

Expanded Variant Panel

Variant cDNA name	Variant protein name	Variant legacy name
c.-9_14del	No protein name	124del23bp
c.1A>C	No protein name	M1L
c.2T>A	No protein name	M1K
c.2T>C	No protein name	M1T
c.2T>G	No protein name	M1R
c.3G>A	No protein name	M1I
c.4C>T	p.Gln2X	Q2X
c.4del	p.Gln2ArgfsX23	c.4delC
c.11C>A	p.Ser4X	S4X
c.25dup	p.Ala9GlyfsX36	156insG
c.38C>T	p.Ser13Phe	S13F
c.40A>T	p.Lys14X	K14X
c.40_44del	p.Lys14PhefsX29	c.40_44delAAACT
c.42del	p.Lys14AsnfsX11	174delA
c.43del	p.Leu15PhefsX10	175delC
c.43dup	p.Leu15ProfsX30	c.43dupC
c.44T>C	p.Leu15Pro	L15P
c.49_50del	p.Phe17GlnfsX27	c.49_50delTT
c.49_50dup	p.Trp19AlafsX7	181_182dup
c.50del	p.Phe17SerfsX8	182delT
c.50dup	p.Ser18GlnfsX27	175insT
c.51del	p.Phe17LeufsX8	183delC
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.56G>A c.57G>A	p.Trp19X	W19X
c.71_72delinsA	p.Leu24X	c.71_72delTGinsA
c.79G>A	p.Gly27Arg	G27R
c.79G>T	p.Gly27X	G27X
c.87dup	p.Gln30ThrfsX15	218insA
c.88C>T	p.Gln30X	Q30X
c.88_89del	p.Gln30AlafsX14	c.88_89delCA
c.97G>T	p.Glu33X	E33X
c.112_113del	p.Tyr38ProfsX6	241delAT
c.114C>G	p.Tyr38X	Y38X
c.137C>A	p.Ala46Asp	A46D
c.142_145del	p.Asn48TyrfsX42	c.142_145delAATC
c.147_150del	p.Ser50LysfsX40	c.147_150delATCT
c.154A>T	p.Lys52X	K52X
c.156del	p.Lys52AsnfsX39	284delA

c.156_163dup	p.Arg55AsnfsX39	295ins8
c.160G>T	p.Glu54X	E54X
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>A	No protein name	296+2T->A
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
c.165-3C>T	No protein name	297-3C->T
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.166G>T	p.Glu56X	E56X
c.169T>G	p.Trp57Gly	W57G
c.170G>A c.171G>A	p.Trp57X	W57X
c.174dup	p.Arg59X	305insT
c.174_177del	p.Asp58GluX32	306delTAGA
c.175dup	p.Arg59LysfsX10	306insA
c.177dup	p.Glu60ArgfsX9	308insA
c.178G>A	p.Glu60Lys	E60K
c.185dup	p.Ser63PhefsX6	317insC
c.202A>T	p.Lys68X	K68X
c.233dup	p.Trp79LeufsX32	365-366insT
c.234del	p.Trp79GlyfsX12	c.234delC
c.234dup	p.Trp79LeufsX32	366insC
c.236G>A	p.Trp79X	W79X
c.243del	p.Phe81LeufsX10	c.243delT
c.248dup	p.Tyr84LeufsX27	379-381insT
c.252T>A	p.Tyr84X	Y84X
c.253G>T	p.Gly85X	G85X
c.263T>A c.263T>G	p.Leu88X	L88X
c.264_268del	p.Leu88PhefsX21	c.264_268delATATT
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	406-1G->C
c.285dup	p.Ala96SerfsX15	415insA
c.293A>G	p.Gln98Arg	Q98R
c.296C>T	p.Pro99Leu	P99L
c.302T>G	p.Leu101X	L101X
c.303dup	p.Leu102ThrfsX9	435insA
c.305T>G	p.Leu102Arg	L102R
c.307G>T	p.Gly103X	G103X
c.310A>T	p.Arg104X	R104X

c.310del	p.Arg104GluX3	442delA
c.313del	p.Ile105SerX2	444delA
c.324del	p.Tyr109MetX15	c.324delC
c.326_327del	p.Tyr109X	458delAT
c.327T>A	p.Tyr109X	Y109X
c.332del	p.Pro111ArgX13	464delC
c.340A>T	p.Lys114X	K114X
c.346G>A	p.Glu116Lys	E116K
c.350G>C	p.Arg117Pro	R117P
c.353del	p.Ser118LeuX6	c.353delC
c.357del	p.Ile119MetX5	489delC
c.367dup	p.Leu123ProX36	499dupC
c.377G>A	p.Gly126Asp	G126D
c.381_382dup	p.Cys128TyrX7	c.381_382dup
c.387del	p.Leu130SerX4	519delT
c.393del	p.Phe131LeuX3	525delT
c.394_398del	p.Ile132GluX25	526_527delAT
c.405_406dup	p.Leu136HisX18	538insAC
c.409del	p.Leu137SerX16	541delC
c.409_412del	p.Leu137TyrX15	541del4
c.410_416del	p.Leu137ProX14	542del7
c.413_415dup	p.Leu138dup	L138ins
c.415_416insTA	p.His139LeuX15	547insTA
c.416A>G	p.His139Arg	H139R
c.424del	p.Ile142PheX11	556delA
c.429del	p.Phe143LeuX10	557delT
c.433del	p.Leu145PheX8	565delC
c.445G>A	p.Gly149Arg	G149R
c.450del	p.Met150IleX3	c.450delG
c.450dup	p.Gln151AlaX8	582insG
c.451C>T	p.Gln151X	Q151X
c.451del	p.Gln151ArgX2	583delC
c.461dup	p.Ala155SerX4	593insT
c.470_483del	p.Phe157X	602del14
c.472dup	p.Ser158LysX5	604insA
c.476dup	p.Leu159PheX4	605insT
c.476T>A	p.Leu159X	L159X
c.481T>G	p.Tyr161Asp	Y161D
c.488del	p.Lys163ArgX3	c.488delA
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.490-1G>A	No protein name	622-1G->A
c.490-1G>C	No protein name	622-1G->C
c.490-1G>T	No protein name	622-1G->T
c.494del	p.Leu165X	624delT

