DCLS Cystic Fibrosis Variant List

Legacy |HGVS Coding | HGVS Protein	Legacy |HGVS Coding | HGVS Protein	Legacy |HGVS Coding | HGVS Protein
124del23bp|c.-9_14del|p.Met1?	E33X|c.97G>T|p.(Glu33Ter)	R75X|c.223C>T|p.(Arg75Ter)
M1L|c.1A>C|p.Met1?	241delAT|c.112_113del|p.(Tyr38ProfsTer6)	365-366insT|c.233dup|p.(Trp79LeufsTer32)
M1V|c.1A>G|p.Met1?	Y38X|c.114C>G|p.(Tyr38Ter)	c.234delC|c.234del|p.(Trp79GlyfsTer12)
M1K|c.2T>A|p.Met1?	Q39X|c.115C>T|p.(Gln39Ter)	366insC|c.234dup|p.(Trp79LeufsTer32)
M1T|c.2T>C|p.Met1?	A46D|c.137C>A|p.(Ala46Asp)	W79X|c.236G>A|p.(Trp79Ter)
M1R|c.2T>G|p.Met1?	c.142_145delAATC|c.142_145del|p.(Asn48TyrfsTer42)	c.243delT|c.243del|p.(Phe81LeufsTer10)
M1I|c.3G>A|p.Met1?	c.147_150delATCT|c.147_150del|p.(Ser50LysfsTer40)	379-381insT|c.248dup|p.(Tyr84LeufsTer27)
c.4delC|c.4del|p.(Gln2ArgfsTer23)	284delA|c.156del|p.(Lys52AsnfsTer39)	Y84X|c.252T>A|p.(Tyr84Ter)
Q2X|c.4C>T|p.(Gln2Ter)	295ins8|c.156_163dup|p.(Arg55AsnfsTer39)	G85X|c.253G>T|p.(Gly85Ter)
S4X|c.11C>A|p.(Ser4Ter)	K52X|c.154A>T|p.(Lys52Ter)	G85E|c.254G>A|p.(Gly85Glu)
P5L|c.14C>T|p.(Pro5Leu)	E54X|c.160G>T|p.(Glu54Ter)	394delTT|c.262_263del|p.(Leu88IlefsTer22)
156insG|c.25dup|p.(Ala9GlyfsTer36)	296+1G->A|c.164+1G>A|p.?	c.264_268delATATT|c.264_268del|p.(Leu88PhefsTer21)
S13F|c.38C>T|p.(Ser13Phe)	296+1G->C|c.164+1G>C|p.?	L88X|c.263T>A|p.(Leu88Ter)
c.40_44delAAACT|c.40_44del|p.(Lys14PhefsTer29)	296+1G->T|c.164+1G>T|p.?	L88X|c.263T>G|p.(Leu88Ter)
174delA|c.42del|p.(Lys14AsnfsTer11)	296+2T->A|c.164+2T>A|p.?	G91R|c.271G>A|p.(Gly91Arg)
K14X|c.40A>T|p.(Lys14Ter)	296+2T->C|c.164+2T>C|p.?	405+1G->A|c.273+1G>A|p.?
c.43dupC|c.43dup|p.(Leu15ProfsTer30)	296+2T->G|c.164+2T>G|p.?	405+2T->G|c.273+2T>G|p.?
175delC|c.43del|p.(Leu15PhefsTer10)	296+3insT|c.164+4dup|p.?	405+3A->C|c.273+3A>C|p.?
175insT|c.50dup|p.(Ser18GlnfsTer27)	296+28A->G|c.164+28A>G|p.?	406-2A->C|c.274-2A>C|p.?
181_182dup|c.49_50dup|p.(Trp19AlafsTer7)	297-3C->T|c.165-3C>T|p.?	406-2A->G|c.274-2A>G|p.?
182delT|c.50del|p.(Phe17SerfsTer8)	297-2A->G|c.165-2A>G|p.?	406-1G->A|c.274-1G>A|p.?
c.49_50delTT|c.49_50del|p.(Phe17GlnfsTer27)	297-1G->A|c.165-1G>A|p.?	406-1G->C|c.274-1G>C|p.?
L15P|c.44T>C|p.(Leu15Pro)	E56K|c.166G>A|p.(Glu56Lys)	E92K|c.274G>A|p.(Glu92Lys)
183delC|c.51del|p.(Phe17LeufsTer8)	300delA|c.168del|p.(Glu56AspfsTer35)	E92X|c.274G>T|p.(Glu92Ter)
185+1G->T|c.53+1G>T|p.?	E56X|c.166G>T|p.(Glu56Ter)	415insA|c.285dup|p.(Ala96SerfsTer15)
185+2T->C|c.53+2T>C|p.?	W57G|c.169T>G|p.(Trp57Gly)	Q98X|c.292C>T|p.(Gln98Ter)
185+2T->G|c.53+2T>G|p.?	W57X|c.170G>A|p.(Trp57Ter)	Q98R|c.293A>G|p.(Gln98Arg)
186-2A->G|c.54-2A>G|p.?	W57X|c.171G>A|p.(Trp57Ter)	P99L|c.296C>T|p.(Pro99Leu)
186-1G->A|c.54-1G>A|p.?	306delTAGA|c.174_177del|p.(Asp58GlufsTer32)	L101X|c.302T>G|p.(Leu101Ter)
W19X|c.56G>A|p.(Trp19Ter)	305insT|c.174dup|p.(Arg59Ter)	435insA|c.303dup|p.(Leu102ThrfsTer9)
W19X|c.57G>A|p.(Trp19Ter)	306insA|c.175dup|p.(Arg59LysfsTer10)	L102R|c.305T>G|p.(Leu102Arg)
c.71_72delTGinsA|c.71_72delinsA|p.(Leu24Ter)	308insA|c.177dup|p.(Glu60ArgfsTer9)	G103X|c.307G>T|p.(Gly103Ter)
G27R|c.79G>A|p.(Gly27Arg)	E60K|c.178G>A|p.(Glu60Lys)	442delA|c.310del|p.(Arg104GlufsTer3)
G27X|c.79G>T|p.(Gly27Ter)	E60X|c.178G>T|p.(Glu60Ter)	R104X|c.310A>T|p.(Arg104Ter)
c.88_89delCA|c.88_89del|p.(Gln30AlafsTer14)	317insC|c.185dup|p.(Ser63PhefsTer6)	444delA|c.313del|p.(Ile105SerfsTer2)
218insA|c.87dup|p.(Gln30ThrfsTer15)	P67L|c.200C>T|p.(Pro67Leu)	c.324delC|c.324del|p.(Tyr109MetfsTer15)
Q30X|c.88C>T|p.(Gln30Ter)	K68X|c.202A>T|p.(Lys68Ter)	458delAT|c.326_327del|p.(Tyr109Ter)
R31L|c.92G>T|p.(Arg31Leu)	R74W|c.220C>T|p.(Arg74Trp)	457TAT->G|c.325_327delinsG|p.(Tyr109GlyfsTer4)

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Y109X	c.327T>A	p.(Tyr109Ter)	602del14	c.470_483del	p.(Phe157Ter)	711+1G->T	c.579+1G>T	p.?
D110H	c.328G>C	p.(Asp110His)	604insA	c.472dup	p.(Ser158LysfsTer5)	711+2T->C	c.579+2T>C	p.?
464delC	c.332del	p.(Pro111ArgfsTer13)	605insT	c.476dup	p.(Leu159PhefsTer4)	711+3A->G	c.579+3A>G	p.?
D110E	c.330C>A	p.(Asp110Glu)	L159X	c.476T>A	p.(Leu159Ter)	711+5G->A	c.579+5G>A	p.?
K114X	c.340A>T	p.(Lys114Ter)	Y161D	c.481T>G	p.(Tyr161Asp)	712-2A->C	c.580-2A>C	p.?
E116K	c.346G>A	p.(Glu116Lys)	c.488delA	c.488del	p.(Lys163ArgfsTer3)	712-2A->G	c.580-2A>G	p.?
R117G	c.349C>G	p.(Arg117Gly)	621+1G->A	c.489+1G>A	p.?	712-1G->T	c.580-1G>T	p.?
