST00000461397, ENST00000470149, ENST00000397022	c.502G>C, c.238G>C, c.238G>C, c.502G>C, c.502G>C, c.502G>C, c.433G>C	p.Glu168Gln, p.Glu80Gln, p.Glu80Gln, p.Glu168Gln, p.Glu168Gln, p.Glu168Gln, p.Glu145Gln	1, 1, 1, 0, 1, 0, 1	2, 0, 1, 2, 2, 2, 1	44,0,0	264,0,0	224,2,0	4897,7,0	7813,7,0
HFE	***missense***	****ENST00000357618, ENST00000349999, ENST00000309234, ENST00000353147, ENST00000461397, ENST00000470149, ENST00000488199, ENST00000352392, ENST00000317896, ENST00000397022, ENST00000336625***	***c.50C>T, c.50C>T, c.50C>T, c.50C>T, c.50C>T, c.50C>T, c.50C>T, c.50C>T, c.50C>T, c.50C>T, c.50C>T***	***p.Thr17Ile, p.Thr17Ile, p.Thr17Ile, p.Thr17Ile, p.Thr17Ile, p.Thr17Ile, p.Thr17Ile, p.Thr17Ile, p.Thr17Ile, p.Thr17Ile, p.Thr17Ile***	***1, 1, 1, 1, 1, 1, 1, 1, 1, 1, 1***	***1, 2, 1, 1, 1, 1, 2, , 2, 2, 2***	***9,1,0***	***45,0,0***	***29,1,0***	***4317,12,0***	***6519,12,0***
HFE	missense	*ENST00000357618, ENST00000336625, ENST00000470149, ENST00000317896, ENST00000309234, ENST00000397022, ENST00000353147, ENST00000488199, ENST00000349999, ENST00000461397	c.670C>T, c.352C>T, c.661C>T, c.394C>T, c.670C>T, c.601C>T, c.130C>T, c.364C>T, c.406C>T, c.628C>T	p.Arg224Trp, p.Arg118Trp, p.Arg221Trp, p.Arg132Trp, p.Arg224Trp, p.Arg201Trp, p.Arg44Trp, p.Arg122Trp, p.Arg136Trp, p.Arg210Trp	1, 1, 1, 1, 1, 1, 1, 1, 1, 1	2, 2, 2, 2, 2, 2, 2, 2, 2, 2	49,1,0	385,1,0	296,0,0	4593,3,0	6795,4,0
HFE	***missense***	****ENST00000357618, ENST00000317896,***	***c.68G>A, c.68G>A,***	***p.Arg23His, p.Arg23His,***	***1, 1, 1, 1,***	***2, 2, 2, 2,***	***7,1,0***	***23,0,0***	***17,0,0***	***4690,4,0***	***7600,9,0***

		ENST00000309234, ENST00000470149, ENST00000336625, ENST00000397022, ENST00000352392, ENST00000353147, ENST00000488199, ENST00000349999, ENST00000461397	***c.68G>A, c.68G>A, c.68G>A, c.68G>A, c.68G>A, c.68G>A, c.68G>A, c.68G>A, c.68G>A***	***p.Arg23His, p.Arg23His, p.Arg23His, p.Arg23His, p.Arg23His, p.Arg23His, p.Arg23His, p.Arg23His, p.Arg23His***	***1, 1, 1, 1, 0, 0, 1***	***2, 2, , 1, 2, 1, 2***					
HFE	missense	*ENST00000357618, ENST00000353147, ENST00000349999, ENST00000309234, ENST00000461397, ENST00000488199, ENST00000336625, ENST00000397022, ENST00000317896, ENST00000470149	c.766G>A, c.226G>A, c.502G>A, c.766G>A, c.724G>A, c.460G>A, c.448G>A, c.697G>A, c.490G>A, c.757G>A	p.Val256Ile, p.Val76Ile, p.Val168Ile, p.Val256Ile, p.Val242Ile, p.Val154Ile, p.Val150Ile, p.Val233Ile, p.Val164Ile, p.Val253Ile	0, 1, 0, 0, 0, 0, 1, 0, 1, 0	0, 0, 0, 0, 0, 1, 0, 0, 0, 0	50,0,0	387,1,0	295,0,0	295,0,0	295,0,0
HFE	missense	*ENST00000357618, ENST00000349999, ENST00000488199, ENST00000317896, ENST00000309234, ENST00000470149, ENST00000397022, ENST00000461397, ENST00000353147, ENST00000336625	c.829G>A, c.565G>A, c.523G>A, c.553G>A, c.829G>A, c.820G>A, c.760G>A, c.787G>A, c.289G>A, c.511G>A	p.Glu277Lys, p.Glu189Lys, p.Glu175Lys, p.Glu185Lys, p.Glu277Lys, p.Glu274Lys, p.Glu254Lys, p.Glu263Lys, p.Glu97Lys, p.Glu171Lys	1, 1, 1, 1, 1, 1, 1, 1, 1, 1	2, 2, 2, 2, 2, 2, 2, 2, 2, 2	50,0,0	382,1,0	299,1,0	4970,8,0	7883,11,0
HFE	missense	*ENST00000357618, ENST00000488199, ENST00000317896, ENST00000470149, ENST00000309234, ENST00000349999,	c.845G>A, c.539G>A, c.569G>A, c.836G>A, c.845G>A, c.581G>A,	p.Cys282Tyr, p.Cys180Tyr, p.Cys190Tyr, p.Cys279Tyr, p.Cys282Tyr, p.Cys194Tyr,	1, 1, 1, 1, 1, 1, 1, 1, 1, 1	1, 2, 2, 1, 1, 1, 2, 1, 2, 2	42,8,0	302,70,3	234,60,7	4345,608,26	7187,681,27

		ENST00000336625, ENST00000461397, ENST00000353147, ENST00000397022	c.527G>A, c.803G>A, c.305G>A, c.776G>A	p.Cys176Tyr, p.Cys268Tyr, p.Cys102Tyr, p.Cys259Tyr							
IFNK	missense	*ENST00000276943	c.263A>G	p.Tyr88Cys	1	2	50,0,0	390,1,0	287,1,0	4961,5,0	7832,50,0
IFNK	synonymous	*ENST00000276943	c.279G>A	p.(=)			50,0,0	391,0,0	287,1,0	287,1,0	287,1,0
IFNK	synonymous	*ENST00000276943	c.282C>T	p.(=)			50,0,0	391,0,0	287,1,0	287,1,0	287,1,0
IFNK	***feature elongation, frameshift***	****ENST00000276943***	***c.30_31insTGTT***	***p.Trp13Phefs X4***			***44,6,0***	***366,21,0***	***269,14,0***	***269,14,0***	***269,14,0***
IFNK	missense	*ENST00000276943	c.397A>G	p.Lys133Glu	0	0	0,0,49	0,4,386	0,2,274	0,83,4870	111,885,6873
IFNK	***stop gained***	****ENST00000276943***	***c.43G>T***	***p.Glu15X***			***46,3,0***	***330,31,0***	***205,37,0***	***205,37,0***	***205,37,0***
IFNK	synonymous	*ENST00000276943	c.486G>C	p.(=)			49,0,0	385,0,0	280,1,0	280,1,0	280,1,0
ITPR2	stop gained	*ENST00000381340	c.1318C>T	p.Arg440X			49,0,0	384,0,0	275,1,0	275,1,0	275,1,0
ITPR2	missense	*ENST00000381340	c.1358C>T	p.Ala453Val	0	0	41,6,1	358,24,0	242,19,1	4318,413,8	6837,446,8
ITPR2	synonymous	*ENST00000381340	c.1674C>T	p.(=)			48,0,0	354,2,0	276,1,0	4396,2,0	6289,2,0
ITPR2	synonymous	*ENST00000381340	c.1857C>T	p.(=)			49,0,0	383,1,0	268,0,0	4702,22,0	7240,23,0
ITPR2	synonymous	*ENST00000381340	c.1905A>G	p.(=)			34,13,1	254,113,13	177,75,7	3241,1332,144	4465,2379,406
ITPR2	synonymous	*ENST00000381340	c.2016A>C	p.(=)			36,14,0	295,83,13	190,89,7	3294,1331,135	5101,1954,275
ITPR2	missense	*ENST00000381340	c.2018T>C	p.Met673Thr	0	0	50,0,0	390,1,0	287,0,0	4382,1,0	6240,1,0
ITPR2	synonymous	*ENST00000381340	c.2055A>G	p.(=)			15,27,8	183,154,51	113,137,33	1984,2198,593	3391,3137,860
ITPR2	synonymous	*ENST00000381340	c.2262A>C	p.(=)			40,9,1	334,52,3	244,60,1	3989,833,50	6660,918,52
ITPR2	synonymous	*ENST00000381340	c.2358C>T	p.(=)			36,14,0	284,83,12	196,92,7	3386,1347,135	5162,2144,306
ITPR2	synonymous	*ENST00000381340	c.2694C>T	p.(=)			30,16,4	223,129,28	141,109,15	2689,1726,310	4946,1980,328
ITPR2	missense	*ENST00000381340	c.2854C>T	p.Pro952Ser	0	0	48,0,0	304,1,0	274,1,0	4496,2,0	6598,2,0
ITPR2	synonymous	*ENST00000381340	c.2883C>T	p.(=)			9,29,6	72,146,94	42,140,54	1234,2385,1211	1857,3718,2048
ITPR2	synonymous	*ENST00000381340	c.3117G>A	p.(=)			47,0,0	368,1,0	245,0,0	4687,2,0	7202,2,0
ITPR2	stop gained	*ENST00000381340	c.3358C>T	p.Arg1120X			49,0,0	363,0,0	256,1,0	256,1,0	256,1,0
ITPR2	synonymous	*ENST00000381340	c.3369A>G	p.(=)			50,0,0	369,10,0	267,8,0	4629,150,1	7222,159,1
ITPR2	missense	*ENST00000381340	c.3485T>G	p.Val1162Gly	0	0	44,1,0	354,1,0	249,0,0	4335,10,0	6173,12,0
ITPR2	missense	*ENST00000381340	c.3539G>A	p.Arg1180Gln	0	0	45,2,0	341,5,0	230,2,0	4631,68,0	7164,78,0
ITPR2	missense	*ENST00000381340	c.3614G>A	p.Arg1205Gln	0	2	50,0,0	370,1,0	266,0,0	266,0,0	266,0,0
ITPR2	missense	*ENST00000381340	c.3635C>T	p.Ala1212Val	0	1	50,0,0	373,0,0	261,3,0	261,3,0	261,3,0

ITPR2	missense	*ENST00000381340	c.3824G>A	p.Arg1275Gln	0	0	47,1,0	367,3,0	256,9,0	4771,51,0	7458,55,0
ITPR2	synonymous	*ENST00000381340	c.4023C>T	p.(=)			50,0,0	388,1,0	293,0,0	293,0,0	293,0,0
ITPR2	synonymous	*ENST00000381340	c.4125C>T	p.(=)			49,1,0	355,32,3	278,21,1	4596,244,6	7288,272,6
ITPR2	synonymous	*ENST00000381340	c.4239C>T	p.(=)			16,29,4	139,183,49	105,133,31	2087,2179,577	4220,2762,624
ITPR2	***synonymous***	****ENST00000381340***	***c.4305A>G***	***p.(=)***			***48,0,0***	***377,1,0***	***248,0,0***	***248,0,0***	***248,0,0***
ITPR2	synonymous	*ENST00000381340	c.4407A>G	p.(=)			17,29,3	144,186,50	106,126,27	2044,2124,557	3976,2691,598
ITPR2	synonymous	*ENST00000381340	c.4482C>T	p.(=)			18,28,2	151,175,43	106,122,25	2194,2023,490	4106,2590,532
ITPR2	splice region, synonymous	*ENST00000381340	c.4962G>A	p.(=)			50,0,0	383,5,0	283,1,0	4804,34,1	7511,39,1
ITPR2	synonymous	*ENST00000381340	c.5118T>C	p.(=)			48,0,0	367,1,0	256,0,0	256,0,0	256,0,0
ITPR2	synonymous	*ENST00000381340	c.5175G>A	p.(=)			27,19,3	227,129,14	166,81,12	2888,1631,230	5279,1829,234
ITPR2	synonymous	*ENST00000381340	c.5331C>T	p.(=)			49,0,0	379,0,0	263,1,0	4360,2,0	6211,2,0
ITPR2	synonymous	*ENST00000381340	c.5569C>T	p.(=)			50,0,0	384,1,0	255,3,0	4698,18,0	7239,19,0
ITPR2	***synonymous***	****ENST00000381340***	***c.570G>C***	***p.(=)***			***48,0,0***	***382,1,0***	***281,3,0***	***281,3,0***	***281,3,0***
ITPR2	missense	*ENST00000381340	c.6022G>A	p.Ala2008Thr	1	2	46,0,0	350,1,0	203,5,0	4302,6,0	6152,6,0
ITPR2	***missense***	****ENST00000381340***	***c.6346G>C***	***p.Ala2116Pro***	***1***	***2***	***48,1,0***	***375,5,0***	***263,4,0***	***263,4,0***	***263,4,0***
ITPR2	missense	*ENST00000381340	c.6389C>T	p.Ser2130Leu	0	0	50,0,0	386,1,0	282,0,0	4404,1,0	6326,1,0
ITPR2	synonymous	*ENST00000381340	c.6390G>A	p.(=)			50,0,0	378,1,0	277,0,0	4402,1,0	6329,1,0
ITPR2	missense	*ENST00000381340	c.6529A>C	p.Lys2177Gln	1	0	50,0,0	390,0,0	278,1,0	4383,7,0	6242,7,0
ITPR2	missense	*ENST00000381340	c.6833T>C	p.Leu2278Pro	1	1	49,1,0	383,0,0	281,1,0	4757,2,0	7318,3,0
ITPR2	synonymous	*ENST00000381340	c.6966T>C	p.(=)			50,0,0	388,3,0	302,2,0	4420,5,0	6317,5,0
ITPR2	splice region, synonymous	*ENST00000381340	c.7401T>G	p.(=)			50,0,0	376,0,0	277,2,0	4357,3,0	6193,3,0
ITPR2	***synonymous***	****ENST00000381340***	***c.7518G>A***	***p.(=)***			***48,0,0***	***377,0,0***	***286,1,0***	***286,1,0***	***286,1,0***
ITPR2	synonymous	*ENST00000381340	c.765A>G	p.(=)			8,32,10	120,175,94	76,152,55	1341,2380,1055	3448,2822,1098
ITPR2	synonymous	*ENST00000381340	c.798G>A	p.(=)			47,2,0	346,30,3	255,13,0	4573,176,1	6327,896,117
ITPR2	missense	*ENST00000381340	c.8002G>A	p.Ala2668Thr	0	0	48,0,0	356,5,0	287,3,0	4884,28,0	7671,33,0
MAPT	missense	*ENST00000344290, ENST00000262410, ENST00000415613, ENST00000571987	c.1108C>T, c.1108C>T, c.1108C>T, c.1108C>T	p.Arg370Trp, p.Arg370Trp, p.Arg370Trp, p.Arg370Trp	1, 1, 1, 1	2, 1, 2, 1	2,2,1	28,37,5	24,44,9	2730,1552,217	5265,1777,229
MAPT	missense	*ENST00000344290, ENST00000415613,	c.1280C>T, c.1280C>T,	p.Ser427Phe, p.Ser427Phe,	1, 1, 1, 1	2, 2, 2, 2	50,0,0	388,3,0	303,4,0	4962,23,0	7874,27,0

