

Veleszületett hypotonia (congenital hypotonia) génpanel v:2022.02.14.

Panel tartalma 1619 gén:

AAAS, AARS, AARS2, AASS, ABAT, ABCA5, ABCA7, ABCB7, ABCC8, ABCC9, ABCD1, ABCD3, ACAD9, ACADM, ACADS, ACADSB, ACAT1, ACMSD, ACO2, ACOT7, ACOX1, ACSF3, ACSL4, ACTB, ACTG1, ACTL6B, ACVR1, ACY1, ADAM22, ADAR, ADAT3, ADCK3, ADCK4, ADD3, ADGRG1, ADK, ADNRP, ADPRHL2, ADRA2B, ADSL, AFF2, AFG3L2, AGA, AGGF1, AGK, AGTPBP1, AGTR2, AHDC1, AHI1, AHSG, AIFM1, AIMP1, AIMP2, AKT1, AKT2, AKT3, ALAD, ALDH18A1, ALDH1B1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALPL, ALX4, AMACR, AMER1, AMPD2, AMT, ANGPTL6, ANK2, ANKH, ANKLE2, ANKRD11, ANO10, ANO3, ANTXR1, AP1S2, AP2S1, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APOA1BP, APOPT1, APTX, AQP2, ARCN1, ARFGEF2, ARG1, ARHGAP31, ARHGDIA, ARHGEF15, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARID2, ARID4A, ARL13B, ARL6IP6, ARMIC9, ARNT2, ARSA, ARV1, ARVCF, ARX, ASAH1, ASCL1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASTN1, ASTN2, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATIC, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2A2, ATP2B3, ATP5A1, ATP5E, ATP6AP1, ATP6AP2, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B2, ATP7A, ATP8A2, ATPAF2, ATR, ATRIP, ATRX, ATXN10, AUH, AUTS2, AVPR2, B3GALNT2, B3GALT1, B4GALT1, B9D1, B9D2, BAZ1B, BAZ2B, BCAP31, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCS1L, BDNF, BMP2, BMP4, BOLA3, BPTF, BRAF, BRAT1, BRCC3, BRD4, BRPF1, BRWD3, BSCL2, BSN, BTBD, BUB1B, BUB3, C10orf2, C11ORF95, C12ORF57, C12ORF65, C19ORF70, C5orf42, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA2D1, CACNA2D2, CACNB4, CACNG2, CAD, CALM1, CAMK2A, CAMK2B, CAMKMT, CAMTA1, CARND, CARS2, CASK, CASP10, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC115, CCDC141, CCDC88A, CCDC88C, CCM2, CCND2, CCR1, CD96, CDC42, CDH11, CDH15, CDK10, CDK13, CDK5, CDK5RAP2, CDK6, CDKL5, CDKN1C, CDON, CECR1, CELF4, CENPE, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP57, CEP63, CERS1, CHAT, CHD1, CHD1L, CHD2, CHD4, CHD7, CHD8, CHKB, CHL1, CHRNA2, CHRNA4, CHRNA7, CHRNA2, CHRN2, CIC, CIT, CKAP2L, CLCF1, CLCN2, CLCN4, CLCNKA, CLCNKB, CLDN16, CLEC7A, CLIC2, CLIP1, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CNKSR2, CNNM2, CNOT3, CNPY3, CNTN2, CNTN4, CNTNAP1, CNTNAP2, COA3, COA5, COA7, COG1, COG2, COG4, COG5, COG6, COG7, COG8, COL13A1, COL18A1, COL4A1, COL4A2, COL4A3BP, COLGALT1, COMT, COPB2, COQ2, COQ4, COQ6, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CPA6, CPLX1, CPOX, CPS1, CPT1A, CPT2, CRADD, CRB2, CRBN, CREBBP, CRELD1, CRH, CRHR1, CRTAP, CRKL, CRLF1, CSF1R, CSMD1, CSNK2A1, CSNK2B, CSPPI, CSTB, CTBP1, CTC1, CTCF, CTH, CTLA4, CTNNB1, CTNND2, CTR9, CTSA, CTSD, CTSF, CTU2, CUL4B, CUX1, CUX2, CXCR4, CYC1, CYFIP2, CYP26C1, CYP27A1, CYP27B1, CYP2C9, CYP2R1, CYP2U1, D2HGDH, DAG1, DARS2, DBH, DBT, DCC, DCHS1, DCX, DDC, DDOST, DDX3X, DEAF1, DENND5A, DEPDC5, DGKD, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHX30, DIAPH1, DIP2B, DIP2C, DIS3L2, DISP1, DLAT, DLD, DLG1, DLG2, DLG3, DLG4, DLGAP2, DLL1, DLL4, DMBX1, DMD,

