

Expanded Variant Panel

| Variant cDNA name (ordered 5' to 3') | Variant Protein Name | Variant Legacy Name |
|--------------------------------------|----------------------|---------------------|
| c.-9_14del | No protein name | 124del23bp |
| c.1A>C | p.Met1Leu | M1L |
| c.2T>A | p.Met1Lys | M1K |
| c.4C>T | p.Gln2* | Q2X |
| c.11C>A | p.Ser4* | S4X |
| c.25dup | p.Ala9Glyfs**36 | 156insG |
| c.38C>T | p.Ser13Phe | S13F |
| c.44T>C | p.Leu15Pro | L15P |
| c.49_50del | p.Phe17Glnfs*27 | c.49_50delTT |
| c.49_50dup | p.Trp19Alafs*7 | 181_182dup |
| c.50del | p.Phe17Serfs*8 | 182delT |
| c.50dup | p.Ser18Glnfs*27 | 175insT |
| c.53+1G>T | No protein name | 185+1G->T |
| c.53+2T>C | No protein name | 185+2T->C |
| c.53+2T>G | No protein name | 185+2T->G |
| c.54-2A>G | No protein name | 186-2A->G |
| c.54-1G>A | No protein name | 186-1G->A |
| c.57G>A | p.Trp19* | W19X |
| c.79G>T | p.Gly27* | G27X |
| c.79G>A | p.Gly27Arg | G27R |
| c.87dup | p.Gln30Thrfs*15 | 218insA |
| c.88C>T | p.Gln30* | Q30X |
| c.112_113del | p.Tyr38Profs*6 | 241delAT |
| c.137C>A | p.Ala46Asp | A46D |
| c.142_145del | p.Asn48Tyrfs*42 | c.142_145delAATC |
| c.154A>T | p.Lys52* | K52X |
| c.156_163dup | p.Arg55Asnfs*39 | 295ins8 |
| c.156del | p.Lys52Asnfs*39 | 284delA |
| c.164+1G>A | No protein name | 296+1G->A |
| c.164+1G>C | No protein name | 296+1G->C |
| c.164+1G>T | No protein name | 296+1G->T |
| c.164+2T>C | No protein name | 296+2T->C |
| c.164+2T>G | No protein name | 296+2T->G |
| c.164+4dup | No protein name | 296+3insT |

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| c.165-2A>G | No protein name | 297-2A->G |
| c.165-1G>A | No protein name | 297-1G->A |
| c.166G>A | p.Glu56Lys | E56K |
| c.168del | p.Glu56Aspfs*35 | 300delA |
| c.169T>G | p.Trp57Gly | W57G |
| c.170G>A or c.171G>A | p.Trp57* | W57X |
| c.174_177del | p.Asp58Gluufs*32 | 306delTAGA |
| c.175dup | p.Arg59Lysfs*10 | 306insA |
| c.177dup | p.Glu60Argfs*9 | 308insA |
| c.178G>A | p.Glu60Lys | E60K |
| c.185dup | p.Ser63Phefs*6 | 317insC |
| c.202A>T | p.Lys68* | K68X |
| c.233dup | p.Trp79Leufs*32 | 365-366insT |
| c.234del | p.Trp79Glyfs*12 | c.234delC |
| c.236G>A | p.Trp79* | W79X |
| c.263T>A or c.263T>G | p.Leu88* | L88X |
| c.271G>A | p.Gly91Arg | G91R |
| c.273+2T>G | No protein name | 405+2T->G |
| c.273+3A>C | No protein name | 405+3A->C |
| c.274-2A>C | No protein name | 406-2A->C |
| c.274-2A>G | No protein name | 406-2A->G |
| c.274-1G>C | No protein name | 406-2G->C |
| c.285dup | p.Ala96Serfs*15 | 415insA |
| c.293A>G | p.Gln98Arg | Q98R |
| c.296C>T | p.Pro99Leu | P99L |
| c.303dup | p.Leu102Thrfs*9 | 435insA |
| c.305T>G | p.Leu102Arg | L102R |
| c.307G>T | p.Gly103* | G103X |
| c.310del | p.Arg104Gluufs*3 | 442delA |
| c.313del | p.Ile105Serfs*2 | 444delA |
| c.327T>A | p.Tyr109* | Y109X |
| c.332del | p.Pro111Argfs*13 | 464delC |
| c.340A>T | p.Lys114* | K114X |
| c.346G>A | p.Glu116Lys | E116K |
| c.350G>C | p.Arg117Pro | R117P |
| c.357del | p.Ile119Metfs*5 | 489delC |
| c.367dup | p.Leu123Profs*36 | 499dupC |
| c.377G>A | p.Gly126Asp | G126D |