		ENST00000571987, ENST00000262410	c.1280C>T, c.1280C>T	p.Ser427Phe, p.Ser427Phe						
MAPT	***missense***	****ENST00000344290, ENST00000415613, ENST00000262410, ENST00000571987***	***c.1306C>A, c.1306C>A, c.1306C>A, c.1306C>A***	***p.Pro436Thr, p.Pro436Thr, p.Pro436Thr, p.Pro436Thr***	***0, 0, 0, 0***	***0, 0, 0, 0***	***50,0,0***	***390,0,0***	***306,1,0***	***306,1,0***	***306,1,0***
MAPT	missense	*ENST00000344290, ENST00000571987, ENST00000415613, ENST00000262410	c.1321T>C, c.1321T>C, c.1321T>C, c.1321T>C	p.Tyr441His, p.Tyr441His, p.Tyr441His, p.Tyr441His	0, 0, 0, 0	0, 0, 0, 0	37,12,1	257,126,8	210,87,9	3204,1578,202	5108,2414,378
MAPT	missense	*ENST00000344290, ENST00000571987, ENST00000415613, ENST00000262410	c.1339T>C, c.1339T>C, c.1339T>C, c.1339T>C	p.Ser447Pro, p.Ser447Pro, p.Ser447Pro, p.Ser447Pro	0, 0, 0, 0	0, 0, 0, 0	36,12,2	250,123,18	190,105,12	3019,1712,254	5642,1992,267
MAPT	***missense***	****ENST00000344290, ENST00000415613, ENST00000351559, ENST00000340799, ENST00000571987, ENST00000420682, ENST00000574436, ENST00000576518, ENST00000431008, ENST00000446361, ENST00000347967, ENST00000535772, ENST00000334239, ENST00000262410***	***c.1405G>A , c.1405G>A , c.454G>A, c.367G>A, c.1405G>A , c.367G>A, c.454G>A, c.247G>A, c.454G>A, c.280G>A, c.172G>A, c.454G>A, c.280G>A, c.1405G>A***	***p.Ala469Thr, p.Ala469Thr, p.Ala152Thr, p.Ala123Thr, p.Ala469Thr, p.Ala123Thr, p.Ala152Thr, p.Ala83Thr, p.Ala152Thr, p.Ala94Thr, p.Ala58Thr, p.Ala152Thr, p.Ala94Thr, p.Ala469Thr***	***0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0***	***0, 0, 0, 0, 0, 0, 0, 1, 0, 0, 1, 0, 0, 0***	***3,0,0***	***36,2,0***	***27,0,0***	***4679,25,1***	***7591,29,1***
MAPT	synonymous	*ENST00000344290, ENST00000576518, ENST00000347967, ENST00000574436, ENST00000420682, ENST00000334239,	c.1479G>A, c.321G>A, c.246G>A, c.528G>A, c.441G>A, c.354G>A,	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=),	, , , , , , , , , , , , ,	, , , , , , , , , , , , ,	4,6,1	37,62,4	37,50,8	2856,1667,250	5518,1910,261

		ENST00000535772, ENST00000262410, ENST00000431008, ENST00000446361, ENST00000351559, ENST00000571987, ENST00000415613, ENST00000340799	c.528G>A, c.1479G>A, c.528G>A, c.354G>A, c.528G>A, c.1479G>A, c.1479G>A, c.441G>A	p.(=), p.(=)							
MAPT	synonymous	*ENST00000344290, ENST00000415613	c.1512T>C, c.1512T>C	p.(=), p.(=)	,	,	27,12,1	154,91,12	146,93,10	2556,1434,215	4607,1643,228
MAPT	synonymous	*ENST00000344290, ENST00000571987, ENST00000351559, ENST00000340799, ENST00000415613, ENST00000334239, ENST00000431008, ENST00000446361, ENST00000576518, ENST00000262410, ENST00000574436, ENST00000347967, ENST00000420682, ENST00000535772	c.1686A>G, c.1632A>G, c.681A>G, c.594A>G, c.1686A>G, c.507A>G, c.681A>G, c.507A>G, c.474A>G, c.1632A>G, c.681A>G, c.399A>G, c.594A>G, c.681A>G	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , , , , , , , , ,	, , , , , , , , , , , , ,	9,7,0	57,31,1	52,24,3	2892,1625,240	5508,1912,253
MAPT	missense	*ENST00000344290, ENST00000347967, ENST00000420682, ENST00000571987, ENST00000334239, ENST00000415613, ENST00000351559, ENST00000262410, ENST00000446361, ENST00000431008, ENST00000340799,	c.1720G>A, c.433G>A, c.628G>A, c.1666G>A, c.541G>A, c.1720G>A, c.715G>A, c.1666G>A, c.541G>A, c.715G>A, c.628G>A,	p.Ala574Thr, p.Ala145Thr, p.Ala210Thr, p.Ala556Thr, p.Ala181Thr, p.Ala574Thr, p.Ala239Thr, p.Ala556Thr, p.Ala181Thr, p.Ala239Thr, p.Ala210Thr,	0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0, 0	18,0,0	92,2,0	66,0,0	4354,12,0	6551,18,0

		ENST00000535772, ENST00000576518, ENST00000574436	c.715G>A, c.508G>A, c.715G>A	p.Ala239Thr, p.Ala170Thr, p.Ala239Thr							
MAPT	synonymous	*ENST00000344290, ENST00000351559, ENST00000446361, ENST00000535772, ENST00000340799, ENST00000334239, ENST00000574436, ENST00000262410, ENST00000576518, ENST00000431008, ENST00000347967, ENST00000571987, ENST00000420682, ENST00000415613	c.1770T>C, c.765T>C, c.591T>C, c.765T>C, c.678T>C, c.591T>C, c.765T>C, c.1716T>C, c.558T>C, c.765T>C, c.483T>C, c.1716T>C, c.678T>C, c.1770T>C	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , , , , , , , , ,	, , , , , , , , , , , , ,	16,5,2	75,60,5	66,52,2	2887,1667,244	5509,1948,257
MAPT	synonymous	*ENST00000344290, ENST00000446361, ENST00000574436, ENST00000351559, ENST00000571987, ENST00000334239, ENST00000420682, ENST00000347967, ENST00000340799, ENST00000576518, ENST00000535772, ENST00000431008, ENST00000415613, ENST00000262410	c.1815G>A, c.636G>A, c.810G>A, c.810G>A, c.1761G>A, c.636G>A, c.723G>A, c.528G>A, c.723G>A, c.603G>A, c.810G>A, c.810G>A, c.1815G>A, c.1761G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , , , , , , , , ,	, , , , , , , , , , , , ,	8,3,0	69,16,0	55,8,0	4319,414,8	7017,631,9
MAPT	synonymous	*ENST00000344290, ENST00000446361, ENST00000571987, ENST00000420682,	c.2178G>A, c.999G>A, c.2124G>A, c.1086G>A,	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=),	, , , , , , , , , , , , ,	, , , , , , , , , , , , ,	45,0,0	325,1,0	287,1,0	4585,3,0	6788,3,0

		ENST00000334239, ENST00000535772, ENST00000576518, ENST00000415613, ENST00000340799, ENST00000351559, ENST00000431008, ENST00000347967, ENST00000262410, ENST00000574436	c.906G>A, c.1080G>A, c.873G>A, c.2178G>A, c.1086G>A, c.1173G>A, c.1080G>A, c.798G>A, c.2124G>A, c.1173G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)							
MAPT	missense	*ENST00000344290, ENST00000431008, ENST00000262410, ENST00000571987, ENST00000535772, ENST00000574436, ENST00000351559, ENST00000415613	c.284C>T, c.284C>T, c.284C>T, c.284C>T, c.284C>T, c.284C>T, c.284C>T, c.284C>T	p.Thr95Met, p.Thr95Met, p.Thr95Met, p.Thr95Met, p.Thr95Met, p.Thr95Met, p.Thr95Met, p.Thr95Met	0, 0, 0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0, 0, 0	10,0,0	49,1,0	50,0,0	4348,1,0	6548,1,0
MAPT	***missense***	****ENST00000344290, ENST00000415613, ENST00000571987, ENST00000262410***	***c.605C>T, c.605C>T, c.605C>T, c.605C>T***	***p.Pro202Leu, p.Pro202Leu, p.Pro202Leu, p.Pro202Leu***	***0, 0, 0, 0***	***1, 1, 1, 1***	***0,2,0***	***0,8,1***	***7,12,1***	***2822,1566,254***	***5405,1839,268***
MAPT	missense	*ENST00000344290, ENST00000262410, ENST00000415613, ENST00000571987	c.671T>G, c.671T>G, c.671T>G, c.671T>G	p.Val224Gly, p.Val224Gly, p.Val224Gly, p.Val224Gly	1, 1, 1, 1	1, 0, 1, 0	8,0,0	52,0,0	47,1,0	4684,39,0	7593,43,0
MAPT	missense	*ENST00000344290, ENST00000415613, ENST00000262410, ENST00000571987	c.689A>G, c.689A>G, c.689A>G, c.689A>G	p.Gln230Arg, p.Gln230Arg, p.Gln230Arg, p.Gln230Arg	1, 1, 1, 1	0, 0, 2, 2	9,3,0	71,9,0	63,6,1	4218,495,9	7028,586,11
MAPT	missense	*ENST00000344290, ENST00000262410, ENST00000415613, ENST00000571987	c.698C>T, c.698C>T, c.698C>T, c.698C>T	p.Pro233Leu, p.Pro233Leu, p.Pro233Leu, p.Pro233Leu	0, 0, 0, 0	2, 0, 2, 0	14,0,0	78,1,0	80,0,0	80,0,0	80,0,0
MAPT	stop gained	*, ENST00000576518	c.7A>T	p.Lys3X			26,10,1	154,88,12	126,75,9	352,205,31	991,272,38

