SUPPORTING INFORMATION

Table S2 Selected variants of unknown significance (VUS)

Family Proband	**Variant 1** (transcript) genomic pos. hg19	**Variant 2** (transcript) genomic pos. hg19	**Variant 3** (transcript) genomic pos. hg19
Family 1	**RBM20 p.(Val535Ile)** c.1603G>A (NM_001134363.2) chr10:112557341-G>A	**DCTN1 p.(Ile196Val)** c.586A>G (NM_004082.4) chr2:074598723-T>C	**MYH14 p.(Arg1858Cys)** c.5572C>T (NM_001145809.2) chr19:050805020-C>T
II-2 (A)	+	+	+
III-3 (A)	-	+	+
II-5 (DP)	-	NT	-
ACMG Classification* (identified criteria)	**Likely Benign** (PP3, PP5, BS2, BP1)	**Benign** (BS1, BS2, BP1, BP4, BP6)	**Likely Benign** (PP3, BS2, BP1)
Family 2	**TRIM63 p.(Met14SerfsTer38)** c.35_38dupATCC (NM_032588.3) chr1:026393947-G>GGGAT	**TTN p.(Ser30125Phe)** c.90374C>T (NM_001256850.1) chr2:179410666-G>A	**ACTC1 p.(Ile371Thr)** c.1112T>C (NM_005159.4) chr15:035082635-A>G
II-1 (A)	+	+	+
II-2 (U)	-	+	-
ACMG Classification* (identified criteria)	**Uncertain Significance** (PM2)	**Benign** (PP3, BS1, BS2, BP1, BP6)	**Uncertain Significance** (PM2, PP3)

A, affected; U, unaffected; DP, patient affected by myopathy with divergent phenotype; NT, not tested
*verdict and identified ACMG criteria are based on Varsome database; PS/PM/PP – pathogenic strong/moderate/supporting; BS/BP – benign strong, supporting.