Supplementary Appendix

Supplement to: Nangalia J, Massie CE, Baxter EJ, et al. Somatic *CALR* mutations in myeloproliferative neoplasms with nonmutated *JAK2*. N Engl J Med 2013;369:2391-405. DOI: 10.1056/NEJMoa1312542

SUPPLEMENTARY APPENDIX

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Supplementary Methods

Patient samples

Samples were obtained from patients with myeloproliferative neoplasms and other cancers following written informed consent and local ethics committee approval. The study was conducted from 2010 to 2013. Tumor samples were obtained by isolating granulocytes from peripheral blood. Constitutional samples were obtained from either buccal swabs, T-cell or cultured T-cell DNA. Empirical testing identified cultured T-cells as having the lowest contamination with known myeloproliferative neoplasm driver mutations eg *JAK2*^{V617F}, particularly, in patients with myelofibrosis (MF) or high clonal burden (Figure S1). All constitutional samples were screened by qPCR for the presence of *JAK2*^{V617F} and/or *MPL*$^{W515L/K}$ mutations known to be present in matched tumor samples, and patients with contaminated buccal or T-cell DNA were prioritized for T-cell cultures. In addition, T-cell cultures were prioritised for patients with high-burden disease to minimise contamination of constitutional material with neoplastic cells. Overall, 88% of constitutional DNA samples for patients with MF were derived from T-cell cultures. CD3-positive T-cells were isolated from MPN patients (CD3 MicroBead kit, Miltenyi Biotec) and stimulated using CD2, CD3 and CD28-coated beads for 72 hours in RPMI supplemented with 10% human albumin solution (HAS) and grown in the same media supplemented with IL-2 for up to 14 days. If further expansions of cell numbers were required this stimulation protocol was repeated. Where constitutional DNA was limited, whole genome amplification (WGA) was performed to supplement unamplified constitutional DNA (Figure S2).

Sequencing and genomic alignment

Matched pairs of constitutional and tumor DNA from each MPN patient were used to generate genomic libraries enriched for protein-coding exons using Agilent SureSelect All Exome 50Mb RNA probe sets, as previously described[1–3]. These Illumina sequencing libraries containing an average insert size of 200-300bp were subjected to 75bp paired-end sequencing on the Illumina HiSeq platform. Sequencing data were mapped to the hg19 reference genome using the BWA algorithm on default settings.

Somatic variant calling

Somatic variants were called by pairwise comparison of tumor and constitutional sequence reads using previously described methods for substitutions (CaVEMan) and insertions or deletions (Pindel)[1–3]. Targeted searches of unfiltered calls were undertaken for genes commonly affected by insertions or deletions (*CALR*, *TET2*, *ASXL1*) to ensure that variants were not missed due to data filtering. In addition, due to poor coverage of exon 9 *CALR*, unmapped mutant reads were recovered by mapping reads to reference mutant *CALR* alleles.

Somatic Variant Validation

Somatic variants were validated using an Agilent SureSelect custom bait-set with tiling coverage of 125bp around the original exome mutation calls. Fresh DNA aliquots were whole genome amplified for bait-capture and sequencing as described above. Mutations were marked as 'confirmed somatic' if they were identified in tumor samples from both exome and validation experiments and if they were absent or >=3-fold lower in constitutional samples in both exome and validation experiments. It was not possible to design capture probes for all regions and at a small proportion of sites sequencing depth was too low to confidently validate somatic mutations, therefore we report manually curated variants with convincing evidence for the mutation based on exome data (high coverage, good quality sequencing, high fraction of reads reporting the variants in the tumor and no reads reporting the variant in matched germline sequencing data).

Testing for enrichment, dependency and mutual exclusivity of mutations

To determine whether the frequency of missense, nonsense and splice site mutations was higher than expected by chance for each gene, we adapted a previously described method[4]. Briefly, the rate of mutations is modeled as a Poisson process, with a rate given by a product of the mutation rate and the impact of selection. In particular, we use 12 parameters to describe the different rates of the 12 possible single nucleotide substitutions, 2 parameters to better account for the CpG effect on C>T transitions in each strand, and 3 selection parameters to measure the observed-over-expected ratio of missense (wMIS), nonsense (wNON) and essential splice site (wSPL) mutations. For example, the expected number of A>C missense mutations is modeled as: $\text{rate}_{\text{misA>C}}$ = (t)*(AtoC)*(wMIS)*($L_{\text{misA>C}}$), $L_{\text{misA>C}}$ being the number of sites that can suffer a missense A>C mutation (which is calculated for any particular sequence). "t" refers to the overall mutation rate or the density of mutations. The likelihood of observing $n_{\text{misA>C}}$ missense A>C mutations given the expected $\text{rate}_{\text{misA>C}}$ is then calculated as Lik = $\text{Pois}(n_{\text{misA>C}} | \text{rate}_{\text{misA>C}})$. The likelihood of the entire model is the product of all individual likelihoods. This allows us to quantify the strength of selection while avoiding the confounding effect of gene length, sequence composition and different rates of each substitution type. To obtain accurate estimates of the relative rates of each substitution type, the 14 rate parameters were estimated from the entire collection of mutations. These rates are shared by all genes and maximum-likelihood estimates for "wMIS", "wNON" and "wSPLICE" are obtained for each gene. Likelihood Ratio Tests are then used to test deviations from neutrality (wMIS = 1, wNON = 1 or wSPL = 1). Owing to the limited number of mutations, mutation rates were assumed constant among genes but an additional Likelihood Ratio Test was performed for each gene to detect violations of this assumption (comparing the observed number of synonymous mutations to the assumed mutation rate). No gene was found to deviate significantly from its estimated mutation rate in this dataset (q>0.05 for all genes). For Indels we test for significant enrichment of indel recurrence within gene coding sequences compared to the expected background rate, under a uniform distribution model. Interactions between mutations were assessed to determine any co-dependence or mutual exclusivity using previously described methods[1].

Clonal analysis of mutations identified by exome sequencing

Plots in Figure S9 were produced using a previously developed Bayesian Dirichlet process to model clusters of clonal and subclonal point mutations, allowing inference of the number of subclones, the fraction of cells within each subclone and the number of mutations present within each subclonal population[5]. Within this model, the number of reads bearing the *i*th mutation, y_i, is drawn from a binomial distribution

$$y_i \sim Bin(N_i, \zeta_i \pi_i), with\ \pi_i \sim DP(\alpha P_0)$$

where N_i is the total number of reads at the mutated base and ζ_i is the expected fraction of reads that would report a mutation present in 100% of cells at that locus. $\pi_i \in (0, 1)$, the fraction of cells carrying the ith mutation, is modelled as coming from a Dirichlet process. We use the stick-breaking representation of the Dirichlet process:

$$\omega_h = V_h \prod_{l<h} (1 - V_l)\text{, with } V_h \sim Beta(1, \alpha)$$

where ω_h is the weight of the hth mutation cluster, i.e. the proportion of all somatic mutations specific to that cluster.

Gibbs sampling was used to estimate the posterior distribution of the parameters of interest, implemented in R, version 2.11.1. The Markov chain was run for 10,000 iterations, of which the first 2,000 were discarded. The median of the density was estimated from π_h, each weighted by the associated value of ω_h, using a Gaussian kernel, implemented in the R libraries *stats*. In Figure S9 the median values are plotted as a purple line and the 95% confidence intervals as a pale blue region. The grey histogram shows the observed frequency density of mutations as a function of the fraction of cells bearing the mutation.

CALR targeted sequencing

Sanger sequencing was used to validate exome calls for *CALR* (in addition to validation of exome sequencing *CALR* variants by custom bait capture and resequencing), test constitutional material and to genotype DNA samples from additional patient samples and normal controls as detailed in Table 1. Sequence trace analysis was performed using Mutation Surveyor (SoftGenetics, State College, Warrington, United Kingdom). Sensitivity of Sanger sequencing was established by testing a mixture of plasmids (pGEM-T Easy vector (Promega)) containing either *CALR* exon 9 wildtype or mutant (L367fs*46) in varying proportions. In addition, we mined publicly available whole genome and exome sequencing data sets of multiple malignancies and a panel of controls for the presence of *CALR* mutations. Germline variants in *CALR* were infrequently found (9 and 12 base pair deletions in exon 9) and were confirmed using data from the 1000-Genomes project, panels of healthy blood exome sequencing data and targeted sequencing of *CALR* mutations in matched constitutional samples (T-cells).

Cell line cultures and transfection

HEK293T and 32D cells were cultured in RPMI supplemented with 10% fetal calf serum, L-glutamine, penicillin/streptomycin as well as IL-3 for 32D cells.

Plasmid construction

CALR-FLAG plasmid constructs were made using PCR fragments amplified from cDNAs derived from patients with either CALR insertion or deletion. The common forward primer for the PCR was 5′-ATCGAAGCTTATGCTGCTATCCGTGCCGC-3′ and the reverse primers were as follows:

Wildtype:	5′-ATCGCTCGAGCAGCTCGTCCTTGGCCTGGC-3′
L367fs*46:	5′-ATCGCTCGAGGGCCTCAGTCCAGCCCTGGAG-3′
K385fs*47:	5′-ATCGCTCGAGGGCCTCAGTCCAGCCCTGGA-3′

The PCR fragments were cloned into the HindIII/XhoI sites in pCDNA5_FRT_3XFLAG-c-term (UK918, David Ron). Similarly, PCR fragments were amplified from cDNAs derived from patients with either *CALR*^{K385fs*47} (common insertion) or *CALR*^{L367fs*46} (common deletion) with the following primers:

Forward primer 5'-ATTATCTAGAGGAGGGTCGTTTTAAAGGGC-3'
Reverse primer 5'-ATTAAGATCTGAGTGGAGGAGGGGAACAAA-3'

PCR fragments were cloned into XbaI/BglII sites of pCDF1-MSC2-EF2-copGFP (System Biosciences).

Immunofluorescence and confocal microscopy

COS-7 cells grown on cover glass were transiently cotransfected with vectors encoding for wildtype or mutant CALR-FLAG and either a Golgi reporter Galt-YFP (β1,3-galactosyltransferase fused to Yellow Fluorescent Protein (YFP)[6] or CALR-GFP[7]. Cells were fixed with 2% paraformaldehyde followed by permeablization in 1% Triton X100. Cells were then stained with a primary mouse anti-FLAG (Sigma, diluted 1:200) and goat anti-mouse DyLight543 conjugated IgG secondary antibody (1:200, Jackson ImmunoResearch Laboratories). Cytospin slides were prepared from myeloid cells of normal or mutant CFU-GM colonies derived from two *CALR*-mutated patients with essential thrombocythemia (ET) - one patient with *CALR*^{K385fs*47} (common insertion) and one patient with *CALR*^{L367fs*46} (common deletion). The cells were fixed in methanol/acetone (1:1) and then co-stained with primary rabbit anti N-term CALR (ab39897, Abcam) and mouse anti PDI (Assay Design). Goat anti rabbit AlexaFluor 555 and anti mouse AlexaFluor 488 conjugated IgG secondary antibodies were used (1:500, Invitrogen). CALR localization was visualized by immunofluorescence using a Zeiss 510 Meta laser scanning confocal system with Plan-Apochromat x63 oil immersion lens (NA = 1.6) to acquire fluorescence microscopy images as described. Experiments were repeated three times and samples were tested in triplicate during each experiment. Representive images are shown.

Protein analysis and immunoblotting

Cells were lysed in SDS-sample buffer and samples run on a SDS polyacrylamide gel. Western blot analyses were performed using the following antibodies: CALR (ab39897, Abcam), Flag (F1804, Sigma), B-Actin (sc47778, Santa Cruz).

Flow cytometric analysis of cell surface calreticulin

Whole peripheral blood lysed for red blood cells and 32D cells were analysed for cell surface CALR by a Becton Dickinson (BD) Fortessa LSR II and data were analysed in Flowjo (Treestar, USA). Antibodies used were as follows: CALR-PE (abcam83220, FMC-75, Abcam), CD15- FITC (BD), CD19 Alexa647 (BD), CD4 APC (BD). White blood cells were gated by forward and side scatter, and for viability (DAPI-negative) prior to assessment of CALR positive cells in viable, granulocytic (CD15 positive) or lymphocytic (CD647 or CD4 positive) compartments. 32D cells were gated on GFP positive and negative populations. Mean fluorescence intensity and percentage positive calreticulin cells were calculated in relation to an IgG1 PE control (BD).

Hematopoietic stem and progenitor cell isolation

Peripheral blood mononuclear cells were depleted of differentiated hematopoietic cells using the EasySep Human Hematopoietic Progenitor enrichment kit (#19056 STEMCELL Technologies) and stained with anti-CD34 Per-CpCy5.5 (clone 581 Biolegend), anti-CD38 FITC (clone HIT2, BD), anti-CD90 APC (clone 5E10, BD), anti-FLK2 PE (clone BV10A4H2, Biolegend), anti-CD10 APC-Cy7 (clone HI10a, Biolegend), and anti-CD45RA Horizon V450 (HI100, BD). HSCs were isolated as lin^{-}CD34^{+}CD38^{-}CD45RA^{-}CD90^{+}, CMPs as lin^{-}CD34^{+}CD38^{+}CD90^{-}CD10^{-}FLK2^{+}CD45RA^{-}, GMPs as lin^{-}CD34^{+}CD38^{+}CD90^{-}CD10^{-}FLK2^{+}CD45RA^{+} and MEPs as lin^{-}CD34^{+}CD38^{+}CD90^{-}CD10^{-}FLK2^{-}CD45RA^{-}. The CMP, MEP and GMP fractions were placed directly into colony forming assays (Methocult 04435; STEMCELL) as described by the manufacturer. Day 10 colonies were picked and genotyped for *CALR* exon 9 was by PCR and Sanger sequencing.

Clonal analysis

Erythroid colonies were grown from patients harbouring *CALR* mutations and clones were genotyped for all mutations identified by exome sequencing to empirically determine the underlying clonal hierarchy. Briefly, peripheral blood mononuclear cells were isolated from fresh patient blood samples, cultured for 10 days in MethoCult (04034; STEMCELL) and single erythroid (BFU-E) or granulocyte-monocyte (CFU-GM) colonies were plucked into separate aliquots of RLT lysis buffer (Qiagen) to allow subsequent DNA extraction. Using Sanger sequencing, we genotyped each colony DNA for all coding mutations identified by exome sequencing and integrated these genotyping results to empirically determine the clonal hierarchy for individual patients.

Statistical analysis

Differences between patients with *CALR* mutations versus *JAK2* mutations were analyzed by *t* test (hemoglobin) where data were normally distributed, Wilcoxon rank-sum test (platelets, white cell count, age at diagnosis) where data distribution were skewed, and Fisher's exact test for categorical data. Comparison of number of somatic mutations between MPN subtypes was performed using a one-way analysis of variance and pairwise *t* tests with multiple hypotheses correction.

PCR and sequencing primers

Sample	Gene	Forward Primer (5'→3')	Reverse Primer (5'→3')
	CALR	CCTGCAGGCAGCAGAGAAAC	ACAGAGACATTATTTGGCGCG
PD4063	*FAT2*	GGTGTGGAGAATTGGGGTAT	GACCCCTATCTTGGGGACAT
PD4063	*EIF5B*	GGGAGAAAGTGAAGGCAGTG	CCTCCGTTTTGCTTTGTCAT
PD4063	*KIAA1244*	TGCTTTTTGGTTTCCAGGTC	AGGCAGACACCTGCAGAAAT
PD4063	*ZNF561*	CCAAAAGGCATTTAGGACAAA	GCGAACTCATACCGGAGAGA
PD8632	*COL1A1*	CTCCGGATGTTCTCGATCTG	CCCCACTCTCTTCCCTCTCT
PD8632	*COL4A2*	GCTGGTTGGCTGATTCTCTC	GCTCCCCCAGGTTATAAAGG
PD8632	*LRTM1*	GCCATGGCACTGACCTAAAT	TGCTCCTGTTTTCCAGTGTG
PD8632	*OR52I2*	GTGGGGGTTATGGCTTTGTA	TCATGAACCCAGGTTGGAAT
PD6563	*KNTC1*	TTGTCTTTTGCAGAGAGTGGTT	AATGGAGCGAGACCCTGTC
PD6563	*ABCC9*	CACCAGGAACTTACTCCAGTTGTG	GCAGATCATCCAAATCGGACTCCA
PD6563	*DIP2C*	CCGGAAACCATATGCTTGAT	GGCTCCGATCAGTGAATACC
PD6563	*ARHGAP32*	AGGCCCTTGACGACTGCTATCTTT	TGCATGGTTTGCTACTCCTCTGGA
PD6563	*B4GALNT2*	TGCTACTGGGCAAGCGACTATGAT	TTGCTGGAACAGAGTGAGAATGGG
PD6563	*IFNAR1*	TCCCTGTGCTGGGAGCAATCATTA	CAGGTCATTCTGACAAATCAGTAAGTAGAG
PD6563	*GTPBP2*	AAACGCTATCAATGGCCTGCCAAG	TGCCTCCTGTTTGGACTGTCTTCA
PD6563	*STX11*	CGTACACGAGCTCTTCTTGCAGAT	AAGACTGAGTTCCGGAAAGGGTTG
PD6561	*CELF5*	TAGGAACAAGCCGAGGACAC	GCCATGCCTACAGCCTCAG
PD6561	*CRYGN*	CCTCCAGCAGGAAGACTCAG	GGGCTTTATGAACCGAGTGA
PD6561	*GK5*	TTCTATGGGGAAGGGGAAGT	AACGTGGCTATGTTGTTGTTG
PD6561	*OR4C6*	AGACACCCTCTCCAAGAGCA	CCTCCTACCATTAGGCAGCA
PD6561	*PDE4B*	ACCCAAGGTGTTAGCAACTGA	TGTTCAGCTTAGTTTGGTTTGG
PD6561	*KIAA0802*	TACCTCACCCTTCGGTGACT	GCTGACTTCAGCTCCTCCTG
PD6561	*KIAA0802*	TACCTCACCCTTCGGTGACT	GCTGACTTCAGCTCCTCCTG
PD7442	CPA4	CCCTTCAGCTCAATTTCTGG	CTTGCAAATCACTCCGGTCT

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6. Avezov E, Frenkel Z, Ehrlich M, Herscovics A, Lederkremer GZ. Endoplasmic reticulum (ER) mannosidase I is compartmentalized and required for N-glycan trimming to Man5-6GlcNAc2 in glycoprotein ER-associated degradation. Mol Biol Cell 2008;19(1):216–25.
7. Shaffer KL, Sharma A, Snapp EL, Hegde RS. Regulation of protein compartmentalization expands the diversity of protein function. Dev Cell 2005;9(4):545–54.

Figure S1

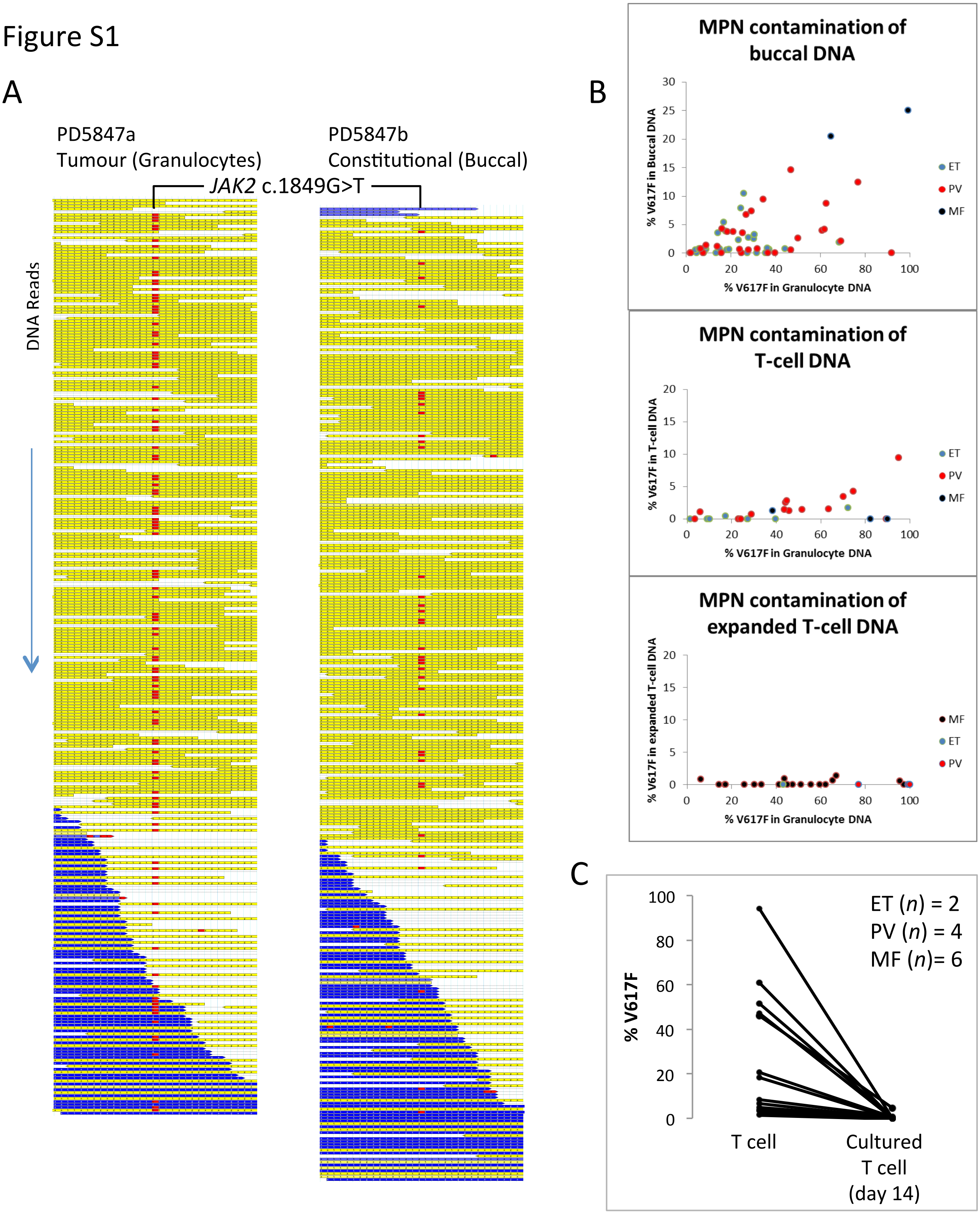

Figure S1 MPN patients with high burden disease frequently show contamination of buccal and T cell DNAs with the *JAK2*^{V617F} mutation. (A) Exome sequencing reads showing the *JAK2*^{V617F} mutation in 47% of reads in a granulocyte DNA sample and 14.6% of reads in the matched constitutional contaminated buccal DNA. Mismatched bases are highlighted in red, forward and reverse sequence reads are shown in blue and yellow, respectively. (B) Scatter plots showing the relationship between JAK2^{V617F} allele burden in DNAs isolated from granulocyte versus DNAs from buccal swabs, isolated T-cells or T-cell cultures (assessed by qPCR); ET (blue), PV (red), MF (black) (C) Paired analysis of *JAK2*^{V617F} contamination in T-cell isolates and T-cells following 14 days of culture i*n vitro*.

Figure S2

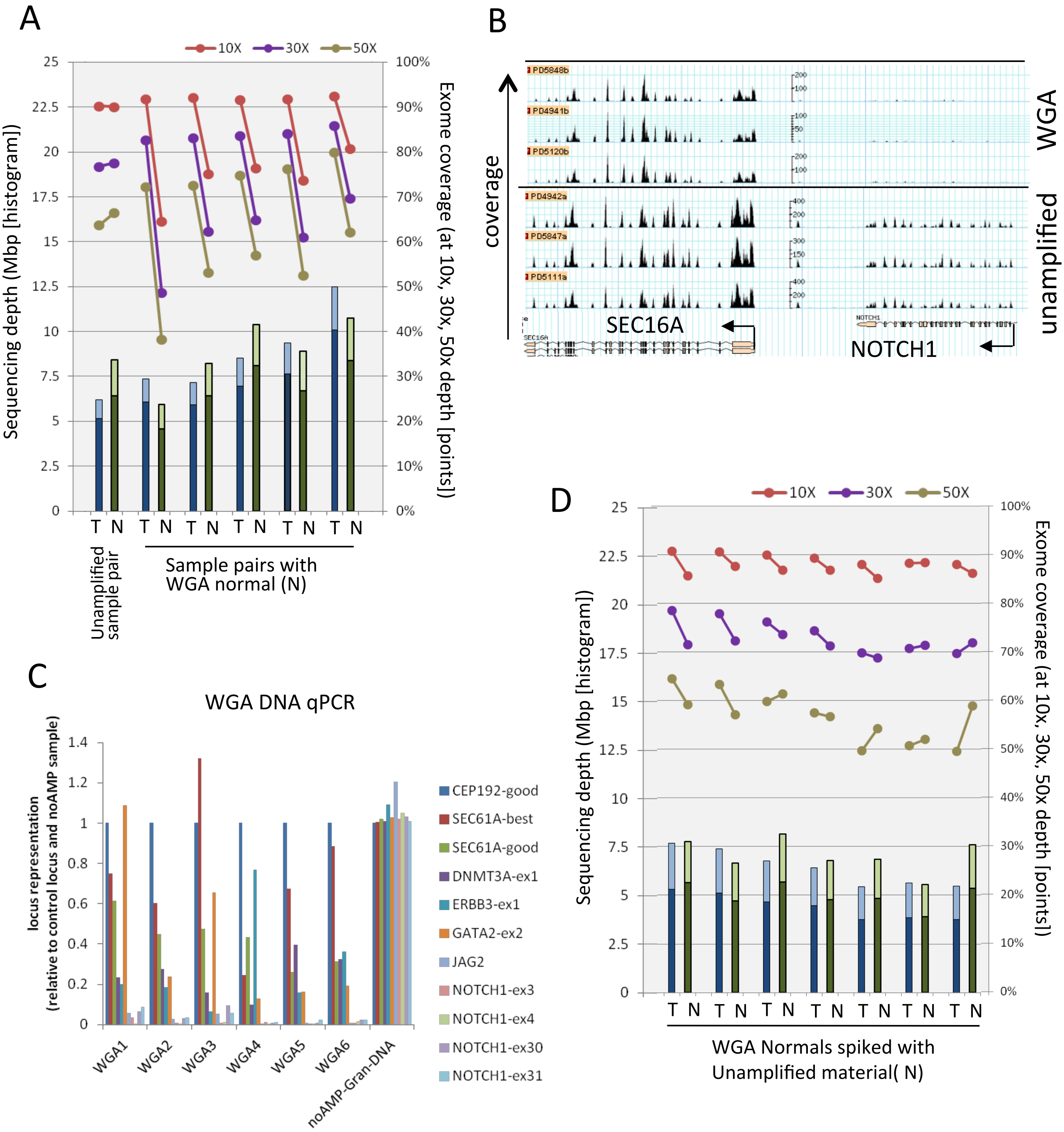

Figure S2 Whole genome amplification (WGA) of DNA prior to exome sequencing consistently results in loss of coverage at specific genomic regions which can be rescued by ‘spiking’ with unamplified DNA. (A) Summary of exome sequencing depth (Mbp, mapped reads) and percentage exome coverage (at 10x, 30x and 50x, as indicated) for one unamplified sample pair and five representative sample pairs with WGA normal samples. Note the consistent loss of coverage in WGA normal samples, which is unrelated to depth of sequencing. (B) Genome browser view of exome sequencing depth for whole genome amplified constitutional DNA samples, compared to unamplified tumour DNAs shows specific regions of lost coverage. (C) Realtime qPCR quantification of loci commonly under-represented in WGA exomes. WGA was performed in 6 independent experiements on the same sample (WGA1-6). Subsequent qPCR highlights regions which are lost stochastically (eg. *GATA2*) and regions always underrepresented by WGA (eg. *JAG2*). (D) Exome sequencing depth and percentage exome coverage for 7 sample pairs with 5-30% unamplified DNA added to normal into WGA normal samples, showing no loss of coverage.

Figure S3

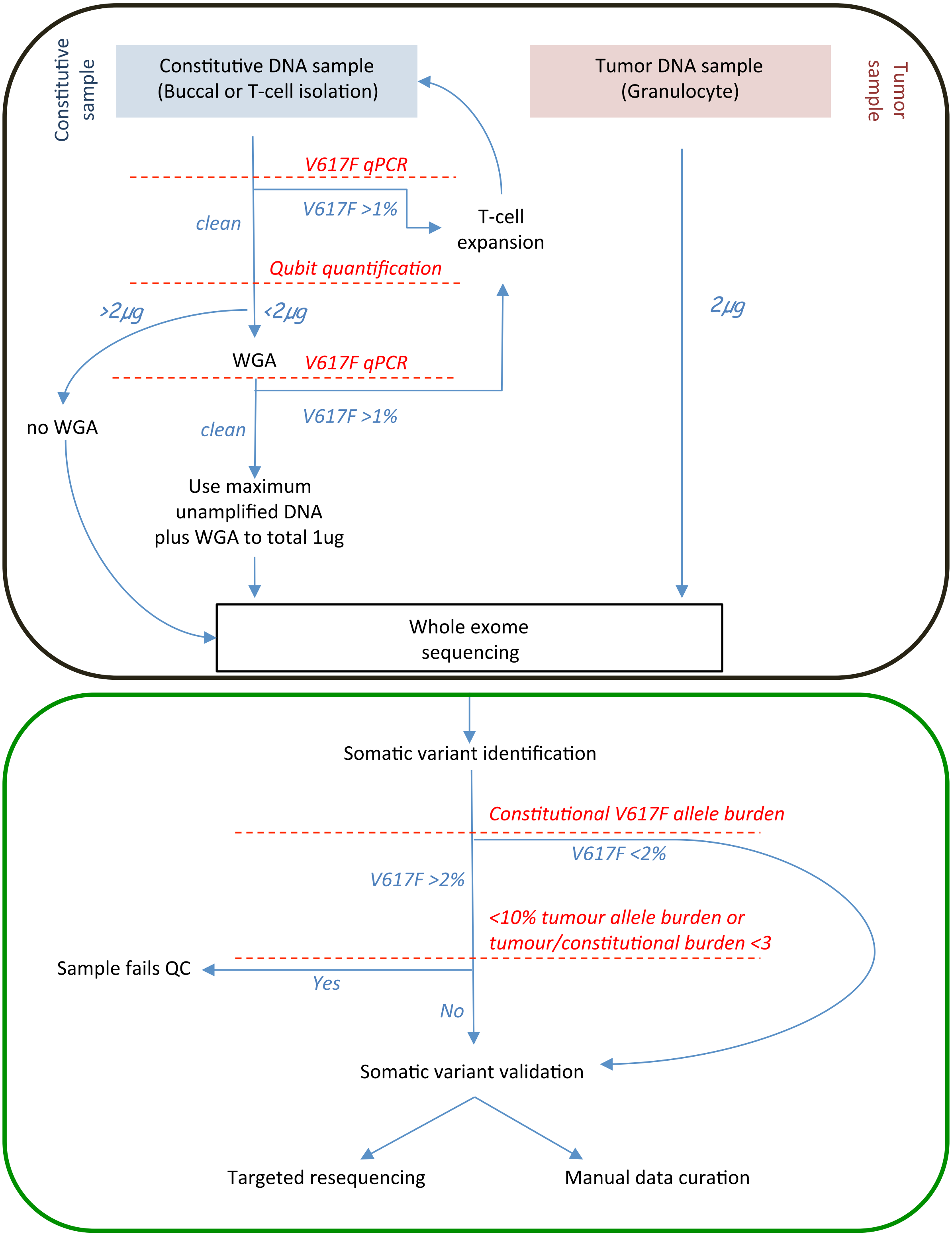

Figure S3 Quality assurance and bioinformatic workflow for all MPN samples prior to submission for exome sequencing (black panel) and following variant identification (green panel) to enable compliation of a high confidence mutation catalogue of MPNs. Tumor DNAs (2ug) were prepared for exome sequencing. For matched constitutional material, buccal, T-cell or cultured T-cell DNAs were prepared. *V617F* allele burden was measured by qPCR at various stages and if levels were >1%, cultured T-cells were prepared. Where constitutional material was limited, whole-genome amplication (WGA) was performed. The maximum amount of unamplified DNA was used and supplemented with WGA DNA to total 2ug. Following exome sequencing and variant identification, further bioinformatic criteria (green panel) as detailed above were applied to ensure only samples where somatic variants could be detected robustly were taken forward for analysis. All variants were validated by either custom bait capture and resequencing, and/or manual curation of the variants.

Figure S4

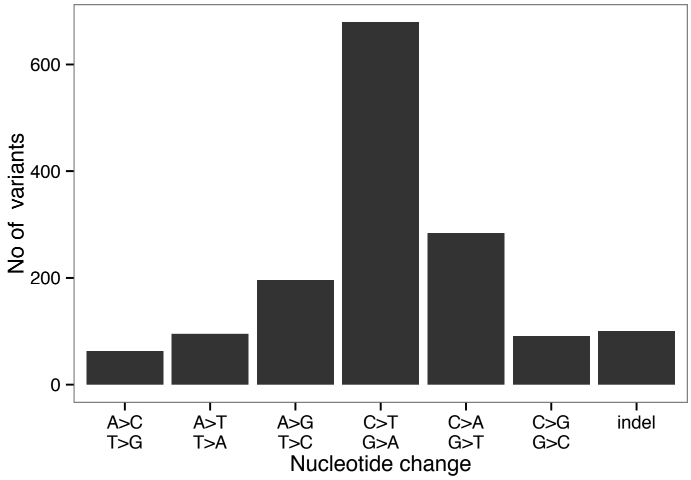

Figure S4 Somatic mutation spectrum in 151 MPN samples. Barchart showing the nucleotide changes of somatic variants identified by exome sequencing. Indel = insertions or deletions.

Figure S5

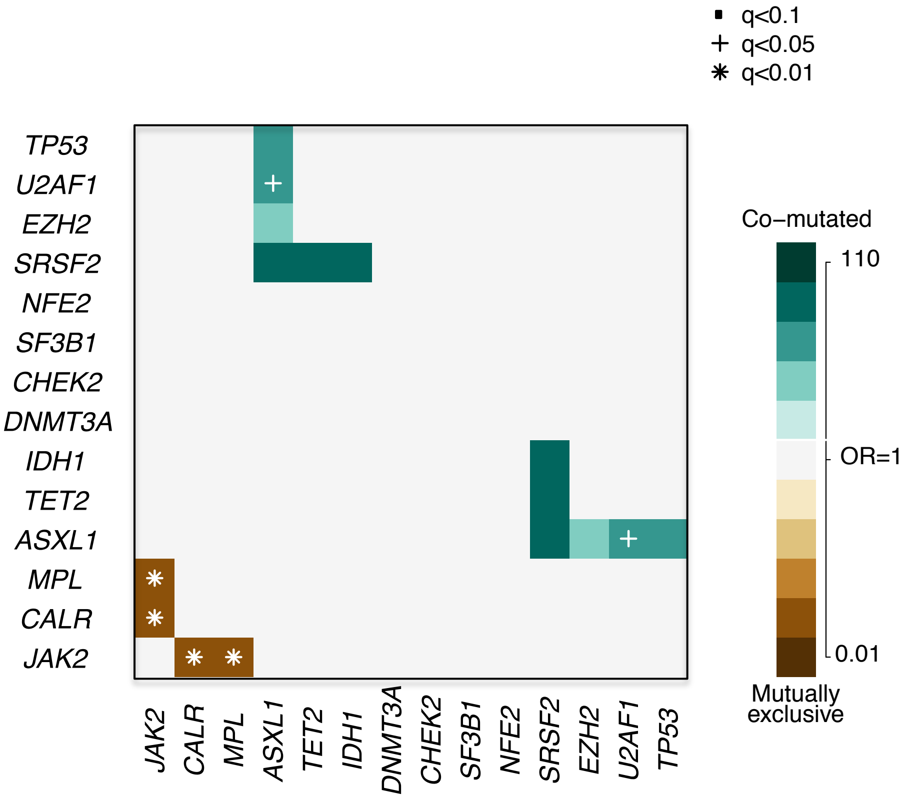

Figure S5 Pairwise associations among MPN cancer genes. Only associations with a p value < 0.05 are shown, and those that have a false discovery rate of less than 0.1 are marked by a white character. Associations are colored by odds ratio. Brown colors depict mutually exclusive gene pairs (one or other mutated, but rarely both together) and blue-green colors gene pairs that are co-mutated more than expected by chance.

Figure S6

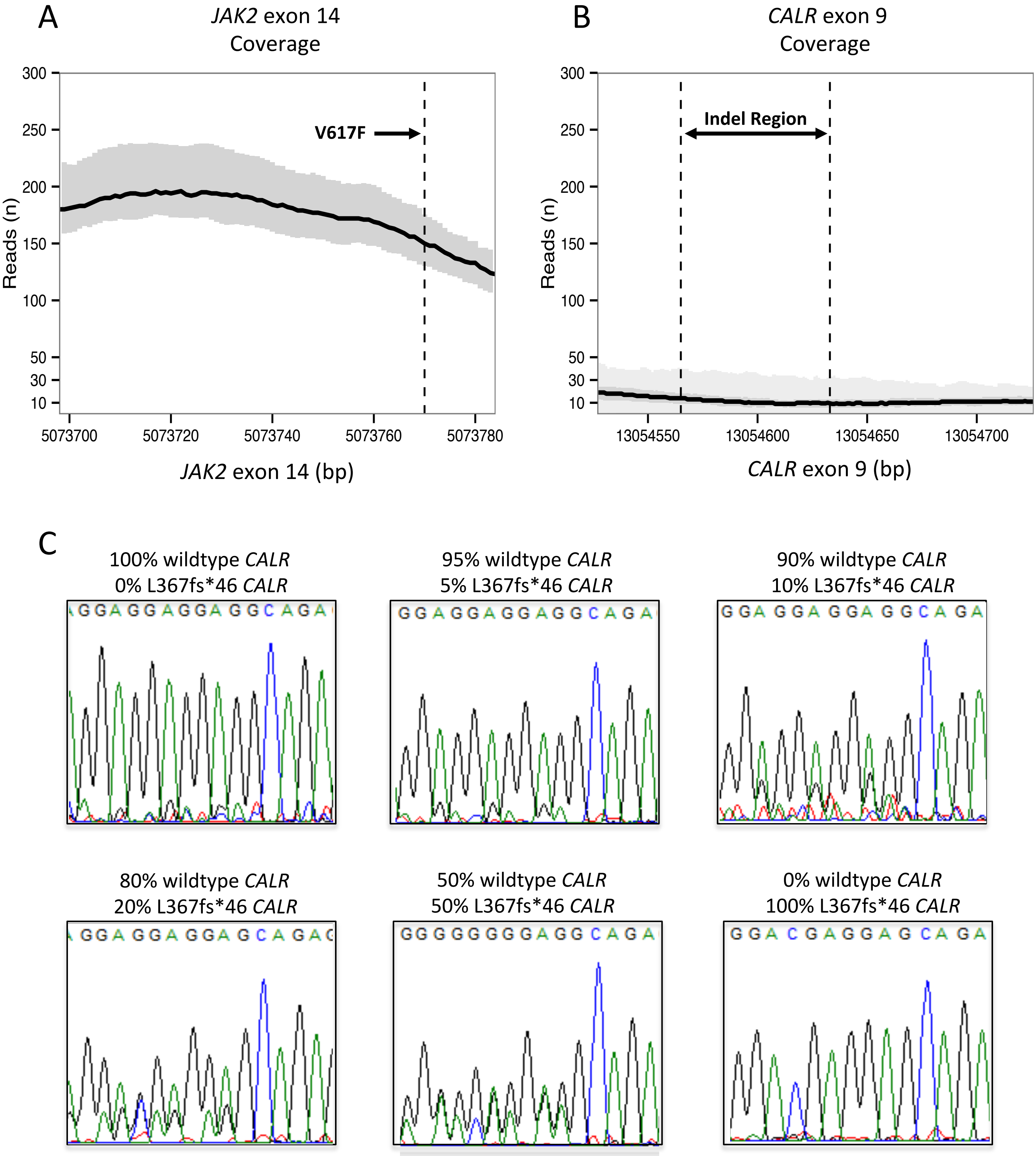

Figure S6 Identification of somatic variants in *CALR*. (a) Exome sequencing coverage across *JAK2*^{V617F} in 151 MPN constitutional samples showing read depth at genomic positions across *JAK2* exon 14. Black line shows median coverage, shaded region shows the interquartile range. Position of *JAK2*^{V617F} is indicated. (b) Exome sequencing coverage across *CALR* exon 9 in 151 MPN constitutional samples, showing low coverage at this locus. (c) Titration analysis to determine the sensitivity of *CALR* mutation screening using PCR and capillary sequencing. Indicated proportions of *CALR* exon 9 plasmids (wildtype and mutant (L367fs*46), were mixed and genotyped. Traces of capillary sequencing show that the mixed sequence (wildtype and mutant) can be seen clearly down to levels of 10% mutant allele fraction.

