

Mutation	Type	Exon	Coding Effect	Clinical/Predicted
c.1A>G	Missense	3	p.M1V	Clinical
c.3G>A	Missense	3	p.M1I	Clinical
c.60delTinsAA	indel	3		Clinical
c.89G>C	Missense	3	p.G30A	Clinical
c.91C>T	Missense	3	p.R31C	Clinical
c.92G>A	Missense	3	p.R31H	Predicted
c.98_194del	Deletion	3	p.W33fsX4	Clinical
c.98+2_+6delTAAGG	Deletion	3		Clinical
c.111G>A	Missense	4	p.W37X	Clinical
c.149C>A	Missense	4	p.A50D	Clinical
c.149C>T ^b	Missense	4	p.A50V	Clinical
c.151C>T	Missense	4	p.P51S	Clinical
c.152C>A	Missense	4	p.P51H	Clinical
c.165C>G	Missense	4	p.Y55X	Predicted
c.176G>T	Missense	4	p.M59R	Clinical
c.185A>T	Missense	4	p.D62V	Clinical
c.203T>C	Missense	4	p.L68P	Clinical
c.208G>T ^b	Missense	4	p.G70C	Clinical
c.232G>A	Missense	4	p.G78R	Predicted
c.265A>T	Missense	4	p.T89S	Predicted
c.278C>T	Missense	4	p.T93M	Clinical
c.292C>T	Missense	4	p.Q98X	Clinical
c.296T>C	Missense	4	p.L99P	Clinical
c.321G>C	Missense	4	p.Q107H	Clinical
c.326T>C	Missense	5	p.L109P	Clinical
c.338T>C	Missense	5	p.S113C	Clinical
c.355delC	Deletion	5	p.H119ifsX8	Clinical
c.356A>T	Missense	5	p.H119L	Clinical
c.356_368del13	Deletion	5	p.H119PfsX23	Clinical
c.374A>G	Missense	5	p.Y125C	Clinical
c.376G>A	Missense	5	p.V126I	Predicted
c.385_412+5del	Deletion	5		Clinical
c.400G>T	Missense	5	p.V134L	Clinical
c.413G>T	Missense	6	p.G138V	Clinical
c.433A>C	Missense	6	p.I145L	Clinical
c.438C>G	Missense	6	p.N146K	Clinical
c.440G>A	Missense	6	p.G147D	Clinical
c.443T>G	Missense	6	p.L148R	Clinical
c.445C>T	Missense	6	p.Q149X	Clinical
c.452G>A	Missense	6	p.W151X	Clinical
c.453G>A	Missense	6	p.W151X	Clinical
c.461C>T	Missense	6	p.T154M	Clinical
c.461C>G	Missense	6	p.T154R	Clinical
c.470T>C	Missense	6	p.L157P	Clinical
c.474G>T	Missense	6	p.W158C	Clinical
c.502T>A	Missense	6	p.F168I	Clinical
c.506C>T	Missense	6	p.S169L	Clinical
c.509C>G	Missense	6	p.P170R	Predicted
c.521T>C	Missense	6	p.F174S	Clinical
c.523G>A	Missense	6	p.D175N	Predicted
c.523G>C	Missense	6	p.D175H	Clinical
c.529T>C	Missense	6	p.W177R	Clinical
c.532A>T	Missense	6	p.I178F	Clinical
c.533T>A	Missense	6	p.I178N	Clinical
c.536C>T	Missense	6	p.P179L	Clinical
c.545G>T	Missense	6	p.W182L	Clinical
c.546G>C	Missense	6	p.W182C	Clinical
c.548G>A	Missense	6	p.C183Y	Clinical
c.575C>T	Missense	6	p.S192F	Clinical
c.577A>C	Missense	6	p.T193P	Clinical
c.592A>G	Missense	6	p.K198E	Clinical

Mutation	Type	Exon	Coding Effect	Clinical/Predicted
