**Supplemental Table 3. Mutations detected by next-generation sequencing in the training cohort of 301 PV patients.**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Patient ID | Gene | Nucleotide nomenclature | Protein consequence | VAF % | dbSNP rs |
| PV1 | JAK2exon14 | c.1849G>T | p.V617F | 46.8 | rs77375493 |
| PV2 | JAK2exon14 | c.1849G>T | p.V617F | 92.4 | rs77375493 |
| PV3 | JAK2exon14 | c.1849G>T | p.V617F | 57.1 | rs77375493 |
| PV4 | JAK2exon14 DNMT3Aexon22 TET2exon10 | c.1849G>T c.2578T>C c.4210C>T | p.V617F p.W860R p.R1404X | 63.5 3.7 42.8 | rs77375493 rs373014701 — |
| PV5 | JAK2exon14 | c.1849G>T | p.V617F | 21.9 | rs77375493 |
| PV6 | JAK2Exon14 ASXL1Exon12 | c.1849G>T c.3187C>T | p.V617F p.Q1063X | 21.5 5 | rs77375493 — |
| PV7 | JAK2exon14 TET2exon6 | c.1849G>T c.3732\_3733del | p.V617F p.Y1245Lfs\*22 | 90.9 47.8 | rs77375493 — |
| PV8 | JAK2exon14 TET2exon3 | c.1849G>T c.1954C>T | p.V617F p.Q652X | 64.1 11.6 | rs77375493 — |
| PV9 | JAK2exon14 | c.1849G>T | p.V617F | 34.6 | rs77375493 |
| PV10 | JAK2exon14 | c.1849G>T | p.V617F | 62.5 | rs77375493 |
| PV11 | JAK2exon14 | c.1849G>T | p.V617F | 35.7 | rs77375493 |
| PV12 | JAK2exon14 DNMT3Aexon23 | c.1849G>T c.2644C>T | p.V617F p.R882C | 32.1 0.58 | rs77375493 rs377577594 |
| PV13 | JAK2exon14 DNMT3Aexon 23 | c.G1849T c.G2645A | p.V617F p.R882H | 84.48 44.78 | rs77375493 rs147001633 |
| PV14 | JAK2exon14 ASXL1exon12 ASXL1exon7 | c.1849G>T c.4285\_4287del c.686C>T | p.V617F p.S1429del p.P229L | 40.1 49.7 49.5 | rs77375493 — rs576523117 |
| PV15 | JAK2exon14 | c.1849G>T | p.V617F | 35.3 | rs77375493 |
| PV16 | JAK2exon12 BCORL1exon3 | c.1624\_1629delAATGAA c.2615T>G | p.N542\_E543delNE p.V872G | 45.7 99.7 | — rs750640150 |
| PV17 | JAK2exon14 ASXL1exon12 DNMT3Aexon19 | c.1849G>T c.2757dupA c.2254T>A | p.V617F p.P920Tfs\*4 p.F752I | 44.6 23 19 | rs77375493 rs771822198 rs776844126 |
| PV18 | JAK2exon14 BCORexon4 | c.1849G>T c.935\_937delAGC | p.V617F p.Q312delQ | 49.4 44.9 | rs77375493 rs3840969 |
| PV19 | JAK2exon14 | c.1849G>T | p.V617F | 83 | rs77375493 |
| PV20 | JAK2exon14 BCORexon13 ASXL1exon12 | c.1849G>T c.4664G>A c.1773C>G | p.V617F p.R1555H p.Y591\* | 59.5 48.3 11.6 | rs77375493 rs367859441 rs371369583 |
| PV21 | JAK2exon14 | c.G1849T | p.V617F | 76.17 | rs77375493 |
| PV22 | JAK2exon14 BCORL1exon7 ASXL1exon12 | c.1849G>T c.4171G>A c.1998\_2011del14 | p.V617F p.G1391R p.D667Lfs\*5 | 79.6 46.6 41.5 | rs77375493 rs199973665 — |
| PV23 | JAK2exon14 NRASexon 2 | c.1849G>T c.G35A | p.V617F p.G12D | 95.77 39.07 | rs77375493 rs121913237 |
| PV24 | JAK2exon14 | c.1849G>T | p.V617F | 86.8 | rs77375493 |
| PV25 | JAK2exon14 TP53exon6 BRCA2exon11 | c.1849G>T c.659A>G c.2950dupG | p.V617F p.Y220C p.E984Gfs\*4 | 61.2 2.1 44.4 | rs77375493 rs121912666 — |
| PV26 | JAK2exon14 | c.1849G>T | p.V617F | 39.2 | rs77375493 |
| PV27 | JAK2exon14 | c.1849G>T | p.V617F | 43.9 | rs77375493 |
| PV28 | JAK2exon14 | c.1849G>T | p.V617F | 61.6 | rs77375493 |
| PV29 | JAK2exon14 ASXL1exon12 | c.1849G>T c.3098A>T | p.V617F p.E1033V | 59 51 | rs77375493 rs192330235 |
| PV30 | JAK2exon14 BCORexon12 | c.1849G>T c.4567A>T | p.V617F p.T1523S | 65 38.7 | rs77375493 — |
