

Macrocephalia (Macrocephaly) génpanel v:2023.10.12.

Panel tartalma 62 gén:

AKT1, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, EED, EIF2B5, EZH2, GFAP, GLI3, GPC3, GPSM2, GRIA3, H19, HEPACAM, HERC1, HUWE1, KDM1A, KIAA0196, KIF7, KPTN, KRAS, L1CAM, MED12, MLC1, MPDZ, MTOR, NF1, NFIA, NFIB, NFIX, NPR2, NSD1, OFD1, PHF6, PIGA, PIK3CA, PIK3R2, PPP2R5D, PTCH1, PTEN, RAB39B, RNF125, RNF135, SETD2, SNX14, SPRED1, SYN1, TBC1D7, TMEM94, TSC1, TSC2, UPF3B, ZBTB20

Forrás:

Invitae Overgrowth and Macrocephaly Syndromes Panel

<https://www.invitae.com/en/providers/test-catalog/test-04501> (Megtekintve: 2023.05.17.)

Macrocephaly / Overgrowth Syndrome Panel

<https://blueprintgenetics.com/tests/panels/malformations/macrocephaly-overgrowth-syndrome-panel/> (Megtekintve: 2023.05.17.)

Macrocephaly/Overgrowth Syndrome NGS Panel

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

Syndromic Macrocephaly/Overgrowth Panel

<https://providers.genedx.com/tests/detail/syndromic-macrocephaly-overgrowth-panel-775>
(Megtekintve: 2023.05.17.)