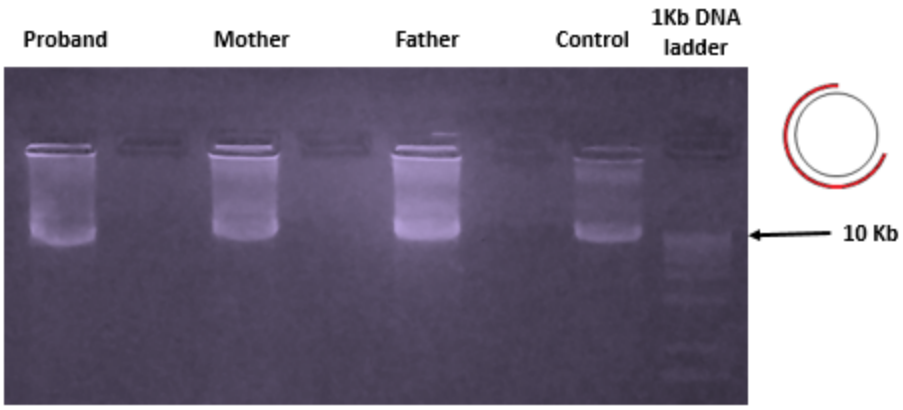

Figure S1: Long-range PCR amplification of the mitochondrial genome from blood was carried out between nucleotide positions 5,462–45 (20 ng total DNA per sample, 30 cycles of PCR; indicated by red line). No deletions in mtDNA was observed in the proband.

Table S1. List of 19 homozygous variants after the variant filtering. The variant in *MGME1* is highlighted in blue.

Gene	cDNA Change	OMIM Phenotype (of Gene)
IRF2BPL	c.368_369ins21	None
CACNA1H	c.4566+5G>A	Epilepsy, childhood absence, susceptibility to, 6
TELO2	c.749G>A	None
GLYR1	c.967A>G	None
SARS2	c.364-7C>A	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis
RSAD2	c.346+1G>T	None
RSAD2	c.827T>G	None
PPP4R3B	c.2518_2520del	None
ZNF638	c.1117T>G	None
DYSF	c.1799G>T	Miyoshi muscular dystrophy 1
MGME1	c.359del	Mitochondrial DNA depletion syndrome 11
MYLK2	c.972+14G>A	Cardiomyopathy, hypertrophic, 1, digenic
ASXL1	c.320A>G	Bohring-Opitz syndrome
SETD4	c.902-13_902-12insCT	None
SETD4	c.902-17_902-16insGT	None
C4ORF45	c.234_237del	None
B4GALT1	c.413-8C>A	Congenital disorder of glycosylation, type IId
SPAG8	c.1039G>A	None
AGTPBP1	c.909+16_909+18del	None