

## Focused Myeloproliferative Neoplasm Targeted Variants List

**Current Version: v1.0**

**Latest Update:**

- **v1.0:** New document 2/1/2026

**Scope:** This document contains a list of the pathogenic variants targeted by the WML Focused Myeloproliferative Mutation Panel FMPN. Variants not on this list are not reported in this test. This list will be periodically updated if/when additional variants are identified as pathogenic or potentially pathogenic.

**Methodology:** The FMPN test uses targeted multiplexed PCR and Next Generation DNA Sequencing (NGS) to identify the presence/absence of 151 pathogenic mutations in the JAK2 (exons 12 and 14), MPL, CALR, and CSF3R genes.

The panel is designed for, and results presented relative to, hg19 (GRCh37).

### Variant List:

| Variant (CDS)        | Protein Change      | Gene (Transcript)     | Exon | Associated Phenotype           |
|----------------------|---------------------|-----------------------|------|--------------------------------|
| c.1849G>A            | p.V617I             | JAK2<br>(NM_004972.4) | 14   | Myeloproliferative<br>Neoplasm |
| c.1849G>T            | p.V617F             |                       |      |                                |
| c.1609_1616delinsAG  | p.F537_K539delinsR  | JAK2<br>(NM_004972.4) | 12   | Myeloproliferative<br>Neoplasm |
| c.1612_1614del       | p.H538del           |                       |      |                                |
| c.1612C>T            | p.H538Y             |                       |      |                                |
| c.1614_1616del       | p.H538_K539delinsQ  |                       |      |                                |
| c.1612_1617delinsTTT | p.H538_K539delinsF  |                       |      |                                |
| c.1612_1616delinsTT  | p.H538_K539delinsL  |                       |      |                                |
| c.1612_1616delinsAT  | p.H538_K539delinsI  |                       |      |                                |
| c.1613_1616delinsT   | p.H538_K539delinsL  |                       |      |                                |
| c.1613_1617delinsTG  | p.H538_K539delinsL  |                       |      |                                |
| c.1614C>A            | p.H538Q             |                       |      |                                |
| c.1614_1616delinsATT | p.H538_K539delinsQL |                       |      |                                |
| c.1614_1616delinsGTT | p.H538_K539delinsQL |                       |      |                                |
| c.1615_1616delinsTT  | p.K539L             |                       |      |                                |
| c.1615_1617delinsTTG | p.K539L             |                       |      |                                |
| c.1615_1617delinsCTT | p.K539L             |                       |      |                                |
| c.1615_1617delinsCTC | p.K539L             |                       |      |                                |
| c.1615_1616delinsCT  | p.K539L             |                       |      |                                |
| c.1615_1617delinsCTG | p.K539L             |                       |      |                                |

|                                   |                     |                                |    |                                |
|-----------------------------------|---------------------|--------------------------------|----|--------------------------------|
| c.1615_1617delinsTTG              | p.K539L             | JAK2<br>(NM_004972.4)<br>Cont. | 12 | Myeloproliferative<br>Neoplasm |
| c.1620_1621del                    | p.I540MfsTer3       |                                |    |                                |
| c.1619_1627delinsAAA              | p.I540_E543delinsKK |                                |    |                                |
| c.1620_1627delinsGA               | p.I540_E543delinsMK |                                |    |                                |
| c.1620_1632delinsGAAA             | p.I540_D544delinsMK |                                |    |                                |
| c.1622_1627del                    | p.R541_E543delinsK  |                                |    |                                |
| c.1624_1629del                    | p.N542_E543del      |                                |    |                                |
| c.1626_1629del                    | p.N542KfsTer3       |                                |    |                                |
| c.1627_1632del                    | p.E543_D544del      |                                |    |                                |
| c.1626_1632delinsC                | p.E543_D544del      |                                |    |                                |
| c.1628A>G                         | p.E543G             |                                |    |                                |
| c.1080_1143delins<br>GGAAGAAGACAA | p.Q361EfsTer52      | CALR<br>(NM_004343.4)          | 9  | Myeloproliferative<br>Neoplasm |
| c.1088delinsTTTGTC                | p.E363VfsTer69      |                                |    |                                |
| c.1088_1089insTTGTC               | p.E363DfsTer69      |                                |    |                                |
| c.1089_1141del                    | p.E364GfsTer7       |                                |    |                                |
| c.1090_1123del                    | p.E364NfsTer55      |                                |    |                                |
| c.1090_1141del                    | p.E364RfsTer49      |                                |    |                                |
| c.1091_1142del                    | p.E364GfsTer49      |                                |    |                                |
| c.1092_1125del                    | p.E364DfsTer55      |                                |    |                                |
| c.1092_1140delinsCCA              | p.E364DfsTer51      |                                |    |                                |
| c.1093_1138del                    | p.Q365RfsTer50      |                                |    |                                |
| c.1094A>T                         | p.Q365L             |                                |    |                                |
| c.1094_1139del                    | p.Q365RfsTer50      |                                |    |                                |
| c.1097_1130del                    | p.R366Kfs*Ter53     |                                |    |                                |
| c.1099C>A                         | p.L367I             |                                |    |                                |
| c.1099_1141del                    | p.L367RfsTer49      |                                |    |                                |
| c.1099_1150del                    | p.L367TfsTer46      |                                |    |                                |
| c.1099_1153delinsAGG              | p.L367RfsTer46      |                                |    |                                |
| c.1099_1156del                    | p.L367RfsTer44      |                                |    |                                |
| c.1100_1133del                    | p.L367RfsTer52      |                                |    |                                |
| c.1100_1134delinsA                | p.L367QfsTer52      |                                |    |                                |
| c.1100_1136delins GGAGGT          | p.L367RfsTer53      |                                |    |                                |
| c.1100_1145del                    | p.L367QfsTer48      |                                |    |                                |
| c.1100_1152del                    | p.L367QfsTer4       |                                |    |                                |
| c.1101_1146del                    | p.K368RfsTer47      |                                |    |                                |
| c.1101_1152del                    | p.K368RfsTer45      |                                |    |                                |
| c.1101_1161del                    | p.K368MfsTer42      |                                |    |                                |
| c.1102_1137delinsCA               | p.K368QfsTer51      |                                |    |                                |
| c.1103A>G                         | p.K368R             |                                |    |                                |
| c.1103A>T                         | p.K368MfsTer42      |                                |    |                                |
| c.1103_1136del                    | p.K368RfsTer51      |                                |    |                                |
| c.1103_1139del                    | p.K368RfsTer50      |                                |    |                                |
| c.1103_1148del                    | p.K368RfsTer47      |                                |    |                                |