R117C	c.349C>T	p.(Arg117Cys)	621+1G->T	c.489+1G>T	p.?	G194R	c.580G>A	p.(Gly194Arg)
R117H	c.350G>A	p.(Arg117His)	621+2T->C	c.489+2T>C	p.?	G194R	c.580G>C	p.(Gly194Arg)
R117P	c.350G>C	p.(Arg117Pro)	621+2T->G	c.489+2T>G	p.?	713delGA	c.581_582del	p.(Gly194AlafsTer63)
R117L	c.350G>T	p.(Arg117Leu)	621+3A->G	c.489+3A>G	p.?	G194X	c.580G>T	p.(Gly194Ter)
c.353delC	c.353del	p.(Ser118LeufsTer6)	622-2A->C	c.490-2A>C	p.?	G194V	c.581G>T	p.(Gly194Val)
489delC	c.357del	p.(Ile119MetfsTer5)	622-1G->A	c.490-1G>A	p.?	c.583delC	c.583del	p.(Ala196HisfsTer19)
A120T	c.358G>A	p.(Ala120Thr)	622-1G->C	c.490-1G>C	p.?	H199Y	c.595C>T	p.(His199Tyr)
Y122X	c.366T>A	p.(Tyr122Ter)	622-1G->T	c.490-1G>T	p.?	733delG	c.601del	p.(Val201CysfsTer14)
499dupC	c.367dup	p.(Leu123ProfsTer36)	624delT	c.494del	p.(Leu165Ter)	c.604_605delTG	c.604_605del	p.(Trp202AspfsTer55)
G126D	c.377G>A	p.(Gly126Asp)	L165X	c.494T>A	p.(Leu165Ter)	V201M	c.601G>A	p.(Val201Met)
c.381_382dup	c.381_382dup	p.(Cys128TyrfsTer7)	L165S	c.494T>C	p.(Leu165Ser)	W202X	c.606G>A	p.(Trp202Ter)
519delT	c.387del	p.(Leu130SerfsTer4)	630delG	c.498del	p.(Lys166AsnfsTer7)	746insC	c.614dup	p.(Leu206PhefsTer52)
525delT	c.393del	p.(Phe131LeufsTer3)	654del5	c.522_526del	p.(Ile175TyrfsTer6)	P205S	c.613C>T	p.(Pro205Ser)
526_527delAT	c.394_398del	p.(Ile132GlufsTer25)	663insT	c.531dup	p.(Gly178TrpfsTer5)	749delT	c.617del	p.(Leu206CysfsTer9)
538insAC	c.405_406dup	p.(Leu136HisfsTer18)	663delT	c.531del	p.(Ile177MetfsTer12)	L206W	c.617T>G	p.(Leu206Trp)
541delC	c.409del	p.(Leu137SerfsTer16)	G178R	c.532G>A	p.(Gly178Arg)	Q207X	c.619C>T	p.(Gln207Ter)
541del4	c.409_412del	p.(Leu137TyrfsTer15)	Q179X	c.535C>T	p.(Gln179Ter)	W216X	c.647G>A	p.(Trp216Ter)
542del7	c.410_416del	p.(Leu137ProfsTer14)	675del4	c.543_546del	p.(Leu183PhefsTer5)	W216X	c.648G>A	p.(Trp216Ter)
L138ins	c.413_415dup	p.(Leu138dup)	675del14	c.543_556del	p.(Ser182GlnfsTer6)	c.650_659del	c.650_659del	p.(Glu217GlyfsTer11)
547insTA	c.415_416insTA	p.(His139LeufsTer15)	680delT	c.548del	p.(Leu183ProfsTer6)	L218X	c.653T>A	p.(Leu218Ter)
H139R	c.416A>G	p.(His139Arg)	c.551_555delTTTCC	c.551_555del	p.(Leu184GlnfsTer7)	Q220X	c.658C>T	p.(Gln220Ter)
556delA	c.424del	p.(Ile142PhefsTer11)	681delC	c.550del	p.(Leu184PhefsTer5)	C225X	c.675T>A	p.(Cys225Ter)
557delT	c.429del	p.(Phe143LeufsTer10)	690delC	c.558del	p.(Asn186LysfsTer3)	L227R	c.680T>G	p.(Leu227Arg)
565delC	c.433del	p.(Leu145PhefsTer8)	c.560delA	c.560del	p.(Asn187ThrfsTer2)	V232D	c.695T>A	p.(Val232Asp)
574delA	c.442del	p.(Ile148LeufsTer5)	F191V	c.571T>G	p.(Phe191Val)	840insT	c.708dup	p.(Gln237SerfsTer21)
G149R	c.445G>A	p.(Gly149Arg)	710_711+5del7	c.578_579+5del	p.?	840delT	c.708del	p.(Gln237ArgfsTer4)
c.450delG	c.450del	p.(Met150IlefsTer3)	D192G	c.575A>G	p.(Asp192Gly)	Q237E	c.709C>G	p.(Gln237Glu)
582insG	c.450dup	p.(Gln151AlafsTer8)	708delT	c.576del	p.(Asp192GlufsTer23)	Q237X	c.709C>T	p.(Gln237Ter)
583delC	c.451del	p.(Gln151ArgfsTer2)	E193K	c.577G>A	p.(Glu193Lys)	845_846insA	c.713_714insA	p.(Gly239TrpfsTer19)
Q151X	c.451C>T	p.(Gln151Ter)	710del4	c.579_579+3del	p.?	L240X	c.714del	p.(Leu240Ter)
593insT	c.461dup	p.(Ala155SerfsTer4)	E193X	c.577G>T	p.(Glu193Ter)	849delG	c.717del	p.(Leu240Ter)

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852del22	c.723_743+1del	p.?
Y247X	c.741C>A	p.(Tyr247Ter)
Y247X	c.741C>G	p.(Tyr247Ter)
875insTACA	c.743_743+1insTACA	p.?
875+1G->A	c.743+1G>A	p.?
875+1G->C	c.743+1G>C	p.?
875+2T->A	c.743+2T>A	p.?
875+2T->C	c.743+2T>C	p.?
876-2A->G	c.744-2A>G	p.?
Q250X	c.748C>T	p.(Gln250Ter)
892delA	c.761del	p.(Lys254ArgfsTer7)
896delT	c.764del	p.(Ile255ThrfsTer6)
R258G	c.772A>G	p.(Arg258Gly)
905delG	c.773del	p.(Arg258AsnfsTer3)
c.775delinsGCTGGGAAGAT	c.775delinsGCTGGGAAGAT	p.(Leu259AlafsTer12)
909delT	c.777del	p.(Val260Ter)
M265R	c.794T>G	p.(Met265Arg)
935delA	c.803del	p.(Asn268IlefsTer17)
936delTA	c.805_806del	p.(Ile269ProfsTer4)
Q270X	c.808C>T	p.(Gln270Ter)
c.811delT	c.811del	p.(Ser271LeufsTer14)
954delA	c.822del	p.(Tyr275ThrfsTer10)
Y275X	c.825C>G	p.(Tyr275Ter)
C276X	c.828C>A	p.(Cys276Ter)
W277X	c.830G>A	p.(Trp277Ter)
W277X	c.831G>A	p.(Trp277Ter)
E279X	c.835G>T	p.(Glu279Ter)
982delA	c.850del	p.(Met284Ter)
977insA	c.850dup	p.(Met284AsnfsTer3)
991del5	c.861_865del	p.(Asn287LysfsTer19)
994del9	c.863_869+2del	p.?
c.865_869delAGACA	c.865_869del	p.(Arg289AsnfsTer17)
R289X	c.865A>T	p.(Arg289Ter)
Q290X	c.868C>T	p.(Gln290Ter)
1001+1G->C	c.869+1G>C	p.?
1001+2T->A	c.869+2T>A	p.?
1001+2T->G	c.869+2T>G	p.?
1002-2A->C	c.870-2A>C	p.?
1002-2A->G	c.870-2A>G	p.?