| PV31 | JAK2exon14 ASXL1exon12 | c.1849G>T c.2254delG | p.V617F p.A752Lfs\*20 | 54.1 30.3 | rs77375493 — |
| PV32 | JAK2Exon14 DNMT3AExon15 | c.1849G>T c.1792C>T | p.V617F p.R598X | 16.2 39.3 | rs77375493 rs568207978 |
| PV33 | JAK2Exon14 | c.1849G>T | p.V617F | 38.5 | rs77375493 |
| PV34 | JAK2exon14 ASXL1exon12 TET2exon6 | c.1849G>T c.1772dupA c.3764dupA | p.V617F p.Y591\*fs\*1 p.Y1255\*fs\*1 | 46.6 47.4 46.7 | rs77375493 rs762036456 rs756805589 |
| PV35 | JAK2exon14 TET2exon6 DNMT3Aexon14 | c.1849G>T c.3781C>T c.1627G>T | p.V617F p.R1261C p.G543C | 25.2 1.1 41.1 | rs77375493 — — |
| PV36 | JAK2exon14 BCORL1exon3 | c.1849G>T c.2669G>A | p.V617F p.R890Q | 6.1 48.8 | rs77375493 rs201843717 |
| PV37 | JAK2exon14 DNMT3Aexon8 | c.1849G>T c.867delC | p.V617F p.G289fs | 86.5 42.9 | rs77375493 — |
| PV38 | JAK2exon14 DNMT3Aexon20 BCORL1exon3 | c.1849G>T c.2351\_2354dup c.2669G>A | p.V617F p.A787Vfs\*12 p.R890Q | 25.6 19.7 51.7 | rs77375493 — rs201843717 |
| PV39 | JAK2exon14 ASXL1exon12 | c.1849G>T c.3098A>T | p.V617F p.E1033V | 8.4 52.9 | rs77375493 rs192330235 |
| PV40 | JAK2exon14 | c.1849G>T | p.V617F | 71.7 | rs77375493 |
| PV41 | JAK2exon14 DNMT3Aexon14 PDS5Bexon20 | c.1849G>T c.1567G>T c.2164dupC | p.V617F p.E523\* p.R724Pfs\*16 | 81.6 1.6 45.7 | rs77375493 — rs746601412 |
| PV42 | JAK2exon14 TET2exon10 | c.1849G>T c.4263C>G | p.V617F p.Y1421\* | 88.5 46.4 | rs77375493 — |
| PV43 | JAK2exon14 DNMT3Aexon17 | c.1849G>T c.2080C>T | p.V617F p.H694Y | 72.8 1.8 | rs77375493 — |
| PV44 | JAK2exon14 TET2exon3 DNMT3Aexon19 | c.1849G>T c.2491delC c.2206C>T | p.V617F p.Q831Rfs\*10 p.R736C | 93.5 13 12.9 | rs77375493 — rs761934754 |
| PV45 | JAK2exon14 | c.1849G>T | p.V617F | 36.2 | rs77375493 |
| PV46 | JAK2exon14 | c.1849G>T | p.V617F | 86.4 | rs77375493 |
| PV47 | JAK2exon14 RBBP6exon18 JAK1exon6 ASXL1exon13 ASXL1exon13 | c.1849G>T c.5214dupC c.599\_600del c.2879G>A c.1927dupG | p.V617F p.K1739Qfs\*4 p.Y200Cfs\*27 p.W960\* p.G646Wfs\*11 | 95.5 35.1 43.1 8 33.3 | rs77375493 — — — rs1085307856 |
| PV48 | JAK2exon14 | c.1849G>T | p.V617F | 82.2 | rs77375493 |
| PV49 | JAK2exon14 BCORL1exon7 | c.1849G>T c.4171G>A | p.V617F p.G1391R | 49.2 100 | rs77375493 rs199973665 |
| PV50 | JAK2exon14 GNB1exon5 | c.1849G>T c.169A>G | p.V617F p.K57E | 56.9 11.1 | rs77375493 rs141326438 |
| PV51 | JAK2exon14 | c.1849G>T | p.V617F | 45.4 | rs77375493 |
| PV52 | JAK2exon14 | c.1849G>T | p.V617F | 39.9 | rs77375493 |
| PV53 | JAK2exon14 | c.1849G>T | p.V617F | 44.7 | rs77375493 |
| PV54 | JAK2exon14 | c.1849G>T | p.V617F | 53 | rs77375493 |
| PV55 | JAK2exon14 TET2exon11 | c.1849G>T c.5618T>C | p.V617F p.I1873T | 37.3 35.5 | rs77375493 rs116519313 |
| PV56 | JAK2exon14 | c.1849G>T | p.V617F | 87 | rs77375493 |
| PV57 | JAK2exon14 | c.1849G>T | p.V617F | 26.4 | rs77375493 |
| PV58 | NA |  |  |  |  |
| PV59 | JAK2exon14 | c.1849G>T | p.V617F | 35.1 | rs77375493 |
| PV60 | JAK2exon14 | c.1849G>T | p.V617F | 33.4 | rs77375493 |
| PV61 | JAK2exon14 | c.1849G>T | p.V617F | 52.9 | rs77375493 |
| PV62 | JAK2exon14 | c.1849G>T | p.V617F | 83 | rs77375493 |
| PV63 | JAK2exon14 TET2exon6 BCORexon4 BCORL1exon7 | c.1849G>T c.3781C>T c.347C>T c.4171G>A | p.V617F p.R1261C p.S116L p.G1391R | 58.5 2.3 51.2 54.9 | rs77375493 — — rs199973665 |