Figure S7

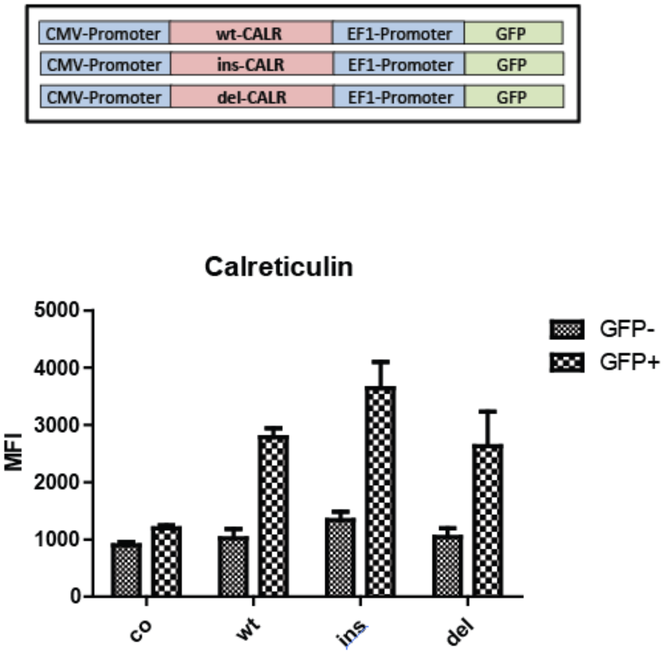

Figure S7. Calreticulin cell surface levels in 32D hematopoietic cells. CALR expression constructs are shown above (co, empty vector; wt, wildtype; ins, CALR K385fs*47; del, CALR L347fs*46). Graph shows flow cytometry analysis for CALR cell surface expression in 32D cells transiently transfected with CALR variants (MFI, mean fluorescent intensity; GFP-, untransfected cells; GFP+, transfected cells). 48h after transfection GFP- and GFP+ cells were detected in each sample. GFP- cells from transfected samples had cell surface CALR levels at similar levels to the empty vector control. Transient expression of CALR variants caused an increase in cell surface CALR with no significant differences between wildtype and mutant CALR proteins. Data shows mean + SEMs of three independent experiments.

Figure S8

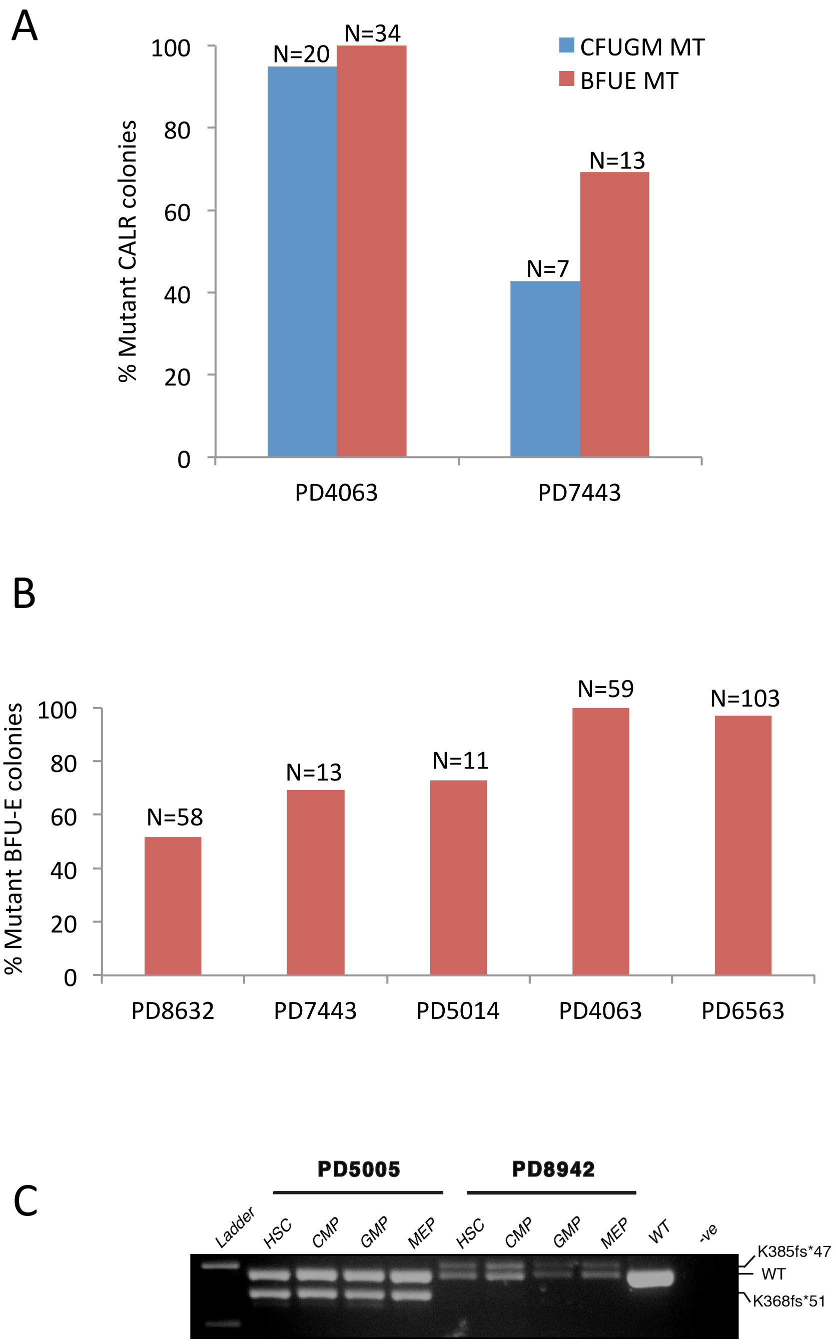

Figure S8 Exome sequencing of granulocytes and hematopoietic colonies *CALR* mutations in multiple lineages. (A) Erythroid (BFU-E) and granulocytic (CFU-GM) colonies were grown from peripheral blood MNCs of two CALR mutated patients and discrete clones were plucked at day-10. Data show *CALR* mutated CFU-GM and BFU-Es in both patients. (B) For 5 *CALR* mutated patients identified by exome sequencing of granulocytes, BFU-E clones were grown and genotyped for *CALR*. Together these data show presence of the mutation in granulocyte and erythroid compartments. (C) PCR analysis of *CALR*-mutated hematopoietic colonies grown from the indicated flow sorted hematopoietic stem and progenitor cell fractions obtained from two patients with *CALR* mutations; PD5005 with a 34bp deletion (K368fs*51); PD8942 with a 5bp insertion (K385fs*47). Colonies from each cell fraction were pooled and genotyped for *CALR* exon 9. HSC, hematopoietic stem cell; CMP, common myeloid progenitor; GMP, granulocytic-macrophage progenitor; MEP, megakaryocyte-erythroid progenitor; WT, wildtype; -ve, no template control

Figure S9

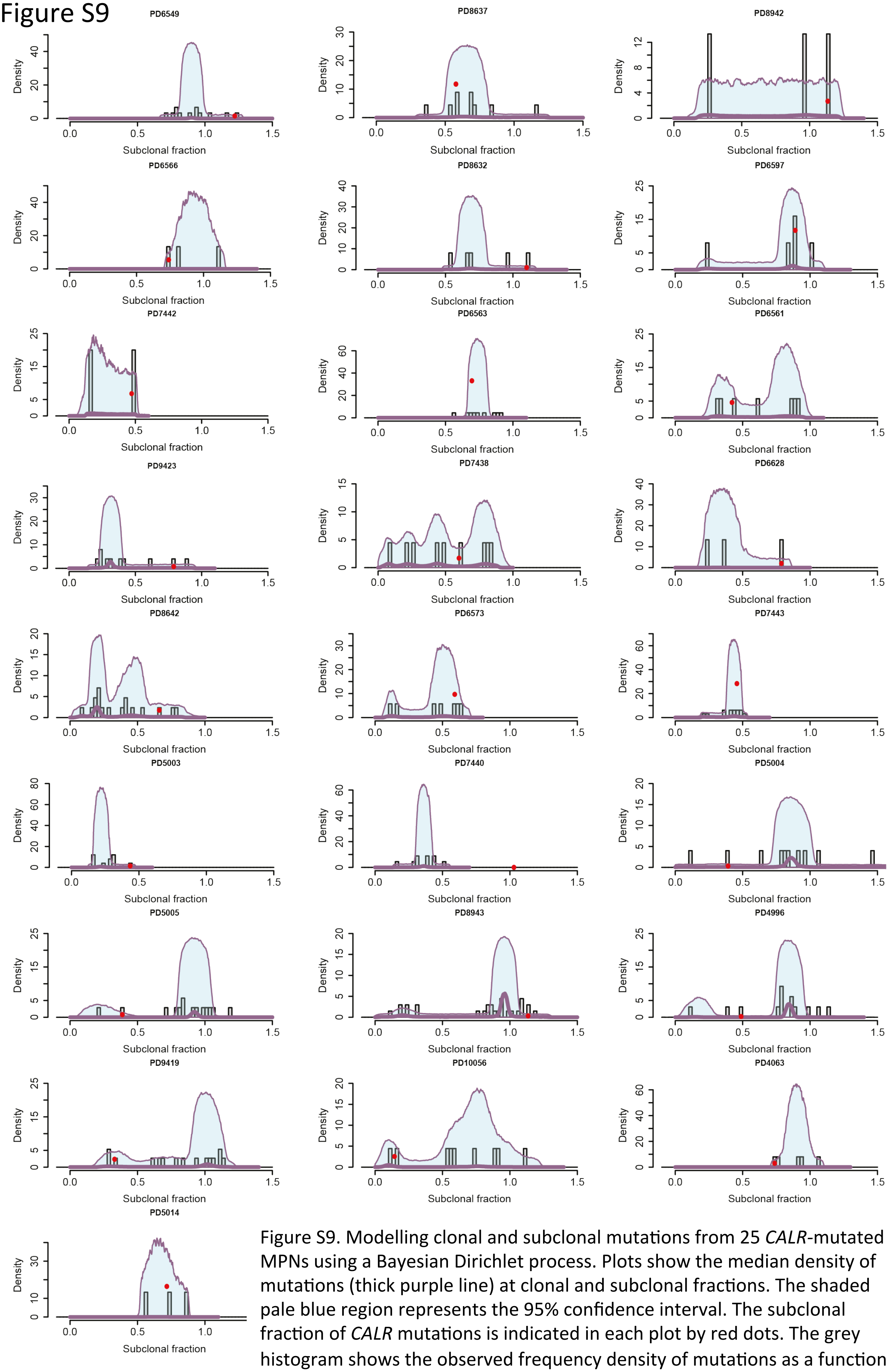

Figure S9. Modelling clonal and subclonal mutations from 25 *CALR*-mutated MPNs using a Bayesian Dirichlet process. Plots show the median density of mutations (thick purple line) at clonal and subclonal fractions. The shaded pale blue region represents the 95% confidence interval. The subclonal fraction of *CALR* mutations is indicated in each plot by red dots. The grey histogram shows the observed frequency density of mutations as a function of the fraction of cells bearing the mutation.

Table S1. Myeloproliferative neoplasm subtype and mutation status of samples for exome sequencing.

Myeloproliferative Neoplasm	*JAK2* mutated	*MPL* mutated	JAK2/*MPL* unmutated	Total
Polycythemia Vera (PV)	48	0	0	48
Essential Thombocythemia (ET)	35	5	22	62
Myelofibrosis (MF)	28	2	9	39
Primary MF	20	1	5	27
Post ET - MF	1	1	4	6
Post PV - MF	7	0	0	6
Myeloproliferative neoplasm_unclassifiable	2	0	0	2
Total	113	7	31	151

Data show number of patients. *JAK2* and *MPL* status obtained from source information was confirmed by exome sequencing.

Table S2 Somatic mutations identified by exome sequencing of 151 Myeloproliferative neoplasm samples

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1	PD10051	PMF	JAK2	9	5073770	G	T	179	0.6	114	95.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
2	PD10051	PMF	JAK2	5	49963910	A	C	102	0.0	74	52.7	PARP8	CCDS3954.1	c.97A>C	p.R33R	silent	M
3	PD10051	PMF	JAK2	7	156555827	T	C	89	0.0	73	49.3	LMBR1	CCDS5945.1	c.594A>G	p.S198S	silent	M
4	PD10051	PMF	JAK2	18	13826416	C	T	144	0.0	255	48.2	MC5R	CCDS11868.1	c.652C>T	p.R218W	missense	M
5	PD10051	PMF	JAK2	11	56058390	C	T	266	1.1	187	48.1	OR8H1	CCDS31526.1	c.149G>A	p.R50H	missense	M
6	PD10051	PMF	JAK2	4	186272630	G	T	188	0.0	144	46.5	SNX25	CCDS34116.1	c.1841G>T	p.S614I	missense	M
7	PD10051	PMF	JAK2	14	76118164	T	C	127	0.0	66	45.5	C14orf1	CCDS9845.1	c.293A>G	p.Y98C	missense	M
8	PD10051	PMF	JAK2	1	167095146	G	T	37	0.0	58	43.1	DUSP27	CCDS30932.1	c.778G>T	p.A260S	missense	M
9	PD10051	PMF	JAK2	7	124511095	T	A	98	0.0	87	41.4	POT1	CCDS5793.1	c.125A>T	p.D42V	missense	M
10	PD10051	PMF	JAK2	4	65275036	G	A	190	0.0	73	37.0	TECRL	CCDS33990.1	c.34C>T	p.R12C	missense	M
11	PD10051	PMF	JAK2	4	126373660	G	A	43	0.0	43	30.2	FAT4	CCDS3732.3	c.11489G>A	p.R3830H	missense	M
12	PD10051	PMF	JAK2	9	125282040	_	delTACTC	58	0.0	40	20.0	OR1J4	CCDS35122.1	c.621_625delTACTC	p.L209fs*22	frameshift	M
13	PD10051	PMF	JAK2	7	43917086	G	A	29	0.0	27	18.5	URGCP	CCDS47578.1	c.1976C>T	p.P659L	missense	M
14	PD10051	PMF	JAK2	18	29867420	G	C	58	0.0	75	12.0	FAM59A	CCDS11905.1	c.1140C>G	p.T380T	silent	M
15	PD10051	PMF	JAK2	8	21996292	C	G	33	0.0	42	9.5	REEP4	CCDS6024.1	c.568G>C	p.G190R	missense	M
16	PD10052	PPV-MF	JAK2	9	5073770	G	T	134	0.0	125	82.4	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
17	PD10052	PPV-MF	JAK2	19	39913981	T	A	20	0.0	20	50.0	PLEKHG2	CCDS33022.2	c.2287T>A	p.S763T	missense	M
18	PD10052	PPV-MF	JAK2	8	143763518	G	A	17	0.0	6	50.0	PSCA	CCDS47925.1	c.340G>A	p.G114S	missense	M
19	PD10052	PPV-MF	JAK2	16	23079452	C	T	105	1.0	83	48.2	USP31	CCDS10607.1	c.3974G>A	p.G1325D	missense	M
20	PD10052	PPV-MF	JAK2	17	5114200	T	G	81	0.0	69	47.8	C17orf87	CCDS42242.1	c.334A>C	p.K112Q	missense	M
21	PD10052	PPV-MF	JAK2	20	50715032	C	T	57	0.0	35	45.7	ZFP64	CCDS13439.1	c.858G>A	p.E286E	silent	M
22	PD10052	PPV-MF	JAK2	17	8395641	T	A	118	0.0	83	38.6	MYH10	CCDS11144.1	c.4552A>T	p.S1518C	missense	M
23	PD10052	PPV-MF	JAK2	16	56686927	C	G	234	0.0	132	37.1	MT1B	CCDS10765.1	c.136C>G	p.Q46E	missense	M
24	PD10052	PPV-MF	JAK2	4	165981237	A	T	195	1.0	192	37.0	TRIM75	CCDS47160.1	c.938A>T	p.K313I	missense	M
25	PD10052	PPV-MF	JAK2	6	33631514	G	A	20	0.0	19	36.8	ITPR3	CCDS4783.1	c.1006-1G>A	p.?	essential splice	M
26	PD10052	PPV-MF	JAK2	13	99582427	_	insA	111	0.0	74	27.0	DOCK9	CCDS45062.1	c.327_328insT	p.T110fs*8	frameshift	M
27	PD10052	PPV-MF	JAK2	2	97285424	_	delGC	33	0.0	31	25.8	KIAA1310	CCDS46361.1	c.461_462delGC	p.R154fs*8	frameshift	M
28	PD10053	PET-MF	JAK2	2	25457242	C	T	45	0.0	51	56.9	DNMT3A	CCDS33157.1	c.2645G>A	p.R882H	missense	T
29	PD10053	PET-MF	JAK2	17	57139957	C	G	72	0.0	50	52.0	TRIM37	CCDS32694.1	c.913G>C	p.G305R	missense	M
30	PD10053	PET-MF	JAK2	6	96651060	G	A	164	0.0	107	50.5	FUT9	CCDS5033.1	c.29G>A	p.R10H	missense	M
31	PD10053	PET-MF	JAK2	2	198266834	T	C	91	1.1	83	49.4	SF3B1	CCDS33356.1	c.2098A>G	p.K700E	missense	M
32	PD10053	PET-MF	JAK2	5	76932764	C	G	56	0.0	78	47.4	OTP	CCDS4039.1	c.329G>C	p.R110P	missense	M
33	PD10053	PET-MF	JAK2	14	64518414	T	C	55	0.0	58	46.6	SYNE2	CCDS9761.2	c.7783T>C	p.L2595L	silent	M
34	PD10053	PET-MF	JAK2	4	106155749	_	delC	91	0.0	43	46.5	TET2	CCDS47120.1	c.650delC	p.V218fs*32	frameshift	M
35	PD10053	PET-MF	JAK2	1	226564918	A	G	450	0.0	292	43.2	PARP1	CCDS1554.1	c.1832T>C	p.I611T	missense	M
36	PD10053	PET-MF	JAK2	9	5073770	G	T	146	0.0	102	43.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
37	PD10053	PET-MF	JAK2	14	73718405	A	G	28	0.0	72	41.7	PAPLN	CCDS32114.1	c.623A>G	p.N208S	missense	M
38	PD10053	PET-MF	JAK2	17	66267152	C	T	71	0.0	86	38.4	SLC16A6	CCDS11675.1	c.1149G>A	p.W383*	nonsense	M
39	PD10053	PET-MF	JAK2	12	40830429	A	T	140	0.0	100	37.0	MUC19	ENST00000425730	c.2039A>T	p.K680I	missense	M
40	PD10053	PET-MF	JAK2	1	181689358	C	T	71	0.0	88	11.4	CACNA1E	ENST00000367573	c.1768C>T	p.R590W	missense	M
41	PD10053	PET-MF	JAK2	11	128333408	T	C	69	1.5	49	10.2	ETS1	CCDS44767.1	c.1238A>G	p.D413G	missense	M
42	PD10053	PET-MF	JAK2	1	248097640	A	G	93	0.0	76	9.2	OR2AJ1	ENST00000318244	c.570A>G	p.A190A	silent	M
43	PD10053	PET-MF	JAK2	5	140589856	C	T	139	0.0	215	7.4	PCDHB12	CCDS4254.1	c.1377C>T	p.F459F	silent	M
44	PD10053	PET-MF	JAK2	5	149792295	G	C	134	0.0	193	6.7	CD74	CCDS47309.1	c.18C>G	p.S6R	missense	M
45	PD10054	ET	JAK2	9	5073770	G	T	163	0.6	130	65.4	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
46	PD10054	ET	JAK2	X	37534942	C	T	47	0.0	26	61.5	LANCL3	CCDS14240.1	Non-coding	r.1524c>u	UTR	M
47	PD10054	ET	JAK2	11	62285353	_	delT	132	0.0	160	36.9	AHNAK	CCDS31584.1	c.16536delA	p.K5512fs*16	frameshift	M
48	PD10054	ET	JAK2	17	18541950	T	C	76	0.0	93	35.5	TBC1D28	CCDS42273.1	c.263A>G	p.Y88C	missense	M
49	PD10054	ET	JAK2	12	119588938	C	T	137	0.0	126	34.1	SRRM4	CCDS44994.1	c.1193C>T	p.T398I	missense	M
50	PD10054	ET	JAK2	2	228148555	G	A	67	0.0	77	33.8	COL4A3	CCDS42829.1	c.2729G>A	p.G910D	missense	M

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
51	PD10054	ET	JAK2	2	232458152	G	A	40	0.0	51	33.3	C2orf57	CCDS2487.1	c.490G>A	p.A164T	missense	M
52	PD10054	ET	JAK2	9	88967850	_	delA	133	0.0	159	28.3	ZCCHC6	CCDS35057.1	c.265delT	p.W89fs*2	frameshift	M
53	PD10054	ET	JAK2	17	79991385	C	T	19	0.0	35	25.7	RAC3	CCDS11798.1	c.358C>T	p.R120C	missense	M
54	PD10054	ET	JAK2	10	37508043	G	A	252	0.0	140	24.3	ANKRD30A	CCDS7193.1	c.3235G>A	p.D1079N	missense	M
55	PD10054	ET	JAK2	12	85674055	_	insAGA	66	0.0	63	23.8	ALX1	CCDS9028.1	c.16_17insAGA	p.K7_F8insK	inframe	M
56	PD10054	ET	JAK2	1	38339763	T	C	59	1.7	51	23.5	INPP5B	CCDS41306.1	c.1853A>G	p.Q618R	missense	M
57	PD10054	ET	JAK2	8	101011507	C	T	47	0.0	78	23.1	RGS22	CCDS43758.1	c.2932G>A	p.A978T	missense	M
58	PD10054	ET	JAK2	11	49032223	C	T	35	0.0	45	20.0	ENSG00000205044	ENST00000341343	c.459G>A	p.A153A	silent	M
59	PD10054	ET	JAK2	15	65962151	C	T	174	0.0	190	19.5	DENND4A	CCDS45285.1	c.4621G>A	p.A1541T	missense	M
60	PD10055	PPV-MF	JAK2	9	5073770	G	T	126	0.0	110	55.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
61	PD10055	PPV-MF	JAK2	6	65612122	C	T	58	0.0	54	50.0	EYS	CCDS47445.1	c.2739-9G>A	p.?	splice	M
62	PD10055	PPV-MF	JAK2	X	133551230	C	A	52	0.0	57	47.4	PHF6	CCDS14639.1	c.866C>A	p.T289N	missense	M
63	PD10055	PPV-MF	JAK2	4	106196705	C	T	126	2.4	196	44.9	TET2	CCDS47120.1	c.5038C>T	p.Q1680*	nonsense	M
64	PD10055	PPV-MF	JAK2	X	70514084	G	A	73	2.7	43	44.2	NONO	CCDS14410.1	c.356G>A	p.R119Q	missense	M
65	PD10055	PPV-MF	JAK2	14	95590575	A	C	46	0.0	66	42.4	DICER1	CCDS9931.1	c.1334T>G	p.I445S	missense	M
66	PD10055	PPV-MF	JAK2	4	106164775	G	T	11	0.0	10	40.0	TET2	CCDS47120.1	c.3643G>T	p.E1215*	nonsense	M
67	PD10055	PPV-MF	JAK2	19	10305546	C	A	17	0.0	10	40.0	DNMT1	CCDS45958.1	c.30G>T	p.V10V	silent	M
68	PD10055	PPV-MF	JAK2	1	158622296	_	insT	146	0.0	106	39.6	SPTA1	CCDS41423.1	c.3335_3336insA	p.D1112fs*10	frameshift	M
69	PD10055	PPV-MF	JAK2	6	63921761	C	T	63	0.0	51	39.2	FKBP1C	ENST00000370659	c.300C>T	p.F100F	silent	M
70	PD10055	PPV-MF	JAK2	8	3245147	C	T	27	0.0	45	37.8	CSMD1	ENST00000318252	c.1838G>A	p.R613H	missense	M
71	PD10055	PPV-MF	JAK2	1	115256535	G	T	102	2.0	112	33.0	NRAS	CCDS877.1	c.176C>A	p.A59D	missense	M
72	PD10056	PET-MF	unmutated	2	29449779	C	G	67	0.0	84	56.0	ALK	CCDS33172.1	c.3067+9G>C	p.?	splice	M
73	PD10056	PET-MF	unmutated	21	45994307	_	delC	122	0.0	138	45.7	KRTAP10-4	CCDS42957.1	c.672delC	p.Q225fs*102	frameshift	M
74	PD10056	PET-MF	unmutated	X	32862919	C	T	45	0.0	40	45.0	DMD	CCDS14233.1	c.245G>A	p.R82Q	missense	M
75	PD10056	PET-MF	unmutated	1	32201977	C	T	160	0.0	95	36.8	BAI2	CCDS346.2	c.3145G>A	p.V1049M	missense	M
76	PD10056	PET-MF	unmutated	16	66434681	G	A	92	0.0	90	28.9	CDH5	CCDS10804.1	c.1599G>A	p.T533T	silent	M
77	PD10056	PET-MF	unmutated	21	36252849	C	A	63	0.0	49	28.6	RUNX1	CCDS13639.1	c.508+5G>T	p.?	essential splice	M
78	PD10056	PET-MF	unmutated	12	69042539	G	A	145	0.0	91	27.5	RAP1B	CCDS8984.1	c.35G>A	p.G12E	missense	M
79	PD10056	PET-MF	unmutated	3	164758870	C	A	182	0.6	129	5.4	SI	CCDS3196.1	c.2017G>T	p.A673S	missense	M
80	PD10056	PET-MF	unmutated	19	13054565	_	delGCAGA<42>AGGAG	13	15.4	13	7.7	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M C
81	PD10057	PMF	JAK2	9	5073770	G	T	155	0.0	125	25.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
82	PD10057	PMF	JAK2	1	236882190	A	T	40	0.0	52	21.2	ACTN2	CCDS1613.1	c.242-4A>T	p.?	splice	M
83	PD10057	PMF	JAK2	5	170834699	A	C	62	0.0	49	20.4	NPM1	CCDS4376.1	c.772-5A>C	p.?	splice	M
84	PD10058	PMF	JAK2	9	5073770	G	T	116	0.0	111	97.3	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
85	PD10058	PMF	JAK2	7	30963734	C	A	12	0.0	11	54.6	AQP1	CCDS5431.1	Non-coding	r.1355c>a	UTR	M
86	PD10058	PMF	JAK2	1	207263650	C	T	39	0.0	36	52.8	C4BPB	CCDS1476.1	c.59-3C>T	p.?	splice	M
87	PD10058	PMF	JAK2	8	41166638	_	delGCT	6	0.0	9	33.3	SFRP1	CCDS34886.1	c.39_41delAGC	p.A14delA	inframe	M
88	PD10058	PMF	JAK2	12	45410053	G	A	85	0.0	77	27.3	DBX2	CCDS31781.1	Non-coding	r.1208c>u	UTR	M
89	PD10058	PMF	JAK2	19	57890074	T	C	141	0.7	76	26.3	ZNF547	CCDS33131.1	Non-coding	r.1923u>c	UTR	M
90	PD10058	PMF	JAK2	9	79634805	G	T	30	0.0	44	22.7	FOXB2	CCDS35045.1	c.235G>T	p.D79Y	missense	M
91	PD10058	PMF	JAK2	17	21825984	G	A	50	0.0	32	21.9	FAM27L	ENST00000426869	c.4G>A	p.V2I	missense	M
92	PD10058	PMF	JAK2	17	40874827	A	T	49	0.0	49	18.4	EZH1	CCDS32659.1	c.473T>A	p.V158D	missense	M
93	PD10058	PMF	JAK2	2	238285995	G	C	54	0.0	56	17.9	COL6A3	CCDS33412.1	c.2498-8C>G	p.?	splice	M
94	PD10058	PMF	JAK2	13	23930028	A	T	60	0.0	59	17.0	SACS	CCDS9300.2	c.723T>A	p.S241R	missense	M
95	PD10058	PMF	JAK2	4	104032115	C	T	177	0.0	142	16.9	CENPE	CCDS34042.1	c.7594G>A	p.G2532S	missense	M
96	PD10058	PMF	JAK2	4	106158463	_	insCT	143	0.0	114	16.7	TET2	CCDS47120.1	c.3364_3365insCT	p.P1123fs*15	frameshift	M
97	PD10058	PMF	JAK2	1	17314721	C	T	50	0.0	76	15.8	ATP13A2	CCDS175.1	c.2771G>A	p.R924H	missense	M
98	PD10058	PMF	JAK2	17	29231299	T	C	77	0.0	59	15.3	C17orf42	CCDS42291.1	c.280A>G	p.R94G	missense	M
99	PD10058	PMF	JAK2	1	10412688	G	A	123	0.0	75	14.7	KIF1B	CCDS111.1	c.3812-1G>A	p.?	essential splice	M
100	PD10058	PMF	JAK2	10	47087574	A	G	65	0.0	60	8.3	PPYR1	CCDS31193.1	c.791A>G	p.N264S	missense	M
101	PD10059	PMF	JAK2	19	45649503	_	insGTAA	1	0.0	6	66.7	LRRC68	ENST00000422370	c.770_771insGTAA	p.259fs*50	frameshift	M
102	PD10059	PMF	JAK2	15	52476780	G	A	62	0.0	69	56.5	GNB5	CCDS10149.1	c.94C>T	p.Q32*	nonsense	M

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
103	PD10059	PMF	JAK2	2	18745146	A	C	80	0.0	84	53.6	NT5C1B	CCDS33150.1	c.1749T>G	p.F583L	missense	M
104	PD10059	PMF	JAK2	9	71679928	A	G	44	2.3	56	51.8	FXN	CCDS6626.1	c.459A>G	p.Q153Q	silent	M
105	PD10059	PMF	JAK2	5	127674688	T	C	82	1.2	73	49.3	FBN2	CCDS34222.1	c.3409A>G	p.S1137G	missense	M
106	PD10059	PMF	JAK2	8	104897565	A	G	71	1.4	45	46.7	RIMS2	ENST00000436393	c.72A>G	p.R24R	silent	M
107	PD10059	PMF	JAK2	3	182756815	T	C	49	0.0	35	45.7	MCCC1	CCDS3241.1	c.1376A>G	p.N459S	missense	M
108	PD10059	PMF	JAK2	19	41095087	_	delGAGGA<16>GGAGG	2	0.0	9	44.4	SHKBP1	CCDS12560.1	c.1589+3_1589+28del26	p.?	frameshift	M
109	PD10059	PMF	JAK2	7	98805086	G	A	162	0.0	108	42.6	KPNA7	CCDS47651.1	c.4C>T	p.P2S	missense	M
110	PD10059	PMF	JAK2	7	45717813	C	T	134	0.0	127	42.5	ADCY1	CCDS34631.1	c.1849C>T	p.L617F	missense	M
111	PD10059	PMF	JAK2	12	111779646	C	T	68	0.0	52	42.3	CUX2	CCDS41837.1	c.3448C>T	p.R1150C	missense	M
112	PD10059	PMF	JAK2	9	114180255	C	T	56	0.0	59	39.0	KIAA0368	CCDS48006.1	c.2125G>A	p.V709I	missense	M
113	PD10059	PMF	JAK2	12	114261086	G	A	39	0.0	36	36.1	RBM19	CCDS9172.1	c.2826C>T	p.I942I	silent	M
114	PD10059	PMF	JAK2	22	29924417	T	C	107	0.0	185	35.1	THOC5	CCDS13859.1	c.964A>G	p.T322A	missense	M
115	PD10059	PMF	JAK2	10	106974205	G	A	147	0.0	81	34.6	SORCS3	CCDS7558.1	c.2381G>A	p.R794Q	missense	M
116	PD10059	PMF	JAK2	21	44524456	G	T	40	0.0	50	34.0	U2AF1	CCDS13694.1	c.101C>A	p.S34Y	missense	M
117	PD10059	PMF	JAK2	9	5073770	G	T	135	0.0	96	33.3	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
118	PD10059	PMF	JAK2	1	27215489	A	G	180	0.0	271	5.2	GPN2	CCDS289.1	c.539T>C	p.M180T	missense	M
119	PD10060	PMF	JAK2	9	5073770	G	T	146	0.0	185	59.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
120	PD10060	PMF	JAK2	5	22078546	C	T	41	0.0	50	48.0	CDH12	CCDS3890.1	c.231+9G>A	p.?	splice	M
121	PD10060	PMF	JAK2	1	46497255	C	T	33	0.0	20	45.0	MAST2	CCDS41326.1	c.3185C>T	p.S1062L	missense	M
122	PD10060	PMF	JAK2	4	185646105	C	A	64	0.0	49	42.9	MLF1IP	CCDS3838.1	c.320+6G>T	p.?	splice	M
123	PD10060	PMF	JAK2	11	17448654	G	T	39	0.0	42	40.5	ABCC8	CCDS31437.1	c.2164C>A	p.L722I	missense	M
124	PD10060	PMF	JAK2	16	70531195	C	T	86	0.0	106	33.0	COG4	CCDS10892.2	c.1410G>A	p.L470L	silent	M
125	PD10060	PMF	JAK2	2	110881429	G	T	35	2.9	48	29.2	NPHP1	CCDS2086.1	c.2141C>A	p.P714H	missense	M
126	PD10060	PMF	JAK2	10	73053163	C	T	45	0.0	44	27.3	UNC5B	CCDS7309.1	c.1774C>T	p.P592S	missense	M
127	PD10060	PMF	JAK2	11	61094359	G	A	193	0.0	119	13.5	DDB1	CCDS31576.1	c.556C>T	p.Q186*	nonsense	M
128	PD10061	PMF	JAK2	X	69498364	G	A	22	0.0	18	77.8	ARR3	CCDS14399.1	c.778G>A	p.A260T	missense	M
129	PD10061	PMF	JAK2	9	5073770	G	T	147	1.4	115	67.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
130	PD10061	PMF	JAK2	4	106164913	C	T	36	2.8	19	63.2	TET2	CCDS47120.1	c.3781C>T	p.R1261C	missense	M
131	PD10061	PMF	JAK2	22	24177101	C	G	21	0.0	24	62.5	DERL3	CCDS46672.1	c.633G>C	p.K211N	missense	M
132	PD10061	PMF	JAK2	3	156763572	G	A	76	0.0	40	47.5	LEKR1	CCDS33883.1	Non-coding	r.2535g>a	UTR	M
133	PD10061	PMF	JAK2	4	186065924	C	G	57	0.0	69	44.9	SLC25A4	CCDS34114.1	c.118C>G	p.H40D	missense	M
134	PD10061	PMF	JAK2	6	110763614	G	A	84	0.0	74	43.2	SLC22A16	CCDS5084.1	c.1016C>T	p.T339I	missense	M
135	PD10061	PMF	JAK2	8	17601285	G	A	67	0.0	63	41.3	MTUS1	CCDS43717.1	c.2115C>T	p.T705T	silent	M
136	PD10061	PMF	JAK2	19	33288793	G	C	60	0.0	44	40.9	TDRD12	ENST00000444215	c.1626G>C	p.R542S	missense	M
137	PD10061	PMF	JAK2	20	31021439	G	T	167	0.6	173	38.7	ASXL1	CCDS13201.1	c.1438G>T	p.E480*	nonsense	M
138	PD10061	PMF	JAK2	5	140531475	G	A	78	0.0	71	38.0	PCDHB6	CCDS4248.1	c.1637G>A	p.R546H	missense	M
139	PD10061	PMF	JAK2	19	50550234	A	T	69	4.4	56	35.7	ZNF473	CCDS33077.1	c.2534A>T	p.Q845L	missense	M
140	PD10061	PMF	JAK2	10	114181736	C	T	74	0.0	77	33.8	ACSL5	CCDS7572.1	c.1587C>T	p.Y529Y	silent	M
141	PD10061	PMF	JAK2	7	123594038	C	A	131	1.5	96	32.3	SPAM1	CCDS5790.1	c.414C>A	p.D138E	missense	M
142	PD10061	PMF	JAK2	17	74732959	G	A	16	0.0	21	28.6	SFRS2	CCDS11749.1	c.284C>T	p.P95L	missense	M
143	PD10061	PMF	JAK2	2	209104604	G	A	77	0.0	64	14.1	IDH1	CCDS2381.1	c.974C>T	p.T325M	missense	M
144	PD10061	PMF	JAK2	4	106180784	G	A	130	0.0	102	9.8	TET2	CCDS47120.1	c.3812G>A	p.C1271Y	missense	M
145	PD10062	PMF	JAK2	9	5073770	G	T	151	1.3	146	38.4	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
146	PD10062	PMF	JAK2	17	27425877	T	C	27	0.0	45	20.0	MYO18A	CCDS45642.1	c.3747A>G	p.V1249V	silent	M
147	PD10062	PMF	JAK2	19	33202719	G	A	56	0.0	63	19.1	NUDT19	CCDS42543.1	c.984G>A	p.K328K	silent	M
148	PD10062	PMF	JAK2	20	37214676	A	C	54	0.0	57	17.5	ADIG	ENST00000416116	c.74A>C	p.S25S	silent	M
149	PD10062	PMF	JAK2	2	180810389	T	C	277	0.0	229	17.5	CWC22	CCDS46465.1	c.2194A>G	p.K732E	missense	M
150	PD10062	PMF	JAK2	1	175372505	C	T	43	0.0	36	13.9	TNR	CCDS1318.1	c.747G>A	p.E249E	silent	M
151	PD10062	PMF	JAK2	12	54686430	G	A	47	0.0	54	13.0	NFE2	CCDS8876.1	c.850C>T	p.R284C	missense	M
152	PD10062	PMF	JAK2	1	43650825	T	C	142	0.7	119	8.4	WDR65	CCDS479.1	c.767T>C	p.I256T	missense	M
153	PD10062	PMF	JAK2	10	124594414	A	T	64	0.0	102	6.9	CUZD1	CCDS7631.1	c.1190T>A	p.M397K	missense	M
154	PD10063	PMF	JAK2	6	45390488	_	delGCGGC<8>CTGCG	6	0.0	18	22.2	RUNX2	CCDS43467.1	c.421_438del18	p.A141_A146delAAAAAA	frameshift	M

