Supplementary table 1 Results of pathogenicity assessment for detected variants

Gene	Nucleotide change*	Pathogenicity†	Rule	Description
PROM1	c.139del	Pathogenic	PVS1	This frameshift variant is expected to result in loss of function by nonsense-mediated mRNA decay.
			PM2	The variant is not found in gnomAD.
PROM1	c.794del	Pathogenic	PVS1	This frameshift variant is expected to result in loss of function by nonsense-mediated mRNA decay.
			PM2	The variant is not found in gnomAD.
PROM1	c.1238T>A	Likely pathogenic	PS2	*De novo* (both maternity and paternity confirmed) in a patient with the disease and no family history.
			PM2	The variant is not found in gnomAD.
			PP3	Multiple lines of computational evidence support a deleterious effect on the gene or gene product.
PROM1	c.1117C>T	Pathogenic	PS3	Functional studies demonstrate a damaging effect with reduced actin binding as well as protein mislocalization.
			PM2	This missense variant is not found in gnomAD.
			PP3	Multiple lines of computational evidence support a deleterious effect on the gene or gene product.
PROM1	c.2110C>T	Pathogenic	PS2	*De novo* (both maternity and paternity confirmed) in a patient with the disease and no family history.
			PM2	The variant is not found in gnomAD.
			PP3	Multiple lines of computational evidence support a deleterious effect on the gene or gene product.
CACNA1F	c.3115G>T	Pathogenic	PVS1	This nonsense variant is expected to result in loss of function by nonsense-mediated mRNA decay.
			PM2	The variant is not found in gnomAD.

*: The variants of *PROM1* and *CACNA1F* gene were presented based on NM_006017.3 and NM_001256789.3, respectively.

†: The pathogenicity of variant was presented according to the ACMG guidelines.