

Veleszületett hypotonia (Congenital hypotonia) génpanel v:2023.10.12.

Panel tartalma 1422 gén:

A2ML1, AAAS, AARS, AASS, ABAT, ABCB7, ABCC8, ABCC9, ABCD4, ABHD11, ACACA, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT2, ACO2, ACOX1, ACP2, ACP6, ACTA1, ACTB, ACTG1, ACTG2, ACY1, ACSL4, ADAMTS2, ADAP2, ADAR, ADAT3, ADCK3, ADCY5, ADCY6, ADGRG1, ADK, ADNP, ADPGK, ADSL, AFF2, AFF4, AFG3L2, AGA, AGK, AGO1, AGRN, AGTPBP1, AHCY, AHDC1, AHI1, AHSB, AIFM1, AIFM3, AIMP1, AK9, AKT1, AKT3, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALPL, AMER1, AMMECR1, AMPD1, AMPD2, AMT, ANK3, ANKRD11, ANOS1, ANTXR2, AP1S1, AP1S2, AP3B2, AP3D1, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APOA1BP, APOPT1, AR, ARFGEF2, ARHGAP11B, ARHGAP31, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARMC9, ARSA, ARV1, ARVCF, ARX, ASAH1, ASCC1, ASCL1, ASH1L, ASNS, ASPA, ASPM, ASXL1, ASXL2, ASXL3, ATAD3A, ATCAY, ATIC, ATL1, ATM, ATP10A, ATP1A2, ATP1A3, ATP2B3, ATP5A1, ATP6V0A2, ATP6V1A, ATP6V1B2, ATP6V1E1, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRX, AUTS2, B3GALNT2, B3GALT6, B3GAT3, B4GALT1, B4GALT7, B4GAT1, B9D1, BAZ1B, BBS1, BCAP31, BCKDHA, BCKDHB, BCL11A, BCL11B, BCOR, BCS1L, BDNF, BICD2, BIN1, BLK, BMP1, BMP2, BMP4, BMPER, BOLA, BPTF, BRAF, BRAT1, BRPF1, BRWD3, BSCL2, BSND, BTBD, BUB1B, BUB3, C10orf2, C11orf73, C12orf4, C12orf57, C12orf65, C21orf2, C5orf42, CA8, CACNA1A, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1S, CAD, CAMK2B, CAMKMT, CAMTA1, CANT1, CAPN3, CARS2, CASK, CASR, CBL, CC2D2A, CCDC115, CCDC141, CCDC174, CCDC22, CCDC47, CCDC78, CCDC88A, CCNF, CD59, CD96, CDC42, CDH23, CDK13, CDKL5, CDKN1C, CDRT15, CEP104, CEP120, CEP290, CEP41, CEP57, CFL2, CFTR, CHAMP1, CHAT, CHD1, CHD2, CHD3, CHD4, CHD7, CHD8, CHKB, CHMP1A, CHRNA1, CHRNA7, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CKAP2L, CLCN4, CLCN7, CLCNKA, CLCNKB, CLIP2, CLP1, CLPB, CLTC, CNKSR2, CNTN1, CNTNAP1, CNTNAP2, COA6, COA7, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL12A1, COL13A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A3BP, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COLQ, COMT, COQ4, COQ7, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX8A, CPLX1, CPS1, CPT1A, CPT2, CREBBP, CRYAB, CTBP1, CTCF, CTNNB1, CTNND2, CTNS, CTU2, CUL4B, CWF19L1, CYB5R3, CYFIP2, CYP24A1, CYP27B1, CYP2R1, CSNK2A1, CSNK2B, CSPP1, CSTB, D2HGDH, DAG1, DAO, DARS, DBT, DCC, DCHS1, DCPS, DCX, DDC, DDOST, DDR2, DDX11, DDX3X, DEAF1, DEPDC5, DGAT1, DGCR14, DGCR2, DGCR6, DGCR8, DGUOK, DHCR7, DHDDS, DHFR, DHTKD1, DHX30, DIAPH1, DIS3L2, DKK1, DLAT, DLD, DLG3, DLG4, DMD, DNA2, DNAJB2, DNAJC12, DNAJC19, DNAJC21, DNM1, DNM2, DNMT3A, DOCK6, DOK7, DOLK, DPAGT1, DPF2, DPH1, DPM1, DPM2, DPM3, DPYD, DSE, DST, DUOX2, DUOXA2, DYNC1H1, DYRK1A, EARS2, EBF3, EBP, ECHS1, EDN1, EDN3, EDNRB, EEF1A2, EFEMP2, EFN1, EFTUD1, EFTUD2, EGR2, EHMT1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELAC2, ELN, ELP2, EMC1, EMD, EP300, EPCAM, EPG5,

