

Alacsonynövés (Short stature) génpanel v:2025-05-23

Panel tartalma 207 gén:

ACAN, ACTB, ACTG1, ALMS1, AMMECR1, ANAPC1, ANKRD11, ARCN1, ARID1A, ARID1B, ATR, ATRIP, B3GAT3, BCS1L, BLM, BMP2, BRAF, BRCA2, BRF1, BRIP1, BTK, CBL, CCDC186, CCDC8, CDC45, CDC6, CDKN1C, CDT1, CENPJ, CEP152, CEP57, CEP63, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREBBP, CRIPT, CUL7, DHCR7, DNA2, DONSON, DVL1, EP300, ERCC4, ERCC6, ERCC8, EVC, EVC2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FBN1, FBXO22, FGD1, FGFR3, FN1, FOXP4, GGPS1, GH1, GHR, GHRHR, GHSR, GLI2, GLI3, GNAS, HDAC8, HESX1, HMGA2, HRAS, HSPG2, IDUA, IGF1, IGF1R, IGF2, IGFALS, IHH, INSR, INTS1, IRS1, KDM3B, KDM6A, KMT2D, KRAS, LARP7, LFNG, LHX3, LHX4, LIG4, LMNA, LRRC8C, LZTR1, MAP2K1, MAP2K2, MAPK1, MATN3, MRAS, MSTO1, MTX2, NBAS, NBN, NF1, NHLRC2, NIPBL, NLRP2, NLRP5, NLRP7, NOTCH2, NPPC, NPR2, NRAS, NSMCE2, OBSL1, ORC1, ORC4, ORC6, OSGEP, OTX2, PADI6, PALB2, PCNT, PDE4D, PIK3R1, PISD, PITX2, PLAG1, PLK4, POC1A, POP1, POU1F1, PPP1CB, PPP3CA, PRKAR1A, PRMT7, PROP1, PTH1R, PTPN11, PUF60, RAD21, RAD51, RAF1, RALA, RAP1B, RASA2, RBBP8, RECQL4, RIT1, RNPC3, RNU4ATAC, ROR2, RPS6KA3, RRAS, RRAS2, RSPRY1, RTTN, SETD5, SGMS2, SHOC2, SHOX, SLC13A1, SLF2, SLX4, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SMC5, SOS1, SOS2, SOX11, SOX2, SOX3, SOX9, SPOUT1, SPRED1, SPRED2, SRCAP, STAT5B, TALDO1, TBX19, TBX2, TBX3, TOP3A, TRIM37, TRMT10A, UBE2T, VPS50, WNT5A, XRCC4, ZFP57, ZNF668

Forrás:

Monogenic short stature (Version 1.17)

<https://panelapp.genomicsengland.co.uk/panels/1471/>

Comprehensive Short Stature Syndrome Panel

<https://blueprintgenetics.com/tests/panels/malformations/comprehensive-short-stature-syndrome-panel/>

Short Stature NGS Panel

<https://www.fulgentgenetics.com/Short-Stature>

(Megtekintve: 2025-05-23)