c.494T>A	p.Leu165X	L165X
c.494T>C	p.Leu165Ser	L165S
c.498del	p.Lys166AsnfsX7	630delG
c.531dup	p.Gly178TrpfsX5	663insT
c.535C>T	p.Gln179X	Q179X
c.543_546del	p.Leu183PhefsX5	675del4
c.543_556del	p.Ser182GlnfsX6	675del14
c.548del	p.Leu183ProfsX6	680delT
c.550del	p.Leu184PhefsX5	681delC
c.551_555del	p.Leu184GlnfsX7	c.551_555delTTTCC
c.558del	p.Asn186LysfsX3	690delC
c.560del	p.Asn187ThrfsX2	c.560delA
c.571T>G	p.Phe191Val	F191V
c.575A>G	p.Asp192Gly	D192G
c.576del	p.Asp192GluX23	708delT
c.577G>A	p.Glu193Lys	E193K
c.577G>T	p.Glu193X	E193X
c.578_579+5del	No protein name	710_711+5del7
c.579_579+3del	No protein name	710del4
c.579+2T>C	No protein name	711+2T->C
c.580-2A>C	No protein name	712-2A->C
c.580-2A>G	No protein name	712-2A->G
c.580G>A c.580G>C	p.Gly194Arg	G194R
c.580G>T	p.Gly194X	G194X
c.581_582del	p.Gly194AlafsX63	713delGA
c.583del	p.Ala196HisfsX19	c.583delC
c.601del	p.Val201CysfsX14	733delG
c.604_605del	p.Trp202AspfsX55	c.604_605delTG
c.606G>A	p.Trp202X	W202X
c.614dup	p.Leu206PhefsX52	746insC
c.617del	p.Leu206CysfsX9	749delT
c.619C>T	p.Gln207X	Q207X
c.647G>A c.648G>A	p.Trp216X	W216X
c.650_659del	p.Glu217GlyfsX11	c.650_659del
c.653T>A	p.Leu218X	L218X
c.675T>A	p.Cys225X	C225X
c.680T>G	p.Leu227Arg	L227R
c.695T>A	p.Val232Asp	V232D
c.708del	p.Gln237ArgfsX4	840delT
c.708dup	p.Gln237SerfsX21	840insT
c.709C>T	p.Gln237X	Q237X
c.713_714insA	p.Gly239TrpfsX19	845_846insA
c.714del	p.Leu240X	L240X
c.717del	p.Leu240X	849delG
c.741C>G c.741C>A	p.Tyr247X	Y247X
c.743_743+1insTACA	No protein name	875insTACA
c.743+1G>A	No protein name	875+1G->A

c.743+1G>C	No protein name	875+1G->C
c.743+2T>A	No protein name	875+2T->A
c.743+2T>C	No protein name	875+2T->C
c.744-2A>G	No protein name	876-2A->G
c.748C>T	p.Gln250X	Q250X
c.761del	p.Lys254ArgfsX7	892delA
c.764del	p.Ile255ThrfsX6	896delT
c.773del	p.Arg258AsnfsX3	905delG
c.775delinsGCTGGGAAGAT	p.Leu259AlafsX12	c.775delinsGCTGGGAAGAT
c.777del	p.Val260X	909delT
c.803del	p.Asn268IlefsX17	935delA
c.805_806del	p.Ile269ProfsX4	936delTA
c.808C>T	p.Gln270X	Q270X
c.811del	p.Ser271LeufsX14	c.811delT
c.822del	p.Tyr275ThrfsX10	954delA
c.825C>G	p.Tyr275X	Y275X
c.828C>A	p.Cys276X	C276X
c.830G>A c.831G>A	p.Trp277X	W277X
c.835G>T	p.Glu279X	E279X
c.850del	p.Met284X	982delA
c.850dup	p.Met284AsnfsX3	977insA
c.861_865del	p.Asn287LysfsX19	991del5
c.863_869+2del	No protein name	994del9
c.865_869del	p.Arg289AsnfsX17	c.865_869delAGACA
c.865A>T	p.Arg289X	R289X
c.868C>T	p.Gln290X	Q290X
c.869+1G>C	No protein name	1001+1G->C
c.869+2T>A	No protein name	1001+2T->A
c.869+2T>G	No protein name	1001+2T->G
c.870-2A>C	No protein name	1002-2A->C
c.870-2A>G	No protein name	1002-2A->G
c.874_875del	p.Glu292ThrfsX15	1006_1007delGA
c.874G>T	p.Glu292X	E292X
c.895del	p.Ala299GlnfsX4	1027delG
c.905_906insA	p.Arg303GlufsX5	1037insA
c.908_911del	p.Arg303ThrfsX24	1040del4
c.912C>G	p.Tyr304X	Y304X
c.912delinsGG	p.Tyr304X	c.912delCinsGG
c.927del	p.Phe310SerfsX18	1058delC
c.933C>A c.933C>G	p.Phe311Leu	F311L
c.937_938del	p.Ser313ArgfsX50	c.937_938delTC
c.942del	p.Phe315SerfsX13	c.942delG
c.959T>A	p.Leu320X	L320X
c.972del	p.Tyr325MetfsX3	c.972delC
c.980del	p.Leu327GlnfsX42	1112delT
c.982del	p.Ile328SerfsX41	c.982delA
c.985A>T	p.Lys329X	K329X