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
155	PD10063	PMF	JAK2	16	30721267	_	delA	71	0.0	44	20.5	SRCAP	CCDS10689.2	c.952delA	p.R318fs*62	frameshift	M
156	PD10063	PMF	JAK2	9	5073770	G	T	133	0.0	116	17.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
157	PD10063	PMF	JAK2	9	130911771	C	G	27	0.0	60	16.7	LCN2	CCDS6892.1	Non-coding	r.204c>g	UTR	M
158	PD10063	PMF	JAK2	14	58718925	A	T	87	0.0	61	16.4	PSMA3	CCDS9731.1	c.193A>T	p.K65*	nonsense	M
159	PD10063	PMF	JAK2	13	52593243	G	A	62	0.0	52	15.4	UTP14C	CCDS31977.1	c.239G>A	p.R80K	missense	M
160	PD10063	PMF	JAK2	12	3728528	C	T	25	0.0	66	15.2	EFCAB4B	CCDS44803.1	c.1943G>A	p.G648E	missense	M
161	PD10063	PMF	JAK2	9	85615130	C	T	86	1.2	113	15.0	RASEF	CCDS6662.1	c.1677G>A	p.M559I	missense	M
162	PD10063	PMF	JAK2	9	109688207	G	A	500	0.2	468	13.7	ZNF462	CCDS35096.1	c.2014G>A	p.V672I	missense	M
163	PD10063	PMF	JAK2	19	47856679	C	T	40	0.0	60	13.3	DHX34	CCDS12700.1	c.392C>T	p.A131V	missense	M
164	PD10063	PMF	JAK2	5	80508237	G	A	15	0.0	32	12.5	RASGRF2	CCDS4052.1	c.3209G>A	p.R1070H	missense	M
165	PD10063	PMF	JAK2	9	139358931	C	T	53	0.0	131	12.2	SEC16A	ENST00000313050	c.3853G>A	p.V1285M	missense	M
166	PD10063	PMF	JAK2	5	140249351	G	A	58	0.0	212	11.3	PCDHA11	CCDS47284.1	c.663G>A	p.E221E	silent	M
167	PD10063	PMF	JAK2	X	106793394	C	G	98	0.0	143	10.5	FRMPD3	ENST00000276185	c.609+8C>G	p.?	splice	M
168	PD10063	PMF	JAK2	4	139980293	T	C	146	0.0	204	10.3	ELF2	CCDS3744.1	c.1590A>G	p.K530K	silent	M
169	PD10063	PMF	JAK2	19	1399170	_	delA	18	0.0	78	10.3	GAMT	CCDS45897.1	c.416delT	p.L139fs*22	frameshift	M
170	PD10063	PMF	JAK2	X	130219590	C	T	421	0.0	429	9.6	ARHGAP36	CCDS14628.1	c.984C>T	p.A328A	silent	M
171	PD10063	PMF	JAK2	2	20838338	_	delCTGT	20	0.0	53	9.4	HS1BP3	CCDS1700.1	c.478_481delACAG	p.T160fs*26	frameshift	M
172	PD10063	PMF	JAK2	16	51174212	C	T	28	0.0	58	8.6	SALL1	CCDS10747.1	c.1921G>A	p.V641I	missense	M
173	PD10063	PMF	JAK2	X	151908806	G	A	278	0.0	204	7.8	CSAG1	CCDS14711.1	c.45G>A	p.G15G	silent	M
174	PD10063	PMF	JAK2	2	102626297	C	G	73	0.0	87	6.9	IL1R2	CCDS2054.1	c.332+9C>G	p.?	splice	M
175	PD4060	PPV-MF	JAK2	9	5073770	G	T	142	0.0	30	100.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
176	PD4060	PPV-MF	JAK2	8	8234772	C	T	19	0.0	27	59.3	ENSG00000182319	CCDS43706.1	c.1147G>A	p.V383M	missense	T
177	PD4060	PPV-MF	JAK2	2	98828414	C	A	96	0.0	66	50.0	VWA3B	CCDS42718.1	c.1759C>A	p.L587M	missense	T
178	PD4060	PPV-MF	JAK2	12	111885596	T	C	57	0.0	101	49.5	SH2B3	CCDS9153.1	c.1373T>C	p.L458P	missense	T
179	PD4060	PPV-MF	JAK2	1	52981650	A	T	90	0.0	97	49.5	ZCCHC11	CCDS30715.1	c.795T>A	p.D265E	missense	T
180	PD4060	PPV-MF	JAK2	4	106164860	A	G	25	0.0	64	48.4	TET2	CCDS47120.1	c.3728A>G	p.K1243R	missense	T C
181	PD4060	PPV-MF	JAK2	8	133895139	G	C	40	0.0	87	48.3	TG	CCDS34944.1	c.970G>C	p.D324H	missense	T
182	PD4060	PPV-MF	JAK2	7	5355647	C	G	37	0.0	25	48.0	TNRC18	CCDS47534.1	c.6802G>C	p.D2268H	missense	T
183	PD4060	PPV-MF	JAK2	19	44536240	T	A	213	0.0	225	46.7	ZNF222	CCDS46098.1	c.533T>A	p.L178*	nonsense	T
184	PD4060	PPV-MF	JAK2	5	94991892	G	A	113	0.9	137	44.5	RFESD	CCDS47248.1	c.512G>A	p.R171K	missense	T
185	PD4060	PPV-MF	JAK2	1	120462033	G	A	48	0.0	108	44.4	NOTCH2	CCDS908.1	c.5683C>T	p.R1895C	missense	T
186	PD4060	PPV-MF	JAK2	19	12246376	_	insC	143	0.0	81	38.3	ZNF20	CCDS45986.1	c.140_141insG	p.T49fs*8	frameshift	T
187	PD4060	PPV-MF	JAK2	4	84352921	C	T	76	0.0	55	27.3	HELQ	CCDS3603.1	c.2191G>A	p.V731I	missense	T
188	PD4060	PPV-MF	JAK2	10	38740351	G	A	16	0.0	23	17.4	ENSG00000215153	ENST00000399699	c.394G>A	p.V132I	missense	T
189	PD4060	PPV-MF	JAK2	9	123906255	C	T	83	0.0	58	10.3	CEP110	CCDS35118.1	c.2946C>T	p.A982A	silent	T
190	PD4060	PPV-MF	JAK2	3	124485118	C	T	74	0.0	110	10.0	ITGB5	CCDS3030.1	c.2092G>A	p.E698K	missense	T
191	PD4063	PET-MF	unmutated	5	150924251	C	T	282	0.0	197	53.3	FAT2	CCDS4317.1	c.6437G>A	p.R2146Q	missense	T
192	PD4063	PET-MF	unmutated	6	138638505	C	T	206	0.0	161	47.2	KIAA1244	CCDS5189.2	c.4463C>T	p.T1488M	missense	T
193	PD4063	PET-MF	unmutated	19	9720950	G	A	211	0.0	166	45.2	ZNF561	CCDS12216	c.1387C>T	p.R463C	missense	T
194	PD4063	PET-MF	unmutated	2	99993053	G	A	312	0.0	247	38.5	EIF5B	CCDS42721.1	c.1796G>A	p.R599Q	missense	T
195	PD4063	PET-MF	unmutated	19	13054565	_	delGCAGA<42>AGGAG	7	0.0	43	37.2	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M C
196	PD4177	PMF	JAK2	2	38298008	G	C	27	0.0	95	50.5	CYP1B1	CCDS1793.1	c.1489C>G	p.P497A	missense	M
197	PD4177	PMF	JAK2	17	46804413	G	A	40	2.5	36	50.0	HOXB13	CCDS11536.1	c.602-8C>T	p.?	splice	M
198	PD4177	PMF	JAK2	4	119234411	G	A	41	2.4	47	44.7	PRSS12	CCDS3709.1	c.1434C>T	p.R478R	silent	T
199	PD4177	PMF	JAK2	4	68606291	G	A	500	0.2	237	43.9	GNRHR	CCDS3517.1	c.894C>T	p.N298N	silent	M
200	PD4177	PMF	JAK2	1	84663455	G	C	224	0.9	173	43.4	PRKACB	CCDS693.1	c.731G>C	p.W244S	missense	M
201	PD4177	PMF	JAK2	16	21698920	A	G	25	0.0	49	38.8	OTOA	CCDS10600.2	c.586A>G	p.I196V	missense	M
202	PD4177	PMF	JAK2	1	146687379	T	C	111	1.8	88	38.6	FMO5	CCDS926.1	c.269A>G	p.E90G	missense	T
203	PD4177	PMF	JAK2	2	102490556	G	A	137	0.7	168	37.5	MAP4K4	ENST00000324219	c.2891G>A	p.C964Y	missense	M
204	PD4177	PMF	JAK2	9	5073770	G	T	99	4.0	106	34.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
205	PD4177	PMF	JAK2	15	55967796	A	G	48	0.0	75	33.3	PRTG	CCDS42040.1	c.1467T>C	p.Y489Y	silent	T
206	PD4177	PMF	JAK2	10	73990160	G	A	34	0.0	52	28.9	C10orf104	CCDS7314.1	c.179G>A	p.S60N	missense	M

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
207	PD4177	PMF	JAK2	X	96203967	C	T	67	0.0	38	23.7	DIAPH2	CCDS14467.1	c.1543C>T	p.Q515*	nonsense	M
208	PD4177	PMF	JAK2	2	189876477	G	A	55	0.0	72	23.6	COL3A1	CCDS2297.1	c.4378G>A	p.V1460I	missense	M
209	PD4177	PMF	JAK2	3	195512081	C	A	11	0.0	500	19.8	MUC4	ENST00000475231	c.6370G>T	p.G2124W	missense	M
210	PD4177	PMF	JAK2	18	47803329	G	T	25	0.0	87	19.5	MBD1	CCDS11943.1	c.265C>A	p.P89T	missense	M
211	PD4177	PMF	JAK2	13	79951583	C	T	46	0.0	58	17.2	RBM26	CCDS9462.1	c.258G>A	p.E86E	silent	M
212	PD4177	PMF	JAK2	22	45281760	T	C	17	0.0	121	7.4	PHF21B	CCDS14061.1	c.1256A>G	p.Y419C	missense	M
213	PD4178	PPV-MF	JAK2	9	5073770	G	T	122	18.9	93	98.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
214	PD4178	PPV-MF	JAK2	6	72806830	C	T	134	6.7	125	44.0	RIMS1	CCDS47449.1	c.424C>T	p.R142C	missense	M
215	PD4178	PPV-MF	JAK2	15	41862902	G	A	31	6.5	53	43.4	TYRO3	CCDS10080.1	c.1579+5G>A	p.?	essential splice	M
216	PD4178	PPV-MF	JAK2	6	32803121	C	T	66	4.6	65	41.5	TAP2	CCDS4755.1	c.755G>A	p.R252Q	missense	M
217	PD4178	PPV-MF	JAK2	X	55042088	A	C	86	4.7	59	39.0	ALAS2	CCDS14366.1	c.1091T>G	p.L364R	missense	M
218	PD4178	PPV-MF	JAK2	5	112889465	T	C	42	4.8	60	38.3	YTHDC2	CCDS4113.1	c.1966T>C	p.Y656H	missense	M
219	PD4178	PPV-MF	JAK2	4	106158455	T	G	147	5.4	112	37.5	TET2	CCDS47120.1	c.3356T>G	p.L1119*	nonsense	M
220	PD4178	PPV-MF	JAK2	12	96180979	G	C	40	0.0	55	12.7	NTN4	CCDS9054.1	c.323C>G	p.S108C	missense	M
221	PD4178	PPV-MF	JAK2	7	140045012	G	A	44	0.0	34	11.8	SLC37A3	CCDS5859.1	c.1174+4C>T	p.?	splice	M
222	PD4178	PPV-MF	JAK2	4	3133019	C	T	42	0.0	51	9.8	HTT	CCDS43206.1	c.1993C>T	p.R665C	missense	M
223	PD4178	PPV-MF	JAK2	9	16215760	T	C	125	0.0	119	7.6	C9orf92	ENST00000380683	c.190+10A>G	p.?	splice	M
224	PD4178	PPV-MF	JAK2	17	78796006	C	G	156	0.6	254	7.5	RPTOR	CCDS11773.1	c.896C>G	p.P299R	missense	M
225	PD4178	PPV-MF	JAK2	10	24822125	_	delGAA	71	0.0	90	5.6	KIAA1217	CCDS31165.1	c.3373_3375delGAA	p.E1125delE	inframe	M
226	PD4772	PET-PV	JAK2	9	5073770	G	T	191	0.5	162	46.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
227	PD4772	PET-PV	JAK2	22	38437084	G	T	31	0.0	500	23.6	POLR2F	ENST00000407936	c.462G>T	p.R154R	silent	T C
228	PD4772	PET-PV	JAK2	2	26437358	C	T	139	0.0	149	21.5	HADHA	CCDS1721.1	c.872G>A	p.R291Q	missense	T C
229	PD4772	PET-PV	JAK2	3	194062558	C	A	12	0.0	80	21.3	CPN2	CCDS33920.1	c.874G>T	p.V292F	missense	T C
230	PD4772	PET-PV	JAK2	12	117914262	C	A	17	0.0	39	20.5	KSR2	ENST00000339824	c.2582+7G>T	p.?	splice	T C
231	PD4772	PET-PV	JAK2	18	5290980	G	A	26	0.0	149	18.8	ZFP161	CCDS11837.1	c.1227C>T	p.N409N	silent	T C
232	PD4772	PET-PV	JAK2	12	70964834	C	T	214	0.0	274	17.2	PTPRB	CCDS44943.1	c.3342G>A	p.Q1114Q	silent	T
233	PD4772	PET-PV	JAK2	6	70071337	A	T	209	0.0	155	16.1	BAI3	CCDS4968.1	c.4172A>T	p.E1391V	missense	T C
234	PD4772	PET-PV	JAK2	22	29120993	T	A	108	0.9	143	16.1	CHEK2	CCDS33629.1	c.693A>T	p.E231D	missense	T C
235	PD4772	PET-PV	JAK2	16	30976208	A	G	36	0.0	237	14.8	SETD1A	CCDS32435.1	c.1145A>G	p.Y382C	missense	T C
236	PD4772	PET-PV	JAK2	15	65917894	G	A	24	0.0	109	14.7	SLC24A1	CCDS45284.1	c.1476G>A	p.E492E	silent	T C
237	PD4772	PET-PV	JAK2	1	33404025	A	C	132	0.0	151	13.3	RNF19B	CCDS372.2	c.1718T>G	p.I573R	missense	T C
238	PD4772	PET-PV	JAK2	10	12043738	T	C	210	0.5	92	13.0	UPF2	CCDS7086.1	c.1591A>G	p.T531A	missense	T C
239	PD4772	PET-PV	JAK2	5	11364981	C	T	36	0.0	102	10.8	CTNND2	CCDS3881.1	c.1199G>A	p.S400N	missense	T C
240	PD4772	PET-PV	JAK2	10	78944618	G	C	262	0.0	197	10.7	KCNMA1	CCDS7352.1	c.659C>G	p.A220G	missense	T C
241	PD4772	PET-PV	JAK2	8	67900698	C	T	46	0.0	57	10.5	LRRC67	CCDS34902.1	c.607G>A	p.G203R	missense	T C
242	PD4772	PET-PV	JAK2	21	38538413	T	C	104	0.0	170	7.1	TTC3L	CCDS13651.1	c.3897T>C	p.V1299V	silent	T C
243	PD4772	PET-PV	JAK2	17	33496956	G	T	24	0.0	93	5.4	UNC45B	CCDS11292.1	c.1547+6G>T	p.?	splice	T C
244	PD4773	PV	JAK2	9	5073770	G	T	136	1.5	174	51.7	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
245	PD4773	PV	JAK2	5	147889325	G	T	97	6.2	73	39.7	HTR4	CCDS34272.1	c.770C>A	p.A257E	missense	T C
246	PD4773	PV	JAK2	X	18911669	G	T	221	2.7	212	39.6	PHKA2	CCDS14190.1	c.3642C>A	p.Y1214*	nonsense	T C
247	PD4773	PV	JAK2	1	203468918	A	T	95	2.1	95	37.9	OPTC	CCDS1439.1	c.671A>T	p.E224V	missense	T
248	PD4773	PV	JAK2	11	61570286	C	T	65	0.0	44	34.1	FADS1	CCDS8011.2	c.1441G>A	p.A481T	missense	T C
249	PD4773	PV	JAK2	14	20216077	T	A	72	1.4	58	32.8	OR4Q3	CCDS32020.1	c.491T>A	p.V164D	missense	T C
250	PD4773	PV	JAK2	15	86205632	C	G	163	0.6	112	28.6	AKAP13	CCDS32320.1	c.4759C>G	p.P1587A	missense	T
251	PD4773	PV	JAK2	X	123554168	C	T	152	2.6	122	27.9	ODZ1	CCDS14609.1	c.4954G>A	p.V1652I	missense	T C
252	PD4773	PV	JAK2	15	74964100	C	T	81	2.5	76	27.6	EDC3	CCDS10267.1	c.180G>A	p.T60T	silent	T
253	PD4773	PV	JAK2	5	55178962	C	T	91	0.0	88	18.2	IL31RA	CCDS3970.2	c.545C>T	p.A182V	missense	T C
254	PD4773	PV	JAK2	2	10264927	T	C	105	0.0	123	16.3	RRM2	CCDS1669.1	c.519T>C	p.S173S	silent	T
255	PD4773	PV	JAK2	22	46677555	G	T	94	2.1	68	13.2	TTC38	CCDS43030.1	c.675G>T	p.E225D	missense	T C
256	PD4773	PV	JAK2	10	67862990	C	T	50	0.0	50	10.0	CTNNA3	CCDS7269.1	c.1902G>A	p.E634E	silent	T
257	PD4773	PV	JAK2	2	198266584	C	A	177	0.0	141	5.7	SF3B1	CCDS33356.1	c.2252G>T	p.G751V	missense	T
258	PD4774	PV	JAK2	9	5073770	G	T	144	3.5	200	70.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
259	PD4774	PV	JAK2	11	55890395	C	T	430	1.9	399	35.6	OR8H3	CCDS31519.1	c.547C>T	p.P183S	missense	T
260	PD4774	PV	JAK2	6	5404914	T	A	124	0.8	76	32.9	FARS2	CCDS4494.1	c.752T>A	p.M251K	missense	T
261	PD4774	PV	JAK2	1	12837518	C	T	198	0.5	266	27.4	PRAMEF12	CCDS41254.1	c.1228C>T	p.L410L	silent	T
262	PD4774	PV	JAK2	8	17823896	T	C	500	0.2	500	24.6	PCM1	CCDS47812.1	c.3135T>C	p.N1045N	silent	T
263	PD4774	PV	JAK2	20	56185321	G	A	108	0.0	213	23.0	ZBP1	CCDS13461.1	c.977C>T	p.S326L	missense	T
264	PD4774	PV	JAK2	1	214818100	C	T	58	3.5	99	21.2	CENPF	CCDS31023.1	c.5187C>T	p.T1729T	silent	T
265	PD4774	PV	JAK2	7	91603160	T	A	224	0.5	144	19.4	AKAP9	CCDS5622.1	c.184T>A	p.C62S	missense	T
266	PD4774	PV	JAK2	19	53989928	G	A	500	0.0	500	19.4	ZNF813	CCDS46172.1	c.58G>A	p.E20K	missense	T
267	PD4774	PV	JAK2	1	205385414	G	T	178	0.6	130	16.2	LEMD1	ENST00000367153	c.216C>A	p.I72I	silent	T
268	PD4774	PV	JAK2	2	220342434	G	A	15	0.0	39	15.4	SPEG	CCDS42824.1	c.4753G>A	p.G1585R	missense	T
269	PD4774	PV	JAK2	18	52896228	C	T	73	0.0	115	13.9	TCF4	CCDS42438.1	c.1729G>A	p.E577K	missense	T
270	PD4774	PV	JAK2	17	63221498	T	C	47	0.0	46	13.0	RGS9	CCDS42373.1	c.1786T>C	p.F596L	missense	T
271	PD4774	PV	JAK2	11	47506081	A	G	107	0.0	129	10.9	CELF1	CCDS31482.1	c.311-6T>C	p.?	splice	T
272	PD4774	PV	JAK2	3	47165976	A	T	183	0.6	188	10.6	SETD2	CCDS2749.2	c.150T>A	p.A50A	silent	T
273	PD4774	PV	JAK2	19	55241196	G	A	500	0.0	216	9.3	KIR3DL3	CCDS12903.1	c.893G>A	p.R298H	missense	T
274	PD4774	PV	JAK2	2	286212	C	T	155	1.3	231	9.1	FAM150B	CCDS46218.1	c.308-9G>A	p.?	splice	T
275	PD4774	PV	JAK2	18	31319793	G	A	124	0.0	240	8.8	ASXL3	CCDS45847.1	c.2425G>A	p.E809K	missense	T
276	PD4774	PV	JAK2	17	20149313	C	T	78	0.0	92	8.7	CYTSB	CCDS32590.1	c.2426C>T	p.T809I	missense	T
277	PD4774	PV	JAK2	10	90512427	C	T	224	0.0	313	8.6	LIPK	CCDS44455.1	c.1114C>T	p.H372Y	missense	T
278	PD4774	PV	JAK2	11	118502727	G	A	54	0.0	71	8.5	PHLDB1	CCDS8401.1	c.2198G>A	p.R733H	missense	T
279	PD4774	PV	JAK2	12	7805341	G	T	65	0.0	94	7.5	APOBEC1	CCDS8579.1	c.135C>A	p.G45G	silent	T
280	PD4774	PV	JAK2	13	36445403	C	G	169	0.0	147	6.1	DCLK1	CCDS9354.1	c.898G>C	p.G300R	missense	T
281	PD4775	PPV-MF	JAK2	9	5073770	G	T	76	25.0	129	99.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
282	PD4775	PPV-MF	JAK2	4	106180838	_	delG	145	3.5	117	77.8	TET2	CCDS47120.1	c.3866delG	p.C1289fs*74	frameshift	M
283	PD4775	PPV-MF	JAK2	13	96242650	T	C	147	3.4	145	50.3	DZIP1	CCDS9478.1	c.1726A>G	p.S576G	missense	M
284	PD4775	PPV-MF	JAK2	1	205589093	C	A	60	6.7	67	49.3	ELK4	CCDS1456.1	c.1080+1G>T	p.?	missense	M
285	PD4775	PPV-MF	JAK2	5	174868910	G	A	34	2.9	61	47.5	DRD1	CCDS4393.1	c.1193C>T	p.S398F	missense	M
286	PD4775	PPV-MF	JAK2	7	127014483	T	C	279	3.2	153	47.1	ZNF800	CCDS5795.1	c.907A>G	p.R303G	missense	M
287	PD4775	PPV-MF	JAK2	2	186665453	G	C	365	2.2	281	44.1	FSIP2	ENST00000424728	c.11420G>C	p.G3807A	missense	M
288	PD4775	PPV-MF	JAK2	13	39587196	G	A	271	2.6	133	40.6	C13orf23	CCDS9368.2	c.2193C>T	p.T731T	missense	M
289	PD4775	PPV-MF	JAK2	20	37270390	G	A	32	0.0	73	27.4	C20orf95	ENST00000373345	c.1142G>A	p.R381H	missense	M
290	PD4775	PPV-MF	JAK2	9	105767103	C	T	249	1.2	87	25.3	CYLC2	CCDS35085.1	c.307C>T	p.R103C	missense	M
291	PD4775	PPV-MF	JAK2	14	62204896	T	A	59	0.0	85	7.1	HIF1A	CCDS9753.1	c.1341T>A	p.N447K	missense	M
292	PD4775	PPV-MF	JAK2	1	35907930	G	A	165	1.8	140	5.7	KIAA0319L	CCDS390.1	c.2787C>T	p.S929S	missense	M
293	PD4776	PV	JAK2	9	5073770	G	T	199	4.0	193	60.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
294	PD4776	PV	JAK2	7	130365837	G	A	106	0.0	141	41.8	TSGA13	CCDS5824.1	c.121C>T	p.Q41*	nonsense	T
295	PD4776	PV	JAK2	15	49034218	T	C	268	0.4	211	40.3	CEP152	CCDS42033.1	c.3747A>G	p.Q1249Q	silent	T
296	PD4776	PV	JAK2	12	64819669	T	C	238	2.1	358	40.2	XPOT	CCDS31852.1	c.1647T>C	p.S549S	silent	T
297	PD4776	PV	JAK2	17	38635928	C	T	19	0.0	75	40.0	TNS4	CCDS11368.1	c.1908G>A	p.R636R	silent	T
298	PD4777	PV	JAK2	9	5073770	G	T	164	0.0	238	40.3	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
299	PD4777	PV	JAK2	12	120152131	G	C	19	0.0	69	30.4	CIT	CCDS9192.1	c.4051C>G	p.R1351G	missense	T
300	PD4777	PV	JAK2	3	164750405	G	T	157	0.0	165	24.2	SI	CCDS3196.1	c.2641C>A	p.L881I	missense	T
301	PD4777	PV	JAK2	13	25910402	T	A	124	0.0	219	23.3	NUPL1	ENST00000381745	c.1487T>A	p.L496H	missense	T
302	PD4777	PV	JAK2	4	114824524	T	A	284	0.0	295	22.4	ARSJ	CCDS43264.1	c.706A>T	p.I236L	missense	T
303	PD4777	PV	JAK2	6	24667050	G	A	30	0.0	156	20.5	TTRAP	CCDS4557.1	c.41C>T	p.A14V	missense	T
304	PD4777	PV	JAK2	2	209113112	C	T	97	0.0	90	16.7	IDH1	CCDS2381.1	c.395G>A	p.R132H	missense	T
305	PD4777	PV	JAK2	5	32239127	G	A	19	0.0	85	11.8	MTMR12	CCDS34138.1	c.1324C>T	p.R442C	missense	T
306	PD4777	PV	JAK2	X	36329024	G	A	18	0.0	52	11.5	CXorf30	ENST00000378653	c.1457G>A	p.R486H	missense	T
307	PD4777	PV	JAK2	5	110411672	G	A	28	0.0	108	9.3	TSLP	CCDS4101.1	c.380G>A	p.R127K	missense	T
308	PD4777	PV	JAK2	2	179427553	G	A	57	0.0	109	8.3	TTN	ENST00000356127	c.75596C>T	p.A25199V	missense	T
309	PD4777	PV	JAK2	4	88767549	G	A	164	0.0	393	4.6	MEPE	CCDS3625.1	c.1529G>A	p.S510N	missense	T
310	PD4778	PV	JAK2	9	5073770	G	T	168	4.2	139	61.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
311	PD4778	PV	JAK2	8	113303924	A	G	126	0.0	159	32.1	CSMD3	CCDS6315.1	c.8789T>C	p.V2930A	missense	T
312	PD4778	PV	JAK2	17	65185786	C	A	66	1.5	112	27.7	HELZ	CCDS42374.1	c.783G>T	p.K261N	missense	T
313	PD4778	PV	JAK2	1	42880441	C	T	84	1.2	87	27.6	RIMKLA	CCDS466.2	c.972C>T	p.N324N	silent	T
314	PD4778	PV	JAK2	18	12814309	T	C	27	0.0	49	26.5	PTPN2	CCDS11865.1	c.751A>G	p.M251V	missense	T
315	PD4778	PV	JAK2	1	214625308	A	G	48	2.1	64	25.0	PTPN14	CCDS1514.1	c.184T>C	p.F62L	missense	T
316	PD4778	PV	JAK2	7	21584775	T	A	154	2.0	223	23.3	DNAH11	ENST00000424253	c.495+8T>A	p.?	splice	T
317	PD4778	PV	JAK2	X	23411329	_	delA	107	0.0	238	23.1	PTCHD1	CCDS35215.2	c.1694delA	p.E565fs*12	frameshift	T
318	PD4778	PV	JAK2	17	27005085	C	A	65	6.2	139	23.0	SUPT6H	CCDS32596.1	c.1000-9C>A	p.?	splice	T
319	PD4778	PV	JAK2	2	234072457	A	G	174	0.0	192	16.7	INPP5D	ENST00000359570	c.1345A>G	p.I449V	missense	T
320	PD4778	PV	JAK2	4	36118699	C	A	45	0.0	48	10.4	ARAP2	CCDS3441.1	c.4022G>T	p.C1341F	missense	T
321	PD4780	PMF	JAK2	9	5073770	G	T	380	20.5	159	64.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
322	PD4780	PMF	JAK2	2	25457242	C	T	19	10.5	61	45.9	DNMT3A	CCDS33157.1	c.2645G>A	p.R882H	missense	T C
323	PD4780	PMF	JAK2	11	94730931	A	T	250	3.2	190	11.6	KDM4D	CCDS8302.1	c.395A>T	p.N132I	missense	T C
324	PD4780	PMF	JAK2	19	47935366	G	C	148	2.0	142	5.6	SLC8A2	ENST00000391903	c.1931C>G	p.T644R	missense	T C
325	PD4780	PMF	JAK2	16	71113806	A	T	163	1.2	160	5.0	HYDIN2	CCDS42189.1	c.1720T>A	p.F574I	missense	T C
326	PD4781	PV	JAK2	9	5073770	G	T	176	0.0	185	91.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
327	PD4781	PV	JAK2	2	160086905	C	G	23	0.0	72	51.4	TANC1	CCDS42766.1	c.4968C>G	p.H1656Q	missense	T
328	PD4781	PV	JAK2	4	106196561	C	T	421	0.0	174	47.1	TET2	CCDS47120.1	c.4894C>T	p.Q1632*	nonsense	T
329	PD4781	PV	JAK2	1	237632393	G	A	218	0.0	303	45.2	RYR2	ENST00000366574	c.1608G>A	p.A536A	silent	T
330	PD4781	PV	JAK2	6	128150732	C	T	224	0.0	229	44.5	THEMIS	CCDS34534.1	c.598G>A	p.V200I	missense	T
331	PD4781	PV	JAK2	11	47498982	A	G	32	0.0	80	43.8	CELF1	CCDS31482.1	c.764-5T>C	p.?	splice	T
332	PD4781	PV	JAK2	3	153840279	T	G	18	0.0	32	43.8	ENSG00000114790	CCDS46938.1	c.498T>G	p.T166T	silent	T
333	PD4781	PV	JAK2	13	27847423	T	C	28	0.0	56	41.1	RASL11A	CCDS9321.1	c.521T>C	p.L174P	missense	T
334	PD4781	PV	JAK2	6	80718118	T	A	136	0.0	108	39.8	TTK	CCDS4993.1	c.378T>A	p.D126E	missense	T
335	PD4781	PV	JAK2	4	48592819	A	G	500	0.0	219	37.9	FRYL	CCDS43227.1	c.1364T>C	p.L455P	missense	T
336	PD4781	PV	JAK2	3	42956783	C	A	90	2.2	124	37.9	ZNF662	CCDS46807.1	c.1296C>A	p.H432Q	missense	T
337	PD4781	PV	JAK2	11	47331140	A	G	96	0.0	136	37.5	MADD	CCDS7930.1	c.4135A>G	p.I1379V	missense	T
338	PD4781	PV	JAK2	15	29346853	C	T	52	0.0	48	37.5	APBA2	CCDS10022.1	c.766C>T	p.P256S	missense	T
339	PD4781	PV	JAK2	10	112541459	C	T	12	0.0	108	37.0	RBM20	CCDS44477.1	c.1092C>T	p.F364F	silent	T
340	PD4781	PV	JAK2	10	133748027	G	A	198	1.0	500	36.8	PPP2R2D	ENST00000455566	c.73G>A	p.G25S	missense	T
341	PD4781	PV	JAK2	10	37488720	C	T	211	0.0	83	25.3	ANKRD30A	CCDS7193.1	c.2610+4C>T	p.?	splice	T
342	PD4781	PV	JAK2	8	146107097	_	delCAAT	12	0.0	42	14.3	ZNF250	CCDS34972.1	c.1483_1486delATTG	p.I495fs*3	frameshift	T
343	PD4781	PV	JAK2	7	540862	G	A	19	0.0	275	6.9	PDGFA	CCDS34578.1	c.471C>T	p.Y157Y	silent	T
344	PD4781	PV	JAK2	17	58740749	C	T	72	0.0	163	5.5	PPM1D	CCDS11625.1	c.1654C>T	p.R552*	nonsense	T
345	PD4781	PV	JAK2	8	30916726	T	A	500	0.0	343	3.5	WRN	CCDS6082.1	c.154T>A	p.S52T	missense	T
346	PD4940	PV	JAK2	7	129666132	G	A	84	1.2	167	31.7	ZC3HC1	CCDS34753.1	c.642C>T	p.I214I	silent	M
347	PD4940	PV	JAK2	14	64532156	G	T	188	7.5	119	28.6	SYNE2	CCDS9761.2	c.10219G>T	p.V3407L	missense	M
348	PD4940	PV	JAK2	9	5073770	G	T	442	6.8	245	26.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
349	PD4940	PV	JAK2	2	27015025	C	T	107	3.7	99	25.3	CENPA	CCDS1729.1	c.127C>T	p.R43W	missense	M
350	PD4941	PV	JAK2	9	5073770	G	T	157	1.2	153	13.7	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
351	PD4941	PV	JAK2	1	109742543	_	insGCAA	44	0.0	44	6.8	KIAA1324	CCDS794.1	c.2569_2570insGCAA	p.P857fs*8	frameshift	T
352	PD4941	PV	JAK2	3	14974221	T	A	207	0.0	268	6.0	FGD5	CCDS46767.1	c.4335T>A	p.D1445E	missense	T
353	PD4941	PV	JAK2	10	20568746	T	A	121	0.0	149	4.7	PLXDC2	CCDS7132.1	c.1588T>A	p.*530K	nonstop	T
354	PD4944	PV	JAK2	7	72892410	G	A	125	0.8	160	31.9	BAZ1B	CCDS5549.1	c.1381C>T	p.P461S	missense	T
355	PD4944	PV	JAK2	9	5073770	G	T	126	0.8	270	28.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
356	PD4944	PV	JAK2	12	95914938	C	T	65	0.0	174	27.6	USP44	CCDS9053.1	c.1774G>A	p.V592I	missense	T
357	PD4944	PV	JAK2	10	55582162	G	A	46	0.0	91	27.5	PCDH15	CCDS44404.1	c.5330C>T	p.S1777F	missense	T
358	PD4944	PV	JAK2	9	84606792	_	delT	266	0.4	448	24.3	ENSG00000214929	ENSG00000214929	c.1407delT	p.W471fs*7	frameshift	T
359	PD4944	PV	JAK2	8	134144057	C	T	60	0.0	91	14.3	TG	CCDS34944.1	c.7864C>T	p.L2622L	silent	T
360	PD4947	PPV-MF	JAK2	9	5073770	G	T	178	0.0	156	76.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
361	PD4947	PPV-MF	JAK2	X	18276358	T	C	44	0.0	42	64.3	SCML2	CCDS14185.1	c.1079A>G	p.Y360C	missense	M
362	PD4947	PPV-MF	JAK2	X	34150174	G	A	73	0.0	48	56.3	FAM47A	CCDS43926.1	c.222C>T	p.D74D	silent	M

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
363	PD4947	PPV-MF	JAK2	9	139277995	_	delGCT	1	0.0	10	50.0	SNAPC4	CCDS6998.1	c.1624_1626delAGC	p.S542delS	inframe	M
364	PD4947	PPV-MF	JAK2	2	20201812	C	T	77	0.0	124	34.7	MATN3	CCDS46226.1	c.946G>A	p.G316R	missense	M
365	PD4947	PPV-MF	JAK2	4	104080185	C	T	93	0.0	90	34.4	CENPE	CCDS34042.1	c.2583G>A	p.L861L	silent	M
366	PD4947	PPV-MF	JAK2	12	121618196	A	G	75	0.0	60	31.7	P2RX7	CCDS9213.1	c.1228A>G	p.R410G	missense	M
367	PD4947	PPV-MF	JAK2	5	7897263	G	A	61	0.0	94	30.9	MTRR	CCDS3874.1	c.1936G>A	p.E646K	missense	M
368	PD4947	PPV-MF	JAK2	12	112673344	G	A	49	0.0	102	30.4	C12orf51	CCDS44978.1	c.4423C>T	p.L1475L	silent	M
369	PD4947	PPV-MF	JAK2	1	62380259	G	T	192	0.0	189	30.2	INADL	CCDS617.2	c.3493G>T	p.V1165F	missense	M
370	PD4947	PPV-MF	JAK2	17	49281241	C	T	148	0.0	245	27.8	MBTD1	CCDS11581.2	c.650G>A	p.G217D	missense	M
371	PD4947	PPV-MF	JAK2	1	111893999	A	G	47	2.1	36	25.0	C1orf88	CCDS833.1	Non-coding	r.991a>g	UTR	M
372	PD4947	PPV-MF	JAK2	20	31022287	_	insA	51	0.0	104	22.1	ASXL1	CCDS13201.1	c.1772_1773insA	p.Y591fs*1	frameshift	M
373	PD4947	PPV-MF	JAK2	2	24484025	C	G	73	0.0	64	21.9	ITSN2	CCDS1710.2	c.2632G>C	p.A878P	missense	M
374	PD4947	PPV-MF	JAK2	3	186760924	C	T	36	0.0	67	19.4	ST6GAL1	CCDS3285.1	c.433C>T	p.R145W	missense	M
375	PD4947	PPV-MF	JAK2	7	796489	C	T	29	0.0	99	19.2	HEATR2	CCDS34580.1	c.1328C>T	p.T443M	missense	M
376	PD4947	PPV-MF	JAK2	1	181549728	C	A	229	0.0	208	16.4	CACNA1E	ENST00000367573	c.770-3C>A	p.?	splice	M
377	PD4947	PPV-MF	JAK2	19	33608878	G	T	39	0.0	50	14.0	GPATCH1	CCDS12428.1	c.2344G>T	p.G782C	missense	M
378	PD4947	PPV-MF	JAK2	17	7577535	C	A	67	0.0	31	12.9	TP53	CCDS11118.1	c.746G>T	p.R249M	missense	M
379	PD4947	PPV-MF	JAK2	X	13626530	A	G	81	0.0	58	12.1	EGFL6	CCDS14155.1	c.743A>G	p.K248R	missense	M
380	PD4947	PPV-MF	JAK2	20	31022656	_	delC	23	0.0	38	10.5	ASXL1	CCDS13201.1	c.2141delC	p.R715fs*10	frameshift	M
381	PD4947	PPV-MF	JAK2	11	114400913	T	C	141	0.0	97	10.3	FAM55A	CCDS8372.1	c.391A>G	p.N131D	missense	M
382	PD4947	PPV-MF	JAK2	11	33108653	G	A	226	0.0	318	9.4	CSTF3	CCDS7883.1	c.1676C>T	p.P559L	missense	M
383	PD4947	PPV-MF	JAK2	4	106158248	C	A	92	0.0	77	9.1	TET2	CCDS47120.1	c.3149C>A	p.S1050*	nonsense	M
384	PD4947	PPV-MF	JAK2	5	33643529	G	A	164	0.0	108	8.3	ADAMTS12	ENST00000504830	c.1526C>T	p.S509F	missense	M
385	PD4947	PPV-MF	JAK2	4	70361595	T	A	307	0.0	240	8.3	UGT2B4	CCDS43234.1	Non-coding	r.32a>u	UTR	M
386	PD4947	PPV-MF	JAK2	18	55287857	C	T	59	0.0	94	7.5	NARS	CCDS32837.1	c.37G>A	p.G13R	missense	M
387	PD4947	PPV-MF	JAK2	12	81528768	C	A	78	0.0	69	7.3	ACSS3	CCDS9022.1	c.630C>A	p.R210R	silent	M
388	PD4990	PMF	JAK2	9	133779655	G	A	51	0.0	72	31.9	FIBCD1	CCDS6937.1	c.1182C>T	p.S394S	silent	T
389	PD4990	PMF	JAK2	3	48664359	G	C	55	0.0	55	30.9	SLC26A6	CCDS43087.1	c.2023C>G	p.L675V	missense	T C
390	PD4990	PMF	JAK2	19	56720317	G	T	102	0.0	72	29.2	ZSCAN5C	ENST00000376267	c.1239G>T	p.T413T	silent	T
391	PD4990	PMF	JAK2	9	35957932	C	T	56	0.0	44	25.0	OR2S2	CCDS6596.2	c.164G>A	p.R55H	missense	T
392	PD4990	PMF	JAK2	10	50531367	G	A	97	0.0	94	20.2	C10orf71	CCDS44387.1	c.777G>A	p.T259T	silent	T
393	PD4990	PMF	JAK2	11	118184577	G	A	28	0.0	30	20.0	CD3E	CCDS31685.1	c.508G>A	p.G170S	missense	T
394	PD4990	PMF	JAK2	2	15493788	T	A	228	0.0	209	18.2	NBAS	CCDS1685.1	c.3978A>T	p.E1326D	missense	T
395	PD4990	PMF	JAK2	1	74797193	C	T	40	0.0	33	18.2	TNNI3K	CCDS44161.1	c.807C>T	p.F269F	silent	T C
396	PD4990	PMF	JAK2	16	53706849	C	T	69	0.0	62	17.7	RPGRIP1L	CCDS32447.1	c.962G>A	p.R321H	missense	T
397	PD4990	PMF	JAK2	2	21363871	C	T	161	0.6	130	16.9	ENSG00000218819	ENST00000405799	c.3532C>T	p.R1178C	missense	T
398	PD4990	PMF	JAK2	10	87362144	G	A	31	0.0	25	16.0	GRID1	CCDS31236.1	c.2916C>T	p.N972N	silent	T C
399	PD4990	PMF	JAK2	X	71804146	C	T	74	0.0	71	15.5	PHKA1	CCDS14421.1	c.3250G>A	p.G1084R	missense	T
400	PD4990	PMF	JAK2	14	24528165	T	C	26	0.0	40	15.0	LRRC16B	CCDS32054.1	c.1541T>C	p.F514S	missense	T
401	PD4990	PMF	JAK2	8	2855593	C	T	201	0.5	181	14.9	CSMD1	ENST00000318252	c.7504G>A	p.V2502M	missense	T
402	PD4990	PMF	JAK2	3	38591839	C	T	71	0.0	68	14.7	SCN5A	CCDS46796.1	c.6024G>A	p.P2008P	silent	T
403	PD4990	PMF	JAK2	9	5073770	G	T	159	0.0	112	14.3	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
404	PD4990	PMF	JAK2	4	88295868	C	T	153	0.0	140	14.3	HSD17B11	CCDS3619.1	c.433G>A	p.V145I	missense	T C
405	PD4990	PMF	JAK2	2	25463184	G	A	73	2.7	92	14.1	DNMT3A	CCDS33157.1	c.2309C>T	p.S770L	missense	T C
406	PD4990	PMF	JAK2	2	25457242	C	T	22	0.0	48	6.3	DNMT3A	CCDS33157.1	c.2645G>A	p.R882H	missense	T C
407	PD4994	PMF	MPL	X	30269088	G	A	11	0.0	10	100.0	MAGEB1	CCDS14222.1	c.478G>A	p.V160I	missense	T
408	PD4994	PMF	MPL	5	163395	C	T	24	0.0	61	59.0	PLEKHG4B	CCDS34124.1	c.2140C>T	p.R714C	missense	T
409	PD4994	PMF	MPL	5	163382	G	T	19	0.0	48	56.3	PLEKHG4B	CCDS34124.1	c.2127G>T	p.Q709H	missense	T
410	PD4994	PMF	MPL	1	43815009	G	T	40	2.5	54	55.6	MPL	CCDS483.1	c.1544G>T	p.W515L	missense	T
411	PD4994	PMF	MPL	12	117968764	G	A	126	1.6	149	53.0	KSR2	ENST00000339824	c.1784C>T	p.P595L	missense	T
412	PD4994	PMF	MPL	17	45364547	G	A	53	0.0	56	50.0	ITGB3	CCDS11511.1	c.889G>A	p.G297R	missense	T
413	PD4994	PMF	MPL	9	35092523	C	G	110	0.9	94	48.9	PIGO	CCDS6575.1	c.1361G>C	p.G454A	missense	T
414	PD4994	PMF	MPL	1	84663454	T	G	147	2.7	134	47.8	PRKACB	CCDS693.1	c.730T>G	p.W244G	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
415	PD4994	PMF	MPL	12	13716667	C	G	25	0.0	44	45.5	GRIN2B	CCDS8662.1	c.3505G>C	p.G1169R	missense	T
416	PD4994	PMF	MPL	20	2969091	G	A	108	1.9	82	45.1	PTPRA	CCDS13038.1	c.709G>A	p.D237N	missense	T
417	PD4994	PMF	MPL	12	53681865	G	T	14	0.0	28	42.9	ESPL1	CCDS8852.1	c.4286G>T	p.R1429M	missense	T
418	PD4994	PMF	MPL	15	57565358	C	T	68	1.5	74	41.9	TCF12	CCDS10160.1	c.1876C>T	p.R626*	nonsense	T
419	PD4994	PMF	MPL	3	73438996	G	A	40	0.0	46	41.3	PDZRN3	CCDS33789.1	c.1387C>T	p.R463C	missense	T
420	PD4994	PMF	MPL	15	99670821	C	A	29	0.0	53	37.7	SYNM	ENST00000336292	c.2253C>A	p.P751P	silent	T
421	PD4994	PMF	MPL	4	106180824	_	delCTT	104	0.0	77	29.9	TET2	CCDS47120.1	c.3852_3854delCTT	p.F1285delF	inframe	T
422	PD4994	PMF	MPL	18	8806981	C	T	11	0.0	17	29.4	KIAA0802	CCDS11841.1	c.2527C>T	p.R843W	missense	T
423	PD4994	PMF	MPL	16	75513433	G	A	24	0.0	28	28.6	CHST6	CCDS10918.1	c.294C>T	p.S98S	silent	T
424	PD4994	PMF	MPL	2	88125234	T	C	17	0.0	34	11.8	RGPD2	CCDS42710.1	Non-coding	r.53a>g	UTR	T
425	PD4996	PET-MF	unmutated	9	91617201	C	T	30	0.0	44	56.8	S1PR3	CCDS6680.1	c.1086C>T	p.S362S	silent	T
426	PD4996	PET-MF	unmutated	3	47278102	G	A	20	0.0	30	53.3	KIF9	CCDS2752.1	c.1932C>T	p.Y644Y	silent	T
427	PD4996	PET-MF	unmutated	4	76714946	G	T	118	0.0	104	49.0	USO1	ENST00000264904	c.1128G>T	p.L376F	missense	T
428	PD4996	PET-MF	unmutated	1	27480515	T	C	42	0.0	59	44.1	SLC9A1	CCDS295.1	c.311A>G	p.E104G	missense	T
429	PD4996	PET-MF	unmutated	3	54913060	G	A	328	0.3	231	43.7	CACNA2D3	ENST00000474759	c.1726G>A	p.V576M	missense	T
430	PD4996	PET-MF	unmutated	3	121575935	A	G	69	0.0	55	43.6	EAF2	CCDS3006.1	c.416A>G	p.N139S	missense	T
431	PD4996	PET-MF	unmutated	4	123663666	G	A	83	0.0	76	39.5	BBS12	CCDS3728.1	c.619G>A	p.E207K	missense	T
432	PD4996	PET-MF	unmutated	21	45504088	T	C	12	0.0	18	38.9	TRAPPC10	CCDS13704.1	c.2324T>C	p.I775T	missense	T
433	PD4996	PET-MF	unmutated	8	139732990	C	T	311	0.0	289	38.8	COL22A1	CCDS6376.1	c.2347G>A	p.G783S	missense	T
434	PD4996	PET-MF	unmutated	5	140482394	T	G	72	0.0	70	38.6	PCDHB3	CCDS4245.1	c.2161T>G	p.S721A	missense	T
435	PD4996	PET-MF	unmutated	20	40730778	C	A	50	0.0	52	19.2	PTPRT	CCDS42874.1	c.3700G>T	p.A1234S	missense	T
436	PD4996	PET-MF	unmutated	1	152383006	_	delT	216	0.0	141	5.7	CRNN	CCDS1010.1	c.552delA	p.Q184fs*23	frameshift	T
437	PD4996	PET-MF	unmutated	19	13054565	_	delGCAGA<42>AGGAG	21	4.8	8	25.0	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M T C
438	PD5003	ET	unmutated	10	44104735	C	A	34	0.0	31	16.1	ZNF485	CCDS7205.2	c.184C>A	p.Q62K	missense	M
439	PD5003	ET	unmutated	8	134050936	C	A	21	0.0	31	16.1	SLA	CCDS47923.1	c.784G>T	p.E262*	nonsense	M
440	PD5003	ET	unmutated	19	12297505	C	A	45	0.0	33	15.2	ZNF136	CCDS32916.1	c.312C>A	p.S104R	missense	M
441	PD5003	ET	unmutated	1	67519621	C	A	44	0.0	43	14.0	SLC35D1	CCDS636.1	c.76G>T	p.E26*	nonsense	M
442	PD5003	ET	unmutated	12	57843673	C	A	83	0.0	36	13.9	INHBC	CCDS8938.1	c.927C>A	p.G309G	silent	M
443	PD5003	ET	unmutated	15	101566186	C	A	19	0.0	52	11.5	LRRK1	CCDS42086.1	c.2249C>A	p.A750D	missense	M
444	PD5003	ET	unmutated	5	156811442	C	A	101	0.0	82	8.5	ENSG00000204823	ENST00000377571	c.366C>A	p.L122L	silent	M
445	PD5003	ET	unmutated	15	42742663	C	A	215	0.0	103	7.8	ZFP106	CCDS32208.1	c.1738G>T	p.E580*	nonsense	M
446	PD5003	ET	unmutated	11	46742363	C	A	35	0.0	104	7.7	F2	CCDS31476.1	c.289C>A	p.R97R	silent	M
447	PD5003	ET	unmutated	19	13054565	_	delGCAGA<42>AGGAG	19	5.3	18	22.2	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M T C
448	PD5004	PMF	unmutated	11	64084957	C	T	13	0.0	11	72.7	TRMT112	CCDS8068.1	c.42G>A	p.R14R	silent	T
449	PD5004	PMF	unmutated	11	48286217	G	A	66	0.0	91	52.8	OR4X1	CCDS31487.1	c.805G>A	p.V269I	missense	T
450	PD5004	PMF	unmutated	19	9088357	A	T	105	0.0	128	48.4	MUC16	ENST00000397910	c.3458T>A	p.I1153N	missense	T
451	PD5004	PMF	unmutated	12	69107542	A	G	187	0.0	79	45.6	NUP107	CCDS8985.1	c.923A>G	p.Q308R	missense	T
452	PD5004	PMF	unmutated	2	189406042	G	C	130	0.0	81	42.0	GULP1	CCDS2295.1	c.396G>C	p.K132N	missense	T
453	PD5004	PMF	unmutated	4	37448315	C	T	106	0.0	98	40.8	KIAA1239	CCDS47040.1	c.4705C>T	p.R1569W	missense	T
454	PD5004	PMF	unmutated	21	44514777	T	G	48	0.0	38	39.5	U2AF1	CCDS13694.1	c.470A>C	p.Q157P	missense	T
455	PD5004	PMF	unmutated	5	150908881	C	T	54	0.0	54	31.5	FAT2	CCDS4317.1	c.9884G>A	p.R3295Q	missense	T
456	PD5004	PMF	unmutated	20	31022662	_	delC	37	0.0	17	5.9	ASXL1	CCDS13201.1	c.2147delC	p.A716fs*9	frameshift	T
457	PD5004	PMF	unmutated	19	13054565	_	delGCAGA<42>AGGAG	37	0.0	50	20.0	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M C
458	PD5005	PMF	unmutated	16	30393447	G	A	23	0.0	10	60.0	SEPT1	CCDS10678.1	c.148C>T	p.L50F	missense	T
459	PD5005	PMF	unmutated	11	124007722	C	T	72	1.4	64	53.1	VWA5A	CCDS8444.1	c.1626C>T	p.N542N	silent	T
460	PD5005	PMF	unmutated	22	30163410	C	T	46	0.0	74	51.4	UQCR10	CCDS46680.1	c.23C>T	p.S8L	missense	T
461	PD5005	PMF	unmutated	3	128344793	T	C	53	1.9	49	51.0	RPN1	CCDS3051.1	c.1199A>G	p.Y400C	missense	T
462	PD5005	PMF	unmutated	18	32374119	T	A	77	1.3	70	50.0	DTNA	CCDS42425.1	c.267T>A	p.F89L	missense	T
463	PD5005	PMF	unmutated	14	63784482	C	T	62	0.0	36	47.2	GPHB5	ENST00000314140	c.82G>A	p.G28R	missense	T
464	PD5005	PMF	unmutated	20	9561377	G	A	166	1.8	106	46.2	PAK7	CCDS13107.1	c.405C>T	p.S135S	silent	T
465	PD5005	PMF	unmutated	14	32034248	C	T	109	1.8	26	42.3	NUBPL	CCDS41940.1	c.285C>T	p.N95N	silent	T
466	PD5005	PMF	unmutated	4	106196347	_	delT	238	3.8	123	42.3	TET2	CCDS47120.1	c.4680delT	p.S1561fs*10	frameshift	T C

