

Table S1. Genomic defects and genes analyzed prior to WES.

Gene	A.III-1	B.II-2
<i>17p dupl.</i>	+	+
<i>PMP22</i>	+	+
<i>MPZ</i>	+	+
<i>GJB1</i>	+	+
<i>NDRG1</i>	+	+
<i>YARS</i>	+	
<i>GARS</i>	+	
<i>AARS</i>	+	
<i>MFN2</i>	+	
<i>GDAP1</i>	+	+
<i>SH3TC2</i>	+	
<i>HINT1</i>	+	
<i>BSCL2</i>	+	
<i>CTDP1</i>	+	
<i>DNM2</i>	+	
<i>FGD4</i>	+	
<i>GAN</i>	+	
<i>HSP22</i> (<i>HSPB8</i>)	+	
<i>HSP27</i> (<i>HSPB1</i>)	+	
<i>IGHMBP2</i>	+	
<i>GNB4</i>	+	
<i>LITAF</i>	+	
<i>MTMR2</i>	+	+
<i>NEFL</i>	+	
<i>NTRK1</i>	+	
<i>PRX</i>	+	
<i>RAB7</i>	+	
<i>SPTLC1</i>	+	
<i>SPTLC2</i>	+	
<i>TRPV4</i>	+	

“+” - screening of the gene performed.

Table S2. Primers used for *PMP2* screening.

Exon	Forward primer	Reverse primer
1	5'-AGCCTTGCAAACTCCCCAG-3'	5'-TTGCTTAGTCCCGACACTGC-3'
2	5'-TGTCTCAGGAGGTACTCC-3'	5'-CCACTGAGCGTAGGATGTGG-3'
3	5'-AAAATCTTGGGTCAGTACTTGA-3'	5'-TGGTTCATACTGAAAGAACTGC-3'
4	5'-ACTGGAGACTGACATACTGAT-3'	5'-CTCCTCTGGCCCCTGTCA-3'

Table S3. Scores from the pathogenicity prediction programs.

cDNA	Protein	SIFT (max score 0)	PolyPhen2 (max score 1)	Mutation Taster (max score 1)	CADD (>20)
c.341T>C	p.Met114Thr	0.01 (Del)	1 (PD)	1 (DC)	27.6
c.344T>C	p.Val115Ala	0.02 (Del)	0.923 (PD)	1 (DC)	25.3

Abbreviations: “Del” – deleterious, “DC” – disease causing, “PD” – probably damaging. A CADD score >20 suggests that the variant is amongst the top 1% of deleterious variants in the human genome.