|                               |                |                                |   |                                |
|-------------------------------|----------------|--------------------------------|---|--------------------------------|
| c.1100_1152del                | p.L367QfsTer4  | CALR<br>(NM_004343.4)<br>Cont. | 9 | Myeloproliferative<br>Neoplasm |
| c.1103_1154del                | p.K368RfsTer45 |                                |   |                                |
| c.1105_1138del                | p.E369Rfs*50   |                                |   |                                |
| c.1105_1147delins AGGAGGCAA   | p.E369Rfs*50   |                                |   |                                |
| c.1105_1150del                | p.E369TfsTer46 |                                |   |                                |
| c.1105_1156del                | p.E369Rfs*44   |                                |   |                                |
| c.1108G>T                     | p.E370Ter      |                                |   |                                |
| c.1108_1138del                | p.E370Rfs*50   |                                |   |                                |
| c.1108_1141del                | p.E370Rfs*49   |                                |   |                                |
| c.1108_1177del                | p.E370Rfs*37   |                                |   |                                |
| c.1109_1160del                | p.E370Vfs*43   |                                |   |                                |
| c.1112A>G                     | p.E371G        |                                |   |                                |
| c.1113A>C                     | p.E371D        |                                |   |                                |
| c.1113_1160del                | p.E371_E386del |                                |   |                                |
| c.1116del                     | p.D373TfsTer57 |                                |   |                                |
| c.1120A>C                     | p.K374Q        |                                |   |                                |
| c.1121_1139del                | p.K374RfsTer50 |                                |   |                                |
| c.1121A>G                     | p.K374R        |                                |   |                                |
| c.1122_1125del                | p.K374NfsTer55 |                                |   |                                |
| c.1123_1125delinsTGTTT        | p.K375CfsTer56 |                                |   |                                |
| c.1124A>G                     | p.K375R        |                                |   |                                |
| c.1137G>C                     | p.E379D        |                                |   |                                |
| c.1137_1154delins CCATCCTTGTC | p.E379DfsTer49 |                                |   |                                |
| c.1141del                     | p.E381Rfs*49   |                                |   |                                |
| c.1142A>C                     | p.E381A        |                                |   |                                |
| c.1142A>G                     | p.E381G        |                                |   |                                |
| c.1142_1151delins CGGCATGTC   | p.E381Afs*49   |                                |   |                                |
| c.1143_1154delins TCCTTGTC    | p.E381DfsTer48 |                                |   |                                |
| c.1144_1220delins69           | p.A382QfsTer57 |                                |   |                                |
| c.1145_1146insGACGC           | p.E383Tfs*49   |                                |   |                                |
| c.1147_1151del                | p.E383Qfs*4    |                                |   |                                |
| c.1147_1154delinsTGTC         | p.E383CfsTer46 |                                |   |                                |
| c.1148_1149ins TCCTTGTC       | p.E383Dfs*50   |                                |   |                                |
| c.1148_1154delinsGAC          | p.E383GfsTer46 |                                |   |                                |
| c.1150_1154delinsTGTC         | p.D384Cfs*46   |                                |   |                                |
| c.1151_1154delinsTTTGTC       | p.D384VfsTer47 |                                |   |                                |
| c.1151_1154delins TATGTC      | p.D384VfsTer47 |                                |   |                                |
| c.1151_1154delins GCAATTGTC   | p.D384GfsTer48 |                                |   |                                |
| c.1152_1157del                | p.D384_K385del |                                |   |                                |
| c.1153_1154delinsTGTC         | p.K385CfsTer46 |                                |   |                                |
| c.1153_1154delins TCTTGTC     | p.K385SfsTer47 |                                |   |                                |
| c.1153_1154insTCTGT           | p.K385IfsTer47 |                                |   |                                |
| c.1154del                     | p.K385Rfs*45   |                                |   |                                |
| c.1154_1155insTTGTC           | p.K385NfsTer47 |                                |   |                                |