c.628A>T	Missense	7	p.K210X	Clinical
c.651C>A	Missense	7	p.Y217X	Clinical
c.655T>G	Missense	7	p.Y219D	Clinical
c.658A>C	Missense	7	p.M220L	Predicted
c.670G>A	Missense	7	p.E224K	Clinical
c.679C>T	Missense	7	p.P227S	Clinical
c.679C>A	Missense	7	p.P227T	Clinical
c.680dupC	Duplication	7		Clinical
c.682C>T	Missense	7	p.R228W	Clinical
c.682insC	Insertion	7	Frameshift	Clinical
c.683G>A	Missense	7	p.R228Q	Predicted
c.700G>T	Missense	7	p.D234Y	Clinical
c.704T>C	Missense	7	p.F235S	Clinical
c.719A>G	Missense	7	p.N240S	Predicted
c.720_735del	Deletion	7	Frameshift	Clinical
c.724C>T	Missense	7	p.R242C	Clinical
c.725G>A	Missense	7	p.R242H	Clinical
c.728C>G	Missense	7	p.P243R	Clinical
c.730G>A	Missense	7	p.G244R	Clinical
c.740C>T	Missense	7	p.A247V	Clinical
c.742T>C	Missense	7	p.W248R	Clinical
c.744G>T	Missense	7	p.W248C	Clinical
c.744G>C	Missense	7	p.W248C	Clinical
c.752T>A	Missense	7	p.I251N	Clinical
c.753C>G	Missense	7	p.I251M	Clinical
c.755A>G	Missense	7	p.N252S	Clinical
c.760T>G	Missense	7	p.S254A	Clinical
c.762insT	Insertion	7	Frameshift	Clinical
c.765C>A	Missense	7	p.F255L	Clinical
c.808A>G	Missense	7	p.M270V	Clinical
c.818T>G	Missense	7	p.V273G	Clinical
c.822C>A	Missense	7	p.N274K	Clinical
c.841G>A	Missense	7	p.V281M	Clinical
c.839A>G	Missense	8	p.Y280C	Clinical
c.849_856insCTTCT	Insertion	8		Clinical
c.852C>A	Missense	8	p.F284L	Clinical
c.854_856delTCT	Deletion	8		Clinical
c.861C>A	Missense	8	p.N287K	Clinical
c.862G>A	Missense	8	p.E288K	Clinical
c.866C>T	Missense	8	p.T289I	Clinical
c.883A>G	Missense	8	p.I295V	Predicted
c.890T>C	Missense	8	p.I297T	Clinical
c.896A>G	Missense	8	p.H299R	Clinical
c.902A>G	Missense	8	p.H301R	Clinical
c.906C>G	Missense	8	p.F302L	Clinical
c.907G>A	Missense	8	p.G303R	Clinical
c.920G>A	Missense	8	p.G307D	Clinical
c.925G>A	Missense	8	p.G309S	Clinical
c.928G>A	Missense	8	p.D310N	Predicted
c.931T>G	Missense	8	p.C311G	Clinical
c.932G>A	Missense	8	p.C311Y	Clinical
c.950T>G	Missense	8	p.L317R	Clinical
c.952T>A	Missense	8	p.Y318N	Clinical
c.956C>T	Missense	8	p.T319M	Clinical
c.957G>A	Missense	8	p.T319A	Clinical
c.964-1G>T	Intronic	8		Clinical
c.964-1G>C	Intronic	8	p.G322KfsX136	Clinical
c.964G>C	Missense	8	p.G322R	Clinical

Mutation	Type	Exon	Coding Effect	Clinical/Predicted
c.976G>T	Missense	9	p.V326L	Clinical
c.986C>T	Missense	9	p.P329L	Clinical
c.988G>A	Missense	9	p.V330M	Clinical
c.1022T>C	Missense	9	p.L341P	Clinical
