

## Neuromuscularis betegségek (Neuromuscular) génpanel (v. 2025-04-23)

Panel tartalma 735 gén:

AARS1, ABCA1, ABCC9, ABCD1, ABHD12, ABHD16A, ABHD5, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACTA1, ACTC1, ACTN2, ADAMTS10, ADAMTS15, ADAR, ADCY6, ADGRG6, ADSL, ADSS1, AFG3L2, AGK, AGL, AGRN, AGTPBP1, AGXT, AHCY, AIFM1, AIMP1, ALDH18A1, ALDH3A2, ALDOA, ALG14, ALG2, ALG3, ALS2, AMACR, AMPD1, AMPD2, AMPD3, ANG, ANO5, ANTXR2, AP1S1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APTX, AR, ARG1, ARHGEF10, ARL6IP1, ARSA, ASAH1, ASCC1, ASCC3, ASXL1, ATAD1, ATL1, ATL3, ATM, ATP13A2, ATP1A1, ATP1A2, ATP2A1, ATP2A2, ATP7A, ATP7B, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B3GALNT2, B4GALNT1, B4GAT1, BAG3, BCKDHB, BET1, BICD2, BIN1, BRAF, BSCL2, BVES, C19orf12, C1QBP, C9orf72, CACNA1A, CACNA1E, CACNA1S, CADM3, CAPN1, CAPN3, CASQ1, CASQ2, CAV1, CAV3, CAVIN1, CCDC47, CCDC78, CD59, CEP55, CFL2, CHAT, CHCHD10, CHKB, CHMP1A, CHMP2B, CHRNA1, CHRN1, CHRND, CHRNE, CHRNG, CHST14, CIAO1, CLCF1, CLCN1, CNBP, CNTN1, CNTNAP1, COA7, COASY, COL12A1, COL13A1, COL25A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ2, COQ4, COQ7, COQ8A, COQ9, COX15, COX20, COX6A1, COX6A2, COX6B1, CPOX, CPT1A, CPT1B, CPT2, CRLF1, CRPPA, CRYAB, CTDP1, CUL4B, CYP27A1, CYP2C8, CYP2U1, CYP7B1, CSRP3, DAG1, DARS1, DARS2, DCTN1, DDC, DDHD1, DDHD2, DEGS1, DES, DGUOK, DHCR24, DHH, DHTKD1, DHX16, DMD, DMPK, DNA2, DNAJB2, DNAJB4, DNAJB6, DNAJC3, DNMT2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DRP2, DSC2, DSG2, DSP, DST, DSTYK, DTNA, DUX4, DYNC1H1, DYSF, EBP, ECEL1, EGR2, ELP1, EMD, EMILIN1, ENO3, ENTPD1, EPG5, ERBB3, ERBB4, ERCC1, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, EXOSC9, FA2H, FAH, FAM111B, FAM126A, FAM20C, FARS2, FBLN5, FBN2, FBP2, FBXL4, FBXO38, FDX2, FGD4, FGF14, FGFR2, FGFR3, FHL1, FIG4, FILIP1, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLNC, FLVCR1, FMR1, FUS, FXN, FXR1, GAA, GAD1, GALC, GAN, GARS1, GATM, GBA, GBA2, GBE1, GBF1, GCH1, GDAP1, GFER, GFM2, GFPT1, GGPS1, GIPC1, GJB1, GJC2, GLA, GLDN, GLE1, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GRN, GSN, GYG1, GYS1, HACD1, HACE1, HADH, HADHA, HADHB, HARS1, HFE, HIKESHI, HINT1, HK1, HMBS, HMGCR, HNRNPA1, HNRNPA2B1, HNRNPDL, HRAS, HSPB1, HSPB3, HSPB8, HSPD1, HSPG2, HTRA2, HTT, IARS2, IBA57, IGHMBP2, INF2, INPP5K, IRF6, ISCU, ITGA7, ITPR1, ITPR3, JAG1, JAG2, JPH2, JUP, KARS1, KAT6B, KBTBD13, KCNA1, KCNA2, KCNC3, KCNJ2, KCNJ5, KCNK3, KDM5C, KIDINS220, KIF1A, KIF1B, KIF1C, KIF21A, KIF5A, KIF5C, KLHL40, KLHL41, KLHL7, KLHL9, KRAS, KY, LICAM, LAMA2, LAMA4, LAMA5, LAMB2, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LETM1, LGI3, LGI4, LIMS2, LITAF, LMNA, LMOD3, LMX1B, LPIN1, LRIF1, LRP12, LRP4, LRSAM1, LYST, MAG, MAGEL2, MAN2B1, MAP1B, MAP2K1, MAP2K2, MAP3K20, MARS1, MARS2, MATR3, MB, MCM3AP, MED12, MED25, MEG3, MEGF10, MET, MFN2, MGME1, MICU1, MLIP, MMACHC, MME, MORC2, MPV17, MPZ, MRE11, MSTO1, MT-CO1, MT-CO2, MTM1, MTMR14, MTMR2, MTPAP, MTRFR, MT-TL1, MTPP, MUSK, MYBPC1, MYBPC3, MYF5, MYF6, MYH1, MYH14, MYH2, MYH3, MYH6, MYH7, MYH8, MYL1, MYL11, MYL2, MYL3, MYMK, MYMX, MYO18B, MYO9A, MYOD1, MYOT, MYPN, NAGA, NAGLU, NALCN, NDRG1, NEB, NEFH, NEFL, NEK1, NEK9, NEXN, NGF, NIPA1, NKX6-2, NOP56, NRAS, NT5C2, NTRK1, NUDT2, NUP88, OBSCN, OPA1, OPA3, OPTN, ORAI1, PABPN1, PAX7, PCDH12, PCYT2, PDHA1, PDK3, PDSS1, PDSS2, PDXK, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGAP1, PGK1, PGM1, PHKA1, PHKB,

