

Families	cDNA (Nucleotide)	Variant (Amino Acid)	Number of Patients Affected	Location
TESS_001	c.655G>A c.1475T>C	p.Gly219Arg p.Leu492Pro	1 boy and 1 girl	USA
TESS_002	c.245A>G c.655G>A	p.Tyr82Cys p.Gly219Arg	1 boy and 1 girl	USA
TESS_003	c.368+1G>A	IVS3+1 G>A (Canonical splice site donor destroyed for intron 3.)	2 girls	USA
TESS_004	c.511delG	p.E171SfsX16	1 boy and 1 girl	USA
TESS_005	c.997C>T c.680C>T	p.Arg333X p.Thr227Met	1 boy	USA
TESS_006	c.389G>A partial gene deletion exon 2-4	p.Gly130 Asp Truncated or no protein	1 girl	USA
TESS_007	Deletion entire sequence c.425C>T	No protein p.Thr142Met	1 girl	USA
TESS_008	c.655G>A	p.Gly219Arg	1 boy	Brazil
TESS_009	c.103-1G>A c.1276-1G>A	splicing mutation (intronic CCDS11079.1) splicing mutation	1 girl and 1 boy	Brazil
TESS_010	c.148T>C	p.Cys50Arg	1 girl and 2 boys (1 boy deceased)	Lebanon
TESS_011	c.680 C > T	p.Thr227Met	1 girl	Netherlands
TESS_012	c.655G>A	p.Gly219Arg	1 girl	Iceland
TESS_014	c.680C>T c.655G>A	p.Thr227Met p.Gly219Arg	1 girl and 1 boy	Unknown
TESS_015	c.1463T>C	p.Leu488Pro	3 (1 girl + 2 boys) 2 boys (1 deceased)	France
TESS_016	c.680C>T c.655G>A	p.Thr227Met p.Gly219Arg	1 (boy)	France
TESS_017	c.1280C>T	p.Ser427Leu	2 boys (1 deceased)	Unknown
TESS_018	c.1022G>A c.1207_1217dup11	p.Trp341* p.Pro407Arfs*12	1 girl and 1 boy	Unknown
TESS_019	c.425C>T c.655G>A	p.Thr142Met p.Gly219Arg	2 girls and 1 boy	Germany
TESS_020	c.680C>T c.1570G>C	p.Thr227Met p.Asp524His	1 girl	Unknown
TESS_021	c.655G>A	p.Gly219Arg	1 girl	Iceland
TESS_022	c.1022G>A c.655G>A	p.Trp341* p.Gly219Arg	1 girl	France
TESS_024	c.655G>A c.1280C>T	p.(Gly219Arg) p.(Ser427Leu)	1 girl and 1 boy deceased	Unknown
TESS_025	c.680C>T c.1280C>T	p.(Thr227Met) p.(Ser427Leu)	1 boy deceased	Unknown
TESS_026	c.655G>A	p.(Gly219Arg)	2 girls	Unknown
TESS_027	c.997C>T	p.Arg333Ter	1 girl and 1 boy	Unknown
TESS_028	c.203C>A c.434C>A	p.Pro68Gln p.Thr145Lys	2 girls	Unknown
TESS_029	c.655G>A c.317A>G	p.Gly219Arg p.His106Arg	2 boys	Unknown
TESS_030	c.1268G>A	p.Gly423Glu	1 girl	Unknown
TESS_031	c.1451 del G c.332T>G	p.G484A fs*13 p.Leu111Arg	1 boy	UK
TESS_032	c.655G>A c.1250G>A	p.Gly219Arg p.Gly417Glu	1 boy	Poland
TESS_033	c.655G>A Deletion exon 1-5	p.Gly219Arg Truncated protein or no protein	1 boy	Italy
TESS_040	c.997C>T c.478G>T	p.(Arg333*) p.(Glu160*)	1 boy	Spain