| PV64 | JAK2exon14 | c.1849G>T | p.V617F | 70.4 | rs77375493 |
| PV65 | JAK2exon14 DNMT3Aexon23 | c.1849G>T c.2675C>G | p.V617F p.S892\* | 79.1 1.9 | rs77375493 — |
| PV66 | JAK2exon14 TET2exon11 | c.1849G>T c.5095C>T | p.V617F p.Q1699\* | 39.6 2.4 | rs77375493 — |
| PV67 | JAK2exon14 TET2exon3 | c.1849G>T c.1796delA | p.V617F p.M600\* | 80.8 2.4 | rs77375493 — |
| PV68 | JAK2exon14 | c.1849G>T | p.V617F | 63.9 | rs77375493 |
| PV69 | JAK2exon14 | c.1849G>T | p.V617F | 13.1 | rs77375493 |
| PV70 | JAK2exon14 | c.1849G>T | p.V617F | 18 | rs77375493 |
| PV71 | JAK2Exon14 | c.1849G>T | p.V617F | 16.3 | rs77375493 |
| PV72 | JAK2exon14 | c.1849G>T | p.V617F | 69 | rs77375493 |
| PV73 | JAK2Exon14 | c.1849G>T | p.V617F | 20.9 | rs77375493 |
| PV74 | JAK2exon14 | c.1849G>T | p.V617F | 51.1 | rs77375493 |
| PV75 | JAK2exon14 | c.1849G>T | p.V617F | 43.4 | rs77375493 |
| PV76 | JAK2exon14 DNMT3Aexon19 ASXL1exon12 | c.1849G>T c.2185C>G c.2090\_2108del | p.V617F p.R729G p.P698Gfs\*21 | 8.6 3 1.7 | rs77375493 rs200018028 — |
| PV77 | JAK2exon14 | c.1849G>T | p.V617F | 56.4 | rs77375493 |
| PV78 | JAK2Exon14 | c.1849G>T | p.V617F | 22 | rs77375493 |
| PV79 | JAK2Exon14 | c.1849G>T | p.V617F | 29.8 | rs77375493 |
| PV80 | JAK2exon14 | c.1849G>T | p.V617F | 80.7 | rs77375493 |
| PV81 | JAK2exon14 | c.1849G>T | p.V617F | 62.5 | rs77375493 |
| PV82 | JAK2exon14 PRF1exon3 | c.1849G>T c.1283G>A | p.V617F p.W428\* | 76.8 48.8 | rs77375493 — |
| PV83 | JAK2exon12 ASXL1exon13 TET2exon3 SF3B1exon15 TET2exon6 | c.1625\_1630del c.1927dupG c.2706\_2709delinsTT c.2218G>A c.3632G>A | p.E543\_D544del p.G646Wfs\*11 p.R902Sfs\*21 p.G740R p.C1211Y | 19.5 2.7 7.5 1.9 2 | — rs1085307856 — — — |
| PV84 | JAK2exon14 | c.1849G>T | p.V617F | 68.2 | rs77375493 |
| PV85 | JAK2exon14 | c.1849G>T | p.V617F | 42.7 | rs77375493 |
| PV86 | JAK2exon14 BCORL1exon2 | c.1849G>T c.148G>A | p.V617F p.E50K | 18.2 100 | rs77375493 — |
| PV87 | JAK2exon14 DNMT3Aexon18 | c.1849G>T c.2099C>T | p.V617F p.P700L | 45.9 3.5 | rs77375493 — |
| PV88 | JAK2exon14 | c.1849G>T | p.V617F | 25.9 | rs77375493 |
| PV89 | JAK2exon14 | c.1849G>T | p.V617F | 50.82 | rs77375493 |
| PV90 | JAK2exon14 | c.1849G>T | p.V617F | 72 | rs77375493 |
| PV91 | JAK2exon14 | c.1849G>T | p.V617F | 61.2 | rs77375493 |
| PV92 | JAK2exon14 | c.1849G>T | p.V617F | 69.7 | rs77375493 |
| PV93 | NA |  |  |  |  |
| PV94 | JAK2exon14 ASXL1exon12 | c.1849G>T c.1945G>T | p.V617F p.G649X | 36.8 19.2 | rs77375493 — |
| PV95 | JAK2exon14 BCORexon4 | c.1849G>T c.935\_937delAGC | p.V617F p.Q312delQ | 31.9 100 | rs77375493 rs3840969 |
| PV96 | JAK2exon14 TET2exon9 | c.1849G>T c.4075C>T | p.V617F p.R1359C | 37 2.6 | rs77375493 — |
| PV97 | JAK2exon14 | c.1849G>T | p.V617F | 74.2 | rs77375493 |
| PV98 | JAK2exon14 | c.1849G>T | p.V617F | 50.1 | rs77375493 |
| PV99 | JAK2exon14 | c.1849G>T | p.V617F | 74 | rs77375493 |
| PV100 | JAK2exon14 | c.1849G>T | p.V617F | 74.2 | rs77375493 |
| PV101 | NA |  |  |  |  |
| PV102 | JAK2exon14 | c.1849G>T | p.V617F | 42.3 | rs77375493 |
| PV103 | JAK2exon14 DNMT3Aexon23 | c.1849G>T c.2644C>T | p.V617F p.R882C | 35.2 3.4 | rs77375493 rs377577594 |
| PV104 | JAK2exon14 | c.1849G>T | p.V617F | 81.6 | rs77375493 |
