

Veleszületett myopathia (Congenital myopathy) génpanel v:2025-05-21

Panel tartalma 119 gén:

ACTA1, ACTN2, ADSS1, ALG2, ANO5, AR, ASCC1, ASCC3, ATP2A1, BAG3, BICD2, BIN1, CACNA1S, CAV3, CCDC78, CFL2, CHKB, CIAO1, CNTN1, COL12A1, COL13A1, COL25A1, COL6A1, COL6A2, COL6A3, COX6A2, CRPPA, CRYAB, DES, DHX16, DMPK, DNAJB4, DNAJB6, DNMT2, DOK7, DYSF, ECEL1, EPG5, FHL1, FKBP14, FKRP, FKTN, FLNC, FXR1, GBE1, GFER, GNE, HACD1, HNRNPA2B1, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LAMA2, LAMP2, LARGE1, LDB3, LETM1, LMNA, LMOD3, MAP3K20, MEGF10, MICU1, MLIP, MTM1, MTMR14, MYBPC1, MYBPC3, MYF5, MYF6, MYH2, MYH3, MYH7, MYL1, MYL2, MYMK, MYMX, MYO18B, MYOD1, MYOT, MYPN, NEB, NEFL, ORAI1, PAX7, PIEZO2, POMGNT1, POMT1, POMT2, PPA2, PYROXD1, RFC4, RYR1, RYR3, SCN4A, SELENON, SLC25A4, SLC25A42, SPEG, SPTBN4, STAC3, STIM1, SVIL, SYNE1, TIA1, TNNC2, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRDN, TRIP4, TTN, UNC45B, VMA21, VWA1, ZC4H2

Forrás:

Congenital myopathy (Version 6.1)

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

Congenital Myopathy NGS Panel

<https://www.fulgentgenetics.com/Congenital-Myopathy>

(Megtekintve: 2025-05-21)