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
467	PD5005	PMF	unmutated	19	39234790	C	T	48	2.1	37	40.5	CAPN12	CCDS12519.1	c.16G>A	p.G6R	missense	T
468	PD5005	PMF	unmutated	6	107390423	G	A	17	0.0	23	39.1	BEND3	CCDS34507.1	c.1972C>T	p.R658C	missense	T
469	PD5005	PMF	unmutated	10	26856423	G	A	19	0.0	28	35.7	APBB1IP	CCDS31167.1	Non-coding	r.2462g>a	UTR	T
470	PD5005	PMF	unmutated	7	99711538	G	T	52	0.0	49	10.2	TAF6	CCDS5686.1	c.196C>A	p.L66I	missense	T
471	PD5005	PMF	unmutated	19	13054575	_	delAAGGA<24>AGAGG	6	0.0	15	20.0	CALR	CCDS12288.1	c.1102_1135del34	p.K368fs*51	frameshift	M T C
472	PD5008	PMF	JAK2	9	5073770	G	T	131	0.0	80	90.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
473	PD5008	PMF	JAK2	1	240656623	C	T	25	0.0	38	57.9	GREM2	CCDS31070.1	c.153G>A	p.E51E	silent	T
474	PD5008	PMF	JAK2	X	53565939	T	C	17	0.0	14	57.1	HUWE1	CCDS35301.1	c.11735A>G	p.K3912R	missense	T
475	PD5008	PMF	JAK2	17	51900732	C	T	103	1.0	104	47.1	KIF2B	CCDS32685.1	c.338C>T	p.T113M	missense	T
476	PD5008	PMF	JAK2	2	113785532	T	G	69	0.0	49	46.9	IL1F8	CCDS2110.1	c.422A>C	p.K141T	missense	T
477	PD5008	PMF	JAK2	15	56207723	T	C	106	0.0	61	45.9	NEDD4	CCDS10156.1	c.1307A>G	p.N436S	missense	T
478	PD5008	PMF	JAK2	X	153594943	G	T	39	0.0	62	45.2	FLNA	CCDS48194.1	c.1052C>A	p.T351K	missense	T
479	PD5008	PMF	JAK2	11	26569053	C	T	193	0.5	122	45.1	ANO3	CCDS31447.1	c.1245C>T	p.C415C	silent	T
480	PD5008	PMF	JAK2	6	117861907	G	A	103	1.0	95	43.2	DCBLD1	CCDS34522.1	c.1178G>A	p.R393Q	missense	T
481	PD5008	PMF	JAK2	3	183029486	G	A	90	0.0	65	38.5	MCF2L2	CCDS3243.1	c.828C>T	p.P276P	silent	T
482	PD5008	PMF	JAK2	19	9071537	C	T	98	0.0	77	37.7	MUC16	ENST00000397910	c.15909G>A	p.Q5303Q	silent	T
483	PD5008	PMF	JAK2	7	141884901	A	G	53	0.0	48	37.5	ENSG00000204987	ENST00000477922	c.3853A>G	p.I1285V	missense	T
484	PD5008	PMF	JAK2	1	43680991	G	C	183	0.0	159	35.2	ENSG00000164012	ENST00000372492	c.103G>C	p.W35C	missense	T
485	PD5008	PMF	JAK2	4	154624391	A	G	70	0.0	47	27.7	TLR2	CCDS3784.1	c.332A>G	p.Y111C	missense	T
486	PD5008	PMF	JAK2	22	17956638	A	C	114	0.0	104	26.9	CECR2	ENST00000262608	c.12A>C	p.E4D	missense	T
487	PD5008	PMF	JAK2	14	50472497	G	T	156	0.6	150	26.0	C14orf182	CCDS41949.1	c.21C>A	p.T7T	silent	T
488	PD5008	PMF	JAK2	1	198665855	A	G	109	0.0	87	25.3	PTPRC	CCDS1397.1	c.109A>G	p.K37E	missense	T
489	PD5008	PMF	JAK2	7	131194320	G	A	54	0.0	81	24.7	PODXL	CCDS34755.1	c.827C>T	p.T276I	missense	T
490	PD5008	PMF	JAK2	12	118298169	T	C	17	0.0	20	20.0	KSR2	ENST00000339824	c.248A>G	p.N83S	missense	T
491	PD5008	PMF	JAK2	9	111617084	C	T	61	0.0	62	16.1	ACTL7B	CCDS6771.1	c.1127G>A	p.R376K	missense	T
492	PD5008	PMF	JAK2	11	134126530	G	A	48	0.0	66	15.2	ACAD8	CCDS8498.1	c.198G>A	p.E66E	silent	T
493	PD5008	PMF	JAK2	7	143826291	T	A	82	1.2	56	12.5	OR2A14	CCDS43672.1	c.86T>A	p.L29H	missense	T
494	PD5008	PMF	JAK2	4	106164862	indel	delCT	29	0.0	27	3.7	TET2	CCDS47120.1	c.3730_3731delCT	p.Y1245fs*22	frameshift	T
495	PD5014	PMF	unmutated	12	56087082	G	A	12	0.0	47	42.6	ITGA7	CCDS8888.1	c.2555C>T	p.S852L	missense	T
496	PD5014	PMF	unmutated	1	233190215	T	G	68	1.5	87	27.6	PCNXL2	CCDS44335.1	c.4150A>C	p.N1384H	missense	T
497	PD5014	PMF	unmutated	19	13054565	_	delGCAGA<42>AGGAG	18	0.0	22	36.4	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M T C
498	PD5027	PMF	JAK2	X	106066631	A	T	64	1.6	47	100.0	TBC1D8B	CCDS14522.1	c.762A>T	p.R254S	missense	T
499	PD5027	PMF	JAK2	3	88040034	_	delC	135	1.0	128	72.0	HTR1F	CCDS2920.1	c.135delC	p.I45fs*6	frameshift	T
500	PD5027	PMF	JAK2	11	7723322	C	T	22	0.0	27	55.6	OVCH2	ENST00000331572	c.500G>A	p.R167Q	missense	T
501	PD5027	PMF	JAK2	9	115421926	G	A	28	0.0	11	54.6	KIAA1958	CCDS35108.1	c.1728G>A	p.A576A	silent	T
502	PD5027	PMF	JAK2	14	64408426	C	T	55	0.0	101	50.5	SYNE2	CCDS9761.2	c.155C>T	p.S52L	missense	T
503	PD5027	PMF	JAK2	1	152280503	C	T	62	1.6	301	49.2	FLG	CCDS30860.1	c.6859G>A	p.E2287K	missense	T
504	PD5027	PMF	JAK2	5	153406840	C	T	41	0.0	49	49.0	FAM114A2	CCDS4323.1	c.685G>A	p.V229M	missense	T
505	PD5027	PMF	JAK2	14	62547699	G	A	38	0.0	66	48.5	SYT16	CCDS45121.1	c.1141G>A	p.D381N	missense	T
506	PD5027	PMF	JAK2	22	29095854	T	C	157	1.3	155	47.7	CHEK2	CCDS33629.1	c.1109A>G	p.Y370C	missense	T
507	PD5027	PMF	JAK2	21	31066267	G	A	69	0.0	76	47.4	GRIK1	CCDS42913.1	c.234C>T	p.T78T	silent	T
508	PD5027	PMF	JAK2	5	64510649	C	T	116	0.0	66	47.0	ADAMTS6	CCDS3983.2	c.2547G>A	p.W849*	nonsense	T
509	PD5027	PMF	JAK2	18	56001092	G	A	88	0.0	114	46.5	NEDD4L	CCDS45872.1	c.1033G>A	p.D345N	missense	T
510	PD5027	PMF	JAK2	16	61854953	G	A	60	0.0	39	46.2	CDH8	CCDS10802.1	c.900C>T	p.A300A	silent	T
511	PD5027	PMF	JAK2	12	111159955	T	C	41	0.0	49	44.9	PPP1CC	CCDS9150.1	c.867A>G	p.L289L	silent	T
512	PD5027	PMF	JAK2	2	71366958	T	C	120	0.0	129	44.2	MPHOSPH10	CCDS1916.1	c.1274T>C	p.L425P	missense	T
513	PD5027	PMF	JAK2	2	71366959	G	T	119	0.0	125	44.0	MPHOSPH10	CCDS1916.1	c.1275G>T	p.L425L	silent	T
514	PD5027	PMF	JAK2	2	71366960	G	T	119	0.0	125	44.0	MPHOSPH10	CCDS1916.1	c.1276G>T	p.E426*	nonsense	T
515	PD5027	PMF	JAK2	9	5073770	G	T	112	0.9	94	43.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
516	PD5027	PMF	JAK2	21	44514777	T	G	44	0.0	53	43.4	U2AF1	CCDS13694.1	c.470A>C	p.Q157P	missense	T
517	PD5027	PMF	JAK2	6	31896514	C	T	37	0.0	33	42.4	C2	CCDS4728.1	c.262C>T	p.R88C	missense	T
518	PD5027	PMF	JAK2	8	55539545	A	C	158	0.0	203	42.4	RP1	CCDS6160.1	c.3103A>C	p.N1035H	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
519	PD5027	PMF	JAK2	13	24861007	G	A	23	0.0	48	41.7	SPATA13	CCDS9305.1	c.711G>A	p.K237K	silent	T
520	PD5027	PMF	JAK2	2	160964243	G	T	70	1.4	68	39.7	ITGB6	CCDS2212.1	c.2215C>A	p.R739S	missense	T
521	PD5027	PMF	JAK2	3	186002449	C	T	63	1.6	76	38.2	DGKG	CCDS3274.1	c.594G>A	p.A198A	silent	T
522	PD5027	PMF	JAK2	20	31022937	_	delC	44	0.0	69	31.0	ASXL1	CCDS13201.1	c.2422delC	p.P808fs*10	frameshift	T
523	PD5029	PPV-MF	JAK2	9	5073770	G	T	134	0.0	135	47.4	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
524	PD5029	PPV-MF	JAK2	2	121995385	C	T	13	0.0	18	27.8	TFCP2L1	CCDS2134.1	c.902G>A	p.G301D	missense	T
525	PD5029	PPV-MF	JAK2	6	117631363	G	A	133	0.8	121	25.6	ROS1	CCDS5116.1	c.6315C>T	p.L2105L	silent	T
526	PD5029	PPV-MF	JAK2	6	159653545	G	A	17	0.0	40	25.0	FNDC1	CCDS47512.1	c.2001G>A	p.R667R	silent	T
527	PD5029	PPV-MF	JAK2	7	31016113	G	T	12	0.0	21	23.8	GHRHR	CCDS5432.1	c.1044G>T	p.L348L	silent	T
528	PD5029	PPV-MF	JAK2	12	7586183	T	C	162	0.6	234	22.2	CD163L1	CCDS8577.1	c.232A>G	p.N78D	missense	T
529	PD5029	PPV-MF	JAK2	8	134472081	C	T	31	0.0	157	19.8	ST3GAL1	CCDS6373.1	c.949G>A	p.D317N	missense	T
530	PD5029	PPV-MF	JAK2	12	11461420	G	A	110	0.0	181	18.8	PRB4	CCDS8641.1	c.497C>T	p.P166L	missense	T
531	PD5029	PPV-MF	JAK2	8	67808529	A	G	64	0.0	85	17.7	C8orf45	CCDS6197.2	c.1404A>G	p.S468S	silent	T
532	PD5029	PPV-MF	JAK2	2	197530377	G	A	46	0.0	150	15.3	CCDC150	CCDS46478.1	c.732G>A	p.M244I	missense	T
533	PD5029	PPV-MF	JAK2	1	10363911	G	A	99	0.0	62	11.3	KIF1B	CCDS112.1	c.2668G>A	p.V890I	missense	T
534	PD5029	PPV-MF	JAK2	1	92647821	_	delAC	131	1.0	72	11.1	KIAA1107	CCDS44172.1	c.3267_3268delAC	p.Q1090fs*17	frameshift	T
535	PD5111	PV	JAK2	9	5073770	G	T	260	2.6	228	44.3	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
536	PD5115	PV	JAK2	9	5073770	G	T	162	4.3	86	74.4	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
537	PD5115	PV	JAK2	17	39296629	G	A	172	1.2	155	41.9	KRTAP4-6	ENST00000345847	c.111C>T	p.R37R	silent	M
538	PD5115	PV	JAK2	5	115351332	G	T	83	0.0	56	39.3	LVRN	CCDS4124.1	c.2626G>T	p.E876*	nonsense	M
539	PD5115	PV	JAK2	2	1312324	C	T	146	0.7	132	37.9	SNTG2	CCDS46220.1	c.1343C>T	p.A448V	missense	M
540	PD5115	PV	JAK2	11	101818851	C	T	76	2.6	35	37.1	KIAA1377	CCDS31658.1	c.484C>T	p.R162C	missense	M
541	PD5115	PV	JAK2	8	56436264	C	T	101	3.0	79	35.4	XKR4	CCDS34893.1	c.1431C>T	p.L477L	silent	M
542	PD5115	PV	JAK2	16	67942955	G	A	37	5.4	43	34.9	PSKH1	CCDS10851.1	c.303G>A	p.K101K	silent	M
543	PD5115	PV	JAK2	16	21261645	C	A	104	5.8	103	30.1	ANKS4B	CCDS42130.1	c.758C>A	p.S253*	nonsense	M
544	PD5115	PV	JAK2	5	175392025	G	A	86	1.2	111	27.0	THOC3	CCDS4397.1	c.558C>T	p.N186N	silent	M
545	PD5115	PV	JAK2	X	13780563	T	A	121	0.0	65	9.2	OFD1	CCDS14157.1	c.2488T>A	p.S830T	missense	M
546	PD5115	PV	JAK2	2	231326080	G	C	86	0.0	99	5.1	SP100	CCDS42832.1	c.924G>C	p.E308D	missense	M
547	PD5117	PV	JAK2	1	196748430	A	C	90	4.4	78	20.5	CFHR3	CCDS30958.1	c.197A>C	p.Y66S	missense	T
548	PD5117	PV	JAK2	6	31632125	T	C	63	0.0	39	10.3	BAT4	CCDS4711.1	c.131A>G	p.Y44C	missense	T
549	PD5117	PV	JAK2	12	57457968	T	C	231	1.7	220	7.3	TMEM194A	CCDS44927.1	c.640A>G	p.K214E	missense	T
550	PD5117	PV	JAK2	6	49483372	G	T	294	2.4	239	6.7	GLYATL3	CCDS47440.1	c.85G>T	p.G29*	nonsense	T
551	PD5117	PV	JAK2	9	5073770	G	T	181	1.1	164	5.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
552	PD5120	PV	JAK2	9	5073770	G	T	212	9.4	259	95.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
553	PD5120	PV	JAK2	4	41984532	T	C	91	6.6	177	48.0	DCAF4L1	CCDS33978.1	c.723T>C	p.N241N	silent	T
554	PD5120	PV	JAK2	6	42633250	A	G	305	2.3	266	47.7	UBR2	CCDS4870.1	c.3802A>G	p.N1268D	missense	T
555	PD5120	PV	JAK2	12	71092090	T	A	300	2.3	143	44.1	PTPRR	CCDS8998.1	c.1234A>T	p.I412F	missense	T
556	PD5120	PV	JAK2	1	74945966	G	A	81	2.5	152	42.1	LRRC53	ENST00000416014	c.775C>T	p.Q259*	nonsense	T
557	PD5120	PV	JAK2	6	41705484	G	A	50	0.0	93	36.6	PGC	CCDS4859.1	c.998C>T	p.S333F	missense	T
558	PD5120	PV	JAK2	5	82360885	A	C	278	3.2	467	31.1	TMEM167A	ENST00000502346	c.55T>G	p.C19G	missense	T
559	PD5120	PV	JAK2	12	120146005	G	A	107	1.9	199	16.6	CIT	CCDS9192.1	c.5064C>T	p.C1688C	silent	T
560	PD5120	PV	JAK2	4	123274060	A	G	227	0.9	247	16.2	KIAA1109	CCDS43267.1	c.13851A>G	p.L4617L	silent	T
561	PD5120	PV	JAK2	5	140772789	G	T	110	0.0	225	16.0	PCDHGA12	CCDS47291.1	c.409G>T	p.D137Y	missense	T
562	PD5120	PV	JAK2	5	16703177	T	C	311	0.0	351	11.1	MYO10	ENST00000427430	c.2400A>G	p.K800K	silent	T
563	PD5120	PV	JAK2	X	133559280	G	A	114	3.5	139	10.8	PHF6	CCDS14639.1	c.1018G>A	p.E340K	missense	T
564	PD5120	PV	JAK2	14	65068268	C	T	30	0.0	78	10.3	ENSG00000238163	ENST00000428403	c.96G>A	p.E32E	silent	T
565	PD5120	PV	JAK2	6	30919290	C	A	161	0.0	193	9.8	DPCR1	ENST00000462446	c.3049C>A	p.P1017T	missense	T
566	PD5120	PV	JAK2	X	37526605	G	T	67	0.0	96	9.4	LANCL3	CCDS14240.1	c.966G>T	p.R322R	silent	T
567	PD5120	PV	JAK2	1	29323832	G	A	260	0.8	243	8.2	EPB41	CCDS332.1	c.159+1G>A	p.?	essential splice	T
568	PD5120	PV	JAK2	5	32089484	C	T	62	0.0	150	8.0	PDZD2	CCDS34137.1	c.5930C>T	p.T1977M	missense	T
569	PD5120	PV	JAK2	14	96924440	C	T	65	1.5	148	5.4	AK7	CCDS9945.1	c.1248C>T	p.N416N	silent	T
570	PD5120	PV	JAK2	9	93641130	C	G	173	0.0	268	4.9	SYK	CCDS6688.1	c.1476C>G	p.H492Q	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
571	PD5120	PV	JAK2	8	39468078	T	A	196	0.5	171	4.7	ADAM18	CCDS6113.1	c.375T>A	p.Y125*	nonsense	T
572	PD5121	PV	JAK2	9	5073770	G	T	160	4.3	130	16.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
573	PD5122	PV	JAK2	9	5073770	G	T	131	0.0	216	24.5	JAK2	CCDS6457.1	c.901G>T	p.G301W	missense	T
574	PD5122	PV	JAK2	3	194373671	C	T	92	4.4	191	13.6	LSG1	CCDS33922.1	c.960G>A	p.T320T	silent	T
575	PD5122	PV	JAK2	9	136083552	C	T	34	0.0	61	11.5	OBP2B	CCDS6961.1	c.245G>A	p.R82Q	missense	T
576	PD5122	PV	JAK2	15	62945497	G	T	70	0.0	115	7.0	TLN2	CCDS32261.1	c.501G>T	p.L167L	silent	T
577	PD5122	PV	JAK2	19	59028367	C	T	19	0.0	180	6.1	ZBTB45	CCDS12984.1	c.674G>A	p.G225D	missense	T
578	PD5125	PV	JAK2	9	5070036	_	delATGAAG	107	0.0	49	10.2	JAK2	CCDS6457.1	c.1625_1630delATGAAG	p.E543D544delED	inframe	T
579	PD5125	PV	JAK2	3	39104548	A	G	192	1.6	148	10.1	WDR48	CCDS33738.1	c.56A>G	p.Y19C	missense	T
580	PD5125	PV	JAK2	15	54006699	G	C	97	0.0	71	8.5	WDR72	CCDS10151.1	c.523C>G	p.L175V	missense	T
581	PD5125	PV	JAK2	2	169707629	T	C	110	0.9	117	6.0	NOSTRIN	CCDS42771.1	c.666T>C	p.L222L	silent	T
582	PD5125	PV	JAK2	2	107002777	G	A	418	0.5	454	4.4	ENSG00000169659	ENST00000324709	c.91G>A	p.A31T	missense	T
583	PD5125	PV	JAK2	9	5073770	G	T	217	0.0	162	1.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
584	PD5134	PV	JAK2	12	125478381	_	insCTG	5	0.0	12	33.3	BRI3BP	CCDS9262.1	c.45_46insCTG	p.L15_L16insL	inframe	M
585	PD5134	PV	JAK2	8	10470010	G	A	25	0.0	61	9.8	RP1L1	CCDS43708.1	c.1598C>T	p.S533L	missense	M
586	PD5134	PV	JAK2	9	5073770	G	T	146	0.0	144	2.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
587	PD5135	PV	JAK2	9	5073770	G	T	158	0.0	127	15.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
588	PD5135	PV	JAK2	19	9085683	C	T	251	0.0	183	9.3	MUC16	ENST00000397910	c.6132G>A	p.R2044R	silent	T
589	PD5135	PV	JAK2	2	234545779	A	C	338	0.3	275	6.9	UGT1A10	CCDS33403.1	c.611A>C	p.K204T	missense	T
590	PD5136	PV	JAK2	9	5073770	G	T	194	9.4	124	34.7	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
591	PD5136	PV	JAK2	14	55819055	G	T	55	1.8	65	16.9	FBXO34	CCDS32086.1	c.1947G>T	p.W649C	missense	T
592	PD5138	PV	JAK2	9	5073770	G	T	169	12.4	156	76.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
593	PD5138	PV	JAK2	2	234708446	C	T	37	5.4	25	48.0	HEATR7B1	ENST00000396517	c.1443C>T	p.H481H	silent	T
594	PD5138	PV	JAK2	9	87563482	G	A	31	3.2	43	37.2	NTRK2	CCDS6671.1	c.1870G>A	p.V624M	missense	T
595	PD5138	PV	JAK2	1	234509418	C	T	98	6.1	101	31.7	C1orf31	ENST00000424237	c.206C>T	p.A69V	missense	T
596	PD5141	ET	JAK2	20	42320934	A	T	85	0.0	68	25.0	MYBL2	CCDS13322.1	c.638A>T	p.Q213L	missense	T
597	PD5141	ET	JAK2	6	117995041	G	A	24	0.0	25	24.0	AL590303.1	ENST00000501802	Non-coding		RNA	T
598	PD5141	ET	JAK2	9	5073770	G	T	195	0.7	186	23.7	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
599	PD5141	ET	JAK2	2	103300686	G	T	165	0.0	145	22.8	SLC9A2	CCDS2062.1	c.1316G>T	p.G439V	missense	T
600	PD5141	ET	JAK2	10	129347805	T	A	150	0.0	129	20.2	NPS	CCDS41577.1	c.61T>A	p.F21I	missense	T
601	PD5141	ET	JAK2	12	42499828	T	A	68	0.0	117	18.0	GXYLT1	CCDS41772.1	c.656A>T	p.D219V	missense	T
602	PD5141	ET	JAK2	10	27448599	G	A	133	0.0	136	17.7	MASTL	CCDS7153.1	c.376G>A	p.G126S	missense	T
603	PD5141	ET	JAK2	21	19169436	C	G	93	0.0	84	16.7	C21orf91	CCDS42907.1	c.128-1G>C	p.?	essential splice	T
604	PD5141	ET	JAK2	12	52883847	C	T	69	0.0	64	14.1	KRT6A	CCDS41786.1	c.1083G>A	p.E361E	silent	T
605	PD5141	ET	JAK2	4	126240009	G	A	87	0.0	116	13.8	FAT4	CCDS3732.3	c.2443G>A	p.V815M	missense	T
606	PD5141	ET	JAK2	3	185146686	A	G	58	0.0	47	12.8	MAP3K13	CCDS3270.1	c.317A>G	p.D106G	missense	T
607	PD5141	ET	JAK2	21	31768892	G	T	71	0.0	66	12.1	KRTAP13-1	CCDS13590.2	c.488G>T	p.R163I	missense	T
608	PD5141	ET	JAK2	12	77423962	T	G	140	0.0	197	8.6	E2F7	CCDS9016.1	c.1533A>C	p.A511A	silent	T
609	PD5141	ET	JAK2	11	70224260	T	C	152	0.0	150	7.3	PPFIA1	CCDS31627.1	c.3509T>C	p.M1170T	missense	T
610	PD5141	ET	JAK2	6	106547343	C	A	108	0.0	83	7.2	PRDM1	CCDS5054.2	c.580C>A	p.Q194K	missense	T
611	PD5141	ET	JAK2	11	62563990	A	T	118	0.0	125	7.2	NXF1	CCDS8037.1	c.1228T>A	p.Y410N	missense	T
612	PD5141	ET	JAK2	14	94088623	G	A	246	0.0	132	6.8	KIAA1409	CCDS9911.2	c.4513G>A	p.A1505T	missense	T
613	PD5141	ET	JAK2	7	138363234	G	A	78	0.0	89	6.7	SVOPL	CCDS47721.1	c.119C>T	p.T40I	missense	T
614	PD5141	ET	JAK2	3	150396244	G	C	149	0.0	100	6.0	FAM194A	CCDS3151.2	c.1209C>G	p.N403K	missense	T
615	PD5141	ET	JAK2	4	88986569	C	T	156	0.0	184	5.4	PKD2	CCDS3627.1	c.2162C>T	p.T721I	missense	T
616	PD5141	ET	JAK2	11	56086430	A	T	153	0.0	99	5.1	OR8K3	CCDS31527.1	c.648A>T	p.L216F	missense	T
617	PD5141	ET	JAK2	11	14825567	T	G	85	0.0	102	4.9	PDE3B	CCDS7817.1	c.1493T>G	p.L498R	missense	T
618	PD5141	ET	JAK2	18	56247362	G	A	213	0.0	249	4.8	ALPK2	CCDS11966.2	c.646C>T	p.L216F	missense	T
619	PD5141	ET	JAK2	19	53304616	C	A	447	0.0	408	4.7	ZNF28	CCDS33093.1	c.323G>T	p.G108V	missense	T
620	PD5148	PV	JAK2	2	170843295	G	C	128	4.7	107	20.6	UBR3	CCDS2238.2	c.3775G>C	p.A1259P	missense	M
621	PD5148	PV	JAK2	16	10274070	C	T	91	4.4	86	17.4	GRIN2A	CCDS10539.1	c.199G>A	p.V67M	missense	M
622	PD5148	PV	JAK2	7	86811566	G	A	149	2.0	131	15.3	DMTF1	CCDS5601.1	c.733G>A	p.D245N	missense	M