c.987del	p.Gly330GlufsX39	1119delA
c.991_995del	p.Ile331ProfsX31	c.991_995del
c.1001G>T	p.Arg334Leu	R334L
c.1006_1007insG	p.Ile336SerfsX28	1138insG
c.1025_1026insTA	p.Cys343ThrfsX27	1157insTA
c.1029del	p.Cys343X	1161delC
c.1037T>C	p.Leu346Pro	L346P
c.1053_1054del	p.Arg352AlafsX11	1185delTC
c.1057C>T	p.Gln353X	Q353X
c.1068_1087del	p.Trp356X	1200_1219del20
c.1068G>A	p.Trp356X	W356X
c.1069del	p.Ala357LeufsX12	1199delG
c.1072_1073del		1204_1205delGT
c.[1075C>A;1079C>A]	p.Gln359_Thr360delinsLysLys	Q359K/T360K
c.1076A>G	p.Gln359Arg	Q359R
c.1080del	p.Trp361GlyfsX8	1212delA
c.1082G>A c.1083G>A	p.Trp361X	W361X
c.1083del	p.Trp361CysfsX8	1215delG
c.1084_1088dup	p.Ser364MetfsX7	c.1084_1088dup
c.1086T>A c.1086T>G	p.Tyr362X	Y362X
c.1093_1094del	p.Leu365TrpfsX16	1221delCT
c.1114C>T	p.Gln372X	Q372X
c.1115del	p.Gln372ArgfsX16	c.1115delA
c.1116_1116+1insATCAA	No protein name	1248insATCAA
c.1116+1G>C	No protein name	1248+1G->C
c.1116+1G>T	No protein name	1248+1G->T
c.1116+2T>A	No protein name	1248+2T->A
c.1116+2T>C	No protein name	1248+2T->C
c.1117-1G>A	No protein name	1249-1G->A
c.1118dup	p.Asp373GlufsX9	1249insA
c.1126C>T	p.Gln376X	Q376X
c.1130del	p.Lys377SerfsX11	1262delA
c.1132C>T	p.Gln378X	Q378X
c.1135G>T	p.Glu379X	E379X
c.1141A>T	p.Lys381X	K381X
c.1155_1156dup	p.Asn386IlefsX3	1288insTA
c.1157_1158insTA	p.Leu387ThrfsX2	1289insTA
c.1157dup	p.Asn386LysfsX25	1288insA
c.1159_1160del	p.Leu387AsnfsX23	1291delTT
c.1162_1163delinsTA	p.Thr388X	T388X
c.1162_1168del	p.Thr388GlnfsX3	1294del7
c.1177del	p.Val393X	1309delG
c.1190dup	p.Thr398AsnfsX13	c.1190dupT
c.1204G>T	p.Glu402X	E402X
c.1208del	p.Glu403GlyfsX39	1340delA
c.1209+1G>C	No protein name	1341+1G->C
c.1209+1G>T	No protein name	1341+1G->T

c.1209+2T>A	No protein name	1341+2T->A
c.1209+2T>G	No protein name	1341+2T->G
c.1210-1del	No protein name	1342-1delG
c.1210-1G>A	No protein name	1342-1G->A
c.1210-1G>C	No protein name	1342-1G->C
c.1210-1G>T	No protein name	1342-1G->T
c.1210-2_1210-1del	No protein name	1342-2delAG
c.1210-2A>C	No protein name	1342-2A->C
c.1211del	p.Gly404AspfsX38	1343delG
c.1219del	p.Glu407AsnfsX35	c.1219delG
c.1219G>T	p.Glu407X	E407X
c.1221del	p.Glu407AspfsX35	1353delA
c.1234del	p.Ala412GlnfsX30	1366delG
c.1240C>T	p.Gln414X	Q414X
c.1243_1247del	p.Asn415X	1367del5
c.1244del	p.Asn415ThrfsX27	c.1244delA
c.1248dup	p.Asn417X	1380insT
c.1262del	p.Thr421IlefsX21	c.1262delC
c.1287del	p.Phe430SerfsX12	1419delC
c.1301_1307del	p.Ser434LeufsX6	1429del7
c.1301C>A c.1301C>G	p.Ser434X	S434X
c.1302del	p.Leu435PhefsX7	1434delA
c.1312dup	p.Thr438AsnfsX8	c.1312dupA
c.1324A>T	p.Lys442X	K442X
c.1329_1350del	p.Asp443GlufsX19	D443fs
c.1330_1331del	p.Ile444X	1460delAT
c.1333_1334insTAAT	p.Asn445IlefsX38	1465_1466insTAAT
c.1340del	p.Lys447ArgfsX2	1471delA
c.1342del	p.Ile448X	1474delA
c.1354C>T	p.Gln452X	Q452X
c.1358T>C	p.Leu453Ser	L453S
c.1360_1387del	p.Leu454AlafsX6	1491-1500del
c.1365_1366del	p.Val456CysfsX25	1497delGG
c.1366del	p.Val456LeufsX13	1498delG
c.1366G>T	p.Val456Phe	V456F
c.1367T>C	p.Val456Ala	V456A
c.1373del	p.Gly458AspfsX11	1504delG
c.1375_1385del	p.Ser459ArgfsX19	c.1375_1385del
c.1392+1del	No protein name	1524+1delG
c.1392+1G>A	No protein name	1524+1G->A
c.1393-1G>C	No protein name	1525-1G->C
c.1393-1G>T	No protein name	1525-1G->T
c.1393-2A>G	No protein name	1525-2A->G
c.1409_1418del	p.Val470GlufsX54	1540del10
c.1420G>A	p.Glu474Lys	E474K
c.1424del	p.Leu475ArgfsX52	1556delT
c.1433_1434del	p.Ser478X	1565delCA

