

Macrocephalia (Macrocephaly) / Túlnövekedés (Overgrowth) génpanel

v:2025-05-22

Panel tartalma 114 gén:

ABCC9, ADK, AKT1, AKT2, AKT3, ANKH, ASPA, ASXL2, BLTP1, BRSK2, BRWD3, CCND2, CDKN1C, CHD3, CHD4, CHD8, CUL4B, D2HGDH, DHCR24, DICER1, DIS3L2, DNMT3A, DVL1, EED, EIF2B5, ERF, ETFA, ETFB, ETFDH, EXT2, EZH2, FBLN5, FBN1, FGFR3, FOXP1, GATAD2B, GCDH, GFAP, GLI3, GPC3, GPC4, GPSM2, GRIA3, H1-4, H19, HEPACAM, HERC1, HRAS, HUWE1, KCNH1, KCNQ1OT1, KDM1A, KIF7, KMT2E, KPTN, KRAS, L1CAM, LAMB1, LBR, MED12, MITF, MLC1, MPDZ, MTM1, MTOR, NF1, NFIA, NFIB, NFIX, NONO, NPR2, NRAS, NSD1, ODC1, OFD1, PDGFRB, PHF21A, PHF6, PIGA, PIGM, PIK3CA, PIK3R2, PPP1CB, PPP2R5D, PTCH1, PTEN, RAB39B, RIN2, RNF125, RNF135, SETD2, SHANK3, SHOC2, SNX14, SOS1, SPRED1, STRADA, SUFU, SUZ12, SYN1, SZT2, TBC1D7, TCF20, TGFB3, TMEM94, TRIO, TRIP12, TSC1, TSC2, UPF3B, WASHC5, WNT5A, ZBTB20, ZBTB7A

Forrás:

Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (Version 1.120)

<https://panelapp.genomicsengland.co.uk/panels/38/>

Macrocephaly / Overgrowth Syndrome Panel

<https://blueprintgenetics.com/tests/panels/malformations/macrocephaly-overgrowth-syndrome-panel/>

Macrocephaly/Overgrowth Syndrome NGS Panel

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

Overgrowth and Macrocephaly Syndromes Panel

<https://www.preventiongenetics.com/testInfo?val=Overgrowth-and-Macrocephaly-Syndromes-Panel>

(Megtekintve: 2025-05-22)