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
623	PD5148	PV	JAK2	14	65240018	C	T	97	0.0	78	7.7	SPTB	CCDS32099.1	c.5098G>A	p.D1700N	missense	M
624	PD5148	PV	JAK2	9	5073770	G	T	158	0.0	164	0.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
625	PD5151	PV	JAK2	9	5073770	G	T	211	2.8	86	44.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
626	PD5151	PV	JAK2	9	34976632	G	A	75	1.3	60	20.0	KIAA1045	CCDS43796.1	c.744G>A	p.A248A	silent	T
627	PD5151	PV	JAK2	16	31419114	G	T	70	2.9	57	19.3	ITGAD	CCDS32438.1	c.886G>T	p.A296S	missense	T
628	PD5151	PV	JAK2	18	46690150	A	T	51	0.0	27	18.5	DYM	CCDS11937.1	c.1468T>A	p.S490T	missense	T
629	PD5151	PV	JAK2	4	160275139	A	G	141	2.1	86	17.4	RAPGEF2	CCDS43277.1	c.4109A>G	p.K1370R	missense	T
630	PD5151	PV	JAK2	2	171715377	C	T	83	3.6	54	16.7	GAD1	CCDS2239.1	c.1585C>T	p.P529S	missense	T
631	PD5151	PV	JAK2	11	57268286	A	C	45	2.2	53	15.1	SLC43A1	CCDS7958.1	c.431T>G	p.F144C	missense	T
632	PD5151	PV	JAK2	5	127599264	G	A	99	0.0	73	13.7	FBN2	CCDS34222.1	c.8045C>T	p.S2682L	missense	T
633	PD5151	PV	JAK2	8	133961162	G	A	177	1.7	117	12.8	TG	CCDS34944.1	c.5375G>A	p.R1792H	missense	T
634	PD5151	PV	JAK2	14	74969477	C	A	74	0.0	79	11.4	LTBP2	CCDS9831.1	c.5049G>T	p.E1683D	missense	T
635	PD5151	PV	JAK2	11	34156762	A	G	195	0.0	146	10.3	NAT10	CCDS7889.1	c.1952A>G	p.Y651C	missense	T
636	PD5151	PV	JAK2	18	59958820	A	G	275	0.4	49	10.2	KIAA1468	CCDS11979.2	c.3415A>G	p.R1139G	missense	T
637	PD5151	PV	JAK2	11	65622897	T	C	37	0.0	51	9.8	CFL1	CCDS8114.1	c.411A>G	p.A137A	silent	T
638	PD5151	PV	JAK2	2	198266822	T	A	151	0.7	99	9.1	SF3B1	CCDS33356.1	c.2110A>T	p.I704F	missense	T
639	PD5151	PV	JAK2	15	77450934	G	T	87	1.2	55	9.1	ENSG00000173517	CCDS42062.1	c.3242C>A	p.S1081Y	missense	T
640	PD5151	PV	JAK2	1	152052876	T	A	390	0.0	382	8.1	ENSG00000215854	ENST00000400998	c.183A>T	p.S61S	silent	T
641	PD5151	PV	JAK2	19	9075010	A	T	444	1.1	210	8.1	MUC16	ENST00000397910	c.12436T>A	p.S4146T	missense	T
642	PD5151	PV	JAK2	10	104140270	C	T	287	1.1	213	8.0	GBF1	CCDS7533.1	c.4997C>T	p.A1666V	missense	T
643	PD5151	PV	JAK2	5	140182165	G	A	200	1.0	193	7.8	PCDHA3	CCDS34248.1	c.1383G>A	p.T461T	silent	T
644	PD5151	PV	JAK2	15	35275201	A	G	126	0.0	68	7.4	ZNF770	CCDS10042.1	c.435T>C	p.S145S	silent	T
645	PD5151	PV	JAK2	X	37029191	T	C	234	0.4	173	5.8	FAM47C	CCDS35227.1	c.2708T>C	p.L903P	missense	T
646	PD5151	PV	JAK2	9	95064006	A	C	165	0.6	125	5.6	NOL8	CCDS47993.1	c.3002T>G	p.F1001C	missense	T
647	PD5151	PV	JAK2	X	106882519	T	C	230	0.0	247	5.3	PRPS1	CCDS14529.1	c.123-6T>C	p.?	splice	T
648	PD5151	PV	JAK2	11	66192341	G	A	158	0.0	211	5.2	NPAS4	CCDS8138.1	c.1980G>A	p.E660E	silent	T
649	PD5151	PV	JAK2	10	123970851	T	A	208	0.0	128	4.7	TACC2	CCDS7626.1	c.6911T>A	p.M2304K	missense	T
650	PD5151	PV	JAK2	15	25425657	G	C	202	0.5	435	3.7	SNORD115-6	ENST00000363942	Non-coding		RNA	T
651	PD5155	PV	JAK2	19	19349164	G	A	25	0.0	72	44.4	NCAN	CCDS12397.1	c.3353G>A	p.R1118H	missense	T
652	PD5155	PV	JAK2	16	28884923	G	A	11	0.0	16	43.8	SH2B1	ENST00000359285	c.2053G>A	p.G685S	missense	T
653	PD5155	PV	JAK2	15	42145608	C	T	10	0.0	44	38.6	SPTBN5	ENST00000320955	c.10018G>A	p.E3340K	missense	T
654	PD5155	PV	JAK2	1	152733326	G	A	52	1.9	79	35.4	KPRP	CCDS30862.1	c.1262G>A	p.R421H	missense	T
655	PD5155	PV	JAK2	10	104160201	G	A	11	0.0	29	31.0	NFKB2	CCDS41564.1	c.1751G>A	p.S584N	missense	T
656	PD5155	PV	JAK2	14	60591026	A	T	140	0.0	236	29.7	C14orf135	ENST00000406854	c.2137A>T	p.R713*	nonsense	T
657	PD5155	PV	JAK2	1	109714510	G	A	47	2.1	53	28.3	KIAA1324	CCDS794.1	c.490G>A	p.D164N	missense	T
658	PD5155	PV	JAK2	9	5073770	G	T	183	0.5	191	27.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
659	PD5155	PV	JAK2	6	22570031	G	A	20	0.0	34	23.5	HDGFL1	CCDS34347.1	c.227G>A	p.S76N	missense	T
660	PD5155	PV	JAK2	2	70486691	A	G	98	0.0	264	21.6	PCYOX1	CCDS1902.1	c.312A>G	p.K104K	silent	T
661	PD5155	PV	JAK2	1	210015695	G	A	68	0.0	118	21.2	C1orf107	CCDS1493.1	c.1571G>A	p.W524*	nonsense	T
662	PD5155	PV	JAK2	2	25463568	A	G	51	0.0	88	19.3	DNMT3A	CCDS33157.1	c.2114T>C	p.I705T	missense	T C
663	PD5155	PV	JAK2	12	117768456	G	A	49	0.0	53	18.9	NOS1	CCDS41842.1	c.419C>T	p.P140L	missense	T
664	PD5155	PV	JAK2	12	110383092	G	C	13	0.0	27	18.5	GIT2	CCDS9138.1	c.1704C>G	p.S568S	silent	T
665	PD5155	PV	JAK2	5	160067574	G	T	129	0.0	207	15.9	ATP10B	CCDS43394.1	c.894C>A	p.N298K	missense	T
666	PD5155	PV	JAK2	12	39740311	A	G	87	0.0	74	14.9	KIF21A	ENST00000395670	c.1674-5T>C	p.?	splice	T
667	PD5155	PV	JAK2	5	178987137	A	T	13	0.0	34	14.7	RUFY1	CCDS4445.2	c.422A>T	p.D141V	missense	T
668	PD5155	PV	JAK2	16	20376784	C	T	172	0.0	135	13.3	PDILT	CCDS10584.1	c.1195G>A	p.V399I	missense	T
669	PD5155	PV	JAK2	16	89764457	C	T	12	0.0	30	13.3	SPATA2L	CCDS10985.1	c.560G>A	p.R187H	missense	M
670	PD5155	PV	JAK2	17	48649313	G	A	259	0.0	129	13.2	CACNA1G	CCDS45730.1	c.661G>A	p.V221I	missense	T
671	PD5155	PV	JAK2	18	12115525	A	C	132	0.0	152	9.9	ANKRD62	ENST00000314074	c.1190A>C	p.E397A	missense	T
672	PD5155	PV	JAK2	9	33319053	C	T	52	0.0	81	8.6	NFX1	CCDS6538.1	c.1834C>T	p.R612W	missense	T
673	PD5155	PV	JAK2	X	26212643	G	T	62	0.0	49	8.2	MAGEB6	CCDS14217.1	c.680G>T	p.R227I	missense	T
674	PD5155	PV	JAK2	7	13940470	G	A	87	0.0	106	7.6	ETV1	ENST00000405358	c.1153-6C>T	p.?	splice	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
675	PD5155	PV	JAK2	9	128064434	C	T	122	0.0	155	7.1	GAPVD1	CCDS35138.1	c.358C>T	p.L120F	missense	T
676	PD5155	PV	JAK2	3	39431020	C	G	132	0.0	109	6.4	SLC25A38	CCDS2685.1	c.104C>G	p.S35C	missense	T
677	PD5155	PV	JAK2	11	14316397	C	T	129	0.8	172	6.4	RRAS2	CCDS7814.1	c.208G>A	p.A70T	missense	T
678	PD5155	PV	JAK2	15	99670279	G	A	58	0.0	169	5.9	SYNM	ENST00000336292	c.1711G>A	p.V571M	missense	T
679	PD5155	PV	JAK2	6	56485273	T	A	92	0.0	141	5.7	DST	CCDS4959.1	c.3559A>T	p.I1187L	missense	T
680	PD5155	PV	JAK2	11	108236087	G	A	66	0.0	144	5.6	ATM	CCDS31669.1	c.9023G>A	p.R3008H	missense	T
681	PD5155	PV	JAK2	1	158631121	C	T	166	0.0	209	4.8	SPTA1	CCDS41423.1	c.2543G>A	p.R848H	missense	T
682	PD5155	PV	JAK2	5	16091158	T	A	69	0.0	132	4.6	MARCH11	CCDS47192.1	c.726A>T	p.K242N	missense	T
683	PD5158	PV	JAK2	2	192250642	A	G	364	8.0	443	46.1	MYO1B	CCDS46477.1	c.1386A>G	p.E462E	silent	T
684	PD5158	PV	JAK2	11	26581277	G	T	236	5.1	222	43.7	ANO3	CCDS31447.1	c.1398G>T	p.L466F	missense	T
685	PD5158	PV	JAK2	4	106196702	_	insT	148	13.6	311	41.8	TET2	CCDS47120.1	c.5035_5036insT	p.Y1679fs*8	frameshift	T C
686	PD5158	PV	JAK2	6	132910335	G	C	80	2.5	68	33.8	TAAR5	CCDS5156.1	c.491C>G	p.A164G	missense	T
687	PD5158	PV	JAK2	8	72975724	T	A	74	1.4	86	19.8	TRPA1	CCDS34908.1	c.635A>T	p.E212V	missense	T
688	PD5158	PV	JAK2	9	5073770	G	T	158	3.8	188	18.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
689	PD5158	PV	JAK2	7	120906394	C	T	296	3.4	146	15.8	C7orf58	CCDS34739.1	c.2424C>T	p.N808N	silent	T
690	PD5158	PV	JAK2	7	139164929	G	T	60	1.7	56	14.3	KLRG2	CCDS5854.1	c.822C>A	p.V274V	silent	T
691	PD5158	PV	JAK2	2	11741065	C	G	104	3.9	153	13.1	GREB1	CCDS42655.1	c.2473C>G	p.Q825E	missense	T
692	PD5158	PV	JAK2	12	9847365	C	T	217	4.2	273	11.4	CLEC2D	CCDS31741.1	c.553C>T	p.P185S	missense	T
693	PD5158	PV	JAK2	14	23313685	G	A	244	1.6	237	7.6	MMP14	CCDS9577.1	c.1117G>A	p.E373K	missense	T
694	PD5158	PV	JAK2	5	31526394	G	C	75	1.3	128	6.3	RNASEN	CCDS47195.1	c.646C>G	p.P216A	missense	T
695	PD5161	PV	JAK2	9	5073770	G	T	137	8.8	129	62.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
696	PD5161	PV	JAK2	4	8589022	C	T	43	0.0	57	36.8	GPR78	CCDS3403.1	c.1024C>T	p.R342C	missense	M
697	PD5161	PV	JAK2	9	99246896	A	T	80	6.3	72	33.3	HABP4	CCDS6719.1	c.980A>T	p.K327M	missense	M
698	PD5161	PV	JAK2	1	158261175	C	T	249	1.6	91	30.8	CD1C	CCDS1175.1	c.313C>T	p.Q105*	nonsense	M
699	PD5161	PV	JAK2	15	75890833	T	C	92	0.0	114	30.7	SNUPN	CCDS10281.1	c.949A>G	p.K317E	missense	M
700	PD5161	PV	JAK2	10	104572845	T	C	48	0.0	69	30.4	C10orf26	CCDS44473.1	c.849T>C	p.H283H	silent	M
701	PD5161	PV	JAK2	2	26653653	G	A	280	0.0	265	30.2	C2orf39	CCDS1723.1	c.679-1G>A	p.?	essential splice	M
702	PD5161	PV	JAK2	6	123319065	G	A	121	0.8	94	29.8	CLVS2	CCDS34525.1	c.143G>A	p.R48H	missense	M
703	PD5161	PV	JAK2	17	57737916	G	A	287	0.7	300	29.3	CLTC	CCDS32696.1	c.1134G>A	p.S378S	silent	M
704	PD5161	PV	JAK2	16	53358543	A	C	152	0.0	192	23.4	CHD9	CCDS45485.1	c.8382A>C	p.P2794P	silent	M
705	PD5163	PV	JAK2	12	57111539	G	A	21	0.0	28	21.4	NACA2	CCDS44925.1	c.3775C>T	p.P1259S	missense	M
706	PD5163	PV	JAK2	9	5073770	G	T	135	0.7	147	6.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	C
707	PD5165	PV	JAK2	9	5073770	G	T	153	2.6	93	49.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
708	PD5165	PV	JAK2	4	31144181	C	T	295	0.7	97	33.0	PCDH7	CCDS43220.1	c.3313C>T	p.P1105S	missense	T
709	PD5165	PV	JAK2	1	90478737	G	A	83	4.8	80	27.5	ZNF326	CCDS727.1	c.867G>A	p.R289R	silent	T
710	PD5165	PV	JAK2	19	8841739	C	T	78	0.0	56	26.8	OR2Z1	CCDS32895.1	c.349C>T	p.L117F	missense	T
711	PD5165	PV	JAK2	2	232327250	G	C	49	2.0	65	24.6	NCL	ENST00000392033	c.170C>G	p.T57S	missense	T
712	PD5165	PV	JAK2	16	2296890	G	A	23	4.4	64	20.3	DCI	CCDS10464.1	c.264C>T	p.D88D	silent	T
713	PD5165	PV	JAK2	19	57325206	G	A	114	2.6	65	18.5	PEG3AS	CCDS12948.1	c.4604C>T	p.P1535L	missense	T
714	PD5165	PV	JAK2	19	16957798	G	A	51	0.0	40	12.5	SIN3B	CCDS32946.1	c.600G>A	p.T200T	silent	T
715	PD5165	PV	JAK2	3	176767859	C	T	188	0.0	125	9.6	TBL1XR1	CCDS46961.1	c.628G>A	p.V210I	missense	T
716	PD5182	PV	JAK2	2	25458575	C	T	58	1.7	104	6.7	DNMT3A	CCDS33157.1	c.2597+1G>A	p.?	essential splice	T C
717	PD5182	PV	JAK2	9	5073770	G	T	156	0.0	223	3.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
718	PD5192	PV	JAK2	6	89975484	C	G	121	5.0	151	21.2	GABRR2	CCDS5020.2	c.812G>C	p.G271A	missense	T
719	PD5192	PV	JAK2	9	5073770	G	T	159	3.8	140	20.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
720	PD5847	PV	JAK2	9	5073770	G	T	254	14.6	234	47.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
721	PD5847	PV	JAK2	4	106155938	_	insT	176	14.8	78	25.6	TET2	CCDS47120.1	c.839_840insT	p.N281fs*0	frameshift	T
722	PD5847	PV	JAK2	5	74892823	T	C	390	8.0	290	20.7	POLK	CCDS4030.1	c.2305T>C	p.C769R	missense	T C
723	PD5847	PV	JAK2	3	171320961	C	T	54	5.6	110	18.2	PLD1	CCDS3216.1	c.3132G>A	p.L1044L	silent	T C
724	PD5847	PV	JAK2	11	64891979	G	T	88	3.4	150	12.7	MRPL49	CCDS8096.1	c.84G>T	p.Q28H	missense	T C
725	PD5847	PV	JAK2	2	25464534	T	A	11	0.0	25	12.0	DNMT3A	CCDS33157.1	c.1979A>T	p.Y660F	missense	T C
726	PD6548	ET	JAK2	4	106157755	C	T	36	5.6	34	17.7	TET2	CCDS47120.1	c.2656C>T	p.Q886*	nonsense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
727	PD6548	ET	JAK2	9	5073770	G	T	111	5.4	107	16.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
728	PD6548	ET	JAK2	5	148748248	C	G	19	0.0	44	11.4	PCYOX1L	CCDS4296.1	Non-coding	r.1578c>g	UTR	M
729	PD6549	ET	unmutated	19	13054627	_	insTTGTC	16	0.0	13	61.5	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M T C
730	PD6549	ET	unmutated	7	150498783	G	A	12	0.0	12	58.3	TMEM176A	CCDS5909.1	c.145G>A	p.G49S	missense	T
731	PD6549	ET	unmutated	5	21783610	A	C	143	0.0	188	52.1	CDH12	CCDS3890.1	c.1257-7T>G	p.?	splice	T
732	PD6549	ET	unmutated	12	97328812	A	G	172	0.6	197	48.7	NEDD1	CCDS44955.1	c.569A>G	p.D190G	missense	T
733	PD6549	ET	unmutated	15	101816741	C	T	109	0.9	118	47.5	ENSG00000131871	ENST00000398226	c.211+5G>A	p.?	essential splice	T
734	PD6549	ET	unmutated	7	143141030	G	A	143	0.0	174	47.1	TAS2R60	CCDS5885.1	c.485G>A	p.G162D	missense	T
735	PD6549	ET	unmutated	1	20671941	C	T	52	0.0	45	44.4	VWA5B1	CCDS44078.1	c.2627-8C>T	p.?	splice	T
736	PD6549	ET	unmutated	14	77880351	G	A	17	0.0	37	40.5	C14orf148	CCDS45142.1	c.275C>T	p.A92V	missense	T
737	PD6549	ET	unmutated	9	127216794	G	A	17	0.0	18	38.9	GPR144	CCDS48016.1	c.1110G>A	p.S370S	silent	T
738	PD6549	ET	unmutated	2	202939644	T	C	104	0.0	134	38.8	ENSG00000182329	ENST00000409515	c.115T>C	p.Y39H	missense	T
739	PD6549	ET	unmutated	17	26920087	G	A	81	1.2	86	38.4	SPAG5	CCDS32594.1	c.178-3C>T	p.?	splice	T
740	PD6549	ET	unmutated	8	52320920	C	T	133	0.0	148	35.8	PXDNL	CCDS47855.1	c.3264G>A	p.P1088P	silent	T
741	PD6561	ET	unmutated	19	3293325	A	T	54	0.0	64	45.3	CELF5	CCDS12106.1	c.1339A>T	p.S447C	missense	T C
742	PD6561	ET	unmutated	7	151135153	C	T	49	0.0	68	44.1	CRYGN	CCDS5926.1	c.199G>A	p.G67S	missense	T C
743	PD6561	ET	unmutated	18	8783965	G	A	191	0.0	215	43.3	KIAA0802	CCDS11841.1	c.855G>A	p.A285A	silent	T C
744	PD6561	ET	unmutated	1	66833673	_	delG	82	0.0	95	30.5	PDE4B	CCDS632.1	c.1587delG	p.K530fs*4	frameshift	T C
745	PD6561	ET	unmutated	11	55433006	G	A	97	0.0	118	17.0	OR4C6	CCDS31506.1	c.364G>A	p.V122M	missense	T C
746	PD6561	ET	unmutated	3	141891234	T	C	112	0.0	111	15.3	GK5	ENST00000392993	c.1208A>G	p.Y403C	missense	T C
747	PD6561	ET	unmutated	19	13054627	_	insTTGTC	17	0.0	14	21.4	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M T C
748	PD6563	ET	unmutated	19	13054565	_	delGCAGA<42>AGGAG	21	9.5	17	35.3	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M C
749	PD6563	ET	unmutated	12	22065906	C	T	83	1.2	108	45.4	ABCC9	CCDS8693.1	c.911G>A	p.R304H	missense	T C
750	PD6563	ET	unmutated	17	47246181	G	A	36	0.0	36	44.4	B4GALNT2	CCDS11544.1	c.1414G>A	p.V472M	missense	T C
751	PD6563	ET	unmutated	6	144508545	G	A	27	0.0	16	43.8	STX11	CCDS5205.1	c.781G>A	p.A261T	missense	M C
752	PD6563	ET	unmutated	6	43591675	A	G	243	1.2	228	39.5	GTPBP2	CCDS4903.1	c.1231T>C	p.F411L	missense	T C
753	PD6563	ET	unmutated	12	123061513	G	A	150	0.0	283	36.4	KNTC1	CCDS45002.1	c.2658G>A	p.A886A	silent	T C
754	PD6563	ET	unmutated	10	410346	G	A	106	0.9	89	34.8	DIP2C	CCDS7054.1	c.2445C>T	p.A815A	silent	M C
755	PD6563	ET	unmutated	11	128851419	C	T	96	1.0	106	33.0	ARHGAP32	CCDS44769.1	c.1630G>A	p.A544T	missense	T C
756	PD6563	ET	unmutated	21	34707883	A	G	109	0.0	123	28.5	IFNAR1	CCDS13624.1	c.130A>G	p.N44D	missense	T C
757	PD6566	ET	unmutated	19	13054627	_	insTTGTC	4	0.0	16	37.5	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M C
758	PD6566	ET	unmutated	4	141543733	C	T	63	6.4	59	55.9	TBC1D9	CCDS47136.1	c.3417G>A	p.S1139S	silent	T
759	PD6566	ET	unmutated	15	26825472	C	T	49	0.0	54	40.7	GABRB3	CCDS10019.1	c.676G>A	p.A226T	missense	T
760	PD6568	PV	JAK2	9	5073770	G	T	158	0.0	145	31.7	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
761	PD6568	PV	JAK2	2	27844314	C	T	61	0.0	76	31.6	ZNF512	CCDS1758.1	c.1690C>T	p.R564*	nonsense	T
762	PD6568	PV	JAK2	1	151508754	G	A	34	2.9	128	20.3	CGN	CCDS999.1	c.3239G>A	p.R1080Q	missense	T
763	PD6568	PV	JAK2	2	71896831	C	T	138	0.0	135	19.3	DYSF	CCDS46328.1	c.5739C>T	p.P1913P	silent	T
764	PD6568	PV	JAK2	19	16035618	C	T	51	0.0	96	16.7	ENSG00000171903	CCDS12337.1	c.600G>A	p.L200L	silent	T
765	PD6568	PV	JAK2	1	179086534	_	delA	84	0.0	142	14.8	ABL2	CCDS41441.1	c.1233delT	p.P412fs*51	frameshift	T
766	PD6568	PV	JAK2	14	20443698	T	A	142	0.0	113	14.2	OR4K15	CCDS32026.1	c.21T>A	p.D7E	missense	T
767	PD6568	PV	JAK2	12	54686911	_	delG	34	0.0	105	10.5	NFE2	CCDS8876.1	c.369delC	p.L124fs*0	frameshift	T
768	PD6568	PV	JAK2	1	229750071	C	T	152	0.0	85	9.4	TAF5L	ENST00000366674	c.159G>A	p.S53S	silent	T
769	PD6568	PV	JAK2	9	139350516	G	A	40	0.0	75	8.0	SEC16A	ENST00000313050	c.5119C>T	p.R1707W	missense	T
770	PD6568	PV	JAK2	9	91940839	_	insT	134	0.0	111	7.2	SECISBP2	CCDS6683.1	c.460_461insT	p.Y154fs*2	frameshift	T
771	PD6568	PV	JAK2	2	179436414	T	C	157	0.0	261	6.1	TTN	ENST00000356127	c.66735A>G	p.P22245P	silent	T
772	PD6568	PV	JAK2	11	71331462	C	T	92	0.0	318	4.7	OR7E87P	ENST00000454336	c.248C>T	p.T83I	missense	T
773	PD6573	ET	unmutated	19	13054627	_	insTTGTC	3	33.3	20	30.0	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M T C
774	PD6573	ET	unmutated	X	70461098	T	C	38	2.6	34	32.4	ZMYM3	CCDS14409.1	c.3899A>G	p.Y1300C	missense	T
775	PD6573	ET	unmutated	1	16455924	C	A	37	0.0	69	30.4	EPHA2	CCDS169.1	c.2825+5G>T	p.?	essential splice	T
776	PD6573	ET	unmutated	4	7398081	C	T	89	3.4	142	23.9	SORCS2	CCDS47008.1	c.547C>T	p.R183W	missense	T
777	PD6573	ET	unmutated	2	32475594	G	T	38	0.0	87	21.8	NLRC4	CCDS33174.1	c.1339C>A	p.Q447K	missense	T
778	PD6573	ET	unmutated	11	128840264	C	T	33	0.0	91	7.7	ARHGAP32	CCDS44769.1	c.4802G>A	p.R1601Q	missense	T

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779	PD6573	ET	unmutated	16	89016802	A	T	99	2.0	222	5.9	ENSG00000205018	ENST00000378347	c.276A>T	p.S92S	silent	T
780	PD6586	ET	JAK2	9	5073770	G	T	114	0.0	177	9.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
781	PD6586	ET	JAK2	4	96761711	C	T	69	0.0	130	8.5	PDHA2	CCDS3644.1	c.410C>T	p.T137M	missense	T
782	PD6591	ET	JAK2	9	5073770	G	T	114	3.5	42	14.3	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
783	PD6591	ET	JAK2	14	86089606	G	A	128	3.1	269	11.5	FLRT2	CCDS9877.1	c.1748G>A	p.R583Q	missense	M
784	PD6593	ET	MPL	1	43815009	G	T	51	0.0	125	4.0	MPL	CCDS483.1	c.1544G>T	p.W515L	missense	T
785	PD6594	ET	JAK2	4	122825591	G	T	85	2.4	68	33.8	TRPC3	CCDS47130.1	c.2139C>A	p.H713Q	missense	T
786	PD6594	ET	JAK2	8	80965612	A	C	169	4.1	130	26.9	TPD52	CCDS34912.1	c.309T>G	p.H103Q	missense	T
787	PD6594	ET	JAK2	16	12371903	G	A	179	1.7	155	25.2	SNX29	CCDS45416.1	c.627+10G>A	p.?	splice	T
788	PD6594	ET	JAK2	13	53418723	G	A	31	0.0	65	12.3	PCDH8	CCDS9438.1	c.3185C>T	p.P1062L	missense	T
789	PD6594	ET	JAK2	17	26108178	G	A	93	0.0	52	9.6	NOS2	CCDS11223.1	c.748C>T	p.R250W	missense	T
790	PD6594	ET	JAK2	11	107948968	G	T	500	0.0	273	8.8	CUL5	CCDS31668.1	c.1178+1G>T	p.?	essential splice	T
791	PD6594	ET	JAK2	16	5134829	C	T	67	0.0	73	8.2	ALG1	CCDS10528.1	c.1342C>T	p.R448*	nonsense	T
792	PD6594	ET	JAK2	18	3193959	G	C	80	1.3	75	8.0	MYOM1	CCDS45824.1	c.291-3C>G	p.?	splice	T
793	PD6594	ET	JAK2	1	172062005	T	C	215	0.0	146	7.5	DNM3	CCDS44276.1	c.1535T>C	p.V512A	missense	T
794	PD6594	ET	JAK2	9	5073770	G	T	183	0.5	120	4.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
795	PD6597	ET	unmutated	19	13054627	_	insTTGTC	2	0.0	20	45.0	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M T C
796	PD6597	ET	unmutated	11	57077693	A	T	15	0.0	102	51.0	TNKS1BP1	CCDS7951.1	c.2492T>A	p.L831H	missense	T
797	PD6597	ET	unmutated	6	149916245	T	G	75	5.3	150	44.7	KATNA1	CCDS5217.1	c.1403A>C	p.K468T	missense	T
798	PD6597	ET	unmutated	6	56473996	T	C	500	2.8	500	41.6	DST	CCDS47444.1	c.3819A>G	p.P1273P	silent	T
799	PD6597	ET	unmutated	12	31269398	G	A	78	3.9	265	12.5	ENSG00000177359	ENST00000266524	c.3900C>T	p.T1300T	silent	T
800	PD6599	ET	JAK2	19	24115392	A	G	67	6.0	61	44.3	ZNF726	ENST00000322487	c.474A>G	p.I158M	missense	M
801	PD6599	ET	JAK2	7	43496045	C	T	29	3.5	46	19.6	HECW1	CCDS5469.2	c.2650C>T	p.R884W	missense	M
802	PD6599	ET	JAK2	7	103131214	C	A	70	0.0	91	9.9	RELN	CCDS47680.1	c.9506G>T	p.C3169F	missense	M
803	PD6599	ET	JAK2	17	76046069	T	C	25	0.0	64	9.4	ENSG00000078687	CCDS45799.1	c.926T>C	p.L309P	missense	M
804	PD6599	ET	JAK2	9	5073770	G	T	90	0.0	206	1.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
805	PD6600	ET	JAK2	9	5073770	G	T	121	2.5	137	29.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
806	PD6602	ET	unmutated	14	95570029	C	T	89	1.1	154	22.1	DICER1	CCDS9931.1	c.3704G>A	p.C1235Y	missense	T
807	PD6602	ET	unmutated	16	31123561	G	C	38	0.0	19	21.1	BCKDK	CCDS10705.1	c.1214G>C	p.G405A	missense	T
808	PD6602	ET	unmutated	1	91406390	A	T	165	4.9	255	20.0	ZNF644	CCDS731.1	c.521T>A	p.V174D	missense	T
809	PD6602	ET	unmutated	5	68661333	G	A	119	0.8	158	17.1	TAF9	CCDS4002.1	c.232C>T	p.R78C	missense	T
810	PD6602	ET	unmutated	5	149277971	T	C	119	7.6	137	16.1	PDE6A	CCDS4299.1	c.1362A>G	p.V454V	silent	T
811	PD6602	ET	unmutated	12	14019063	C	T	64	1.6	50	10.0	GRIN2B	CCDS8662.1	c.80G>A	p.R27H	missense	T
812	PD6603	ET	JAK2	9	5073770	G	T	197	0.0	220	9.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
813	PD6603	ET	JAK2	10	43293968	G	A	139	0.0	155	6.5	BMS1	CCDS7199.1	c.2142G>A	p.G714G	silent	T
814	PD6603	ET	JAK2	1	36474509	A	C	134	0.0	147	5.4	EIF2C3	CCDS399.1	c.893A>C	p.Q298P	missense	T
815	PD6603	ET	JAK2	3	128292319	_	delTC	90	0.0	102	4.9	C3orf27	CCDS3050.1	c.253_254delGA	p.D85fs*0	frameshift	T
816	PD6619	ET	JAK2	9	139751004	C	A	18	0.0	52	42.3	MAMDC4	ENST00000392881	c.1946C>A	p.T649K	missense	T
817	PD6619	ET	JAK2	1	16456031	G	A	47	0.0	19	42.1	EPHA2	CCDS169.1	c.2723C>T	p.T908M	missense	T
818	PD6619	ET	JAK2	1	53377235	T	C	82	0.0	53	41.5	ECHDC2	CCDS571.1	c.277+3A>G	p.?	splice	T
819	PD6619	ET	JAK2	4	140280987	A	T	64	0.0	75	40.0	NAA15	CCDS43270.1	c.1348A>T	p.K450*	nonsense	T
820	PD6619	ET	JAK2	18	74536368	C	T	43	2.3	95	37.9	ZNF236	CCDS42447.1	c.49+6C>T	p.?	splice	T
821	PD6619	ET	JAK2	9	5073770	G	T	139	0.7	162	37.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
822	PD6619	ET	JAK2	18	74536367	G	A	44	2.3	96	36.5	ZNF236	CCDS42447.1	c.49+5G>A	p.?	essential splice	T
823	PD6619	ET	JAK2	17	17062183	A	G	33	0.0	72	34.7	MPRIP	CCDS42268.1	c.1913A>G	p.Q638R	missense	T
824	PD6619	ET	JAK2	1	204518404	G	A	98	0.0	123	34.2	MDM4	CCDS1447.1	c.1067G>A	p.G356E	missense	T
825	PD6619	ET	JAK2	20	31022675	_	delC	30	0.0	48	33.3	ASXL1	CCDS13201.1	c.2160delC	p.L721fs*4	frameshift	T
826	PD6619	ET	JAK2	1	216390859	G	A	64	0.0	58	29.3	USH2A	CCDS31025.1	c.3027C>T	p.A1009A	silent	T
827	PD6619	ET	JAK2	18	9887213	C	T	286	0.0	245	26.5	TXNDC2	CCDS42414.1	c.737C>T	p.T246I	missense	T
828	PD6619	ET	JAK2	19	51827026	A	G	24	0.0	30	16.7	IGLON5	CCDS46158.1	c.269A>G	p.N90S	missense	T
829	PD6619	ET	JAK2	8	95178104	G	A	62	0.0	71	9.9	CDH17	CCDS6260.1	c.1167C>T	p.P389P	silent	T
830	PD6619	ET	JAK2	16	4751105	G	A	85	0.0	182	7.7	ANKS3	CCDS10520.1	c.1150C>T	p.R384C	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
831	PD6619	ET	JAK2	5	90049557	C	T	144	0.0	151	7.3	GPR98	CCDS47246.1	c.11288C>T	p.S3763F	missense	T
832	PD6619	ET	JAK2	6	148848683	C	T	106	0.0	111	6.3	SASH1	CCDS5212.1	c.1379C>T	p.T460M	missense	T
833	PD6619	ET	JAK2	12	95650963	G	T	161	0.0	203	4.4	VEZT	CCDS44954.1	c.206G>T	p.S69I	missense	T
834	PD6626	ET	JAK2	9	5073770	G	T	174	1.7	164	72.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
835	PD6626	ET	JAK2	19	50958380	G	A	61	0.0	96	39.6	MYBPC2	CCDS46152.1	c.2030G>A	p.R677Q	missense	T
836	PD6626	ET	JAK2	5	169535413	C	T	35	0.0	37	37.8	FOXI1	CCDS4372.1	c.935C>T	p.T312M	missense	T
837	PD6626	ET	JAK2	6	125619850	C	T	51	0.0	59	37.3	HDDC2	CCDS43503.1	c.309+10G>A	p.?	splice	T
838	PD6626	ET	JAK2	21	35254619	G	A	50	0.0	68	36.8	ITSN1	CCDS33545.1	c.4414G>A	p.A1472T	missense	T
839	PD6626	ET	JAK2	16	67696071	C	T	10	0.0	32	31.3	PARD6A	CCDS10843.1	c.562C>T	p.R188W	missense	T
840	PD6626	ET	JAK2	6	126320661	T	C	278	0.0	122	30.3	TRMT11	CCDS35496.1	c.581T>C	p.I194T	missense	T
841	PD6626	ET	JAK2	11	55419131	C	T	381	0.3	209	29.7	OR4S2	CCDS31505.1	c.752C>T	p.P251L	missense	T
842	PD6626	ET	JAK2	3	185636158	G	A	145	0.0	144	23.6	TRA2B	CCDS33905.1	c.851C>T	p.S284L	missense	T
843	PD6626	ET	JAK2	6	56819403	G	A	12	0.0	26	23.1	DST	ENST00000281662	c.2C>T	p.P1L	missense	T
844	PD6626	ET	JAK2	19	49253923	G	A	51	0.0	135	14.1	FUT1	CCDS12733.1	c.616C>T	p.R206C	missense	T
845	PD6626	ET	JAK2	12	41949546	G	T	268	0.0	149	9.4	PDZRN4	CCDS8739.1	c.575G>T	p.G192V	missense	T
846	PD6626	ET	JAK2	12	41949545	G	A	275	0.0	152	8.6	PDZRN4	CCDS8739.1	c.574G>A	p.G192R	missense	T
847	PD6626	ET	JAK2	1	53504619	G	A	200	0.0	97	7.2	SCP2	CCDS572.1	c.1369G>A	p.G457S	missense	T
848	PD6626	ET	JAK2	19	9058926	G	A	157	0.0	248	4.8	MUC16	ENST00000331986	c.15419C>T	p.T5140M	missense	T
849	PD6626	ET	JAK2	11	10876254	G	C	191	0.0	166	4.2	ZBED5	ENST00000432999	c.239C>G	p.S80C	missense	T
850	PD6628	ET	unmutated	19	13054627	_	insTTGTC	13	15.4	10	40.0	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M T C
851	PD6628	ET	unmutated	7	148507424	C	G	60	5.0	39	18.0	EZH2	CCDS5891.1	c.2029+1G>C	p.?	essential splice	M
852	PD6628	ET	unmutated	7	142989778	C	A	60	0.0	42	11.9	CASP2	CCDS5879.1	c.462C>A	p.L154L	silent	M
853	PD6629	ET	JAK2	12	58112814	G	A	297	5.4	191	48.2	OS9	CCDS31843.1	c.1449G>A	p.E483E	silent	T C
854	PD6629	ET	JAK2	11	129993661	G	A	103	1.0	107	46.7	APLP2	CCDS8486.1	c.1077G>A	p.V359V	silent	T C
855	PD6629	ET	JAK2	9	5073770	G	T	123	0.8	208	44.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
856	PD6629	ET	JAK2	15	42564273	T	C	91	3.3	109	42.2	TMEM87A	CCDS32205.1	c.193A>G	p.I65V	missense	T C
857	PD6629	ET	JAK2	2	198511265	G	A	84	3.6	239	37.2	RFTN2	CCDS2323.1	c.265C>T	p.R89*	nonsense	T C
858	PD6629	ET	JAK2	2	25457242	C	T	56	0.0	67	34.3	DNMT3A	CCDS33157.1	c.2645G>A	p.R882H	missense	T C
859	PD6629	ET	JAK2	4	106157326	_	insA	70	0.0	97	11.3	TET2	CCDS47120.1	c.2227_2228insA	p.Q744fs*10	frameshift	T C
860	PD6637	ET	MPL	18	32374073	C	T	54	7.4	37	29.7	DTNA	CCDS42425.1	c.221C>T	p.T74I	missense	M
861	PD6637	ET	MPL	19	34819061	C	A	18	0.0	36	16.7	KIAA0355	CCDS12436.1	c.1104+5C>A	p.?	essential splice	M
862	PD6637	ET	MPL	X	53590752	C	A	19	0.0	32	15.6	HUWE1	CCDS35301.1	c.7060G>T	p.E2354*	nonsense	M
863	PD6637	ET	MPL	17	71166765	C	A	18	0.0	42	14.3	SSTR2	CCDS11691.1	Non-coding	r.1667c>a	UTR	M
864	PD6637	ET	MPL	12	73046869	C	A	66	0.0	54	13.0	TRHDE	CCDS9004.1	c.2782C>A	p.R928R	silent	M
865	PD6637	ET	MPL	7	99697209	C	A	28	0.0	64	10.9	MCM7	CCDS5683.1	c.276+3G>T	p.?	splice	M
866	PD6637	ET	MPL	18	23713967	C	A	75	0.0	65	10.8	PSMA8	CCDS32808.1	c.38C>A	p.S13Y	missense	M
867	PD6637	ET	MPL	6	2890534	C	A	66	0.0	80	10.0	SERPINB9	CCDS4478.1	c.994G>T	p.A332S	missense	M
868	PD6637	ET	MPL	13	36765945	C	A	135	0.0	123	7.3	SOHLH2	CCDS9355.1	c.517G>T	p.D173Y	missense	M
869	PD6637	ET	MPL	5	63894228	C	A	180	0.0	161	6.8	RGS7BP	CCDS34170.1	c.648C>A	p.L216L	silent	M
870	PD6637	ET	MPL	1	43815009	G	T	27	0.0	42	4.8	MPL	CCDS483.1	c.1544G>T	p.W515L	missense	T
871	PD6642	ET	JAK2	9	5073770	G	T	95	10.5	143	25.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
872	PD6642	ET	JAK2	2	80136837	C	T	29	0.0	46	13.0	CTNNA2	CCDS42703.2	c.970C>T	p.R324*	nonsense	T
873	PD6642	ET	JAK2	4	2175745	C	T	140	0.7	149	10.1	POLN	CCDS3360.1	c.1311G>A	p.V437V	silent	T
874	PD6642	ET	JAK2	X	134428048	T	A	122	0.0	211	7.6	ZNF75D	CCDS14648.1	c.19A>T	p.N7Y	missense	T
875	PD6642	ET	JAK2	1	176853475	C	T	43	0.0	113	7.1	ASTN1	CCDS1319.1	c.3226G>A	p.E1076K	missense	T
876	PD6644	ET	JAK2	17	42090488	G	A	25	0.0	27	14.8	TMEM101	CCDS11474.1	c.354C>T	p.G118G	silent	T
877	PD6644	ET	JAK2	9	5073770	G	T	237	0.0	126	13.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
878	PD6646	ET	JAK2	22	38212690	G	A	22	0.0	38	60.5	GCAT	CCDS13957.1	c.1225G>A	p.V409M	missense	T C
879	PD6646	ET	JAK2	X	153209030	G	A	19	0.0	37	59.5	RENBP	CCDS14738.2	c.430C>T	p.L144L	silent	T C
880	PD6646	ET	JAK2	5	139744100	T	A	20	0.0	26	53.9	SLC4A9	CCDS47278.1	c.1541T>A	p.I514N	missense	T C
881	PD6646	ET	JAK2	1	40767003	C	G	23	0.0	23	52.2	COL9A2	CCDS450.1	c.1921G>C	p.G641R	missense	T C
882	PD6646	ET	JAK2	2	21362519	A	G	58	1.7	89	48.3	ENSG00000218819	ENST00000405799	c.2180A>G	p.N727S	missense	T C