MAPT	missense	*ENST00000344290, ENST00000571987, ENST00000262410, ENST00000415613	c.853G>A, c.853G>A, c.853G>A, c.853G>A	p.Asp285Asn, p.Asp285Asn, p.Asp285Asn, p.Asp285Asn	0, 0, 0, 0	0, 0, 0, 0	20,9,2	120,70,13	91,68,12	2910,1684,255	5532,1963,270
MAPT	synonymous	*ENST00000344290, ENST00000262410, ENST00000571987, ENST00000415613	c.855C>T, c.855C>T, c.855C>T, c.855C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	27,7,0	147,48,2	125,45,5	3756,1021,76	6277,1393,99
MAPT	missense	*ENST00000344290, ENST00000415613, ENST00000262410, ENST00000571987	c.866T>C, c.866T>C, c.866T>C, c.866T>C	p.Val289Ala, p.Val289Ala, p.Val289Ala, p.Val289Ala	0, 0, 0, 0	0, 0, 0, 0	24,9,2	124,74,14	119,80,11	2938,1696,254	5560,1976,268
MOB3B	synonymous	*ENST00000262244	c.414C>T	p.(=)			28,19,3	243,128,12	195,84,13	3304,1488,178	5880,1815,191
MOB3B	synonymous	*ENST00000262244	c.615G>A	p.(=)			50,0,0	390,0,0	303,1,0	303,1,0	303,1,0
MOB3B	missense	*ENST00000262244	c.98G>A	p.Arg33Gln	0	0	50,0,0	354,0,0	277,1,0	277,1,0	277,1,0
NEFH	missense	*ENST00000310624	c.1054C>A	p.Arg352Ser	1		31,0,0	184,2,0	142,10,0	4778,52,0	7687,59,0
NEFH	missense	*ENST00000310624	c.1105G>A	p.Ala369Thr	0		43,0,0	254,0,0	175,3,0	175,3,0	175,3,0
NEFH	synonymous	*ENST00000310624	c.1200C>T	p.(=)			28,16,0	172,91,7	125,62,18	3464,1310,109	6039,1631,129
NEFH	missense	*ENST00000310624	c.1387G>A	p.Glu463Lys	0		45,5,0	324,52,3	244,45,1	4066,852,50	6892,940,52
NEFH	missense	*ENST00000310624	c.1580C>T	p.Pro527Leu	0		47,0,0	354,1,0	279,2,0	279,2,0	279,2,0
NEFH	synonymous	*ENST00000310624	c.1740C>T	p.(=)			50,0,0	370,6,0	291,8,0	4857,119,1	7763,129,1
NEFH	missense	*ENST00000310624	c.1844C>T	p.Pro615Leu	1	2	30,17,2	217,147,12	170,109,13	3028,1715,227	5081,2490,315
NEFH	***missense***	****ENST00000310624***	***c.1933G>A***	***p.Glu645Lys***	***0***	***2***	***41,2,0***	***314,30,0***	***253,10,0***	***253,10,0***	***253,10,0***
NEFH	***feature elongation, frameshift***	****ENST00000310624***	***c.1935_1936insG***	***p.Ala646GlyfsX5***			***34,9,0***	***237,63,0***	***198,44,0***	***198,44,0***	***198,44,0***
NEFH	***synonymous***	****ENST00000310624***	***c.1935A>G***	***p.(=)***			***39,5,0***	***292,41,0***	***246,21,0***	***246,21,0***	***246,21,0***
NEFH	***synonymous***	****ENST00000310624***	***c.1938A>C***	***p.(=)***			***27,18,0***	***163,147,0***	***137,115,0***	***137,115,0***	***137,115,0***
NEFH	***synonymous***	****ENST00000310624***	***c.1965A>T***	***p.(=)***			***16,29,0***	***118,206,0***	***129,125,0***	***129,125,0***	***129,125,0***
NEFH	***inframe deletion***	****ENST00000310624***	***c.1970_1975delAGGAAG***	***p.Glu658_Glu659del***			***24,23,0***	***189,148,0***	***187,79,0***	***187,79,0***	***187,79,0***
NEFH	***missense***	****ENST00000310624***	***c.1973A>C***	***p.Glu658Ala***	***0***	***0***	***40,1,0***	***298,26,0***	***227,32,0***	***227,32,0***	***227,32,0***
NEFH	***missense***	****ENST00000310624***	***c.1974A>C***	***p.Glu658Asp***	***0***	***2***	***45,1,0***	***323,15,0***	***248,14,0***	***248,14,0***	***248,14,0***
NEFH	***missense***	****ENST00000310624***	***c.1975G>A***	***p.Glu659Lys***	***0***	***2***	***46,1,0***	***325,12,0***	***258,11,0***	***258,11,0***	***258,11,0***

NEFH	***synonymous***	****ENST00000310624***	***c.1989T>A***	***p.(=)***			***43,2,0***	***332,22,0***	***300,0,0***	***300,0,0***	***300,0,0***
NEFH	***synonymous***	****ENST00000310624***	***c.2082C>T***	***p.(=)***			***47,0,0***	***330,0,1***	***304,0,0***	***304,0,0***	***304,0,0***
NEFH	synonymous	*ENST00000310624	c.2232T>C	p.(=)			2,18,29	14,145,216	12,107,179	230,1711,3035	321,2459,5112
NEFH	***inframe deletion***	****ENST00000310624***	***c.2368_2370delAAG***	***p.Lys790del***			***48,0,0***	***324,1,0***	***278,0,0***	***278,0,0***	***278,0,0***
NEFH	missense	*ENST00000310624	c.2414A>C	p.Glu805Ala	1	1	31,14,0	200,92,4	177,62,10	3518,1307,101	5785,1900,157
NEFH	synonymous	*ENST00000310624	c.2646C>T	p.(=)			37,0,0	281,0,0	199,2,1	199,2,1	199,2,1
NEFH	missense	*ENST00000310624	c.2740G>A	p.Val914Met	0		41,0,0	301,1,0	258,1,0	258,1,0	258,1,0
NEFH	synonymous	*ENST00000310624	c.2757C>T	p.(=)			39,1,0	266,6,0	201,3,0	4827,55,0	7724,74,0
NEFH	synonymous	*ENST00000310624	c.2784A>G	p.(=)			2,16,23	9,129,182	9,98,156	221,1706,3014	308,2459,5090
NEFH	missense	*ENST00000310624	c.2977A>G	p.Lys993Glu	1		38,0,0	315,0,0	265,1,0	265,1,0	265,1,0
NIPA1	***missense***	****ENST00000337435, ENST00000437912, ENST00000561183***	***c.233T>G, c.8T>G, c.8T>G***	***p.Val78Gly, p.Val3Gly, p.Val3Gly***	***0, 1, 1***	***0, 0, 0***	***32,1,0***	***174,51,0***	***200,46,0***	***200,46,0***	***200,46,0***
NIPA1	synonymous	*ENST00000337435, ENST00000437912, ENST00000561183	c.441A>G, c.216A>G, c.216A>G	p.(=), p.(=), p.(=)	, ,	, ,	1,17,31	32,141,214	17,120,159	357,1937,2680	1027,3268,3595
OPTN	synonymous	*ENST00000263036, ENST00000378764, ENST00000378757, ENST00000378747, ENST00000378752, ENST00000378748	c.102G>A, c.102G>A, c.102G>A, c.102G>A, c.102G>A, c.102G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	24,16,3	120,112,12	104,102,14	2564,1980,354	5049,2389,376
OPTN	missense	*ENST00000263036, ENST00000378764, ENST00000378747, ENST00000378752, ENST00000378748, ENST00000378757	c.1192C>G, c.1174C>G, c.1192C>G, c.1174C>G, c.1192C>G, c.1192C>G	p.Gln398Glu, p.Gln392Glu, p.Gln398Glu, p.Gln392Glu, p.Gln398Glu, p.Gln398Glu	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0	50,0,0	367,1,0	243,0,0	243,0,0	243,0,0
OPTN	synonymous	*ENST00000263036, ENST00000378764, ENST00000378757, ENST00000378747, ENST00000378748, ENST00000378752	c.123G>A, c.123G>A, c.123G>A, c.123G>A, c.123G>A, c.123G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	39,2,0	225,8,0	224,9,0	4806,104,1	7710,116,1

OPTN	synonymous	*ENST00000263036, ENST00000378748, ENST00000378764, ENST00000378747, ENST00000378752, ENST00000378757	c.1569G>A, c.1569G>A, c.1551G>A, c.1569G>A, c.1551G>A, c.1569G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	48,0,0	341,1,0	279,0,0	279,0,0	279,0,0
OPTN	***missense***	****ENST00000263036, ENST00000378747, ENST00000378764, ENST00000378748, ENST00000430081, ENST00000378757, ENST00000378752***	***c.187C>A, c.187C>A, c.187C>A, c.187C>A, c.16C>A, c.187C>A, c.187C>A***	***p.Gln63Lys, p.Gln63Lys, p.Gln63Lys, p.Gln63Lys, p.Gln6Lys, p.Gln63Lys, p.Gln63Lys***	***0, 0, 0, 0, 1, 0, 0***	***2, 2, 2, 2, 2, 2, 2***	***49,0,0***	***361,1,0***	***271,1,0***	***271,1,0***	***271,1,0***
OPTN	synonymous	*ENST00000263036, ENST00000378757, ENST00000430081, ENST00000378752, ENST00000378747, ENST00000378748, ENST00000378764	c.213G>A, c.213G>A, c.42G>A, c.213G>A, c.213G>A, c.213G>A, c.213G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , ,	, , , , , ,	49,1,0	384,0,0	284,0,0	284,0,0	284,0,0
OPTN	missense	*ENST00000263036, ENST00000378764, ENST00000378747, ENST00000430081, ENST00000378757, ENST00000378752, ENST00000378748	c.287G>A, c.287G>A, c.287G>A, c.116G>A, c.287G>A, c.287G>A, c.287G>A	p.Arg96His, p.Arg96His, p.Arg96His, p.Arg39His, p.Arg96His, p.Arg96His, p.Arg96His	0, 0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0, 0	47,0,0	368,0,0	264,1,0	642,1,0	1354,2,0
OPTN	missense	*ENST00000263036, ENST00000378764, ENST00000378752, ENST00000430081, ENST00000378757, ENST00000378748, ENST00000378747	c.293T>A, c.293T>A, c.293T>A, c.122T>A, c.293T>A, c.293T>A, c.293T>A	p.Met98Lys, p.Met98Lys, p.Met98Lys, p.Met41Lys, p.Met98Lys, p.Met98Lys, p.Met98Lys	0, 0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0, 0	48,2,0	357,21,1	267,18,1	4654,303,7	6956,885,39
OPTN	synonymous	*ENST00000263036,	c.489A>G,	p.(=), p.(=),	, , , ,	, , , ,	50,0,0	388,1,0	306,0,0	4968,16,0	7881,19,0

		ENST00000378764, ENST00000378748, ENST00000430081, ENST00000378757, ENST00000378747, ENST00000378752	c.489A>G, c.489A>G, c.318A>G, c.489A>G, c.489A>G, c.489A>G	p.(=), p.(=), p.(=), p.(=), p.(=)	, ,	, ,					
OPTN	synonymous	*ENST00000263036, ENST00000378748, ENST00000430081, ENST00000378757, ENST00000378747, ENST00000378764, ENST00000378752	c.513C>T, c.513C>T, c.342C>T, c.513C>T, c.513C>T, c.513C>T, c.513C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , ,	, , , , , ,	50,0,0	384,0,0	299,1,0	4598,2,0	6801,2,0
OPTN	missense	*ENST00000263036, ENST00000378748, ENST00000378757, ENST00000378747, ENST00000378752, ENST00000378764	c.964A>G, c.964A>G, c.964A>G, c.964A>G, c.946A>G, c.946A>G	p.Lys322Glu, p.Lys322Glu, p.Lys322Glu, p.Lys322Glu, p.Lys316Glu, p.Lys316Glu	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0	0,0,44	0,0,294	0,1,195	0,12,4862	2,138,7650
PARK7	synonymous	*ENST00000338639, ENST00000469225, ENST00000377491, ENST00000377488, ENST00000493678, ENST00000493373	c.234C>T, c.117C>T, c.234C>T, c.234C>T, c.234C>T, c.234C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	48,0,0	345,1,0	240,0,0	4906,12,0	7317,490,27
PARK7	missense	*ENST00000338639, ENST00000493678, ENST00000377493, ENST00000469225, ENST00000377488, ENST00000493373, ENST00000377491	c.293G>A, c.293G>A, c.233G>A, c.176G>A, c.293G>A, c.293G>A, c.293G>A	p.Arg98Gln, p.Arg98Gln, p.Arg78Gln, p.Arg59Gln, p.Arg98Gln, p.Arg98Gln, p.Arg98Gln	0, 0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0, 0	46,0,0	294,3,0	239,6,0	4809,113,1	7715,123,1
PARK7	***missense***	****ENST00000338639, ENST00000493373, ENST00000377493,***	***c.49A>C, c.49A>C, c.49A>C,***	***p.Met17Leu, p.Met17Leu, p.Met17Leu,***	***1, 1, 1, 1, 1, 1***	***0, 0, 0, 0, 0, 0***	***49,0,0***	***375,1,0***	***283,0,0***	***661,0,0***	***1373,1,0***

		ENST00000493678, ENST00000377488, ENST00000377491	***c.49A>C, c.49A>C, c.49A>C***	***p.Met17Leu, p.Met17Leu, p.Met17Leu***							
PARK7	synonymous	*ENST00000338639, ENST00000493373, ENST00000469225, ENST00000377493, ENST00000377488, ENST00000377491, ENST00000493678	c.501A>G, c.501A>G, c.414A>G, c.441A>G, c.501A>G, c.501A>G, c.501A>G	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , , ,	, , , , , ,	50,0,0	383,1,0	305,2,0	4967,18,0	7881,20,0
PARK7	missense	*ENST00000338639, ENST00000377493, ENST00000377491, ENST00000469225, ENST00000377488, ENST00000493373, ENST00000493678	c.535G>A, c.475G>A, c.535G>A, c.448G>A, c.535G>A, c.535G>A, c.535G>A	p.Ala179Thr, p.Ala159Thr, p.Ala179Thr, p.Ala150Thr, p.Ala179Thr, p.Ala179Thr, p.Ala179Thr	0, 0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0, 0	50,0,0	378,0,0	303,1,0	4976,6,0	7890,8,0
PON1	missense	*ENST00000222381, ENST00000542556	c.163T>A, c.163T>A	p.Leu55Met, p.Leu55Met	1, 1	0, 2	18,26,6	159,176,54	109,128,43	1982,2264,712	4041,3035,798
PON1	***feature elongation, frameshift***	****ENST00000222381, ENST00000542556***	***c.391dupG, c.391dupG***	***p.Val131Glyfs X15, p.Val131Glyfs X15***	***,***	***,***	***50,0,0***	***391,0,0***	***282,1,0***	***282,1,0***	***282,1,0***
PON1	synonymous	*ENST00000222381, ENST00000542556	c.468A>G, c.468A>G	p.(=), p.(=)	,	,	50,0,0	388,1,0	280,0,0	280,0,0	280,0,0
PON1	missense	*ENST00000222381, ENST00000542556	c.575A>G, c.575A>G	p.Gln192Arg, p.Gln192Arg	0, 0	0, 0	24,23,3	211,152,28	150,115,21	2544,2013,407	2905,3306,1669
PON1	missense	*ENST00000222381, ENST00000542556	c.602C>T, c.602C>T	p.Ala201Val, p.Ala201Val	0, 0	0, 0	50,0,0	386,2,0	278,3,0	4931,28,0	7844,31,0
PON1	synonymous	*ENST00000222381, ENST00000542556	c.603G>A, c.603G>A	p.(=), p.(=)	,	,	50,0,0	386,4,0	283,1,0	4946,16,0	7860,18,0
PON1	missense	*ENST00000222381, ENST00000542556	c.953C>T, c.953C>T	p.Thr318Ile, p.Thr318Ile	1, 1	0, 0	50,0,0	390,0,0	289,1,0	4588,2,0	6791,2,0
PON1, PON3	synonymous	*ENST00000265627, ENST00000427422,	c.63C>T, c.63C>T,	p.(=), p.(=), p.(=)	, ,	, ,	1,3,0	10,34,1	12,28,1	2607,1810,302	3945,3085,605

