Families	cDNA (Nucleotide)	Variant (Amino Acid)	Number of Patients Affected	Location
TESS_001	c.655G>A	p.Gly219Arg	1 boy and 1 girl	USA
	c.1475T>C	p.Leu492Pro		
TESS_002	c.245A>G	p.Tyr82Cys	1 boy and 1 girl	USA
	c.655G>A	p.Gly219Arg		
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	p.Arg333X	1 boy	USA
	c.680C>T	p.Thr227Met		
TESS_006	c.389G>A	p.Gly130 Asp	1 girl	USA
	partial gene deletion exon 2-4	Truncated or no protein		
TESS_007	Deletion entire sequence	No protein	1 girl	USA
	c.425C>T	p.Thr142Met		
TESS_008	c.655G>A	p.Gly219Arg	1 boy	Brazil
TESS_009	c.103-1G>A	splicing mutation (intronic CCDS11079.1)	1 girl and 1 boy	Brazil
	c.1276-1G>A	splicing mutation		
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	p.Thr227Met	1 girl and 1 boy	Unknown
	c.655G>A	p.Gly219Arg		
TESS_015	c.1463T>C	p.Leu488Pro	3 (1 girl + 2 boys)	France
			2 boys (1 deceased)	
TESS_016	c.680C>T	p.Thr227Met	1 (boy)	France
	c.655G>A	p.Gly219Arg		
TESS_017	c.1280C>T	p.Ser427Leu	2 boys (1 deceased)	Unknown
TESS_018	c.1022G>A	p.Trp341*	1 girl and 1 boy	Unknown
	c.1207_1217dup11	p.Pro407Argfs*12		
TESS_019	c.425C>T	p.Thr142Met	2 girls and 1 boy	Germany
	c.655G>A	p.Gly219Arg		
TESS_020	c.680C>T	p.Thr227Met	1 girl	Unknown
	c.1570G>C	p.Asp524His		
TESS_021	c.655G>A	p.Gly219Arg	1 girl	Iceland
TESS_022	c.1022G>A	p.Trp341*	1 girl	France
	c.655G>A	p.Gly219Arg		
TESS_024	c.655G>A	p.(Gly219Arg)	1 girl and 1 boy deceased	Unknown
	c.1280C>T	p.(Ser427Leu)		

TESS_025	c.680C>T	p.(Thr227Met)	1 boy deceased	Unknown
	c.1280C>T	p.(Ser427Leu)		
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	p.Pro68Gln	2 girls	Unknown
	c.434C>A	p.Thr145Lys		
TESS_029	c.655G>A	p.Gly219Arg	2 boys	Unknown
	c.317A>G	p.His106Arg		
TESS_030	c.1268G>A	p.Gly423Glu	1 girl	Unknown
TESS_031	c.1451 del G	p.G484A fs*13	1 boy	UK
	c.332T>G	p.Leu111Arg		
TESS_032	c.655G>A	p.Gly219Arg	1 boy	Poland
	c.1250G>A	p.Gly417Glu		
TESS_033	c.655G>A	p.Gly219Arg	1 boy	Italy
	Deletion exon 1-5	Truncated protein or no protein		
TESS_040	c.997C>T	p.(Arg333*)	1 boy	Spain
	c.478G>T	p.(Glu160*)		
TESS_043	c.997C>T	(p.Arg333*)	1 boy	USA
	c.1514C>T	(p.Pro505Leu)		
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	p.Pro487Leu	1 boy	USA
	c.232-2A>G			
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_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				

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Last modified: 3/6/2020

cDNA (Nucleotide)	Variant (Amino Acid)	Number of Patients with this Mutation (Include Deceased)
c.655G>A	p.Gly219Arg	22
c.680C>T	p.Thr227Met	7
c.997C>T	p.Arg333Ter	6
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.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.3G>A (initiator codon)	p.Met1?	1
c.232-2A>G		1
c.332T>G	p.Leu111Arg	1
c.389G>A	p.Gly130 Asp	1
c.478G>T	p.(Glu160*)	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.1514C>T	(p.Pro505Leu)	1
c.1516T>A	p.Phe506Ile	1
c.1570G>C	p.Asp524His	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.1511delT	p.L504CfsX23	1
36 different known mutations		