Supplementary Table 1. Mutations and genes included in each genetic panel.

Most common pathogenic variants in mtDNA and *POLG*
MT-RNR1 m.1555A>G *MT-TL1* m.3243A>G *MT-ND1* m.3460G>A (p.Ala52Thr) *MT-TK* m.8344A>G *MT-ATP6* m.8993T>G (p.Leu156Arg) *MT-ATP6* m.8993T>C (p.Leu156Pro) *MT-ATP6* m.9176T>C (p.Leu217Pro) *MT-ATP6* m.9176T>G (p.Leu217Arg) *MT-ND3* m.10158T>C (p.Ser34Pro) *MT-ND3* m.10191T>C (p.Ser45Pro) *MT-ND4* m.11777C>A (p.Arg340Ser) *MT-ND4* m.11778G>A (p.Arg340His) *MT-ND4* m.11832G>A (p.Trp358*) *MT-ND5* m.13513G>A (p.Asp393Asn) *MT-ND5* m.13514A>G (p.Asp393Gly) *MT-ND6* m.14459G>A (p.Ala72Val) *MT-ND6* m.14482C>A (p.Met64Ile) *MT-ND6* m.14482C>G (p.Met64Ile) *MT-ND6* m.14484T>C (p.Met64Val) *MT-ND6* m.14487T>C (p.Met63Val) *POLG* c.1399G>A (p.Ala467Thr)
mtDNA maintenance panel
DGUOK, MFN2, MPV17, OPA1, POLG, POLG2, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK, TYMP
Metabolic myopathies panel
ABHD5, ACADL, ACADM, ACADS, ACADVL, AGL, ALDOA, AMPD1, ATP2A1, CPT1B, CPT2, ENO3, ETFA, ETFB, ETFDH, GBE1, GYG1, GYS1, HADHA, HADHB, LDHA, LPIN1, PFKM, PGAM2, PGM1, PGK1, PHKA1, PHKB, PNPLA2, PYGM, SLC22A5, SLC25A20