DNA2, DNAJC12, DNAJC19, DNAJC5, DNAJC6, DNASE1L3, DNM1, DNM1L, DNMT3A, DOCK6, DOCK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DSCAM, DSTYK, DYNC1H1, DYRK1A, EARS2, EBF3, EBP, ECE1, ECHS1, ECM1, EDC3, EEF1A2, EFHC1, EFHC2, EFTUD2, EGF, EHHADH, EHMT1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, ELMO2, ELOVL4, ELP4, EMC1, EMG1, EML1, EMX2, EOGT, EP300, EPB41L1, EPG5, EPM2A, EPRS, EPT1, ERBB2IP, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERF, ERLIN2, ERMARD, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EXOC6B, EXOSC3, EXOSC8, EXT2, EXTL3, EZH2, FA2H, FADD, FAM111A, FAM126A, FAM183A, FAM58A, FAR1, FARS2, FARSB, FASLG, FASN, FASTKD2, FAT4, FBP1, FBXL4, FBXO11, FBXO28, FBXO31, FDXR, FGD1, FGF12, FGF13, FGF23, FGF8, FGFR1, FGFR2, FGFR3, FGFR1L, FH, FIG4, FKR1, FKTN, FLII, FLNA, FLRT3, FLVCR1, FLVCR2, FMN2, FMR1, FOLR1, FOXG1, FOXH1, FOXP1, FOXRED1, FRMPD4, FRRS1L, FTL, FTO, FTSJ1, FUCA1, FUK, FXR1, G6PC, GABBR2, GABRA1, GABRA3, GABRA5, GABRA6, GABRB1, GABRB2, GABRB3, GABRD, GABRE, GABRG2, GABRG3, GABRQ, GABRR1, GABRR3, GAD1, GAL, GALT, GAMT, GAS1, GATA3, GATA6, GATAD2B, GATM, GBA, GCDH, GCH1, GCM2, GCSH, GDI1, GDNF, GEMIN4, GFAP, GFER, GFM1, GFM2, GJA1, GJC2, GK, GLB1, GLDC, GLE1, GLI2, GLI3, GLRA1, GLRB, GLRX5, GLS, GLTSCR1, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAI1, GNAO1, GNAQ, GNAS, GNB1, GNB5, GNE, GNPTAB, GNPTG, GNRH1, GNS, GOLGA2, GOSR2, GP9, GPAA1, GPC3, GPC4, GPHN, GPR161, GPSM2, GPT2, GRIA2, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GRM7, GRM8, GRN, GSPT2, GSS, GTF2E2, GTF2H5, GTF2IRD1, GTPBP2, GTPBP3, GUCY1A3, GUF1, GUSB, GYS1, GYS2, H3F3A, H3F3B, HACE1, HADH, HARS2, HAX1, HCCS, HCFC1, HCN1, HCN2, HDAC1, HDAC4, HDAC8, HECW2, HEPACAM, HERC1, HERC2, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HIC1, HIP1, HIRA, HIVEP2, HK1, HLCS, HMBS, HMGCL, HMGCS2, HNRNP2, HNRNP1, HNRNP3, HNRNP4, HOXA1, HPD, HRAS, HSD17B10, HSD17B4, HSPD1, HTR1A, HTRA2, HUWE1, HYLS1, IARS, IBA57, ICK, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT122, IFT140, IL10, IL12A, IL12B, IL12RB2, IL17F, IL17RA, IL1RAPL1, IL23R, IL27RA, ILF2, IMPAD1, INO80, INPP4A, INPP5E, INPP5K, INS, INSR, INTS6, INVS, IQCB1, IQSEC1, IQSEC2, IRF2BPL, ISCU, ISG15, ISPD, ITGB6, ITPA, ITPR1, IVD, JAM3, JMJD1C, JNK, KANK1, KANS1, KARS, KAT2B, KAT6A, KAT6B, KATNB1, KCNA1, KCNA2, KCNAB1, KCNAB2, KCNB1, KCNC1, KCNC3, KCND2, KCND3, KCNH1, KCNJ1, KCNJ10, KCNJ11, KCNJ2, KCNJ6, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCNV2, KCTD3, KCTD7, KDM2B, KDM5B, KDM5C, KDM6A, KIAA0226, KIAA0556, KIAA0586, KIAA0753, KIAA1033, KIAA1109, KIAA1279, KIAA1715, KIAA2022(NEXMIF), KIF11, KIF1A, KIF2A, KIF4A, KIF5A, KIF5C, KIF7, KIRREL3, KISS1, KLF13, KLHL15, KLHL7, KLLN, KLRC4, KMT2A(MLL), KMT2C, KMT2D, KPNA7, KPTN, KRAS, KRIT1, L1CAM, L2HGDH, LAGE3, LAMA2, LAMB1, LAMC3, LARGE, LARS, LARS2, LAS1L, LBR, LEO1, LETM1, LGI1, LGI4, LHX1, LIAS, LIG4, LIMK1, LINGO1, LIPT1, LMAN2L, LMBRD1, LMNB2, LONP1, LRFN2, LRP2, LRPPRC, LYRM7, LYST, MACF1, MAF, MAFB, MAGEL2, MAGI2, MAN1B1, MANBA, MAOA, MAP1B, MAP2K1, MAP2K2, MAPK1, MAPK10, MAPK3, MAPRE2, MARS2, MAT1A, MATN4, MBD5, MBOAT7, MBTPS2, MC2R, MCCC1, MCCC2, MCEE, MCM4, MCOLN1, MCPH1, MCTP2, MDH2, ME2, MECP2, MED12, MED13, MED13L, MED17, MED23, MED25, MED27, MEF2C, MEGF10, MEIS2, METTL23, MFF, MFHAS1, MFSD2A, MFSD8, MGAT2, MGME1, MGP, MICAL1, MID1, MID2, MIPEP, MKS1, MLC1, MLLT3, MLX, MLYCD, MMAA,