1006_1007delGA	c.874_875del	p.(Glu292ThrfsTer15)
E292X	c.874G>T	p.(Glu292Ter)
1027delG	c.895del	p.(Ala299GlnfsTer4)
1037insA	c.905_906insA	p.(Arg303GlufsTer5)
1040del4	c.908_911del	p.(Arg303ThrfsTer24)
c.912delCinsGG	c.912delinsGG	p.(Tyr304Ter)
Y304X	c.912C>G	p.(Tyr304Ter)
1058delC	c.927del	p.(Phe310SerfsTer18)
F312del	c.935_937del	p.(Phe312del)
F311L	c.933C>A	p.(Phe311Leu)
F311L	c.933C>G	p.(Phe311Leu)
c.937_938delTC	c.937_938del	p.(Ser313ArgfsTer50)
c.942delG	c.942del	p.(Phe315SerfsTer13)
G314E	c.941G>A	p.(Gly314Glu)
1078delT	c.948del	p.(Phe316LeufsTer12)
L320X	c.959T>A	p.(Leu320Ter)
c.972delC	c.972del	p.(Tyr325MetfsTer3)
1112delT	c.980del	p.(Leu327GlnfsTer42)
c.982delA	c.982del	p.(Ile328SerfsTer41)
1119delA	c.987del	p.(Gly330GlufsTer39)
K329X	c.985A>T	p.(Lys329Ter)
G330X	c.988G>T	p.(Gly330Ter)
c.991_995del	c.991_995del	p.(Ile331ProfsTer31)
R334W	c.1000C>T	p.(Arg334Trp)
R334Q	c.1001G>A	p.(Arg334Gln)
R334L	c.1001G>T	p.(Arg334Leu)
1138insG	c.1006_1007insG	p.(Ile336SerfsTer28)
I336K	c.1007T>A	p.(Ile336Lys)
T338I	c.1013C>T	p.(Thr338Ile)
1154insTC	c.1021_1022dup	p.(Phe342HisfsTer28)
S341P	c.1021T>C	p.(Ser341Pro)
1157insTA	c.1025_1026insTA	p.(Cys343ThrfsTer27)
1161delC	c.1029del	p.(Cys343Ter)
L346P	c.1037T>C	p.(Leu346Pro)
R347H	c.1040G>A	p.(Arg347His)
R347P	c.1040G>C	p.(Arg347Pro)
A349V	c.1046C>T	p.(Ala349Val)
1185delTC	c.1053_1054del	p.(Arg352AlafsTer11)
R352W	c.1054C>T	p.(Arg352Trp)
R352Q	c.1055G>A	p.(Arg352Gln)
Q353X	c.1057C>T	p.(Gln353Ter)
1200_1219del20	c.1068_1087del	p.(Trp356Ter)
1199delG	c.1069del	p.(Ala357LeufsTer12)
W356X	c.1068G>A	p.(Trp356Ter)
1204_1205delGT	c.1072_1073del	p.(Val358ThrfsTer5)
Q359R	c.1076A>G	p.(Gln359Arg)
1212delA	c.1080del	p.(Trp361GlyfsTer8)
1213delT	c.1081del	p.(Trp361GlyfsTer8)
1215delG	c.1083del	p.(Trp361CysfsTer8)
W361X	c.1082G>A	p.(Trp361Ter)
W361X	c.1083G>A	p.(Trp361Ter)
c.1084_1088dup	c.1084_1088dup	p.(Ser364MetfsTer7)
Y362X	c.1086T>A	p.(Tyr362Ter)
Y362X	c.1086T>G	p.(Tyr362Ter)
1221delCT	c.1093_1094del	p.(Leu365TrpfsTer16)
c.1115delA	c.1115del	p.(Gln372ArgfsTer16)
Q372X	c.1114C>T	p.(Gln372Ter)
1248insATCAA	c.1116_1116+1insATCAA	p.?
1248+1G->A	c.1116+1G>A	p.?
1248+1G->C	c.1116+1G>C	p.?
1248+1G->T	c.1116+1G>T	p.?
1248+2T->A	c.1116+2T>A	p.?
1248+2T->C	c.1116+2T>C	p.?
1249-1G->A	c.1117-1G>A	p.?
1249insA	c.1118dup	p.(Asp373GlufsTer9)
1262delA	c.1130del	p.(Lys377SerfsTer11)
1259insA	c.1130dup	p.(Gln378AlafsTer4)
Q376X	c.1126C>T	p.(Gln376Ter)
Q378X	c.1132C>T	p.(Gln378Ter)

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E379X	c.1135G>T	p.(Glu379Ter)	S434X	c.1301C>A	p.(Ser434Ter)	1576insT	c.1446dup	p.(Lys483Ter)
K381X	c.1141A>T	p.(Lys381Ter)	1434delA	c.1302del	p.(Leu435PhefsTer7)	S489X	c.1466C>A	p.(Ser489Ter)
1288insTA	c.1155_1156dup	p.(Asn386IlefsTer3)	S434X	c.1301C>G	p.(Ser434Ter)	S489X	c.1466C>G	p.(Ser489Ter)
1288insA	c.1157dup	p.(Asn386LysfsTer25)	c.1312dupA	c.1312dup	p.(Thr438AsnfsTer8)	1601delT	c.1469del	p.(Phe490SerfsTer37)
1289insTA	c.1157_1158insTA	p.(Leu387ThrfsTer2)	D443fs	c.1329_1350del	p.(Asp443GlufsTer19)	1601delTC	c.1470_1471del	p.(Phe490LeufsTer13)
1291delTT	c.1159_1160del	p.(Leu387AsnfsTer23)	K442X	c.1324A>T	p.(Lys442Ter)	S492F	c.1475C>T	p.(Ser492Phe)
1294del7	c.1162_1168del	p.(Thr388GlnfsTer3)	1461ins4	c.1327_1330dup	p.(Ile444ArgfsTer3)	1609delCA	c.1477_1478del	p.(Gln493ValfsTer10)
T388X	c.1162_1163delinsTA	p.(Thr388Ter)	1460delAT	c.1330_1331del	p.(Ile444Ter)	Q493X	c.1477C>T	p.(Gln493Ter)
1309delG	c.1177del	p.(Val393Ter)	D443Y	c.1327G>T	p.(Asp443Tyr)	c.1486delT	c.1486del	p.(Trp496GlyfsTer31)
c.1190dupT	c.1190dup	p.(Thr398AsnfsTer13)	1465_1466insTAAT	c.1333_1334insTAAT	p.(Asn445IlefsTer38)	W496X	c.1487G>A	p.(Trp496Ter)
W401X	c.1202G>A	p.(Trp401Ter)	1471delA	c.1340del	p.(Lys447ArgfsTer2)	I502T	c.1505T>C	p.(Ile502Thr)
W401X	c.1203G>A	p.(Trp401Ter)	1474delA	c.1342del	p.(Ile448Ter)	E504X	c.1510G>T	p.(Glu504Ter)
E402X	c.1204G>T	p.(Glu402Ter)	Q452X	c.1354C>T	p.(Gln452Ter)	I507del	c.1519_1521del	p.(Ile507del)
1340delA	c.1208del	p.(Glu403GlyfsTer39)	L453S	c.1358T>C	p.(Leu453Ser)	F508del	c.1521_1523del	p.(Phe508del)
1341+1G->A	c.1209+1G>A	p.?	1491-1500del	c.1360_1387del	p.(Leu454AlafsTer6)	1660delG	c.1528del	p.(Val510PhefsTer17)
1341+1G->C	c.1209+1G>C	p.?	A455E	c.1364C>A	p.(Ala455Glu)	c.1530_1531delTT	c.1530_1531del	p.(Ser511LeufsTer2)
1341+1G->T	c.1209+1G>T	p.?	1498delG	c.1366del	p.(Val456LeufsTer13)	D513G	c.1538A>G	p.(Asp513Gly)
1341+2T->A	c.1209+2T>A	p.?	1497delGG	c.1365_1366del	p.(Val456CysfsTer25)	1675_1687del13	c.1543_1555del	p.(Tyr515AlafsTer8)
1341+2T->G	c.1209+2T>G	p.?	V456F	c.1366G>T	p.(Val456Phe)	E514X	c.1540G>T	p.(Glu514Ter)
5T;TG13	c.1210-11delinsGTG	p.?	V456A	c.1367T>C	p.(Val456Ala)	1677delTA	c.1545_1546del	p.(Tyr515Ter)
5T;TG12	c.1210-11T>G	p.?	1504delG	c.1373del	p.(Gly458AspfsTer11)	c.1547_1548delGA	c.1547_1548del	p.(Arg516IlefsTer51)
1342-2delAG	c.1210-2_1210-1del	p.?	c.1375_1385del	c.1375_1385del	p.(Ser459ArgfsTer19)	R516G	c.1546A>G	p.(Arg516Gly)
1342-2A->C	c.1210-2A>C	p.?	1524+1delG	c.1392+1del	p.?	Y517X	c.1551C>A	p.(Tyr517Ter)
1343delG	c.1211del	p.(Gly404AspfsTer38)	1524+1G->A	c.1392+1G>A	p.?	Y517X	c.1551C>G	p.(Tyr517Ter)
1342-1G->A	c.1210-1G>A	p.?	1525-2A->G	c.1393-2A>G	p.?	V520F	c.1558G>T	p.(Val520Phe)
1342-1G->C	c.1210-1G>C	p.?	1525-1G->A	c.1393-1G>A	p.?	c.1573delC	c.1573del	p.(Gln525AsnfsTer2)
1342-1G->T	c.1210-1G>T	p.?	1525-1G->C	c.1393-1G>C	p.?	C524X	c.1572C>A	p.(Cys524Ter)
c.1219delG	c.1219del	p.(Glu407AsnfsTer35)	1525-1G->T	c.1393-1G>T	p.?	1710del17bp	c.1579_1584+11del	p.?