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| c.387del | p.Leu130Serfs*4 | 519delT |
| c.393del | p.Phe131Leufs*3 | 525delT |
| c.409del | p.Leu137Serfs*16 | 541delC |
| c.409_412del | p.Leu137Tyrfs*15 | 541del4 |
| c.410_416del | p.Leu137Profs*14 | 542del7 |
| c.413_415dup | p.Leu138dup | L138ins |
| c.416A>G | p.His139Arg | H139R |
| c.424del | p.Ile142Phefs*11 | 556delA |
| c.429del | p.Phe143Leufs*10 | 557delT |
| c.445G>A | p.Gly149Arg | G149R |
| c.450dup | p.Gln151Alafs*8 | 582insG |
| c.461dup | p.Ala155Serfs*4 | 593insT |
| c.470_483del | p.Phe157* | 602del14 |
| c.476dup | p.Leu159Phefs*4 | 605insT |
| c.476T>A | p.Leu159* | L159X |
| c.481T>G | p.Tyr161Asp | Y161D |
| c.489+1G>A | No protein name | 621+1G->A |
| c.489+2T>C | No protein name | 621+2T->C |
| c.489+2T>G | No protein name | 621+2T->G |
| c.490-2A>C | No protein name | 622-2A->C |
| c.494T>C | p.Leu165Ser | L165S |
| c.498del | p.Lys166Asnfs*7 | 630delG |
| c.522_526del | p.Ile175Tyrfs*6 | 654del5 |
| c.531dup | p.Gly178Trpfs*5 | 663insT |
| c.543_546del | p.Leu183Phefs*5 | 675del4 |
| c.543_556del | p.Ser182Glnfs*6 | 675del14 |
| c.548del | p.Leu183Profs*6 | 680delT |
| c.550del | p.Leu184Phefs*5 | 681delC |
| c.551_555del | p.Leu184Glnfs*7 | c.551_555delTTTCC |
| c.571T>G | p.Phe191Val | F191V |
| c.575A>G | p.Asp192Gly | D192G |
| c.577G>A | p.Glu193Lys | E193K |
| c.577G>T | p.Glu193* | E193X |
| c.578_579+5del | No protein name | 710_711+5del7 |
| c.579_579+3del | No protein name | 710del4 |
| c.580-2A>G | No protein name | 712-2A->G |
| c.580G>A | p.Gly194Arg | G194R |
| c.580G>T | p.Gly194* | G194X |

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| c.581_582del | p.Gly194Alafs*63 | 713delGA |
| c.601del | p.Val201Cysfs*14 | 733delG |
| c.604_605del | p.Trp202Aspfs*55 | c.604_605delTG |
| c.606G>A | p.Trp202* | W202X |
| c.614dup | p.Leu206Phefs*52 | 746insC |
| c.617del | p.Leu206Cysfs*9 | 749delT |
| c.619C>T | p.Gln207* | Q207X |
| c.647G>A | p.Trp216* | W216X |
| c.653T>A | p.Leu218* | L218X |
| c.675T>A | p.Cys225* | C225X |
| c.680T>G | p.Leu227Arg | L227R |
| c.695T>A | p.Val232Asp | V232D |
| c.708dup | p.Gln237Serfs*21 | 840insT |
| c.717del | p.Leu240* | 849delG |
| c.743+1G>A | No protein name | 875+1G->A |
| c.743+1G>C | No protein name | 875+1G->C |
| c.743+2T>C | No protein name | 875+2T->C |
| c.744-2A>G | No protein name | 876-2A->G |
| c.748C>T | p.Gln250* | Q250X |
| c.761del | p.Lys254Argfs*7 | 892delA |
| c.764del | p.Ile255Thrfs*6 | 896delT |
| c.773del | p.Arg258Asnfs*3 | 905delG |
| c.777del | p.Val260* | 909delT |
| c.803del | p.Asn268Ilefs*17 | 935delA |
| c.805_806del | p.Ile269Profs*4 | 936delTA |
| c.825C>G | p.Tyr275* | Y275X |
| c.828C>A | p.Cys276* | C276X |
| c.830G>A | p.Trp277* | W277X |
| c.850del | p.Met284* | 982delA |
| c.850dup | p.Met284Asnfs*3 | 977insA |
| c.861_865del | p.Asn287Lysfs*19 | 991del5 |
| c.865A>T | p.Arg289* | R289X |
| c.868C>T | p.Gln290* | Q290X |
| c.869+1G>C | No protein name | 1001+1G->C |
| c.874_875del | p.Glu292Thrfs*15 | 1006_1007delGA |
| c.905_906insA | p.Arg303Glufs*5 | 1037insA |
| c.908_911del | p.Arg303Thrfs*24 | 1040del4 |
| c.927del | p.Phe310Serfs*18 | 1058delC |

