

Mutations	Changed Amino acid	Exon/ Intron	Number	Novel mutations
c.76C>T	p.R26C	Exon 3	2	N
c.77G>A	p.R26H	Exon 3	2	N
c.92C>T	p.A31V	Exon 4	1	N
c.181G>A	p.D61N	Exon 5	1	N
c.181G>T	p.D61Y	exon 5	1	N
c.229T>G	p.F77V	exon 6	1	Y
c.293A>C	p.K98T	Exon 7	1	Y
c.331G>A	p.G111R	exon 7	2	N
c.334G>C	p.A112P	exon 7	1	N
c.364G>C	p.R116W	exon 8	1	N
c.386T>A	p.V123D	Exon 8	1	Y
c.517C>T	p.R173W	Exon 10	7	N
c.518G>A	P.R173E	exon 10	2	N
c.653G>A	P.G218E	exon 12	1	Y
c.662G>A	p. G221D	exon 12	1	N
c.739T>C	p.C247R	exon 12	1	N
c.771G>A	del205-257 amino acids	exon 12	1	N
c.806C>G	p.T269R	exon 12	2	N
c.962G>A	p.R321H	exon 15	1	N
c.445C>T	p.R149X	exon 9	2	N
c.469A>T	p.K157X	exon 9	2	Y
c.673C>T	p.R225X	exon 12	8	N
c.973C>T	p.R325X	exon 15	3	N
c.261delC	p.V87Vfs45	exon 6	1	Y
c.582-586del CGCA	p.Q195fs*10	exon 10	1	Y
c.597dupC	p.H200fs8	exon 10	1	Y
c.673_674del	p.R225Dfs*7	exon 12	1	Y
c.726_727del	p.E242Dfs*8	exon 12	1	Y
c.728_729del	p.L244Afs*5	exon 12	1	N
c.730-731delCT	p.L244fs*6	exon 12	1	N
c.734-741delTTCGCTGC	p.L245Hfs*3	exon 12	1	Y
c.847_848delT	p.W283Efs*7	exon 14	1	N
c.1045_1046delAA	p.N349Hfs*9	exon 15	1	Y
c.760-771+2delCTGAGGCACCTG		exon 12	1	Y
TinsGCTGCATCGCTGAA				
c.33+5G>C		intron 1	1	N
c.88-1 G>C		Intron 4	1	Y
c.160+3G>T		intron 4	1	y
c.267-1G>C		intron 6	3	N

c.372-2A>G	intron 7	1	N
c.423-2A>G	intron 7	1	N
c.771+1G>C	Intron 1	1	N
	2		
c.772-17A>G	Intron 1	1	N
	2		
c.913-9_914del	Intron 1	1	Y
	4		
c.913-2A>G	Intron 1	1	N
	4		
c.912+1G>C	Intron 1	1	Y
	4		

Mutation	Change of Amino acid	Ex on	Possible pathogenic mechanism	reference
c.76C>T	p.R26C	3	M	[10]
c.77G>A	p.R26H	3	M	[10]
c.92C>T	p.A31 V	4	M	Located in the center of enzyme activity, affects p
c.517C>T	p.R173 W	10	M	yrrole ring elongation [11-14]
c.518G>A	p.R173E	10	M	[11-14]
c.181G>T	p.D61Y	5	M	[3,15]
			Diminishes enzyme stability	
c.364G>C	p.R116 W	8	M	[16-17]
c.739T>C	p.C247R	12	M	[18]
c.662G>A	p. G221 D	12	M	Limits the movement of activity center and affect pyrrole ring elongation [19]
c.331G>A	p.G111R	7	M	Forms the β -folds of domain 1 and affects hinge motion [2,15]
c.334G>C	p.A112P	7	M	Affects PBGD folding [20]
			The hydrophobic center has difficulty accommoda	[21,22]

c.806C>G	p.T269R	12	M	ting the large side chain after mutation, and stability is reduced	
c.962G>A	p.R321H	15	M	Reduces the movement between domains	[1]
c.973C>T	p.R325X	15	M		[23]
c.771G>A	del205-257 th amino acids	12	M		[24]
Truncated inactivate protein					
c.445C>T	p.R149X	9	N		
c.469A>T	p.K157X	9	N		
c.673C>T	p.R225X	12	N		
c.261delC	p.V87 Vfs45	6	F		
c.728_729del	p.L244Afs*5	12	F		
c.730-731delCT	p.L244fs*6	12	F		[25]
c.847_848delT	p.W283Efs*7	14	F		[11,22,26]