Izomdisztrófia/Myopathia (Muscular dystrophy/Myopathy) génpanel v:2023.10.12.

Panel tartalma 386 gén:

AARS1, ABHD5, ACAD9, ACADM, ACADVL, ACTA1, ACTG2, ACTN2, ADCK3, ADGRG6, AGL, AGRN, AHCY, AIFM1, ALDOA, ALG14, ALG2, AMPD1, ANO5, ARHGEF9, ASAH1, ASCC1, ATAD1, 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, CNTNAP1, COL12A1, COL13A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COX6A1, CPT2, CRLF1, CRPPA, CRYAB, CTDP1, CSRP3, DAG1, DCTN1, DES, DGUOK, DHCR24, DHTKD1, DMD, DMPK, DNA2, DNAJB2, DNAJB6, DNM2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DUX4, DYNC1H1, DYSF, ECEL1, EGR2, ELP1, EMD, ENO3, ERCC5, ERCC6, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FBLN5, FBN2, FBXO38, FDX1L, FDX2, FGD4, FHL1, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNC, GAA, GAN, GARS1, GBA, GBE1, GDAP1, GFPT1, GGPS1, GJB1, GLDN, GLE1, GLRA1, GLRB, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GPHN, GYG1, GYS1, HADHA, HADHB, HINT1, HK1, HNRNPA2B1, HNRNPDL, HOXD10, HRAS, HSPB1, HSPB3, HSPB8, HSPG2, IGHMBP2, INF2, INPP5K, ISCU, ISPD, ITGA7, JAG2, KARS1, KAT6B, KBTBD13, KCNA1, KCNE3, KCNJ2, KIF1A, KIF1B, KIF5A, KLHL40, KLHL41, KLHL7, KY, LAMA2, LAMB2, LAMP2, LARGE, LARGE1, LDB3, LDHA, LGI4, LIMS2, LITAF, LMNA, LMOD3, LPIN1, LRIF1, LRP4, LRSAM1, MAGEL2, MAMLD1, MAP3K20, MARS1, MATR3, MED25, MEGF10, MFN2, MICU1, MME, MOCS1, MPV17, MPZ, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTM1, MTMR14, MTMR2, MUSK, MYBPC1, MYBPC3, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NALCN, NDRG1, NEB, NTRK1, OPA1, OPA3, ORAI1, PABPN1, PAX7, PDK3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PIP5K1C, PLEC, PLEKHG5, PLOD2, PMM2, PMP22, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PREPL, PRKAG2, PRPS1, PRX, PYGM, PYROXD1, QARS1, RAB7A, RAPSN, RBCK1, REEP1, RETREG1, RRM2B, RXYLT1, RYR1, SBF1, SBF2, SCN4A, SELENON, SEPT9, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIL1, SLC12A6, SLC16A1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SLC6A5, SMCHD1, SMN1, SMN2, SMPD4, SPEG, SPG11, SPTBN4, SPTLC1, SPTLC2, STAC3, STIM1, SUCLA2, SUCLG1, SYNE1, SYNE2, SYT2, TAFAZZIN, TANGO2, TBCK, TCAP, TFG, TGFB3, TIA1, TK2, TMEM126B, TMEM43, TMEM5, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM2, TRIM32, TRIP4, TRPV4, TSEN2, TSFM, TTN, TWNK, TYMP, UBA1, VAMP1, VAPB, VCP, VIPAS39, VMA21, VPS13A, VPS33B, VRK1, WNK1, XK, YARS1, ZC4H2

Forrás:

Congenital muscular dystrophy (Version 4.9)

https://panelapp.genomicsengland.co.uk/panels/207/ (Megtekintve: 2023.05.16.)

Limb girdle muscular dystrophies, myofibrillar myopathies and distal myopathies (Version 4.20)

https://panelapp.genomicsengland.co.uk/panels/185/ (Megtekintve: 2023.05.16.)

Comprehensive Muscular Dystrophy / Myopathy Panel

https://blueprintgenetics.com/tests/panels/neurology/comprehensive-muscular-dystrophy-myopathy-panel/ (Megtekintve: 2023.05.16.)

Neuromuscular panel

https://www.centogene.com/diagnostics/our-tests/ngs-panels/neurology (Megtekintve: 2023.07.19.)

Invitae Comprehensive Muscular Dystrophy Panel

https://www.invitae.com/en/providers/test-catalog/test-03291 (Megtekintve: 2023.05.16.)