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| c.933C>A or c.933C>G | p.Phe311Leu | F311L |
| c.959T>A | p.Leu320* | L320X |
| c.980del | p.Leu327Glnfs*42 | 1112delT |
| c.982del | p.Ile328Serfs*41 | c.982delA |
| c.987del | p.Gly330Glufs*39 | 1119delA |
| c.1001G>T | p.Arg334Leu | R334L |
| c.1006_1007insG | p.Ile336Serfs*28 | 1138insG |
| c.1025_1026insTA | p.Cys343Thrfs*27 | 1157insTA |
| c.1029del | p.Cys343* | 1161delC |
| c.1037T>C | p.Leu346Pro | L346P |
| c.1053_1054del | p.Arg352Alafs*11 | 1185delTC |
| c.1057C>T | p.Gln353* | Q353X |
| c.1068G>A | p.Trp356* | W356X |
| c.1069del | p.Ala357Leufs*12 | 1199delG |
| c.1075_1079delinsAAAAA | p.[Gln359Lys;Thr360Lys] | Q359K/T360K |
| c.1082G>A or c.1083G>A | p.Trp361* | W361X |
| c.1083del | p.Trp361Cysfs*8 | 1215delG |
| c.1086T>A or c.1086T>G | p.Tyr362* | Y362X |
| c.1093_1094del | p.Leu365Trpfs*16 | 1221delCT |
| c.1116_1116+1insATCAA | No protein name | 1248insATCAA |
| c.1117-1G>A | No protein name | 1249-1G->A |
| c.1118dup | p.Asp373Glufs*9 | 1249insA |
| c.1130del | p.Lys377Serfs*11 | 1262delA |
| c.1132C>T | p.Gln378* | Q378X |
| c.1135G>T | p.Glu379* | E379X |
| c.1155_1156dup | p.Asn386Ilefs*3 | 1288insTA |
| c.1157dup | p.Asn386Lysfs*25 | 1288insA |
| c.1159_1160del | p.Leu387Asnfs*23 | 1291delTT |
| c.1162_1163delinsTA | p.Thr388* | T388X |
| c.1162_1168del | p.Thr388Glnfs*3 | 1294del7 |
| c.1209+1G>C | No protein name | 1341+1G->C |
| c.1209+1G>T | No protein name | 1341+1G->T |
| c.1209+2T>G | No protein name | 1341+2T->G |
| c.1210-2A>C | | 1342-2A->C |
| c.1210-1G>C | No protein name | 1342-1G->C |
| c.1210-1G>T | No protein name | 1342-1G->T |
| c.1211del | p.Gly404Aspfs*38 | 1343delG |
| c.1221del | p.Glu407Aspfs*35 | 1353delA |

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| c.1234del | p.Ala412Glnfs*30 | 1366delG |
| c.1240C>T | p.Gln414* | Q414X |
| c.1243_1247del | p.Asn415* | 1367del5 |
| c.1248dup | p.Asn417* | 1380insT |
| c.1287del | p.Phe430Serfs*12 | 1419delC |
| c.1301C>A or c.1301C>G | p.Ser434* | S434X |
| c.1301_1307del | p.Ser434Leufs*6 | 1429del7 |
| c.1302del | p.Leu435Phefs*7 | 1434delA |
| c.1324A>T | p.Lys442* | K442X |
| c.1330_1331del | p.Ile444* | 1460delAT |
| c.1340del | p.Lys447Argfs*2 | 1471delA |
| c.1358T>C | p.Leu453Ser | L453S |
| c.1360_1387del | p.Leu454Alafs*6 | 1491-1500del |
| c.1365_1366del | p.Val456Cysfs*25 | 1497delGG |
| c.1366del | p.Val456Leufs*13 | 1498delG |
| c.1366G>T | p.Val456Phe | V456F |
| c.1367T>C | p.Val456Ala | V456A |
| c.1373del | p.Gly458Aspfs*11 | 1504delG |
| c.1375_1385del | p.Ser459Argfs*19 | c.1375_1385del |
| c.1392+1del | No protein name | 1524+1delG |
| c.1392+1G>A | No protein name | 1524+1G->A |
| c.1393-2A>G | No protein name | 1525-2A->G |
| c.1409_1418del | p.Val470Glufs*54 | 1540del10 |
| c.1420G>A | p.Glu474Lys | E474K |
| c.1424del | p.Leu475Argfs*52 | 1556delT |
| c.1439del | p.Gly480Valfs*47 | 1571delG |
| c.1446dup | p.Lys483* | 1576insT |
| c.1470_1471del | p.Phe490Leufs*13 | 1601delITC |
| c.1477_1478del | p.Gln493Valfs*10 | 1609delCA |
| c.1487G>A | p.Trp496* | W496X |
| c.1505T>C | p.Ile502Thr | I502T |
| c.1510G>T | p.Glu504* | E504X |
| c.1538A>G | p.Asp513Gly | D513G |
| c.1540G>T | p.Glu514* | E514X |
| c.1546A>G | p.Arg516Gly | R516G |
| c.1551C>A or c.1551C>G | p.Tyr517* | Y517X |
| c.1572C>A | p.Cys524* | C524X |
| c.1582G>T | p.Glu528* | E528X |