		ENST00000542556	c.63C>T								
PON1, PON3	stop gained	*ENST00000265627, ENST00000427422, ENST00000542556	c.94C>T, c.94C>T, c.94C>T	p.Arg32X, p.Arg32X, p.Arg32X	, ,	, ,	48,0,0	371,3,0	280,0,0	4937,21,0	7849,25,0
PON2	***feature truncation, frameshift***	****ENST00000222572, ENST00000433091, ENST00000536183***	***c.286delA, c.286delA, c.349delA***	***p.Arg96Glyfs X5, p.Arg96Glyfs X5, p.Arg117Glyf sX5***	***, ,***	***, ,***	***49,1,0***	***386,2,0***	***290,1,0***	***290,1,0***	***290,1,0***
PON2	missense	*ENST00000222572, ENST00000433091, ENST00000536183	c.359T>G, c.359T>G, c.422T>G	p.Ile120Arg, p.Ile120Arg, p.Ile141Arg	1, 1, 0	0, 0, 1	50,0,0	389,0,0	287,1,0	287,1,0	287,1,0
PON2	missense	*ENST00000222572, ENST00000536183, ENST00000433091	c.443C>G, c.506C>G, c.407C>G	p.Ala148Gly, p.Ala169Gly, p.Ala136Gly	1, 1, 1	0, 0, 0	31,17,0	205,158,22	155,90,20	2924,1756,263	4445,2939,475
PON2	missense	*ENST00000222572, ENST00000433091, ENST00000536183	c.661T>G, c.625T>G, c.724T>G	p.Ser221Ala, p.Ser209Ala, p.Ser242Ala	0, 0, 0	0, 0, 0	49,0,0	380,1,0	257,0,0	257,0,0	257,0,0
PON2	missense	*ENST00000222572, ENST00000433091, ENST00000536183	c.827C>T, c.791C>T, c.890C>T	p.Ser276Leu, p.Ser264Leu, p.Ser297Leu	1, 1, 1	0, 0, 0	50,0,0	381,1,0	273,1,0	4572,2,0	6775,2,0
PON2	missense	*ENST00000222572, ENST00000536183, ENST00000433091	c.913C>T, c.976C>T, c.877C>T	p.Arg305Cys, p.Arg326Cys, p.Arg293Cys	1, 1, 1	2, 2, 2	48,1,0	384,2,0	276,0,0	4573,3,0	6776,3,0
PON2	missense	*ENST00000222572, ENST00000433091, ENST00000536183	c.932C>G, c.896C>G, c.995C>G	p.Ser311Cys, p.Ser299Cys, p.Ser332Cys	1, 1, 1	0, 0, 0	32,17,1	210,157,23	166,95,22	2934,1762,265	4447,2950,480
PON3	missense	*ENST00000265627, ENST00000427422	c.262A>G, c.262A>G	p.Met88Val, p.Met88Val	1, 0	0, 0	50,0,0	383,1,0	280,1,0	4943,16,0	7859,16,0
PON3	synonymous	*ENST00000265627, ENST00000427422	c.297G>A, c.297G>A	p.(=), p.(=)	,	,	17,20,12	93,199,91	85,121,72	1446,2454,1056	1930,3805,2137
PON3	missense	*ENST00000265627, ENST00000427422	c.408G>T, c.408G>T	p.Met136Ile, p.Met136Ile	0, 0	0, 0	49,0,0	380,1,0	259,2,0	4554,7,0	6756,8,0
PON3	synonymous	*ENST00000265627,	c.609T>C,	p.(=), p.(=)	,	,	49,1,0	379,12,0	281,7,0	4908,57,1	7814,67,1

		ENST00000427422	c.609T>C								
PON3	***synonymous***	****ENST00000265627***	***c.819T>C***	***p.(=)***			***47,1,0***	***330,28,0***	***247,23,0***	***247,23,0***	***247,23,0***
PON3	synonymous	*ENST00000265627	c.954C>T	p.(=)			49,0,0	376,1,0	285,0,0	4584,1,0	6787,1,0
PON3	missense	*ENST00000265627	c.971G>A	p.Gly324Asp	1	2	49,0,0	382,0,0	292,1,0	4935,35,0	7850,36,0
PRPH	***synonymous***	****ENST00000257860, ENST00000532332***	***c.1083C>G, c.268C>G***	***p.(=), p.(=)***	***,***	***,***	***0,0,0***	***1,2,0***	***1,0,0***	***4535,129,0***	***7383,188,0***
PRPH	***synonymous***	****ENST00000257860, ENST00000532332***	***c.1104A>G , c.289A>G***	***p.(=), p.(=)***	***,***	***,***	***0,0,0***	***9,0,0***	***7,0,0***	***4305,1,0***	***6507,1,0***
PRPH	***synonymous***	****ENST00000257860, ENST00000532332***	***c.1107A>G , c.292A>G***	***p.(=), p.(=)***	***,***	***,***	***0,5,0***	***6,21,0***	***4,12,2***	***3018,1458,219***	***4431,2637,543***
PRPH	***missense***	****ENST00000257860, ENST00000532332***	***c.1225G>A , c.410G>A***	***p.Val409Met, p.Val138Met***	***0, 0***	***0, 0***	***41,0,0***	***271,2,0***	***176,7,0***	***176,7,0***	***176,7,0***
PRPH	***missense***	****ENST00000257860, ENST00000532332***	***c.1231G>A , c.416G>A***	***p.Val411Ile, p.Val140Ile***	***0, 0***	***0, 0***	***43,0,0***	***271,0,0***	***191,5,0***	***191,5,0***	***191,5,0***
PRPH	missense	*ENST00000257860, ENST00000532332	c.1303C>T, c.488C>T	p.Arg435Trp, p.Arg164Trp	1, 1	2, 1	41,0,0	244,1,0	215,1,0	4889,5,0	7802,8,0
PRPH	missense	*ENST00000257860	c.26G>A	p.Arg9Gln	1	1	18,1,0	43,2,0	31,2,0	4585,111,1	7459,145,2
PRPH	***missense***	****ENST00000257860, ENST00000451891***	***c.322T>C, c.79T>C***	***p.Phe108Leu, p.Phe27Leu***	***1, 1***	***1, 2***	***2,0,0***	***4,1,0***	***5,0,0***	***5,0,0***	***5,0,0***
PRPH	synonymous	*ENST00000257860	c.63C>T	p.(=)			24,0,0	74,7,0	45,5,0	4516,169,0	7272,309,2
PRPH	***missense***	****ENST00000257860, ENST00000532332***	***c.829G>A, c.14G>A***	***p.Ala277Thr, p.Ala6Thr***	***0, 0***	***0, 0***	***2,0,0***	***2,0,0***	***2,0,0***	***4511,169,0***	***7276,319,1***
SETX	synonymous	*ENST00000224140, ENST00000372169, ENST00000393220	c.1077T>C, c.1077T>C, c.1077T>C	p.(=), p.(=), p.(=)	, ,	, ,	0,15,35	6,104,276	6,64,187	110,1280,3544	588,2589,4673
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.1655A>C, c.1655A>C, c.1655A>C	p.Gln552Pro, p.Gln552Pro, p.Gln552Pro	0, 0, 0	1, 1, 1	50,0,0	390,0,0	290,1,0	290,1,0	290,1,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.1750C>G, c.1750C>G, c.1750C>G	p.Leu584Val, p.Leu584Val, p.Leu584Val	1, 1, 1	2, 2, 2	50,0,0	390,1,0	278,2,0	278,2,0	278,2,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.1869A>C, c.1869A>C, c.1869A>C	p.Glu623Asp, p.Glu623Asp, p.Glu623Asp	1, 1, 1	2, 2, 2	50,0,0	385,0,0	267,1,0	4564,4,0	6767,4,0
SETX	synonymous	*ENST00000224140,	c.192A>G,	p.(=), p.(=),	, ,	, ,	48,1,0	377,5,0	256,2,1	4910,24,1	7824,26,1

		ENST00000393220, ENST00000372169	c.192A>G, c.192A>G	p.(=)							
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169	c.1979C>G, c.1979C>G, c.1979C>G	p.Ala660Gly, p.Ala660Gly, p.Ala660Gly	0, 0, 0	0, 0, 0	43,7,0	339,44,1	234,33,0	4384,539,21	6103,1543,214
SETX	missense	*, ENST00000436441, ENST00000372169	c.2095C>T, c.7369C>T	p.Leu699Phe, p.Leu2457Phe	0, 0	0, 0	6,0,0	58,3,0	72,3,0	2409,34,1	3995,37,1
SETX	synonymous	*ENST00000224140, ENST00000393220, ENST00000372169	c.234G>A, c.234G>A, c.234G>A	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	384,0,0	262,2,0	4559,5,0	6761,6,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.2411T>C, c.2411T>C, c.2411T>C	p.Leu804Ser, p.Leu804Ser, p.Leu804Ser	1, 1, 1	0, 0, 0	49,0,0	378,1,0	253,0,0	4928,2,0	7843,2,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.2479A>G, c.2479A>G, c.2479A>G	p.Lys827Glu, p.Lys827Glu, p.Lys827Glu	0, 0, 0	0, 0, 0	49,0,0	378,1,0	246,0,0	4914,8,0	7828,9,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.2717C>T, c.2717C>T, c.2717C>T	p.Ser906Leu, p.Ser906Leu, p.Ser906Leu	0, 0, 0	0, 0, 0	49,0,0	380,1,0	258,0,0	4557,1,0	6760,1,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.2755G>C, c.2755G>C, c.2755G>C	p.Val919Leu, p.Val919Leu, p.Val919Leu	0, 0, 0	0, 0, 0	50,0,0	383,1,0	266,0,0	266,0,0	266,0,0
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169	c.2842C>A, c.2842C>A, c.2842C>A	p.Pro948Thr, p.Pro948Thr, p.Pro948Thr	0, 0, 0	0, 0, 0	49,1,0	387,0,0	282,0,0	282,0,0	282,0,0
SETX	synonymous	*ENST00000224140, ENST00000372169, ENST00000393220	c.2964A>G, c.2964A>G, c.2964A>G	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	389,0,0	278,1,0	4577,2,0	6780,2,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.2975A>G, c.2975A>G, c.2975A>G	p.Lys992Arg, p.Lys992Arg, p.Lys992Arg	1, 1, 1	0, 0, 0	48,1,1	375,13,0	277,3,0	4799,159,0	7698,176,0
SETX	synonymous	*ENST00000224140, ENST00000393220, ENST00000372169	c.3147C>T, c.3147C>T, c.3147C>T	p.(=), p.(=), p.(=)	, ,	, ,	42,8,0	336,53,1	228,36,2	4199,718,27	6767,1048,45

SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.3221A>G, c.3221A>G, c.3221A>G	p.Glu1074Gly, p.Glu1074Gly, p.Glu1074Gly	1, 1, 1	2, 2, 2	50,0,0	384,0,0	264,1,0	264,1,0	264,1,0
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169	c.3229G>A, c.3229G>A, c.3229G>A	p.Asp1077Asn, p.Asp1077Asn, p.Asp1077Asn	1, 1, 1	1, 1, 1	50,0,0	383,2,0	267,0,0	4936,9,0	7850,10,0
SETX	synonymous	*ENST00000224140, ENST00000393220, ENST00000372169	c.3336T>C, c.3336T>C, c.3336T>C	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	383,4,0	267,2,0	4940,7,0	7853,10,0
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169	c.3455T>G, c.3455T>G, c.3455T>G	p.Phe1152Cys, p.Phe1152Cys, p.Phe1152Cys	1, 1, 1	1, 1, 1	47,3,0	362,25,0	263,13,0	4629,320,5	7166,679,25
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169	c.3576T>G, c.3576T>G, c.3576T>G	p.Asp1192Glu, p.Asp1192Glu, p.Asp1192Glu	0, 0, 0	0, 0, 0	0,15,35	5,93,286	3,66,203	93,1168,3689	533,2448,4885
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169	c.3754G>A, c.3754G>A, c.3754G>A	p.Gly1252Arg, p.Gly1252Arg, p.Gly1252Arg	0, 0, 0	0, 0, 0	0,15,35	6,107,276	8,70,212	118,1289,3561	1198,2588,4098
SETX	***feature elongation, frameshift***	****ENST00000224140, ENST00000372169, ENST00000393220***	***c.3930dupA, c.3930dupA, c.3930dup***	***p.Arg1311ThrfsX2, p.Arg1311ThrfsX2, p.Arg1311Thr***	***, ,***	***, ,***	***49,1,0***	***391,0,0***	***293,0,0***	***293,0,0***	***293,0,0***