| PV105 | JAK2exon14 ASXL1exon12 SF3B1exon15 | c.1849G>T c.1900\_1922del23 c.2098A>G | p.V617F p.E635Rfs\*15 p.K700E | 19.8 30.4 1.8 | rs77375493 rs766433101 rs559063155 |
| PV106 | JAK2exon14 | c.1849G>T | p.V617F | 46.2 | rs77375493 |
| PV107 | JAK2Exon14 | c.1849G>T | p.V617F | 52 | rs77375493 |
| PV108 | JAK2exon14 | c.1849G>T | p.V617F | 92.5 | rs77375493 |
| PV109 | JAK2exon14 TET2exon10 | c.1849G>T c.4302delT | p.V617F p.Q1435Rfs\*13 | 89.6 45.8 | rs77375493 — |
| PV110 | JAK2exon14 ASXL1exon13 TET2exon9 TET2exon11 | c.1849G>T c.1749G>A c.4075C>T c.5148dupT | p.V617F p.W583\* p.R1359C p.H1717Sfs\*12 | 27.6 41.1 41.9 6.2 | rs77375493 — — — |
| PV111 | JAK2exon14 TET2exon6 TET2exon9 | c.1849G>T c.3619G>T c.4133G>A | p.V617F p.E1207\* p.C1378Y | 69 43.4 40.5 | rs77375493 — rs765853421 |
| PV112 | JAK2exon14 | c.1849G>T | p.V617F | 49.4 | rs77375493 |
| PV113 | JAK2exon14 BCORL1exon3 | c.1849G>T c.3332C>T | p.V617F p.T1111M | 8.5 100 | rs77375493 rs144988023 |
| PV114 | JAK2exon14 BCORexon4 | c.1849G>T c.1448C>T | p.V617F p.P483L | 31.3 48.2 | rs77375493 rs587778096 |
| PV115 | JAK2exon14 TET2exon8 ETV6exon4 | c.1849G>T c.3992delC c.403\_404insT | p.V617F p.T1331Ifs\*32 p.H135Lfs\*19 | 48.9 22.1 1.8 | rs77375493 — — |
| PV116 | JAK2exon12 | c.1624\_1629delAATGAA | p.N542\_E543delNE | 10 | — |
| PV117 | NA |  |  |  |  |
| PV118 | JAK2exon14 CUX1exon22 | c.1849G>T c.3535C>T | p.V617F p.R1179X | 85.5 38.2 | rs77375493 — |
| PV119 | JAK2exon14 | c.1849G>T | p.V617F | 47.2 | rs77375493 |
| PV120 | JAK2exon12 DNMT3Aexon21 | c.1624\_1629del c.2477A>G | p.N542\_E543delNE p.K826R | 27.2 28.3 | — — |
| PV121 | JAK2exon14 | c.1849G>T | p.V617F | 42.2 | rs77375493 |
| PV122 | JAK2exon14 | c.1849G>T | p.V617F | 86 | rs77375493 |
| PV123 | JAK2exon14 | c.1849G>T | p.V617F | 75.6 | rs77375493 |
| PV124 | JAK2exon14 | c.1849G>T | p.V617F | 60.3 | rs77375493 |
| PV125 | JAK2exon14 | c.1849G>T | p.V617F | 82.7 | rs77375493 |
| PV126 | JAK2exon14 DNMT3Aexon19 | c.1849G>T c.2230A>T | p.V617F p.K744X | 17.4 3.4 | rs77375493 — |
| PV127 | JAK2exon14 JAK2exon12 | c.1849G>T c.1615\_1616delinsTT | p.V617F p.K539L | 1.1 30.8 | rs77375493 rs121912473 |
| PV128 | JAK2exon14 | c.G1849T | p.V617F | 14.5 | rs77375493 |
| PV129 | JAK2exon14 | c.1849G>T | p.V617F | 53.5 | rs77375493 |
| PV130 | JAK2exon14 | c.1849G>T | p.V617F | 24.8 | rs77375493 |
| PV131 | JAK2exon14 JAK3exon4 TET2exon3 | c.1849G>T c.394C>A c.2601T>G | p.V617F p.P132T p.Y867\* | 23 0.49 3.1 | rs77375493 rs3212723 rs145844118 |
| PV132 | JAK2exon14 | c.1849G>T | p.V617F | 43.3 | rs77375493 |
| PV133 | JAK2exon14 | c.1849G>T | p.V617F | 61.2 | rs77375493 |
| PV134 | JAK2exon14 | c.1849G>T | p.V617F | 91.5 | rs77375493 |
| PV135 | JAK2exon14 CALRexon9 | c.1849G>T c.1191\_1199del9 | p.V617F p.E398\_D400delEED | 76.9 52.5 | rs77375493 — |
| PV136 | JAK2exon14 TET2exon11 | c.1849G>T c.5650A>G | p.V617F p.T1884A | 26.2 24.9 | rs77375493 rs76428136 |
| PV137 | JAK2exon14 | c.1849G>T | p.V617F | 72.4 | rs77375493 |
| PV138 | JAK2exon14 | c.1849G>T | p.V617F | 47.8 | rs77375493 |
| PV139 | JAK2exon14 | c.1849G>T | p.V617F | 19.9 | rs77375493 |
| PV140 | JAK2exon14 | c.1849G>T | p.V617F | 72.1 | rs77375493 |
| PV141 | JAK2exon14 | c.G1849T | p.V617F | 17.41 | rs77375493 |