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| c.1584+1G>A | No protein name | 1716+1G->A |
| c.1584+2T>C | No protein name | 1716+2T->C |
| c.1585-1G>T | No protein name | 1717-1G->T |
| c.1585-2A>G | No protein name | 1717-2A->G |
| c.1616_1617dup | p.Val540* | 1749insTA |
| c.1642_1643del | p.Leu548Glufs*19 | 1774delCT |
| c.1647T>A | p.Ser549Arg | S549R |
| c.1648G>T | p.Gly550* | G550X |
| c.1650del | p.Gly551Valfs*8 | 1782delA |
| c.1651G>A | p.Gly551Ser | G551S |
| c.1670del | p.Ser557Phefs*2 | 1802delC |
| c.1673T>C | p.Leu558Ser | L558S |
| c.1674del | p.Ala559Glnfs*13 | 1806delA |
| c.1679+1G>A | No protein name | 1811+1G->A |
| c.1679+1G>C | No protein name | 1811+1G->C |
| c.1679+2T>C | No protein name | 1811+2T->C |
| c.1680-877G>T | No protein name | 1811+1643G->T |
| c.1680-2A>C | No protein name | 1812-2A->C |
| c.1680-1G>C | No protein name | IVS11-1G->C |
| c.1680A>C | p.Arg560Ser | R560S |
| c.1682C>A | p.Ala561Glu | A561E |
| c.1682dup | p.Val562Serfs*6 | 1813insC |
| c.1687T>A | p.Tyr563Asn | Y563N |
| c.1687T>G | p.Tyr563Asp | Y563D |
| c.1689C>A | p.Tyr563* | Y563X |
| c.1692del | p.Asp565Metfs*7 | 1824delA |
| c.1703del | p.Leu568Cysfs*4 | 1833delT |
| c.1703T>A | p.Leu568* | L568X |
| c.1705T>G | p.Tyr569Asp | Y569D |
| c.1721C>A | p.Pro574His | P574H |
| c.1731C>A | p.Tyr577* | Y577X |
| c.1741_1742dup | p.Leu581Phefs*2 | 1874_1875insTT |
| c.1742dup | p.Leu581Phefs*8 | 1874insT |
| c.1766+1G>C | No protein name | 1898+1G->C |
| c.1766+1G>T | No protein name | 1898+1G->T |
| c.1766+2del | No protein name | 1898+2delT |
| c.1766+2T>A | No protein name | 1898+2T->A |
| c.1766+2T>C | No protein name | 1898+2T->C |

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| c.1766+5G>T | No protein name | 1898+5G->T |
| c.1786_1787del | p.Ala596* | 1918delGC |
| c.1792_1798del | p.Lys598Glyfs*11 | 1924del7 |
| c.1792A>T | p.Lys598* | K598X |
| c.1800del | p.Ile601Phefs*10 | 1932delG |
| c.1801A>T | p.Ile601Phe | I601F |
| c.1807del | p.Val603Serfs*8 | c.1807delG |
| c.1810_1826del | p.Thr604Phefs*5 | 1942del17 |
| c.1820_1903del | p.Met607_Gln634del | 1949del84 |
| c.1826A>G | p.His609Arg | H609R |
| c.1837G>A | p.Ala613Thr | A613T |
| c.1846del | p.Ile616Tyrfs*2 | 1978delA |
| c.1846dup | p.Ile616Asnfs*6 | 1978dupA |
| c.1874_1875del | p.Tyr625Phefs*16 | 2005delTA |
| c.1882G>C or c.1882G>A | p.Gly628Arg | G628R |
| c.1900C>T | p.Gln634* | Q634X |
| c.1911del | p.Gln637Hisf**26 | 2043delG |
| c.1920_1921dup | p.Ser641Ilefs*23 | 2053insTA |
| c.1923_1931delinsA | p.Ser641Argfs*5 | 2055del9->A |
| c.1936G>T | p.Gly646* | G646X |
| c.1943del | p.Asp648Valfs*15 | 2075delA |
| c.1954C>T | p.Gln652* | Q652X |
| c.1966G>T | p.Glu66* | E66X |
| c.1972dup | p.Arg658Lysfs*7 | 2104insA |
| c.1973_1985delinsAGAAA | p.Arg658Lysfs*4 | 2105- 2117del13insAGAAA |
| c.1976del | p.Asn659Ilefs*4 | 2108delA |
| c.1977_1986del | p.Asn659Lysfs*10 | 2109-2118del10 |
| c.1981del | p.Ile661Serfs*2 | 2113delA |
| c.1986_1989del | p.Thr663Argfs*8 | 2118del4 |
| c.1987_2000del | p.Thr663Profs*21 | 2118del14 |
| c.1990G>T | p.Glu664* | E664X |
| c.2000_2001del | p.His667Profs*21 | 2132delAC |
| c.2017G>T | p.Gly673* | G673X |
| c.2036G>A | p.Trp679* | W679X |
| c.2044dup | p.Thr682Asnfs*7 | 2175insA |
| c.2045dup | p.Gln685Thrfs*4 | 2176insC |
| c.2051_2052del | p.Lys684Thrfs*4 | 2183delAA |