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
883	PD6646	ET	JAK2	2	219691670	C	T	32	0.0	40	45.0	PRKAG3	CCDS2424.1	c.1149G>A	p.L383L	silent	T C
884	PD6646	ET	JAK2	11	9519238	T	A	73	1.4	62	43.6	ZNF143	CCDS7799.2	c.858T>A	p.S286R	missense	T C
885	PD6646	ET	JAK2	5	38957800	A	G	118	0.0	117	42.7	RICTOR	CCDS34148.1	c.2453T>C	p.L818P	missense	T C
886	PD6646	ET	JAK2	1	20829715	C	T	24	0.0	58	41.4	MUL1	CCDS208.1	c.208G>A	p.G70R	missense	T C
887	PD6646	ET	JAK2	X	119388450	_	delA	138	0.0	146	41.1	ZBTB33	CCDS14596.1	c.1180delA	p.I394fs*0	frameshift	T
888	PD6646	ET	JAK2	3	39227114	G	A	35	0.0	36	38.9	XIRP1	CCDS2683.1	c.3823C>T	p.R1275C	missense	T C
889	PD6646	ET	JAK2	5	154394878	G	A	161	0.0	173	36.4	KIF4B	CCDS47324.1	c.1459G>A	p.A487T	missense	T C
890	PD6646	ET	JAK2	9	5073770	G	T	114	0.0	139	36.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
891	PD6646	ET	JAK2	2	238742968	G	A	93	0.0	88	34.1	RBM44	CCDS46554.1	c.3083G>A	p.G1028D	missense	T C
892	PD6646	ET	JAK2	15	72144573	A	T	49	0.0	73	32.9	MYO9A	CCDS10239.1	c.6375T>A	p.Y2125*	nonsense	T C
893	PD6646	ET	JAK2	2	25469624	_	delTCCCC<13>CCACC	52	0.0	47	17.0	DNMT3A	CCDS33157.1	c.1123-1_1144del23	p.?	frameshift	T C
894	PD6646	ET	JAK2	15	26812851	G	A	87	0.0	65	7.7	GABRB3	CCDS10019.1	c.712C>T	p.R238W	missense	T C
895	PD6646	ET	JAK2	3	38888444	A	G	72	1.4	159	6.3	SCN11A	CCDS33737.1	c.5117T>C	p.M1706T	missense	T C
896	PD6646	ET	JAK2	11	119148981	T	A	91	0.0	123	4.9	CBL	CCDS8418.1	c.1201T>A	p.C401S	missense	T C
897	PD6647	ET	JAK2	3	48999113	G	A	85	1.2	54	48.2	ARIH2	CCDS2780.1	c.323+1G>A	p.?	essential splice	M
898	PD6647	ET	JAK2	15	100672279	A	T	23	0.0	23	39.1	ADAMTS17	CCDS10383.1	c.1654T>A	p.W552R	missense	M
899	PD6647	ET	JAK2	9	123476543	_	delCGGCGG	2	0.0	16	25.0	MEGF9	CCDS48010.1	c.65_70delCCGCCG	p.A22_A23delAA	inframe	M
900	PD6647	ET	JAK2	1	157668287	T	G	77	2.6	91	23.1	FCRL3	CCDS1167.1	c.185A>C	p.E62A	missense	M
901	PD6647	ET	JAK2	9	5073770	G	T	165	0.0	119	10.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
902	PD6647	ET	JAK2	8	25287389	G	A	216	0.0	192	9.9	KCTD9	CCDS6048.1	c.1154C>T	p.S385L	missense	M
903	PD6647	ET	JAK2	4	148582072	C	A	148	0.0	74	9.5	PRMT10	CCDS3771.1	c.1071G>T	p.M357I	missense	M
904	PD6647	ET	JAK2	1	213046037	C	A	81	0.0	91	8.8	FLVCR1	CCDS1510.1	c.901C>A	p.R301R	silent	M
905	PD6647	ET	JAK2	3	57454685	C	T	140	0.0	134	8.2	DNAH12	CCDS46857.1	c.2127G>A	p.L709L	silent	M
906	PD6647	ET	JAK2	14	75514023	G	A	101	1.0	144	7.6	MLH3	CCDS32123.1	c.2336C>T	p.T779I	missense	M
907	PD6647	ET	JAK2	1	203786225	_	delT	106	0.0	80	7.5	ZC3H11A	CCDS30978.1	c.27delT	p.F12fs*20	frameshift	M
908	PD6648	ET	JAK2	15	41961052	G	A	143	0.7	136	19.9	MGA	ENST00000219905	Non-coding	r.141g>a	start gained	T
909	PD6648	ET	JAK2	9	5073770	G	T	163	0.6	187	18.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
910	PD6648	ET	JAK2	20	34022174	G	A	47	0.0	66	18.2	GDF5	CCDS13254.1	c.1039C>T	p.R347C	missense	T
911	PD6648	ET	JAK2	12	21954026	C	G	56	0.0	102	15.7	ABCC9	CCDS8693.1	c.4602G>C	p.L1534F	missense	T
912	PD6648	ET	JAK2	15	60740283	T	C	298	1.3	168	15.5	NARG2	CCDS10176.1	c.2181A>G	p.E727E	silent	T
913	PD6648	ET	JAK2	2	197674092	C	T	122	0.0	111	13.5	C2orf66	CCDS2317.1	c.20G>A	p.R7H	missense	T
914	PD6648	ET	JAK2	X	31152268	C	T	52	3.9	71	12.7	DMD	CCDS14233.1	c.10965G>A	p.G3655G	silent	T
915	PD6650	ET	JAK2	8	113236797	G	A	13	0.0	8	50.0	CSMD3	CCDS6315.1	Non-coding	r.11572c>u	UTR	M
916	PD6650	ET	JAK2	8	73850113	G	C	86	0.0	75	42.7	KCNB2	CCDS6209.1	c.2523G>C	p.G841G	silent	M
917	PD6650	ET	JAK2	1	185153997	G	A	186	0.5	161	41.0	C1orf26	CCDS1367.1	c.1363G>A	p.D455N	missense	M
918	PD6650	ET	JAK2	7	150555973	G	A	17	0.0	40	40.0	ABP1	CCDS43679.1	c.1693G>A	p.A565T	missense	M
919	PD6650	ET	JAK2	7	51111193	C	T	63	0.0	93	38.7	COBL	CCDS34637.1	c.1293G>A	p.K431K	silent	M
920	PD6650	ET	JAK2	17	10409167	G	A	71	0.0	73	38.4	MYH1	CCDS11155.1	c.2136C>T	p.G712G	silent	M
921	PD6650	ET	JAK2	2	77745645	G	A	255	0.8	201	37.3	LRRTM4	CCDS46346.1	c.1350C>T	p.Y450Y	silent	M
922	PD6650	ET	JAK2	6	79713494	C	A	96	0.0	51	35.3	PHIP	CCDS4987.1	c.1606G>T	p.G536*	nonsense	M
923	PD6650	ET	JAK2	8	133146533	C	T	98	0.0	61	31.2	KCNQ3	CCDS34943.1	c.1799+4G>A	p.?	splice	M
924	PD6650	ET	JAK2	9	5073770	G	T	208	0.0	99	27.3	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
925	PD6650	ET	JAK2	1	235657991	C	T	176	0.6	146	26.7	B3GALNT2	CCDS1606.1	c.260G>A	p.R87H	missense	M
926	PD6650	ET	JAK2	X	53239664	G	T	33	0.0	29	20.7	KDM5C	CCDS14351.1	c.1678C>A	p.Q560K	missense	M
927	PD6650	ET	JAK2	6	111912621	_	insC	54	0.0	78	9.0	TRAF3IP2	CCDS5092.1	c.695_696insG	p.Y232fs*1	frameshift	M
928	PD6650	ET	JAK2	3	38407165	C	G	12	0.0	50	8.0	XYLB	CCDS2678.1	c.445C>G	p.L149V	missense	M
929	PD6653	ET	JAK2	9	5073770	G	T	153	3.3	121	30.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
930	PD6653	ET	JAK2	20	31389221	C	T	38	0.0	45	11.1	DNMT3B	CCDS13205.1	c.2134C>T	p.R712W	missense	T
931	PD6653	ET	JAK2	15	101432816	C	A	30	0.0	68	10.3	ALDH1A3	CCDS10389.1	c.447C>A	p.D149E	missense	T
932	PD6654	ET	JAK2	5	140711266	G	T	81	0.0	96	44.8	PCDHGA1	CCDS34256.1	c.1015G>T	p.D339Y	missense	T
933	PD6654	ET	JAK2	17	7577121	G	C	42	2.4	33	42.4	TP53	CCDS11118.1	c.817C>G	p.R273G	missense	T
934	PD6654	ET	JAK2	11	34979073	C	T	68	0.0	98	37.8	PDHX	CCDS7896.1	c.485C>T	p.S162L	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
935	PD6654	ET	JAK2	9	5073770	G	T	116	0.8	150	36.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
936	PD6654	ET	JAK2	2	152563482	A	T	242	0.8	115	35.7	NEB	CCDS46424.1	c.1065T>A	p.N355K	missense	T
937	PD6654	ET	JAK2	3	96706460	C	T	350	0.3	500	35.6	EPHA6	CCDS46876.1	c.737C>T	p.A246V	missense	T
938	PD6654	ET	JAK2	10	118896116	C	A	32	0.0	32	34.4	VAX1	CCDS44483.1	c.296G>T	p.R99L	missense	T
939	PD6654	ET	JAK2	7	94043209	G	A	43	0.0	43	32.6	COL1A2	CCDS34682.1	c.1615G>A	p.V539I	missense	T
940	PD6654	ET	JAK2	20	40044124	T	A	29	0.0	43	25.6	CHD6	CCDS13317.1	c.6641A>T	p.E2214V	missense	T
941	PD6654	ET	JAK2	2	234294602	G	T	28	0.0	76	21.1	DGKD	ENST00000447484	c.12G>T	p.S4S	silent	T
942	PD6654	ET	JAK2	21	10916395	T	C	415	0.0	369	14.4	TPTE	CCDS13560.2	c.1251A>G	p.K417K	silent	T
943	PD6654	ET	JAK2	3	52536076	G	A	21	0.0	43	14.0	STAB1	CCDS33768.1	c.386G>A	p.G129D	missense	T
944	PD6654	ET	JAK2	7	126882891	A	G	56	0.0	87	13.8	GRM8	CCDS5794.1	c.368T>C	p.L123S	missense	T
945	PD6654	ET	JAK2	2	207452084	A	G	234	0.0	143	6.3	ADAM23	CCDS2369.1	c.1773A>G	p.A591A	silent	T
946	PD6654	ET	JAK2	20	31022284	_	insT	99	0.0	133	3.8	ASXL1	CCDS13201.1	c.1769_1770insT	p.Y591fs*28	frameshift	M
947	PD6656	ET	JAK2	9	5073770	G	T	102	7.8	49	24.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
948	PD6656	ET	JAK2	5	9629623	T	C	60	3.3	74	14.9	TAS2R1	CCDS3876.1	c.522A>G	p.I174M	missense	T
949	PD6661	ET	JAK2	2	25457242	C	T	30	0.0	74	41.9	DNMT3A	CCDS33157.1	c.2645G>A	p.R882H	missense	T C
950	PD6661	ET	JAK2	11	61295466	G	A	23	0.0	124	30.7	SYT7	CCDS31577.1	c.543C>T	p.P181P	silent	T
951	PD6661	ET	JAK2	18	32418142	T	C	47	4.3	95	30.5	DTNA	CCDS42425.1	c.1172+7T>C	p.?	splice	T
952	PD6661	ET	JAK2	2	17884509	T	A	51	5.9	192	29.2	SMC6	CCDS1690.1	c.2057A>T	p.Q686L	missense	T
953	PD6661	ET	JAK2	19	9087213	A	T	124	0.0	500	26.0	MUC16	ENST00000397910	c.4602T>A	p.T1534T	silent	T
954	PD6661	ET	JAK2	9	5073770	G	T	68	0.0	246	23.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
955	PD6661	ET	JAK2	19	50204031	G	A	69	1.5	182	23.1	CPT1C	CCDS12779.1	c.372G>A	p.V124V	silent	T
956	PD6661	ET	JAK2	18	34853037	C	T	13	0.0	42	21.4	CELF4	CCDS32818.1	c.891G>A	p.Q297Q	silent	T
957	PD6661	ET	JAK2	5	16818188	G	A	19	0.0	96	20.8	MYO10	ENST00000427430	c.209C>T	p.S70F	missense	T
958	PD6661	ET	JAK2	5	175717147	G	A	39	0.0	157	18.5	C5orf25	ENST00000443967	c.563G>A	p.C188Y	missense	T
959	PD6661	ET	JAK2	9	104189866	C	T	26	0.0	141	14.9	ALDOB	CCDS6756.1	c.438G>A	p.G146G	silent	T
960	PD6661	ET	JAK2	17	40996845	A	T	73	0.0	298	8.4	AOC2	CCDS11443.1	c.202A>T	p.M68L	missense	T
961	PD6661	ET	JAK2	1	150234627	_	delC	63	0.0	212	8.0	CA14	CCDS947.1	c.327delC	p.L110fs*38	frameshift	T
962	PD7268	ET	JAK2	9	5073770	G	T	131	0.8	118	5.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
963	PD7269	ET	JAK2	9	5073770	G	T	103	3.9	113	17.7	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
964	PD7269	ET	JAK2	8	20022628	T	C	36	0.0	70	8.6	SLC18A1	CCDS6013.1	c.859-10A>G	p.?	splice	T
965	PD7270	ET	JAK2	19	8651554	G	T	52	1.9	98	31.6	ADAMTS10	CCDS12206.1	c.2291C>A	p.P764H	missense	T
966	PD7270	ET	JAK2	9	5073770	G	T	153	0.0	236	27.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
967	PD7271	ET	JAK2	9	5073770	G	T	147	0.0	166	4.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
968	PD7272	ET	JAK2	9	5073770	G	T	145	0.7	201	9.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
969	PD7272	ET	JAK2	16	112802	C	T	41	0.0	126	7.1	RHBDF1	CCDS32344.1	c.766G>A	p.D256N	missense	T
970	PD7272	ET	JAK2	4	139980385	T	C	137	0.7	173	6.4	ELF2	CCDS3744.1	c.1498A>G	p.M500V	missense	T
971	PD7272	ET	JAK2	1	32498884	T	A	99	0.0	124	4.8	KHDRBS1	CCDS350.1	c.720T>A	p.A240A	silent	T
972	PD7273	ET	JAK2	1	1635577	A	G	12	0.0	23	26.1	CDK11B	CCDS44042.1	c.1695T>C	p.D565D	silent	M
973	PD7273	ET	JAK2	1	1635574	A	G	12	0.0	22	22.7	CDK11B	CCDS44042.1	c.1698T>C	p.F566F	silent	M
974	PD7273	ET	JAK2	9	5073770	G	T	93	1.1	125	8.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
975	PD7274	ET	JAK2	3	75787646	_	delAA	10	1.0	13	46.2	ZNF717	CCDS46871.1	c.1127_1128delTT	p.F376fs*3	frameshift	M
976	PD7274	ET	JAK2	22	17680532	G	A	18	0.0	28	39.3	CECR1	CCDS13743.1	Non-coding	r.142c>u	start gained	M
977	PD7274	ET	JAK2	1	204124264	G	A	50	2.0	34	38.2	REN	CCDS30981.1	c.1101C>T	p.H367H	silent	M
978	PD7274	ET	JAK2	16	50104098	G	A	39	2.6	37	37.8	HEATR3	CCDS10739.1	c.409G>A	p.G137R	missense	M
979	PD7274	ET	JAK2	X	54013571	C	T	73	0.0	42	35.7	PHF8	CCDS14355.1	c.1935G>A	p.A645A	silent	M
980	PD7274	ET	JAK2	7	103008053	C	T	104	0.0	52	28.9	PSMC2	CCDS5731.1	c.1038C>T	p.P346P	silent	M
981	PD7274	ET	JAK2	6	157714009	A	C	105	0.0	108	28.7	C6orf35	CCDS43519.1	c.350T>G	p.M117R	missense	M
982	PD7274	ET	JAK2	9	37732388	C	T	64	0.0	70	28.6	FRMPD1	CCDS6612.1	c.946C>T	p.R316W	missense	M
983	PD7274	ET	JAK2	14	69925152	T	C	128	0.0	174	25.9	SLC39A9	CCDS9795.1	c.766T>C	p.Y256H	missense	M
984	PD7274	ET	JAK2	X	105281066	T	A	117	0.0	75	24.0	SERPINA7	CCDS14518.1	Non-coding	r.320a>u	UTR	M
985	PD7274	ET	JAK2	22	17443634	C	T	18	0.0	25	24.0	GAB4	CCDS42976.1	c.1714G>A	p.A572T	missense	M
986	PD7274	ET	JAK2	22	32113131	_	delG	248	2.0	193	19.7	C22orf30	CCDS13900.2	c.694delC	p.Q232fs*3	frameshift	M

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
987	PD7274	ET	JAK2	X	100129303	C	T	27	0.0	26	19.2	NOX1	CCDS14474.1	Non-coding	r.32g>a	UTR	M
988	PD7274	ET	JAK2	9	5073770	G	T	151	0.7	146	19.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
989	PD7274	ET	JAK2	7	94540180	T	C	37	0.0	48	12.5	PPP1R9A	CCDS34683.1	c.755T>C	p.L252P	missense	M
990	PD7274	ET	JAK2	4	106157212	C	T	112	0.0	89	7.9	TET2	CCDS47120.1	c.2113C>T	p.Q705*	nonsense	M
991	PD7275	PET_PV	JAK2	9	5073770	G	T	172	0.0	88	39.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
992	PD7275	PET_PV	JAK2	X	107436891	C	T	251	0.8	203	18.7	COL4A6	CCDS14541.1	c.1042G>A	p.V348I	missense	M
993	PD7275	PET_PV	JAK2	15	102300097	_	ins_delGC	16	0.0	58	8.6	AC107977.4	ENSG00000237157	Non-coding		RNA	M
994	PD7275	PET_PV	JAK2	1	10355727	A	G	134	0.0	84	7.1	KIF1B	CCDS111.1	c.1542A>G	p.Q514Q	silent	M
995	PD7276	PET-PV	JAK2	1	11848080	G	A	63	6.4	12	50.0	C1orf167	ENST00000433342	c.3896G>A	p.S1299N	missense	M
996	PD7276	PET-PV	JAK2	9	5073770	G	T	139	0.7	127	15.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
997	PD7276	PET-PV	JAK2	11	48510924	G	T	159	0.6	134	14.2	OR4A47	CCDS31490.1	c.580G>T	p.G194C	missense	M
998	PD7278	PET-PV	JAK2	9	5073770	G	T	128	2.3	119	23.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
999	PD7278	PET-PV	JAK2	11	92714701	C	T	253	0.0	108	17.6	MTNR1B	CCDS8290.1	c.312C>T	p.D104D	silent	M
1000	PD7278	PET-PV	JAK2	12	112371734	C	T	68	0.0	62	16.1	TMEM116	CCDS9157.1	c.413G>A	p.R138Q	missense	M
1001	PD7278	PET-PV	JAK2	20	31395706	A	G	104	1.0	55	12.7	DNMT3B	CCDS13205.1	c.2559A>G	p.E853E	silent	M
1002	PD7278	PET-PV	JAK2	7	128632094	C	T	108	0.0	129	12.4	TNPO3	CCDS5809.1	c.1317G>A	p.A439A	silent	M
1003	PD7278	PET-PV	JAK2	X	77224411	G	T	211	0.5	110	10.0	PGAM4	CCDS35338.1	c.725C>A	p.A242D	missense	M
1004	PD7278	PET-PV	JAK2	1	45798841	C	T	37	2.7	70	10.0	MUTYH	CCDS44129.1	c.390G>A	p.V130V	silent	M
1005	PD7279	PET-PV	JAK2	9	5073770	G	T	210	1.9	108	68.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1006	PD7279	PET-PV	JAK2	14	91942197	A	T	167	1.8	30	50.0	SMEK1	CCDS9895.1	c.1224T>A	p.D408E	missense	T
1007	PD7279	PET-PV	JAK2	X	12939245	C	T	139	1.4	92	38.0	TLR8	CCDS14152.1	c.2086C>T	p.R696C	missense	T
1008	PD7279	PET-PV	JAK2	16	89825010	T	G	174	5.2	74	37.8	FANCA	CCDS32515.1	c.2956A>C	p.N986H	missense	T
1009	PD7279	PET-PV	JAK2	5	167674668	C	T	63	1.6	85	37.7	ODZ2	ENST00000388903	c.6223C>T	p.R2075C	missense	T
1010	PD7279	PET-PV	JAK2	18	2891624	G	A	109	4.6	67	32.8	EMILIN2	CCDS11828.1	c.1499G>A	p.R500H	missense	T
1011	PD7279	PET-PV	JAK2	8	91033209	A	G	62	3.2	61	32.8	DECR1	CCDS6250.1	c.490A>G	p.I164V	missense	T
1012	PD7279	PET-PV	JAK2	12	53216930	C	T	95	2.1	49	32.7	KRT79	CCDS8839.1	c.1237G>A	p.V413M	missense	T
1013	PD7279	PET-PV	JAK2	X	118724851	C	T	194	1.0	143	32.2	NKRF	CCDS35375.1	c.537G>A	p.A179A	silent	T
1014	PD7279	PET-PV	JAK2	11	118014557	C	T	32	6.3	23	30.4	SCN4B	CCDS8389.1	c.454G>A	p.V152I	missense	T
1015	PD7279	PET-PV	JAK2	1	209963860	C	G	69	0.0	55	29.1	IRF6	CCDS1492.1	c.1040G>C	p.C347S	missense	T
1016	PD7279	PET-PV	JAK2	4	106196641	G	T	142	0.0	116	27.6	TET2	CCDS47120.1	c.4974G>T	p.L1658L	silent	T
1017	PD7280	PV	JAK2	9	5073770	G	T	170	3.5	126	25.4	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1018	PD7280	PV	JAK2	19	17411794	G	A	70	0.0	78	14.1	ABHD8	CCDS12355.1	c.632C>T	p.A211V	missense	M
1019	PD7280	PV	JAK2	6	29599327	G	A	59	0.0	72	8.3	GABBR1	CCDS4663.1	c.135C>T	p.G45G	silent	M
1020	PD7281	PV	JAK2	9	5073770	G	T	104	0.0	123	99.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1021	PD7281	PV	JAK2	5	154275775	C	T	53	0.0	73	54.8	GEMIN5	CCDS4330.1	c.3474G>A	p.E1158E	silent	M
1022	PD7281	PV	JAK2	20	45801371	G	T	51	0.0	56	53.6	EYA2	CCDS13403.1	c.1054G>T	p.A352S	missense	M
1023	PD7281	PV	JAK2	19	20727943	G	C	171	0.0	171	52.1	ZNF737	ENST00000344519	c.1063C>G	p.L355V	missense	M
1024	PD7281	PV	JAK2	2	220409562	C	T	42	0.0	50	52.0	TMEM198	CCDS33385.1	c.113C>T	p.P38L	missense	T
1025	PD7281	PV	JAK2	3	195498647	T	A	37	0.0	36	50.0	MUC4	CCDS3310.1	c.410A>T	p.Y137F	missense	M
1026	PD7281	PV	JAK2	2	136594655	T	C	109	0.0	102	50.0	LCT	CCDS2178.1	c.85A>G	p.I29V	missense	M
1027	PD7281	PV	JAK2	12	108589739	G	A	88	0.0	85	48.2	WSCD2	CCDS41828.1	c.130G>A	p.A44T	missense	M
1028	PD7281	PV	JAK2	17	45993985	C	T	28	0.0	25	48.0	SP2	CCDS11521.2	c.548C>T	p.P183L	missense	M
1029	PD7281	PV	JAK2	2	242524037	T	C	129	0.0	105	47.6	THAP4	CCDS2551.1	c.1718A>G	p.K573R	missense	T
1030	PD7281	PV	JAK2	6	129835661	C	T	110	0.0	118	46.6	LAMA2	CCDS5138.1	c.9132C>T	p.N3044N	silent	M
1031	PD7281	PV	JAK2	7	6150943	C	A	104	0.0	110	46.4	USP42	CCDS47535.1	c.179C>A	p.A60D	missense	M
1032	PD7281	PV	JAK2	X	100086592	G	A	102	0.0	96	44.8	CSTF2	CCDS14473.1	c.978G>A	p.P326P	silent	M
1033	PD7281	PV	JAK2	X	135443653	A	C	93	0.0	100	43.0	GPR112	CCDS35409.1	c.7184A>C	p.Y2395S	missense	M
1034	PD7281	PV	JAK2	18	44260149	C	T	59	0.0	57	42.1	ST8SIA5	CCDS11930.1	c.987G>A	p.S329S	silent	M
1035	PD7281	PV	JAK2	6	56475247	C	T	66	0.0	69	36.2	DST	CCDS47441.1	c.5162G>A	p.G1721E	missense	M
1036	PD7281	PV	JAK2	1	162381753	C	T	63	0.0	91	31.9	SH2D1B	CCDS30928.1	c.54G>A	p.L18L	silent	M
1037	PD7281	PV	JAK2	16	53404748	C	T	53	0.0	54	29.6	ENSG00000236139	ENST00000416703	c.547G>A	p.G183R	missense	M
1038	PD7281	PV	JAK2	5	884052	C	T	44	0.0	37	27.0	BRD9	CCDS34127.1	c.678+1G>A	p.?	essential splice	M

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1039	PD7281	PV	JAK2	16	1837704	G	A	61	0.0	57	22.8	NUBP2	CCDS10445.1	c.361G>A	p.V121M	missense	M
1040	PD7281	PV	JAK2	2	42553332	T	C	269	0.0	272	15.4	EML4	CCDS1807.1	c.2281T>C	p.S761P	missense	M
1041	PD7281	PV	JAK2	X	123185054	T	A	89	0.0	56	14.3	STAG2	CCDS43990.1	c.1101T>A	p.F367L	missense	M
1042	PD7281	PV	JAK2	11	113857734	C	T	43	0.0	42	14.3	HTR3A	CCDS8366.1	c.1200C>T	p.A400A	silent	M
1043	PD7281	PV	JAK2	7	129037063	G	A	31	0.0	38	13.2	AHCYL2	CCDS5812.1	c.721G>A	p.V241M	missense	M
1044	PD7281	PV	JAK2	3	57898950	G	T	79	0.0	94	12.8	SLMAP	CCDS33774.1	c.1984G>T	p.E662*	nonsense	M
1045	PD7281	PV	JAK2	19	37903967	G	A	110	0.0	99	12.1	ZNF569	CCDS12503.1	c.1593C>T	p.F531F	silent	T
1046	PD7281	PV	JAK2	5	55247394	A	C	101	0.0	81	8.6	IL6ST	CCDS3971.1	c.1738T>G	p.S580A	missense	M
1047	PD7394	PET-MF	MPL	6	135510928	G	C	120	6.7	316	26.6	MYB	CCDS47481.1	c.214-1G>C	p.?	essential splice	T
1048	PD7394	PET-MF	MPL	17	39191011	G	C	50	0.0	105	9.5	KRTAP1-3	CCDS42323.1	c.63C>G	p.G21G	silent	T
1049	PD7394	PET-MF	MPL	1	43818310	G	A	35	2.9	64	9.4	MPL	CCDS483.1	c.1775G>A	p.R592Q	missense	T
1050	PD7394	PET-MF	MPL	1	208206700	C	T	28	3.6	103	8.7	PLXNA2	CCDS31013.1	c.5019G>A	p.V1673V	silent	T
1051	PD7394	PET-MF	MPL	X	118717178	G	C	99	3.0	237	6.3	UBE2A	CCDS14580.1	c.419G>C	p.R140P	missense	T
1052	PD7395	PV	JAK2	9	5073770	G	T	159	1.3	185	46.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1053	PD7395	PV	JAK2	16	86544980	G	A	11	0.0	22	31.8	FOXF1	CCDS10957.2	c.805G>A	p.A269T	missense	T
1054	PD7395	PV	JAK2	16	2813025	A	G	472	0.9	398	24.1	SRRM2	CCDS32373.1	c.2496A>G	p.R832R	silent	T
1055	PD7395	PV	JAK2	1	207039688	T	A	221	0.5	226	21.2	IL20	CCDS1470.1	c.204T>A	p.T68T	silent	T
1056	PD7395	PV	JAK2	9	136577783	A	G	30	0.0	53	20.8	SARDH	CCDS6978.1	c.1286T>C	p.I429T	missense	T
1057	PD7395	PV	JAK2	2	73677832	A	C	122	1.6	179	18.4	ALMS1	CCDS42697.1	c.4175A>C	p.Q1392P	missense	T
1058	PD7395	PV	JAK2	16	18861780	_	insT	118	0.0	133	18.0	SMG1	CCDS45430.1	c.5061_5062insA	p.I1688fs*7	frameshift	T
1059	PD7395	PV	JAK2	X	32486696	T	A	90	1.1	84	14.3	DMD	CCDS14233.1	c.3081A>T	p.G1027G	silent	T
1060	PD7396	MPN_u	JAK2	9	5073770	G	T	185	0.0	65	23.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1061	PD7396	MPN_u	JAK2	6	39838188	C	T	105	0.0	167	9.0	DAAM2	ENST00000398904	c.946C>T	p.L316F	missense	T
1062	PD7396	MPN_u	JAK2	20	37153481	G	A	196	0.0	125	8.8	RALGAPB	CCDS13305.1	c.1680G>A	p.L560L	silent	T
1063	PD7396	MPN_u	JAK2	3	10359778	C	A	50	0.0	76	6.6	SEC13	CCDS2599.1	c.4G>T	p.V2L	missense	T
1064	PD7397	PV	JAK2	9	5073770	G	T	187	1.6	176	63.6	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1065	PD7397	PV	JAK2	9	112144048	C	T	70	0.0	65	40.0	PTPN3	CCDS6776.1	c.2548G>A	p.G850S	missense	T
1066	PD7397	PV	JAK2	17	39464493	C	T	40	2.5	53	34.0	KRTAP16-1	ENST00000391352	c.1013G>A	p.R338H	missense	T
1067	PD7397	PV	JAK2	3	1415683	G	A	164	0.6	110	33.6	CNTN6	CCDS2557.1	c.2021G>A	p.R674H	missense	T
1068	PD7397	PV	JAK2	2	219695533	G	T	89	0.0	102	28.4	PRKAG3	CCDS2424.1	c.165C>A	p.A55A	silent	T
1069	PD7397	PV	JAK2	19	49703901	G	A	49	0.0	58	24.1	TRPM4	CCDS33073.1	c.2812G>A	p.G938S	missense	T
1070	PD7397	PV	JAK2	3	54922025	T	G	198	0.5	152	21.7	CACNA2D3	ENST00000474759	c.2096T>G	p.V699G	missense	T
1071	PD7397	PV	JAK2	3	125295185	T	C	87	0.0	76	21.1	OSBPL11	CCDS3033.1	c.514A>G	p.S172G	missense	T
1072	PD7397	PV	JAK2	1	99772258	G	A	49	0.0	54	20.4	ENSG00000117600	CCDS757.1	c.1984G>A	p.E662K	missense	T
1073	PD7397	PV	JAK2	7	92098285	G	A	89	0.0	83	19.3	ERVWE1	ENST00000493463	c.1411C>T	p.L471F	missense	T
1074	PD7397	PV	JAK2	1	44159715	A	G	64	0.0	51	15.7	KDM4A	CCDS491.1	c.2426A>G	p.N809S	missense	T
1075	PD7397	PV	JAK2	4	187524079	G	A	168	0.0	214	5.6	FAT1	CCDS47177.1	c.11460C>T	p.S3820S	silent	T
1076	PD7397	PV	JAK2	9	113171090	A	T	235	0.4	332	4.5	SVEP1	CCDS48004.1	c.6790T>A	p.C2264S	missense	T
1077	PD7398	ET	JAK2	9	5073770	G	T	138	2.1	166	68.7	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	M
1078	PD7398	ET	JAK2	1	33944913	A	C	100	1.0	89	50.6	ZSCAN20	CCDS41300.1	c.24A>C	p.Q8H	missense	M
1079	PD7398	ET	JAK2	3	56115005	G	A	82	2.4	131	50.4	ERC2	CCDS46851.1	c.1481C>T	p.A494V	missense	M
1080	PD7398	ET	JAK2	1	50941357	G	A	28	0.0	28	50.0	FAF1	CCDS554.1	c.1654-6C>T	p.?	splice	M
1081	PD7398	ET	JAK2	3	124351541	C	T	115	0.0	132	47.7	KALRN	CCDS3028.1	c.360C>T	p.S120S	silent	M
1082	PD7398	ET	JAK2	15	90631934	C	T	87	5.8	77	46.8	IDH2	CCDS10359.1	c.419G>A	p.R140Q	missense	T
1083	PD7398	ET	JAK2	6	160679401	G	A	134	0.0	98	43.9	SLC22A2	CCDS5276.1	c.389C>T	p.T130M	missense	T
1084	PD7398	ET	JAK2	17	76166929	A	G	95	1.1	140	43.6	SYNGR2	CCDS11753.1	c.131A>G	p.Y44C	missense	M
1085	PD7398	ET	JAK2	2	87013080	G	T	234	1.7	228	42.1	CD8A	CCDS1992.1	c.671C>A	p.S224*	nonsense	T
1086	PD7398	ET	JAK2	10	135204920	G	A	27	0.0	49	38.8	PAOX	CCDS7682.1	c.1339G>A	p.V447M	missense	T
1087	PD7398	ET	JAK2	12	88440625	C	A	155	0.0	106	38.7	C12orf29	CCDS31866.1	c.661C>A	p.H221N	missense	M
1088	PD7398	ET	JAK2	5	140431959	C	T	83	1.2	107	37.4	PCDHB1	CCDS4243.1	c.904C>T	p.R302*	nonsense	M
1089	PD7398	ET	JAK2	14	74824762	C	T	61	0.0	81	14.8	C14orf115	CCDS9830.1	c.1276C>T	p.R426C	missense	M
1090	PD7399	PV	JAK2	9	5073770	G	T	135	7.4	198	28.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1091	PD7399	PV	JAK2	11	66190678	C	T	11	0.0	21	28.6	NPAS4	CCDS8138.1	c.783C>T	p.H261H	silent	T
1092	PD7399	PV	JAK2	13	52991208	G	A	45	4.4	59	22.0	VPS36	CCDS9434.1	c.974C>T	p.A325V	missense	T
1093	PD7399	PV	JAK2	17	76832207	G	A	62	4.8	78	21.8	USP36	CCDS32755.1	c.239C>T	p.P80L	missense	T
1094	PD7399	PV	JAK2	6	42141493	C	A	152	4.0	187	21.4	GUCA1A	CCDS4864.1	c.142C>A	p.L48M	missense	T
1095	PD7399	PV	JAK2	5	13829681	C	T	149	6.0	137	19.0	DNAH5	CCDS3882.1	c.6382G>A	p.V2128I	missense	T
1096	PD7399	PV	JAK2	6	76558241	C	T	101	6.9	120	18.3	MYO6	CCDS34487.1	c.1071C>T	p.S357S	silent	T
1097	PD7399	PV	JAK2	1	222902998	G	T	98	1.0	88	17.1	C1orf58	CCDS1534.1	c.793G>T	p.D265Y	missense	T
1098	PD7399	PV	JAK2	3	189587194	C	A	104	5.8	90	16.7	TP63	CCDS3293.1	c.1211C>A	p.P404Q	missense	T
1099	PD7399	PV	JAK2	11	4881667	C	T	274	2.9	161	15.5	OR51H1P	ENST00000322059	c.128G>A	p.G43E	missense	T
1100	PD7399	PV	JAK2	7	116937852	C	T	198	2.5	152	14.5	WNT2	CCDS5771.1	c.667G>A	p.A223T	missense	T
1101	PD7399	PV	JAK2	17	45232056	G	C	147	2.0	143	13.3	CDC27	CCDS45720.1	c.939C>G	p.T313T	silent	T
1102	PD7399	PV	JAK2	17	11833352	A	G	58	1.7	42	11.9	DNAH9	CCDS11160.1	c.12047A>G	p.N4016S	missense	T
1103	PD7399	PV	JAK2	10	105906091	G	T	73	2.7	120	11.7	C10orf79	CCDS31281.1	c.3785C>A	p.A1262D	missense	T
1104	PD7399	PV	JAK2	4	39905790	T	C	116	4.3	193	8.8	PDS5A	CCDS47045.1	c.1255A>G	p.M419V	missense	T
1105	PD7399	PV	JAK2	3	40231666	C	T	35	2.9	70	8.6	MYRIP	CCDS2689.1	c.1377C>T	p.S459S	silent	T
1106	PD7400	PV	JAK2	9	5073770	G	T	147	1.3	199	9.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1107	PD7400	PV	JAK2	1	55349366	G	A	130	0.0	169	8.9	DHCR24	CCDS600.1	c.312C>T	p.V104V	silent	T
1108	PD7400	PV	JAK2	11	6262735	G	T	209	3.4	149	8.1	CNGA4	CCDS31408.1	c.992G>T	p.R331L	missense	T
1109	PD7436	ET	JAK2	7	48411838	C	T	402	0.3	250	24.0	ABCA13	CCDS47584.1	c.10877C>T	p.T3626I	missense	T
1110	PD7436	ET	JAK2	7	43591845	G	A	221	0.0	105	21.9	HECW1	CCDS5469.2	c.4420G>A	p.V1474M	missense	T
1111	PD7436	ET	JAK2	4	56473507	A	G	47	2.1	69	21.7	NMU	CCDS3501.1	c.318T>C	p.F106F	silent	T
1112	PD7436	ET	JAK2	4	125590313	C	A	283	0.0	229	21.4	ANKRD50	CCDS34060.1	c.4119G>T	p.Q1373H	missense	T
1113	PD7436	ET	JAK2	22	32828527	C	G	43	0.0	80	21.3	BPIL2	CCDS13906.1	c.982G>C	p.A328P	missense	T
1114	PD7436	ET	JAK2	15	45848048	C	T	302	1.0	265	21.1	C15orf21	ENST00000396645	c.398C>T	p.A133V	missense	T
1115	PD7436	ET	JAK2	16	20548587	G	A	500	0.0	418	21.1	ACSM2B	CCDS10586.1	c.1727C>T	p.A576V	missense	T
1116	PD7436	ET	JAK2	13	114542721	C	T	27	0.0	38	21.1	GAS6	CCDS45072.1	c.446G>A	p.G149E	missense	T
1117	PD7436	ET	JAK2	X	114425813	T	A	12	0.0	15	20.0	RBMXL3	ENST00000424776	c.1809T>A	p.S603R	missense	T
1118	PD7436	ET	JAK2	3	52786237	G	A	318	0.0	363	19.8	NEK4	CCDS2863.1	c.1079C>T	p.A360V	missense	T
1119	PD7436	ET	JAK2	2	26257516	C	T	47	0.0	94	19.2	RAB10	CCDS1720.1	c.39C>T	p.L13L	silent	T
1120	PD7436	ET	JAK2	12	6806545	T	C	37	0.0	42	19.1	C12orf53	CCDS44818.1	c.431A>G	p.N144S	missense	T
1121	PD7436	ET	JAK2	8	23106826	C	G	29	0.0	32	18.8	CHMP7	CCDS6040.1	c.403C>G	p.L135V	missense	T
1122	PD7436	ET	JAK2	6	84056052	G	A	38	5.3	65	18.5	ME1	CCDS34492.1	c.440C>T	p.A147V	missense	T
1123	PD7436	ET	JAK2	2	206592724	C	A	46	2.2	65	18.5	NRP2	CCDS2364.1	c.1100C>A	p.T367N	missense	T
1124	PD7436	ET	JAK2	7	2594058	G	C	18	0.0	28	17.9	C7orf27	CCDS5334.1	c.8C>G	p.P3R	missense	T
1125	PD7436	ET	JAK2	12	59307770	G	T	49	0.0	68	17.7	LRIG3	CCDS8960.1	c.376C>A	p.L126I	missense	T
1126	PD7436	ET	JAK2	2	44553960	C	A	116	1.7	140	17.1	PREPL	CCDS33190.1	c.1637G>T	p.G546V	missense	T
1127	PD7436	ET	JAK2	2	25457242	C	T	42	0.0	30	16.7	DNMT3A	CCDS33157.1	c.2645G>A	p.R882H	missense	T C
1128	PD7436	ET	JAK2	7	99695778	G	A	20	0.0	36	16.7	MCM7	CCDS5683.1	c.856C>T	p.R286*	nonsense	T
1129	PD7436	ET	JAK2	X	41598667	A	G	328	0.3	142	16.2	CASK	CCDS14257.1	c.399T>C	p.H133H	silent	T
1130	PD7436	ET	JAK2	1	197071594	G	A	144	0.0	190	15.3	ASPM	CCDS1389.1	c.6787C>T	p.H2263Y	missense	T
1131	PD7436	ET	JAK2	1	248129264	G	T	40	0.0	59	15.3	OR2AK2	CCDS31102.1	c.631G>T	p.E211*	nonsense	T
1132	PD7436	ET	JAK2	10	82126661	A	C	265	0.0	337	15.1	DYDC2	CCDS7367.1	c.488A>C	p.H163P	missense	T
1133	PD7436	ET	JAK2	5	142662140	A	G	80	0.0	110	14.6	NR3C1	CCDS34258.1	c.2177T>C	p.M726T	missense	T
1134	PD7436	ET	JAK2	5	118525568	A	G	369	0.0	249	14.1	DMXL1	CCDS4125.1	c.7301A>G	p.Y2434C	missense	T
1135	PD7436	ET	JAK2	10	5969443	C	G	99	1.0	105	13.3	FBXO18	CCDS7073.1	c.2921C>G	p.T974S	missense	T
1136	PD7436	ET	JAK2	9	73479380	G	A	157	0.0	148	12.2	TRPM3	CCDS43835.1	c.225C>T	p.V75V	silent	T
1137	PD7436	ET	JAK2	10	73571109	C	T	26	0.0	36	11.1	CDH23	ENST00000398860	c.9130C>T	p.R3044W	missense	T
1138	PD7436	ET	JAK2	12	91396234	A	C	215	0.0	155	5.2	EPYC	CCDS31870.1	c.109T>G	p.L37V	missense	T
1139	PD7436	ET	JAK2	9	5073770	G	T	149	0.0	147	0.7	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
1140	PD7437	ET	MPL	1	43815009	G	T	49	0.0	56	39.3	MPL	CCDS483.1	c.1544G>T	p.W515L	missense	T C
1141	PD7437	ET	MPL	1	76378423	G	A	52	0.0	67	37.3	MSH4	CCDS670.1	c.2662G>A	p.V888M	missense	T C
1142	PD7437	ET	MPL	2	217724631	G	A	158	0.0	148	23.7	TNP1	CCDS2406.1	c.127C>T	p.R43W	missense	T C

