Supplementary Table 1

Summary of Clinical and laboratory findings

Patient :	**Skeletal muscle:**	**Heart:**	**Liver:**	**CNS:**
Family A, Patient A.II.1: Male, born 2008	Proximal, axial, pharyngeal, and distal weakness in upper extremities, abdomen, hands. Increased fatiguability. Cannot walk on heels. Difficulty to swallow. Slight scoliosis (Cobb angle 17°). p-CK 1200-2400 U/L (2-5 times upper normal reference level) p-Myoglobin 646-803 μg/L (9 -11 times upper normal reference level). EMG normal. Muscle biopsy: normal morphology. No increase of lysosomes or peroxisomes.	Echocardiography: Increased trabeculation left ventricle. Discrete left ventricle hypertrophy. Normal systolic and diastolic function. ECG normal.	Neonatal transient non-conjugated hyperbilirubimemia. s-ALAT 480-660 U/L (16-21 times upper normal reference level). s-ASAT 412-509 U/L Liver scan: slight to medium fibrosis. Normal values of p-ALP, p-albumin, p-bilirubin, PK, APTT, p-GT. Liver biopsy: normal morphology. No deposits of glycogen or other material. Electron microscopy: Increased and dilated ER. Disrupted structure of mitochondrial cristae. Deranged mitochondria with crystalline deposits.	Attention deficit disorder (ADD). Cognitive skills at lower average to borderline range.
Family A, Patient A.II.2: Male, born 2016	Increased fatiguability. Normal gross motor function. p-CK 420-1500 U/L (1-3 times upper normal reference level) Muscle biopsy not performed.	Echocardiography: discrete signs increased trabeculation left ventricle. Normal systolic and diastolic function. ECG normal.	s-ALAT: 240-540 U/L (8-17 times upper normal reference level). Normal values of p-ALP, p-albumin, p-bilirubin, PK, APTT, p-GT. Ultrasound liver normal. Liver biopsy not performed.	Late language development.

Family B, Patient 3 Male, Born Dec 2016	p-CK: 700-1000U/L LDH:1200-1500U/L Myoglobin: 481 Muscle biopsy not preformed	Prenatal suspicion of anomalous pulmonary vein opening Cardiac ultrasound after birth showed dilated superior Vena cava, cardiomegaly with large coronary arteries, a large PDA, a small aortic isthmus stenosis but was normal in the subsequent visit	Elevation of transaminase first noted at age of 2.5 years old p-ALAT: 220-640U/L p-ASAT:150-370U/L Ultrasound liver showed hepatopathy	At age of 3, lower vocabulary (approx. 10 Words) Slightly hypotonic, increased patellar stretch reflex on both sides Inconspicuous EEG and cranial MRI