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| c.2053dup | p.Gln685Profs*84 | 2185insC |
| c.2053C>T | p.Gln685* | Q685X |
| c.2057_2058delinsA | p.Ser686Tyrfs*36 | 2189CT->A |
| c.2062A>T | p.Lys688* | K688X |
| c.2074G>T | p.Glu692* | E692X |
| c.2083del | p.Glu695Lysfs*27 | 2215delG |
| c.2089dup | p.Arg697Lysfs*33 | 2221insA |
| c.2091del | p.Lys698Argfs*24 | 2222delG |
| c.2143C>T | p.Gln715* | Q715X |
| c.2145_2146delinsGT | p.Lys716* | K716X |
| c.2148del | p.Thr717Leufs*5 | c.2148delG |
| c.2156T>A | p.Leu719* | L719X |
| c.2158C>T | p.Gln720* | Q720X |
| c.2188G>T | p.Glu730* | E730X |
| c.2203del | p.Arg735Glyfs*4 | 2335delA |
| c.2233G>T | p.Gly745* | G745X |
| c.2241_2248del | p.Ile748Serfs*28 | 2372del8 |
| c.2261del | p.Val754Glyfs*17 | c.2261delT |
| c.2277del | p.Thr760Argfs*11 | 2409delC |
| c.2299C>T | p.Gln767* | Q767X |
| c.2303dup | p.Val769Cysfs*10 | 2435insC |
| c.2324_2325del | p.His775Leufs*3 | 2456delAC |
| c.2335C>T | p.Gln779* | Q779X |
| c.2341C>T | p.Gln781* | Q781X |
| c.2349dup | p.His784Serfs*21 | 2481-2482insT |
| c.2353C>T | p.Arg785* | R785X |
| c.2374C>T | p.Arg792* | R792X |
| c.2380del | p.Val794Cysfs*9 | 2512delG |
| c.2393dup | p.Gln799Serfs*6 | 2522insC |
| c.2423_2424dup | p.Ser809Ilefs*13 | 2556insAT |
| c.2425del | p.Ser809Glnfs*12 | 2557delT |
| c.2440C>T | p.Gln814* | Q814X |
| c.2443G>T | p.Glu815* | E815X |
| c.2454_2455insT | p.Glu819* | 2586-2687insT |
| c.2463_2464del | p.Ser821Argfs*4 | 2594delGT |
| c.2488A>T | p.Lys830* | K830X |
| c.2489dup | p.Glu831Glyfs*5 | c.2489_2490insA |
| c.2491-2A>G | No protein name | 2623-2A->G |

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| c.2502del | p.Phe834Leufs*10 | 2634delT |
| c.2502dup | p.Asp835* | 2634insT |
| c.2508del | p.Asp836Glufs*8 | 2640delT |
| c.2523_2548del | p.Ala842Serfs*45 | 2655del26 |
| c.2547C>A | p.Tyr849* | Y849X |
| c.2562del | p.Val855Serfs*5 | 2694delT |
| c.2576_2588del | p.Leu859* | 2708del13 |
| c.2589_2599del | p.Ile864Serfs*28 | 2721del11 |
| c.2592_2593del | p.Ile864Metfs*31 | 2723delTT |
| c.2601dup | p.Val868Serfs*28 | 2732insA |
| c.2615del | p.Ala872Glufs*34 | 2747delC |
| c.2619+2T>A | No protein name | 2751+2T->A |
| c.2619+2T>C | No protein name | 2751+2T->C |
| c.2620-2A>G | No protein name | 2752-2A->G |
| c.2620-1G>T | No protein name | 2752-1G->T |
| c.2644_2645dup | p.Trp882Cysfs*25 | 2777insTG |
| c.2645G>A | p.Trp882* | W882X |
| c.2655_2670del | p.Asn886Thrfs*15 | 2787del16 |
| c.2658-2A>G | No protein name | 2790-2A->G |
| c.2658-1G>C | No protein name | 2790-1G->C |
| c.2705del | p.Ser902Thrfs*4 | 2837delG |
| c.2732_2733insA | p.Ser911Argfs*64 | c.2732_2733insA |
| c.2733_2734insA | p.Ser912Ilefs*63 | c.2733insA |
| c.2735C>A | p.Ser912* | S912X |
| c.2737_2738insG | p.Tyr913* | 2869insG |
| c.2739T>A | p.Tyr913* | Y913X |
| c.2763_2764dup | p.Val922Glufs*2 | 2896insAG |
| c.2776_2777del | p.Leu926Alafs*48 | 2907delTT |
| c.2777del | p.Leu926Cysfs*16 | 2909delT |
| c.2803_2813del | p.Leu935Alafs*36 | 2935del11 |
| c.2805_2810del | p.Pro936_Leu937del | 2937_2942delinsTC AGA |
| c.2810dup | p.Val938Glyfs*37 | 2942insT |
| c.2816del | p.His939Leufs*3 | 2948delA |
| c.2819_2820insA | p.Leu941Serfs*34 | 2951insA |
| c.2819_2823del | p.Thr940Asnfs*33 | 2949del5 |
| c.2822del | p.Leu941Glnfs*27 | 2954delT |
| c.2825del | p.Ile942Thrfs*26 | 2957delT |

