

Mozgászavarok (Movement disorders) génpanel v:2023.10.12.

Panel tartalma 1016 gén:

AAAS, AARS1, AARS2, AASS, ABCA1, ABCA2, ABCA7, ABCB7, ABCC8, ABCD1, ABHD12, ABHD5, ACAT2, ACBD5, ACO2, ACOX1, ACOX2, ACP5, ACTB, ACTL6B, ADA2, ADAMTS13, ADAR, ADCY5, ADCY6, ADD3, ADGRG1, ADH1C, ADPRS, ADSL, AFG3L2, AGA, AGTPBP1, AHI1, AIFM1, AIMP1, ALAD, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALG6, ALS2, AMACR, AMPD2, ANG, ANK3, ANKLE2, ANO10, ANO3, ANOS1, AP1S2, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOB, APP, APTX, ARCN1, ARFGEF2, ARG1, ARL13B, ARL3, ARL6, ARL6IP1, ARMC9, ARNT2, ARSA, ARV1, ARX, ASAH1, ASL, ASNS, ASS1, ATCAY, ATG5, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP2B4, ATP5F1E, ATP6AP2, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRX, ATXN1, ATXN2, ATXN3, ATXN7, AUH, B3GALT6, B4GALNT1, B4GAT1, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCL11B, BCOR, BCS1L, BDNF, BICD2, BOLA3, BRAT1, BSCL2, BTB, C12orf4, C19orf12, C2CD3, C5orf42, C9orf72, CA8, CACNA1A, CACNA1B, CACNA1D, CACNA1G, CACNA2D2, CACNB4, CAMTA1, CAPN1, CARS2, CASK, CAV1, CBY1, CC2D2A, CCDC28B, CCDC88C, CCT5, CDKL5, CENPF, CEP104, CEP120, CEP152, CEP290, CEP41, CFH, CHAMP1, CHCHD10, CHCHD2, CHMP1A, CHMP2B, CHP1, CIT, CIZ1, CKAP2L, CLCN2, CLIC2, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CNTN2, CNTNAP1, CNTNAP2, COA3, COA7, COA8, COASY, COG2, COG4, COG5, COL18A1, COL4A1, COL4A2, COL6A3, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX8A, CP, CPLANE1, CPS1, CPT1C, CRB2, CREBBP, CRLF1, CRPPA, CTBP1, CTC1, CTDP1, CTNNB1, CTSD, CTSF, CUL4B, CWF19L1, CYB5R3, CYC1, CYP27A1, CYP2U1, CYP7B1, CSF1R, CSPP1, CSTB, DAB1, DAO, DARS1, DARS2, DBH, DBT, DCAF10, DCAF17, DCC, DCPS, DCTN1, DCX, DDC, DDHD1, DDHD2, DDX3X, DDX59, DEAF1, DHCR24, DHCR7, DHFR, DHPS, DKC1, DLAT, DLD, DMXL2, DNA2, DNAJB2, DNAJC12, DNAJC13, DNAJC19, DNAJC3, DNAJC5, DNAJC6, DNMI1L, DNMT1, DOCK3, DOCK8, DPM1, DPYD, DPYS, DRD2, DRD3, DRD5, DYNC1H1, DYRK1A, EARS2, EBF3, ECHS1, EEF2, EGR2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELOVL1, ELOVL4, ELOVL5, ELP2, EMC1, EML1, ENTPD1, EPM2A, ERAL1, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ETHE1, EVC, EVC2, EXOC3L2, EXOC8, EXOSC3, EXOSC8, EXOSC9, EXT2, EZH2, FA2H, FAM149B1, FAR1, FARS2, FASTKD2, FAT2, FBXL4, FBXO7, FGD4, FGF12, FGF14, FGFR1, FIG4, FITM2, FKRP, FKTN, FLNA, FLNC, FLVCR1, FOLR1, FOXG1, FOXP2, FOXRED1, FRMD4A, FRRS1L, FTL, FUCA1, FUS, FXN, GABRB1, GABRB2, GABRB3, GAD1, GALC, GAMT, GAN, GATAD2B, GBA, GBA1, GBA2, GBE1, GCDH, GCH1, GCLC, GDAP2, GEMIN4, GFAP, GFM1, GFM2, GIGYF2, GJA1, GJB1, GJC2, GLB1, GLI3, GLRA1, GLRB, GLRX5, GLS, GLUD2, GLYCTK, GM2A, GMPPA, GMPPB, GNAL, GNAO1, GOSR2, GPAA1, GPR88, GRIA3, GRID2, GRIK2, GRIN1, GRIN2B, GRIN2D, GRM1, GRN, GSS, GTPBP2, HACE1, HARS1, HARS2, HCFC1, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HK1, HMBS, HNRNPA1, HNRNPA2B1, HNRNPH2, HPCA, HPDL, HPRT1, HSD17B10,