| PV142 | JAK2exon14 | c.1849G>T | p.V617F | 34.1 | rs77375493 |
| PV143 | JAK2exon14 | c.1849G>T | p.V617F | 29.5 | rs77375493 |
| PV144 | JAK2exon14 GNASexon8 | c.1849G>T c.2531G>A | p.V617F p.R844H | 23.7 2.3 | rs77375493 rs121913495 |
| PV145 | NA |  |  |  |  |
| PV146 | JAK2exon14 | c.1849G>T | p.V617F | 23.6 | rs77375493 |
| PV147 | JAK2exon14 JAK2exon12 BCORL1exon7 SRSF2exon1 | c.1849G>T c.1614\_1616delinsATT c.4171G>A c.284C>T | p.V617F p.H538\_K539delinsQL p.G1391R p.P95L | 2.5 12.2 49.2 7.9 | rs77375493 — rs199973665 — |
| PV148 | JAK2exon14 | c.1849G>T | p.V617F | 29 | rs77375493 |
| PV149 | JAK2exon14 | c.1849G>T | p.V617F | 17.3 | rs77375493 |
| PV150 | JAK2exon14 | c.1849G>T | p.V617F | 23.6 | rs77375493 |
| PV151 | JAK2exon14 | c.1849G>T | p.V617F | 47.2 | rs77375493 |
| PV152 | JAK2exon14 | c.G1849T | p.V617F | 85.12 | rs77375493 |
| PV153 | JAK2exon14 | c.1849G>T | p.V617F | 28.1 | rs77375493 |
| PV154 | JAK2exon14 | c.1849G>T | p.V617F | 51.9 | rs77375493 |
| PV155 | JAK2exon14 TET2exon3 DNMT3Aexon18 CBLexon8 | c.1849G>T c.1381C>T c.2173+1G>A c.1223G>C | p.V617F p.Q461\* — p.W408S | 58.6 6.7 38.1 2.3 | rs77375493 — rs763716866 — |
| PV156 | NA |  |  |  |  |
| PV157 | JAK2exon14 | c.1849G>T | p.V617F | 83 | rs77375493 |
| PV158 | JAK2exon14 | c.1849G>T | p.V617F | 70.5 | rs77375493 |
| PV159 | JAK2exon14 | c.1849G>T | p.V617F | 51.8 | rs77375493 |
| PV160 | JAK2exon14 TET2exon3 | c.1849G>T c.949C>T | p.V617F p.Q317X | 65.7 37.1 | rs77375493 — |
| PV161 | JAK2exon14 | c.G1849T | p.V617F | 57.14 | rs77375493 |
| PV162 | JAK2exon14 | c.1849G>T | p.V617F | 89.5 | rs77375493 |
| PV163 | JAK2exon14 | c.1849G>T | p.V617F | 61.7 | rs77375493 |
| PV164 | NA |  |  |  |  |
| PV165 | JAK2exon14 | c.1849G>T | p.V617F | 52.7 | rs77375493 |
| PV166 | JAK2exon14 | c.1849G>T | p.V617F | 39.5 | rs77375493 |
| PV167 | JAK2exon14 | c.1849G>T | p.V617F | 14.2 | rs77375493 |
| PV168 | NA |  |  |  |  |
| PV169 | JAK2exon14 | c.1849G>T | p.V617F | 77 | rs77375493 |
| PV170 | JAK2exon12 | c.1624\_1629delAATGAA | p.N542\_E543delNE | 11.1 | — |
| PV171 | JAK2exon14 | c.1849G>T | p.V617F | 59.5 | rs77375493 |
| PV172 | JAK2exon14 | c.1849G>T | p.V617F | 63.2 | rs77375493 |
| PV173 | JAK2exon14 | c.1849G>T | p.V617F | 44.9 | rs77375493 |
| PV174 | JAK2exon14 | c.1849G>T | p.V617F | 26.6 | rs77375493 |
| PV175 | JAK2exon14 | c.1849G>T | p.V617F | 46.51 | rs77375493 |
| PV176 | JAK2exon14 | c.1849G>T | p.V617F | 73.5 | rs77375493 |
| PV177 | JAK2exon14 | c.1849G>T | p.V617F | 48.5 | rs77375493 |
| PV178 | JAK2exon14 | c.1849G>T | p.V617F | 32.2 | rs77375493 |
| PV179 | JAK2exon14 BCORexon14 | c.1849G>T c.4815delT | p.V617F p.F1605Lfs\*35 | 60.3 25.8 | rs77375493 — |
| PV180 | JAK2exon14 TET2Intron4 | c.1849G>T c.3501-2A>G | p.V617F — | 70.3 32.3 | rs77375493 — |
| PV181 | JAK2exon14 | c.1849G>T | p.V617F | 66.4 | rs77375493 |
| PV182 | JAK2exon14 | c.1849G>T | p.V617F | 47 | rs77375493 |
| PV183 | JAK2exon14 | c.1849G>T | p.V617F | 74.4 | rs77375493 |
| PV184 | JAK2exon14 | c.1849G>T | p.V617F | 16.03 | rs77375493 |
| PV185 | JAK2exon14 | c.1849G>T | p.V617F | 18.7 | rs77375493 |
| PV186 | JAK2exon14 | c.1849G>T | p.V617F | 69.7 | rs77375493 |