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| c.2836A>T | p.Lys946* | K946X |
| c.2859_2890del | p.Leu953Phefs*11 | 2991del32 |
| c.2879del | p.Pro960Leufs*8 | 3011delC |
| c.2883_2886dup | p.Thr963Valfs*13 | 3015_3018dupGTCA |
| c.2885C>A c.2885C>G | p.Ser962* | S962X |
| c.2896del | p.Thr966Argfs*2 | 3028delA |
| c.2897del | p.Thr966Serfs*2 | 3029delC |
| c.2899_2900delinsA | p.Leu967Argfs*14 | 3031-3032delinsA |
| c.2908+1G>A | No protein name | 3040+1G->A |
| c.2909del | p.Gly970Valfs*11 | 3041delG |
| c.2909G>A | p.Gly970Asp | G970D |
| c.2924_2925del | p.Arg975Ilefs*10 | 3056delGA |
| c.2932A>T | p.Lys978* | K978X |
| c.2936A>T | p.Asp979Val | D979V |
| c.2964dup | p.Leu989Serfs*5 | 3095insT |
| c.2982_2988+2del | No protein name | c.2982_2988+2delC ATCCAGGT |
| c.2986C>T | p.Gln996* | Q996X |
| c.2989-2A>C | No protein name | 3121-2A->C |
| c.2989-2A>G | No protein name | 3121-2A->G |
| c.2989-2A>T | No protein name | 3121-2A->T |
| c.2989-1G>T | No protein name | 3121-1G->T |
| c.2997_3000del | p.Ile1000* | 3129del4 |
| c.2998del | p.Ile1000Leufs*2 | 3130delA |
| c.3002_3003del | p.Val1001Aspfs*45 | 3132delTG |
| c.3007G>T | p.Gly1003* | G1003X |
| c.3008del | p.Gly1003Glufs*3 | 3139delG |
| c.3011_3019del | p.Ala1004_Ala1006del | 3143del9 |
| c.3011del | p.Ala1004Valfs*2 | 3143delC |
| c.3017C>A | p.Ala1006Glu | A1006E |
| c.3022del | p.Val1008Serfs*15 | 3154delG |
| c.3033dup | p.Gln1012Thrfs*35 | c.3033dup |
| c.3039dup | p.Tyr1014Leufs*33 | 3171insC |
| c.3039del | p.Tyr1014Thrfs*9 | 3171delC |
| c.3067_3072del | p.Ile1023_Val1024del | 3199del6 |
| c.3081dup | p.Met1028Tyrfs*19 | 3213-3214insT |
| c.3103C>T | p.Gln1035* | Q1035X |
| c.3106del | p.Thr1036Profs*24 | 3238delA |