c.1435G>T	p.Glu479X	E479X
c.1439del	p.Gly480ValfsX47	1571delG
c.1446dup	p.Lys483X	1576insT
c.1469del	p.Phe490SerfsX37	1601delT
c.1470_1471del	p.Phe490LeufsX13	1601delTC
c.1477_1478del	p.Gln493ValfsX10	1609delCA
c.1486del	p.Trp496GlyfsX31	c.1486delT
c.1487G>A	p.Trp496X	W496X
c.1505T>C	p.Ile502Thr	I502T
c.1510G>T	p.Glu504X	E504X
c.1528del	p.Val510PhefsX17	1660delG
c.1530_1531del	p.Ser511LeufsX2	c.1530_1531delTT
c.1538A>G	p.Asp513Gly	D513G
c.1540G>T	p.Glu514X	E514X
c.1543_1555del	p.Tyr515AlafsX8	1675_1687del13
c.1546A>G	p.Arg516Gly	R516G
c.1547_1548del	p.Arg516IlefsX51	c.1547_1548delGA
c.1551C>A c.1551C>G	p.Tyr517X	Y517X
c.1572C>A	p.Cys524X	C524X
c.1573del	p.Gln525AsnfsX2	c.1573delC
c.1579_1584+11del	No protein name	1710del17bp
c.1579G>T	p.Glu527X	E527X
c.1581dup	p.Glu528ArgfsX40	c.1581dupA
c.1582G>T	p.Glu528X	E528X
c.1584+1G>A	No protein name	1716+1G->A
c.1584+1G>T	No protein name	1716+1G->T
c.1584+2T>C	No protein name	1716+2T->C
c.1584+2del	No protein name	1716+2delT
c.1585-2A>G	No protein name	1717-2A->G
c.1585-1G>T	No protein name	1717-1G->T
c.1592_1593delinsG	p.Ser531X	S531X
c.1606A>T	p.Lys536X	K536X
c.1608del	p.Asp537ThrfsX3	c.1608delA
c.1616_1617dup	p.Val540X	1749insTA
c.1627del	p.Glu543LysfsX6	1759delG
c.1642_1643del	p.Leu548GlufsX19	1774delCT
c.1648G>T	p.Gly550X	G550X
c.1650del	p.Gly551ValfsX8	1782delA
c.1651G>A	p.Gly551Ser	G551S
c.1652del	p.Gly551ValfsX8	1784delG
c.1652_1653insATCAT	p.Gln552SerfsX9	1784insATCAT
c.1656del	p.Gln552HisfsX7	1787delA
c.1663A>T	p.Arg555X	R555X
c.1670del	p.Ser557PhefsX2	1802delC
c.1673T>C	p.Leu558Ser	L558S
c.1674del	p.Ala559GlnfsX13	1806delA
c.1675del	p.Ala559GlnfsX13	1807delG

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.1680-1G>C	No protein name	1812-1G->C
c.1680-2A>C	No protein name	1812-2A->C
c.1680A>C c.1680A>T	p.Arg560Ser	R560S
c.1682C>A	p.Ala561Glu	A561E
c.1682dup	p.Val562SerfsX6	1813insC
c.1687T>A	p.Tyr563Asn	Y563N
c.1687T>G	p.Tyr563Asp	Y563D
c.1689C>A	p.Tyr563X	Y563X
c.1692del	p.Asp565MetfsX7	1824delA
c.1703del	p.Leu568CysfsX4	1833delT
c.1703T>A	p.Leu568X	L568X
c.1705T>G	p.Tyr569Asp	Y569D
c.1707T>A	p.Tyr569X	Y569X
c.1708_1712del	p.Leu570ArgfsX17	c.1708_1712delTTATT
c.1709T>A	p.Leu570X	L570X
c.1714_1715del	p.Asp572LeufsX16	1845delAG/1846delGA
c.1721C>A	p.Pro574His	P574H
c.1725_1727delinsAT	p.Phe575LeufsX4	c.1725_1727delinsAT
c.1725del	p.Phe575LeufsX4	1857delT
c.1731C>A	p.Tyr577X	Y577X
c.1738del	p.Val580PhefsX2	1870delG
c.1741_1742dup	p.Leu581PhefsX2	1874_1875insTT
c.1742dup	p.Leu581PhefsX8	1874insT
c.1742T>A	p.Leu581X	L581X
c.1753del	p.Glu585LysfsX10	c.1753delG
c.1762G>T	p.Glu588X	E588X
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
c.1766+5G>T	No protein name	1898+5G->T
c.1767-2A>G	No protein name	1898-2A->G
c.1767-1G>A	No protein name	1899-1G->A
c.1786_1787del	p.Ala596X	1918delGC
c.1792_1798del	p.Lys598GlyfsX11	1924del7
c.1792A>T	p.Lys598X	K598X
c.1800del	p.Ile601PhefsX10	1932delG
c.1801A>T	p.Ile601Phe	I601F
c.1807del	p.Val603SerfsX8	c.1807delG
c.1810_1826del	p.Thr604PhefsX5	1942del17
c.1820_1903del	p.Met607_Gln634del	1949del84
c.1826A>G	p.His609Arg	H609R
c.1837G>A	p.Ala613Thr	A613T

c.1846del	p.Ile616TyrfsX2	1978delA
c.1846dup	p.Ile616AsnfsX6	1978dupA
c.1853_1863del	p.Ile618ArgfsX2	c.1853_1863del
c.1853T>C	p.Ile618Thr	I618T
c.1871_1878del	p.Ser624IlefsX15	2003del8
c.1874_1875del	p.Tyr625PhefsX16	2005delTA
c.1882G>A c.1882G>C	p.Gly628Arg	G628R
c.1892C>G	p.Ser631X	S631X
c.1893dup	p.Glu632ArgfsX10	2025dupA
c.1894_1895del	p.Glu632ThrfsX9	c.1894_1895del
c.1900C>T	p.Gln634X	Q634X
c.1904del	p.Asn635IlefsX28	2036delA
c.1911del	p.Gln637HisfsX26	2043delG
c.1920_1921dup	p.Ser641IlefsX23	2053insTA
c.1923_1931delinsA	p.Ser641ArgfsX5	2055del9->A
c.1936G>T	p.Gly646X	G646X
c.1943del	p.Asp648ValfsX15	2075delA
c.1954C>T	p.Gln652X	Q652X
c.1966G>T	p.Glu656X	E656X
c.1969A>T	p.Arg657X	R657X
c.1970del	p.Arg657LysfsX6	c.1970delG
c.1972dup	p.Arg658LysfsX7	2104insA
c.1973_1985delinsAGAAA	p.Arg658LysfsX4	2105-2117del13insAGAAA
c.1976del	p.Asn659IlefsX4	2108delA
c.1977_1986del	p.Asn659LysfsX10	2109-2118del10
c.1979C>A c.1979C>G	p.Ser660X	S660X
c.1981del	p.Ile661SerfsX2	2113delA
c.1982del	p.Ile661ThrfsX2	2114delT
c.1986_1989del	p.Thr663ArgfsX8	2118del4
c.1987_2000del	p.Thr663ProfsX21	2118del14
c.1990G>T	p.Glu664X	E664X
c.2000_2001del	p.His667ProfsX21	2132delAC
c.2017G>T	p.Gly673X	G673X
c.2032_2033del	p.Ser678LeufsX10	c.2032_2033delTC
c.2036G>A c.2037G>A	p.Trp679X	W679X
c.2044dup	p.Thr682AsnfsX7	2175insA
c.2045del	p.Thr682LysfsX40	c.2045delC
c.2045dup	p.Gln685ThrfsX4	2176insC
c.2051_2052del	p.Lys684ThrfsX4	2183delAA
c.2052_2055del	p.Lys684AsnfsX37	2184del4
c.2053_2054insAA	p.Ser686AsnfsX37	c.2053_2054insAA
c.2053C>T	p.Gln685X	Q685X
c.2053del	p.Gln685AsnfsX37	c.2053delC
c.2053dup	p.Gln685ProfsX4	2185insC
c.2057_2058delinsA	p.Ser686TyrfsX36	2189CT->A
c.2058_2061dup	p.Lys688PhefsX2	2193ins4
c.2062A>T	p.Lys688X	K688X

