

Expanded Variant Panel

Variant cDNA name (ordered 5' to 3')	Variant Protein Name	Variant Legacy Name
c.-9_14del23	No protein name	124del23bp
c.4C>T	p.Gln2*	Q2X
c.11C>A	p.Ser4*	S4X
c.38C>T	p.Ser13Phe	S13F
c.44T>C	p.Leu15Pro	L15P
c.50delT	p.Phe17Serfs*8	182delT
c.53+1G>T	No protein name	185+1G->T
c.57G>A	p.Trp19*	W19X
c.79G>T	p.Gly27*	G27X
c.79G>A	p.Gly27Arg	G27R
c.88C>T	p.Gln30*	Q30X
c.137C>A	p.Ala46Asp	A46D
c.164+1G>A	No protein name	296+1G->A
c.164+1G>T	No protein name	296+1G->T
c.164+2T>C	No protein name	296+2T->C
c.164+3_164+4insT	No protein name	296+3insT
c.165-1G>A	No protein name	297-1G->A
c.166G>A	p.Glu56Lys	E56K
c.168delA	p.Glu56Aspfs*35	300delA
c.169T>G	p.Trp57Gly	W57G
c.170G>A or c.171G>A	p.Trp57*	W57X
c.175dupA	p.Arg59Lysfs*10	306insA
c.174_177delTAGA	p.Asp58Glufs*32	306delTAGA
c.178G>A	p.Glu60Lys	E60K
c.233dupT	p.Trp79Leufs*32	365-366insT
c.263T>A or c.263T>G	p.Leu88*	L88X
c.271G>A	p.Gly91Arg	G91R
c.273+3A>C	No protein name	405+3A->C
c.274-2A>G	No protein name	406-2A->G
c.293A>G	p.Gln98Arg	Q98R
c.296C>T	p.Pro99Leu	P99L
c.305T>G	p.Leu102Arg	L102R
c.310delA	p.Arg104Glufs*3	442delA
c.313delA	p.Ile105Serfs*2	444delA

c.346G>A	p.Glu116Lys	E116K
c.350G>C	p.Arg117Pro	R117P
c.377G>A	p.Gly126Asp	G126D
c.409delC	p.Leu137Serfs*16	541delC
c.413_415dupTAC	p.Leu138dup	L138ins
c.416A>G	p.His139Arg	H139R
c.424delA	p.Ile142Phefs*11	556delA
c.429delT	p.Phe143Leufs*10	557delT
c.470_483del14	p.Phe157*	602del14
c.481T>G	p.Tyr161Asp	Y161D
c.494T>C	p.Leu165Ser	L165S
c.543_546delTAGT	p.Leu183Phefs*5	675del4
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.580-2A>G		712-2A->G
c.580G>A	p.Gly194Arg	G194R
c.647G>A	p.Trp216*	W216X
c.653T>A	p.Leu218*	L218X
c.680T>G	p.Leu227Arg	L227R
c.695T>A	p.Val232Asp	V232D
c.717delG	p.Leu240*	849delG
c.744-2A>G		876-2A->G
c.761delA	p.Lys254Argfs*7	892delA
c.803delA	p.Asn268Ilefs*17	935delA
c.825C>G	p.Tyr275*	Y275X
c.828C>A	p.Cys276*	C276X
c.850dupA	p.Met284Asnfs*3	977insA
c.861_865delCTTAA	p.Asn287Lysfs*19	991del5
c.933C>G	p.Phe311Leu	F311L
c.987delA	p.Gly330Glufs*39	1119delA
c.1001G>T	p.Arg334Leu	R334L
c.1006_1007insG	p.Ile336Serfs*28	1138insG
c.1029delC	p.Cys343*	1161delC
c.1037T>C	p.Leu346Pro	L346P
c.1075_1079delCAAACinsAAAAA	p.[Gln359Lys;Thr360Lys]	Q359K/T360K
c.1117-1G>A	No protein name	1249-1G->A