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| c.3107C>A | p.Thr1036Asn | T1036N |
| c.3124C>T | p.Gln1042* | Q1042X |
| c.3136G>T | p.Glu1046* | E1046X |
| c.3139_3139+1del | p.Gly1047Glnfs*28 | 3271delGG |
| c.3139+2T>C | No protein name | 3271+2T->C |
| c.3140-1G>A | No protein name | 3272-1G->A |
| c.3160C>G | p.His1054Asp | H1054D |
| c.3161del | p.His1054Leufs*6 | 3293delA |
| c.3176T>G | p.Leu1059* | L1059X |
| c.3181G>C | p.Gly1061Arg | G1061R |
| c.3189G>A | p.Trp1063* | W1063X |
| c.3217dup | p.Tyr1073Leufs*3 | 3349insT |
| c.3227_3231del | p.Thr1076Ilefs*78 | 3359delCTCTG |
| c.3229_3230del | p.Leu1077Valfs*78 | 3359delCT |
| c.3254A>C | p.His1085Pro | H1085P |
| c.3264del | p.Trp1089Glyfs*13 | 3396delC |
| c.3275_3290del | p.Tyr1092Serfs*5 | 3407_3422del16 |
| c.3290_3305del | p.Arg1097Glnfs*2 | 3422del16 |
| c.3292T>C | p.Trp1098Arg | W1098R |
| c.3293G>A or c.3294G>A | p.Trp1098* | W1098X |
| c.3294del | p.Trp1098Cysfs*4 | 3425delG |
| c.3294G>C or c.3294G>T | p.Trp1098Cys | W1098C |
| c.3302T>G | p.Met1101Arg | M1101R |
| c.3304A>T | p.Arg1102* | R1102X |
| c.3315del | p.Met1105Ilefs*16 | 3447delG |
| c.3324del | p.Ile1109Serfs*12 | 3456delC |
| c.3353C>T | p.Ser1118Phe | S1118F |
| c.3365del | p.Thr1122Lysfs*12 | 3497delC |
| c.3367+1G>T | No protein name | 3499+1G->T |
| c.3367+2T>C | No protein name | 3499+2T->C |
| c.3368-2A>G | No protein name | 3500-2A->G |
| c.3368-1G>A | No protein name | 3500-1G->A |
| c.3382A>T | p.Arg1128* | R1128X |
| c.3384_3388del | p.Val1129Tyrfs*25 | 3516del5 |
| c.3389_3402del | p.Gly1130Valfs*21 | 3521del14 |
| c.3397del | p.Leu1133* | 3528delC |
| c.3400_3401delinsGTA | p.Thr1134Valfs*22 | 3532AC->GTA |
| c.3407_3422del | p.Ala1136Valfs*7 | c.3407_3422del16 |

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| c.3421_3424dup | p.Thr1142Lysfs*15 | 3556insAGTA |
| c.3430C>T | p.Gln1144* | Q1144X |
| c.3435G>A | p.Trp1145* | W1145X |
| c.3468G>A | P.Leu1156Leu | 3600G->A |
| c.3468+1G>A | No protein name | 3600+1G->A |
| c.3468+1G>T | No protein name | 3600+1G->T |
| c.3468+2dup | No protein name | 3600+2insT |
| c.3468+5G>A | No protein name | 3600+5G->A |
| c.3469-2A>G | No protein name | 3601-2A->G |
| c.3469-1G>A | No protein name | 3601-1G->A |
| c.3475T>C | p.Ser1159Pro | S1159P |
| c.3476C>T | p.Ser1159Phe | S1159F |
| c.3492dup | p.Lys1165* | 3622insT |
| c.3493A>T | p.Lys1165* | K1165X |
| c.3497del | p.Phe1166Serfs*26 | 3629delT |
| c.3530del | p.Lys1177Serfs*15 | 3662delA |
| c.3532_3535dup | p.Thr1179Ilefs*17 | 3667ins4 |
| c.3533C>A or c.3533C>G | p.Ser1178* | S1178X |
| c.3536_3539del | p.Thr1179Asnfs*12 | 3667del4 |
| c.3539_3554del | p.Lys1180Thrfs*7 | c.3539_3554del |
| c.3546C>G | p.Tyr1182* | Y1182X |
| c.3556C>T | p.Gln1186* | Q1186X |
| c.3592del | p.Val1198* | 3724delG |
| c.3598delinsTCT | p.Lys1200Serfs*12 | 3730A->TCT |
| c.3599_3600delinsG | p.Lys1200Argfs*11 | 3731-3732AA->G |
| c.3600del | p.Asp1201Metfs*10 | 3732delA |
| c.3605del | p.Asp1202Alafs*9 | 3737delA |
| c.3615del | p.Ser1206Glnfs*5 | 3747delC |
| c.3617C>G | p.Ser1206* | S1206X |
| c.3618_3619del | p.Gly1208Profs*56 | 3750delAG |
| c.3623dup | p.Gln1209Profs*56 | 3755dupG |
| c.3658dup | p.Thr1220Asnfs*45 | 3789insA |
| c.3691del | p.Ser1231Profs*4 | 3821delT |
| c.3700A>G | p.Ile1234Val | I1234V |
| c.3708del | p.Gly1237Alafs*22 | 3840delT |
| c.3717G>A | p.Arg1239Arg | 3849G->A |
| c.3717+4A>G | No protein name | 3849+4A->G |
| c.3717+5G>A | No protein name | 3849+5G->A |

