

Túlnövekedés (Overgrowth) génpanel v:2023.10.12.

Panel tartalma 66 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, HIST1H1E, HUWE1, IGF2, KCNQ1OT1, KDM1A, KIAA0196, KIF7, KPTN, KRAS, L1CAM, MED12, MLC1, MPDZ, MTOR, NF1, NFIB, NFIX, NPR2, NSD1, OFD1, PDGFRB, PHF6, PIGA, PIK3CA, PIK3R2, PPP2R5D, PTCH1, PTEN, RAB39B, RNF125, RNF135, SETD2, SPRED1, SUZ12, SYN1, TBC1D7, TMEM94, TSC1, TSC2, UPF3B, ZBTB20, ZBTB7A

Forrás:

Beckwith-Wiedemann syndrome (BWS) and other congenital overgrowth disorders (Version 1.119)

<https://panelapp.genomicsengland.co.uk/panels/38/> (Megtekintve: 2023.05.18.)

Invitae Overgrowth and Macrocephaly Syndromes Panel

<https://www.invitae.com/en/providers/test-catalog/test-04501> (Megtekintve: 2023.05.18.)

Macrocephaly / Overgrowth Syndrome Panel

<https://blueprintgenetics.com/tests/panels/malformations/macrocephaly-overgrowth-syndrome-panel/> (Megtekintve: 2023.05.18.)

Macrocephaly/Overgrowth Syndrome NGS Panel

<https://www.fulgentgenetics.com/Macrocephaly> (Megtekintve: 2023.05.18.)