|                       |                     |                                |    |                                |
|-----------------------|---------------------|--------------------------------|----|--------------------------------|
| c.1154_1155insATGTC   | p.E386CfsTer46      | CALR<br>(NM_004343.4)<br>Cont. | 9  | Myeloproliferative<br>Neoplasm |
| c.1155_1156insTGTCG   | p.E386VfsTer46      |                                |    |                                |
| c.1155_1156insTGTCG   | p.E386VfsTer46      |                                |    |                                |
| c.1154delinsTGTGTC    | p.K385MfsTer47      |                                |    |                                |
| c.1154delinsTTTGTC    | p.K385IfsTer47      |                                |    |                                |
| c.1154delinsCTTGTC    | p.K385TfsTer47      |                                |    |                                |
| c.1154delinsTCTGTC    | p.K385IfsTer47      |                                |    |                                |
| c.1154delinsTATGTC    | p.K385Ifs*47        |                                |    |                                |
| c.1156G>T             | p.E386*             |                                |    |                                |
| c.1162del             | p.D388Mfs*42        |                                |    |                                |
| c.1165G>T             | p.E389*             |                                |    |                                |
| c.1207G>T             | p.E403Ter           |                                |    |                                |
| c.1211_1217del        | p.D404Gfs*24        |                                |    |                                |
| c.1212T>A             | p.D404E             |                                |    |                                |
| c.1213G>C             | p.E405Q             |                                |    |                                |
| c.1214del             | p.E405Gfs*25        |                                |    |                                |
| c.1214_1225del        | p.E405_D408del      |                                |    |                                |
| c.1215G>T             | p.E405D             |                                |    |                                |
| c.1221del             | p.D408Mfs*22        |                                |    |                                |
| c.1502T>C             | p.V501A             | MPL (NM_005373.3)              | 10 | Myeloproliferative<br>Neoplasm |
| c.1513A>T             | p.S505C             |                                |    |                                |
| c.1514G>A             | p.S505N             |                                |    |                                |
| c.1542_1544delinsAGC  | p.W515A             |                                |    |                                |
| c.1543T>C             | p.W515R             |                                |    |                                |
| c.1543T>A             | p.W515R             |                                |    |                                |
| c.1543_1544delinsAA   | p.W515K             |                                |    |                                |
| c.1543_1545delinsAAA  | p.W515K             |                                |    |                                |
| c.1543_1552delinsAAAA | p.W515_P518delinsKT |                                |    |                                |
| c.1543_1544delinsGC   | p.W515A             |                                |    |                                |
| c.1544G>T             | p.W515L             |                                |    |                                |
| c.1544G>C             | p.W515S             |                                |    |                                |
| c.1853C>T             | p.T618I             | CSF3R<br>(NM_000760.4)         | 14 | Myeloproliferative<br>Neoplasm |
| c.1843A>G             | p.T615A             |                                |    |                                |
| c.1844C>A             | p.T615N             |                                |    |                                |
| c.1919C>A             | p.T640N             |                                |    |                                |
| c.1919C>T             | p.T640I             |                                |    |                                |
| c.2215C>T             | p.Q739Ter           |                                |    |                                |
| c.2221C>T             | p.Q741Ter           |                                |    |                                |
| c.2256T>A             | p.Y752Ter           |                                |    |                                |
| c.2256T>G             | p.Y752Ter           |                                |    |                                |
| c.2346insC            | p.S783fs            |                                |    |                                |
| c.2372G>A             | p.W791Ter           |                                |    |                                |
| c.2373G>A             | p.W791Ter           |                                |    |                                |