HSD17B4, HSPD1, HSPG2, HTRA1, HTRA2, HTT, HTT_CAG, HYLS1, IBA57, ICK, IFIH1, IFRD1, IFT140, IFT172, IFT27, IFT74, INPP5E, INTS8, IPPK, IRF2BPL, ISCA2, ISG15, ITM2B, ITPR1, IVD, JAM3, JPH3, KANK1, KARS1, KATNIP, KCNA1, KCNA2, KCNA4, KCNC1, KCNC3, KCND3, KCNJ10, KCNJ6, KCNK18, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD17, KCTD7, KDM5C, KIAA0556, KIAA0586, KIAA0753, KIAA1161, KIDINS220, KIF14, KIF1A, KIF1B, KIF1C, KIF2A, KIF5A, KIF5C, KIF7, KLC2, KMT2B, KY, L1CAM, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB2, LARGE1, LIAS, LIPT1, LIPT2, LMNB1, LMNB2, LRP4, LRPPRC, LRRK2, LRSAM1, LYRM7, LYST, MAG, MAN2B1, MAOA, MAPT, MARS1, MARS2, MAT1A, MATR3, MBD5, MCCC2, MCOLN1, MDH2, MECP2, MECP2, MECP2, MED13L, MED17, MED25, MFF, MFN2, MFSD2A, MFSD8, MGME1, MICU1, MIPEP, MKS1, MLC1, MMACHC, MMADHC, MME, MOCS1, MOCS2, MORC2, MPDU1, MPV17, MPZ, MR1, MRE11, MRPS22, MRPS34, MT-ATP6, MT-ND1, MT-ND6, MTO1, MTPAP, MTRFR, MTRFR, MUT, MVK, MYBPC1, MYH14, MYO7A, NADK2, NAGA, NAGLU, NALCN, NANS, NAT8L, NAXE, NDE1, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEFH, NEFL, NEU1, NEXMIF, NF1, NF2, NFASC, NGLY1, NHLRC1, NIPA1, NKX2-1, NKX6-2, NMNAT1, NOL3, NOS3, NOTCH3, NPC1, NPC2, NPHP1, NPHP3, NR4A2, NSUN2, NT5C2, NUBPL, NUP62, OCLN, OFD1, OPA1, OPA3, OPHN1, OPTN, OSGEP, OTC, PAFAH1B1, PANK2, PARK7, PARN, PARS2, PC, PCCA, PCCB, PCDH12, PCDH19, PCLO, PCNA, PDE10A, PDE2A, PDE6D, PDE8B, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFN1, PGAP1, PGK1, PGM3, PHGDH, PHKA1, PHYH, PIBF1, PIEZO2, PIGA, PIGG, PIGN, PIGQ, PIGS, PIK3R5, PINK1, PITRM1, PITX3, PLA2G6, PLAA, PLCB1, PLD3, PLEKHG2, PLEKHG4, PLP1, PLXND1, PMM2, PMP22, PMPCA, PNKD, PNKP, PNP, PNPLA6, PNPLA8, PNPT1, POC1B, PODXL, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PON1, PPP1R15B, PPP2R2B, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM8, PRF1, PRICKLE1, PRICKLE2, PRKCG, PRKN, PRKRA, PRNP, PROK2, PROKR2, PRPH, PRPS1, PRRT2, PRX, PSAP, PSAT1, PSEN1, PSEN2, PSMB8, PTEN, PTF1A, PTPN23, PTRH2, PTRHD1, PTS, PUM1, PURA, PYCR2, PYROXD1, QARS1, QDPR, RAB18, RAB27A, RAB39B, RAB3GAP1, RABL2A, RAD50, RARS1, RARS2, REEP1, REEP2, RETREG1, RFT1, RHOBTB2, RLIM, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF168, RNF170, RNF216, ROGDI, RORA, RPGRIP1L, RPIA, RPL10, RRM2B, RTN2, RTN4IP1, RTTN, RUBCN, SACS, SAMD9L, SAMHD1, SARS1, SCARB2, SCLT1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SCO1, SCO2, SCP2, SCYL1, SDHA, SDHAF1, SDHB, SDHD, SELENOI, SEPSECS, SERAC1, SETX, SFXN4, SGCE, SGPL1, SH3TC2, SHMT2, SIGMAR1, SIL1, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC18A2, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A4, SLC25A46, SLC2A1, SLC30A10, SLC30A9, SLC33A1, SLC39A14, SLC39A4, SLC41A1, SLC44A1, SLC46A1, SLC52A2, SLC52A3, SLC6A1, SLC6A17, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC9A1, SLC9A6, SMAD4, SMC1A, SMG9, SMPD1, SNAP25, SNCA, SNCAIP, SNX14, SOD1, SOX10, SOX2, SOX6, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2,