c.2074G>T	p.Glu692X	E692X
c.2083del	p.Glu695LysfsX27	2215delG
c.2089del	p.Arg697GlyfsX25	c.2089delA
c.2089dup	p.Arg697LysfsX33	2221insA
c.2091del	p.Lys698ArgfsX24	2222delG
c.2143C>T	p.Gln715X	Q715X
c.2145_2146delinsGT c.2146A>T	p.Lys716X	K716X
c.2148del	p.Thr717LeufsX5	c.2148delG
c.2156T>A	p.Leu719X	L719X
c.2158_2173del	p.Gln720LysfsX8	2290del16
c.2158C>T	p.Gln720X	Q720X
c.2188G>T	p.Glu730X	E730X
c.2203del	p.Arg735GlyfsX4	2335delA
c.2233G>T	p.Gly745X	G745X
c.2250del	p.Arg751AlafsX4	c.2250delT
c.2261del	p.Val754GlyfsX17	c.2261delT
c.2274_2275delinsT	p.Thr760ArgfsX11	2406_2407delinsT
c.2277del	p.Thr760ArgfsX11	2409delC
c.2298del	p.Arg766SerfsX5	2429delG
c.2299C>T	p.Gln767X	Q767X
c.2303dup	p.Val769CysfsX10	2435insC
c.2324_2325del	p.His775LeufsX3	2456delAC
c.2335C>T	p.Gln779X	Q779X
c.2335del	p.Gln779LysfsX24	2466delC
c.2337del	p.Gly780ValfsX23	c.2337delA
c.2340_2346del	p.Gln781PhefsX20	2472_2478del7
c.2341C>T	p.Gln781X	Q781X
c.2341del	p.Gln781ArgfsX22	c.2341delC
c.2349dup	p.His784SerfsX21	2481-2482insT
c.2353C>T	p.Arg785X	R785X
c.2357dup	p.Thr787AspfsX18	c.2357dupA
c.2374C>T	p.Arg792X	R792X
c.2378_2379del	p.Lys793SerfsX11	2510delAA
c.2380del	p.Val794CysfsX9	2512delG
c.2393dup	p.Gln799SerfsX6	2522insC
c.2395C>T	p.Gln799X	Q799X
c.2423_2424dup	p.Ser809IlefsX13	2556insAT
c.2425del	p.Ser809GlnfsX12	2557delT
c.2428A>T	p.Arg810X	R810X
c.2433_2437delinsATA	p.Leu812TyrfsX10	c.2433_2437delinsATA
c.2435T>A	p.Leu812X	L812X
c.2440C>T	p.Gln814X	Q814X
c.2443G>T	p.Glu815X	E815X
c.2454_2455insT	p.Glu819X	2586-2687insT
c.2463_2464del	p.Ser821ArgfsX4	2594delGT
c.2467_2470del	p.Glu823LeufsX6	c.2467_2470delGAAA
c.2472del	p.Asn825ThrfsX5	2603delT

c.2476G>T	p.Glu826X	E826X
c.2488A>T	p.Lys830X	K830X
c.2489dup	p.Glu831GlyfsX5	c.2489_2490insA
c.2491-2A>G	No protein name	2623-2A->G
c.2491-1G>C	No protein name	2623-1G->C
c.2493del	p.Glu831AspfsX13	c.2493delG
c.2502del	p.Phe834LeufsX10	2634delT
c.2502dup	p.Asp835X	2634insT
c.2508del	p.Asp836GlufsX8	2640delT
c.2523_2548del	p.Ala842SerfsX45	2655del26
c.2538del	p.Trp846X	c.2538delG
c.2540del	p.Asn847ThrfsX13	2672delA
c.2547C>A	p.Tyr849X	Y849X
c.2554dup	p.Tyr852LeufsX44	2686dupT
c.2556dup	p.Ile853TyrfsX43	c.2556dupT
c.2562del	p.Val855SerfsX5	2694delT
c.2573del	p.Ser858ThrfsX2	c.2573delG
c.2576_2588del	p.Leu859X	2708del13
c.2589_2599del	p.Ile864SerfsX28	2721del11
c.2592_2593del	p.Ile864MetfsX31	2723delTT
c.2601dup	p.Val868SerfsX28	2732insA
c.2605del	p.Ile869PhefsX37	2737delA
c.2615del	p.Ala872GlufsX34	2747delC
c.2619+1G>A	No protein name	2751+1G->A
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.Trp882CysfsX25	2777insTG
c.2645G>A c.2646G>A	p.Trp882X	W882X
c.2655_2657+13del	No protein name	2787del16
c.2658-2A>G	No protein name	2790-2A->G
c.2658-1G>C	No protein name	2790-1G->C
c.2658-1G>T	No protein name	2790-1G->T
c.2705del	p.Ser902ThrfsX4	2837delG
c.2732_2733insA	p.Ser911ArgfsX64	c.2732_2733insA
c.2733_2734insA	p.Ser912IlefsX63	c.2733insA
c.2735C>A	p.Ser912X	S912X
c.2737_2738insG	p.Tyr913X	2869insG
c.2739T>A	p.Tyr913X	Y913X
c.2745_2746del	p.Phe916LeufsX58	c.2745_2746delGT
c.2763_2764dup	p.Val922GlufsX2	2896insAG
c.2789del	p.Gly930AspfsX12	c.2789delG
c.2803_2813del	p.Leu935AlafsX36	2935del11
c.2805_2810del	p.Pro936_Leu937del	2937_2942delinsTCAGA
c.2805_2810delinsTCAGA	p.Pro936GlnfsX6	c.2805_2810delinsTCAGA
c.2810dup	p.Val938GlyfsX37	2942insT