			A	***fsX2***							
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.4156A>G, c.4156A>G, c.4156A>G	p.Ile1386Val, p.Ile1386Val, p.Ile1386Val	0, 0, 0	0, 0, 0	0,15,35	6,107,277	8,68,191	112,1292,3541	1190,2591,4080
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.4660T>G, c.4660T>G, c.4660T>G	p.Cys1554Gly , p.Cys1554Gly , p.Cys1554Gly	1, 1, 1	2, 2, 2	49,1,0	385,6,0	284,1,0	4922,41,0	7831,48,0
SETX	synonymous	*ENST00000224140, ENST00000372169, ENST00000393220	c.4707T>C, c.4707T>C, c.4707T>C	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	391,0,0	288,1,0	288,1,0	288,1,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.472T>G, c.472T>G, c.472T>G	p.Leu158Val, p.Leu158Val, p.Leu158Val	1, 1, 1	2, 2, 2	50,0,0	383,3,0	280,5,0	4910,53,0	7821,58,0
SETX	synonymous	*ENST00000224140, ENST00000393220, ENST00000372169	c.4755T>G, c.4755T>G, c.4755T>G	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	384,8,0	287,3,0	4888,80,0	7796,88,0
SETX	***missense***	****ENST00000224140, ENST00000393220, ENST00000372169***	***c.506G>A, c.506G>A, c.506G>A***	***p.Arg169His, p.Arg169His, p.Arg169His***	***1, 1, 1***	***2, 2, 2***	***49,0,0***	***372,3,0***	***249,2,0***	***249,2,0***	***249,2,0***
SETX	synonymous	*ENST00000224140, ENST00000393220, ENST00000372169	c.5271A>G, c.5271A>G, c.5271A>G	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	387,1,0	268,1,0	268,1,0	268,1,0
SETX	synonymous	*ENST00000224140, ENST00000436441, ENST00000372169, ENST00000393220	c.5283A>G, c.9A>G, c.5283A>G, c.5283A>G	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	49,0,0	375,1,0	263,0,0	4558,4,0	6761,4,0
SETX	synonymous	*ENST00000224140, ENST00000372169, ENST00000393220	c.540A>G, c.540A>G, c.540A>G	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	385,1,0	279,0,0	279,0,0	279,0,0
SETX	missense	*ENST00000224140, ENST00000436441, ENST00000372169, ENST00000393220	c.5563A>G, c.289A>G, c.5563A>G, c.5563A>G	p.Thr1855Ala, p.Thr97Ala, p.Thr1855Ala, p.Thr1855Ala	0, 0, 0, 0	0, 0, 0, 0	34,15,1	275,107,9	197,72,8	3541,1301,113	4078,2603,1190

SETX	missense	*ENST00000224140, ENST00000372169, ENST00000436441, ENST00000393220	c.5587A>G, c.5587A>G, c.313A>G, c.5587A>G	p.Thr1863Ala, p.Thr1863Ala, p.Thr105Ala, p.Thr1863Ala	1, 1, 1, 1	2, 2, 2, 2	49,1,0	391,0,0	278,0,0	278,0,0	278,0,0
SETX	synonymous	*ENST00000224140, ENST00000372169, ENST00000393220, ENST00000436441	c.5811T>C, c.5811T>C, c.5811T>C, c.537T>C	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	33,15,1	274,106,8	191,69,8	3532,1301,113	4070,2603,1189
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000436441, ENST00000393220	c.5842A>G, c.5842A>G, c.568A>G, c.5842A>G	p.Met1948Val, p.Met1948Val, p.Met190Val, p.Met1948Val	1, 1, 1, 1	2, 2, 2, 2	50,0,0	387,1,0	279,0,0	279,0,0	279,0,0
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169, ENST00000436441	c.5998C>G, c.5998C>G, c.5998C>G, c.724C>G	p.Gln2000Glu, p.Gln2000Glu, p.Gln2000Glu, p.Gln242Glu	1, 1, 1, 0	1, 0, 0, 0	49,1,0	386,2,0	285,0,0	4959,4,0	7874,5,0
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169	c.59G>A, c.59G>A, c.59G>A	p.Arg20His, p.Arg20His, p.Arg20His	0, 0, 0	0, 0, 0	49,0,0	380,5,0	288,9,0	4866,108,1	7771,119,1
SETX	***missense***	****ENST00000224140, ENST00000393220, ENST00000372169, ENST00000436441***	***c.6005G>A, c.6005G>A, c.6005G>A, c.731G>A***	***p.Arg2002His, p.Arg2002His, p.Arg2002His, p.Arg244His***	***1, 1, 1, 1***	***2, 2, 2, 2***	***50,0,0***	***375,1,0***	***268,2,0***	***268,2,0***	***268,2,0***
SETX	synonymous	*ENST00000224140, ENST00000393220, ENST00000372169, ENST00000436441	c.6024T>C, c.6024T>C, c.6024T>C, c.750T>C	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	389,0,0	285,1,0	285,1,0	285,1,0
SETX	***missense***	****ENST00000224140,***	***c.6049A>G***	***p.Met2017Va***	***0, 0,***	***2, 2,***	***50,0,0***	***390,1,0***	***285,0,0***	***285,0,0***	***285,0,0***

		ENST00000393220, ENST00000436441, ENST00000372169	***, c.6049A>G, c.775A>G, c.6049A>G***	***l, p.Met2017Val, p.Met259Val, p.Met2017Val***	***0, 0***	***1, 1***					
SETX	synonymous	*ENST00000224140, ENST00000436441, ENST00000372169, ENST00000393220	c.6507G>A, c.1233G>A, c.6507G>A, c.6507G>A	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	383,7,0	279,16,0	4760,208,5	7653,231,5
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169	c.654G>C, c.654G>C, c.654G>C	p.Lys218Asn, p.Lys218Asn, p.Lys218Asn	1, 1, 1	2, 2, 2	50,0,0	391,0,0	279,1,0	4951,7,0	7867,7,0
SETX	***missense***	****ENST00000224140, ENST00000393220, ENST00000436441, ENST00000372169***	***c.6848C>A, c.6848C>A, c.1574C>A, c.6848C>A***	***p.Thr2283Lys, p.Thr2283Lys, p.Thr525Lys, p.Thr2283Lys***	***1, 1, 1, 1***	***2, 2, 2, 2***	***48,0,0***	***357,3,0***	***241,8,0***	***241,8,0***	***241,8,0***
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169	c.710A>G, c.710A>G, c.710A>G	p.Tyr237Cys, p.Tyr237Cys, p.Tyr237Cys	1, 1, 1	2, 2, 2	50,0,0	386,1,0	272,1,0	4570,3,0	6773,3,0
SETX	synonymous	*ENST00000224140, ENST00000372169, ENST00000436441	c.7119T>C, c.7119T>C, c.1845T>C	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	389,1,0	293,0,0	293,0,0	293,0,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000436441	c.7139G>A, c.7139G>A, c.1865G>A	p.Arg2380Gln, p.Arg2380Gln, p.Arg622Gln	0, 0, 0	2, 1, 1	49,1,0	387,0,0	283,2,0	4582,3,0	6785,3,0
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000372169, ENST00000436441	c.7406T>C, c.7307T>C, c.7493T>C, c.2219T>C	p.Leu2469Pro, p.Leu2436Pro, p.Leu2498Pro, p.Leu740Pro	0, 0, 0, 0	0, 0, 0, 0	50,0,0	390,0,0	302,1,0	4601,2,0	6804,2,0

SETX	synonymous	*ENST00000224140, ENST00000372169, ENST00000436441, ENST00000393220	c.7524A>G, c.7611A>G, c.2337A>G, c.7425A>G	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	390,0,0	303,1,0	303,1,0	303,1,0
SETX	synonymous	*ENST00000224140, ENST00000436441, ENST00000372169, ENST00000393220	c.7590T>A, c.2403T>A, c.7677T>A, c.7491T>A	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	388,1,0	303,0,0	303,0,0	303,0,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000436441, ENST00000393220	c.7640T>C, c.7727T>C, c.2453T>C, c.7541T>C	p.Ile2547Thr, p.Ile2576Thr, p.Ile818Thr, p.Ile2514Thr	0, 0, 0, 0	0, 0, 0, 0	50,0,0	378,10,0	305,0,0	4913,69,1	7824,74,1
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000436441, ENST00000372169	c.7645G>A, c.7546G>A, c.2458G>A, c.7732G>A	p.Val2549Ile, p.Val2516Ile, p.Val820Ile, p.Val2578Ile	1, 1, 0, 0	0, 0, 0, 0	50,0,0	387,2,0	305,0,0	4605,0,0	6807,1,0
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000436441, ENST00000372169	c.7682C>T, c.7583C>T, c.2495C>T, c.7769C>T	p.Ser2561Leu, p.Ser2528Leu, p.Ser832Leu, p.Ser2590Leu	0, 0, 0, 0	0, 0, 0, 0	50,0,0	356,2,0	253,0,0	4552,1,0	6755,1,0
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000436441, ENST00000372169	c.7709C>T, c.7610C>T, c.2522C>T, c.7796C>T	p.Pro2570Leu, p.Pro2537Leu, p.Pro841Leu, p.Pro2599Leu	1, 1, 0, 1	0, 0, 0, 0	50,0,0	362,0,0	297,1,0	297,1,0	297,1,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220, ENST00000436441	c.7759A>G, c.7846A>G, c.7660A>G, c.2572A>G	p.Ile2587Val, p.Ile2616Val, p.Ile2554Val, p.Ile858Val	0, 0, 0, 0	0, 0, 0, 0	22,20,4	160,154,28	145,115,15	2472,2065,416	2770,3162,1937
SETX	***missense***	****ENST00000224140, ENST00000372169, ENST00000436441,***	***c.7787C>G, c.7874C>G, c.2600C>G,***	***p.Ala2596Gly, p.Ala2625Gly***	***1, 1, 0, 1***	***0, 0, 0, 0***	***9,4,0***	***125,19,0***	***142,16,0***	***142,16,0***	***142,16,0***

		ENST00000393220	***c.7688C>G***	***, p.Ala867Gly, p.Ala2563Gly***							
SETX	missense	*ENST00000224140, ENST00000393220, ENST00000436441, ENST00000372169	c.7834A>G, c.7735A>G, c.2647A>G, c.7921A>G	p.Ser2612Gly, p.Ser2579Gly, p.Ser883Gly, p.Ser2641Gly	0, 0, 0, 0	0, 0, 0, 0	34,2,0	202,19,1	176,15,0	4611,252,6	6595,1076,114
SETX	synonymous	*ENST00000224140, ENST00000393220, ENST00000436441, ENST00000372169	c.7944C>T, c.7845C>T, c.2757C>T, c.8031C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	37,0,0	220,1,0	180,0,0	4479,1,0	6682,1,0
SETX	missense	*ENST00000224140, ENST00000372169, ENST00000393220	c.814C>G, c.814C>G, c.814C>G	p.His272Asp, p.His272Asp, p.His272Asp	1, 1, 1	2, 2, 2	50,0,0	386,1,0	280,0,0	280,0,0	280,0,0
SIGMAR 1	missense	*ENST00000277010, ENST00000378892, ENST00000477726	c.595C>T, c.328C>T, c.502C>T	p.Leu199Phe, p.Leu110Phe, p.Leu168Phe	0, 0, 1	0, 0, 0	14,0,0	169,0,0	139,1,0	4437,3,0	6640,3,0
SIGMAR 1	missense	*ENST00000277010, ENST00000378892, ENST00000477726	c.622C>T, c.355C>T, c.529C>T	p.Arg208Trp, p.Arg119Trp, p.Arg177Trp	1, 1, 1	2, 2, 2	13,0,0	161,1,0	127,2,0	4788,19,0	7599,123,1
SPG11	missense	*ENST00000261866, ENST00000559193, ENST00000535302, ENST00000558319, ENST00000427534	c.1108G>A, c.1108G>A, c.1108G>A, c.1108G>A, c.1108G>A	p.Glu370Lys, p.Glu370Lys, p.Glu370Lys, p.Glu370Lys, p.Glu370Lys	0, 0, 0, 0, 0	0, 1, 0, 1, 2	50,0,0	376,13,0	280,11,0	4768,198,1	7661,216,1
SPG11	synonymous	*ENST00000261866, ENST00000535302, ENST00000559193, ENST00000557866, ENST00000427534, ENST00000558319	c.1347C>T, c.1347C>T, c.1347C>T, c.45C>T, c.1347C>T, c.1347C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	50,0,0	387,3,0	288,5,0	4925,44,0	7720,155,5
SPG11	missense	*ENST00000261866, ENST00000558319, ENST00000559193, ENST00000557866,	c.1348A>G, c.1348A>G, c.1348A>G, c.46A>G,	p.Ile450Val, p.Ile450Val, p.Ile450Val, p.Ile16Val,	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0	50,0,0	387,3,0	289,5,0	4925,45,0	7715,161,5

