Izomdisztrófia/Myopathia (Muscular dystrophy/Myopathy) génpanel v:2022.01.28.

Panel tartalma 359 gén:

AARS1, ABHD5, ACAD9, ACADM, ACADVL, ACTA1, ACTN2, ADCK3, ADSSL1, AGL, AGRN, AHCY, AIFM1, ALDOA, ALG14, ALG2, AMPD1, ANO5, ARHGEF10, ARHGEF9, ASAH1, ASCC3, ATL1, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BET1, BICD2, BIN1, BSCL2, BVES, CACNA1S, CAPN3, CASK, CASQ1, CAV1, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCN1, CNTN1, COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ2, COX6A1, CPT2, CRPPA, CRYAB, CTDP1, CSRP3, DAG1, DCTN1, DES, DGUOK, DHCR24, DHTKD1, DHX16, DMD, DMPK, DNA2, DNAJB2, DNAJB6, DNM2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DUX4, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, ENO3, EPG5, ERCC5, ERCC6, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FAM111B, FBLN5, FBN2, FBXO38, FDX1L, FGD4, FHL1, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GAN, GARS1, GBA, GBE1, GDAP1, GFER, GFPT1, GGPS1, GJB1, GLE1, GLRA1, GLRB, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GPHN, GYG1, GYS1, HACD1, HADHA, HADHB, HINT1, HK1, HNRNPA1, HNRNPA2B1, HNRNPDL, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, HSPG2, HTRA2, IGHMBP2, INF2, INPP5K, ISCU, ISPD, ITGA7, JAG2, KARS1, KAT6B, KBTBD13, KCNA1, KCNE3, KIF1A, KIF1B, KIF5A, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMB2, LAMP2, LARGE, LARGE1, LDB3, LDHA, LGI4, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRIF1, LRSAM1, MAGEL2, MAMLD1, MAP3K20, MARS1, MATR3, MED25, MEGF10, MFN2, MICU1, MME, MPV17, MPZ, MSTO1, MTM1, MTMR14, MTMR2, MUSK, MYBPC1, MYBPC3, MYF5, MYF6, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYOD1, MYOT, MYPN, NALCN, NDRG1, NEB, NEFL, NTRK1, OPA1, OPA3, ORAI1, PABPN1, PAX7, PDK3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PLEC, PLEKHG5, PLOD2, PMM2, PMP22, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PPA2, PREPL, PRKAG2, PRPS1, PRX, PUS1, PYGM, PYROXD1, QARS1, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RRM2B, RXYLT1, RYR1, RYR3, SBF1, SBF2, SCN4A, SELENON, SEPT9, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIL1, SLC12A6, SLC16A1, SLC22A5, SLC25A1, SLC25A20, SLC25A4, SLC25A42, SLC25A46, SLC5A7, SLC6A5, SMCHD1, SMN1, SMN2, SPEG, SPG11, SPTBN4, SPTLC1, SPTLC2, SRPK3, STAC3, STIM1, STIM2, SUCLA2, SUCLG1, SUN1, SUN2, SVIL, SYNE1, SYNE2, TANGO2, TAZ, TCAP, TFG, TGFB3, TIA1, TK2, TMEM126B, TMEM43, TMEM5, TNNC2, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM2, TRIM32, TRIP4, TRPV4, TSEN2, TSFM, TTN, TWNK, TYMP, UBA1, UNC45B, VAMP1, VAPB, VCP, VIPAS39, VMA21, VPS13A, VPS33B, VRK1, WNK1, XK, YARS1, YARS2, ZC4H2.

Forrás:

Congenital myopathy (Version 2.70)
https://panelapp.genomicsengland.co.uk/panels/225/ (Megtekintve: 2022.01.28.)

Congenital muscular dystrophy (Version 2.21)
https://panelapp.genomicsengland.co.uk/panels/207/ (Megtekintve: 2022.01.28.)

Limb girdle muscular dystrophy (Version 2.33)
https://panelapp.genomicsengland.co.uk/panels/185/ (Megtekintve: 2022.01.28.)

Comprehensive Muscular Dystrophy / Myopathy Panel
https://blueprintgenetics.com/tests/panels/neurology/comprehensive-muscular-dystrophy-myopathy-panel/ (Megtekintve: 2022.01.28.)

Neuromuscular panel
https://www.centogene.com/science/centopedia/ngs-panel-genetic-testing-for-muscular-dystrophy.html (Megtekintve: 2022.01.28.)

Invitae Comprehensive Muscular Dystrophy Panel
https://www.invitae.com/pt/physician/tests/03291/ (Megtekintve: 2022.01.28.)