

Microcephalia (Microcephaly) génpanel v:2022.02.09.

Panel tartalma 295 gén:

AARS1, ACBD6, ADARB1, AGMO, AKT3, AMPD2, ANKLE2, AP4B1, AP4E1, AP4M1, AP4S1, ARCN1, ARF3, ARFGEF2, ARPC4, ASPM, ASXL1, ASXL3, ATP1A2, ATP6V0A1, ATP6V0C, ATP9A, ATR, ATRIP, ATRX, BLM, BPTF, BRCA2, BRD4, BRIP1, BUB1, BUB1B, CAMK2B, CAMSAP1, CASK, CCDC47, CCDC88A, CCND2, CDK5RAP2, CDK6, CDT1, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP55, CEP57, CEP63, CHAMP1, CHKA, CIT, CKAP2L, COASY, COG3, COPB1, COX7B, CPSF3, CREBBP, CRIPT, CTCF, CTNNB1, CTU2, CSNK2A1, DDX11, DHCR7, DIAPH1, DNA2, DNMT1L, DNMT3A, DOHH, DONSON, DPM1, DROSHA, DYNC1H1, DYNC1I2, DYRK1A, EEF1D, EFTUD2, EIF2S3, EIF5A, ERCC4, ERCC5, ERCC6, ERCC8, EXOC7, EXOSC3, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FBRSL1, FILIP1, FOXG1, FRA10AC1, GFM1, GINS3, GMNN, GPT2, GRM7, GTF2E2, GTF3C3, H4C3, HDAC8, HHAT, HMGB1, HMGB3, HPDL, IARS1, IER3IP1, IGF1, IGF1R, INTS11, KANSL1, KATNB1, KIF11, KIF14, KIFBP, KMT2B, KNL1, LAGE3, LARP7, LHX2, LIG4, LMNB1, LMNB2, LRRC8C, MBD5, MCM7, MCPH1, MECP2, MED11, MED17, METTL5, MFSD2A, MINPP1, MIPEP, MIR17HG, MORC2, MPLKIP, MRE11, MRE11, MSMO1, MYCN, MYO18B, NAA20, NAPB, NARS1, NBN, NCAPD2, NCAPD3, NDE1, NHEJ1, NIN, NIPBL, NR2E1, NSD2, NSRP1, NUP107, NUP188, NUP214, OPHN1, ORC1, ORC4, ORC6, OSGEP, PAFAH1B1, PALB2, PCDH12, PCDHGC4, PCLO, PCNT, PDCD6IP, PDHA1, PHC1, PHGDH, PLEKHG2, PLK4, PNKP, PNPLA8, POC1A, POGZ, POMT1, PPFIBP1, PPIL1, PPP1R15B, PQBP1, PRIM1, PRUNE1, PSMC3, PTPMT1, PTPN23, PUF60, PUS7, QARS1, QARS1, RAD21, RAD50, RAD51, RAD51C, RARS2, RBBP8, RMI1, RNU4-2, RNU4ATAC, RPL10, RRP7A, RTTN, SARS1, SASS6, SEPSECS, SLC1A4, SLC25A19, SLC38A3, SLC4A10, SLC9A6, SLF2, SLX4, SMARCA2, SMARCA5, SMARCE1, SMC1A, SMC3, SMC5, SMG8, SOX11, SPATA5, SPATA5L1, SPOUT1, STAG2, STAMPB, STIL, SVBP, TAF13, TBC1D20, TBC1D23, THOC6, TMTC3, TMX2, TNPO2, TOE1, TOP3A, TP53RK, TRA2B, TRAI, TRAPPC10, TRAPPC12, TRAPPC14, TRAPPC6B, TRAPPC9, TRIO, TRIP13, TRMT10A, TSEN15, TSEN2, TSEN34, TSEN54, TTC5, TTI1, TUBB, TUBB2B, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, UBA5, UBE3A, UBE3B, UFC1, UFM1, UGP2, UNC80, VARS1, VPS50, VRK1, WDR11, WDR37, WDR4, WDR47, WDR62, WDR73, WLS, XRCC4, YIF1B, YIPF5, ZEB2, ZNF148, ZNF335, ZNF526, ZNF668

Forrás:

Microcephaly and Pontocerebellar Hypoplasia Panel

<https://blueprintgenetics.com/tests/panels/malformations/microcephaly-and-pontocerebellar-hypoplasia-panel/>

Microcephaly NGS Panel

<https://www.fulgentgenetics.com/Microcephaly>

Severe microcephaly (Version 8.2)

<https://panelapp.genomicsengland.co.uk/panels/162/>

(Megtekintve: 2025-05-22)