

PMID	Discovery method	Zygoty	cNomen	pNomen	NM_
26612766	Whole exome sequencing	homozygous	c.270delA	p.Lys90Asnfs8*	NM_025000
26440089	sanger	compound heterozygous	c.459-7_499del / c.1238delA	p.? / nonsense mutation	NM_025000.3
24015686	sanger	homozygous	c.436delC	p.Ala147Hisfs*9	not given
21963443	sanger	homozygous	c.321 + 1 G > A	p.?	not given
21044051	sanger	homozygous	c.906 G>A	p.Trp302*	not given
19026396	sanger	homozygous	c.436 delC	p.Ala147Hisfs*9	NM_025000.2
31152917	sanger	heterozygous	c.270 dup	p.Cys91Metfs × 28	NM_025000.4
34732557	sanger	homozygous	c.436delC	fs	not given
29574468	Whole exome sequencing	homozygous	c.436delC	p.Ala147Hisfs*9	not given
34590781	sanger, Whole exome sequencing	homozygous	c.436delC	p.Ala147Hisfs*9	not given
31347785	sanger	homozygous	c.1091+2T>C		not given
34630532	Whole exome sequencing	homozygous	c.1111delA	p.(Ile371Term)	not given
20507343	sanger	homozygous?	c.341C>A	p.Ser114*	not given
20507343	sanger	homozygous?	c..387G>A	p.Trp129*	not given
20507343	sanger	homozygous?	c.906G>A	p.Trp302*	not given
27240811	sanger	homozygous	c.436delC	p.Ala147Hisfs*9	not given
33819414	Whole exome sequencing	homozygous	c.1091+1G>A	p.?	not given