| PV187 | JAK2exon14 ASXL1exon12 | c.1849G>T c.2110G>A | p.V617F p.G704R | 22.9 2 | rs77375493 rs151317625 |
| PV188 | JAK2exon14 | c.1849G>T | p.V617F | 89.1 | rs77375493 |
| PV189 | JAK2exon14 | c.1849G>T | p.V617F | 86.9 | rs77375493 |
| PV190 | JAK2exon14 | c.1849G>T | p.V617F | 44.3 | rs77375493 |
| PV191 | JAK2exon14 | c.1849G>T | p.V617F | 80.1 | rs77375493 |
| PV192 | JAK2exon14 BCORexon4 | c.1849G>T c.215T>C | p.V617F p.M72T | 84.3 51.9 | rs77375493 — |
| PV193 | JAK2exon14 | c.1849G>T | p.V617F | 24 | rs77375493 |
| PV194 | JAK2exon14 TET2exon3 TET2exon7 | c.1849G>T c.3252\_3259del c.3882C>G | p.V617F p.Q1084Hfs\*17 p.Y1294X | 38.3 42.7 3.2 | rs77375493 rs755531331 — |
| PV195 | JAK2exon14 CDKN2Aexon2 | c.1849G>T c.340C>T | p.V617F p.P114S | 52.3 0.43 | rs77375493 rs104894104 |
| PV196 | JAK2exon14 | c.1849G>T | p.V617F | 15 | rs77375493 |
| PV197 | JAK2exon14 SRP72exon1 | c.1849G>T c.25dupG | p.V617F p.V9Gfs\*9 | 38.4 47.2 | rs77375493 rs763130641 |
| PV198 | JAK2exon14 | c.1849G>T | p.V617F | 66 | rs77375493 |
| PV199 | JAK2exon14 | c.1849G>T | p.V617F | 49.3 | rs77375493 |
| PV200 | JAK2exon14 BCORexon4 | c.1849G>T c.2633T>C | p.V617F p.V878A | 40.7 53.1 | rs77375493 — |
| PV201 | NA |  |  |  |  |
| PV202 | JAK2exon14 | c.1849G>T | p.V617F | 49.3 | rs77375493 |
| PV203 | JAK2exon14 TET2exon10 | c.1849G>T c.4393C>T | p.V617F p.R1465X | 25.4 1.7 | rs77375493 — |
| PV204 | JAK2exon14 | c.1849G>T | p.V617F | 73.8 | rs77375493 |
| PV205 | JAK2exon14 | c.1849G>T | p.V617F | 57.8 | rs77375493 |
| PV206 | JAK2exon14 | c.1849G>T | p.V617F | 31.36 | rs77375493 |
| PV207 | JAK2exon14 | c.1849G>T | p.V617F | 14.9 | rs77375493 |
| PV208 | JAK2exon14 SF3B1exon15 | c.1849G>T c.2098A>G | p.V617F p.K700E | 72 4.4 | rs77375493 rs559063155 |
| PV209 | NA |  |  |  |  |
| PV210 | JAK2exon14 | c.1849G>T | p.V617F | 67.5 | rs77375493 |
| PV211 | JAK2exon14 | c.1849G>T | p.V617F | 38.3 | rs77375493 |
| PV212 | JAK2exon12 | c.1615\_1616delinsTT | p.K539L | 29.5 | rs121912473 |
| PV213 | JAK2exon14 ASXL1exon11 | c.1849G>T c.1210C>T | p.V617F p.R404X | 26.6 27.5 | rs77375493 rs373145711 |
| PV214 | JAK2exon14 | c.1849G>T | p.V617F | 77.5 | rs77375493 |
| PV215 | JAK2exon14 DNMT3AIntron8 | c.1849G>T c.1015-2A>G | p.V617F — | 75.5 42.3 | rs77375493 — |
| PV216 | JAK2exon14 | c.1849G>T | p.V617F | 25.6 | rs77375493 |
| PV217 | JAK2exon14 | c.1849G>T | p.V617F | 91.8 | rs77375493 |
| PV218 | JAK2exon14 | c.1849G>T | p.V617F | 33.4 | rs77375493 |
| PV219 | JAK2exon14 | c.1849G>T | p.V617F | 28 | rs77375493 |
| PV220 | JAK2exon14 | c.1849G>T | p.V617F | 32.3 | rs77375493 |
| PV221 | JAK2exon14 | c.1849G>T | p.V617F | 54.6 | rs77375493 |
| PV222 | JAK2exon14 | c.1849G>T | p.V617F | 54.8 | rs77375493 |
| PV223 | JAK2exon14 | c.1849G>T | p.V617F | 63.3 | rs77375493 |
| PV224 | JAK2exon14 | c.1849G>T | p.V617F | 40.7 | rs77375493 |
| PV225 | JAK2exon14 BCORL1exon3 CBLexon8 | c.1849G>T c.233G>T c.1142G>A | p.V617F p.G78V p.C381Y | 37.6 48 2.5 | rs77375493 — rs768898787 |
| PV226 | JAK2exon14 BCORexon15 IDH1exon4 | c.1849G>T c.5138G>C c.395G>A | p.V617F p.S1713T p.R132H | 2.4 100 1.21 | rs77375493 rs201002764 rs121913500 |
| PV227 | JAK2exon14 ASXL1exon1 | c.1849G>T c.56T>G | p.V617F p.L19R | 54.5 1.8 | rs77375493 — |