PHKG1, PHKG2, PHYH, PI4KA, PIEZO2, PIGS, PIP5K1C, PKP2, PLAAT3, PLEC, PLEKHG5, PLN, PLOD1, PLOD2, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA2, PNPLA6, PNPLA8, POGLUT1, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, POR, PPA2, PPOX, PPP2R2B, PRDM12, PREPL, PRG4, PRKAG2, PRKCG, PRNP, PRPS1, PRX, PSEN1, PTEN, PTPN11, PTRH2, PUS1, PYGM, PYROXD1, RAB3GAP2, RAB7A, RAF1, RAPSN, RBCK1, RBM20, REEP1, REEP2, RETREG1, RFC4, RIPK4, RIT1, RNASEH1, RNASEH2B, RRM2B, RTN2, RXYLT1, RYR1, RYR2, RYR3, SACS, SARS2, SBF1, SBF2, SCARB2, SCARF2, SCN10A, SCN11A, SCN1A, SCN4A, SCN5A, SCN9A, SCP2, SCYL1, SCYL2, SDHA, SELENOI, SELENON, SEPTIN9, SERAC1, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SIGMAR1, SIL1, SKI, SLC12A6, SLC16A1, SLC16A2, SLC18A3, SLC1A3, SLC1A4, SLC22A12, SLC22A5, SLC25A1, SLC25A19, SLC25A20, SLC25A3, SLC25A4, SLC25A42, SLC25A46, SLC29A3, SLC2A1, SLC2A9, SLC33A1, SLC35A3, SLC52A1, SLC52A2, SLC52A3, SLC5A6, SLC5A7, SLC6A9, SMAD3, SMAD4, SMCHD1, SMN1, SMN2, SMPD4, SMPX, SNAP25, SNRPN, SOD1, SORD, SOS1, SOX10, SPART, SPAST, SPEG, SPG11, SPG21, SPG7, SPTAN1, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SQSTM1, SRPK3, STAC3, STIM1, STIM2, SUCLA2, SUCLG1, SURF1, SVIL, SYNE1, SYNE2, SYT15, SYT2, TAFAZZIN, TAMM41, TANGO2, TARDBP, TBCD, TBK1, TBP, TCAP, TDP1, TECPR2, TFG, TGFB2, TGFB3, TGFBR1, TGFBR2, TIA1, TK2, TMEM43, TNNC1, TNNC2, TNNI2, TNNI3, TNNT1, TNNT2, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM1, TPM2, TPM3, TRAPPC11, TRDN, TRIM2, TRIM32, TRIP4, TRMT5, TRPA1, TRPV4, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTN, TTPA, TTR, TUBA4A, TUBB3, TUBB4A, TWNK, TYMP, UBA1, UBAP1, UBQLN1, UBQLN2, UNC13A, UNC45B, VAMP1, VAPB, VCL, VCP, VDR, VIPAS39, VMA21, VPS13A, VPS33B, VRK1, VWA1, WARS1, WASHC5, WDR45B, WNK1, WWTR1, XK, XPA, YARS1, YARS2, ZC4H2, ZEB2, ZFH2, ZFYVE26, ZMPSTE24, ZNF335

Források:

Other rare neuromuscular disorders (Version 26.33)

<https://panelapp.genomicsengland.co.uk/panels/465/>

Invitae Comprehensive Neuromuscular Disorders Panel

<https://www.ncbi.nlm.nih.gov/gtr/tests/551683/>

Neuromuscular Disorders Panel (GeneDX)

<https://providers.genedx.com/tests/detail/neuromuscular-disorders-panel-819>

Neuromuscular Diseases (Cegat)

<https://cegat.com/diagnostics/rare-diseases/neuromuscular-diseases/>

Neuromuscular NGS Panel (Fulgent)

<https://www.fulgentgenetics.com/neuromuscular>

Neuromuscular disorders panel Upgraded

<https://www.centportal.com/order/new/products/analysis-method?queryType=TEST&query=Neuromuscular%20Disorders%20Upgraded>

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