c.2816del	p.His939LeufsX3	2948delA
c.2818_2831del	p.Thr940ValfsX30	c.2818_2831del
c.2819_2820insA	p.Leu941SerfsX34	2951insA
c.2819_2823del	p.Thr940AsnfsX33	2949del5
c.2822del	p.Leu941GlnfsX27	2954delT
c.2825del	p.Ile942ThrfsX26	2957delT
c.2835del	p.Ile947PhefsX21	2967delG
c.2836A>T	p.Lys946X	K946X
c.2839del	p.Ile947PhefsX21	c.2839delA
c.2843del	p.Leu948TyrfsX20	2975delT
c.2851A>T	p.Lys951X	K951X
c.2859_2890del	p.Leu953PhefsX11	2991del32
c.2879_2882del	p.Pro960ArgfsX7	c.2879_2882delCTAT
c.2879del	p.Pro960LeufsX8	3011delC
c.2880del	p.Met961CysfsX7	3012delT
c.2883_2886dup	p.Thr963ValfsX13	3015_3018dupGTCA
c.2885C>A c.2885C>G	p.Ser962X	S962X
c.2896del	p.Thr966ArgfsX2	3028delA
c.2897del	p.Thr966SerfsX2	3029delC
c.2899_2900delinsA	p.Leu967ArgfsX14	3031-3032delinsA
c.2908+1G>A	No protein name	3040+1G->A
c.2908+1G>T	No protein name	3040+1G->T
c.2909_2924dup	p.Phe976TrpfsX4	c.2909_2924dup16
c.2909-1G>A	No protein name	3041-1G->A
c.2909-1G>C	No protein name	3041-1G->C
c.2909del	p.Gly970ValfsX11	3041delG
c.2909G>A	p.Gly970Asp	G970D
c.2924_2925del	p.Arg975IlefsX10	3056delGA
c.2929_2930insA	p.Ser977TyrfsX9	3061insA
c.2932A>T	p.Lys978X	K978X
c.2936A>T	p.Asp979Val	D979V
c.2958_2961del	p.Pro988LeufsX11	3090del4
c.2959_2960dup	p.Pro988CysfsX13	3089insTC
c.2964dup	p.Leu989SerfsX5	3095insT
c.2982_2988+2del	No protein name	c.2982_2988+2delCATCCAGGT
c.2986C>T	p.Gln996X	Q996X
c.2988+1G>C	No protein name	3120+1G->C
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.2994del	p.Leu998PhefsX2	3126delA
c.2997_3000del	p.Ile1000X	3129del4
c.2998del	p.Ile1000LeufsX2	3130delA
c.3002_3003del	p.Val1001AspfsX45	3132delTG
c.3007G>T	p.Gly1003X	G1003X
c.3008del	p.Gly1003GlnfsX3	3139delG

c.3011_3019del	p.Ala1004_Ala1006del	3143del9
c.3011del	p.Ala1004ValfsX2	3143delC
c.3017C>A	p.Ala1006Glu	A1006E
c.3021del	p.Val1008SerfsX15	3152delT
c.3022del	p.Val1008SerfsX15	3154delG
c.3032T>G	p.Leu1011X	L1011X
c.3033dup	p.Gln1012ThrfsX35	3165dupA
c.3034C>T	p.Gln1012X	Q1012X
c.3039del	p.Tyr1014ThrfsX9	3171delC
c.3039dup	p.Tyr1014LeufsX33	3171insC
c.3067_3072del	p.Ile1023_Val1024del	3199del6
c.3081dup	p.Met1028TyrfsX19	3213-3214insT
c.3103C>T	p.Gln1035X	Q1035X
c.3107C>A	p.Thr1036Asn	T1036N
c.3110C>A	p.Ser1037X	S1037X
c.3112C>T	p.Gln1038X	Q1038X
c.3123dup	p.Gln1042ThrfsX5	Q1042TfsX5
c.3124C>T	p.Gln1042X	Q1042X
c.3130G>T	p.Glu1044X	E1044X
c.3136G>T	p.Glu1046X	E1046X
c.3139_3139+1del	No protein name	3271delGG
c.3139+1del	No protein name	3271+1delG
c.3139+1G>A	No protein name	3271+1G->A
c.3139+1G>C	No protein name	3271+1G->C
c.3139+1G>T	No protein name	3271+1G->T
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.His1054LeufsX6	3293delA
c.3176T>G	p.Leu1059X	L1059X
c.3180del	p.Gly1061AspfsX22	c.3180delA
c.3181G>A c.3181G>C	p.Gly1061Arg	G1061R
c.3189G>A	p.Trp1063X	W1063X
c.3199del	p.Ala1067ProfsX16	c.3199delG
c.3217dup	p.Tyr1073LeufsX3	3349insT
c.3227_3231del	p.Thr1076IlefsX78	3359delCTCTG
c.3227del	p.Thr1076IlefsX7	3359delC
c.3229_3230del	p.Leu1077ValfsX78	3359delCT
c.3231_3232del	p.Phe1078ProfsX77	c.3231_3232delGT
c.3254A>C	p.His1085Pro	H1085P
c.3264del	p.Trp1089GlyfsX13	3396delC
c.3290_3305del	p.Arg1097GlnfsX2	3422del16
c.3292T>C	p.Trp1098Arg	W1098R
c.3293G>A c.3294G>A	p.Trp1098X	W1098X
c.3294G>C c.3294G>T	p.Trp1098Cys	W1098C
c.3298C>T	p.Gln1100X	Q1100X
c.3302T>G	p.Met1101Arg	M1101R