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1143	PD7437	ET	MPL	16	31382405	C	T	65	0.0	103	20.4	ITGAX	CCDS10711.1	c.1711C>T	p.R571W	missense	T C
1144	PD7437	ET	MPL	2	128886594	T	C	100	0.0	222	19.8	UGGT1	CCDS2154.1	c.1227-9T>C	p.?	splice	T C
1145	PD7437	ET	MPL	3	10443892	G	A	128	0.0	265	18.9	ATP2B2	CCDS33701.1	c.538C>T	p.R180W	missense	T C
1146	PD7437	ET	MPL	5	55265528	T	G	108	0.0	117	18.0	IL6ST	CCDS3971.1	c.220A>C	p.I74L	missense	T C
1147	PD7437	ET	MPL	6	79668297	C	T	59	0.0	113	16.8	PHIP	CCDS4987.1	c.3677G>A	p.W1226*	nonsense	T C
1148	PD7437	ET	MPL	2	25457242	C	T	30	0.0	49	14.3	DNMT3A	CCDS33157.1	c.2645G>A	p.R882H	missense	T C
1149	PD7437	ET	MPL	1	215972270	C	G	176	0.0	71	12.7	USH2A	CCDS31025.1	c.9937G>C	p.V3313L	missense	T C
1150	PD7437	ET	MPL	6	124979623	T	G	197	0.0	205	4.9	NKAIN2	ENST00000368416	c.565T>G	p.*189E	nonstop	T C
1151	PD7438	ET	unmutated	12	133277885	C	T	100	2.0	186	41.4	PGAM5	CCDS9279.1	c.449C>T	p.P150L	missense	T
1152	PD7438	ET	unmutated	6	46826302	T	C	38	5.3	87	40.2	GPR116	CCDS4919.1	c.3338A>G	p.Y1113C	missense	T
1153	PD7438	ET	unmutated	15	35083473	C	T	102	2.0	149	39.6	ACTC1	CCDS10041.1	c.832G>A	p.E278K	missense	T
1154	PD7438	ET	unmutated	X	17743974	G	A	70	1.4	70	24.3	NHS	CCDS14181.1	c.1685G>A	p.R562H	missense	T
1155	PD7438	ET	unmutated	2	106739466	T	C	183	1.1	372	22.0	UXS1	CCDS46378.1	c.704A>G	p.K235R	missense	T
1156	PD7438	ET	unmutated	10	69881306	C	T	75	2.7	95	12.6	MYPN	CCDS7275.1	c.111C>T	p.P37P	silent	T
1157	PD7438	ET	unmutated	11	59626652	C	T	177	0.0	196	10.7	TCN1	CCDS7978.1	c.645G>A	p.K215K	silent	T
1158	PD7438	ET	unmutated	14	58955535	A	C	209	0.5	243	4.1	KIAA0586	CCDS45115.1	c.3496A>C	p.K1166Q	missense	T
1159	PD7438	ET	unmutated	19	13054569	_	delAGGCT<24>ACGCA	15	0.0	23	30.4	CALR	CCDS12288.1	c.1096_1129del34	p.R366fs*53	frameshift	M T C
1160	PD7440	ET	unmutated	13	23907724	C	T	121.00	3.3	139	21.6	SACS	CCDS9300.2	c.10291G>A	p.V3431I	missense	M
1161	PD7440	ET	unmutated	10	85984824	G	A	121.00	4.1	182	21.4	LRIT2	CCDS31234.1	c.157C>T	p.L53F	missense	M
1162	PD7440	ET	unmutated	11	6648209	G	A	65.00	4.6	97	19.6	DCHS1	CCDS7771.1	c.6061C>T	p.R2021C	missense	M
1163	PD7440	ET	unmutated	1	10364041	A	G	118	5.1	137	19.0	KIF1B	CCDS112.1	c.2798A>G	p.Q933R	missense	M
1164	PD7440	ET	unmutated	12	109998861	G	A	35.00	0.0	44	15.9	MMAB	CCDS9131.1	c.568C>T	p.R190C	missense	M
1165	PD7440	ET	unmutated	7	100549502	C	T	232.00	5.6	271	15.1	ENSG00000228273	ENST00000379458	c.83C>T	p.S28F	missense	M
1166	PD7440	ET	unmutated	5	140348426	A	G	77.00	2.6	104	14.4	PCDHAC2	CCDS4242.1	c.2075A>G	p.Q692R	missense	M
1167	PD7440	ET	unmutated	12	70352300	C	T	71.00	2.8	59	8.5	C12orf28	ENST00000299350	c.817C>T	p.R273C	missense	M
1168	PD7440	ET	unmutated	19	13054627	_	insTTGTC	11	0.0	25	52.0	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M T C
1169	PD7441	ET	unmutated	3	52027854	_	delCCTTGG	4.00	0.0	24	20.8	RPL29	CCDS2845.1	c.386_391delCCAAGG	p.A129_K130delAK	inframe	T
1170	PD7442	ET	unmutated	7	129939220	C	T	123	0.0	90	7.8	CPA4	CCDS5818.1	c.261C>T	p.Y87Y	silent	T C
1171	PD7442	ET	unmutated	19	13054627	_	insTTGTC	2	0.0	25	24.0	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M T C
1172	PD7443	ET	unmutated	19	13054565	_	delGCAGA<42>AGGAG	23	0.0	30	23.3	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M T C
1173	PD7443	ET	unmutated	20	42972021	C	T	117	0.0	87	25.3	R3HDML	CCDS13329.1	c.385C>T	p.R129W	missense	T
1174	PD7443	ET	unmutated	9	135203790	T	G	279	0.0	214	24.8	SETX	CCDS6947.1	c.3195A>C	p.E1065D	missense	T
1175	PD7443	ET	unmutated	15	59506831	G	C	363	0.3	289	23.9	MYO1E	CCDS32254.1	c.1188+8C>G	p.?	splice	T
1176	PD7443	ET	unmutated	19	50550026	C	T	155	0.0	104	23.1	ZNF473	CCDS33077.1	c.2326C>T	p.H776Y	missense	T
1177	PD7443	ET	unmutated	4	84205996	T	A	60	0.0	68	22.1	COQ2	ENST00000311469	c.72A>T	p.G24G	silent	T
1178	PD7443	ET	unmutated	1	40367080	A	G	51	0.0	55	21.8	MYCL1	CCDS30682.1	c.27T>C	p.Y9Y	silent	T
1179	PD7443	ET	unmutated	2	145162549	G	A	87	1.2	57	21.1	ZEB2	CCDS2186.1	c.446C>T	p.A149V	missense	T
1180	PD7443	ET	unmutated	18	10487660	C	T	117	0.0	92	20.7	APCDD1	CCDS11849.1	c.1170C>T	p.C390C	silent	T
1181	PD7443	ET	unmutated	7	5257594	G	A	399	0.0	285	18.6	WIPI2	CCDS5339.1	c.618G>A	p.A206A	silent	T
1182	PD7443	ET	unmutated	19	47278828	T	C	168	0.0	140	18.6	SLC1A5	CCDS12692.1	c.1565A>G	p.K522R	missense	T
1183	PD7443	ET	unmutated	17	1561620	T	C	158	0.0	138	11.6	PRPF8	CCDS11010.1	c.5432A>G	p.N1811S	missense	T
1184	PD7443	ET	unmutated	11	18955840	C	G	203	0.0	211	10.9	MRGPRX1	CCDS7846.1	c.492G>C	p.L164L	silent	T
1185	PD8624	PV	JAK2	X	12734270	G	A	167	0.0	157	92.4	FRMPD4	CCDS35201.1	c.1692G>A	p.G564G	silent	T
1186	PD8624	PV	JAK2	X	130420627	C	T	78	0.0	52	90.4	IGSF1	CCDS14629.1	c.22G>A	p.E8K	missense	T
1187	PD8624	PV	JAK2	9	5073770	G	T	191	0.0	151	89.4	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1188	PD8624	PV	JAK2	16	47345182	C	T	55	0.0	53	52.8	ITFG1	CCDS10728.1	c.1039G>A	p.A347T	missense	T
1189	PD8624	PV	JAK2	5	98228280	A	G	132	0.0	101	45.5	CHD1	CCDS34204.1	c.2129T>C	p.V710A	missense	T
1190	PD8624	PV	JAK2	8	143960835	G	A	20	0.0	33	45.5	CYP11B1	ENST00000377675	c.301C>T	p.R101C	missense	T
1191	PD8624	PV	JAK2	5	70308219	C	T	104	0.0	98	42.9	ENSG00000249437	CCDS4009.1	c.524G>A	p.G175E	missense	T
1192	PD8624	PV	JAK2	4	82369281	A	T	234	0.0	173	39.9	RASGEF1B	CCDS34022.1	c.596T>A	p.I199N	missense	T
1193	PD8624	PV	JAK2	1	146751866	T	G	193	0.0	58	37.9	CHD1L	CCDS927.1	c.1705+2T>G	p.?	essential splice	T
1194	PD8624	PV	JAK2	16	2336765	C	T	45	0.0	78	29.5	ABCA3	CCDS10466.1	c.3208G>A	p.A1070T	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1195	PD8624	PV	JAK2	6	32015790	C	T	20	0.0	41	29.3	TNXB	CCDS47407.1	c.10040-1G>A	p.?	essential splice	T
1196	PD8625	PV	JAK2	9	5073770	G	T	265	0.7	173	31.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T C
1197	PD8625	PV	JAK2	8	26365706	G	A	181	0.0	95	17.9	PNMA2	ENST00000494213	c.566C>T	p.T189M	missense	T C
1198	PD8625	PV	JAK2	X	65253670	G	A	182	1.7	90	13.3	VSIG4	CCDS14383.1	c.58C>T	p.R20C	missense	T C
1199	PD8625	PV	JAK2	5	422892	C	T	26	0.0	65	12.3	AHRR	CCDS43297.1	c.502C>T	p.R168C	missense	T C
1200	PD8625	PV	JAK2	4	47589194	C	G	128	0.0	129	8.5	ATP10D	CCDS3476.1	c.3912C>G	p.I1303M	missense	T C
1201	PD8625	PV	JAK2	9	113169278	C	T	157	1.3	228	8.3	SVEP1	CCDS48004.1	c.8602G>A	p.A2868T	missense	T C
1202	PD8625	PV	JAK2	2	228221829	G	A	227	1.3	142	5.6	MFF	CCDS2465.1	c.1025G>A	p.R342H	missense	T C
1203	PD8626	PV	JAK2	9	5073770	G	T	201	1.5	138	43.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1204	PD8626	PV	JAK2	4	73942011	T	C	59	0.0	30	16.7	ANKRD17	CCDS34004.1	c.7753-4A>G	p.?	splice	T
1205	PD8626	PV	JAK2	16	818666	C	A	14	0.0	36	11.1	MSLN	CCDS32356.1	c.1826C>A	p.P609H	missense	T
1206	PD8628	ET	JAK2	15	91769745	G	C	53	1.9	38	42.1	SV2B	CCDS10370.1	c.252G>C	p.R84S	missense	T
1207	PD8628	ET	JAK2	15	48539175	G	A	500	8.8	350	39.1	SLC12A1	CCDS10129.2	c.1522G>A	p.A508T	missense	T
1208	PD8628	ET	JAK2	2	133538788	G	T	64	1.6	74	37.8	NCKAP5	CCDS46418.1	c.4886C>A	p.A1629E	missense	T
1209	PD8628	ET	JAK2	19	33355032	C	T	21	4.8	34	35.3	SLC7A9	CCDS12425.1	c.448G>A	p.V150I	missense	T
1210	PD8628	ET	JAK2	1	154680715	G	T	500	4.2	340	31.2	KCNN3	CCDS30880.1	c.1933C>A	p.L645I	missense	T
1211	PD8628	ET	JAK2	9	5073770	G	T	182	2.7	142	27.5	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1212	PD8628	ET	JAK2	5	138240097	G	A	147	2.7	84	27.4	CTNNA1	CCDS34243.1	c.1356G>A	p.M452I	missense	T
1213	PD8628	ET	JAK2	4	41263917	G	A	41	2.4	44	27.3	UCHL1	CCDS5295.1	c.436G>A	p.V146M	missense	T
1214	PD8628	ET	JAK2	15	57730653	G	A	126	0.0	130	23.1	CGNL1	CCDS10161.1	c.456G>A	p.L152L	silent	T
1215	PD8628	ET	JAK2	1	169519234	G	A	98	0.0	81	18.5	F5	CCDS1281.1	c.1416C>T	p.I472I	silent	T
1216	PD8628	ET	JAK2	10	50916646	_	insAGGG	161	0.0	128	10.2	C10orf53	CCDS31202.1	c.457_458insAGGG	p.G153fs*37	frameshift	T
1217	PD8628	ET	JAK2	9	74526679	G	A	246	1.2	393	4.8	C9orf85	CCDS6639.1	c.29G>A	p.R10H	missense	T
1218	PD8629	ET	JAK2	17	40871212	A	C	245	0.4	211	26.5	EZH1	CCDS32659.1	c.678T>G	p.S226R	missense	T
1219	PD8629	ET	JAK2	18	19085865	C	T	163	0.0	160	26.3	GREB1L	CCDS45836.1	c.4323C>T	p.H1441H	silent	T
1220	PD8629	ET	JAK2	2	236715959	C	T	84	0.0	112	24.1	AGAP1	CCDS33408.1	c.1034C>T	p.A345V	missense	T
1221	PD8629	ET	JAK2	8	17814836	T	C	500	0.0	229	23.6	PCM1	CCDS47812.1	c.1710T>C	p.S570S	silent	T
1222	PD8629	ET	JAK2	6	87967906	C	T	78	0.0	81	23.5	ZNF292	CCDS47457.1	c.4559C>T	p.A1520V	missense	T
1223	PD8629	ET	JAK2	22	40704607	G	A	191	0.0	197	22.8	TNRC6B	CCDS46713.1	c.3882G>A	p.E1294E	silent	T
1224	PD8629	ET	JAK2	X	73963048	G	T	159	0.0	137	22.6	KIAA2022	CCDS35337.1	c.1344C>A	p.I448I	silent	T
1225	PD8629	ET	JAK2	X	71694531	C	T	92	0.0	106	21.7	HDAC8	ENST00000373561	c.787G>A	p.A263T	missense	T
1226	PD8629	ET	JAK2	11	36297713	C	T	35	0.0	43	20.9	COMMD9	CCDS7900.1	c.430G>A	p.V144I	missense	T
1227	PD8629	ET	JAK2	10	44112369	C	T	43	0.0	54	20.4	ZNF485	CCDS7205.2	c.878C>T	p.T293I	missense	T
1228	PD8629	ET	JAK2	2	71839789	C	T	93	0.0	70	20.0	DYSF	CCDS46328.1	c.4240C>T	p.L1414F	missense	T
1229	PD8629	ET	JAK2	X	150345326	G	A	192	0.0	198	19.7	GPR50	CCDS44012.1	c.133G>A	p.G45S	missense	T
1230	PD8629	ET	JAK2	5	54619984	C	G	54	0.0	36	19.4	SKIV2L2	CCDS3967.1	c.297C>G	p.V99V	silent	T
1231	PD8629	ET	JAK2	6	12120176	C	T	500	0.2	321	17.5	HIVEP1	CCDS43426.1	c.148C>T	p.R50C	missense	T
1232	PD8629	ET	JAK2	9	5073770	G	T	202	0.5	92	17.4	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1233	PD8629	ET	JAK2	1	207648508	G	A	85	0.0	69	17.4	CR2	CCDS31007.1	c.2663G>A	p.R888K	missense	T
1234	PD8629	ET	JAK2	18	77893846	_	delAACTC<10>CTAAG	68	0.0	52	17.3	ADNP2	CCDS32853.1	c.550_569del20	p.S185fs*0	frameshift	T
1235	PD8629	ET	JAK2	14	95670757	G	A	110	0.0	106	16.0	CLMN	CCDS9933.1	c.929C>T	p.T310I	missense	T
1236	PD8629	ET	JAK2	5	79032617	_	delAG	368	0.0	271	5.9	CMYA5	CCDS47238.1	c.8029_8030delAG	p.E2678fs*36	frameshift	T
1237	PD8630	PV	JAK2	4	106157653	G	T	89	6.3	79	32.9	TET2	CCDS47120.1	c.2554G>T	p.E852*	nonsense	T
1238	PD8630	PV	JAK2	12	55969008	G	A	147	4.1	137	30.7	OR2AP1	ENST00000321688	c.810G>A	p.K270K	silent	T
1239	PD8630	PV	JAK2	1	11115942	G	T	13	0.0	10	30.0	SRM	CCDS125.1	c.661C>A	p.R221R	silent	T
1240	PD8630	PV	JAK2	9	5073770	G	T	200	0.0	165	24.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1241	PD8630	PV	JAK2	12	49691801	T	C	42	4.8	33	15.2	PRPH	CCDS8783.1	c.1328T>C	p.I443T	missense	T
1242	PD8630	PV	JAK2	3	168834115	T	C	160	0.6	149	14.8	MECOM	CCDS3205.1	c.981A>G	p.L327L	silent	T
1243	PD8630	PV	JAK2	3	58149042	G	A	29	3.5	44	13.6	FLNB	CCDS2885.1	c.7183G>A	p.E2395K	missense	T
1244	PD8630	PV	JAK2	12	101799685	C	T	73	0.0	67	11.9	ARL1	CCDS44958.1	c.79G>A	p.G27R	missense	T
1245	PD8630	PV	JAK2	19	46733615	G	T	211	1.0	185	11.9	IGFL1	CCDS46123.1	c.164G>T	p.C55F	missense	T
1246	PD8630	PV	JAK2	1	150776560	G	A	260	0.0	183	10.9	CTSK	CCDS969.1	c.555C>T	p.A185A	silent	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1247	PD8630	PV	JAK2	11	63486810	C	A	103	3.9	111	10.8	RTN3	CCDS8050.1	c.779C>A	p.T260N	missense	T
1248	PD8630	PV	JAK2	3	132360937	G	A	144	0.0	128	8.6	ACAD11	CCDS3074.1	c.416C>T	p.P139L	missense	T
1249	PD8630	PV	JAK2	1	155921296	T	C	103	1.9	75	8.0	ARHGEF2	CCDS1125.1	c.2153A>G	p.Y718C	missense	T
1250	PD8630	PV	JAK2	7	149480728	G	A	90	0.0	80	7.5	SSPO	ENST00000378016	c.2434G>A	p.G812S	missense	T
1251	PD8630	PV	JAK2	5	140181545	G	A	63	0.0	81	7.4	PCDHA3	CCDS34248.1	c.763G>A	p.A255T	missense	T
1252	PD8630	PV	JAK2	4	106158509	G	A	176	0.6	123	7.3	TET2	CCDS47120.1	c.3409+1G>A	p.?	essential splice	T
1253	PD8630	PV	JAK2	11	56237766	A	T	126	0.0	131	5.3	OR5M3	CCDS31532.1	c.208T>A	p.W70R	missense	T
1254	PD8630	PV	JAK2	15	43438717	G	A	343	0.0	302	5.0	TMEM62	CCDS32210.1	c.503G>A	p.G168D	missense	T
1255	PD8631	PV	JAK2	9	5073770	G	T	114	0.0	117	36.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1256	PD8631	PV	JAK2	10	118310706	G	A	36	0.0	63	20.6	PNLIP	CCDS7594.1	c.421G>A	p.V141M	missense	T
1257	PD8631	PV	JAK2	15	49308749	G	C	119	0.0	98	19.4	SECISBP2L	CCDS32234.1	c.1426+8C>G	p.?	splice	T
1258	PD8631	PV	JAK2	4	71099842	C	T	207	0.0	190	17.4	C4orf7	CCDS3537.1	c.196C>T	p.R66C	missense	T
1259	PD8631	PV	JAK2	11	9871702	A	C	122	0.0	151	15.9	SBF2	CCDS31427.1	c.2674T>G	p.L892V	missense	T
1260	PD8631	PV	JAK2	1	197111833	C	G	288	0.0	220	14.6	ASPM	CCDS1389.1	c.1549G>C	p.A517P	missense	T
1261	PD8631	PV	JAK2	12	78598873	A	T	60	0.0	71	14.1	NAV3	CCDS41815.1	c.6927A>T	p.T2309T	silent	T
1262	PD8631	PV	JAK2	17	34236448	G	T	76	1.3	82	12.2	AC015849.4	CCDS31216.1	Non-coding		RNA	T
1263	PD8631	PV	JAK2	10	118202630	C	T	57	0.0	92	12.0	PNLIPRP3	CCDS31292.1	c.268C>T	p.R90C	missense	T
1264	PD8631	PV	JAK2	7	148508779	A	G	76	1.3	62	11.3	EZH2	CCDS5891.1	c.1885T>C	p.W629R	missense	T
1265	PD8631	PV	JAK2	1	39352328	C	T	141	0.0	168	10.1	RHBDL2	CCDS30680.1	c.760G>A	p.G254R	missense	T
1266	PD8631	PV	JAK2	19	22496698	T	A	210	0.0	61	9.8	ZNF729	ENST00000357491	c.479T>A	p.M160K	missense	T
1267	PD8631	PV	JAK2	4	152060988	G	A	26	0.0	51	9.8	SH3D19	CCDS34077.2	c.1464C>T	p.H488H	silent	T
1268	PD8631	PV	JAK2	4	164247072	G	A	52	0.0	65	7.7	NPY1R	CCDS34089.1	c.635C>T	p.T212I	missense	T
1269	PD8631	PV	JAK2	2	132238024	C	T	41	0.0	185	6.5	TUBA3D	CCDS33290.1	c.758C>T	p.T253M	missense	T
1270	PD8631	PV	JAK2	19	5739421	A	C	58	0.0	66	6.1	TMEM146	CCDS12149.2	c.544A>C	p.S182R	missense	T
1271	PD8631	PV	JAK2	5	180374517	T	A	153	0.0	300	4.7	BTNL8	CCDS43413.1	c.679T>A	p.F227I	missense	T
1272	PD8632	ET	unmutated	19	13054627	_	insTTGTC	5	0.0	9	55.6	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M C
1273	PD8632	ET	unmutated	13	111080938	T	C	18	0.0	44	47.7	COL4A2	CCDS41907.1	c.477+8T>C	p.?	splice	T
1274	PD8632	ET	unmutated	11	4609000	C	G	427	0.9	261	34.1	OR52I2	CCDS31355.1	c.958C>G	p.P320A	missense	T
1275	PD8632	ET	unmutated	17	48264235	C	T	29	0.0	100	33.0	COL1A1	CCDS11561.1	c.3580G>A	p.A1194T	missense	T
1276	PD8632	ET	unmutated	3	54959184	C	T	62	0.0	67	26.9	LRTM1	CCDS2876.1	c.66G>A	p.P22P	silent	T
1277	PD8633	PV	JAK2	19	8550858	_	delGAGCG<11>CCATC	15	0.0	43	14.0	HNRNPM	CCDS12203.1	c.1546_1566del21	p.E516_I522delERMGPAI	inframe	M
1278	PD8633	PV	JAK2	7	31146163	C	T	128	0.0	88	10.2	ADCYAP1R1	CCDS5433.1	c.1272C>T	p.F424F	silent	M
1279	PD8633	PV	JAK2	9	5073770	G	T	173	0.0	154	7.8	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1280	PD8635	ET	unmutated	13	52345553	_	delATT	4	1.0	24	45.8	DHRS12	CCDS31976.1	c.636_638delAAT	p.I212delI	inframe	M
1281	PD8635	ET	unmutated	2	47641560	_	delAAAAAAAA	40	1.0	20	20.0	MSH2	CCDS1834.1	c.942+3_942+10delaaaaaaaa	p.?	frameshift	M
1282	PD8637	ET	unmutated	19	13054565	_	delGCAGA<42>AGGAG	17	0.0	17	29.4	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M C
1283	PD8637	ET	unmutated	15	23811321	G	A	66	0.0	77	58.4	MKRN3	CCDS10013.1	c.392G>A	p.G131D	missense	T
1284	PD8637	ET	unmutated	19	54403681	C	T	14	0.0	53	41.5	PRKCG	CCDS12867.1	c.1382C>T	p.A461V	missense	T
1285	PD8637	ET	unmutated	17	44145030	C	A	56	0.0	51	35.3	KIAA1267	CCDS11503.1	c.1537G>T	p.E513*	nonsense	T
1286	PD8637	ET	unmutated	2	26667723	C	T	85	0.0	66	34.9	C2orf39	CCDS1723.1	c.1303C>T	p.P435S	missense	T
1287	PD8637	ET	unmutated	6	34101001	G	A	29	0.0	29	34.5	GRM4	CCDS4787.1	c.273C>T	p.N91N	silent	T
1288	PD8637	ET	unmutated	8	143623641	C	T	14	0.0	51	29.4	BAI1	ENST00000323289	c.4046C>T	p.P1349L	missense	T
1289	PD8637	ET	unmutated	11	57100173	A	G	40	0.0	73	27.4	SSRP1	CCDS7952.1	c.694T>C	p.Y232H	missense	T
1290	PD8637	ET	unmutated	X	53588740	A	G	96	0.0	81	18.5	HUWE1	CCDS35301.1	c.7484T>C	p.F2495S	missense	T
1291	PD8640	ET	MPL	22	29837831	G	T	104	6.7	83	51.8	RFPL1	CCDS13857.2	c.674G>T	p.R225L	missense	T
1292	PD8640	ET	MPL	19	48382367	C	T	179	2.8	167	49.7	SULT2A1	CCDS12707.1	c.493G>A	p.D165N	missense	T
1293	PD8640	ET	MPL	5	140615008	T	C	196	1.5	206	48.1	PCDHB18	ENST00000274705	c.723T>C	p.H241H	silent	T
1294	PD8640	ET	MPL	17	45560429	G	T	221	1.8	130	46.9	ENSG00000228782	ENST00000425159	c.387C>A	p.A129A	silent	T
1295	PD8640	ET	MPL	1	43815009	G	T	62	3.2	67	46.3	MPL	CCDS483.1	c.1544G>T	p.W515L	missense	T
1296	PD8640	ET	MPL	2	25463302	A	C	38	2.6	50	44.0	DNMT3A	CCDS33157.1	c.2191T>G	p.F731V	missense	T C
1297	PD8640	ET	MPL	3	161220904	G	T	30	0.0	37	43.2	ENSG00000182447	CCDS46948.1	c.608G>T	p.C203F	missense	T
1298	PD8640	ET	MPL	4	30726223	G	A	29	0.0	39	41.0	PCDH7	CCDS43220.1	c.3033+5G>A	p.?	essential splice	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1299	PD8640	ET	MPL	2	100029344	A	G	46	0.0	118	40.7	REV1	CCDS2045.1	c.2021T>C	p.M674T	missense	T
1300	PD8640	ET	MPL	13	115052022	C	G	42	2.4	47	40.4	UPF3A	CCDS9543.1	c.549C>G	p.T183T	silent	T
1301	PD8640	ET	MPL	20	31023437	C	G	46	2.2	89	39.3	ASXL1	CCDS13201.1	c.2922C>G	p.Y974*	nonsense	T
1302	PD8640	ET	MPL	15	28090177	G	A	61	4.9	74	35.1	OCA2	CCDS10020.1	c.2360C>T	p.A787V	missense	T
1303	PD8640	ET	MPL	11	58034551	C	T	125	0.8	72	34.7	OR10W1	CCDS7968.1	c.780G>A	p.K260K	silent	T
1304	PD8640	ET	MPL	8	48885458	C	G	69	0.0	42	31.0	MCM4	CCDS6143.1	c.1970C>G	p.A657G	missense	T
1305	PD8640	ET	MPL	22	32110551	_	delA	178	0.6	171	24.0	C22orf30	CCDS13900.2	c.3274delT	p.S1092fs*37	frameshift	T
1306	PD8640	ET	MPL	2	97276503	T	A	376	0.0	251	4.8	KIAA1310	CCDS46361.1	c.1279A>T	p.N427Y	missense	T
1307	PD8642	ET	unmutated	19	13054565	_	delGCAGA<42>AGGAG	9	0.0	30	33.3	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M C
1308	PD8642	ET	unmutated	1	183091306	C	T	239	4.2	107	39.3	LAMC1	CCDS1351.1	c.2321C>T	p.P774L	missense	T
1309	PD8642	ET	unmutated	16	1961985	C	T	20	0.0	39	38.5	HS3ST6	CCDS45381.1	c.542G>A	p.G181D	missense	T
1310	PD8642	ET	unmutated	14	95912380	G	A	68	0.0	163	26.4	C14orf49	CCDS9935.1	c.1498C>T	p.Q500*	nonsense	T
1311	PD8642	ET	unmutated	11	105758298	G	C	135	1.5	35	22.9	GRIA4	CCDS8333.1	c.726G>C	p.L242F	missense	T
1312	PD8642	ET	unmutated	12	6692423	C	T	134	0.0	246	21.1	CHD4	CCDS8552.1	c.4001G>A	p.G1334D	missense	T
1313	PD8642	ET	unmutated	6	93964450	G	A	69	0.0	68	20.6	EPHA7	CCDS5031.1	c.2447C>T	p.A816V	missense	T
1314	PD8642	ET	unmutated	6	90573428	C	G	133	0.0	177	19.2	CASP8AP2	ENST00000455594	c.2000C>G	p.T667S	missense	T
1315	PD8642	ET	unmutated	9	125281519	A	C	264	0.4	177	14.1	OR1J4	CCDS35122.1	c.100A>C	p.M34L	missense	T
1316	PD8642	ET	unmutated	11	74994323	T	C	31	0.0	53	11.3	ARRB1	CCDS44684.1	c.354+8A>G	p.?	splice	T
1317	PD8642	ET	unmutated	1	94996094	C	T	98	2.0	89	11.2	F3	ENST00000370207	c.650G>A	p.W217*	nonsense	T
1318	PD8642	ET	unmutated	17	39846180	A	C	43	0.0	86	10.5	EIF1	CCDS11403.1	c.182A>C	p.K61T	missense	T
1319	PD8642	ET	unmutated	12	56848342	G	A	54	0.0	134	10.5	MIP	CCDS8919.1	c.56C>T	p.A19V	missense	T
1320	PD8642	ET	unmutated	7	114269863	C	A	126	3.2	96	9.4	FOXP2	CCDS43635.1	c.475C>A	p.Q159K	missense	T
1321	PD8642	ET	unmutated	2	179464144	T	A	207	2.9	260	8.9	TTN	ENST00000356127	c.48666A>T	p.I16222I	silent	T
1322	PD8642	ET	unmutated	16	56717865	T	C	101	2.0	269	8.6	MT1X	CCDS10768.1	c.95-5T>C	p.?	splice	T
1323	PD8642	ET	unmutated	13	78235580	C	G	219	2.3	500	4.6	ENSG00000215419	ENST00000400287	c.957G>C	p.R319S	missense	T
1324	PD8643	ET	JAK2	9	5073770	G	T	153	0.0	210	8.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1325	PD8643	ET	JAK2	7	156451184	G	C	182	0.6	161	5.0	RNF32	CCDS5944.1	c.604G>C	p.V202L	missense	T
1326	PD8644	ET	JAK2	8	27927128	T	G	346	6.4	370	32.2	C8orf80	CCDS47833.1	c.190A>C	p.T64P	missense	T
1327	PD8644	ET	JAK2	11	19192021	T	C	28	0.0	32	28.1	ZDHHC13	CCDS44550.1	c.1538T>C	p.I513T	missense	T
1328	PD8644	ET	JAK2	9	5073770	G	T	300	0.0	223	26.9	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1329	PD8644	ET	JAK2	3	48694538	G	A	24	0.0	38	26.3	CELSR3	CCDS2775.1	c.3992C>T	p.S1331L	missense	T
1330	PD8644	ET	JAK2	2	70130314	G	A	271	5.9	216	23.2	SNRNP27	CCDS33219.1	c.350G>A	p.G117D	missense	T
1331	PD8644	ET	JAK2	12	118610293	C	T	140	0.0	217	17.5	TAOK3	CCDS9188.1	c.1868G>A	p.R623Q	missense	T
1332	PD8644	ET	JAK2	X	40568684	G	C	111	0.0	86	17.4	MED14	CCDS14254.1	c.1201C>G	p.L401V	missense	T
1333	PD8644	ET	JAK2	1	82421647	T	C	140	0.0	147	17.0	LPHN2	CCDS689.1	c.1869T>C	p.T623T	silent	T
1334	PD8644	ET	JAK2	2	137928444	C	T	242	0.0	188	14.9	THSD7B	ENST00000272643	c.1659C>T	p.F553F	silent	T
1335	PD8644	ET	JAK2	19	17312945	A	T	72	0.0	156	14.1	MYO9B	CCDS46010.1	c.4669A>T	p.K1557*	nonsense	T
1336	PD8644	ET	JAK2	2	101902564	G	A	231	0.0	205	12.7	RNF149	CCDS2051.1	c.939C>T	p.V313V	silent	T
1337	PD8644	ET	JAK2	22	29108001	C	T	101	0.0	96	12.5	CHEK2	CCDS33629.1	c.817G>A	p.A273T	missense	T
1338	PD8644	ET	JAK2	15	41011043	A	C	89	2.3	130	10.0	RAD51	CCDS10062.1	c.476A>C	p.Y159S	missense	T
1339	PD8644	ET	JAK2	3	120977900	C	T	169	0.0	128	9.4	STXBP5L	CCDS43137.1	c.1843C>T	p.R615W	missense	T
1340	PD8644	ET	JAK2	3	122591345	G	C	363	0.8	354	5.7	DIRC2	CCDS3018.1	c.1222G>C	p.V408L	missense	T
1341	PD8644	ET	JAK2	X	119387380	T	C	500	0.0	500	4.2	ZBTB33	CCDS14596.1	c.110T>C	p.I37T	missense	T
1342	PD8942	ET	unmutated	19	13054627	_	insTTGTC	4	0.0	7	57.1	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M T C
1343	PD8942	ET	unmutated	1	112997118	G	A	257	5.1	240	48.3	CTTNBP2NL	CCDS845.1	c.378G>A	p.T126T	silent	M
1344	PD8942	ET	unmutated	7	122153323	G	A	73	0.0	69	13.0	CADPS2	CCDS47691.1	c.1522C>T	p.R508C	missense	M
1345	PD8943	PET-MF	unmutated	1	17736549	T	C	19	0.0	22	77.3	RCC2	CCDS181.1	c.1387-2A>G	p.?	essential splice	M
1346	PD8943	PET-MF	unmutated	8	108264028	G	A	58	0.0	37	59.5	ANGPT1	CCDS6306.1	Non-coding	r.2049c>u	UTR	M
1347	PD8943	PET-MF	unmutated	3	24009427	A	G	102	0.0	74	56.8	NR1D2	CCDS33718.1	c.1456A>G	p.M486V	missense	M
1348	PD8943	PET-MF	unmutated	2	219305522	G	T	24	0.0	42	54.8	VIL1	CCDS2417.1	c.2307G>T	p.L769L	silent	M
1349	PD8943	PET-MF	unmutated	2	203162177	A	G	134	0.0	98	54.1	NOP58	CCDS2353.1	c.1147A>G	p.M383V	missense	M
1350	PD8943	PET-MF	unmutated	19	22363093	G	A	195	0.5	115	53.9	ZNF676	CCDS42539.1	c.1426C>T	p.P476S	missense	M

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1351	PD8943	PET-MF	unmutated	17	29559200	T	G	82	0.0	47	53.2	NF1	CCDS42292.1	c.3307T>G	p.F1103V	missense	M
1352	PD8943	PET-MF	unmutated	12	75444967	T	A	111	0.0	100	51.0	KCNC2	CCDS9007.1	c.818A>T	p.Q273L	missense	M
1353	PD8943	PET-MF	unmutated	3	52734472	G	C	96	0.0	80	50.0	GLT8D1	CCDS2862.1	c.5C>G	p.S2*	nonsense	M
1354	PD8943	PET-MF	unmutated	10	71716795	C	A	46	0.0	57	47.4	COL13A1	CCDS44420.1	c.2148C>A	p.N716K	missense	M
1355	PD8943	PET-MF	unmutated	15	89825042	T	C	94	0.0	104	47.1	FANCI	CCDS45346.1	c.1559T>C	p.V520A	missense	M
1356	PD8943	PET-MF	unmutated	10	30653708	G	A	31	0.0	41	46.3	MTPAP	ENST00000358107	c.474C>T	p.A158A	silent	M
1357	PD8943	PET-MF	unmutated	5	80552759	T	C	52	0.0	46	45.7	CKMT2	CCDS4053.1	c.695T>C	p.V232A	missense	M
1358	PD8943	PET-MF	unmutated	17	39645706	G	C	36	0.0	107	43.9	KRT36	CCDS11395.1	c.411C>G	p.C137W	missense	M
1359	PD8943	PET-MF	unmutated	5	23527933	T	A	15	0.0	23	43.5	PRDM9	CCDS43307.1	Non-coding	r.2918u>a	UTR	M
1360	PD8943	PET-MF	unmutated	11	85979541	A	G	193	0.0	87	42.5	EED	CCDS8273.1	c.904A>G	p.R302G	missense	M
1361	PD8943	PET-MF	unmutated	6	50803774	T	C	251	0.0	118	42.4	TFAP2B	CCDS4934.2	c.602T>C	p.V201A	missense	M
1362	PD8943	PET-MF	unmutated	12	67705481	A	G	59	0.0	60	41.7	CAND1	CCDS8977.1	c.3369A>G	p.T1123T	silent	M
1363	PD8943	PET-MF	unmutated	5	156670701	G	A	151	0.0	94	40.4	ITK	CCDS4336.1	c.1129G>A	p.V377M	missense	M
1364	PD8943	PET-MF	unmutated	20	31022403	_	delCACCA<13>GCGGC	10	0.0	25	16.0	ASXL1	CCDS13201.1	c.1888_1910del23	p.E635fs*15	frameshift	M
1365	PD8943	PET-MF	unmutated	17	38189431	C	A	17	0.0	32	15.6	MED24	CCDS11359.1	c.700G>T	p.E234*	nonsense	M
1366	PD8943	PET-MF	unmutated	15	40917018	C	A	40	0.0	40	12.5	CASC5	CCDS42023.1	c.4634C>A	p.A1545D	missense	M
1367	PD8943	PET-MF	unmutated	5	150282754	C	A	108	0.0	40	12.5	ZNF300	ENST00000446148	c.22-10G>T	p.?	splice	M
1368	PD8943	PET-MF	unmutated	20	24944532	C	A	27	0.0	61	9.8	C20orf3	CCDS13166.1	c.1168G>T	p.E390*	nonsense	M
1369	PD8943	PET-MF	unmutated	11	46907700	C	A	86	0.0	65	9.2	LRP4	CCDS31478.1	c.2448G>T	p.E816D	missense	M
1370	PD8943	PET-MF	unmutated	7	154379692	A	C	45	0.0	86	5.8	DPP6	ENST00000406326	c.960A>C	p.P320P	silent	M
1371	PD8943	PET-MF	unmutated	19	13054627	_	insTTGTC	3	0.0	7	57.1	CALR	CCDS12288.1	c.1154_1155insTTGTC	p.K385fs*47	frameshift	M T C
1372	PD8944	ET	MPL	4	100871390	G	T	16	0.0	25	28.0	H2AFZ	CCDS3654.1	Non-coding	r.156c>a	UTR	M
1373	PD8944	ET	MPL	1	43815009	G	T	71	4.2	67	16.4	MPL	CCDS483.1	c.1544G>T	p.W515L	missense	T
1374	PD8944	ET	MPL	16	23678523	T	G	42	0.0	33	15.2	DCTN5	CCDS10615.1	Non-coding	r.758u>g	UTR	M
1375	PD8945	ET	unmutated	12	21926351	A	G	54	0.0	41	12.2	KCNJ8	CCDS8692.1	c.200T>C	p.L67P	missense	M
1376	PD9409	PMF	JAK2	9	5073770	G	T	144	0.0	131	45.0	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1377	PD9409	PMF	JAK2	3	52817062	G	A	32	0.0	83	27.7	ITIH1	CCDS2864.1	c.1020G>A	p.W340*	nonsense	M
1378	PD9409	PMF	JAK2	10	118969313	C	T	34	0.0	58	24.1	KCNK18	CCDS7598.1	c.658C>T	p.R220C	missense	T
1379	PD9409	PMF	JAK2	13	41524011	G	A	118	0.0	75	21.3	ELF1	CCDS9374.1	c.460C>T	p.Q154*	nonsense	T
1380	PD9409	PMF	JAK2	17	48271336	C	T	17	0.0	35	20.0	COL1A1	CCDS11561.1	c.1735G>A	p.V579M	missense	M
1381	PD9409	PMF	JAK2	17	79517543	G	A	19	0.0	47	10.6	C17orf70	CCDS32765.1	c.524C>T	p.A175V	missense	M
1382	PD9410	PMF	JAK2	9	5073770	G	T	140	0.0	111	17.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1383	PD9410	PMF	JAK2	21	44514777	T	G	45	0.0	48	14.6	U2AF1	CCDS13694.1	c.470A>C	p.Q157P	missense	T
1384	PD9410	PMF	JAK2	16	4016686	G	A	57	0.0	135	13.3	ADCY9	CCDS32382.1	c.3152C>T	p.S1051F	missense	T
1385	PD9410	PMF	JAK2	20	31022712	C	T	27	0.0	38	13.2	ASXL1	CCDS13201.1	c.2197C>T	p.Q733*	nonsense	T
1386	PD9410	PMF	JAK2	12	122090661	C	A	49	0.0	35	11.4	MORN3	CCDS31917.1	c.702G>T	p.R234S	missense	T
1387	PD9411	PMF	JAK2	8	124659198	G	A	145	0.0	198	63.1	KLHL38	CCDS43766.1	c.1407C>T	p.N469N	silent	T
1388	PD9411	PMF	JAK2	19	57329187	G	A	19	0.0	16	62.5	PEG3AS	CCDS12948.1	c.789C>T	p.D263D	silent	T
1389	PD9411	PMF	JAK2	1	157803029	C	T	117	0.0	78	51.3	CD5L	CCDS1171.1	c.992G>A	p.G331E	missense	T
1390	PD9411	PMF	JAK2	20	31021250	C	T	71	0.0	90	51.1	ASXL1	CCDS13201.1	c.1249C>T	p.R417*	nonsense	T
1391	PD9411	PMF	JAK2	12	108169147	G	T	19	0.0	11	45.5	ASCL4	CCDS31894.2	c.155G>T	p.R52L	missense	T
1392	PD9411	PMF	JAK2	18	52896219	G	A	133	0.0	110	40.9	TCF4	CCDS42438.1	c.1738C>T	p.R580W	missense	T
1393	PD9411	PMF	JAK2	17	39258401	G	A	21	0.0	44	40.9	ENSG00000241241	ENST00000440582	c.61C>T	p.P21S	missense	T
1394	PD9411	PMF	JAK2	10	5435809	C	T	41	0.0	36	38.9	TUBAL3	CCDS7066.2	c.1012G>A	p.A338T	missense	T
1395	PD9411	PMF	JAK2	11	65113527	C	T	95	0.0	94	37.2	DPF2	CCDS8100.1	c.902C>T	p.S301L	missense	T
1396	PD9411	PMF	JAK2	7	148523590	C	T	53	0.0	31	29.0	EZH2	CCDS5891.1	c.863G>A	p.R288Q	missense	T
1397	PD9411	PMF	JAK2	8	15531331	C	T	121	0.0	129	27.1	TUSC3	CCDS5994.1	c.784C>T	p.H262Y	missense	T
1398	PD9411	PMF	JAK2	5	93120106	G	A	155	0.0	82	11.0	FAM172A	CCDS4069.1	c.1006C>T	p.R336*	nonsense	T
1399	PD9412	PMF	JAK2	Y	15435532	C	A	41	2.4	33	100.0	UTY	CCDS14783.1	c.2888G>T	p.G963V	missense	T
1400	PD9412	PMF	JAK2	9	5073770	G	T	136	0.0	169	62.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1401	PD9412	PMF	JAK2	9	34729283	G	A	211	1.4	243	61.3	ENSG00000205108	ENST00000378788	c.142C>T	p.R48W	missense	T
1402	PD9412	PMF	JAK2	12	45810576	C	T	42	0.0	63	60.3	ANO6	CCDS44865.1	c.2106C>T	p.D702D	silent	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1403	PD9412	PMF	JAK2	4	170912850	C	T	58	0.0	43	53.5	MFAP3L	CCDS34103.1	c.909G>A	p.S303S	silent	T
1404	PD9412	PMF	JAK2	3	86010782	G	A	66	0.0	72	52.8	CADM2	CCDS33792.1	c.934G>A	p.V312I	missense	T
1405	PD9412	PMF	JAK2	11	810334	G	T	16	0.0	27	48.2	RPLP2	CCDS7717.1	c.100G>T	p.A34S	missense	T
1406	PD9412	PMF	JAK2	1	115128224	T	C	80	0.0	50	44.0	DENND2C	CCDS875.1	c.2613A>G	p.K871K	silent	T
1407	PD9412	PMF	JAK2	17	38340509	C	T	106	0.0	117	42.7	RAPGEFL1	CCDS11363.1	c.25C>T	p.R9W	missense	T
1408	PD9412	PMF	JAK2	1	248757047	A	G	78	0.0	70	34.3	OR2T10	CCDS31121.1	c.23T>C	p.L8P	missense	T
1409	PD9412	PMF	JAK2	3	130287404	G	A	182	0.6	187	34.2	COL6A6	CCDS46911.1	c.2357G>A	p.R786H	missense	T
1410	PD9412	PMF	JAK2	1	153657511	G	A	35	0.0	45	31.1	NPR1	CCDS1051.1	c.1556G>A	p.S519N	missense	T
1411	PD9412	PMF	JAK2	1	217947788	C	T	215	0.5	184	25.0	SPATA17	CCDS1519.1	c.632C>T	p.S211F	missense	T
1412	PD9412	PMF	JAK2	3	128200131	_	ins_del	56	0.0	30	23.3	GATA2	CCDS3049.1	c.1162_1173delATGAAGAAGGAAinsTTC	p.M388_E391delinF	frameshift	T
1413	PD9412	PMF	JAK2	19	11486314	_	delAGACCC	105	0.0	34	14.0	C19orf39	CCDS12259.1	c.312_317delAGACCC	p.D105P106delDP	inframe	T
1414	PD9412	PMF	JAK2	17	30267491	_	delAACA	23	0.0	15	6.0	SUZ12P	CCDS11270.1	c.372_375delAACA	p.N126fs*15	frameshift	T
1415	PD9415	PMF	JAK2	12	116421136	C	T	78	0.0	64	60.9	MED13L	CCDS9177.1	c.4741G>A	p.G1581S	missense	T
1416	PD9415	PMF	JAK2	1	228430956	C	T	56	0.0	48	60.4	OBSCN	ENST00000422127	c.3002C>T	p.T1001M	missense	T
1417	PD9415	PMF	JAK2	3	123512526	G	A	29	0.0	34	55.9	MYLK	CCDS46896.1	c.163C>T	p.R55W	missense	T
1418	PD9415	PMF	JAK2	6	33371816	G	A	100	0.0	85	54.1	KIFC1	CCDS34430.1	c.666G>A	p.T222T	silent	T
1419	PD9415	PMF	JAK2	4	3318020	C	G	26	0.0	53	52.8	RGS12	CCDS3366.1	c.123C>G	p.P41P	silent	T
1420	PD9415	PMF	JAK2	5	82835692	G	A	81	0.0	77	52.0	VCAN	CCDS4060.1	c.6870G>A	p.V2290V	silent	T
1421	PD9415	PMF	JAK2	17	74732959	G	T	18	0.0	16	50.0	SFRS2	CCDS11749.1	c.284C>A	p.P95H	missense	T
1422	PD9415	PMF	JAK2	20	9319661	G	A	47	2.1	71	49.3	PLCB4	CCDS13104.1	c.346G>A	p.A116T	missense	T
1423	PD9415	PMF	JAK2	17	48602274	C	T	48	0.0	53	49.1	MYCBPAP	CCDS32680.2	c.1801C>T	p.R601C	missense	T
1424	PD9415	PMF	JAK2	9	116175797	G	A	39	0.0	49	49.0	C9orf43	CCDS6796.1	c.24G>A	p.Q8Q	silent	T
1425	PD9415	PMF	JAK2	5	79054625	C	A	174	0.0	144	48.6	CMYA5	CCDS47238.1	c.11160C>A	p.S3720R	missense	T
1426	PD9415	PMF	JAK2	4	106197285	T	C	51	0.0	33	48.5	TET2	CCDS47120.1	c.5618T>C	p.I1873T	missense	T
1427	PD9415	PMF	JAK2	10	24790464	G	A	41	0.0	44	47.7	KIAA1217	CCDS31165.1	c.1991G>A	p.R664Q	missense	T
1428	PD9415	PMF	JAK2	17	56573571	C	T	47	0.0	36	47.2	MTMR4	CCDS11608.1	c.1932G>A	p.L644L	silent	T
1429	PD9415	PMF	JAK2	17	34951522	G	A	96	0.0	67	46.3	DHRS11	CCDS11315.2	c.269G>A	p.G90D	missense	T
1430	PD9415	PMF	JAK2	6	5771526	C	T	42	0.0	31	45.2	FARS2	CCDS4494.1	c.1220C>T	p.T407M	missense	T
1431	PD9415	PMF	JAK2	6	63990252	C	T	104	1.0	113	45.1	LGSN	CCDS4964.1	c.1204G>A	p.G402S	missense	T
1432	PD9415	PMF	JAK2	20	31023261	A	T	55	0.0	71	43.7	ASXL1	CCDS13201.1	c.2746A>T	p.R916*	nonsense	T
1433	PD9415	PMF	JAK2	5	110439576	G	A	132	0.0	56	42.9	WDR36	CCDS4102.1	c.857G>A	p.R286H	missense	T
1434	PD9415	PMF	JAK2	9	5073770	G	T	179	0.0	119	41.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1435	PD9415	PMF	JAK2	8	67478976	A	G	204	0.5	121	40.5	MYBL1	CCDS47867.1	c.1893T>C	p.N631N	silent	T
1436	PD9415	PMF	JAK2	19	2403080	G	A	14	0.0	13	38.5	TMPRSS9	CCDS12088.1	c.455G>A	p.G152D	missense	T
1437	PD9415	PMF	JAK2	14	23586984	C	T	41	0.0	34	38.2	CEBPE	CCDS9589.1	c.558G>A	p.A186A	silent	T
1438	PD9415	PMF	JAK2	1	205689762	_	delT	74	0.0	43	37.2	NUCKS1	CCDS30987.1	c.249delA	p.E84fs*54	frameshift	T
1439	PD9416	MPN_u	JAK2	19	45923652	C	T	17	0.0	17	58.8	ERCC1	CCDS12663.1	c.355G>A	p.V119M	missense	T
1440	PD9416	MPN_u	JAK2	5	126753474	T	C	44	0.0	29	58.6	MEGF10	CCDS4142.1	c.1275T>C	p.T425T	silent	T
1441	PD9416	MPN_u	JAK2	14	50625345	A	C	116	0.0	111	52.3	SOS2	CCDS9697.1	c.1872T>G	p.T624T	silent	T
1442	PD9416	MPN_u	JAK2	9	5073770	G	T	148	0.0	131	51.2	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1443	PD9416	MPN_u	JAK2	4	53468066	C	T	55	0.0	76	43.4	USP46	CCDS47053.1	c.877G>A	p.D293N	missense	T
1444	PD9416	MPN_u	JAK2	8	18413811	G	A	132	0.0	114	41.2	PSD3	CCDS43720.1	c.2836C>T	p.L946L	silent	T
1445	PD9416	MPN_u	JAK2	4	106888582	G	A	33	0.0	23	34.8	NPNT	CCDS3796.1	c.1586G>A	p.R529Q	missense	T
1446	PD9416	MPN_u	JAK2	4	106157814	_	insA	66	0.0	54	33.3	TET2	CCDS47120.1	c.2715_2716insA	p.M906fs*18	frameshift	T
1447	PD9416	MPN_u	JAK2	9	136596438	G	A	14	0.0	24	33.3	SARDH	CCDS6978.1	c.679C>T	p.R227*	nonsense	T
1448	PD9416	MPN_u	JAK2	2	25141256	G	A	14	0.0	24	33.3	ADCY3	CCDS1715.1	c.601C>T	p.H201Y	missense	T
1449	PD9416	MPN_u	JAK2	16	70913204	G	A	40	0.0	39	28.2	HYDIN2	CCDS42189.1	c.10550C>T	p.A3517V	missense	T
1450	PD9416	MPN_u	JAK2	17	67305421	T	C	77	0.0	92	22.8	ABCA5	CCDS11685.1	c.451A>G	p.I151V	missense	T
1451	PD9417	PMF	JAK2	15	85327544	G	A	61	0.0	56	30.4	ZNF592	CCDS32317.1	c.1638G>A	p.K546K	silent	T
1452	PD9417	PMF	JAK2	9	5073770	G	T	135	0.0	142	30.3	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1453	PD9417	PMF	JAK2	6	93965550	G	A	54	0.0	48	29.2	EPHA7	CCDS5031.1	c.2378C>T	p.T793I	missense	T
1454	PD9417	PMF	JAK2	2	217343018	A	C	111	0.9	108	25.0	SMARCAL1	CCDS2403.1	c.2621A>C	p.Y874S	missense	T

