

Agyi malformáció (Cerebral malformation) génpanel v:2025-05-15

Panel tartalma 228 gén:

ACTB, ACTG1, ADGRG1, ADNP, AHDC1, AKT1, AKT3, AMPD2, APC2, ARF1, ARFGEF2, ARID1A, ARID1B, ARX, ASNS, ASPM, ATP1A2, ATP1A3, ATP6V0A2, ATP7A, B3GALNT2, B3GNT2, B4GAT1, BMP4, C19orf12, CASK, CASP2, CCM2, CCND2, CDH2, CDK13, CDK5, CDON, CEP85L, CHMP1A, CIT, CNOT1, CNOT3, COASY, COL18A1, COL3A1, COL4A1, COL4A2, CP, CRADD, CRPPA, CTNNA2, CUL4B, CSNK2A1, DAG1, DCHS1, DCX, DEPDC5, DHCR7, DIAPH1, DISP1, DLL1, DMXL2, DPF2, DPYSL5, DYNC1H1, EIF2AK2, EMC1, EML1, EMX2, ERMARD, EXOSC3, FA2H, FAT4, FGF8, FGFR1, FIG4, FKR1, FKTN, FLNA, FOXA2, FTL, GAS1, GLI2, GMPPB, GPSM2, GRIN1, GRIN2B, H3-3A, HECTD4, HIVEP2, HS2ST1, IER3IP1, IQSEC2, KATNB1, KCNMA1, KIF11, KIF2A, KIF5C, KIF7, KIFBP, KMT2D, KMT2E, KRIT1, L1CAM, LAMA1, LAMA2, LAMB1, LAMC3, LARGE1, LRP2, MACF1, MAP1B, MAPK8IP3, MAX, MED12, MED17, MFSD2A, MN1, MOCS1, MRE11, MTOR, NDE1, NEDD4L, NPRL2, NPRL3, NSRP1, OCLN, OPHN1, OSGEP, PAFAH1B1, PANK2, PDCD10, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHGDH, PI4KA, PIDD1, PIK3CA, PIK3R2, PLA2G6, PLCH1, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1R12A, PQBP1, PTCH1, PTEN, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RAC3, RAD21, RARS2, RBM10, RELN, RERE, RTTN, RXYLT1, SCN3A, SEPSECS, SHH, SIN3A, SIX3, SLC25A19, SLC35A2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMO, SNAP29, SON, SOX11, SOX2, SRD5A3, STAG2, STAMPB, STIL, SUFU, TBC1D20, TBC1D32, TGIF1, TMTC3, TMX2, TOE1, TP73, TRRAP, TSC1, TSEN2, TSEN34, TSEN54, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP6, UBE3B, USP7, USP9X, VLDLR, VPS13A, VPS50, VRK1, WDR45, WDR62, WNK3, YWHAE, ZBTB18, ZBTB20, ZIC2, ZMIZ1

Forrás:

Cerebral malformation (Version 15.7)

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