c.3304A>T	p.Arg1102X	R1102X
c.3315del	p.Met1105IlefsX16	3447delG
c.3321del	p.Phe1107LeufsX14	3453delT
c.3321dup	p.Val1108CysfsX48	c.3321dup
c.3322_3323del	p.Val1108HisfsX47	c.3322_3323delGT
c.3322del	p.Val1108SerfsX13	3454delG
c.3324del	p.Ile1109SerfsX12	3456delC
c.3325del	p.Ile1109SerfsX12	c.3325delA
c.3344_3345insA	p.Phe1116LeufsX40	c.3344_3345insA
c.3353C>T	p.Ser1118Phe	S1118F
c.3365del	p.Thr1122LysfsX12	3497delC
c.3367+1G>A	No protein name	3499+1G->A
c.3367+1G>T	No protein name	3499+1G->T
c.3367+2T>A	No protein name	3499+2T->A
c.3367+2T>C	No protein name	3499+2T->C
c.3368-1G>A	No protein name	3500-1G->A
c.3368-2A>G	No protein name	3500-2A->G
c.3368-2A>T	No protein name	3500-2A->T
c.3380_3383del	p.Gly1127GlufsX6	c.3380_3383del
c.3382A>T	p.Arg1128X	R1128X
c.3384_3388del	p.Val1129TyrfsX25	3516del5
c.3389_3402del	p.Gly1130ValfsX21	3521del14
c.3397del	p.Leu1133X	3528delC
c.3400_3401delinsGTA	p.Thr1134ValfsX22	3532AC->GTA
c.3407_3422del	p.Ala1136ValfsX7	3539del16
c.3411_3414del	p.Met1137IlefsX3	c.3411_3414delGAAT
c.3421_3424dup	p.Thr1142LysfsX15	3556insAGTA
c.3426_3427insGTAA	p.Leu1143ValfsX14	c.3426_3427insGTAA
c.3430C>T	p.Gln1144X	Q1144X
c.3434G>A c.3435G>A	p.Trp1145X	W1145X
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.3468G>A	p.Leu1156=	3600G->A
c.3469-1G>A	No protein name	3601-1G->A
c.3469-1G>C	No protein name	3601-1G->C
c.3469-1G>T	No protein name	3601-1G->T
c.3469-2A>C	No protein name	3601-2A->C
c.3469-2A>G	No protein name	3601-2A->G
c.3475T>C	p.Ser1159Pro	S1159P
c.3476C>T	p.Ser1159Phe	S1159F
c.3477del	p.Val1160X	c.3477delT
c.3481_3486delinsGG	p.Ser1161GlyfsX30	3613AGCCGA->GG
c.3486_3487del	p.Val1163LeufsX2	3617delGA
c.3492dup	p.Lys1165X	3622insT
c.3493A>T	p.Lys1165X	K1165X

c.3495del	p.Lys1165AsnfsX27	c.3495delG
c.3497del	p.Phe1166SerfsX26	3629delT
c.3524del	p.Pro1175LeufsX17	c.3524delC
c.3525_3537del	p.Thr1176AsnfsX12	c.3525_3537del
c.3530del	p.Lys1177SerfsX15	3662delA
c.3532_3535dup	p.Thr1179IlefsX17	3667ins4
c.3533C>A c.3533C>G	p.Ser1178X	S1178X
c.3536_3539del	p.Thr1179AsnfsX12	3667del4
c.3539_3554del	p.Lys1180ThrfsX7	c.3539_3554del
c.3546C>G	p.Tyr1182X	Y1182X
c.3556C>T	p.Gln1186X	Q1186X
c.3563del	p.Ser1188X	3695delC
c.3569_3570del	p.Val1190AspfsX4	c.3569_3570delTT
c.3592del	p.Val1198X	3724delG
c.3598delinsTCT	p.Lys1200SerfsX12	3730A->TCT
c.3599_3600delinsG	p.Lys1200ArgfsX11	3731-3732AA->G
c.3600del	p.Asp1201MetfsX10	3732delA
c.3605del	p.Asp1202AlafsX9	3737delA
c.3617C>G	p.Ser1206X	S1206X
c.3618_3619del	p.Gly1208ProfsX56	3750delAG
c.3618del	p.Gly1208AlafsX3	3750delA
c.3623del	p.Gly1208AlafsX3	3755delG
c.3623dup	p.Gln1209ProfsX56	3755dupG
c.3635del	p.Val1212AlafsX16	c.3635delT
c.3658dup	p.Thr1220AsnfsX45	3789insA
c.3678del	p.Leu1227X	c.3678delA
c.3691del	p.Ser1231ProfsX4	3821delT
c.3700A>G	p.Ile1234Val	I1234V
c.3708del	p.Gly1237AlafsX22	3840delT
c.3715del	p.Arg1239GlyfsX20	3847delA
c.3717G>A	p.Arg1239=	3849G->A
c.3717+1G>A	No protein name	3849+1G->A
c.3717+4A>G	No protein name	3849+4A->G
c.3717+5G>A	No protein name	3849+5G->A
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.Leu1242SerfsX17	3856delC
c.3728T>A	p.Leu1243X	L1243X
c.3739G>T	p.Gly1247X	G1247X
c.3743C>A c.3743C>G	p.Ser1248X	S1248X
c.3745G>A c.3745G>C	p.Gly1249Arg	G1249R
c.3747del	p.Lys1250ArgfsX9	3878delG
c.3754dup	p.Thr1252AsnfsX13	3886insA
c.3758dup	p.Leu1253PhefsX12	3889dupT
c.3760_3761del	p.Leu1254IlefsX10	3892delTT
c.3761T>A c.3761T>G	p.Leu1254X	L1254X

