

Families	cDNA (Nucleotide)	Variant (Amino Acid)	Zygoty	Number of Patients Affected	Reference
Family 1	<b>c.511delG</b>	<b>p.E171SfsX16</b>	homozygous	1 boy and 1 girl	Published Klotz et al., 2016
Family 2	<b>c.245 A&gt;G</b> <b>c.655 G&gt;A</b>	<b>Y82C</b> <b>G219R</b>	heterozygous heterozygous	1 boy and 1 girl	Published Klotz et al 2016
Family 3	<b>c368+1G&gt;A</b>	<b>IVS3+1 G&gt;A (Cannonical splice site donor destroyed for intron 3.)</b>	homozygous	2 girls	Not published
Family 4	<b>c.655G&gt;A</b> <b>c.1475T&gt;C</b>	<b>p.G219R</b> <b>p.L492P</b>	heterozygous	1 boy and 1 girl	Published Klotz et al., 2016
Family 5	<b>c. 997C&gt;T</b> <b>c. 680C&gt;T</b>	<b>p.R333X</b> <b>p.T227M</b>	heterozygous	1 boy	Published Bainbridge et al, 2017
Family 6	<b>c.655G&gt;A</b>	<b>p. Gly219Arg</b>	homozygous	1 boy	Published Klotz et al., 2016
Family 7	<b>c. 103-1G&gt;A</b> <b>c. 1276-1G&gt;A</b>	<b>splicing mutation (intronic CCDS11079.1)</b> <b>splicing mutation</b>	heterozygous	1 girl and 1 boy	Published Klotz et al., 2016
Family 8	<b>c.148T&gt;C</b>	<b>p.Cys50Arg</b>	homozygous	1 girl and 2 boys (1 boy deceased)	Not Published
Family 9	<b>c.680 C &gt; T</b>	<b>p. T227M</b>	homozygous	1 girl	Published Klotz et al., 2016
Family 10	<b>c.655G&gt;A</b>	<b>p.Gly219Arg</b>	homozygous	1 girl	Not Published
Family 12	<b>c.680C&gt;T</b> <b>c.655G&gt;A</b>	<b>p.Thr227Met</b> <b>p.Gly219Arg</b>	compound heterozygous	1 girl and 1 boy	Published Thevenon et al., 2014
Family 13	<b>c.1463T&gt;C</b>	<b>p.Leu488Pro</b>	homozygous	3 (1 girl + 2 boys)	Published Thevenon et al., 2014

				2 boys (1 deceased)	
Family 14	c.680C>T c.655G>A	p.Thr227Met p.Gly219Arg	compound heterozygous	1 (boy)	Published Thevenon et al., 2014
Family 15	<b>c.1280C&gt;T</b>	<b>p.Ser427Leu</b>	homozygous	2 boys (1 half sibling deceased)	Published Hardies et al., 2015
Family 16	<b>c.1022G&gt;A</b> <b>c.1207_1217dup11</b>	<b>p.Trp341*</b> <b>p.Pro407Argfs*12</b>	heterozygous	1 girl and 1 boy	Published Hardies et al., 2015
Family 17	<b>c.425C&gt;T</b> c.655G>A	<b>p.Thr142Met</b> p.Gly219Arg	heterozygous	2 girls and 1 boy	Published Hardies et al., 2015
Family 18	c.680C>T <b>c.1570G&gt;C</b>	p.Thr227Met <b>p.Asp524His</b>	heterozygous	1 girl	Published Hardies et al., 2015
Family 19	<b>c.389G&gt;A</b> <b>partial gene deletion exon 2 -4</b>	<b>p.Gly130 Asp</b>	heterozygous	1 girl	Not Published
Family 20	c.655G>A	p.Gly219Arg	homozygous	1 girl	Not Published
Family 21	c.1022G>A c.655G>A	p.Trp341* p.Gly219Arg	heterozygous	1 girl	Not Published
Family 23	c.655G>A c.1280C>T	p.(Gly219Arg) p.(Ser427Leu)	compound heterozygous	1 girl and 1 boy deceased	Published Weeke at al., 2016
Family 24	c.680C>T c.1280C>T	p.(Thr227Met) p.(Ser427Leu)	compound heterozygous	1 boy deceased	Published Weeke at al., 2016
Family 25	c.655G>A	p.(Gly219Arg)	homozygous	2 girls	Published Weeke at al., 2016
Family 26	c.997C>T	p. Arg333Ter	homozygous	1 girl and 1 boy	Published Schossig et al., 2017
Family 27	<b>c. 203C&gt;A</b> <b>c. 434C&gt;A</b>	<b>p. Pro68Gln</b> <b>p. Thr145Lys</b>	heterozygous	2 girls	Published Schossig et al., 2017
Family 28	c. 655G>A <b>c. 317A&gt;G</b>	p.Gly219Arg <b>p.His106Arg</b>	Heterozygous	2 boys	DECIPHER ID: 270740 <a href="https://decipher.sanger.ac.uk/search?q=SLC13A5#consented-patients/results">https://decipher.sanger.ac.uk/search?q=SLC13A5#consented-patients/results</a>

Family 29	<b>c.1268G&gt;A</b>	<b>p.Gly423Glu</b>	Homozygous	1 girl	DECIPHER ID: 264842 <a href="https://decipher.sanger.ac.uk/search?q=SLC13A5#consented-patients/results">https://decipher.sanger.ac.uk/search?q=SLC13A5#consented-patients/results</a>
	<b>c.231+2T&gt;G</b>	<b>splice donor mutation/variation</b>			<b>HGMD database exclusive mutation</b>
	<b>c.1227dupC</b>	<b>p.Ile410Hisfs</b>			<b>ClinVar exclusive pathogenic variants</b>
	<b>c.548-2A&gt;G</b>	<b>Splice acceptor variant</b>			<b>ClinVar exclusive pathogenic variants</b>
	<b>c. 716+5G&gt;A</b>	<b>intron variant</b>			<b>ClinVar exclusive pathogenic variants</b>