| PV228 | JAK2exon14 | c.1849G>T | p.V617F | 65.4 | rs77375493 |
| PV229 | JAK2exon14 | c.1849G>T | p.V617F | 52.8 | rs77375493 |
| PV230 | JAK2exon14 | c.1849G>T | p.V617F | 48.5 | rs77375493 |
| PV231 | JAK2exon14 CHD8exon14 | c.1849G>T c.2904\_2905insAC | p.V617F p.L969Tfs\*17 | 74.9 9.4 | rs77375493 — |
| PV232 | JAK2exon12 | c.1613\_1616delinsT | p.H538\_K539delinsL | 20 | rs77375493 |
| PV233 | JAK2exon14 | c.1849G>T | p.V617F | 14.7 | rs77375493 |
| PV234 | JAK2Exon14 | c.1849G>T | p.V617F | 57.5 | rs77375493 |
| PV235 | JAK2exon14 | c.1849G>T | p.V617F | 94.4 | rs77375493 |
| PV236 | NA |  |  |  |  |
| PV237 | JAK2Exon14 DNMT3AExon18 | c.1849G>T c.2146G>A | p.V617F p.V716I | — | rs77375493 — |
| PV238 | JAK2exon14 | c.G1849T | p.V617F | 28.2 | rs77375493 |
| PV239 | JAK2Exon14 BCORExon4 TP53Exon8 | c.1849G>T c.2930dupT c.868C>T | p.V617F p.K978Qfs\*40 p.R290C | 22.3 31.8 50 | rs77375493 — — |
| PV240 | JAK2Exon14 ASXL1Exon12 TET2Exon3 | c.1849G>T c.1900\_1922del23 c.2422delG | p.V617F p.E635Rfs\*15 p.E808Kfs\*5 | 30.3 8.7 25.2 | rs77375493 rs766433101 — |
| PV241 | JAK2exon14 | c.1849G>T | p.V617F | 12.2 | rs77375493 |
| PV242 | JAK2Exon14 | c.1849G>T | p.V617F | 14.5 | rs77375493 |
| PV243 | JAK2exon14 | c.1849G>T | p.V617F | 49.8 | rs77375493 |
| PV244 | JAK2exon14 TET2exon3 | c.1849G>T c.1245delT | p.V617F p.P416Lfs\*11 | 87.4 18.4 | rs77375493 — |
| PV245 | JAK2exon14 ASXL1exon12 TP53exon7 EZH2exon16 | c.1849G>T c.2791G>T c.733G>A c.1890delG | p.V617F p.E931\* p.G245S p.I631Ffs\*44 | 40.5 18.3 0.67 14.2 | rs77375493 — rs28934575 — |
| PV246 | JAK2exon14 TET2exon3 | c.1849G>T c.1493delT | p.V617F p.P499Hfs\*34 | 94.7 48.3 | rs77375493 — |
| PV247 | JAK2exon14 | c.1849G>T | p.V617F | 86.3 | rs77375493 |
| PV248 | JAK2exon14 | c.1849G>T | p.V617F | 72.2 | rs77375493 |
| PV249 | JAK2exon14 | c.1849G>T | p.V617F | 35.2 | rs77375493 |
| PV250 | JAK2exon12 TET2exon3 | c.1622\_1627del c.2158C>T | p.R541\_E543delinsK p.Q720\* | 20.5 24.5 | — — |
| PV251 | JAK2exon14 | c.1849G>T | p.V617F | 56.4 | rs77375493 |
| PV252 | JAK2exon14 ASXL1exon12 DNMT3Aexon19 KMT2Cexon18 | c.1849G>T c.1927dupG c.2245C>T c.2880T>A | p.V617F p.G646Wfs\*11 p.R749C p.C960\* | 61.9 8.2 3.6 6.4 | rs77375493 rs756958159 rs754613602 rs754055527 |
| PV253 | JAK2exon14 | c.1849G>T | p.V617F | 51.4 | rs77375493 |
| PV254 | JAK2exon14 | c.1849G>T | p.V617F | 16 | rs77375493 |
| PV255 | NA |  |  |  |  |
| PV256 | JAK2exon14 | c.1849G>T | p.V617F | 16.7 | rs77375493 |
| PV257 | JAK2exon14 TET2exon3 | c.1849G>T c.2495dupT | p.V617F p.S833Ffs\*13 | 68.5 20.2 | rs77375493 — |
| PV258 | JAK2exon14 | c.1849G>T | p.V617F | 7.4 | rs77375493 |
| PV259 | JAK2exon14 | c.1849G>T | p.V617F | 79.4 | rs77375493 |
| PV260 | JAK2exon14 | c.1849G>T | p.V617F | 29.9 | rs77375493 |
| PV261 | JAK2exon14 | c.1849G>T | p.V617F | 31.5 | rs77375493 |
| PV262 | JAK2exon14 ASXL1exon12 | c.1849G>T c.1754dupT | p.V617F p.K586fs\*1 | 88.7 43 | rs77375493 — |
| PV263 | JAK2exon14 TP53exon5 | c.1849G>T c.537T>G | p.V617F p.H179Q | 41.7 71.4 | rs77375493 — |
| PV264 | JAK2exon14 | c.1849G>T | p.V617F | 92.5 | rs77375493 |
| PV265 | JAK2exon14 | c.1849G>T | p.V617F | 95.9 | rs77375493 |
