

Szkeletális diszplázia (Skeletal dysplasia) génpanel v:2022.02.08.

Panel tartalma 668 gén:

ABCC9, ABL1, ACAN, ACP5, ACVR1, ACVR2B, ADAMTS10, ADAMTS17, ADAMTSL2, ADGRV1, ADI1, AFF3, AFF4, AGA, AGPS, AHI1, AIFM1, AIPL1, AKT1, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANAPC1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARSB, ARSE, ASCC1, ASPM, ASXL1, ASXL2, ATP6V0A2, ATP7A, ATR, ATRIP, ATXN10, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B9D1, B9D2, BANF1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, C16orf62, C21orf2, C2CD3, C2orf71, C5orf42, CA2, CANT1, CASR, CC2D2A, CCDC28B, CCDC39, CCDC40, CCDC8, CD96, CDC45, CDC6, CDH23, CDH3, CDK5RAP2, CDKN1C, CDT1, CENPJ, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP63, CEP97, CFAP410, CFTR, CHST14, CHST3, CHSY1, CHUK, CKAP2L, CLCN5, CLCN7, CLRN1, COG1, COG4, COL10A1, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL27A1, COL2A1, COL5A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COMP, COPB2, CRB1, CREB3L1, CREBBP, CRELD1, CRIPT, CRTAP, CRX, CTNS, CTSA, CTSC, CTSK, CUL7, CWC27, CYP26B1, CYP27B1, CYP2R1, CSF1R, CSGALNACT1, CSNK1G1, CSPP1, DACT1, DCC, DDR2, DDRGK1, DHCR24, DHCR7, DHODH, DIP2C, DIS3L2, DLL1, DLL3, DLL4, DLX3, DLX5, DLX6, DMP1, DMRT2, DNA2, DNAAF1, DNAAF2, DNAAF3, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DNMT3A, DOCK6, DOLPP1, DONSON, DPAGT1, DPM1, DPM2, DPM3, DSPP, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, EBP, EED, EFTUD2, EIF2AK3, ENPP1, EOGT, EP300, ERF, ESCO2, ESR1, ETF1, EVC, EVC2, EXOC6B, EXOSC2, EXT1, EXT2, EXTL3, EZH2, FAH, FAM111A, FAM20C, FAM46A, FAM58A, FAR1, FAT4, FBLIM1, FBLN1, FBN1, FBN2, FBXW4, FERMT3, FGF10, FGF16, FGF23, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FMN1, FN1, FOXC1, FOXH1, FTO, FUCA1, FZD2, GALNS, GALNT3, GDF1, GDF3, GDF5, GDF6, GHR, GHRHR, GHRSR, GJA1, GLB1, GLI3, GLIS2, GMNN, GNAS, GNE, GNPAT, GNPAT1, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GREM1, GSC, GUCY2D, GUSB, GZF1, HDAC4, HDAC5, HDAC8, HES7, HGSNAT, HHAT, HNRNPK, HOXA11, HOXA13, HOXD11, HOXD13, HPGD, HS2ST1, HSPG2, HYAL1, HYLS1, IARS2, ICK, IDH1, IDH2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IFT88, IGF1, IGF2, IHH, IKBKG, IL11RA, IL1RN, IMPAD1, IMPDH1, INPPL1, INTU, INVS, IQCB1, JAG1, KAT6B, KCNJ13, KDELR2, KIAA0586, KIAA0753, KIAA1217, KIF22, KIF7, KL, KMT2A, KMT2D, LARP7, LBR, LCA5, LEFTY2, LEMD3, LFNG, LIFR, LIG4, LMBR1, LMNA, LMX1B, LONP1, LOXL3, LPIN2, LRAT, LRP4, LRP5, LRP6, LRRK1, LTBP1, LTBP2, LTBP3, MAFB, MAN2B1, MAN2C1, MANBA, MAP3K7, MASP1, MATN3, MBTPS1, MBTPS2, MCM5, MCPH1, MEGF8, MEOX1, MESD, MESDC2, MESP2, MGP, MIA3, MIR17HG, MKKS, MKS1, MMP13, MMP14, MMP2, MMP9, MNX1, MPDU1, MSX2, MTAP, MTX2, MYCN, MYH3, MYO18B, MYO1H, MYO7A, NAGLU, NANS, NBAS, NEK1, NEK8, NEU1, NF1, NFIX, NIN, NIPBL, NKX2-5, NKX3-2, NLRP3, NME8, NODAL, NOG, NOTCH1, NOTCH2, NPHP1, NPHP3, NPHP4, NPPC, NPR2, NPR3, NSD1, NSDHL, NSMCE2, NTRK1, NXN, OAT, OBSL1, OCRL, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PAX3, PCDH15, PCGF2, PCNT, PCYT1A, PDE3A, PDE4D, PDIA6, PEX5, PEX7, PFN1,

