

Craniosynostosis génpanel v:2022.01.26.

Panel tartalma 125 gén:

ABCC9, ADAMTSL4, AHDC1, ALPL, ALX1, ALX3, ALX4, ARSB, ASXL1, ATR, AXIN2, B3GAT3, BBS9, BMP4, BRAF, CCBE1, CD96, CDC45, CDT1, CEP120, CHST3, COLEC10, COLEC11, CRTAP, CTSK, CYP26B1, DHRS3, EDNRB, EFNA4, EFNBB1, ERF, ESCO2, FAM20C, FBN1, FGF3, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FLNB, FREM1, GDF5, GLI3, GNAS, GNPTAB, GPC3, HUWE1, IDS, IDUA, IFT122, IFT140, IFT43, IGF1R, IHH, IL11RA, IMPAD1, IRX5, JAG1, KANSL1, KANSL1-AS1, KAT6A, KAT6B, KDM6A, KMT2D, KRAS, LMX1B, LRP5, MASP1, MEGF8, MSX2, NFIA, NFIX, NOG, ORC1, ORC4, ORC6, OSTM1, P4HB, PAX3, PHEX, POR, PPP1CB, PPP3CA, PRRX1, PTPN11, PTPRD, RAB23, RECQL4, RSPRY1, RUNX2, SCARF2, SCN4A, SEC24D, SH3PXD2B, SHOC2, SIX1, SIX2, SKI, SLC25A24, SLC3A2, SMAD2, SMAD3, SMAD6, SMO, SOX10, SOX6, SPECC1L, STAT3, TCF12, TCOF1, TFAP2B, TGFB2, TGFB3, TGFB1, TGFB2, TICRR, TLK2, TMC01, TWIST1, TWIST2, WDR19, WDR35, ZEB2, ZIC1.

Forrás:

Craniosynostosis (Version 2.61)

<https://panelapp.genomicsengland.co.uk/panels/168/> (Megtekintve: 2022.01.26.)

Craniosynostosis Panel

<https://blueprintgenetics.com/tests/panels/malformations/craniosynostosis-panel/>

(Megtekintve: 2022.01.26.)

Invitae Craniosynostosis Panel

<https://www.invitae.com/pt/physician/tests/04423/> (Megtekintve: 2022.01.26.)