| PV266 | JAK2exon14 SF3B1exon15 TP53exon5 | c.1849G>T c.2098A>G c.524G>A | p.V617F p.K700E p.R175H | 78.8 43.4 6.9 | rs77375493 rs559063155 rs28934578 |
| PV267 | JAK2exon14 | c.1849G>T | p.V617F | 84.8 | rs77375493 |
| PV268 | JAK2exon14 TET2exon4 | c.1849G>T c.3412C>T | p.V617F p.Q1138X | 86.7 45.7 | rs77375493 — |
| PV269 | JAK2exon14 | c.1849G>T | p.V617F | 63 | rs77375493 |
| PV270 | JAK2exon14 | c.1849G>T | p.V617F | 22.9 | rs77375493 |
| PV271 | JAK2exon14 | c.1849G>T | p.V617F | 7.3 | rs77375493 |
| PV272 | JAK2exon14 | c.1849G>T | p.V617F | 33.1 | rs77375493 |
| PV273 | JAK2exon14 | c.1849G>T | p.V617F | 93.3 | rs77375493 |
| PV274 | JAK2exon14 ASXL1exon12 ASXL1exon12 | c.1849G>T c.2077C>T c.2332C>T | p.V617F p.R693X p.Q778X | 84.5 3.7 2.3 | rs77375493 rs373221034 — |
| PV275 | JAK2exon14 TP53exon6 DNMT3Aexon23 | c.1849G>T c.659A>G c.2645G>A | p.V617F p.Y220C p.R882H | 58.3 38.7 41.3 | rs77375493 rs121912666 rs147001633 |
| PV276 | JAK2exon14 BCORexon4 | c.1849G>T c.1448C>T | p.V617F p.P483L | 40.2 100 | rs77375493 rs587778096 |
| PV277 | JAK2exon14 | c.1849G>T | p.V617F | 54.4 | rs77375493 |
| PV278 | JAK2exon14 | c.1849G>T | p.V617F | 89.4 | rs77375493 |
| PV279 | JAK2exon14 | c.1849G>T | p.V617F | 94.5 | rs77375493 |
| PV280 | JAK2exon14 | c.1849G>T | p.V617F | 36.5 | rs77375493 |
| PV281 | JAK2exon14 TET2exon3 | c.1849G>T c.679G>T | p.V617F p.E227X | 17.9 8.1 | rs77375493 — |
| PV282 | JAK2exon12 | c.1611\_1616delinsCAT | p.H538\_K539delinsI | 82.2 | — |
| PV283 | JAK2exon14 | c.1849G>T | p.V617F | 53.6 | rs77375493 |
| PV284 | JAK2exon14 ASXL1exon12 ASXL1exon12 SETBP1exon4 ATMexon41 | c.1849G>T c.1904\_1905delAG c.3612C>G c.2602G>A c.6067G>A | p.V617F p.E635Gfs\*22 p.C1204W p.D868N p.G2023R | 96.8 64.7 18.4 48.4 47.2 | rs77375493 — rs201397030 rs267607042 rs11212587 |
| PV285 | JAK2exon14 | c.1849G>T | p.V617F | 68.1 | rs77375493 |
| PV286 | JAK2exon14 RUNX1exon4 | c.1849G>T c.253C>A | p.V617F p.H85N | 66.5 53.4 | rs77375493 rs121912500 |
| PV287 | JAK2exon14 | c.1849G>T | p.V617F | 88.5 | rs77375493 |
| PV288 | JAK2exon14 | c.1849G>T | p.V617F | 93.6 | rs77375493 |
| PV289 | JAK2exon14 TET2exon3 TET2exon3 | c.1849G>T c.727C>T c.2839C>T | p.V617F p.Q243X p.Q947X | 52.6 2.3 3.4 | rs77375493 — — |
| PV290 | JAK2exon14 SETBP1exon4 | c.1849G>T c.2608G>A | p.V617F p.G870S | 83.9 37.5 | rs77375493 rs267607040 |
| PV291 | JAK2exon14 | c.G1849T | p.V617F | 94.08 | rs77375493 |
| PV292 | JAK2exon14 DNMT3Aexon23 | c.1849G>T c.2644C>T | p.V617F p.R882C | 7.4 0.68 | rs77375493 rs377577594 |
| PV293 | JAK2exon14 | c.1849G>T | p.V617F | 58.5 | rs77375493 |
| PV294 | JAK2exon14 TET2exon3 | c.1849G>T c.3195delG | p.V617F p.T1066Lfs\*16 | 89.9 44.3 | rs77375493 — |
| PV295 | NA |  |  |  |  |
| PV296 | JAK2exon14 | c.1849G>T | p.V617F | 53.4 | rs77375493 |
| PV297 | JAK2exon14 | c.1849G>T | p.V617F | 94.8 | rs77375493 |
| PV298 | JAK2exon14 | c.1849G>T | p.V617F | 89 | rs77375493 |
| PV299 | JAK2exon14 TET2exon3 | c.1849G>T c.3343delC | p.V617F p.P1115Lfs\*2 | 87.8 40.5 | rs77375493 rs867474555 |
| PV300 | JAK2exon14 | c.1849G>T | p.V617F | 83 | rs77375493 |
| PV301 | JAK2exon14 | c.1849G>T | p.V617F | 59.4 | rs77375493 |

NA represents patients without relevant mutations. VAF, variant allele frequency.