1353delA	c.1221del	p.(Glu407AspfsTer35)	S466X	c.1397C>A	p.(Ser466Ter)	Q525X	c.1573C>T	p.(Gln525Ter)
E407X	c.1219G>T	p.(Glu407Ter)	S466X	c.1397C>G	p.(Ser466Ter)	c.1581dupA	c.1581dup	p.(Glu528ArgfsTer40)
1366delG	c.1234del	p.(Ala412GlnfsTer30)	L467P	c.1400T>C	p.(Leu467Pro)	E527X	c.1579G>T	p.(Glu527Ter)
1367del5	c.1243_1247del	p.(Asn415Ter)	1540del10	c.1409_1418del	p.(Val470GlufsTer54)	E528X	c.1582G>T	p.(Glu528Ter)
c.1244delA	c.1244del	p.(Asn415ThrfsTer27)	1548delG	c.1418del	p.(Gly473GlufsTer54)	1716+1G->A	c.1584+1G>A	p.?
Q414X	c.1240C>T	p.(Gln414Ter)	E474K	c.1420G>A	p.(Glu474Lys)	1716+1G->T	c.1584+1G>T	p.?
1380insT	c.1248dup	p.(Asn417Ter)	1556delT	c.1424del	p.(Leu475ArgfsTer52)	1716+2delT	c.1584+2del	p.?
c.1262delC	c.1262del	p.(Thr421IlefsTer21)	1565delCA	c.1433_1434del	p.(Ser478Ter)	1716+2T->C	c.1584+2T>C	p.?
1419delC	c.1287del	p.(Phe430SerfsTer12)	E479X	c.1435G>T	p.(Glu479Ter)	1717-8G->A	c.1585-8G>A	p.?
1429del7	c.1301_1307del	p.(Ser434LeufsTer6)	1571delG	c.1439del	p.(Gly480ValfsTer47)	1717-2A->G	c.1585-2A>G	p.?

Legacy	HGVS Coding	HGVS Protein	Legacy	HGVS Coding	HGVS Protein	Legacy	HGVS Coding	HGVS Protein
1717-1G->A|c.1585-1G>A|p.?			1812-2A->C|c.1680-2A>C|p.?			1898+2delT|c.1766+2del|p.?		
1717-1G->T|c.1585-1G>T|p.?			1812-1G->A|c.1680-1G>A|p.?			1898+2T->A|c.1766+2T>A|p.?		
S531X|c.1592_1593delinsG|p.(Ser531Ter)			1812-1G->C|c.1680-1G>C|p.?			1898+2T->C|c.1766+2T>C|p.?		
c.1608delA|c.1608del|p.(Asp537ThrfsTer3)			R560S|c.1680A>C|p.(Arg560Ser)			1898+3A->G|c.1766+3A>G|p.?		
K536X|c.1606A>T|p.(Lys536Ter)			R560S|c.1680A>T|p.(Arg560Ser)			1898+5G->T|c.1766+5G>T|p.?		
1749insTA|c.1616_1617dup|p.(Val540Ter)			1813insC|c.1682dup|p.(Val562SerfsTer6)			1898-2A->G|c.1767-2A>G|p.?		
G542X|c.1624G>T|p.(Gly542Ter)			A561E|c.1682C>A|p.(Ala561Glu)			1899-1G->A|c.1767-1G>A|p.?		
1759delG|c.1627del|p.(Glu543LysfsTer6)			Y563N|c.1687T>A|p.(Tyr563Asn)			1918delGC|c.1786_1787del|p.(Ala596Ter)		
1774delCT|c.1642_1643del|p.(Leu548GlufsTer19)			Y563D|c.1687T>G|p.(Tyr563Asp)			1924del7|c.1792_1798del|p.(Lys598GlyfsTer11)		
S549R|c.1645A>C|p.(Ser549Arg)			Y563X|c.1689C>A|p.(Tyr563Ter)			K598X|c.1792A>T|p.(Lys598Ter)		
S549N|c.1646G>A|p.(Ser549Asn)			1824delA|c.1692del|p.(Asp565MetfsTer7)			1932delG|c.1800del|p.(Ile601PhefsTer10)		
S549R|c.1647T>A|p.(Ser549Arg)			1833delT|c.1703del|p.(Leu568CysfsTer4)			I601F|c.1801A>T|p.(Ile601Phe)		
S549R|c.1647T>G|p.(Ser549Arg)			L568X|c.1703T>A|p.(Leu568Ter)			c.1807delG|c.1807del|p.(Val603SerfsTer8)		
G550X|c.1648G>T|p.(Gly550Ter)			Y569D|c.1705T>G|p.(Tyr569Asp)			1942del17|c.1810_1826del|p.(Thr604PhefsTer5)		
1782delA|c.1650del|p.(Gly551ValfsTer8)			c.1708_1712delTTATT|c.1708_1712del|p.(Leu570ArgfsTer17)			H609R|c.1826A>G|p.(His609Arg)		
1784delG|c.1652del|p.(Gly551ValfsTer8)			Y569X|c.1707T>A|p.(Tyr569Ter)			A613T|c.1837G>A|p.(Ala613Thr)		
G551S|c.1651G>A|p.(Gly551Ser)			L570X|c.1709T>A|p.(Leu570Ter)			D614G|c.1841A>G|p.(Asp614Gly)		
G551D|c.1652G>A|p.(Gly551Asp)			1845delAG/1846delGA|c.1714_1715del|p.(Asp572LeufsTer16)			1978delA|c.1846del|p.(Ile616TyrfsTer2)		
1784insATCAT|c.1652_1653insATCAT|p.(Gln552SerfsTer9)			P574H|c.1721C>A|p.(Pro574His)			1978dupA|c.1846dup|p.(Ile616AsnfsTer6)		
1787delA|c.1656del|p.(Gln552HisfsTer7)			1857delT|c.1725del|p.(Phe575LeufsTer4)			c.1853_1863del|c.1853_1863del|p.(Ile618ArgfsTer2)		
Q552X|c.1654C>T|p.(Gln552Ter)			F575Y|c.1724T>A|p.(Phe575Tyr)			I618T|c.1853T>C|p.(Ile618Thr)		
R553X|c.1657C>T|p.(Arg553Ter)			c.1725_1727delinsAT|c.1725_1727delinsAT|p.(Phe575LeufsTer4)			G622D|c.1865G>A|p.(Gly622Asp)		
R555X|c.1663A>T|p.(Arg555Ter)			Y577X|c.1731C>A|p.(Tyr577Ter)			2003del8|c.1871_1878del|p.(Ser624IlefsTer15)		
1802delC|c.1670del|p.(Ser557PhefsTer2)			D579G|c.1736A>G|p.(Asp579Gly)			2005delTA|c.1874_1875del|p.(Tyr625PhefsTer16)		
1806delA|c.1674del|p.(Ala559GlnfsTer13)			1870delG|c.1738del|p.(Val580PhefsTer2)			G628R|c.1882G>A|p.(Gly628Arg)		
L558S|c.1673T>C|p.(Leu558Ser)			1874insT|c.1742dup|p.(Leu581PhefsTer8)			G628R|c.1882G>C|p.(Gly628Arg)		
1807delG|c.1675del|p.(Ala559GlnfsTer13)			1874_1875insTT|c.1741_1742dup|p.(Leu581PhefsTer2)			c.1894_1895del|c.1894_1895del|p.(Glu632ThrfsTer9)		
A559T|c.1675G>A|p.(Ala559Thr)			L581X|c.1742T>A|p.(Leu581Ter)			2025dupA|c.1893dup|p.(Glu632ArgfsTer10)		
R560K|c.1679G>A|p.(Arg560Lys)			c.1753delG|c.1753del|p.(Glu585LysfsTer10)			S631X|c.1892C>G|p.(Ser631Ter)		
R560T|c.1679G>C|p.(Arg560Thr)			E585X|c.1753G>T|p.(Glu585Ter)			2036delA|c.1904del|p.(Asn635IlefsTer28)		
1811+1G->A|c.1679+1G>A|p.?			E588X|c.1762G>T|p.(Glu588Ter)			Q634X|c.1900C>T|p.(Gln634Ter)		
1811+1G->C|c.1679+1G>C|p.?			E588V|c.1763A>T|p.(Glu588Val)			2043delG|c.1911del|p.(Gln637HisfsTer26)		
1811+2T->C|c.1679+2T>C|p.?			1898+1G->A|c.1766+1G>A|p.?			2053insTA|c.1920_1921dup|p.(Ser641IlefsTer23)		
1811+1634A->G|c.1680-886A>G|p.?			1898+1G->C|c.1766+1G>C|p.?			2055del9->A|c.1923_1931delinsA|p.(Ser641ArgfsTer5)		
1811+1643G->T|c.1680-877G>T|p.?			1898+1G->T|c.1766+1G>T|p.?			G646X|c.1936G>T|p.(Gly646Ter)		

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Legacy |HGVS Coding | HGVS Protein	Legacy |HGVS Coding | HGVS Protein	Legacy |HGVS Coding | HGVS Protein
2075delA|c.1943del|p.(Asp648ValfsTer15)	2189CT->A|c.2057_2058delinsA|p.(Ser686TyrfsTer36)	2466delC|c.2335del|p.(Gln779LysfsTer24)
