

Centrum för Medfödda Metabola Sjukdomar

Giltig från 2023-12-21

Genpanel MOVE (Movement disorders), version 20 – 431 gener

Gener kopplade till medfödda rörelsесjukdomar hos barn eller andra monogena sjukdomar med liknande klinisk bild

AAAS, AARS1, AARS2, ABAT, ABCB7, ABCD1, ABHD12, ACER3, ACOX1, ACP5, ACTB, ADAR, ADCY5, ADD3, ADPRS, AFG2A, AFG2B, AFG3L2, AGTPBP1, AH11, ALDH18A1, ALDH3A2, ALDH6A1, ALG6, ALS2, AMPD2, ANO10, ANO3, AOPEP, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARID1B, ARL13B, ARL6IP1, ARSA, ARV1, ARX, ATAD3A, ATCAY, ATG7, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP5F1B, ATP5MC3, ATP6AP2, ATP7A, ATP7B, ATP8A2, ATXN10, ATXN2, ATXN3, ATXN7, AUH, B4GALNT1, B4GALT1, BCAP31, BCS1L, BICD2, BSCL2, C19orf12, CA8, CACNA1A, CACNA1B, CACNA1E, CACNA1G, CACNA2D2, CACNB4, CAMK2A, CAMTA1, CAPN1, CARS2, CASK, CCT5, CDKL5, CEP104, CHMP1A, CLCN2, CLN3, CLN5, CLP1, CLTC, CNTNAP2, COA7, COASY, COG4, COG5, COL6A3, COQ2, COQ8A, COX10, COX15, COX20, CP, CPT1C, CSF1R, CSPPI, CSTB, CTBP1, CTC1, CTDP1, CTNNB1, CWF19L1, CYP27A1, CYP2U1, CYP7B1, DARS2, DCAF17, DDC, DDHD1, DDHD2, DEGS1, DHCR24, DHDDS, DHFR, DKC1, DLAT, DLD, DNAJC12, DNAJC19, DNAJC5, DNAJC6, DNM1, DNM1L, DNM2, DNMT1, DSTYK, EARS2, EBF3, ECHS1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, ELOVL1, ELOVL4, EMC1, ENTPD1, ERLIN1, ERLIN2, EXOSC9, FA2H, FAM126A, FAR1, FARS2, FBXL4, FBXO7, FBXW11, FGF14, FLVCR1, FOXG1, FRRS1L, FTL, FUS, FXN, GABBR2, GABRA2, GAD1, GALC, GAMT, GAN, GATM, GBA, GBA2, GCDH, GCH1, GFAP, GJA1, GJB1, GJC2, GLB1, GLRA1, GLRB, GM2A, GNAL, GNAO1, GNB1, GOSR2, GPA1, GPT2, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRM1, GTPBP2, HARS1, HEPACAM, HEXA, HIBCH, HPCA, HPDL, HPRT1, HSPD1, HTRA2, HTT, IARS1, IBA57, IFIH1, IMPDH2, INPP5E, INPP5K, IREB2, ITPR1, JAM3, KANK1, KCNA1, KCNA2, KCNC3, KCND3, KCNMA1, KCNN2, KCNQ2, KCTD17, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KMT2B, KPNA3, LICAM, L2HGDH, LAMA1, LIPT1, LIPT2, MADD, MAG, MAPK8IP3, MARS2, MCOLN1, MDH2, MECP2, MECR, MICU1, MLC1, MRE11, MSTO1, MTFMT, MTRFR, MYORG, NBEA, NDUFA12, NDUFAF2, NDUFAF6, NDUFS1, NDUFS8, NEFL, NFASC, NIPA1, NKX2-1, NKX6-2, NPC1, NPC2, NT5C2, NUP62, OPA3, OPHN1, PANK2, PARK7, PCDH12, PCYT2, PDE10A, PDE2A, PDGFB, PDGFRB, PDHA1, PEX10, PEX2, PEX7, PGAP1, PGK1, PGM3, PINK1, PLA2G6, PLEKHG2, PLP1, PMPCA, PNKD, PNKP, PNPLA6, POLG, POLR1C, POLR3A, POLR3B, POU4F1, PPP2R2B, PRDX3, PRICKLE1, PRKCG, PRKN, PRKRA, PRPS1, PRRT2, PSAP, PTS, QDPR, RAB39B, RAC1, RARS1, REEP1, REEP2, RELN, RHOBTB2, RNASEH2B, RNASET2, RNF170, RNF216, RORA, RTN2, RUBCN, SACS, SAMD9L, SAMHD1, SCN2A, SCN8A, SCP2, SCYL1, SELENO1, SEPSECS, SERAC1, SETX, SGCE, SIGMAR1, SIL1, SLC16A2, SLC18A2, SLC19A3, SLC1A2, SLC1A3, SLC20A2, SLC25A15, SLC2A1, SLC30A10, SLC30A9, SLC33A1, SLC39A14, SLC6A3, SLC6A5, SLC6A8, SLC9A1, SLC9A6, SMPD1, SNAP25, SNCA, SNORD11B, SPART, SPAST, SPG11, SPG21, SPR, SPTAN1, SPTBN2, SPTLC1, SPTLC2, SQSTM1, STAMB, STUB1, STXBP1, SUMF1, SURF1, SYNE1, SYNJ1, SYT1, TACO1, TAF1, TBC1D24, TBP, TECPR2, TENM4,

TFG, TGM6, TH, THAP1, THG1L, TIMM8A, TMEM240, TMEM63C, TOE1, TOR1A, TPK1, TPP1, TRAPPC11, TREXI, TSEN54, TSPOAP1, TTBK2, TTC19, TTPA, TUBB4A, TWNK, UBA5, UBAP1, UBE3A, UBQLN2, UCHL1, VAC14, VAMP1, VAMP2, VLDR, VPS11, VPS13A, VPS13B, VPS13C, VPS13D, VPS16, VPS37A, VPS41, WARS2, WASHC5, WDR45, WDR73, WDR81, WFS1, WWOX, XPR1, YY1, ZFYVE26, ZNF142.