

Izomdisztrófia/Myopathia (Muscular dystrophy/Myopathy) génpanel v:2025-05-22

Panel tartalma 205 gén:

ABHD5, ACAD9, ACADVL, ACTA1, ACTN2, ADSS1, ANO5, AR, ASCC1, ASCC3, ATP2A1, B3GALNT2, B4GAT1, BAG3, BET1, BICD2, BIN1, BVES, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHKB, CIAO1, CNTN1, COL12A1, COL13A1, COL25A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COQ2, COQ8A, COX6A2, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DHX16, DMD, DMPK, DNAJB4, DNAJB6, DNM2, DOK7, DOLK, DPM1, DPM2, DPM3, DTNA, DYSF, ECEL1, EMD, EPG5, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GBE1, GFER, GGPS1, GMPPB, GNE, GOLGA2, GOSR2, GYG1, HACD1, HMGCR, HNRNPA2B1, HNRNPDL, INPP5K, ISCU, ITGA7, JAG2, KBTBD13, KLHL40, KLHL41, KY, LAMA2, LAMP2, LARGE1, LDB3, LETM1, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, MAP3K20, MEGF10, MICU1, MLIP, MME, MSTO1, MTM1, MTMR14, MYBPC1, MYBPC3, MYF5, MYH2, MYH3, MYH7, MYL1, MYL2, MYMK, MYMX, MYO18B, MYOD1, MYOT, MYPN, NEB, NEFL, ORAI1, PABPN1, PAX7, PFKM, PGK1, PHKA1, PHKB, PIEZO2, PLEC, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PPA2, PYGM, PYROXD1, RBCK1, RFC4, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SELENON, SEPTIN9, SGCA, SGCB, SGCD, SGCG, SIL1, SLC22A5, SLC25A20, SLC25A4, SLC25A42, SMCHD1, SMN1, SMN2, SNUPN, SPEG, SPTBN4, STAC3, STIM1, SUCLA2, SUCLG1, SVIL, SYNE1, SYNE2, TANGO2, TCAP, TIA1, TK2, TMEM126B, TMEM43, TNNC2, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM32, TRIP4, TSFM, TTN, TYMP, UNC45B, VCP, VMA21, VPS13A, VWA1, ZC4H2

Forrás:

Congenital myopathy (Version 6.1)

<https://panelapp.genomicsengland.co.uk/panels/225/>

Congenital muscular dystrophy (Version 6.1)

<https://panelapp.genomicsengland.co.uk/panels/207/>

Limb girdle muscular dystrophies, myofibrillar myopathies and distal myopathies (Version 5.3)

<https://panelapp.genomicsengland.co.uk/panels/185/>

Comprehensive Muscular Dystrophy / Myopathy Panel

<https://blueprintgenetics.com/tests/panels/neurology/comprehensive-muscular-dystrophy-myopathy-panel/>

Invitae Comprehensive Muscular Dystrophy Panel

<https://www.invitae.com/pt/physician/tests/03291/>

(Megtekintve: 2025-05-22)