Variant No	PDID	MPN Subtype	JAK2/MPL mutation	Chr	Position	Ref base	Mut base	Germline reads	Germline allele burden (%)	Tumour reads	Tumour allele burden (%)	Gene	CCDS	DNA Change	Protein Change	Type of Change	Validation Method
1455	PD9417	PMF	JAK2	19	9346728	A	G	112	0.9	113	23.9	ENSG00000188314	ENST00000344049	c.594A>G	p.R198R	silent	T
1456	PD9417	PMF	JAK2	17	3370847	G	A	54	0.0	21	23.8	SPATA22	CCDS11027.1	c.45C>T	p.G15G	silent	T
1457	PD9417	PMF	JAK2	17	7577114	C	T	45	0.0	32	21.9	TP53	CCDS11118.1	c.824G>A	p.C275Y	missense	T
1458	PD9417	PMF	JAK2	X	129154967	A	G	48	0.0	40	10.0	BCORL1	CCDS14616.1	c.3449A>G	p.K1150R	missense	T
1459	PD9418	PMF	JAK2	4	106156035	_	insA	79	1.3	22	31.8	TET2	CCDS47120.1	c.936_937insA	p.T313fs*18	frameshift	M
1460	PD9418	PMF	JAK2	20	44520238	_	delCTG	11	3.0	28	25.0	CTSA	CCDS13385.2	c.85_87delCTG	p.L29delL	inframe	T
1461	PD9418	PMF	JAK2	1	109276151	T	A	71	1.4	48	20.8	FNDC7	CCDS44185.1	c.2137T>A	p.S713T	missense	T
1462	PD9418	PMF	JAK2	20	35884838	G	A	17	0.0	36	19.4	GHRH	CCDS13292.1	c.147C>T	p.S49S	silent	T
1463	PD9418	PMF	JAK2	18	22040794	T	C	153	2.0	126	18.3	HRH4	CCDS11887.1	c.102T>C	p.A34A	silent	T
1464	PD9418	PMF	JAK2	1	25785241	G	A	229	2.2	196	16.3	TMEM57	CCDS30638.1	c.1012G>A	p.A338T	missense	T
1465	PD9418	PMF	JAK2	8	609907	G	A	14	0.0	49	10.2	ENSG00000206100	ENST00000382819	c.56C>T	p.A19V	missense	T
1466	PD9418	PMF	JAK2	9	5073770	G	T	116	0.9	98	6.1	JAK2	CCDS6457.1	c.1849G>T	p.V617F	missense	T
1467	PD9419	PMF	unmutated	3	134085206	G	A	97	2.1	207	56.5	AMOTL2	CCDS33860.1	c.1105C>T	p.R369C	missense	T
1468	PD9419	PMF	unmutated	18	51750421	A	C	10	0.0	27	55.6	MBD2	CCDS11953.1	c.509T>G	p.L170R	missense	T
1469	PD9419	PMF	unmutated	12	3686127	T	C	150	0.0	135	55.6	PRMT8	CCDS8521.2	c.803T>C	p.V268A	missense	T
1470	PD9419	PMF	unmutated	11	100211267	G	A	200	1.5	177	52.5	CNTN5	ENST00000279463	c.2755G>A	p.V919I	missense	T
1471	PD9419	PMF	unmutated	3	121647917	C	G	181	0.0	140	52.1	SLC15A2	CCDS3007.1	c.1430C>G	p.T477S	missense	T
1472	PD9419	PMF	unmutated	6	27776252	C	A	56	1.8	41	51.2	HIST1H2AI	CCDS4626.1	c.265C>A	p.R89S	missense	T
1473	PD9419	PMF	unmutated	20	31022712	C	T	32	0.0	30	46.7	ASXL1	CCDS13201.1	c.2197C>T	p.Q733*	nonsense	T
1474	PD9419	PMF	unmutated	13	48955389	_	delC	46	0.0	24	41.7	RB1	CCDS31973.1	c.1505delC	p.T502fs*17	frameshift	T
1475	PD9419	PMF	unmutated	1	190068069	G	A	85	0.0	72	38.9	FAM5C	CCDS1373.1	c.1380C>T	p.C460C	silent	T
1476	PD9419	PMF	unmutated	16	57723866	C	A	39	0.0	32	34.4	GPR97	ENST00000327655	c.1064C>A	p.S355Y	missense	T
1477	PD9419	PMF	unmutated	20	57569190	A	G	56	0.0	45	33.3	TH1L	CCDS13473.1	c.1634A>G	p.Y545C	missense	T
1478	PD9419	PMF	unmutated	6	151907162	C	T	36	0.0	33	30.3	C6orf97	CCDS43515.1	c.1231C>T	p.L411L	silent	T
1479	PD9419	PMF	unmutated	7	148506443	C	T	168	0.0	125	14.4	EZH2	CCDS5891.1	c.2069G>A	p.R690H	missense	T
1480	PD9419	PMF	unmutated	6	31721211	G	A	45	0.0	35	14.3	MSH5	CCDS34409.1	c.997G>A	p.V333M	missense	T
1481	PD9419	PMF	unmutated	19	13054565	_	delGCAGA<42>AGGAG	23	0.0	18	16.7	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M T C
1482	PD9420	PMF	unmutated	14	23652114	C	A	119	0.0	89	47.2	SLC7A8	CCDS9590.1	c.10G>T	p.G4*	nonsense	T
1483	PD9420	PMF	unmutated	2	196737130	C	A	153	0.0	82	43.9	DNAH7	CCDS42794.1	c.6477G>T	p.L2159L	silent	T
1484	PD9420	PMF	unmutated	8	8239236	T	C	24	0.0	21	42.9	ENSG00000182319	CCDS43706.1	c.22A>G	p.N8D	missense	T
1485	PD9420	PMF	unmutated	11	55451219	C	T	345	0.0	234	42.7	OR4P1P	ENST00000345013	c.233C>T	p.T78M	missense	T
1486	PD9420	PMF	unmutated	13	53624898	_	delC	54	0.0	62	38.7	OLFM4	CCDS9440.1	c.1525delC	p.Q510fs*7	frameshift	T
1487	PD9420	PMF	unmutated	3	12963735	C	T	18	0.0	21	28.6	IQSEC1	CCDS33703.1	c.1780G>A	p.V594M	missense	T
1488	PD9420	PMF	unmutated	11	108143537	A	G	82	0.0	63	20.6	ATM	CCDS31669.1	c.3242A>G	p.N1081S	missense	T
1489	PD9423	ET	unmutated	19	13054565	_	delGCAGA<42>AGGAG	26	0.0	10	40.0	CALR	CCDS12288.1	c.1092_1143del52	p.L367fs*46	frameshift	M C
1490	PD9423	ET	unmutated	18	64178840	G	A	120	0.0	91	44.0	CDH19	CCDS11994.1	c.1541C>T	p.T514I	missense	T
1491	PD9423	ET	unmutated	10	104596833	G	A	41	0.0	32	31.3	CYP17A1	CCDS7541.1	c.286C>T	p.R96W	missense	T
1492	PD9423	ET	unmutated	11	32636148	G	A	143	0.0	66	21.2	CCDC73	CCDS41630.1	c.1716C>T	p.N572N	silent	T
1493	PD9423	ET	unmutated	14	96707595	C	T	17	0.0	35	20.0	BDKRB2	CCDS9942.1	c.930C>T	p.I310I	silent	T
1494	PD9423	ET	unmutated	7	11082383	C	A	194	0.0	130	15.4	PHF14	CCDS47541.1	c.2254C>A	p.L752I	missense	T
1495	PD9423	ET	unmutated	2	10104439	C	T	40	0.0	60	15.0	GRHL1	CCDS33144.1	c.412C>T	p.R138W	missense	T
1496	PD9423	ET	unmutated	16	68847239	C	T	134	0.0	117	12.0	CDH1	CCDS10869.1	c.1161C>T	p.N387N	silent	T
1497	PD9423	ET	unmutated	17	10402008	A	C	71	0.0	95	11.6	MYH1	CCDS11155.1	c.4116T>G	p.V1372V	silent	T
1498	PD9423	ET	unmutated	21	45798945	C	T	27	0.0	38	10.5	TRPM2	CCDS13710.1	c.1080C>T	p.A360A	silent	T

JAK2/MPL status was defined by source data and confirmed by exome sequencing. Valdiation method; T (custom bait capture and targetted resequencing), M (manual curation), C (Sanger sequencing).

Table S3. Significance of recurrently mutated genes in 151 MPN samples.

gene_name	cds_ID	n_syn	n_mis	n_non	wMIS	wNON	pMIS	pNON	qMIS	qNON	qSPLICE	qUNIFMUT
TET2	CCDS47120	1.0000	4.0000	8.0000	43.2281	1122.9864	0.0000	0.0000	0.0078	0.0000	1.0000	1.0000
ASXL1	CCDS13201	0.0000	0.0000	6.0000	0.0000	1141.5622	0.6827	0.0000	1.0000	0.0000	1.0000	1.0000
PHIP	CCDS4987	0.0000	0.0000	2.0000	0.0000	242.2355	0.6761	0.0000	1.0000	0.1505	1.0000	1.0000
JAK2	CCDS6457	0.0000	112.0000	0.0000	2171.6949	0.0000	0.0000	0.9252	0.0000	1.0000	1.0000	1.0000
DNMT3A	CCDS33157	0.0000	11.0000	0.0000	185.1983	0.0000	0.0000	0.9365	0.0000	1.0000	1.0000	1.0000
MPL	CCDS483	0.0000	7.0000	0.0000	208.9336	0.0000	0.0000	0.9369	0.0000	1.0000	1.0000	1.0000
U2AF1	CCDS13694	0.0000	4.0000	0.0000	224.9228	0.0000	0.0000	0.9648	0.0000	1.0000	1.0000	1.0000
TP53	CCDS11118	0.0000	3.0000	0.0000	130.2332	0.0000	0.0000	0.9588	0.0057	1.0000	1.0000	1.0000
CHEK2	CCDS33629	0.0000	3.0000	0.0000	105.0720	0.0000	0.0000	0.9497	0.0079	1.0000	1.0000	1.0000
EZH2	CCDS5891	0.0000	3.0000	0.0000	77.3616	0.0000	0.0000	0.9402	0.0179	1.0000	1.0000	1.0000
SF3B1	CCDS33356	0.0000	3.0000	0.0000	47.2070	0.0000	0.0000	0.9179	0.0733	1.0000	1.0000	1.0000
PHF6	CCDS14639	0.0000	2.0000	0.0000	122.8925	0.0000	0.0001	0.9494	0.1873	1.0000	1.0000	1.0000
PRKACB	CCDS693	0.0000	2.0000	0.0000	112.9548	0.0000	0.0001	0.9569	0.2033	1.0000	1.0000	1.0000
IDH1	CCDS2381	0.0000	2.0000	0.0000	95.8897	0.0000	0.0002	0.9573	0.2366	1.0000	1.0000	1.0000
SRSF2	CCDS11749	0.0000	2.0000	0.0000	93.6919	0.0000	0.0002	0.9668	0.2366	1.0000	1.0000	1.0000
ZNF485	CCDS7205	0.0000	2.0000	0.0000	95.0616	0.0000	0.0002	0.9540	0.2366	1.0000	1.0000	1.0000
FARS2	CCDS4494	0.0000	2.0000	0.0000	82.5813	0.0000	0.0002	0.9547	0.2880	1.0000	1.0000	1.0000
GABRB3	CCDS10018	0.0000	2.0000	0.0000	78.6749	0.0000	0.0002	0.9525	0.2990	1.0000	1.0000	1.0000
EZH1	CCDS32659	0.0000	2.0000	0.0000	52.8312	0.0000	0.0005	0.9388	0.6308	1.0000	1.0000	1.0000
DTNA	CCDS42425	0.0000	2.0000	0.0000	52.1854	0.0000	0.0006	0.9429	0.6308	1.0000	1.0000	1.0000
TCF4	CCDS58631	0.0000	2.0000	0.0000	49.6090	0.0000	0.0006	0.9415	0.6649	1.0000	1.0000	1.0000
IL6ST	CCDS3971	0.0000	2.0000	0.0000	48.2155	0.0000	0.0007	0.9353	0.6707	1.0000	1.0000	1.0000
ZNF473	CCDS33077	0.0000	2.0000	0.0000	43.7881	0.0000	0.0008	0.9386	0.7825	1.0000	1.0000	1.0000
EPHA7	CCDS5031	0.0000	2.0000	0.0000	41.0193	0.0000	0.0009	0.9317	0.8571	1.0000	1.0000	1.0000
HUWE1	CCDS35301	0.0000	2.0000	1.0000	8.8069	67.0359	0.0232	0.0112	1.0000	1.0000	1.0000	1.0000
SARDH	CCDS6978	0.0000	1.0000	1.0000	16.2575	313.1889	0.0544	0.0021	1.0000	1.0000	1.0000	1.0000
MPHOSPH10	CCDS1916	1.0000	1.0000	1.0000	32.1569	394.6692	0.0253	0.0016	1.0000	1.0000	1.0000	1.0000
MUC16	CCDS54212	3.0000	3.0000	0.0000	4.5380	0.0000	0.0360	0.8060	1.0000	1.0000	1.0000	1.0000
HECW1	CCDS5469	0.0000	2.0000	0.0000	21.0022	0.0000	0.0038	0.9114	1.0000	1.0000	1.0000	1.0000
TG	CCDS34944	1.0000	2.0000	0.0000	13.2801	0.0000	0.0099	0.8834	1.0000	1.0000	1.0000	1.0000
SYNE2	CCDS9761	1.0000	2.0000	0.0000	6.3895	0.0000	0.0443	0.8180	1.0000	1.0000	1.0000	1.0000
ASPM	CCDS1389	0.0000	2.0000	0.0000	12.5545	0.0000	0.0112	0.8633	1.0000	1.0000	1.0000	1.0000
ABCC9	CCDS8693	0.0000	2.0000	0.0000	27.1517	0.0000	0.0022	0.9194	1.0000	1.0000	1.0000	1.0000
FAT2	CCDS4317	0.0000	2.0000	0.0000	8.9551	0.0000	0.0224	0.8771	1.0000	1.0000	1.0000	1.0000
SI	CCDS3196	0.0000	2.0000	0.0000	23.7487	0.0000	0.0029	0.9035	1.0000	1.0000	1.0000	1.0000
DICER1	CCDS9931	0.0000	2.0000	0.0000	22.0816	0.0000	0.0034	0.9084	1.0000	1.0000	1.0000	1.0000
COL1A1	CCDS11561	0.0000	2.0000	0.0000	20.7844	0.0000	0.0039	0.9216	1.0000	1.0000	1.0000	1.0000
ARHGAP32	CCDS44769	0.0000	2.0000	0.0000	18.7072	0.0000	0.0049	0.9082	1.0000	1.0000	1.0000	1.0000
FBN2	CCDS34222	0.0000	2.0000	0.0000	12.5647	0.0000	0.0111	0.8859	1.0000	1.0000	1.0000	1.0000
MUC4	CCDS54700	0.0000	2.0000	0.0000	6.9626	0.0000	0.0373	0.8906	1.0000	1.0000	1.0000	1.0000
SEC16A	CCDS55351	0.0000	2.0000	0.0000	14.4955	0.0000	0.0083	0.9050	1.0000	1.0000	1.0000	1.0000
ATM	CCDS31669	0.0000	2.0000	0.0000	14.7026	0.0000	0.0080	0.8768	1.0000	1.0000	1.0000	1.0000
GRIN2B	CCDS8662	0.0000	2.0000	0.0000	23.7904	0.0000	0.0029	0.9275	1.0000	1.0000	1.0000	1.0000
SACS	CCDS9300	0.0000	2.0000	0.0000	9.7036	0.0000	0.0190	0.8582	1.0000	1.0000	1.0000	1.0000
PLEKHG4B	CCDS34124	0.0000	2.0000	0.0000	26.2325	0.0000	0.0024	0.9280	1.0000	1.0000	1.0000	1.0000
CACNA2D3	CCDS54598	0.0000	2.0000	0.0000	34.7681	0.0000	0.0013	0.9316	1.0000	1.0000	1.0000	1.0000
SVEP1	CCDS48004	0.0000	2.0000	0.0000	10.8832	0.0000	0.0150	0.8749	1.0000	1.0000	1.0000	1.0000
PDZRN4	CCDS53777	0.0000	2.0000	0.0000	32.0658	0.0000	0.0016	0.9276	1.0000	1.0000	1.0000	1.0000
KSR2		0.0000	2.0000	0.0000	34.6530	0.0000	0.0013	0.9393	1.0000	1.0000	1.0000	1.0000
SGK223	CCDS43706	0.0000	2.0000	0.0000	22.5478	0.0000	0.0033	0.9311	1.0000	1.0000	1.0000	1.0000
CSMD1	CCDS55189	0.0000	2.0000	0.0000	10.4146	0.0000	0.0164	0.8746	1.0000	1.0000	1.0000	1.0000
FAT4	CCDS3732	0.0000	2.0000	0.0000	7.9911	0.0000	0.0282	0.8700	1.0000	1.0000	1.0000	1.0000
TDRD15		0.0000	2.0000	0.0000	24.3721	0.0000	0.0028	0.9085	1.0000	1.0000	1.0000	1.0000
USH2A	CCDS31025	1.0000	1.0000	0.0000	3.9756	0.0000	0.2610	0.8492	1.0000	1.0000	1.0000	1.0000
DNMT3B	CCDS13205	1.0000	1.0000	0.0000	19.3668	0.0000	0.0447	0.9287	1.0000	1.0000	1.0000	1.0000
CIT	CCDS55891	1.0000	1.0000	0.0000	8.7558	0.0000	0.1091	0.8975	1.0000	1.0000	1.0000	1.0000
ANO3	CCDS31447	1.0000	1.0000	0.0000	21.2902	0.0000	0.0402	0.9271	1.0000	1.0000	1.0000	1.0000
DYSF	CCDS46328	1.0000	1.0000	0.0000	8.1101	0.0000	0.1188	0.9097	1.0000	1.0000	1.0000	1.0000
CENPE	CCDS34042	1.0000	1.0000	0.0000	8.9780	0.0000	0.1060	0.8851	1.0000	1.0000	1.0000	1.0000

MYO10	CCDS54834	1.0000	1.0000	0.0000	8.7128	0.0000	0.1097	0.8992	1.0000	1.0000	1.0000	1.0000
LANCL3	CCDS55398	1.0000	1.0000	0.0000	34.3428	0.0000	0.0235	0.9581	1.0000	1.0000	1.0000	1.0000
TTN	CCDS54424	2.0000	1.0000	0.0000	0.6201	0.0000	0.6037	0.6310	1.0000	1.0000	1.0000	1.0000
SOGA2	CCDS11841	1.0000	1.0000	0.0000	10.1826	0.0000	0.0921	0.9164	1.0000	1.0000	1.0000	1.0000
SYNM		1.0000	1.0000	0.0000	9.9696	0.0000	0.0943	0.9169	1.0000	1.0000	1.0000	1.0000
PEG3	CCDS12948	1.0000	1.0000	0.0000	12.2464	0.0000	0.0748	0.9177	1.0000	1.0000	1.0000	1.0000
DMD	CCDS14233	2.0000	1.0000	0.0000	5.8133	0.0000	0.1721	0.8555	1.0000	1.0000	1.0000	1.0000
PCDHA3	CCDS54915	1.0000	1.0000	0.0000	16.1985	0.0000	0.0546	0.9422	1.0000	1.0000	1.0000	1.0000
CDK11A	CCDS44042	2.0000	0.0000	0.0000	0.0000	0.0000	0.7561	0.9385	1.0000	1.0000	1.0000	1.0000
PCM1	CCDS47812	2.0000	0.0000	0.0000	0.0000	0.0000	0.6685	0.9010	1.0000	1.0000	1.0000	1.0000
MYH1	CCDS11155	2.0000	0.0000	0.0000	0.0000	0.0000	0.6606	0.9107	1.0000	1.0000	1.0000	1.0000
PRKAG3	CCDS2424	2.0000	0.0000	0.0000	0.0000	0.0000	0.8147	0.9599	1.0000	1.0000	1.0000	1.0000
NPAS4	CCDS8138	2.0000	0.0000	0.0000	0.0000	0.0000	0.7707	0.9462	1.0000	1.0000	1.0000	1.0000

Gene	CDSlength	Indels	wIND	Pval	Qval
TET2	6049	11.0000	632.3250	0.0000	0.0000
ASXL1	4686	7.0000	519.4300	0.0000	0.0000
CALR	1294	25.0000	6717.9537	0.0000	0.0000

Genes with mutations in greater than 1 MPN sample are shown. Top table shows significance of missense, nonsense and splice site mutations. Lower panel shows insertion/deletion variants. Green shading highlights significantly mutated genes. Dark green, q<0.05; Light green, q<0.20.

Table S4. Clinical characteristics of *JAK2* and *CALR* mutated essential thrombocythemia and myelofibrosis.

	JAK2 mutated	*CALR* mutated	Significance
Essential thrombocythemia			
Number of patients	45	21	
Age (years, median, range)	60 (21-87)	60 (32-85)	0.5
Female (median, %)	24 (53)	10 (48)	0.8
Hemoglobin (g/L, mean, SD)	142 (14)	132 (16)	**0.02***
White-cell count (10^9/L, mean, SD)	10.4 (5.3)	8.3 (2.2)	0.2
Platelet count (10^9/L, mean, SD)	833 (300)	1193 (504)	**0.0003*****
Disease follow up (years, median, range)	7 (1-21)	7 (1-21)	0.5
Complications			
Thrombosis (%)	10 (22)	5 (24)	1
Transformation to myelofibrosis (%)	1 (2)	4 (19)	**0.03***
Transformation to polycythemia vera (%)	7 (16)	0 (0)	0.09
Death	3 (7)	0 (0)	0.5
Therapy with hydroxyurea (%)	34 (76)	17 (81)	0.8
Therapy with anagrelide (%)	7 (16)	6 (29)	0.3
Myelofibrosis			
Number of patients	20	5	
Age (years, median, range)	63 (41-84)	66 (41-70)	0.9
Female (%)	8 (40)	0	0.1
Hemoglobin (g/L, mean, SD)	116 (23)	107 (15)	0.4
White-cell count (10^9/L)	11.5 (9.0)	6.9 (4.5)	0.1
Platelet count (10^9/L, mean, SD)	275 (150)	201 (116)	0.4
Disease follow up (years, median, range)	4 (1-14)	11 (5-16)	**0.02***
Complications			
Thrombosis (%)	4 (20)	2 (40)	0.6
Transformation to acute leukemia (%)	1(5)	0	1
Death	3 (15)	1 (20)	1

Clinical characteristics of patients with essential thromobocythemia (ET), primary myelofibrosis and post-ET myelofibrosis from the exome sequencing cohort are shown. Average values for blood indices are shown from the time of diagnosis. Diagnostic blood counts and follow up information were not available in 2 and 1 patient respectively. Data refer to number of patients (%), unless otherwise stated. The large number of post ET transformations to polycythaemia vera were due to specific inclusion in the study. * $p<0.05$; ** $p<0.01$; *** $p<0.001$

Table S5. Cell lines screened for mutations in *CALR* exon 9

GDSC description	Number of cell lines	Name
acute_myeloid_leukemia	27	CESS, CMK, CTV-1, GDM-1, HEL, HL-60, KASUMI-1, KG-1, KMOE-2, KO52, ME-1, ML-2, MOLM-13, MONO-MAC-6, NB-4, NKM-1, NOMO-1, OCI-AML2, OCI-AML3, OCI-AML5, OCI-M1, P31-FUJ, PL-21, QIMR-WIL, SIG-M5, THP-1, MOLM-16
adrenal_gland	1	
anaplastic_large_cell_lymphoma	3	
B_cell_leukemia	13	
B_cell_lymphoma	35	
biliary_tract	5	
Bladder	20	
bone_other	3	
breast	52	
Burkitt_lymphoma	13	
cervix	15	
chondrosarcoma	5	
chronic_myeloid_leukaemia	10	
digestive_system_other	1	
endometrium	11	
ewings_sarcoma	22	
fibrosarcoma	3	
glioma	53	
hematopoietic_neoplasm other	6	AMO-1, EoL-1-cell, JJN-3, KARPAS-620, MC-CAR, SKM-1
hairy_cell_leukaemia	4	
head and neck	45	
Hodgkin_lymphoma	9	
kidney	34	
large_intestine	52	
leukemia	4	KY821, MV-4-11, MY-M12, RS4-11
liver	17	
lung_NSCLC_adenocarcinoma	68	
lung_NSCLC_carcinoid	4	
lung_NSCLC_large cell	14	
lung_NSCLC_not specified	11	
lung_NSCLC_squamous_cell_carcinoma	15	
Lung_other	1	
lung_small_cell_carcinoma	66	
lymphoblastic_leukemia	12	
lymphoblastic_T_cell_leukaemia	9	
lymphoid_neoplasm other	11	
medulloblastoma	4	
melanoma	59	
mesothelioma	21	
myeloma	13	
nervous_system_other	1	
neuroblastoma	34	
oesophagus	35	
osteosarcoma	10	
ovary	43	
pancreas	32	
prostate	8	
rhabdomyosarcoma	9	
skin_other	3	
soft_tissue_other	9	
stomach	29	
T_cell_leukemia	4	
testis	3	
thyroid	17	
urogenital_system_other	4	
uterus	3	
Total	**1015**	

GDSC - Genome damage and stability centre database. Specific cell lines tested have been listed for hematopoietic groups.

Table S6. *CALR* mutations in exome and follow up cohorts.

Study	Sample	Start(bp)	End (bp)	Length (bp)	Mutation type	DNA change	Hematological disorder	Protein
Exome	PD10056	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Exome	PD4063	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Exome	PD4996	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Exome	PD5003	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Exome	PD5004	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Exome	PD5005	13054575	13054608	34	Deletion	aagga<24>agagg	PMF	p.K368fs*51
Exome	PD5014	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Exome	PD6549	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD6561	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD6563	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Exome	PD6566	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD6573	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD6597	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD6602	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD6628	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD7438	13054569	13054602	34	Deletion	aggct<24>acgca	ET	p.R366fs*53
Exome	PD7440	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD7442	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD7443	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Exome	PD8632	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD8637	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Exome	PD8642	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Exome	PD8942	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Exome	PD8943	13054627	13054628	5	Insertion	TTGTC	PET-MF	p.K385fs*47
Exome	PD9419	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Exome	PD9423	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	CU2258	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	E2737	13054565	13054616	52	Deletion	gcaga<42>aggag	aCML	p.L367fs*46
Follow_up	ED86	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	IF0903	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	JC1148	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	JM83	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	JY91	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	KT03_AML_phase	13054627	13054628	5	Insertion	TTGTC	AML	p.K385fs*47
Follow_up	KT03_PMF_phase	13054627	13054628	5	Insertion	TTGTC	PMF	p.K385fs*47
Follow_up	LT99	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	MD08	13054575	13054608	34	Deletion	aagga<24>agagg	ET	p.K368fs*51
Follow_up	MP69_AML_phase	13054627	13054628	5	Insertion	TTGTC	AML	p.K385fs*47
Follow_up	MP69_ET_phase	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	SD38	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10095c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10105c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10155c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10156c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10160c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10225c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10250c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10313c	13054627	13054628		Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10328c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10368c	13054584	13054614	31	Deletion	gaaga<21>ggagg	ET	p.E371fs*49
Follow_up	WGA-PD10369c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10390c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10397d	13054573	13054633	61	Deletion	ttaag<51>gagga	ET	p.K368fs*43
Follow_up	WGA-PD10433c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10445c	13054568	13054613	46	Deletion	gaggc<36>aggag	ET	p.L367fs*48
Follow_up	WGA-PD10505c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10506c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10514c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10517c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10522c	13054582	13054618	37	Deletion	aggaa<27>gaggc	ET	p.E371fs*47
Follow_up	WGA-PD10538c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10574c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10595c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10611c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10626c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD10671c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD10676c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD11836a	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Follow_up	WGA-PD11838a	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Follow_up	WGA-PD11852a	13054627	13054628	5	Insertion	GTGTC	PMF	p.K385fs*47
Follow_up	WGA-PD11879a	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Follow_up	WGA-PD11882a	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Follow_up	WGA-PD11889a	13054627	13054628	5	Insertion	TTGTC	PET-MF	p.K385fs*47
Follow_up	WGA-PD11898a	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Follow_up	WGA-PD11901a	13054582	13054633	52	Deletion	aggaa<42>gagga	PMF	p.E370fs*42
Follow_up	WGA-PD11902a	13054627	13054628	5	Insertion	TTGTC	PMF	p.K385fs*47
Follow_up	WGA-PD11920a	13054571	13054604	34	Deletion	gctta<24>gcaaa	PMF	p.L367fs*52
Follow_up	WGA-PD11927a	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Follow_up	WGA-PD11935a	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Follow_up	WGA-PD11939a	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46

Study	Sample	Start(bp)	End (bp)	Length (bp)	Mutation type	DNA change	Hematological disorder	Protein
Follow_up	WGA-PD11950a	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Follow_up	WGA-PD11951a	13054627	13054628	5	Insertion	TTGTC	PET-MF	p.K385fs*47
Follow_up	WGA-PD11971a	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Follow_up	WGA-PD11988a	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Follow_up	WGA-PD11994a	13054627	13054628	5	Insertion	TTGTC	PMF	p.K385fs*47
Follow_up	WGA-PD12001a	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Follow_up	WGA-PD12005a	13054627	13054628	5	Insertion	TTGTC	PET-MF	p.K385fs*47
Follow_up	WGA-PD12006a	13054565	13054616	52	Deletion	gcaga<42>aggag	PET-MF	p.L367fs*46
Follow_up	WGA-PD12012a	13054581	13054617	37	Deletion	gagga<27>ggagg	PET-MF	p.E370fs*48
Follow_up	WGA-PD12281a	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD12285a	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD12786a	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD12789a	13054616	13054628	4	Complex	ggcagaggacaa_TCCTTGTC	ET	p.E381fs*48
Follow_up	WGA-PD12904a	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Follow_up	WGA-PD12914a	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD12919a	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Follow_up	WGA-PD12934a	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD12940a	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD12941a	13054627	13054628	5	Insertion	TTGTC	PMF	p.K385fs*47
Follow_up	WGA-PD12943a	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Follow_up	WGA-PD12956a	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD12980a	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD12982a	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD12985a	13054565	13054616	52	Deletion	gcaga<42>aggag	PMF	p.L367fs*46
Follow_up	WGA-PD12986a	13054568	13054613	46	Deletion	gaggc<36>aggag	ET	p.L367fs*48
Follow_up	WGA-PD13007a	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD13019a	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD13028a	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD13039a	13054599	13054602	2	Complex	TTTGC_gca	PMF	p.R376fs*55
Follow_up	WGA-PD13040a	13054627	13054628	5	Insertion	TTGTC	PMF	p.K385fs*47
Follow_up	WGA-PD13042a	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5292h	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5323c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5324c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5331c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5349c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5361c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5363c	13054580	13054649	70	Deletion	ggagg<60>aagat	ET	p.E370fs*37
Follow_up	WGA-PD5364c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5373c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5377c	13054575	13054608	34	Deletion	aagga<24>agagg	ET	p.K368fs*51
Follow_up	WGA-PD5388d	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5395c	13054625	13054628	5	Complex	TCTTGTC_aa	ET	p.K385fs*47
Follow_up	WGA-PD5397c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5402c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5407c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5412c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5415c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5430c	13054588	13054618	31	Deletion	aagac<21>gaggc	ET	p.D373fs*48
Follow_up	WGA-PD5435c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5437c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5446c	13054593	13054602	10	Deletion	aagaa<0>acgca	ET	p.K374fs*53
Follow_up	WGA-PD5448c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5451c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5455c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD5457c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5458c	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47
Follow_up	WGA-PD5477c	13054565	13054616	52	Deletion	gcaga<42>aggag	ET	p.L367fs*46
Follow_up	WGA-PD6260a	13054565	13054616	52	Deletion	gcaga<42>aggag	RA	p.L367fs*46
Follow_up	WGA-PD6265a	13054565	13054616	52	Deletion	gcaga<42>aggag	RAEB	p.L367fs*46
Follow_up	WGA-PD6295a	13054627	13054628	5	Insertion	TTGTC	RARS	p.K385fs*47
Follow_up	WGA-PD6296a	13054565	13054616	52	Deletion	gcaga<42>aggag	RARS	p.L367fs*46
Follow_up	WGA-PD6308a	13054627	13054628	5	Insertion	TTGTC	RAEB	p.K385fs*47
Follow_up	WGA-PD6313a	13054627	13054628	5	Insertion	TTGTC	RARS	p.K385fs*47
Follow_up	WGA-PD6476a	13054568	13054628	61	Deletion	gaggc<51>acaag	RA	p.L367fs*44
Follow_up	WGA-PD6484a	13054627	13054628	5	Insertion	TTGTC	CMML	p.K385fs*47
Follow_up	WGA-PD6503a	13054565	13054616	52	Deletion	gcaga<42>aggag	RA	p.L367fs*46
Follow_up	WGA-PD6506a	13054627	13054628	5	Insertion	TTGTC	RA	p.K385fs*47
Follow_up	WGA-PD6515a	13054565	13054616	52	Deletion	gcaga<42>aggag	RA	p.L367fs*46
Follow_up	WGA-PD6575e	13054627	13054628	5	Insertion	TTGTC	ET	p.K385fs*47

All *CALR* mutations identified in the exome sequencing cohort and follow up study are listed. For deletions, base pair positions are of the first and last nucleotide of the deleted sequence; for insertions the base pair positions are of nucleotides between which the insertion occurs. 'DNA change' shows the exact nucleotide change that occurs with deleted nucleotides in lower case and inserted nucleotides in upper case. ET, essential thrombocythemia; PET-MF, post-ET myelofibrosis; PMF, primary myelofibrosis; RA, refractory anemia; RARS, refractory anemia with ringed sideroblasts; RAEB, refractory anemia with excess blasts; CMML, chronic myelomonocytic leukemia; aCML, atypical chronic myeloid leukemia.