c.1135G>T	p.Glu379*	E379X
c.1155_115dupTA	p.Asn386lIefs*3	1288insTA
c.1210-2A>C		1342-2A->C
c.1211delG	p.Gly404Aspfs*38	1343delG
c.1240C>T	p.Gln414*	Q414X
c.1301C>A or c.1301C>G	p.Ser434*	S434X
c.1301_1307delCACTTCT	p.Ser434Leufs*6	1429del7
c.1330_1331delAT	p.Ile444*	1460delAT
c.1340delA	p.Lys447Argfs*2	1471delA
c.1358T>C	p.Leu453Ser	L453S
c.1365_1366delGG	p.Val456Cysfs*25	1497delGG
c.1367T>C	p.Val456Ala	V456A
c.1373delG	p.Gly458Aspfs*11	1504delG
c.1393-2A>G	No protein name	1525-2A->G
c.1420G>A	p.Glu474Lys	E474K
c.1477_1478delCA	p.Gln493Valfs*10	1609delCA
c.1487G>A	p.Trp496*	W496X
c.1505T>C	p.Ile502Thr	I502T
c.1538A>G	p.Asp513Gly	D513G
c.1572C>A	p.Cys524*	C524X
c.1584+1G>A	No protein name	1716+1G->A
c.1647T>A	p.Ser549Arg	S549R
c.1648G>T	p.Gly550*	G550X
c.1650delA	p.Gly551Valfs*8	1782delA
c.1651G>A	p.Gly551Ser	G551S
c.1670delC	p.Ser557Phefs*2	1802delC
c.1673T>C	p.Leu558Ser	L558S
c.1679+1G>A	No protein name	1811+1G->A
c.1679+1G>C	No protein name	1811+1G->C
c.1680-877G>T	No protein name	1811+1643G->T
c.1680A>C	p.Arg560Ser	R560S
c.1682C>A	p.Ala561Glu	A561E
c.1687T>A	p.Tyr563Asn	Y563N
c.1687T>G	p.Tyr563Asp	Y563D
c.1692delA	p.Asp565Metfs*7	1824delA
c.1703delT	p.Leu568Cysfs*4	1833delT
c.1705T>G	p.Tyr569Asp	Y569D
c.1721C>A	p.Pro574His	P574H

c.1766+1G>C	No protein name	1898+1G->C
c.1766+1G>T	No protein name	1898+1G->T
c.1766+5G>T	No protein name	1898+5G->T
c.1792_1798delAAAATA	p.Lys598Glyfs*11	1924del7
c.1801A>T	p.Ile601Phe	I601F
c.1820_1903del84	p.Met607_Gln634del	1949del84
c.1826A>G	p.His609Arg	H609R
c.1837G>A	p.Ala613Thr	A613T
c.1882G>C or c.1882G>A	p.Gly628Arg	G628R
c.1911delG	p.Gln637Hisfx*26	2043delG
c.1923_1931del9insA	p.Ser641Argfs*5	2055del9->A
c.1943delA	p.Asp648Valfs*15	2075delA
c.1966G>T	p.Glu66*	E656X
c.1973_1985del13insAGAAA	p.Arg658Lysfs*4	2105- 2117del13insAGAAA
c.1986_1989delAACT	p.Thr663Argfs*8	2118del4
c.1990G>T	p.Glu664*	E664X
c.2017G>T	p.Gly673*	G673X
c.2053dupC	p.Gln685Profs*84	2185insC
c.2053C>T	p.Gln685*	Q685X
c.2143C>T	p.Gln715*	Q715X
c.2158C>T	p.Gln720*	Q720X
c.2233G>T	p.Gly745*	G745X
c.2241_2248delGATACTGC	p.Ile748Serfs*28	2372del8
c.2353C>T	p.Arg785*	R785X
c.2374C>T	p.Arg792*	R792X
c.2423_2424dupAT	p.Ser809Ilefs*13	2556insAT
c.2463_2464delTG	p.Ser821Argfs*4	2594delGT
c.2502dupT	p.Asp835*	2634insT
c.2547C>A	p.Tyr849*	Y849X
c.2589_2599delAATTTGGTGCT	p.Ile864Serfs*28	2721del11
c.2601dupA	p.Val868Serfs*28	2732insA
c.2645G>A	p.Trp882*	W882X
c.2658-1G>C	No protein name	2790-1G->C
c.2735C>A	p.Ser912*	S912X
c.2737_2738insG	p.Tyr913*	2869insG
c.2739T>A	p.Tyr913*	Y913X
c.2763_2764dupAG	p.Val922Glufs*2	2896insAG