		ENST00000535302, ENST00000427534	c.1348A>G, c.1348A>G	p.Ile450Val, p.Ile450Val							
SPG11	missense	*ENST00000261866, ENST00000535302, ENST00000559193, ENST00000558319, ENST00000557866, ENST00000427534	c.1388T>C, c.1388T>C, c.1388T>C, c.1388T>C, c.86T>C, c.1388T>C	p.Phe463Ser, p.Phe463Ser, p.Phe463Ser, p.Phe463Ser, p.Phe29Ser, p.Phe463Ser	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0	22,21,7	119,211,57	81,158,55	1494,2435,1041	2163,3866,1852
SPG11	missense	*ENST00000261866, ENST00000535302, ENST00000427534, ENST00000558319, ENST00000559193, ENST00000557866	c.1529G>A, c.1529G>A, c.1529G>A, c.1529G>A, c.1529G>A, c.227G>A	p.Ser510Asn, p.Ser510Asn, p.Ser510Asn, p.Ser510Asn, p.Ser510Asn, p.Ser76Asn	0, 0, 0, 0, 0, 0	2, 2, 2, 1, 2, 2	50,0,0	388,1,0	282,0,0	282,0,0	282,0,0
SPG11	splice region, synonymous	*ENST00000261866, ENST00000535302, ENST00000427534, ENST00000559193, ENST00000558319, ENST00000557866	c.1605C>T, c.1605C>T, c.1605C>T, c.1605C>T, c.1605C>T, c.303C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	48,0,0	356,2,0	236,0,0	4909,3,0	7820,3,0
SPG11	missense	*ENST00000261866, ENST00000427534, ENST00000557866, ENST00000535302, ENST00000559193, ENST00000558319	c.1698T>G, c.1698T>G, c.396T>G, c.1698T>G, c.1698T>G, c.1698T>G	p.Asp566Glu, p.Asp566Glu, p.Asp132Glu, p.Asp566Glu, p.Asp566Glu, p.Asp566Glu	0, 0, 0, 0, 0, 0	0, 2, 0, 0, 1, 1	46,2,0	359,17,0	234,8,0	4740,177,0	7629,197,1
SPG11	missense	*ENST00000261866, ENST00000535302, ENST00000558319, ENST00000559193, ENST00000427534	c.1930A>T, c.1930A>T, c.1930A>T, c.1930A>T, c.1930A>T	p.Thr644Ser, p.Thr644Ser, p.Thr644Ser, p.Thr644Ser, p.Thr644Ser	0, 0, 0, 0, 0	0, 1, 2, 1, 2	48,1,0	382,0,0	265,0,0	265,0,0	265,0,0
SPG11	missense	*ENST00000261866, ENST00000535302, ENST00000427534, ENST00000559193,	c.2083G>A, c.2083G>A, c.2083G>A, c.2083G>A,	p.Ala695Thr, p.Ala695Thr, p.Ala695Thr, p.Ala695Thr,	1, 1, 1, 1, 1	2, 2, 2, 2, 2	48,0,0	343,11,0	234,7,1	4786,131,1	7681,147,1

		ENST00000558319	c.2083G>A	p.Ala695Thr							
SPG11	missense, splice region	*ENST00000261866, ENST00000535302, ENST00000558319, ENST00000427534	c.2318T>G, c.2318T>G, c.2318T>G, c.2318T>G	p.Val773Gly, p.Val773Gly, p.Val773Gly, p.Val773Gly	1, 1, 1, 1	2, 2, 2, 2	29,0,0	272,0,0	166,1,0	4812,4,0	7683,4,0
SPG11	***missense***	****ENST00000261866, ENST00000558319, ENST00000427534, ENST00000535302***	***c.2377G>A , c.2377G>A , c.2377G>A***	***p.Val793Met, p.Val793Met, p.Val793Met, p.Val793Met***	***1, 1, 1, 0***	***0, 0, 2, 0***	***44,0,0***	***345,2,0***	***212,3,0***	***212,3,0***	***212,3,0***
SPG11	missense	*ENST00000261866, ENST00000558319, ENST00000535302, ENST00000427534	c.2577A>C, c.2577A>C, c.2577A>C, c.2577A>C	p.Gln859His, p.Gln859His, p.Gln859His, p.Gln859His	0, 0, 0, 0	0, 0, 1, 0	50,0,0	379,1,0	267,0,0	267,0,0	267,0,0
SPG11	synonymous	*ENST00000261866, ENST00000558319, ENST00000535302, ENST00000427534	c.2887A>C, c.2887A>C, c.2887A>C, c.2887A>C	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	49,1,0	387,1,0	279,2,0	4943,14,0	7852,16,0
SPG11	missense, splice region	*ENST00000261866, ENST00000535302, ENST00000558319, ENST00000427534	c.3037A>G, c.3037A>G, c.3037A>G, c.3037A>G	p.Lys1013Glu, p.Lys1013Glu, p.Lys1013Glu, p.Lys1013Glu	0, 0, 0, 0	0, 0, 0, 0	49,1,0	375,7,0	269,2,0	4811,135,1	7712,145,1
SPG11	synonymous	*ENST00000261866, ENST00000559754, ENST00000558988, ENST00000427534, ENST00000535302, ENST00000558319	c.3420G>A, c.149G>A, c.147G>A, c.3420G>A, c.3420G>A, c.3420G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	44,6,0	355,35,0	259,25,2	4455,490,17	7301,554,18
SPG11	missense	*ENST00000261866, ENST00000559754, ENST00000427534, ENST00000558319, ENST00000558988,	c.3446T>C, c.175T>C, c.3446T>C, c.3446T>C, c.173T>C,	p.Leu1149Pro , p.Leu59Pro, p.Leu1149Pro , p.Leu1149Pro	1, 1, 1, 1, 1, 1	2, 2, 2, 2, 2, 2	50,0,0	391,0,0	285,1,0	285,1,0	285,1,0

		ENST00000535302	c.3446T>C	, p.Leu58Pro, p.Leu1149Pro							
SPG11	missense	*ENST00000261866, ENST00000427534, ENST00000558988, ENST00000535302, ENST00000558319	c.3680A>G, c.3680A>G, c.407A>G, c.3680A>G, c.3680A>G	p.Lys1227Arg, p.Lys1227Arg, p.Lys136Arg, p.Lys1227Arg, p.Lys1227Arg	0, 0, 1, 0, 0	0, 2, 0, 0, 2	49,1,0	390,0,0	302,0,0	302,0,0	302,0,0
SPG11	missense	*ENST00000261866, ENST00000535302, ENST00000558319, ENST00000427534, ENST00000559193	c.394A>G, c.394A>G, c.394A>G, c.394A>G, c.394A>G	p.Ser132Gly, p.Ser132Gly, p.Ser132Gly, p.Ser132Gly, p.Ser132Gly	0, 0, 0, 0, 0	0, 0, 0, 0, 0	50,0,0	390,1,0	279,0,0	279,0,0	279,0,0
SPG11	missense	*ENST00000261866, ENST00000427534, ENST00000558319, ENST00000535302	c.3980T>C, c.3980T>C, c.3980T>C, c.3980T>C	p.Ile1327Thr, p.Ile1327Thr, p.Ile1327Thr, p.Ile1327Thr	1, 1, 1, 1	0, 2, 0, 0	50,0,0	369,2,0	263,2,0	263,2,0	263,2,0
SPG11	missense	*ENST00000261866, ENST00000558319, ENST00000535302, ENST00000427534	c.3988C>G, c.3988C>G, c.3988C>G, c.3988C>G	p.Gln1330Glu , p.Gln1330Glu , p.Gln1330Glu	0, 0, 0, 0	0, 0, 0, 1	49,0,0	364,3,0	265,1,0	4562,2,0	6760,2,0
SPG11	synonymous	*ENST00000261866, ENST00000535302, ENST00000427534, ENST00000558319	c.4026A>G, c.4026A>G, c.4026A>G, c.4026A>G	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	388,3,0	287,2,0	4576,11,0	6774,11,0
SPG11	synonymous	*ENST00000261866, ENST00000558319, ENST00000427534, ENST00000535302	c.4260C>T, c.4260C>T, c.4260C>T, c.4260C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	389,0,0	295,1,0	4590,4,0	6788,4,0
SPG11	missense	*ENST00000261866, ENST00000427534, ENST00000535302, ENST00000558319	c.4261G>A, c.4261G>A, c.4261G>A, c.4261G>A	p.Asp1421Asn, p.Asp1421Asn,	0, 0, 0, 0	0, 0, 0, 0	50,0,0	389,0,0	288,2,0	666,2,0	1378,3,0

				p.Asp1421Asn, p.Asp1421Asn							
SPG11	missense	*ENST00000261866, ENST00000427534, ENST00000558319, ENST00000535302	c.4343G>A, c.4343G>A, c.4343G>A, c.4343G>A	p.Cys1448Tyr, p.Cys1448Tyr, p.Cys1448Tyr, p.Cys1448Tyr	1, 1, 1, 1	1, 0, 1, 1	50,0,0	387,1,0	300,0,0	300,0,0	300,0,0
SPG11	missense, splice region	*ENST00000261866, ENST00000558319, ENST00000427534, ENST00000535302	c.5121G>T, c.5121G>T, c.5121G>T, c.5121G>T	p.Glu1707Asp, p.Glu1707Asp, p.Glu1707Asp, p.Glu1707Asp	1, 1, 1, 1	1, 1, 2, 1	50,0,0	389,0,0	302,1,0	4968,11,0	7878,12,0
SPG11	synonymous	*ENST00000261866, ENST00000558319, ENST00000427534, ENST00000535302, ENST00000559511	c.5361C>T, c.5361C>T, c.5361C>T, c.5361C>T, c.209C>T	p.(=), p.(=), p.(=), p.(=), p.(=)	, , , ,	, , , ,	46,1,0	340,1,0	243,0,0	4916,3,0	7826,4,0
SPG11	***feature truncation, frameshift***	****ENST00000261866, ENST00000558319, ENST00000427534, ENST00000535302, ENST00000559511***	***c.5757_5758delAG, c.5757_5758delAG, c.5757_5758delAG, c.5757_5758delAG, c.605_606delAG***	***p.Glu1921SerfsX2, p.Glu1921SerfsX2, p.Glu1921SerfsX2, p.Glu1921SerfsX2, p.Glu204SerfsX2***	***, , , ,***	***, , , ,***	***50,0,0***	***389,1,0***	***302,0,0***	***302,0,0***	***302,0,0***
SPG11	synonymous	*ENST00000261866, ENST00000427534,	c.5796T>C, c.5796T>C,	p.(=), p.(=), p.(=), p.(=),	, , , ,	, , , ,	50,0,0	380,0,0	299,1,0	4596,2,0	6794,2,0

		ENST00000559511, ENST00000535302, ENST00000558319	c.644T>C, c.5796T>C, c.5796T>C	p.(=)							
SPG11	missense	*ENST00000261866, ENST00000558319, ENST00000427534	c.5911A>G, c.5911A>G, c.5911A>G	p.Asn1971Asp, p.Asn1971Asp, p.Asn1971Asp	0, 0, 0	0, 0, 0	50,0,0	383,0,0	293,1,0	293,1,0	293,1,0
SPG11	missense	*ENST00000261866, ENST00000427534, ENST00000558138, ENST00000535302	c.6224A>G, c.6224A>G, c.23A>G, c.5885A>G	p.Asn2075Ser, p.Asn2075Ser, p.Asn8Ser, p.Asn1962Ser	0, 0, 0, 0	0, 0, 0, 0	50,0,0	387,1,0	296,3,0	4925,50,0	7830,56,0
SPG11	synonymous	*ENST00000261866, ENST00000558138, ENST00000427534, ENST00000535302	c.6258G>T, c.57G>T, c.6258G>T, c.5919G>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	49,1,0	385,4,0	294,3,0	4916,57,0	7816,68,0
SPG11	synonymous	*ENST00000261866, ENST00000535302, ENST00000558138, ENST00000427534	c.6330G>A, c.5991G>A, c.129G>A, c.6330G>A	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	385,3,0	289,5,0	4917,53,0	7690,182,9
SPG11	missense	*ENST00000261866, ENST00000427534, ENST00000559511, ENST00000535302	c.6497T>C, c.6497T>C, c.868T>C, c.6158T>C	p.Ile2166Thr, p.Ile2166Thr, p.Ile290Thr, p.Ile2053Thr	1, 1, 1, 1	2, 2, 2, 1	50,0,0	386,1,0	303,0,0	4598,1,0	6794,1,0
SPG11	missense	*ENST00000261866, ENST00000535302, ENST00000559511, ENST00000427534, ENST00000558138	c.6632G>A, c.6293G>A, c.1003G>A, c.6632G>A, c.323G>A	p.Arg2211His, p.Arg2098His, p.Arg335His, p.Arg2211His, p.Arg108His	0, 0, 0, 0, 0	0, 0, 0, 2, 0	48,1,0	359,3,0	294,0,0	4577,15,0	6775,15,0
SPG11	missense	*ENST00000261866, ENST00000535302, ENST00000559511, ENST00000558138	c.6872G>A, c.6533G>A, c.1243G>A, c.563G>A	p.Cys2291Tyr, p.Cys2178Tyr, p.Cys415Tyr,	1, 1, 1, 1	2, 2, 2, 2	50,0,0	383,0,0	302,1,0	302,1,0	302,1,0

				p.Cys188Tyr							
SPG11	***feature elongation, frameshift***	****ENST00000261866, ENST00000558138, ENST00000535302, ENST00000559511***	***c.6952dupC, c.643dupC, c.6613dupC, c.1323dupC***	***p.Arg2318ProfsX22, p.Arg215ProfsX22, p.Arg2205ProfsX22, p.Arg442ProfsX22***	***, , ,***	***, , ,***	***30,5,0***	***156,51,0***	***166,44,0***	***166,44,0***	***166,44,0***
SPG11	synonymous	*ENST00000261866, ENST00000558138, ENST00000535302, ENST00000559511	c.7023C>T, c.714C>T, c.6684C>T, c.1394C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	46,4,0	353,30,0	257,18,0	4652,299,0	7533,328,1
SPG11	missense	*ENST00000261866, ENST00000535302, ENST00000559511	c.7069C>T, c.6730C>T, c.1440C>T	p.Leu2357Phe, p.Leu2244Phe, p.Leu481Phe	0, 0, 1	2, 2, 2	49,1,0	383,2,0	275,1,0	4929,23,0	7834,29,0
SPG11	synonymous	*ENST00000261866, ENST00000559511, ENST00000535302	c.7197G>A, c.1568G>A, c.6858G>A	p.(=), p.(=), p.(=)	, ,	, ,	50,0,0	382,7,0	278,4,0	4838,120,0	7740,129,0
SPG11	missense	*ENST00000261866, ENST00000535302, ENST00000559511	c.7324G>C, c.6985G>C, c.1695G>C	p.Ala2442Pro, p.Ala2329Pro, p.Ala566Pro	1, 1, 1	2, 2, 2	50,0,0	390,1,0	282,0,0	282,0,0	282,0,0
SPG11	missense	*ENST00000261866, ENST00000427534, ENST00000559193, ENST00000535302, ENST00000558319	c.808G>A, c.808G>A, c.808G>A, c.808G>A, c.808G>A	p.Val270Ile, p.Val270Ile, p.Val270Ile, p.Val270Ile, p.Val270Ile	0, 0, 0, 0, 0	1, 2, 2, 1, 2	50,0,0	389,1,0	282,4,0	4865,97,0	7729,144,0
SPG11	missense	*ENST00000261866, ENST00000559193, ENST00000427534, ENST00000558319, ENST00000535302	c.979C>G, c.979C>G, c.979C>G, c.979C>G, c.979C>G	p.Leu327Val, p.Leu327Val, p.Leu327Val, p.Leu327Val, p.Leu327Val	0, 0, 0, 0, 0	0, 1, 0, 0, 0	50,0,0	386,0,0	274,1,0	651,2,0	1364,2,0

