

Szkeletális diszplázia (Skeletal dysplasia) génpanel v:2025-05-23

Panel tartalma 562 gén:

ABCC9, ABL1, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AFF3, AFF4, AGA, AGPS, AIFM1, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANAPC1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARL6, ARSB, ARSK, ARSL, ASCC1, ASPM, ASXL1, ASXL2, ATP6V0A2, ATP7A, ATR, ATRIP, AXIN1, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B9D1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, BTRC, C2CD3, CA2, CANT1, CASR, CBF3, CC2D2A, CCDC8, CCN2, CCN6, CCNQ, CDC45, CDC6, CDH3, CDK5RAP2, CDKN1C, CDT1, CENPE, CENPJ, CEP120, CEP135, CEP152, CEP290, CEP63, CEP97, CFAP410, CHST14, CHST3, CHSY1, CHUK, CILK1, CKAP2L, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRIPT, CRTAP, CTNS, CTSA, CTSC, CTSK, CUL7, CWC27, CYP27B1, CYP2R1, CSF1R, CSGALNACT1, CSNK1G1, CSPP1, DCC, DDR2, DDRGK1, DHCR24, DHCR7, DHODH, DIP2C, DIS3L2, DLL1, DLL3, DLL4, DLX3, DLX5, DMP1, DMRT2, DNA2, DNMT3A, DOCK6, DONSON, DPAGT1, DPM1, DROSHA, DVL1, DVL2, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EED, EFL1, EFN1, EFTUD2, EIF2AK3, EN1, ENPP1, EOGT, ERF, ERI1, ESCO2, EVC, EVC2, EXOC6B, EXOSC2, EXT1, EXT2, EXTL3, EZH2, FAH, FAM111A, FAM20C, FAR1, FAT4, FBLN1, FBN1, FBN2, FBXW11, FBXW4, FERMT3, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FMN1, FN1, FTO, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GHRHR, GHSR, GJA1, GLB1, GLI3, GMNN, GNAS, GNE, GNPAT, GNPAT1, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GREM1, GSC, GUSB, GZF1, HAAO, HDAC4, HDAC8, HEATR3, HES7, HGSNAT, HHAT, HNRNPK, HOXA11, HOXA13, HOXD13, HPGD, HS2ST1, HSPA9, HSPG2, HYAL1, IARS2, IDH1, IDH2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IGF1, IGF2, IHH, IKBKG, IL11RA, IL1RN, INPPL1, INTU, JAG1, KAT6B, KDELR2, KIAA0586, KIAA0753, KIF22, KIF24, KIF5B, KIF7, KL, KMT2A, KMT2D, KYNU, LARP7, LBR, LEMD3, LFNG, LIFR, LIG4, LMBR1, LMNA, LMX1B, LONP1, LOXL3, LPIN2, LRP4, LRP5, LRRC8C, LRRK1, LTBP1, LTBP2, LTBP3, MAFB, MAN2B1, MANBA, MAP3K7, MASP1, MATN3, MBTPS1, MBTPS2, MCM5, MCPH1, MEGF8, MEOX1, MESD, MESP2, MGP, MIR17HG, MKKS, MKS1, MMP13, MMP14, MMP2, MMP9, MNX1, MPDU1, MSX2, MTX2, MYCN, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEPRO, NEU1, NF1, NFIX, NIN, NIPBL, NKX3-2, NLRP3, NMNAT1, NOG, NOTCH1, NOTCH2, NPPC, NPR2, NPR3, NRCAM, NSD1, NSDHL, NSMCE2, NT5E, NTRK1, NXN, OBSL1, OCRL, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PAX3, PCGF2, PCNT, PCYT1A, PDE3A, PDE4D, PDIA6, PEX5, PEX7, PFN1, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PKDCC, PLEKHM1, PLK4, PLOD2, PLS3, POC1A, POLR1A, POLR1B, POLR1C, POLR1D, POP1, POR, PPIB, PPP3CA, PRKAR1A, PRKG2, PRMT7, PSAT1, PSMC3, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RAD21, RASGRP2, RBBP8, RBM8A, RBPJ, RECQL4, RFT1, RINT1, RIPPLY2, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RSPO2, RSPRY1, RTTN, RUNX2, SALL1, SALL4, SBDS, SC5D, SCARF2, SCUBE3, SEC24D, SERPINF1, SERPINH1, SETBP1, SETD2, SETD5, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOX, SKI, SLC10A7, SLC13A1, SLC17A5, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35B2, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SLCO5A1, SMAD3, SMAD4, SMAD6, SMARCA1,

SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, SQSTM1, SRCAP, STT3A, SUCO, SULF1, SUMF1, TAB2, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFB2, THPO, TMC01, TMEM165, TMEM216, TMEM231, TMEM251, TMEM38B, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TOMM7, TONSL, TP63, TRAF3IP1, TRAIIP, TRAPPC2, TREM2, TRIM37, TRIP11, TRIP4, TRMT10A, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TUBGCP4, TUBGCP6, TWIST1, TYROBP, UBA2, UBE3B, UFSP2, UNC45A, VAC14, VDR, VPS33A, VPS35L, WBP11, WDPCP, WDR19, WDR35, WDR4, WNT1, WNT10B, WNT3, WNT3A, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZMPSTE24, ZNF687, ZSWIM6

Forrás:

Skeletal dysplasia (Version 8.4)

<https://panelapp.genomicsengland.co.uk/panels/309/>

Skeletal Dysplasias Core Panel

<https://blueprintgenetics.com/tests/panels/malformations/skeletal-dysplasias-core-panel/>

Skeletal Dysplasias NGS Panel

<https://fulgentgenetics.com/Skeletal-Dysplasias>

Invitae Skeletal Disorders Panel

<https://www.invitae.com/en/physician/tests/89100/>

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