EPHA4, ERCC1, ERCC2, ERCC5, ERCC6, ERCC6L2, ERCC8, ERF, ERMARD, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXOSC9, EXT2, EXTL3, EZH2, FAM126A, FAM134B, FANCG, FAR1, FARS2, FARSB, FASTKD2, FAT4, FBN1, FBN2, FBP1, FBXL4, FBXO11, FDX1L, FDXR, FEZF1, FGF12, FGF17, FGF8, FGFR1, FGFR2, FGFR3, FGFR1L, FH, FHL1, FIG4, FKBP14, FKRP, FKTN, FLAD1, FLG, FLII, FLNA, FLNB, FLRT3, FLVCR1, FMN2, FMR1, FOXC1, FOXE1, FOXG1, FOXP1, FOXP2, FOXRED1, FRG1, FRRS1L, FUCA1, G6PD, GAA, GABRA1, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GALC, GALE, GALNS, GAMT, GAN, GARS, GATA1, GATA4, GATA6, GATAD2B, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDI1, GDNF, GFAP, GFER, GFM1, GFPT1, GJA1, GJB2, GJC2, GK, GLB1, GLDC, GLE1, GLRA1, GLRA4, GLRX5, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNA11, GNAI3, GNAO1, GNAS, GNB1, GNB5, GNE, GNPAT, GNPTAB, GORAB, GP1BB, GPAA1, GPC3, GPC4, GPHN, GPT2, GPX4, GRIA3, GRIA4, GRID2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM1, GTF2I, GTF2IRD1, GTPBP2, GTPBP3, GUCY2D, GUSB, HACE1, HADH, HADHA, HADHB, HBA1, HBA2, HCFC1, HCN1, HDAC4, HDAC8, HECW2, HEPACAM, HERC1, HERC2, HESX1, HEXA, HFE, HIBCH, HINT1, HIRA, HIST1H1E, HIVEP2, HLCS, HMBS, HMGA2, HMGCS2, HNF1A, HNF4A, HNRNPH2, HNRNPK, HNRNPU, HPD, HPRT1, HRAS, HS6ST1, HSD17B10, HSD17B4, HSPB1, HSPB8, HSPD1, HTRA2, HUWE1, HYLS1, IARS, IBA57, IDH2, IDUA, IER3IP1, IFIH1, IFT140, IFT81, IGF2, IGHMBP2, IKBKAP, IL17RD, IL1RAPL1, INPP5E, INPP5K, INPPL1, INS, INSR, IQCB1, IQSEC2, ISCA2, ISCU, ISPD, ITGA3, ITGA7, ITGB6, ITPA, ITPR1, IYD, JAG1, JMJD1C, KANK1, KANSL1, KAT6A, KAT6B, KBTBD13, KCNA2, KCNAB2, KCNB1, KCNC1, KCNC3, KCNE1L, KCNH1, KCNJ10, KCNJ11, KCNJ2, KCNK9, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCTD7, KDM1A, KDM5C, KDM6A, KIAA0196, KIAA0556, KIAA0586, KIAA0753, KIAA1109, KIAA1279, KIAA2022, KIDINS220, KIF11, KIF1A, KIF21A, KIF22, KIF2A, KIF5A, KIF5C, KIF7, KISS1R, KLF13, KLHL40, KLHL41, KLHL7, KMT2B, KMT2C, KMT2D, KPTN, KRAS, L1CAM, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LAMB2, LAMP2, LARGE, LARP7, LARS, LAS1L, LEMD3, LETM1, LGI4, LHX3, LHX4, LIAS, LIFR, LIMK1, LIMS2, LINS, LIPT1, LMBR1, LMBRD1, LMNA, LMOD3, LMX1B, LONP1, LPL, LRP4, LRP5, LRPPRC, LTBP4, LTC4S, LZTR1, LYRM4, LYRM7, MAF, MAG, MAGEL2, MALL, MAN1B1, MAN2B1, MANBA, MAP2K1, MAP2K2, MAP3K7, MAPRE2, MARS2, MBD5, MBOAT7, MBTPS2, MCCC1, MCCC2, MCM3AP, MCOLN1, MECAP2, MED12, MED13, MED13L, MED25, MEF2C, MEFV, MEGF10, MEIS2, METTL23, MFF, MFN2, MFSD2A, MFSD8, MGAT2, MICU1, MIPEP, MKS1, MLL, MLXIPL, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOGS, MORC2, MPC1, MPDU1, MPI, MPLKIP, MPV17, MPZ, MRE11A, MRPS16, MRPS22, MRPS34, MSTO1, MTFMT, MTM1, MTMR14, MTO1, MTOR, MTR, MTRR, MTPP, MUSK, MUT, MVK, MYBPC3, MYCN, MYF6, MYH3, MYH7, MYL2, MYO18B, MYO1H, MYO5A, MYO7A, MYO9A, MYOT, MYPN, MYT1L, NAA10, NAA15, NACC1, NADK2, NAGA, NALCN, NANS, NARS2, NAT8L, NBAS, NDN, NDNL2, NDP, NDST1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA9, NDUFAF2, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NECAP1, NEDD4L, NEFL, NEK1, NELFA, NEU1, NF1, NFIA, NFIB, NFIX, NGLY1, NHLRC2, NHS, NIPBL, NKX2-1, NKX2-5, NKX6-2, NLRC4, NMNAT1, NONO, NOTCH3, NPAP1, NPC1, NPC2, NPHP1, NPHS1, NPRL3, NR2F1, NR2F2, NR3C2, NRAS, NRXN1, NSD1, NSDHL, NSMF, NSUN2, NTRK2, NUBPL, NUP107, NUP62,