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| c.3718-3T>G | No protein name | 3850-3T->G |
| c.3718-1G>A | No protein name | 3850-1G->A |
| c.3719T>G | p.Val1240Gly | V1240G |
| c.3724del | p.Leu1242Serfs*17 | 3856delC |
| c.3743C>A or c.3743C>G | p.Ser1248* | S1248X |
| c.3745G>A | p.Gly1249Arg | G1249R |
| c.3747del | p.Lys1250Argfs*9 | 3878delG |
| c.3754dup | p.Thr1252Asnfs*13 | 3886insA |
| c.3758dup | p.Leu1253Phefs*12 | 3889dupT |
| c.3761T>G | p.Leu1254* | L1254X |
| c.3763T>C | p.Ser1255Pro | S1255P |
| c.3764C>A | p.Ser1255* | S1255X |
| c.3767dup | p.Leu1258Phefs*7 | 3898insC |
| c.3773del | p.Leu1258* | 3905delT |
| c.3796G>T | p.Glu1266* | E1266X |
| c.3806T>A | p.Ile1269Asn | I1269N |
| c.3808del | p.Asp1270Metfs*8 | 3940delG |
| c.3816_3817del | p.Ser1273Leufs*28 | 3944delGT |
| c.3822G>A | p.Trp1274* | W1274X |
| c.3827del | p.Ser1276* | 3959delC |
| c.3829del | p.Ile1277* | 3960-3961delA |
| c.3838C>T | p.Gln1280* | Q1280X |
| c.3841C>T | p.Gln1281* | Q1281X |
| c.3848G>T | p.Arg1283Met | R1283M |
| c.3873+2T>C | No protein name | 4005+2T->C |
| c.3874-1G>A | No protein name | 4006-1G->A |
| c.3876del | p.Val1293Tyrfs*35 | 4006delA |
| c.3883_3886del | p.Ile1295Phefs*32 | 4010del4 |
| c.3883del | p.Ile1295Phefs*33 | 4015delA |
| c.3889dup | p.Ser1297Phefs*5 | 4016dupT |
| c.3891dup | p.Gly1298Trpfs*4 | 4022insT |
| c.3904A>T | p.Lys1302* | K1302X |
| c.3908del | p.Asn1303Thrfs*25 | 4040delA |
| c.3917_3918dup | p.Tyr1307Profs*22 | 4048insCC |
| c.3921T>A | p.Tyr1307* | Y1307X |
| c.3925C>T | p.Gln1309* | Q1309X |
| c.3929G>A | p.Trp1310* | W1310X |

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| c.3932_3933delinsAATATG | p.Ser1311Lysfs*12 | 4064-4065delinsAATATG |
| c.3947G>A | p.Trp1316* | W1316X |
| c.3957del | p.Asp1320Metfs*8 | 4089delA |
| c.3963+1G>A | No protein name | 4095+1G->A |
| c.3963+1G>C | No protein name | 4095+1G->C |
| c.3971T>C | p.Leu1324Pro | L1324P |
| c.3976del | p.Ser1326Leufs*2 | 4108delT |
| c.3988C>T | p.Gln1330* | Q1330X |
| c.4004T>C | p.Leu1335Pro | L1335P |
| c.4028dup | p.Cys1344Leufs*15 | 4160insG |
| c.4033_4034del | p.Val1345Profs*13 | 4165delGT |
| c.4035_4038dup | p.Ser1347Profs*13 | p.S1347PfsX13 |
| c.4036_4042del | p.Leu1346Metfs*6 | 4168delCTAAGCC |
| c.4040_4041del | p.Ser1347Thrfs*11 | 4172delGC |
| c.4046del | p.Gly1349Alafs*5 | 4177delG |
| c.4046G>A | p.Gly1349Asp | G1349D |
| c.4051A>T | p.Lys1351* | K1351X |
| c.4065_4066del | p.Leu1356Glyfs*2 | 4197_4198delCT |
| c.4071_4073delinsAA | p.Arg1358Asnfs*22 | 4203TAG->AA |
| c.4086dup | p.Lys1363* | 4218insT |
| c.4090del | p.Ala1364Argfs*16 | 4222delG |
| c.4097T>A | p.Ile1366Asn | I1366N |
| c.4111G>T | p.Glu1371* | E1371X |
| c.4124A>C | p.His1375Pro | H1375P |
| c.4127_4131del | p.Leu1376Serfs*8 | 4259del5 |
| c.4139del | p.Thr1380Asnfs*4 | 4271delC |
| c.4144C>T | p.Gln1382* | Q1382X |
| c.4147dup | p.Ile1383Asnfs*3 | 4279insA |
| c.4168C>T | p.Gln1390* | Q1390X |
| c.4170del | p.Ala1391Hisfs*7 | 4301delA |
| c.4197_4198del | p.Cys1400* | 4326delTC |
| c.4231C>T | p.Gln1411* | Q1411X |
| c.4234C>T | p.Gln1412* | Q1412X |
| c.4242+1G>A | No protein name | 4374+1G->A |
| c.4242+1G>T | No protein name | 4374+1G->T |
| c.4300_4301dup | p.Ser1435Glyfs*14 | 4428insGA |