c.2810dupT	p.Val938Glyfs*37	2942insT
c.2822delT	p.Leu941Glnfs*27	2954delT
c.2825delT	p.Ile942Thrfs*26	2957delT
c.2859_2890delACATTCTGTTCTTCAAGCACCTATGTCAACCC	p.Leu953Phefs*11	2991del32
c.2896delA	p.Thr966Argfs*2	3028delA
c.2909G>A	p.Gly970Asp	G970D
c.2936A>T	p.Asp979Val	D979V
c.2989-2A>G	No protein name	3121-2A->G
c.3002_3003delTG	p.Val1001Aspfs*45	3132delTG
c.3011_3019delCTATAGCAG	p.Ala1004_Ala1006del	3143del9
c.3017C>A	p.Ala1006Glu	A1006E
c.3039dupC	p.Tyr1014Leufs*33	3171insC
c.3039delC	p.Tyr1014Thrfs*9	3171delC
c.3067_3072delATAGTG	p.Ile1023_Val1024del	3199del6
c.3103C>T	p.Gln1035*	Q1035X
c.3107C>A	p.Thr1036Asn	T1036N
c.3124C>T	p.Gln1042*	Q1042X
c.3139_3139+1delGG	p.Gly1047Glnfs*28	3271delGG
c.3160C>G	p.His1054Asp	H1054D
c.3181G>C	p.Gly1061Arg	G1061R
c.3217dupT	p.Tyr1073Leufs*3	3349insT
c.3292T>C	p.Trp1098Arg	W1098R
c.3293G>A or c.3294G>A	p.Trp1098*	W1098X
c.3294G>C or c.3294G>T	p.Trp1098Cys	W1098C
c.3302T>G	p.Met1101Arg	M1101R
c.3304A>T	p.Arg1102*	R1102X
c.3353C>T	p.Ser1118Phe	S1118F
c.3368-2A>G	No protein name	3500-2A->G
c.3382A>T	p.Arg1128*	R1128X
c.3435G>A	p.Trp1145*	W1145X
c.3468G>A	P.Leu1156Leu	3600G->A
c.3468+2dupT	No protein name	3600+2insT
c.3468+5G>A	No protein name	3600+5G->A
c.3475T>C	p.Ser1159Pro	S1159P
c.3476C>T	p.Ser1159Phe	S1159F
c.3532_3535dupTCAA	p.Thr1179Ilefs*17	3667ins4
c.3600delA	p.Asp1201Metfs*10	3732delA

c.3605delA	p.Asp1202Alafs*9	3737delA
c.3691delT	p.Ser1231Profs*4	3821delT
c.3700A>G	p.Ile1234Val	I1234V
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
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.3745G>A	p.Gly1249Arg	G1249R
c.3747delG	p.Lys1250Argfs*9	3878delG
c.3761T>G	p.Leu1254*	L1254X
c.3763T>C	p.Ser1255Pro	S1255P
c.3764C>A	p.Ser1255*	S1255X
c.3806T>A	p.Ile1269Asn	I1269N
c.3822G>A	p.Trp1274*	W1274X
c.[3846G>A;3848G>T]	p.[Trp1282*;Arg1283Met]	W1282X;R1283M
c.3848G>T	p.Arg1283Met	R1283M
c.3873+2T>C	No protein name	4005+2T->C
c.3883_3886delATTT	p.Ile1295Phefs*32	4010del4
c.3883delA	p.Ile1295Phefs*33	4015delA
c.3899dupT	p.Ser1297Phefs*5	4021dupT
c.3908delA	p.Asn1303Thrfs*25	4040delA
c.3929G>A	p.Trp1310*	W1310X
c.3971T>C	p.Leu1324Pro	L1324P
c.3988C>T	p.Gln1330*	Q1330X
c.4004T>C	p.Leu1335Pro	L1335P
c.4036_4042del	p.Leu1346Metfs*6	4168delCTAAGCC
c.4046G>A	p.Gly1349Asp	G1349D
c.4086dupT	p.Lys1363*	4218insT
c.4097T>A	p.Ile1366Asn	I1366N
c.4111G>T	p.Glu1371*	E1371X
c.4124A>C	p.His1375Pro	H1375P
c.4127_4131delTGGAT	p.Leu1376Serfs*8	4259del5
c.4144C>T	p.Gln1382*	Q1382X
c.4147dupA	p.Ile1383Asnfs*3	4279insA
c.4197_4198delICT	p.Cys1400*	4326delICT
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