c.1025T>C	Missense	9	p.L342P	Predicted
c.1030G>C	Missense	9	p.G344R	Clinical
c.1031G>A	Missense	9	p.G344D	Clinical
c.1039G>A	Missense	9	p.G347S	Clinical
c.1054C>T	Missense	9	p.R352W	Clinical
c.1055G>A	Missense	9	p.R352Q	Clinical
c.1055G>T	Missense	9	p.R352L	Clinical
c.1057Gdel	Deletion	9	Frameshift	Clinical
c1068_1070del	Deletion	9	p.H356del	Clinical
c.1058T>C	Missense	9	p.V353A	Clinical
c.1063A>G	Missense	9	p.N355D	Clinical
c.1079T>C	Missense	9	p.L360P	Clinical
c.1083C>G	Missense	9	p.F361L	Clinical
c.1084C>T	Missense	9	p.R362C	Predicted
c.1085G>A	Missense	9	p.R362H	Predicted
c.1085G>A	Missense	9	p.R363H	Clinical
c.1087C>T	Missense	9	p.R363C	Clinical
c.1097G>T	Missense	9	p.G366V	Clinical
c.1088G>A	Missense	9	p.R363H	Clinical
c.1127_1128delA	Deletion	9	p.K376RfsX37	Clinical
c.1138T>A	Missense	9	p.C380S	Clinical
c.1138T>C	Missense	9	p.C380R	Clinical
c.1139G>C	Missense	9	p.C380S	Clinical
c.1139G>A	Missense	9	p.C380Y	Clinical
c.1156G>A	Missense	9	p.D386N	Predicted
c.1187T>A	Missense	9	p.V396E	Clinical
c.1190C>T	Missense	9	p.S397L	Clinical
c.1210C>T	Missense	9	p.R404C	Clinical
c.1210C>A	Missense	9	p.R404S	Clinical
c.1213C>T	Missense	9	p.H405Y	Clinical
c.1219A>T	Missense	9	p.N407Y	Clinical
c.1222T>C	Missense	9	p.Y408H	Clinical
c.1228G>C	Missense	9	p.G410R	Clinical
c.1228G>A	Missense	9	p.G410S	Clinical
c.1231G>A	Missense	9	p.D411N	Predicted
c.1270G>T	Missense	9	p.G424C	Predicted
c.1277A>C	Missense	9	p.H426P	Clinical
c.1289A>G	Missense	9	p.Y432C	Clinical
c.1290C>G	Missense	9	p.Y430X	Predicted
c.1295A>G	Missense	9	p.Y432C	Clinical
c.1327C>T	Missense	9	p.R443C	Clinical
c.1328G>A	Missense	9	p.R443H	Clinical
c.1331G>A	Missense	9	p.C444Y	Clinical
c.1336C>T	Missense	9	p.R446W	Clinical
c.1337G>A	Missense	9	p.R446Q	Clinical
c.1342G>C	Missense	9	p.E448Q	Clinical
c.1342G>A	Missense	9	p.E448K	Clinical
c.1349G>T	Missense	9	p.R450L	Clinical
c.1351T>C	Missense	9	p.C451R	Clinical
c.1366G>A	Missense	9	p.G456S	Predicted
c.1369C>T	Missense	9	p.R457W	Predicted
c.1381C>T	Missense	9	p.R461C	Predicted
c.1384T>C	Missense	9	p.Y462H	Clinical
c.1396G>A	Missense	9	p.V466M	Clinical
c.1397T>C	Missense	9	p.V466A	Clinical
c.1400C>T	Missense	9	p.P467L	Clinical
c.1405C>T	Missense	9	p.R469C	Predicted
c.1406G>C	Missense	9	p.R469P	Clinical
c.1406G>A	Missense	9	p.R469H	Predicted
c.1409T>A	Missense	9	p.L470Q	Clinical
c.1423T>C	Missense	9	p.F475S	Clinical
c.1426T>A	Missense	9	p.X476Q	Clinical