TARDBP	synonymous	*ENST00000240185, ENST00000439080	c.1122T>C, c.774T>C	p.(=), p.(=)	,	,	50,0,0	385,0,0	301,1,0	4601,1,0	6803,2,0
TARDBP	missense	*ENST00000240185, ENST00000439080	c.1129T>A, c.781T>A	p.Ser377Thr, p.Ser261Thr	0, 0	1, 1	50,0,0	386,0,0	302,1,0	302,1,0	302,1,0
TARDBP	synonymous	*ENST00000240185, ENST00000473118, ENST00000315091, ENST00000476201	c.312C>T, c.312C>T, c.312C>T, c.435C>T	p.(=), p.(=), p.(=), p.(=)	, , ,	, , ,	50,0,0	382,1,0	270,0,0	270,0,0	270,0,0
TARDBP	missense	*ENST00000240185, ENST00000439080	c.859G>A, c.511G>A	p.Gly287Ser, p.Gly171Ser	0, 0	0, 0	50,0,0	388,2,0	300,0,0	300,0,0	300,0,0
TARDBP	synonymous	*ENST00000240185, ENST00000439080	c.975C>T, c.627C>T	p.(=), p.(=)	,	,	49,0,0	363,1,0	272,2,0	272,2,0	272,2,0
UNC13A	missense	*ENST00000519716, ENST00000428389, ENST00000252773, ENST00000552293, ENST00000551649, ENST00000550896	c.1031T>C, c.1295T>C, c.1031T>C, c.1031T>C, c.1031T>C, c.1031T>C	p.Leu344Pro, p.Leu432Pro, p.Leu344Pro, p.Leu344Pro, p.Leu344Pro, p.Leu344Pro	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 2	15,2,0	109,4,0	96,2,0	4219,15,0	6313,16,0
UNC13A	***inframe deletion***	****ENST00000519716, ENST00000252773, ENST00000428389, ENST00000552293, ENST00000551649, ENST00000550896***	***c.1049_1051delAGG, c.1049_1051delAGG, c.1313_1315delAGG, c.1049_1051delAGG, c.1049_1051delAGG, c.1049_1051delAGG***	***p.Glu350del, p.Glu350del, p.Glu438del, p.Glu350del, p.Glu350del, p.Glu350del***	***, , , , , ***	***, , , , , ***	***14,0,0***	***89,6,0***	***67,7,0***	***67,7,0***	***67,7,0***
UNC13A	missense	*ENST00000519716, ENST00000551649, ENST00000428389, ENST00000252773, ENST00000550896,	c.1075G>A, c.1075G>A, c.1339G>A, c.1075G>A, c.1075G>A,	p.Ala359Thr, p.Ala359Thr, p.Ala447Thr, p.Ala359Thr, p.Ala359Thr,	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 1, 0	7,14,3	38,85,17	32,70,10	1562,2142,979	3649,2792,1092

		ENST00000552293	c.1075G>A	p.Ala359Thr							
UNC13A	***synonymous***	****ENST00000519716, ENST00000428389, ENST00000550896, ENST00000252773, ENST00000551649, ENST00000552293***	***c.1188C>T, c.1452C>T, c.1188C>T, c.1188C>T, c.1188C>T, c.1188C>T***	***p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)***	***, , , , ,***	***, , , , ,***	***1,0,0***	***5,0,0***	***3,0,0***	***4155,6,0***	***6144,6,0***
UNC13A	synonymous	*ENST00000519716, ENST00000551649, ENST00000552293, ENST00000428389, ENST00000252773, ENST00000550896	c.1368C>T, c.1368C>T, c.1368C>T, c.1632C>T, c.1368C>T, c.1368C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	20,2,0	81,0,0	71,2,0	4334,20,0	6519,20,0
UNC13A	splice region, synonymous	*ENST00000519716, ENST00000552293, ENST00000550896, ENST00000428389, ENST00000551649, ENST00000252773	c.153C>T, c.153C>T, c.153C>T, c.417C>T, c.153C>T, c.153C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	48,1,0	360,2,0	302,3,0	4424,12,0	6330,12,0
UNC13A	synonymous	*ENST00000519716, ENST00000551649, ENST00000428389, ENST00000550896, ENST00000552293, ENST00000252773	c.1767C>G, c.1767C>G, c.2031C>G, c.1761C>G, c.1767C>G, c.1767C>G	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	25,5,0	108,38,3	64,12,3	3943,781,33	6017,1536,119
UNC13A	missense	*ENST00000519716, ENST00000252773, ENST00000551649, ENST00000552293, ENST00000550896, ENST00000428389	c.182C>T, c.182C>T, c.182C>T, c.182C>T, c.182C>T, c.446C>T	p.Thr61Met, p.Thr61Met, p.Thr61Met, p.Thr61Met, p.Thr61Met, p.Thr149Met	0, 0, 0, 0, 0, 0	1, 2, 1, 1, 2, 2	50,0,0	366,1,0	296,1,0	4810,19,0	7475,24,0
UNC13A	***missense***	****ENST00000519716, ENST00000551649, ENST00000428389, ENST00000252773,***	***c.2068G>A , c.2068G>A ,***	***p.Ala690Thr, p.Ala690Thr, p.Ala778Thr, p.Ala690Thr,***	***1, 1, 1, 1, 1, 1***	***2, 2, 2, 2, 1, 2***	***45,0,0***	***291,0,0***	***269,1,0***	***269,1,0***	***269,1,0***

		ENST00000550896, ENST00000552293	***c.2332G>A , c.2068G>A , c.2062G>A , c.2068G>A***	***p.Ala688Thr, p.Ala690Thr***							
UNC13A	synonymous	*ENST00000519716, ENST00000428389, ENST00000252773, ENST00000552293, ENST00000551649, ENST00000550896	c.2289T>C, c.2553T>C, c.2289T>C, c.2289T>C, c.2289T>C, c.2283T>C	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	48,0,0	333,1,0	294,0,0	4559,1,0	6709,1,0
UNC13A	***missense***	****ENST00000519716, ENST00000552293, ENST00000428389, ENST00000550896, ENST00000551649, ENST00000252773***	***c.2626C>A, c.2626C>A, c.2890C>A, c.2620C>A, c.2626C>A, c.2626C>A***	***p.Gln876Lys, p.Gln876Lys, p.Gln964Lys, p.Gln874Lys, p.Gln876Lys, p.Gln876Lys***	***1, 1, 1, 1, 1, 1***	***2, 2, 2, , 2, 2***	***36,2,0***	***214,15,0***	***180,16,0***	***180,16,0***	***180,16,0***
UNC13A	missense, splice region	*ENST00000519716, ENST00000552293, ENST00000252773, ENST00000551649, ENST00000428389, ENST00000550896	c.3080C>T, c.3080C>T, c.3080C>T, c.3080C>T, c.3344C>T, c.3074C>T	p.Pro1027Leu , p.Pro1027Leu , p.Pro1027Leu , p.Pro1027Leu , p.Pro1115Leu , p.Pro1025Leu	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 2	45,2,0	336,2,0	253,4,0	4383,58,0	6369,65,0
UNC13A	missense	*ENST00000519716, ENST00000551649, ENST00000428389, ENST00000550896, ENST00000552293,	c.3098T>A, c.3098T>A, c.3362T>A, c.3092T>A, c.3098T>A,	p.Val1033Asp , p.Val1033Asp , p.Val1121Asp	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0	49,0,0	371,1,0	286,0,0	286,0,0	286,0,0

		ENST00000252773	c.3098T>A	, p.Val1031Asp , p.Val1033Asp , p.Val1033Asp							
UNC13A	missense	*ENST00000519716, ENST00000551649, ENST00000428389, ENST00000550896, ENST00000252773, ENST00000552293	c.3101T>C, c.3101T>C, c.3365T>C, c.3095T>C, c.3101T>C, c.3101T>C	p.Leu1034Pro , p.Leu1034Pro , p.Leu1122Pro , p.Leu1032Pro , p.Leu1034Pro , p.Leu1034Pro	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 0, 0	0,0,50	0,0,373	0,0,287	0,0,665	0,0,1378
UNC13A	synonymous	*ENST00000519716, ENST00000551649, ENST00000428389, ENST00000550896, ENST00000552293, ENST00000252773	c.3108G>A, c.3108G>A, c.3372G>A, c.3102G>A, c.3108G>A, c.3108G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	46,4,0	345,29,3	255,37,0	4245,548,16	6795,624,17
UNC13A	missense	*ENST00000519716, ENST00000552293, ENST00000252773, ENST00000551649, ENST00000550896, ENST00000428389	c.3397G>T, c.3397G>T, c.3397G>T, c.3397G>T, c.3391G>T, c.3661G>T	p.Ala1133Ser, p.Ala1133Ser, p.Ala1133Ser, p.Ala1133Ser, p.Ala1131Ser, p.Ala1221Ser	0, 0, 0, 0, 0, 0	0, 0, 0, 0, 1, 0	40,9,1	306,67,0	260,42,0	4122,784,28	6898,851,28
UNC13A	***missense***	****ENST00000519716, ENST00000428389, ENST00000552293, ENST00000550896, ENST00000252773, ENST00000551649***	***c.3461A>G , c.3725A>G , c.3461A>G ,***	***p.Asp1154Gly , p.Asp1242Gly , p.Asp1154Gly ,***	***1, 1, 1, 1, 1, 1***	***1, 0, 0, 1, 1, 1***	***37,0,0***	***215,3,0***	***231,7,0***	***231,7,0***	***231,7,0***

			c.3455A>G , c.3461A>G , c.3461A>G	***p.Asp1152Gly , p.Asp1154Gly , p.Asp1154Gly***							
UNC13A	synonymous	*ENST00000519716, ENST00000428389, ENST00000550896, ENST00000252773, ENST00000552293, ENST00000551649	c.3552A>G, c.3816A>G, c.3546A>G, c.3552A>G, c.3552A>G, c.3552A>G	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	49,1,0	377,1,0	301,2,0	4864,44,0	7694,49,0
UNC13A	synonymous	*ENST00000519716, ENST00000551649, ENST00000552293, ENST00000252773, ENST00000428389, ENST00000550896	c.3576T>C, c.3576T>C, c.3576T>C, c.3576T>C, c.3840T>C, c.3570T>C	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	24,23,3	182,162,44	140,130,35	2152,2111,638	2520,3078,2119
UNC13A	synonymous	*ENST00000519716, ENST00000252773, ENST00000552293, ENST00000550896, ENST00000428389, ENST00000551649	c.3687C>T, c.3687C>T, c.3687C>T, c.3681C>T, c.3951C>T, c.3687C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	50,0,0	346,33,1	273,31,0	4328,552,17	7029,650,17
UNC13A	synonymous	*ENST00000519716, ENST00000552293, ENST00000252773, ENST00000550896, ENST00000428389, ENST00000551649	c.4029C>T, c.4029C>T, c.4029C>T, c.4023C>T, c.4293C>T, c.4029C>T	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	40,0,0	237,30,1	153,26,1	4251,541,18	7061,602,18
UNC13A	***synonymous***	****ENST00000519716, ENST00000428389, ENST00000252773, ENST00000551649***	***c.4251G>A , c.4515G>A , c.4251G>A ,***	***p.(=), p.(=), p.(=), p.(=)***	***, , ,***	***, , ,***	***33,1,0***	***171,20,0***	***123,19,0***	***123,19,0***	***123,19,0***

			c.4251G>A								
UNC13A	***missense***	****ENST00000519716, ENST00000428389, ENST00000252773, ENST00000551649***	***c.4259C>A, c.4523C>A, c.4259C>A, c.4259C>A***	***p.Pro1420Gln , p.Pro1508Gln , p.Pro1420Gln , p.Pro1420Gln***	***0, 0, 0, 0***	***0, 0, 0, 0***	***30,7,0***	***114,86,1***	***63,117,0***	***63,117,0***	***63,117,0***
UNC13A	synonymous	*ENST00000519716, ENST00000428389, ENST00000252773, ENST00000551649, ENST00000552293, ENST00000550896	c.4380G>C, c.4644G>C, c.4380G>C, c.4380G>C, c.4305G>C, c.4299G>C	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	45,0,0	299,0,0	255,1,0	255,1,0	255,1,0
UNC13A	synonymous	*ENST00000519716, ENST00000552293, ENST00000550896, ENST00000428389, ENST00000252773, ENST00000551649	c.4497G>A, c.4422G>A, c.4416G>A, c.4761G>A, c.4497G>A, c.4497G>A	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	24,19,4	165,148,28	161,99,23	2827,1789,286	5371,2036,296
UNC13A	synonymous	*ENST00000519716, ENST00000552293, ENST00000252773, ENST00000550896, ENST00000428389, ENST00000551649	c.474G>C, c.474G>C, c.474G>C, c.474G>C, c.738G>C, c.474G>C	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	1,10,39	12,100,270	4,61,233	89,1095,3618	214,1818,5405
UNC13A	***synonymous***	****ENST00000519716, ENST00000552293, ENST00000551649, ENST00000252773, ENST00000550896, ENST00000428389***	***c.5079C>A, c.5061C>A, c.5136C>A, c.5079C>A, c.4998C>A, c.5343C>A***	***p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)***	***, , , , , , , , , , , ,***	***, , , , , , , , , , , ,***	***0,0,0***	***0,1,0***	***0,0,0***	***3702,654,30***	***5596,1285,89***
UNC13A	missense	*ENST00000519716, ENST00000550896, ENST00000552293,	c.694C>T, c.694C>T, c.694C>T,	p.Pro232Ser, p.Pro232Ser, p.Pro232Ser,	0, 0, 0, 0, 0, 0	0, 1, 0, 0, 0, 0	50,0,0	367,0,0	305,1,0	305,1,0	305,1,0