Q652X|c.1954C>T|p.(Gln652Ter)	2193ins4|c.2058_2061dup|p.(Lys688PhefsTer2)	c.2337delA|c.2337del|p.(Gly780ValfsTer23)
E656X|c.1966G>T|p.(Glu656Ter)	K688X|c.2062A>T|p.(Lys688Ter)	Q779X|c.2335C>T|p.(Gln779Ter)
c.1970delG|c.1970del|p.(Arg657LysfsTer6)	E692X|c.2074G>T|p.(Glu692Ter)	2472_2478del7|c.2340_2346del|p.(Gln781PhefsTer20)
R657X|c.1969A>T|p.(Arg657Ter)	2215delG|c.2083del|p.(Glu695LysfsTer27)	c.2341delC|c.2341del|p.(Gln781ArgfsTer22)
2104insA|c.1972dup|p.(Arg658LysfsTer7)	c.2089delA|c.2089del|p.(Arg697GlyfsTer25)	Q781X|c.2341C>T|p.(Gln781Ter)
2105-2117del13insAGAAA|c.1973_1985delinsAGAAA|p.(Arg658LysfsTer4)	2221insA|c.2089dup|p.(Arg697LysfsTer33)	2481-2482insT|c.2349dup|p.(His784SerfsTer21)
2108delA|c.1976del|p.(Asn659IlefsTer4)	2222delG|c.2091del|p.(Lys698ArgfsTer24)	R785X|c.2353C>T|p.(Arg785Ter)
2109-2118del10|c.1977_1986del|p.(Asn659LysfsTer10)	R709X|c.2125C>T|p.(Arg709Ter)	c.2357dupA|c.2357dup|p.(Thr787AspfsTer18)
S660X|c.1979C>A|p.(Ser660Ter)	K710X|c.2128A>T|p.(Lys710Ter)	R792X|c.2374C>T|p.(Arg792Ter)
2113delA|c.1981del|p.(Ile661SerfsTer2)	Q715X|c.2143C>T|p.(Gln715Ter)	2510delAA|c.2378_2379del|p.(Lys793SerfsTer11)
S660X|c.1979C>G|p.(Ser660Ter)	K716X|c.2145_2146delinsGT|p.(Lys716Ter)	2512delG|c.2380del|p.(Val794CysfsTer9)
2114delT|c.1982del|p.(Ile661ThrfsTer2)	K716X|c.2146A>T|p.(Lys716Ter)	2522insC|c.2393dup|p.(Gln799SerfsTer6)
2118del4|c.1986_1989del|p.(Thr663ArgfsTer8)	c.2148delG|c.2148del|p.(Thr717LeufsTer5)	Q799X|c.2395C>T|p.(Gln799Ter)
2118del14|c.1987_2000del|p.(Thr663ProfsTer21)	L719X|c.2156T>A|p.(Leu719Ter)	2556insAT|c.2423_2424dup|p.(Ser809IlefsTer13)
E664X|c.1990G>T|p.(Glu664Ter)	2290del16|c.2158_2173del|p.(Gln720LysfsTer8)	2557delT|c.2425del|p.(Ser809GlnfsTer12)
2132delAC|c.2000_2001del|p.(His667ProfsTer21)	Q720X|c.2158C>T|p.(Gln720Ter)	R810X|c.2428A>T|p.(Arg810Ter)
2143delT|c.2012del|p.(Leu671Ter)	2307insA|c.2175dup|p.(Glu726ArgfsTer4)	c.2433_2437delinsATAÂ |c.2433_2437delinsATA|p.(Leu812TyrfsTer10)
G673X|c.2017G>T|p.(Gly673Ter)	E730X|c.2188G>T|p.(Glu730Ter)	L812X|c.2435T>A|p.(Leu812Ter)
c.2032_2033delTC|c.2032_2033del|p.(Ser678LeufsTer10)	L732X|c.2195T>G|p.(Leu732Ter)	Q814X|c.2440C>T|p.(Gln814Ter)
W679X|c.2036G>A|p.(Trp679Ter)	2335delA|c.2203del|p.(Arg735GlyfsTer4)	E815X|c.2443G>T|p.(Glu815Ter)
W679X|c.2037G>A|p.(Trp679Ter)	2347delG|c.2215del|p.(Val739TyrfsTer16)	2585delT|c.2453del|p.(Leu818TrpfsTer3)
2175insA|c.2044dup|p.(Thr682AsnfsTer7)	G745X|c.2233G>T|p.(Gly745Ter)	2586-2687insT|c.2454_2455insT|p.(Glu819Ter)
2176insC|c.2045dup|p.(Gln685ThrfsTer4)	2372del8|c.2241_2248del|p.(Ile748SerfsTer28)	2594delGT|c.2463_2464del|p.(Ser821ArgfsTer4)
c.2045delC|c.2045del|p.(Thr682LysfsTer40)	P750L|c.2249C>T|p.(Pro750Leu)	c.2467_2470delGAAA|c.2467_2470del|p.(Glu823LeufsTer6)
2183delAA|c.2051_2052del|p.(Lys684ThrfsTer4)	c.2250delT|c.2250del|p.(Arg751AlafsTer4)	E822X|c.2464G>T|p.(Glu822Ter)
2184delA|c.2052del|p.(Lys684AsnfsTer38)	c.2261delT|c.2261del|p.(Val754GlyfsTer17)	2603delT|c.2472del|p.(Asn825ThrfsTer5)
2184insA|c.2052dup|p.(Gln685ThrfsTer4)	2406_2407delinsT|c.2274_2275delinsT|p.(Thr760ArgfsTer11)	E826X|c.2476G>T|p.(Glu826Ter)
2184del4|c.2052_2055del|p.(Lys684AsnfsTer37)	2409delC|c.2277del|p.(Thr760ArgfsTer11)	c.2489_2490insA|c.2489dup|p.(Glu831GlyfsTer5)
2183AA->G|c.2051_2052delinsG|p.(Lys684SerfsTer38)	R764X|c.2290C>T|p.(Arg764Ter)	K830X|c.2488A>T|p.(Lys830Ter)
2185insC|c.2053dup|p.(Gln685ProfsTer4)	2429delG|c.2298del|p.(Arg766SerfsTer5)	2622+1G->A|c.2490+1G>A|p.?
c.2053delC|c.2053del|p.(Gln685AsnfsTer37)	Q767X|c.2299C>T|p.(Gln767Ter)	2623-2A->G|c.2491-2A>G|p.?
c.2053_2054insAA|c.2054_2055dup|p.(Ser686AsnfsTer37)	2435insC|c.2303dup|p.(Val769CysfsTer10)	2623-1G->C|c.2491-1G>C|p.?
Q685X|c.2053C>T|p.(Gln685Ter)	2456delAC|c.2324_2325del|p.(His775LeufsTer3)	E831X|c.2491G>T|p.(Glu831Ter)

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c.2493delG|c.2493del|p.(Glu831AspfsTer13)	2790-1G->T|c.2658-1G>T|p.?	3007delG|c.2875del|p.(Ala959HisfsTer9)
2634insT|c.2502dup|p.(Asp835Ter)	Q890X|c.2668C>T|p.(Gln890Ter)	3011delC|c.2879del|p.(Pro960LeufsTer8)
2634delT|c.2502del|p.(Phe834LeufsTer10)	2837delG|c.2705del|p.(Ser902ThrfsTer4)	c.2879_2882delCTAT|c.2879_2882del|p.(Pro960ArgfsTer7)
2640delT|c.2508del|p.(Asp836GlufsTer8)	c.2732_2733insA|c.2732_2733insA|p.(Ser911ArgfsTer64)	3012delT|c.2880del|p.(Met961CysfsTer7)
2655del26|c.2523_2548del|p.(Ala842SerfsTer45)	2733insA|c.2733_2734insA|p.(Ser912IlefsTer63)	3015_3018dupGTCA|c.2883_2886dup|p.(Thr963ValfsTer13)
c.2538delG|c.2538del|p.(Trp846Ter)	S912X|c.2735C>A|p.(Ser912Ter)	S962X|c.2885C>A|p.(Ser962Ter)
W846X|c.2537G>A|p.(Trp846Ter)	S912L|c.2735C>T|p.(Ser912Leu)	S962X|c.2885C>G|p.(Ser962Ter)
W846X|c.2538G>A|p.(Trp846Ter)	2869insG|c.2737_2738insG|p.(Tyr913Ter)	3028delA|c.2896del|p.(Thr966ArgfsTer2)
2672delA|c.2540del|p.(Asn847ThrfsTer13)	Y913X|c.2739T>A|p.(Tyr913Ter)	3029delC|c.2897del|p.(Thr966SerfsTer2)
Y849X|c.2547C>A|p.(Tyr849Ter)	c.2745_2746delGT|c.2745_2746del|p.(Phe916LeufsTer58)	3031-3032delinsA|c.2899_2900delinsA|p.(Leu967ArgfsTer14)
R851X|c.2551C>T|p.(Arg851Ter)	2896insAG|c.2763_2764dup|p.(Val922GlufsTer2)	G970R|c.2908G>C|p.(Gly970Arg)
2686dupT|c.2554dup|p.(Tyr852LeufsTer44)	D924N|c.2770G>A|p.(Asp924Asn)	3040+1G->A|c.2908+1G>A|p.?
c.2556dupT|c.2556dup|p.(Ile853TyrfsTer43)	2909delT|c.2777del|p.(Leu926CysfsTer16)	3040+1G->T|c.2908+1G>T|p.?