MMACHC, MMADHC, MN1, MOCS1, MOCS2, MOGS, MPC1, MPDU1, MPDZ, MPLKIP, MPP7, MPV17, MRAP, MRI1, MRPL44, MRPS16, MRPS22, MSL3, MSX2, MTFMT, MTHFS, MTO1, MTOR, MTPAP, MTR, MTRR, MUT, MVK, MYH1, MYH3, MYO1H, MYO5A, MYO7B, MYO9A, MYOCD, MYT1L, NAA10, NAA15, NACC1, NADK2, NAGA, NAGLU, NAGS, NALCN, NANS, NARS2, NAT8L, NBAS, NBEA, NCOR1, NDE1, NDN, NDP, NDST1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEDD4L, NEK9, NELFA, NEU1, NEUROD2, NF1, NFASC, NFIA, NFIB, NFIX, NFU1, NGLY1, NHEJ1, NHLRC1, NID1, NIN, NIPA1, NIPA2, NIPBL, NKX6-2, NLGN1, NLGN4X, NMNAT1, NNT, NOL3, NONO, NOTCH3, NPAP1, NPC1, NPC2, NPHP1, NPHP3, NPRL2, NPRL3, NR2F1, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, NTNG1, NTRK2, NUBPL, NUP107, NUP133, NUS1, OCLN, OCRL, OFD1, OGT, OPA1, OPA3, OPHN1, OSGEP, OSTM1, OTC, OTUD6B, OTX2, PACS1, PACS2, PAFAH1B1, PAH, PAK3, PANK2, PARS2, PAX2, PAX6, PBX1, PC, PCCA, PCCB, PCDH12, PCDH19, PCDH7, PCDHB4, PCK1, PCLO, PCNT, PDCD10, PDE10A, PDE4D, PDE6D, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PHC1, PHF21A, PHF3, PHF6, PHGDH, PI4KA, PIBF1, PIEZO2, PIGA, PIGC, PIGG, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGV, PIGW, PIGY, PIK3AP1, PIK3CA, PIK3R2, PLA2G6, PLAA, PLAGL1, PLCB1, PLEKHG2, PLK4, PLP1, PLXND1, PMM2, PMPCA, PNKD, PNKP, PNPLA8, PNPO, PNPT1, PODXL, POGZ, POLA1, POLE, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU1F1, PPA2, PPM1B, PPM1D, PPM1K, PPOX, PPP1R15B, PPP1R3C, PPP2CA, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM8, PRDX1, PREPL, PRF1, PRICKLE1, PRICKLE2, PRIMA1, PRKAR1A, PRKDC, PRKG2, PRMT7, PRODH, PROP1, PROSC, PRPS1, PRRT2, PRSS12, PRTN3, PRUNE, PSAP, PSAT1, PSMD12, PSPH, PTCH1, PTCHD1, PTEN, PTF1A, PTH, PTPN11, PTPN22, PTPN23, PTPN4, PTPRD, PTS, PUF60, PUM1, PURA, PUS1, PVRL1, PYCR2, QARS, QDPR, QRIC1, RAB11A, RAB11B, RAB18, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAC1, RAD21, RAF1, RAI1, RALGAPB, RANBP2, RAP1A, RAP1B, RARS2, RASGRP1, RBBP8, RBFOX1, RBFOX3, RBM10, RBM8A, RBPJ, RECQL4, REEP1, RELA, RELN, RERE, RFT1, RGR, RHOBTB2, RILP, RIT1, RLBP1, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF13, RNF168, ROBO1, ROBO3, ROGDI, RORA, RORB, RPGRIP1L, RPIA, RPL10, RPS20, RPS6KA3, RREB1, RRM2B, RTN4IP1, RTTN, RYR2, RYR3, SACS, SAMD9, SAMHD1, SARDH, SARS2, SASS6, SATB2, SC5D, SCARB2, SCN10A, SCN1A, SCN1B, SCN2A, SCN3A, SCN7A, SCN8A, SCNM1, SCO1, SCO2, SDCCAG8, SDHA, SDHAF1, SEC24C, SEC24D, SEPSECS, SERAC1, SERPINI1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SGCE, SGSH, SH2B1, SHANK1, SHANK3, SHH, SHROOM4, SIK1, SIN3A, SIX3, SKI, SLC12A1, SLC12A2, SLC12A3, SLC12A5, SLC12A6, SLC13A5, SLC16A1, SLC16A2, SLC17A5, SLC18A2, SLC19A2, SLC19A3, SLC1A1, SLC1A2, SLC1A3, SLC1A4, SLC20A2, SLC22A5, SLC25A1, SLC25A10, SLC25A12, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC26A1, SLC2A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC37A4, SLC39A8, SLC45A1, SLC46A1, SLC4A10, SLC5A7, SLC6A1, SLC6A17, SLC6A19, SLC6A5, SLC6A8, SLC7A5, SLC7A7, SLC9A1, SLC9A6, SLC9A9, SMAD2, SMAD4,

SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMC1A, SMC3, SMCHD1, SMG9, SMPD1, SMS, SNAP25, SNAP29, SNIP1, SNRPN, SNX14, SNX27, SON, SOS1, SOX10, SOX11, SOX2, SOX3, SOX5, SPAST, SPATA5, SPEN, SPG11, SPG20, SPINK5, SPR, SPRED1, SPTAN1, SPTBN4, SQSTM1, SRCAP, SRD5A3, SRPX2, SRSF11, SSR4, SSTR5, ST3GAL3, ST3GAL5, ST5, ST7, ST8SIA2, STAG1, STAG2, STAMBP, STAR, STARD9, STAT1, STAT4, STIL, STRA6, STRADA, STT3A, STT3B, STUB1, STX11, STX16, STX1B, STXBP1, STXBP2, STYXL1, SUCLA2, SUCLG1, SUCO, SUMF1, SUOX, SURF1, SUV420H1, SV2A, SYN1, SYN2, SYNGAP1, SYNJ1, SYP, SYT14, SYT2, SZT2, TAC3, TACO1, TAF1, TANGO2, TAOK2, TAT, TAZ, TBC1D20, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBR1, TBX1, TBX18, TBX19, TCF4, TCIRG1, TCTN1, TCTN2, TCTN3, TDGF1, TDP2, TECPR2, TECR, TFAP2A, TGDS, TGFBR3, TGIF1, TH, THAP11, THOC2, TIMM50, TIMM8A, TIMMDC1, TK2, TKT, TLK2, TMEM106B, TMEM126B, TMEM138, TMEM165, TMEM216, TMEM231, TMEM237, TMEM240, TMEM5, TMEM67, TMEM70, TMLHE, TMTC3, TNFRSF11A, TNKS, TNRC6B, TOE1, TP53RK, TPK1, TPP1, TPRKB, TRAF3IP1, TRAF3IP2, TRAF7, TRAK1, TRAPPC11, TRAPPC2L, TRAPPC6B, TRAPPC9, TREX1, TRIM3, TRIM8, TRIO, TRIP12, TRIP13, TRIT1, TRMT1, TRMT10A, TRNT1, TRPM6, TRPS1, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TSHZ3, TSPAN7, TTC19, TUBA1A, TUBA3E, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TUFM, TUSC3, TWIST1, TXN2, TYMP, TYROBP, UBA5, UBAC2, UBB, UBE2A, UBE3A, UBN2, UBR5, UBTF, UFC1, UFD1L, UGDH, UMPS, UNC13D, UNC80, UPB1, UPF3B, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, USP15, USP18, USP7, USP9X, VAMP2, VANG1, VARS, VARS2, VCP, VDR, VLDLR, VPS11, VPS13A, VPS13B, VPS53, VRK1, WAC, WARS2, WDR19, WDR26, WDR4, WDR45, WDR45B, WDR62, WDR73, WDR81, WFS1, WHSC1, WIPI2, WWOX, XK, XPA, XPNPEP3, XPR1, XRCC1, YAP1, YWHAE, YWHAG, ZBTB18, ZBTB20, ZC3H14, ZC4H2, ZDHHC15, ZDHHC9, ZEB2, ZFP57, ZFYVE26, ZIC2, ZMYND11, ZMYND12, ZNF335, ZNF41, ZNF423, ZNF592, ZNF674, ZNF711, ZNF81, ZNHIT3, ZSWIM6.

Forrás:

Congenital Hypotonia Xpanded Panel

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(Megtekintve: 2022.02.14.)