PGM3, PHEX, PHF6, PHGDH, PIGT, PIGV, PIK3C2A, PIK3CA, PIK3R1, PIN1, PIR, PISD, PITX1, PKD2, PKDCC, PKHD1, PLCB3, PLEKHM1, PLK4, PLOD1, PLOD2, PLS3, POC1A, POLR1A, POLR1B, POLR1C, POLR1D, POP1, POR, PPIB, PPP3CA, PRKAR1A, PRKG2, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PTPRQ, PUF60, PYCR1, RAB23, RAB33B, RAB3GAP2, RAD21, RASGRP2, RBBP8, RBM8A, RBPJ, RD3, RDH12, RECQL4, RFT1, RINT1, RIPPLY2, RMRP, RNU4ATAC, ROR2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RPL13, RSPH4A, RSPH9, RSPO2, RSPRY1, RTTN, RUNX2, SALL1, SALL4, SBDS, SC5D, SCARF2, SCNN1A, SCNN1B, SCNN1G, SCUBE3, SDCCAG8, SEC24D, SEM1, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SLCO5A1, SMAD3, SMAD4, SMAD6, SMARCA2, SMARCA4, SMARCA1, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX11, SOX9, SP7, SPARC, SPATA7, SPECC1L, SQSTM1, SRCAP, SUCO, SULF1, SUMF1, TAB2, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTEX1D2, TCTN1, TCTN2, TCTN3, TDP2, TERT, TGDS, TGFB1, TGFB2, TGFB2, THPO, TMC01, TMEM138, TMEM165, TMEM216, TMEM231, TMEM237, TMEM251, TMEM38B, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TNXB, TONSL, TOPORS, TP63, TRAF3IP1, TRAIP, TRAPPC2, TREM2, TRIM32, TRIM37, TRIP11, TRIP4, TRMT10A, TRPS1, TRPV4, TRPV6, TSC1, TSC2, TTC21B, TTC8, TUBGCP4, TUBGCP6, TULP1, TWIST1, TYROBP, UBA2, UBE3B, UFSP2, UMOD, UNC45A, USH1C, USH1G, USH2A, USP9X, VAC14, VDR, VHL, VPS33A, WBP11, WDPCP, WDR19, WDR34, WDR35, WDR4, WDR60, WHRN, WISP3, WNT1, WNT10B, WNT3, WNT3A, WNT5A, WNT7A, WRN, XPNPEP3, XRCC4, XYLT1, XYLT2, YY1, ZBTB16, ZIC3, ZMPSTE24, ZNF423, ZNF687, ZSWIM6.

Forrás:

Skeletal dysplasia (Version 2.170)

<https://panelapp.genomicsengland.co.uk/panels/309/> (Megtekintve: 2022.02.08.)

Skeletal Dysplasias Core Panel

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

(Megtekintve: 2022.02.08.)

Skeletal Dysplasias NGS Panel

<https://fulgentgenetics.com/Skeletal-Dysplasias> (Megtekintve: 2022.02.08.)

Invitae Skeletal Disorders Panel

<https://www.invitae.com/en/physician/tests/89100/> (Megtekintve: 2022.02.08.)