2694delT|c.2562del|p.(Val855SerfsTer5)	2907delTT|c.2776_2777del|p.(Leu926AlafsTer48)	c.2909_2924dup16|c.2909_2924dup|p.(Phe976TrpfsTer4)
c.2573delG|c.2573del|p.(Ser858ThrfsTer2)	L927P|c.2780T>C|p.(Leu927Pro)	3041delG|c.2909del|p.(Gly970ValfsTer11)
2708del13|c.2576_2588del|p.(Leu859Ter)	c.2789delG|c.2789del|p.(Gly930AspfsTer12)	3041-1G->A|c.2909-1G>A|p.?
2711delT|c.2583del|p.(Phe861LeufsTer3)	R933G|c.2797A>G|p.(Arg933Gly)	3041-1G->C|c.2909-1G>C|p.?
2721del11|c.2589_2599del|p.(Ile864SerfsTer28)	2935del11|c.2803_2813del|p.(Leu935AlafsTer36)	G970D|c.2909G>A|p.(Gly970Asp)
2723delTT|c.2592_2593del|p.(Ile864MetfsTer31)	2937_2942delinsTCAGA|c.2805_2810del|p.(Pro936_Leu937del)	3056delGA|c.2924_2925del|p.(Arg975IlefsTer10)
2732insA|c.2601dup|p.(Val868SerfsTer28)	c.2805_2810delinsTCAGA|c.2805_2810delinsTCAGA|p.(Pro936GlnfsTer6)	3061insA|c.2929_2930insA|p.(Ser977TyrfsTer9)
2737delA|c.2605del|p.(Ile869PhefsTer37)	2942insT|c.2810dup|p.(Val938GlyfsTer37)	S977F|c.2930C>T|p.(Ser977Phe)
2747delC|c.2615del|p.(Ala872GlufsTer34)	2948delA|c.2816del|p.(His939LeufsTer3)	K978X|c.2932A>T|p.(Lys978Ter)
2751+1G->A|c.2619+1G>A|p.?	2949del5|c.2819_2823del|p.(Thr940AsnfsTer33)	D979V|c.2936A>T|p.(Asp979Val)
2751+2T->A|c.2619+2T>A|p.?	c.2818_2831del|c.2818_2831del|p.(Thr940ValfsTer30)	3090del4|c.2958_2961del|p.(Pro988LeufsTer11)
2751+2T->C|c.2619+2T>C|p.?	2951insA|c.2819_2820insA|p.(Leu941SerfsTer34)	3089insTC|c.2959_2960dup|p.(Pro988CysfsTer13)
2752-2A->G|c.2620-2A>G|p.?	2954delT|c.2822del|p.(Leu941GlnfsTer27)	3095insT|c.2964dup|p.(Leu989SerfsTer5)
2752-1G->T|c.2620-1G>T|p.?	2957delT|c.2825del|p.(Ile942ThrfsTer26)	c.2982_2988+2delCATCCAGGT|c.2982_2988+2del|p.?
2777insTG|c.2644_2645dup|p.(Trp882CysfsTer25)	2967delG|c.2835del|p.(Ile947PhefsTer21)	Q996X|c.2986C>T|p.(Gln996Ter)
W882X|c.2645G>A|p.(Trp882Ter)	S945L|c.2834C>T|p.(Ser945Leu)	3120G->A|c.2988G>A|p.(Gln996=)
W882X|c.2646G>A|p.(Trp882Ter)	c.2839delA|c.2839del|p.(Ile947PhefsTer21)	3120+1G->A|c.2988+1G>A|p.?
2787del16|c.2655_2657+13del|p.?	K946X|c.2836A>T|p.(Lys946Ter)	3120+1G->C|c.2988+1G>C|p.?
2789+2insA|c.2657+2_2657+3insA|p.?	2975delT|c.2843del|p.(Leu948TyrfsTer20)	3121-2A->C|c.2989-2A>C|p.?
2789+5G->A|c.2657+5G>A|p.?	K951X|c.2851A>T|p.(Lys951Ter)	3121-2A->G|c.2989-2A>G|p.?
2790-2A->G|c.2658-2A>G|p.?	M952T|c.2855T>C|p.(Met952Thr)	3121-2A->T|c.2989-2A>T|p.?
2790-1G->C|c.2658-1G>C|p.?	2991del32|c.2859_2890del|p.(Leu953PhefsTer11)	3121-1G->A|c.2989-1G>A|p.?

Legacy |HGVS Coding | HGVS Protein	Legacy |HGVS Coding | HGVS Protein	Legacy |HGVS Coding | HGVS Protein
3121-1G->T|c.2989-1G>T|p.?	3271+2T->C|c.3139+2T>C|p.?	W1098X|c.3294G>A|p.(Trp1098Ter)
3126delA|c.2994del|p.(Leu998PhefsTer2)	3272-26A->G|c.3140-26A>G|p.?	W1098C|c.3294G>C|p.(Trp1098Cys)
3129del4|c.2997_3000del|p.(Ile1000Ter)	3272-1G->A|c.3140-1G>A|p.?	W1098C|c.3294G>T|p.(Trp1098Cys)
3130delA|c.2998del|p.(Ile1000LeufsTer2)	F1052V|c.3154T>G|p.(Phe1052Val)	F1099L|c.3297C>A|p.(Phe1099Leu)
3132delTG|c.3002_3003del|p.(Val1001AspfsTer45)	3293delA|c.3161del|p.(His1054LeufsTer6)	Q1100X|c.3298C>T|p.(Gln1100Ter)
3139delG|c.3008del|p.(Gly1003GlufsTer3)	H1054D|c.3160C>G|p.(His1054Asp)	M1101K|c.3302T>A|p.(Met1101Lys)
G1003X|c.3007G>T|p.(Gly1003Ter)	c.3180delA|c.3180del|p.(Gly1061AspfsTer22)	M1101R|c.3302T>G|p.(Met1101Arg)
3143del9|c.3011_3019del|p.(Ala1004_Ala1006del)	L1059X|c.3176T>G|p.(Leu1059Ter)	R1102X|c.3304A>T|p.(Arg1102Ter)
3143delC|c.3011del|p.(Ala1004ValfsTer2)	G1061R|c.3181G>A|p.(Gly1061Arg)	E1104X|c.3310G>T|p.(Glu1104Ter)
A1006E|c.3017C>A|p.(Ala1006Glu)	G1061R|c.3181G>C|p.(Gly1061Arg)	3447delG|c.3315del|p.(Met1105IlefsTer16)
3152delT|c.3021del|p.(Val1008SerfsTer15)	W1063X|c.3189G>A|p.(Trp1063Ter)	c.3321dup|c.3321dup|p.(Val1108CysfsTer48)
3154delG|c.3022del|p.(Val1008SerfsTer15)	L1065P|c.3194T>C|p.(Leu1065Pro)	3453delT|c.3321del|p.(Phe1107LeufsTer14)
L1011X|c.3032T>G|p.(Leu1011Ter)	R1066C|c.3196C>T|p.(Arg1066Cys)	c.3322_3323delGT|c.3322_3323del|p.(Val1108HisfsTer47)
3165dupA|c.3033dup|p.(Gln1012ThrfsTer35)	R1066H|c.3197G>A|p.(Arg1066His)	3454delG|c.3322del|p.(Val1108SerfsTer13)
Q1012X|c.3034C>T|p.(Gln1012Ter)	c.3199delG|c.3199del|p.(Ala1067ProfsTer16)	3456delC|c.3324del|p.(Ile1109SerfsTer12)
3171insC|c.3039dup|p.(Tyr1014LeufsTer33)	G1069R|c.3205G>A|p.(Gly1069Arg)	c.3325delA|c.3325del|p.(Ile1109SerfsTer12)
3171delC|c.3039del|p.(Tyr1014ThrfsTer9)	R1070W|c.3208C>T|p.(Arg1070Trp)	c.3344_3345insA|c.3344_3345insA|p.(Phe1116LeufsTer40)
Y1014C|c.3041A>G|p.(Tyr1014Cys)	R1070Q|c.3209G>A|p.(Arg1070Gln)	S1118F|c.3353C>T|p.(Ser1118Phe)
F1016S|c.3047T>C|p.(Phe1016Ser)	3349insT|c.3217dup|p.(Tyr1073LeufsTer3)	3497delC|c.3365del|p.(Thr1122LysfsTer12)
3199del6|c.3067_3072del|p.(Ile1023_Val1024del)	F1074L|c.3220T>C|p.(Phe1074Leu)	3499+1G->A|c.3367+1G>A|p.?