		ENST00000551649, ENST00000252773, ENST00000428389	c.694C>T, c.694C>T, c.958C>T	p.Pro232Ser, p.Pro232Ser, p.Pro320Ser							
UNC13A	synonymous	*ENST00000519716, ENST00000428389, ENST00000550896, ENST00000252773, ENST00000552293, ENST00000551649	c.771C>G, c.1035C>G, c.771C>G, c.771C>G, c.771C>G, c.771C>G	p.(=), p.(=), p.(=), p.(=), p.(=), p.(=)	, , , , ,	, , , , ,	6,0,0	49,3,0	35,3,0	4432,85,0	7098,97,0
UNC13A	***missense***	****ENST00000519716, ENST00000550896, ENST00000552293, ENST00000551649, ENST00000252773, ENST00000428389***	***c.905A>C, c.905A>C, c.905A>C, c.905A>C, c.905A>C, c.1169A>C***	***p.His302Pro, p.His302Pro, p.His302Pro, p.His302Pro, p.His302Pro, p.His390Pro***	***0, 0, 0, 0, 0, 0***	***0, 1, 0, 2, 0, 1***	***14,1,0***	***75,5,0***	***76,3,0***	***76,3,0***	***76,3,0***
UNC13A	***missense***	****ENST00000519716, ENST00000551649, ENST00000552293, ENST00000252773, ENST00000550896, ENST00000428389***	***c.907T>C, c.907T>C, c.907T>C, c.907T>C, c.907T>C, c.1171T>C***	***p.Ser303Pro, p.Ser303Pro, p.Ser303Pro, p.Ser303Pro, p.Ser303Pro, p.Ser391Pro***	***0, 0, 0, 0, 0, 0***	***0, 2, 1, 2, 1, 1***	***11,1,0***	***70,8,0***	***72,8,0***	***72,8,0***	***72,8,0***
VAPB	***inframe deletion***	****ENST00000475243***	***c.474_476 delTTC***	***p.Ser159del***			***50,0,0***	***386,2,0***	***278,0,0***	***278,0,0***	***278,0,0***
VAPB	missense	*ENST00000475243	c.510G>A	p.Met170Ile	0	0	50,0,0	385,4,0	279,1,0	4939,19,0	7854,20,0
VCP	***synonymous***	****ENST00000358901***	***c.1641G>A***	***p.(=)***			***50,0,0***	***386,1,0***	***299,0,0***	***299,0,0***	***299,0,0***
VCP	synonymous	*ENST00000358901	c.1704A>G	p.(=)			48,1,0	373,5,0	298,5,0	4930,50,1	7835,61,1
VCP	***missense***	****ENST00000358901***	***c.2092G>C***	***p.Ala698Pro***	***1***	***2***	***49,0,0***	***341,7,0***	***272,7,0***	***272,7,0***	***272,7,0***
VCP	missense	*ENST00000358901	c.2249A>G	p.Asn750Ser	0	0	50,0,0	388,1,0	306,0,0	306,0,0	306,0,0
VCP	***synonymous***	****ENST00000358901***	***c.2406T>C***	***p.(=)***			***45,0,0***	***282,0,0***	***262,0,0***	***4559,3,0***	***6762,3,0***

* Denotes canonical transcript

SIFT predition values of “0” and “1” denote “tolerated” and “deleterious” predictions respectively

PolyPhen prediction values of “0”, “1” and “2” denote “benign”, “possibly damaging” and “probably damaging” predictions respectively

Mutations failing variant quality control (supplementary materials and methods) are shown in bold/ italics. It should be noted that these are anticipated to include true variants in addition to sequencing artefacts.

Table S3: Co-occurrence of Mendelian ALS gene variants

*Frequency Threshold	Cases with 1 mutation	Cases with 2 mutations	Controls with 1 mutation	Controls with 2 mutations	**p
0	55	3	12	0	0.36
0.0005	63	4	17	0	0.45
0.005	79	7	34	0	0.76
0.05	114	16	49	7	0.72

*Denotes the cut-off carrier frequency for variants among European and Global reference cohorts. Variants exceeding a given threshold were excluded from analysis. Reference cohorts included individuals resequenced by NHLBI exome sequencing and 1000 genomes project only. Only nonsynonymous and splice variants were included in analyses.

** Probability that the observed frequency of cases carrying multiple Mendelian ALS gene variants is in excess of chance expectation (see Materials and Methods for details)

Total number of cases=444, total number of controls =311

Table S4: Co-occurrence of ALS gene variant

*Frequency Threshold	Cases with 1 variant	Cases with 2 variants	Controls with 1 variant	Controls with 2 variants	**p
0	77	6	34	4	0.85
0.0005	103	9	50	7	0.99
0.005	151	30	95	13	1.00
0.05	247	90	160	72	1.00

*Denotes the cut-off carrier frequency for variants among European and Global reference cohorts. Variants exceeding a given threshold were excluded from analysis. Reference cohorts included individuals resequenced by NHLBI exome sequencing and 1000 genomes project only. Only nonsynonymous and splice variants were included in analyses.

** Probability that the observed frequency of cases carrying multiple ALS gene variants is in excess of chance expectation (see Materials and Methods for details)

Total number of cases=444, total number of controls =311

Table S5: Carriers of established and possible ALS variants

Gene	Mutation	C9orf72 Repeat Expansion	Family History	Gender	Site Of Onset	Cognitive Status	Age Of Onset (Years)	Survival (Months)
ALS2	c.2098A>G(p.Thr700Ala)	-	-	M	spinal	NA	67	39
ALS2	c.3094C>T(p.Arg1032Cys)	-	-	M	bulbar	co-morbid FTD	57	>61
ALS2	c.2408A>G(p.Lys803Arg)	-	-	M	spinal	NA	58	>44
ALS2	c.2606A>C(p.Gln869Pro)	-	-	M	spinal	normal	57	>55
ALS2, SETX	c.2098A>G(p.Thr700Ala), c.7682C>T(p.Ser2561Leu)	-	-	M	spinal	NA	84	58
C9orf72		+	+	M	spinal	NA	35	70
C9orf72		+	+	M	bulbar	NA	NA	NA
C9orf72		+	-	M	both	NA	34	67
C9orf72		+	-	F	spinal	NA	51	13
C9orf72		+	-	F	bulbar	NA	60	20
C9orf72		+	-	M	bulbar	NA	47	30
C9orf72		+	-	M	spinal	NA	66	8
C9orf72		+	-	M	spinal	NA	44	23
C9orf72		+	-	F	bulbar	NA	43	36
C9orf72		+	-	F	bulbar	NA	52	17
C9orf72		+	-	M	spinal	NA	68	NA
C9orf72		+	+	F	bulbar	NA	82	12
C9orf72		+	-	F	spinal	NA	42	54
C9orf72		+	-	M	spinal	NA	NA	NA
C9orf72		+	-	F	spinal	normal	55	30
C9orf72		+	+	F	both	normal	44	50
C9orf72		+	-	M	spinal	cognitively impaired	55	24

C9orf72		+	-	M	spinal	co-morbid FTD	62	52
C9orf72		+	+	M	spinal	co-morbid FTD	56	16
C9orf72		+	-	F	spinal	normal	44	37
C9orf72		+	+	F	spinal	co-morbid FTD	65	16
C9orf72		+	+	M	spinal	co-morbid FTD	65	25
C9orf72		+	-	M	spinal	behaviourally impaired	48	>66
C9orf72		+	-	F	bulbar	normal	63	14
C9orf72		+	-	F	spinal	cognitively impaired	49	37
C9orf72		+	-	F	spinal	cognitively impaired	62	18
C9orf72		+	+	M	bulbar	normal	59	19
C9orf72		+	+	F	bulbar	normal	64	41
C9orf72		+	+	F	bulbar	cognitively impaired	65	33
C9orf72		+	+	M	bulbar	normal	66	32
C9orf72		+	-	M	spinal	co-morbid FTD	36	46
C9orf72		+	-	M	bulbar	NA	59	25
C9orf72		+	-	F	spinal	NA	72	>87
C9orf72		+	-	M	bulbar	No	44	29
C9orf72		+	+	M	spinal	cognitively and behaviourally impaired	59	17
C9orf72		+	+	F	spinal	NA	52	>41
C9orf72, CHMP2B	c.123G>T(p.Gln41His)	+	-	M	spinal	NA	55	48
C9orf72, SETX	c.2842C>A(p.Pro948Thr)	+	+	M	bulbar	co-morbid FTD	58	17
C9orf72, SPG11	c.3680A>G(p.Lys1227Arg)	+	+	M	spinal	NA	65	43
CHMP2B	c.118A>G(p.Lys40Glu)	-	-	M	spinal	NA	61	NA

DCTN1	c.2887-2A>G()	-	-	M	spinal	NA	63	26
DCTN1, SPG11	c.2887-2A>G(), c.1529G>A(p.Ser510Asn)	-	-	M	spinal	NA	63	42
DPP6	c.883G>A(p.Glu295Lys)	-	-	F	bulbar	normal	62	21
ELP3	c.326G>A(p.Cys109Tyr)	NA	-	F	spinal	NA	68	12
ELP3	c.206G>T(p.Arg69Leu)	-	-	F	bulbar	cognitively impaired	68	21
ELP3	c.326G>A(p.Cys109Tyr)	-	+	M	bulbar	normal	67	NA
FGGY	c.1716G>A(p.Met572Ile)	-	-	M	spinal	NA	75	11
FUS	c.1574C>T(p.Pro525Leu)	NA	-	M	spinal	NA	13	17
FUS	c.1574C>T(p.Pro525Leu)	-	-	F	bulbar	normal	21	11
HFE	c.766G>A(p.Val256Ile)	-	-	M	spinal	cognitively impaired	76	7
ITPR2	c.3614G>A(p.Arg1205Gln)	-	-	F	bulbar	NA	79	19
MAPT	c.698C>T(p.Pro233Leu)	-	-	M	spinal	normal	57	>55
MAPT	c.284C>T(p.Thr95Met)	-	-	F	spinal	normal	81	>48
OPTN	c.1192C>G(p.Gln398Glu)	-	-	F	spinal	NA	NA	NA
PON2	c.661T>G(p.Ser221Ala)	-	-	F	spinal	co-morbid FTD	58	12
SETX	c.7645G>A(p.Val2549Ile)	-	-	F	spinal	NA	64	24
SETX	c.5842A>G(p.Met1948Val)	-	-	M	spinal	NA	47	NA
SETX	c.2755G>C(p.Val919Leu)	-	-	M	bulbar	co-morbid FTD	68	15
SETX	c.7682C>T(p.Ser2561Leu)	-	-	M	spinal	cognitively impaired	57	33
SETX	c.5587A>G(p.Thr1863Ala)	-	+	F	other	NA	61	>103
SETX	c.7645G>A(p.Val2549Ile)	-	-	F	bulbar	cognitively and behaviourally impaired	76	26
SETX	c.2975A>G(p.Lys992Arg)	-	+	M	bulbar	NA	78	14
SPG11	c.4343G>A(p.Cys1448Tyr)	-	-	M	spinal	NA	63	33

SPG11	c.1930A>T(p.Thr644Ser)	-	+	F	bulbar	NA	68	27
SPG11	c.394A>G(p.Ser132Gly)	-	-	M	spinal	normal	51	>42
SPG11	c.2577A>C(p.Gln859His)	-	-	F	bulbar	NA	59	NA
SPG11	c.7324G>C(p.Ala2442Pro)	-	-	M	spinal	NA	60	21
TARDBP, ALS2	c.859G>A(p.Gly287Ser), c.2566A>G(p.Thr856Ala)	-	-	M	bulbar	normal	67	>51
TARDBP, SETX	c.859G>A(p.Gly287Ser), c.814C>G(p.His272Asp)	-	-	F	bulbar	NA	66	49
UNC13A	c.3098T>A(p.Val1033Asp)	-	-	M	spinal	NA	80	25
VCP	c.2249A>G(p.Asn750Ser)	-	-	F	bulbar	NA	63	>35

ALS2 - ENST00000264276; *CHMP2B* - ENST00000263780; *DCTN1* - ENST00000361874; *DPP6* - ENST00000377770; *ELP3* - ENST00000256398; *FGGY* - ENST00000371218; *FUS* - ENST00000254108; *HFE* - ENST00000357618; *ITPR2* - ENST00000381340; *MAPT* - ENST00000344290; *OPTN* - ENST00000263036; *PON2* - ENST00000222572; *SETX* - ENST00000224140; *SPG11* - ENST00000261866; *TARDBP* - ENST00000240185; *UNC13A* - ENST00000519716; *VCP* - ENST00000358901