

Agyi malformáció (Cerebral malformation) génpanel v:2022.02.08.

Panel tartalma 266 gén:

ACTB, ACTG1, ADGRG1, ADNP, AHDC1, AHI1, AKT1, AKT2, AKT3, AMPD2, APC2, ARF1, ARFGEF2, ARID1A, ARID1B, ARL13B, ARX, ASNS, ASPM, ATP1A2, ATP1A3, ATP6V0A2, B3GALNT2, B3GNT2, B4GAT1, B9D1, B9D2, BMP4, C19orf12, C5orf42, CASK, CC2D2A, CCM2, CCND2, CDH2, CDK13, CDK5, CDON, CEP104, CEP120, CEP290, CEP41, CEP85L, CHMP1A, CIT, CNOT1, CNOT3, COASY, COL18A1, COL3A1, COL4A1, COL4A2, CP, CRADD, CTNNA2, CUL4B, CSNK2A1, CSPP1, DAG1, DCHS1, DCX, DEPDC5, DHCR7, DIAPH1, DISP1, DLL1, DMXL2, DPF2, DPYSL5, DYNC1H1, EIF2AK2, EMC1, EML1, EMX2, ENO1, EOMES, ERMARD, EXOSC3, FA2H, FAT4, FGF8, FGFR1, FIG4, FKRP, FKTN, FLNA, FOXA2, FOXH1, FTL, GAS1, GCM2, GLI2, GMPBP, GPBM2, GRIN1, GRIN2B, H3F3A, HIVEP2, HRAS, HS2ST1, IER3IP1, IFT172, INPP5E, IQSEC2, ISPD, KATNB1, KCNMA1, KIAA0586, KIF11, KIF1BP, KIF2A, KIF5C, KIF7, KIFBP, KMT2D, KMT2E, KRAS, KRIT1, L1CAM, LAMA1, LAMA2, LAMB1, LAMC3, LARGE1, LRP2, MACF1, MAP1B, MAPK8IP3, MCF2, MED12, MED17, MFSD2A, MKS1, MN1, MRE11, MTOR, NDE1, NEDD4L, NODAL, NPHP1, NPHP3, NPRL2, NPRL3, NRAS, OCLN, OFD1, OPHN1, OSGEP, PAFAH1B1, PANK2, PDCD10, PEX1, PEX10, PEX11A, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PI4KA, PIDD1, PIK3CA, PIK3R2, PLA2G6, PLCH1, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1R12A, PQBP1, PTCH1, PTEN, RAB11B, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RARS2, RBM10, RELN, RERE, RPGRIP1L, RTTN, RXYLT1, SCN3A, SEPSECS, SHH, SIN3A, SIX3, SLC25A19, SMAD2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMO, SNAP29, SON, SOX2, SRD5A3, SRPX2, STAG2, STAMBP, STIL, SUFU, TBC1D20, TBC1D32, TBC1D7, TCTN1, TCTN2, TCTN3, TGIF1, TMEM138, TMEM216, TMEM231, TMEM237, TMEM5, TMEM67, TMTC3, TMX2, TOE1, TP73, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TTC21B, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP6, UBE3B, USP7, USP9X, VLDLR, VPS13A, VPS50, VPS53, VRK1, WDR45, WDR62, YWHAE, ZBTB18, ZBTB20, ZIC2, ZMIZ1.

Forrás:

Cerebral malformations (Version 5.169)

<https://panelapp.genomicsengland.co.uk/panels/491/> (Megtekintve: 2022.02.08.)

Cerebral Cortical Malformation NGS Panel

<https://www.fulgentgenetics.com/Cerebral-Cortical-Malformation> (Megtekintve: 2022.02.08.)

Comprehensive Brain Malformations Panel

<https://www.genedx.com/test-catalog/available-tests/comprehensive-brain-malformations-panel-2/> (Megtekintve: 2022.02.08.)

Invitae Brain Malformations Panel

<https://www.invitae.com/en/physician/tests/55006/> (Megtekintve: 2022.02.08.)

Brain Malformation Panel

<https://www.preventiongenetics.com/testInfo?val=Brain+Malformation+Panel>
(Megtekintve: 2022.02.08.)