3213-3214insT|c.3081dup|p.(Met1028TyrfsTer19)	F1074L|c.3222T>A|p.(Phe1074Leu)	3499+1G->T|c.3367+1G>T|p.?
Y1032C|c.3095A>G|p.(Tyr1032Cys)	F1074L|c.3222T>G|p.(Phe1074Leu)	3499+2T->A|c.3367+2T>A|p.?
3238delA|c.3106del|p.(Thr1036ProfsTer24)	3359delC|c.3227del|p.(Thr1076IlefsTer7)	3499+2T->C|c.3367+2T>C|p.?
Q1035X|c.3103C>T|p.(Gln1035Ter)	3359delCT|c.3229_3230del|p.(Leu1077ValfsTer78)	3500-2A->G|c.3368-2A>G|p.?
T1036N|c.3107C>A|p.(Thr1036Asn)	3359delCTCTG|c.3227_3231del|p.(Thr1076IlefsTer78)	3500-2A->T|c.3368-2A>T|p.?
S1037X|c.3110C>A|p.(Ser1037Ter)	c.3231_3232delGT|c.3231_3232del|p.(Phe1078ProfsTer77)	3500-1G->A|c.3368-1G>A|p.?
Q1038X|c.3112C>T|p.(Gln1038Ter)	L1077P|c.3230T>C|p.(Leu1077Pro)	c.3380_3383del|c.3380_3383del|p.(Gly1127GlufsTer6)
Q1042TfsX5|c.3123dup|p.(Gln1042ThrfsTer5)	H1085P|c.3254A>C|p.(His1085Pro)	3516del5|c.3384_3388del|p.(Val1129TyrfsTer25)
Q1042X|c.3124C>T|p.(Gln1042Ter)	3396delC|c.3264del|p.(Trp1089GlyfsTer13)	R1128X|c.3382A>T|p.(Arg1128Ter)
E1044X|c.3130G>T|p.(Glu1044Ter)	W1089X|c.3266G>A|p.(Trp1089Ter)	3521del14|c.3389_3402del|p.(Gly1130ValfsTer21)
E1046X|c.3136G>T|p.(Glu1046Ter)	Y1092X|c.3276C>A|p.(Tyr1092Ter)	3528delC|c.3397del|p.(Leu1133Ter)
3271+1delG|c.3139+1del|p.?	Y1092X|c.3276C>G|p.(Tyr1092Ter)	3532AC->GTA|c.3400_3401delinsGTA|p.(Thr1134ValfsTer22)
3271delGG|c.3139_3139+1del|p.?	3422del16|c.3290_3305del|p.(Arg1097GlnfsTer2)	3539del16|c.3407_3422del|p.(Ala1136ValfsTer7)
3271+1G->A|c.3139+1G>A|p.?	W1098R|c.3292T>C|p.(Trp1098Arg)	c.3411_3414delGAAT|c.3411_3414del|p.(Met1137IlefsTer3)
3271+1G->C|c.3139+1G>C|p.?	3425delG|c.3294del|p.(Trp1098CysfsTer4)	3556insAGTA|c.3421_3424dup|p.(Thr1142LysfsTer15)
3271+1G->T|c.3139+1G>T|p.?	W1098X|c.3293G>A|p.(Trp1098Ter)	c.3426_3427insGTAA|c.3426_3427insGTAA|p.(Leu1143ValfsTer14)

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Q1144X|c.3430C>T|p.(Gln1144Ter)	Q1186X|c.3556C>T|p.(Gln1186Ter)	L1243X|c.3728T>A|p.(Leu1243Ter)
W1145X|c.3434G>A|p.(Trp1145Ter)	3695delC|c.3563del|p.(Ser1188Ter)	G1244E|c.3731G>A|p.(Gly1244Glu)
W1145X|c.3435G>A|p.(Trp1145Ter)	c.3569_3570delTT|c.3569_3570del|p.(Val1190AspfsTer4)	T1246I|c.3737C>T|p.(Thr1246Ile)
D1152H|c.3454G>C|p.(Asp1152His)	S1196X|c.3587C>A|p.(Ser1196Ter)	G1247X|c.3739G>T|p.(Gly1247Ter)
V1153E|c.3458T>A|p.(Val1153Glu)	S1196X|c.3587C>G|p.(Ser1196Ter)	S1248X|c.3743C>A|p.(Ser1248Ter)
3600G->A|c.3468G>A|p.(Leu1156=)	3724delG|c.3592del|p.(Val1198Ter)	3876delA|c.3744del|p.(Lys1250ArgfsTer9)
3600+1G->A|c.3468+1G>A|p.?	3732delA|c.3600del|p.(Asp1201MetfsTer10)	S1248X|c.3743C>G|p.(Ser1248Ter)
3600+2insT|c.3468+2dup|p.?	3730A->TCT|c.3598delinsTCT|p.(Lys1200SerfsTer12)	3878delG|c.3747del|p.(Lys1250ArgfsTer9)
3600+1G->T|c.3468+1G>T|p.?	3731-3732AA->G|c.3599_3600delinsG|p.(Lys1200ArgfsTer11)	G1249R|c.3745G>A|p.(Gly1249Arg)
3600+5G->A|c.3468+5G>A|p.?	3737delA|c.3605del|p.(Asp1202AlafsTer9)	G1249R|c.3745G>C|p.(Gly1249Arg)
3601-2A->C|c.3469-2A>C|p.?	W1204X|c.3611G>A|p.(Trp1204Ter)	S1251N|c.3752G>A|p.(Ser1251Asn)
3601-2A->G|c.3469-2A>G|p.?	W1204X|c.3612G>A|p.(Trp1204Ter)	3886insA|c.3754dup|p.(Thr1252AsnfsTer13)
3601-1G->A|c.3469-1G>A|p.?	3747delC|c.3615del|p.(Ser1206GlnfsTer5)	3889dupT|c.3758dup|p.(Leu1253PhefsTer12)
3601-1G->C|c.3469-1G>C|p.?	3750delA|c.3618del|p.(Gly1208AlafsTer3)	3892delTT|c.3760_3761del|p.(Leu1254IlefsTer10)
3601-1G->T|c.3469-1G>T|p.?	3750delAG|c.3618_3619del|p.(Gly1208ProfsTer56)	L1254X|c.3761T>A|p.(Leu1254Ter)
R1158X|c.3472C>T|p.(Arg1158Ter)	S1206X|c.3617C>G|p.(Ser1206Ter)	L1254X|c.3761T>G|p.(Leu1254Ter)
S1159P|c.3475T>C|p.(Ser1159Pro)	3755dupG|c.3623dup|p.(Gln1209ProfsTer56)	S1255P|c.3763T>C|p.(Ser1255Pro)
S1159F|c.3476C>T|p.(Ser1159Phe)	3755delG|c.3623del|p.(Gly1208AlafsTer3)	S1255X|c.3764C>A|p.(Ser1255Ter)
c.3477delT|c.3477del|p.(Val1160Ter)	c.3635delT|c.3635del|p.(Val1212AlafsTer16)	3898insC|c.3767dup|p.(Leu1258PhefsTer7)
3613AGCCGA->GG|c.3481_3486delinsGG|p.(Ser1161GlyfsTer30)	3789insA|c.3658dup|p.(Thr1220AsnfsTer45)	3905insT|c.3773dup|p.(Leu1258PhefsTer7)
3617delGA|c.3486_3487del|p.(Val1163LeufsTer2)	3791delC|c.3659del|p.(Thr1220LysfsTer8)	3905delT|c.3773del|p.(Leu1258Ter)
R1162X|c.3484C>T|p.(Arg1162Ter)	c.3678delA|c.3678del|p.(Leu1227Ter)	L1253X|c.3773T>A|p.(Leu1258Ter)
3622insT|c.3492dup|p.(Lys1165Ter)	3821delT|c.3691del|p.(Ser1231ProfsTer4)	E1266X|c.3796G>T|p.(Glu1266Ter)
K1165X|c.3493A>T|p.(Lys1165Ter)	I1234V|c.3700A>G|p.(Ile1234Val)	I1269N|c.3806T>A|p.(Ile1269Asn)
c.3495delG|c.3495del|p.(Lys1165AsnfsTer27)	3840delT|c.3708del|p.(Gly1237AlafsTer22)	3940delG|c.3808del|p.(Asp1270MetfsTer8)
3629delT|c.3497del|p.(Phe1166SerfsTer26)	3847delA|c.3715del|p.(Arg1239GlyfsTer20)	D1270N|c.3808G>A|p.(Asp1270Asn)