NUS1, OCLN, OCRL, OFD1, OGDH, OPA1, OPHN1, ORAI1, OSGEP, OSTM1, OTUD6B, OTX2, P4HB, PACS1, PACS2, PAFAH1B1, PAH, PAM16, PANK2, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH19, PCLO, PCYT1A, PDE10A, PDE4D, PDE6D, PDHA1, PDHB, PDHX, PDP1, PDSS2, PDX1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGM3, PHF21A, PHF6, PHGDH, PHIP, PHKA2, PHKB, PHKG2, PHOX2B, PIBF1, PIEZO2, PIGA, PIGG, PIGH, PIGL, PIGN, PIGO, PIGS, PIGT, PIGV, PIGY, PIK3CA, PIK3R1, PIK3R2, PKD1, PKHD1, PLA2G6, PLAA, PLAGL1, PLCB1, PLCB4, PLEKHG2, PLEKHG5, PLOD1, PLP1, PLXND1, PMM2, PMP22, PMPCA, PNKP, PNP, PNPLA2, PNPLA6, PNPLA8, PNPO, PNPT1, POC1A, POGZ, POLG, POLG2, POLR3A, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PON1, PON2, PON3, POU1F1, PPA2, PPARGC1A, PPM1B, PPM1D, PPP1CB, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM12, PRDM16, PRDM5, PRDX1, PREPL, PRF1, PRKAR1A, PRKCG, PRKD1, PRMT7, PRODH, PROK2, PROKR2, PROP1, PROSC, PRPH, PRPS1, PRRT2, PRUNE, PRX, PSAP, PSAT1, PSMD12, PTCH1, PTCHD1, PTDSS1, PTEN, PTPLA, PTPN11, PTRF, PTRH2, PTS, PUF60, PURA, PUS1, PUS3, PYCR1, PYCR2, PYGM, PYROXD1, QARS, QDPR, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RAC1, RAD21, RAF1, RAI1, RAP1A, RAP1B, RAPSN, RARB, RARS, RARS2, RASA2, RBM10, REEP1, RELN, RERE, RET, REV3L, RFC2, RFT1, RHOBTB2, RIN2, RIT1, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNF125, RORA, RPGRIP1L, RPL10, RPS23, RPS6KA3, RRAS, RREB1, RRM2B, RSPRY1, RUSC2, RYR1, RYR2, SACS, SALL1, SAMD9, SAMHD1, SARS2, SATB2, SBDS, SC5D, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SDHD, SEC24C, SEC24D, SEMA3A, SEMA3E, SEMA5A, SEPN1, SEPSECS, SERAC1, SERPINA1, SERPINH1, SET, SETBP1, SETD1B, SETD2, SETD5, SETX, SFXN4, SGCA, SGCB, SGCD, SGCE, SGCG, SGPL1, SGSH, SH2B1, SH3TC2, SHANK2, SHANK3, SHOC2, SIGMAR1, SIK1, SIL1, SIM1, SIN3A, SIX3, SKI, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC18A2, SLC18A3, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A24, SLC25A26, SLC25A3, SLC25A4, SLC25A46, SLC26A2, SLC26A4, SLC29A3, SLC2A1, SLC2A10, SLC30A9, SLC33A1, SLC34A1, SLC34A3, SLC35A2, SLC35A3, SLC35C1, SLC39A14, SLC39A8, SLC3A1, SLC45A1, SLC46A1, SLC52A2, SLC52A3, SLC5A5, SLC5A7, SLC6A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SLC9A6, SMAD2, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMG9, SMN1, SMPD1, SMS, SNAP25, SNAP29, SNIP1, SNRPN, SNX14, SON, SOS1, SOS2, SOX10, SOX11, SOX2, SOX5, SOX9, SPARC, SPAST, SPATA5, SPECC1L, PEG, SPG20, SPR, SPRED1, SPRY4, SPTAN1, SPTBN2, SPTBN4, SRCAP, SRD5A3, SRP54, SSR4, ST3GAL3, ST3GAL5, STAMBP, STAT1, STAT3, STIM1, STRA6, STRADA, STT3A, STT3B, STX11, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SUV420H1, SYNE1, SYNE2, SYNGAP1, SYNJ1, SYT1, SYT2, SZT2, TAB2, TACO1, TACR3, TAF1, TAF6, TANGO2, TARS2, TAZ, TBC1D20, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBL2, TBR1, TBX1, TBX4, TCAP, TCF20, TCF4, TCTN1, TCTN2, TCTN3, TDP2, TECPR2, TELO2, TFAP2B, TG, TGFB3, TGFBR1, TGFBR2, TH, THOC2, THRA, THRB, TIMM50, TK2, TKT, TLK2, TMCO1, TMEM106B, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM5, TMEM67, TMEM70, TMEM8C, TNFRSF11A, TNNT1, TNPO3, TNXB, TOE1, TOR1A, TOR1AIP1, TP53RK, TP63, TPI1, TPK1, TPM2, TPM3, TPO, TPRKB, TRAF7,

