

## Genpanel avseende hjärnmissbildningar v4.0 551 gener

AARS1, AARS2, ABCD1, ACOX1, ACTB, ACTG1, ADAR, ADGRG1, ADPRS, AIFM1, AIMP1, AIMP2, AKT1, AKT2, AKT3, ALDH3A2, AMPD2, ANKLE2, ANKRD11, AP4B1, AP4E1, AP4M1, AP4S1, APC2, ARF1, ARFGF2, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARMC9, ARSA, ARX, ASNS, ASPA, ASPM, ASXL1, ASXL2, ASXL3, ATP1A2, ATP1A3, ATP6V0A2, ATR, B3GALNT2, B4GAT1, B9D1, B9D2, BCAP31, BLTP1, BOLA3, BRAF, BRWD3, C2CD3, CARS2, CASK, CC2D2A, CCDC47, CCDC88C, CCND2, CDH2, CDK13, CDK5, CDK5RAP2, CDK6, CDKN1C, CDON, CENPE, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP290, CEP41, CEP63, CEP85L, CHD3, CHD4, CHD7, CHD8, CHMP1A, CIT, CLCN2, CLP1, COA7, COA8, COASY, COL4A1, COL4A2, COX10, COX15, COX6B1, CPLANE1, CRADD, CRIPTO, CRPPA, CSF1R, CSNK2A1, CSPP1, CTC1, CTNNA2, CTU2, CUL4B, CYP27A1, D2HGDH, DAG1, DARS1, DARS2, DCHS1, DCX, DDX3X, DEGS1, DEPDC5, DHCR24, DIS3L2, DNMT3A, DONSON, DPF2, DPYSL5, DYNC1H1, DYRK1A, EARS2, EED, EFTUD2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EMC1, EML1, EMX2, EPRS1, ERCC6, ERCC8, ERMARD, EXOSC3, EXOSC8, EXOSC9, EYA1, EZH2, FA2H, FARS2, FAT4, FDX2, FGF8, FGFR1, FH, FIBP, FKRP, FKTN, FLNA, FLVCR2, FOLR1, FOXG1, FOXH1, FOXRED1, FUCA1, GALC, GAN, GBE1, GCDH, GEMIN4, GFAP, GFMI, GJC2, GLA, GLBI, GLI2, GLI3, GLRX5, GMPPB, GPC3, GPSM2, GPT2, GRIA3, GRIN1, GRIN2B, HI-4, H3-3A, HDAC8, HEPACAM, HERC1, HEXA, HIBCH, HIKESHI, HRAS, HSD17B4, HSPD1, HTRA1, HUWE1, HYCC1, IBA57, IDS, IER3IP1, IFIH1, IGF2, INPP5E, IRF6, ISCA1, ISCA2, KANSL1, KARS1, KAT6A, KATNB1, KATNIP, KCNT1, KDM1A, KDM6A, KIAA0586, KIF11, KIF14, KIF2A, KIF5A, KIF5C, KIF7, KIFBP, KMT2D, KNL1, KPTN, KRAS, L1CAM, L2HGDH, LAMA2, LAMB1, LAMC3, LARGE1, LIG4, LIPT2, LMNB1, LRPPRC, LYRM7, MACF1, MAP1B, MAP2K1, MAP2K2, MAPK8IP3, MARS2, MBD5, MCOLN1, MCPHI, MECP2, MED12, MED17, MEF2C, MFS2D, MIPEP, MKSI, MLC1, MNI, MPDZ, MPV17, MRE11, MRPL44, MTAT8P1, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTHFS, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTOR, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYCN, MYO18B, NACCI, NARS2, NAXE, NCAPD2, NCAPD3, NCAPH, NDE1, NDUFAF5, NDUFS1, NDUFV1, NEDD4L, NEU1, NFIB, NFIX, NFUI, NHEJ1, NIN, NIPBL, NKX6-2, NODAL, NOTCH3, NPC1, NPC2, NPHP1, NPRL2, NPRL3, NRAS, NSD1, NSDHL, NSMCE2, NSRP1, NT5C2, NUBPL, NUP37, OCLN, OFD1, OPHN1, OSGEP, PAFAH1B1, PAK1, PC, PCDH12, PCLO, PCNT, PDE6D, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHC1, PHF6, PHGDH, PI4KA, PIBF1, PIDD1, PIGA, PIGN, PIGT, PIK3CA, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLK4, PLP1, PMPCB, PNKP, POLG, POLRIC, POLR3A, POLR3B, POLR3K, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1CB, PPP1R12A, PPP1R15B, PPP2R5B, PPP2R5C, PPP2R5D, PPT1, PQBP1, PRUNE1, PSAP, PSAT1, PTCH1, PTEN, PYCR2, QARS1, RAB11B, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAC3, RAD21, RARS1, RARS2, RBBP8, RELN, RIN2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF125, RNF135, RNF216, RPGRIP1L, RTTN, RXYLT1, SAMHD1, SASS6, SCN3A, SCO1, SCP2, SDHA, SDHAF1, SEPSECS, SERAC1, SETD2, SHH, SIX3, SLC16A2, SLC17A5, SLC1A4, SLC25A12, SLC25A19, SLC25A46, SLC33A1, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMO, SNAP29, SNORD118, SOS1, SOX10, SOX11, SPG11, SPTAN1, SRPX2, STAG2, STAMPB, STIL, STNI, STRADA, SUFU, SUMF1, SURF1, SYN1, TACO1, TARS2, TBC1D20, TBC1D23, TBC1D32, TBC1D7, TBCD, TBCK, TCTN1, TCTN2, TCTN3, TGFB1, TGIF1, THOC6, TMEM106B, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM94, TMTC3, TMX2, TOE1, TOP3A, TP73, TPPI, TRAI, TRAPPC12, TRAPPC6B, TRAPPC9, TREM2, TREX1, TRMT10A, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TTC19, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, TUFM, TYMP, TYROBP, UBE2A, UBE3B, UBT, UFC1, UFM1, UPF3B, VARS1, VARS2, VLDLR, VPS11, VPS53, VRK1, WARS2, WASHC5, WDR45, WDR45B, WDR62, WDR73, WDR81, XRCC4, YWHAE, ZBTB20, ZFYVE26, ZIC2, ZNF148, ZNF335, ZNF423, ZNHIT3