

Microcephalia (Microcephaly) génpanel v:2023.10.12.

Panel tartalma 307 gén:

AARS, ADARB1, AGMO, AKT3, AMPD2, ANKLE2, AP4B1, AP4E1, AP4M1, AP4S1, ARCN1, ARF3, ARFGEF2, ARPC4, ASPM, ASXL1, ASXL3, ATP11A, ATP1A2, ATP6V0A1, ATP6V0C, ATP9A, ATR, ATRIP, ATRX, BLM, BPTF, BRAT1, BRCA2, BRD4, BRIP1, BUB1B, C7orf43, CAMK2B, CASK, CCDC47, CCDC88A, CCND2, CDC6, CDK5RAP2, CDK6, CDKL5, CDT1, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP55, CEP57, CEP63, CHAMP1, CHKA, CIT, CKAP2L, COASY, COPB1, COPB2, COX7B, CPSF3, CREBBP, CRIP1, CTCF, CTNNB1, CTSF, CTU2, CSNK2A1, DDX11, DDX3X, DHCR7, DIAPH1, DNA2, DNMT1, DNMT3A, DOHH, DONSON, DPM1, DPP6, DROSHA, DYNC1H1, DYNC1I2, DYRK1A, EFTUD2, EIF2S3, EIF5A, EOMES, EP300, ERCC4, ERCC5, ERCC6, ERCC8, EXOC7, EXOSC3, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBRSL1, FOXG1, GFM1, GINS2, GINS3, GMNN, GPT2, GTF2E2, HDAC8, HHAT, HIKESHI, HIST1H4C, HMGB1, HMGB3, HPDL, IARS, IER3IP1, IGF1, IGF1R, KANSL1, KAT6A, KATNB1, KIF11, KIF14, KIF1A, KIF1BP, KMT2D, KNL1, LAGE3, LARP7, LIG4, LMNB1, LMNB2, MBD5, MCM7, MCPH1, MECP2, MED17, METTL5, MFSD2A, MINPP1, MIPEP, MIR17HG, MORC2, MPLKIP, MRE11, MRE11A, MSPO1, MYCN, MYO18B, NAA20, NAPB, NARS, NBN, NCAPD2, NCAPD3, NCAPH, NDE1, NHEJ1, NIN, NIPBL, NR2E1, NSD2, NSMCE2, NSRP1, NUF2, NUP107, NUP188, NUP214, OPHN1, ORC1, ORC4, ORC6, OSGEP, PAFAH1B1, PALB2, PCDH12, PCDHGC4, PCLO, PCNT, PDCD6IP, PDHA1, PHC1, PHGDH, PLAA, PLEKHG2, PLK4, PNKP, POC1A, POGZ, POMT1, PPIL1, PPP1R15B, PPP1R35, PQBP1, PRIM1, PRUNE1, PTPN23, PUF60, PUS7, QARS, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RAD50, RAD51, RAD51C, RARS2, RBBP8, RING1, RMI1, RNASEH2C, RNU4ATAC, RPL10, RRP7A, RTTN, RUSC2, SARS, SASS6, SEPSECS, SLC1A4, SLC25A19, SLC38A3, SLC9A6, SLX4, SMARCA2, SMARCA5, SMARCE1, SMC1A, SMC3, SMG8, SMO, SNAP29, SOX11, SPATA5, SPATA5L1, STAG2, STAMBP, STIL, SVBP, TAF13, TBC1D20, TBC1D23, TCF4, THOC6, TMTC3, TMX2, TNPO2, TOE1, TOP3A, TP53RK, TPR, TPRKB, TRAIP, TRAPPC10, TRAPPC12, TRAPPC6B, TRAPPC9, TRIO, TRIP13, TRMT1, TRMT10A, TSEN15, TSEN2, TSEN34, TSEN54, TTC5, TUBA1A, TUBB, TUBB2B, TUBB3, TUBG1, TUBGCP2, TUBGCP3, TUBGCP4, TUBGCP6, UBA5, UBE3A, UBE3B, UFC1, UFM1, UGP2, UNC80, VARS, VPS13B, VPS50, VRK1, WDFY3, WDR11, WDR37, WDR4, WDR62, WDR73, XRCC4, YIF1B, YIPF5, ZEB2, ZNF148, ZNF335, ZNF526, ZNF668, ZNHIT3, ZPR1

Forrás:

Microcephaly and Pontocerebellar Hypoplasia Panel

<https://blueprintgenetics.com/tests/panels/malformations/microcephaly-and-pontocerebellar-hypoplasia-panel/> (Megtekintve: 2023.05.17.)

Microcephaly NGS Panel

<https://www.fulgentgenetics.com/Microcephaly> (Megtekintve: 2023.05.17.)

Microcephaly Panel

<https://www.genedx.com/tests/detail/microcephaly-panel-771> (Megtekintve: 2023.05.17.)

Severe microcephaly (Version 4.12)

<https://panelapp.genomicsengland.co.uk/panels/162/> (Megtekintve: 2023.05.17.)