c.3525_3537del|c.3525_3537del|p.(Thr1176AsnfsTer12)	3849G->A|c.3717G>A|p.(Arg1239=)	3944delGT|c.3816_3817del|p.(Ser1273LeufsTer28)
c.3524delC|c.3524del|p.(Pro1175LeufsTer17)	3849+1G->A|c.3717+1G>A|p.?	W1274X|c.3822G>A|p.(Trp1274Ter)
3659delC|c.3528del|p.(Lys1177SerfsTer15)	3849+4A->G|c.3717+4A>G|p.?	3959delC|c.3827del|p.(Ser1276Ter)
3662delA|c.3530del|p.(Lys1177SerfsTer15)	3849+5G->A|c.3717+5G>A|p.?	3960-3961delA|c.3829del|p.(Ile1277Ter)
3667ins4|c.3532_3535dup|p.(Thr1179IlefsTer17)	3849+40A->G|c.3717+40A>G|p.?	3967delTT|c.3835_3836del|p.(Leu1279AlafsTer22)
3667del4|c.3536_3539del|p.(Thr1179AsnfsTer12)	3849+10kbC->T|c.3718-2477C>T|p.?	Q1280X|c.3838C>T|p.(Gln1280Ter)
S1178X|c.3533C>A|p.(Ser1178Ter)	3850-3T->G|c.3718-3T>G|p.?	Q1281X|c.3841C>T|p.(Gln1281Ter)
S1178X|c.3533C>G|p.(Ser1178Ter)	3850-1G->A|c.3718-1G>A|p.?	W1282X|c.3845G>A|p.(Trp1282Ter)
c.3539_3554del|c.3539_3554del|p.(Lys1180ThrfsTer7)	V1240G|c.3719T>G|p.(Val1240Gly)	W1282X|c.3846G>A|p.(Trp1282Ter)
Y1182X|c.3546C>G|p.(Tyr1182Ter)	3856delC|c.3724del|p.(Leu1242SerfsTer17)	R1283M|c.3848G>T|p.(Arg1283Met)

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G1287X|c.3859G>T|p.(Gly1287Ter)	4096-1G->A|c.3964-1G>A|p.?	c.4163_4167delTAAAA|c.4163_4167del|p.(Leu1388ProfsTer5)
Q1291X|c.3871C>T|p.(Gln1291Ter)	L1324P|c.3971T>C|p.(Leu1324Pro)	4301delA|c.4170del|p.(Ala1391HisfsTer7)
Q1291R|c.3872A>G|p.(Gln1291Arg)	4108delT|c.3976del|p.(Ser1326LeufsTer2)	Q1390X|c.4168C>T|p.(Gln1390Ter)
Q1291H|c.3873G>C|p.(Gln1291His)	4120delCA|c.3988_3989del|p.(Gln1330ValfsTer6)	c.4187delC|c.4187del|p.(Thr1396LysfsTer2)
4005+1G->A|c.3873+1G>A|p.?	Q1330X|c.3988C>T|p.(Gln1330Ter)	4326delTC|c.4197_4198del|p.(Cys1400Ter)
4005+1G->T|c.3873+1G>T|p.?	c.3999delG|c.3999del|p.(Lys1334SerfsTer13)	4332delTG|c.4200_4201del|p.(Cys1400Ter)
4005+2T->C|c.3873+2T>C|p.?	L1335P|c.4004T>C|p.(Leu1335Pro)	C1400X|c.4200T>A|p.(Cys1400Ter)
4006-2A->G|c.3874-2A>G|p.?	c.4028delG|c.4028del|p.(Gly1343AlafsTer4)	E1401X|c.4201G>T|p.(Glu1401Ter)
4006-1G->A|c.3874-1G>A|p.?	4160insG|c.4028dup|p.(Cys1344LeufsTer15)	E1409X|c.4225G>T|p.(Glu1409Ter)
4006delA|c.3876del|p.(Val1293TyrfsTer35)	4165delGT|c.4033_4034del|p.(Val1345ProfsTer13)	C1410X|c.4230C>A|p.(Cys1410Ter)
4010del4|c.3883_3886del|p.(Ile1295PhefsTer32)	4168delCTAAGCC|c.4036_4042del|p.(Leu1346MetfsTer6)	Q1411X|c.4231C>T|p.(Gln1411Ter)
4015delA|c.3883del|p.(Ile1295PhefsTer33)	c.4036dupC|c.4036dup|p.(Leu1346ProfsTer13)	Q1412X|c.4234C>T|p.(Gln1412Ter)
c.3883_3884insG|c.3883_3884insG|p.(Ile1295SerfsTer7)	p.S1347PfsX13|c.4035_4038dup|p.(Ser1347ProfsTer13)	4374+1G->A|c.4242+1G>A|p.?
4016insT|c.3889dup|p.(Ser1297PhefsTer5)	4168delCTAAG|c.4037_4041del|p.(Leu1346ProfsTer11)	4374+1G->T|c.4242+1G>T|p.?
c.3889delT|c.3889del|p.(Ser1297LeufsTer31)	4172delGC|c.4040_4041del|p.(Ser1347ThrfsTer11)	4382delA|c.4251del|p.(Glu1418ArgfsTer14)
4022insT|c.3891dup|p.(Gly1298TrpfsTer4)	4177delG|c.4046del|p.(Gly1349AlafsTer5)	4428insGA|c.4300_4301dup|p.(Ser1435GlyfsTer14)
c.3893delG|c.3893del|p.(Gly1298GlufsTer30)	G1349D|c.4046G>A|p.(Gly1349Asp)	S1455X|c.4364C>G|p.(Ser1455Ter)
4040delA|c.3908del|p.(Asn1303ThrfsTer25)	K1351X|c.4051A>T|p.(Lys1351Ter)	Q1476X|c.4426C>T|p.(Gln1476Ter)
4040dupA|c.3908dup|p.(Asn1303LysfsTer6)	4197_4198delCT|c.4065_4066del|p.(Leu1356GlyfsTer2)	L1480P|c.4439T>C|p.(Leu1480Pro)
K1302X|c.3904A>T|p.(Lys1302Ter)	4203TAG->AA|c.4071_4073delinsAA|p.(Arg1358AsnfsTer22)	
N1303K|c.3909C>G|p.(Asn1303Lys)	4209TGTT->AA|c.4077_4080delinsAA|p.(Val1360ThrfsTer3)	
4048insCC|c.3917_3918dup|p.(Tyr1307ProfsTer22)	c.4078delG|c.4078del|p.(Val1360PhefsTer20)	
Y1307X|c.3921T>A|p.(Tyr1307Ter)	4218insT|c.4086dup|p.(Lys1363Ter)	
E1308X|c.3922G>T|p.(Glu1308Ter)	4222delG|c.4090del|p.(Ala1364ArgfsTer16)	
Q1309X|c.3925C>T|p.(Gln1309Ter)	I1366N|c.4097T>A|p.(Ile1366Asn)	
W1310X|c.3929G>A|p.(Trp1310Ter)	4237-4242delinsAGAA|c.4105_4110delinsAGAA|p.(Leu1369ArgfsTer16)	
W1310X|c.3930G>A|p.(Trp1310Ter)	E1371X|c.4111G>T|p.(Glu1371Ter)	
4064-4065delinsAATATG|c.3932_3933delinsAATATG|p.(Ser1311LysfsTer12)	H1375P|c.4124A>C|p.(His1375Pro)	
Q1313X|c.3937C>T|p.(Gln1313Ter)	4259del5|c.4127_4131del|p.(Leu1376SerfsTer8)	
W1316X|c.3947G>A|p.(Trp1316Ter)	4268+2T->G|c.4136+2T>G|p.?	
4089delA|c.3957del|p.(Asp1320MetfsTer8)	4271delC|c.4139del|p.(Thr1380AsnfsTer4)	
4089ins4|c.3957_3958insAGGG|p.(Asp1320ArgfsTer3)	Y1381X|c.4143C>A|p.(Tyr1381Ter)	
4095+1G->A|c.3963+1G>A|p.?	Y1381X|c.4143C>G|p.(Tyr1381Ter)	
4095+1G->C|c.3963+1G>C|p.?	4279insA|c.4147dup|p.(Ile1383AsnfsTer3)	
4095+1G->T|c.3963+1G>T|p.?	Q1382X|c.4144C>T|p.(Gln1382Ter)	