

Craniosynostosis génpanel v:2023.10.12.

Panel tartalma 150 gén:

ABCC9, ACTB, ACTG1, ACTG2, ADAMTSL4, AHDC1, ALPL, ALX1, ALX3, ALX4, ARID1B, ARSB, ASXL1, ASXL3, ATR, AXIN2, B3GAT3, BBS9, BCL11B, BMP4, BRAF, CCBE1, CD96, CDC45, CDK13, CDT1, CEP120, CHD5, CHD7, CHST3, COLEC10, COLEC11, CRTAP, CTSK, CYP26B1, DHRS3, DPF2, DPH1, EDNRB, EFNA4, EFNBB1, ERF, ESCO2, FAM20C, FBN1, FBXO11, FGF10, FGF3, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FLNB, FREM1, GDF5, GINS2, GLI3, GNAS, GNPTAB, GPC3, HNRNPK, HUWE1, IDS, IDUA, IFT122, IFT140, IFT43, IGF1R, IHH, IL11RA, IL6ST, IMPAD1, IRX5, JAG1, KANSL1, KANSL1-AS1, KAT6A, KAT6B, KDM6A, KMT2D, KRAS, LMX1B, LRP5, LTBP1, MAN2B1, MASP1, MEGF8, MSX2, NFIA, NFIX, NOG, OGT, ORC1, ORC4, ORC6, OSTM1, P4HB, PAX3, PHEX, PJA1, POR, PPP1CB, PPP3CA, PRRX1, PTCH1, PTPN11, PTPRD, RAB23, RECQL4, RNU12, RSPRY1, RUNX2, SCARF2, SCN4A, SEC24D, SH3PXD2B, SHOC2, SIX1, SIX2, SKI, SLC25A24, SLC3A2, SMAD2, SMAD3, SMAD6, SMO, SOX10, SOX6, SPECC1L, SPRY1, STAT3, TCF12, TCOF1, TFAP2B, TGFB2, TGFB3, TGFB1, TGFB2, TICRR, TLK2, TMC01, TRAF7, TWIST1, TWIST2, WDR19, WDR35, ZEB2, ZIC1, ZNF462

Forrás:

Rare syndromic craniosynostosis or isolated multisuture synostosis (Version 4.97)

<https://panelapp.genomicsengland.co.uk/panels/168/> (Megtekintve: 2023.05.16.)

Craniosynostosis Panel

<https://blueprintgenetics.com/tests/panels/malformations/craniosynostosis-panel/>
(Megtekintve: 2023.05.16.)

Invitae Craniosynostosis Panel

<https://www.invitae.com/en/providers/test-catalog/test-04423> (Megtekintve: 2023.05.16.)