SPTBN4, SPTLC2, SQSTM1, STAMBP, STN1, STUB1, STXBP1, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SVBP, SYNE1, SYNJ1, SYT14, SYT2, TACO1, TAF1, TAF2, TANGO2, TAPT1, TARDBP, TBC1D20, TBC1D23, TBC1D24, TBC1D32, TBCD, TBCE, TBK1, TBP, TCF4, TCN2, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TELO2, TENM4, TFG, TGM6, TH, THAP1, THG1L, THOC2, TIMM8A, TIMMDC1, TINF2, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM240, TMEM67, TMEM70, TMX2, TOGARAM1, TOR1A, TP53RK, TPI1, TPK1, TPM2, TPP1, TRAPPC11, TRAPPC12, TRAPPC4, TREM2, TREX1, TRIM32, TRNT1, TRPC3, TRPM7, TSEN15, TSEN2, TSEN54, TSFM, TTBK2, TTC19, TTC21B, TTPA, TTR, TUBA1A, TUBA4A, TUBB, TUBB3, TUBB4A, TUBG1, TWNK, TXN2, TXNDC15, TYMP, TYROBP, UBA5, UBE3A, UBQLN2, UBR4, UBTF, UCHL1, UFC1, UNC80, UPB1, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRFS1, UQCRQ, UROC1, USP18, VAC14, VAMP1, VAMP2, VAPB, VARS2, VCP, VLDLR, VPS11, VPS13A, VPS13B, VPS13C, VPS13D, VPS16, VPS35, VPS37A, VPS41, VPS51, VPS53, VRK1, VWA3B, WARS2, WASHC5, WDR26, WDR45, WDR62, WDR63, WDR73, WDR81, WFS1, WNK1, WWOX, XK, XPA, XPNPEP3, XPR1, XRCC1, XRCC4, YME1L1, YY1, ZC4H2, ZFYVE26, ZFYVE27, ZNF142, ZNF335, ZNF423, ZNHIT3, ZSWIM6

Forrás:

Comprehensive Movement Disorders Panel

<https://www.preventiongenetics.com/testInfo?val=Comprehensive-Movement-Disorders-Panel> (Megtekintve: 2023.05.17.)

Movement disorders - childhood onset (Version 7.10)

<https://panelapp.genomicsengland.co.uk/panels/494/> (Megtekintve: 2023.05.17.)

Adult onset dystonia, chorea or related movement disorder (Version 3.1)

<https://panelapp.genomicsengland.co.uk/panels/540/> (Megtekintve: 2023.05.17.)