TRAK1, TRAPPC11, TRAPPC9, TREX1, TRHR, TRIM2, TRIM32, TRIM37, TRIO, TRIP12, TRIP13, TRIP4, TRMT1, TRMT10C, TRMT5, TRMU, TRNT1, TRPS1, TRPV4, TSC1, TSEN15, TSEN2, TSEN54, TSFM, TSHB, TSHR, TTN, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUFM, TXN2, TYMP, UBA1, UBA2, UBA5, UBE2A, UBE3A, UBE3B, UBR1, UBTF, UFD1L, UNC13A, UNC80, UPB1, UPF3B, UQCC2, UQCC3, UQCRQ, USP18, USP7, USP9X, VAC14, VAMP1, VAPB, VARS2, VDR, VIPAS39, VLDLR, VMA21, VPS11, VPS13B, VPS13D, VPS33B, VPS53, VRK1, WAC, WARS2, WDR11, WDR26, WDR4, WDR45, WDR62, WDR73, WDR81, WFS1, WHSC1, WNK1, WNT10A, WWOX, XYLT1, XYLT2, YARS2, YME1L1, YWHAE, YWHAG, YY1, ZBTB18, ZBTB20, ZC4H2, ZDHHC15, ZDHHC9, ZEB2, ZFP57, ZIC1, ZMYND11, ZNF148, ZNF423, ZNF592, ZNHIT3, ZSWIM6

Forrás:

Congenital Hypotonia Xpanded Panel

<https://www.genedx.com/tests/detail/congenital-hypotonia-xpanded-panel-1126> (Megtekintve: 2023.05.18.)