

S1 : Supplementary genetic features.

Patient	Mutation (NM_001127221.1)	Protein change	Previous published cases	Domain	Type	GnomAD database	CADD score
10	c.835C>T	p.(Arg279Cys)	Maksemous <i>et al.</i> 2016, Angelini <i>et al.</i> 2018	I S5 - I S6	Missense	Absent	26,2
11	c.1082G>A ‡	p.(Gly361Glu)	-	III S5 - III S6	Missense	Absent	33
6, 7, 8	c.1747C>T ‡	p.(Arg583*)	-	II S4, t	Non-sense	Absent	38
14	c.1847G>C ‡	p.(Ser616Thr)	-	II S5, t	Missense	Absent	25,7
4	c.2006A>C ‡	p.(Asp669Ala)	-	II S5 - II S6	Missense	Absent	24
2, 3, 12, 16	c.2137G>A	p.(Ala713Thr)	Epi4k 2016, Balck <i>et al.</i> 2017, Jiang <i>et al.</i> 2019	II S6, t	Missense	Absent	29,3
9	c.2815_2816del ‡	p.(Gly939Glnfs*128)	-	II S6 - III S1	Non-sense	Absent	-
13	c.4046G>A	p.(Arg1349Gln)	Knienim <i>et al.</i> 2011	III S4, t	Missense	Absent	32
15	c.4177G>A	p.(Val1393Met)	Butler <i>et al.</i> 2017	III S5, t	Missense	Absent	28
17	c.4273G>T ‡	p.(Glu1425*)	-	II S6 - III S1	Non-sense	Absent	44
18	c.4890G>A ‡	p.(Trp1630*)	-	IV S3, t	Non-sense	Absent	46
5	c.4990C>T	p.(Arg1664*)	Mantuano <i>et al.</i> 2010	IV S4, t	Non-sense	Absent	39
1	c.5083G>C ‡	p.(Val1695Leu)	-	IV S5, t	Missense	Absent	23,3

‡, not previously described; t, transmembrane domain.