c.3763T>C	p.Ser1255Pro	S1255P
c.3764C>A	p.Ser1255X	S1255X
c.3767dup	p.Leu1258PhefsX7	3898insC
c.3773del	p.Leu1258X	3905delT
c.3773T>A	p.Leu1253X	L1253X
c.3796G>T	p.Glu1266X	E1266X
c.3806T>A	p.Ile1269Asn	I1269N
c.3808del	p.Asp1270MetfsX8	3940delG
c.3816_3817del	p.Ser1273LeufsX28	3944delGT
c.3822G>A	p.Trp1274X	W1274X
c.3827del	p.Ser1276X	3959delC
c.3829del	p.Ile1277X	3960-3961delA
c.3835_3836del	p.Leu1279AlafsX22	3967delTT
c.3838C>T	p.Gln1280X	Q1280X
c.3841C>T	p.Gln1281X	Q1281X
c.[3846G>A;3848G>T]		W1282X;R1283M
c.3848G>T	p.Arg1283Met	R1283M
c.3859G>T	p.Gly1287X	G1287X
c.3871C>T	p.Gln1291X	Q1291X
c.3873+1G>T	No protein name	4005+1G->T
c.3873+2T>C	No protein name	4005+2T->C
c.3874-2A>G	No protein name	4006-2A->G
c.3874-1G>A	No protein name	4006-1G->A
c.3876del	p.Val1293TyrfsX35	4006delA
c.3883_3884insG	p.Ile1295SerfsX7	c.3883_3884insG
c.3883_3886del	p.Ile1295PhefsX32	4010del4
c.3883del	p.Ile1295PhefsX33	4015delA
c.3889del	p.Ser1297LeufsX31	c.3889delT
c.3891dup	p.Gly1298TrpfsX4	4022insT
c.3893del	p.Gly1298GlufsX30	c.3893delG
c.3904A>T	p.Lys1302X	K1302X
c.3908del	p.Asn1303ThrfsX25	4040delA
c.3908dup	p.Asn1303LysfsX6	4040dupA
c.3917_3918dup	p.Tyr1307ProfsX22	4048insCC
c.3921T>A	p.Tyr1307X	Y1307X
c.3922G>T	p.Glu1308X	E1308X
c.3925C>T	p.Gln1309X	Q1309X
c.3929G>A c.3930G>A	p.Trp1310X	W1310X
c.3932_3933delinsAATATG	p.Ser1311LysfsX12	4064-4065delinsAATATG
c.3947G>A	p.Trp1316X	W1316X
c.3957_3958insAGGG	p.Asp1320ArgfsX3	4089ins4
c.3957del	p.Asp1320MetfsX8	4089delA
c.3963+1G>A	No protein name	4095+1G->A
c.3963+1G>C	No protein name	4095+1G->C
c.3963+1G>T	No protein name	4095+1G->T
c.3964-1G>A	No protein name	4096-1G->A
c.3971T>C	p.Leu1324Pro	L1324P

c.3976del	p.Ser1326LeufsX2	4108delT
c.3988_3989del	p.Gln1330ValfsX6	4120delCA
c.3988C>T	p.Gln1330X	Q1330X
c.3999del	p.Lys1334SerfsX13	c.3999delG
c.4004T>C	p.Leu1335Pro	L1335P
c.4028del	p.Gly1343AlafsX4	c.4028delG
c.4028dup	p.Cys1344LeufsX15	4160insG
c.4033_4034del	p.Val1345ProfsX13	4165delGT
c.4035_4038dup	p.Ser1347ProfsX13	p.S1347PfsX13
c.4036_4042del	p.Leu1346MetfsX6	4168delCTAAGCC
c.4036dup	p.Leu1346ProfsX13	c.4036dupC
c.4037_4041del	p.Leu1346ProfsX11	4168delCTAAG
c.4040_4041del	p.Ser1347ThrfsX11	4172delGC
c.4046del	p.Gly1349AlafsX5	4177delG
c.4046G>A	p.Gly1349Asp	G1349D
c.4051A>T	p.Lys1351X	K1351X
c.4065_4066del	p.Leu1356GlyfsX2	4197_4198delCT
c.4071_4073delinsAA	p.Arg1358AsnfsX22	4203TAG->AA
c.4078del	p.Val1360PhefsX20	c.4078delG
c.4086dup	p.Lys1363X	4218insT
c.4090del	p.Ala1364ArgfsX16	4222delG
c.4097T>A	p.Ile1366Asn	I1366N
c.4105_4110delinsAGAA	p.Leu1369ArgfsX16	4237-4242delinsAGAA
c.4111G>T	p.Glu1371X	E1371X
c.4124A>C	p.His1375Pro	H1375P
c.4127_4131del	p.Leu1376SerfsX8	4259del5
c.4136+2T>G	No protein name	4268+2T->G
c.4139del	p.Thr1380AsnfsX4	4271delC
c.4143C>A c.4143C>G	p.Tyr1381X	Y1381X
c.4144C>T	p.Gln1382X	Q1382X
c.4147dup	p.Ile1383AsnfsX3	4279insA
c.4163_4167del	p.Leu1388ProfsX5	c.4163_4167delTAAAA
c.4168C>T	p.Gln1390X	Q1390X
c.4170del	p.Ala1391HisfsX7	4301delA
c.4187del	p.Thr1396LysfsX2	c.4187delC
c.4197_4198del	p.Cys1400X	4326delTC
c.4200_4201del	p.Cys1400X	4332delTG
c.4200T>A	p.Cys1400X	C1400X
c.4201G>T	p.Glu1401X	E1401X
c.4225G>T	p.Glu1409X	E1409X
c.4230C>A	p.Cys1410X	C1410X
c.4231C>T	p.Gln1411X	Q1411X
c.4234C>T	p.Gln1412X	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.Ser1435GlyfsX14	4428insGA