TESS_043	c.997C>T c.1514C>T	(p.Arg333*) (p.Pro505Leu)	1 boy	USA
TESS_045	c.655G>A c.1190delT	p.Gly219Arg p.Leu397ArqfsX5	1 girl	Canada
TESS_046	c.997C>T	(p.Arg333*)	1 boy	Italy
TESS_052	c.1098dupC	p.Ile367fs	2 patients	Unknown
TESS_053	c.1516T>A	p.Phe506Ile	1 patient	Unknown
TESS_054	c.3G>A (initiator codon)	p.Met1?	1 boy	Unknown
TESS_055	c.1460C>T c.232-2A>G	p.Pro487Leu	1 boy	USA
TESS_058	c.1511delT	p.L504CfsX23	1 boy	USA
TESS_059	Partial gene deletion at least exon 1-3	Partial gene deletion at least exon 1- 3	1 girl, 1 boy	USA
TESS_061	c.997 C>T	p.Arg333Ter	1 boy	USA
TESS_062	c.1514C>T	p.Pro505Leu	1 boy	Brazil
TESS_063	c.1412T>C c.997C>T	p.Leu471Ser p.Arg333Ter	1 boy	Italy
TESS_065	c.655G>A	(p.Gly219Arg)	1 boy	United States
TESS_067	c.644C>T	(p.Ala215Val)	1 girl	India
TESS_068	c.650G>A c.1599C>A	(p.Ser217Asn) (p.Asn533Lys)	1 girl	United States
TESS_013, 023, 034, 035, 036, 037, 038, 039, 041, 042, 044, 045, 046, 047, 048, 049, 050, 051, 056, 057, 060 = no data or het carriers				

cDNA (Nucleotide)	Variant (Amino Acid)	Number of Patients with this Mutation (Include Deceased)
c.655G>A	p.Gly219Arg	23
c.680C>T	p.Thr227Met	7
c.997C>T	p.Arg333Ter	8
c.1463T>C	p.Leu488Pro	5
c.1280C>T	p.Ser427Leu	5
c.425C>T	p.Thr142Met	4
c.1022G>A	p.Trp341*	3
c.148T>C	p.Cys50Arg	3
c.103-1G>A	splicing mutation (intronic CCDS11079.1)	2
c.203C>A	p.Pro68Gln	2
c.245A>G	p.Tyr82Cys	2
c.317A>G	p.His106Arg	2
c.368+1G>A	IVS3+1 G>A (Canonical splice site donor destroyed for intron 3.)	2
c.434C>A	p.Thr145Lys	2
c.511delG	p.E171SfsX16	2
c.650G>A	(p.Ser217Asn)	1
c.1098dupC	p.Ile367fs	2
c.1207_1217dup11	p.Pro407Argfs*12	2
c.1276-1G>A	splicing mutation	2
c.1475T>C	p.Leu492Pro	2
Partial gene deletion at least exon 1-3	Partial gene deletion at least exon 1-3	2
c.1514C>T	(p.Pro505Leu)	2
c.3G>A (initiator codon)	p.Met1?	1
c.232-2A>G		1
c.332T>G	p.Leu111Arg	1
c.644C>T	(p.Ala215Val)	1
c.389G>A	p.Gly130 Asp	1
c.478G>T	p.(Glu160*)	1
c.1190delT	p.Leu397ArgfsX5	1
c.1250G>A	p.Gly417Glu	1
c.1268G>A	p.Gly423Glu	1
c.1451 del G	p.G484A fs*13	1
c.1460C>T	p.Pro487Leu	1
c.1516T>A	p.Phe506Ile	1
c.1570G>C	p.Asp524His	1
c.1599C>A	(p.Asn533Lys)	1
Deletion entire sequence	No protein	1
Deletion exon 1-5	Truncated protein or no protein	1
Partial gene deletion exon 2-4	Truncated or no protein	1
c.1412T>C	p.Leu471Ser	1
c.1511delT	p.L504CfsX23	1
41 different known mutations		