



## Seznam genů zahrnutých ve virtuálním panelu – Kostní dysplázie (FocusedExome)

### Virtuální panel zahrnuje analýzu 489 genů:

ABCC9, ABL1, ACAN, ACP5, ACVR1, ACVR2B, ADAMTS10, ADAMTS17, ADAMTSL2, AFF3, AGA, AGPS, AHI1, AIPL1, AKT1, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARSB, ARSE, ASXL1, ATP6V0A2, ATP7A, ATXN10, ATXN10, B3GALT6, B3GAT3, B4GALT7, B9D1, B9D2, BANF1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, C21orf2, C2orf71, C5orf42, CA2, CANT1, CASR, CC2D2A, CCDC28B, CCDC39, CCDC40, CCDC8, CD96, CDC6, CDH23, CDH3, CDKN1C, CDT1, CEP120, CEP164, CEP290, CEP41, CFTR, CLCN5, CLCN7, CLRN1, COG1, COL10A1, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL2A1, COL5A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, CRB1, CREBBP, CRELD1, CRTAP, CRX, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP26B1, CYP27B1, CYP2R1, DACT1, DCC, DDR2, DHCR24, DHCR7, DHODH, DIS3L2, DLL3, DLX3, DLX5, DLX6, DMP1, DNAAF1, DNAAF2, DNAAF3, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DNMT3A, DOCK6, DPAGT1, DPM1, DPM2, DPM3, DSPP, DYM, DYNC2H1, EBP, EFN1, EFTUD2, EIF2AK3, ENPP1, EP300, ERF, ESCO2, ESR1, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FAM58A, FBLIM1, FBLN1, FBN1, FBN2, FBXW4, FERMT3, FGF10, FGF23, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FMN1, FN1, FOXC1, FOXH1, FUCA1, GALNS, GALNT3, GDF1, GDF3, GDF5, GDF6, GHR, GJA1, GLB1, GLI3, GLIS2, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GREM1, GSC, GUCY2D, GUSB, HDAC4, HDAC8, HES7, HGSNAT, HOXA11, HOXA13, HOXD13, HPGD, HSPG2, HYLS1, CHST14, CHST3, CHSY1, ICK, IDH1, IDH2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT80, IGF1R, IHH, IKBKG, IL11RA, IL1RN, IMPAD1, IMPDH1, INVS, IQCB1, KAT6B, KCNJ13, KIF22, KIF7, KMT2D, LBR, LCA5, LEFTY2, LEMD3, LFNG, LIFR, LMBR1, LMNA, LMX1B, LOXL3, LPIN2, LRAT, LRP4, LRP5, LRP6, LTBP2, LTBP3, MAFB, MAN2B1, MANBA, MASP1, MATN3, MCM5, MEOX1, MESP2, MGP, MIR17HG, MKKS, MKS1, MMP13, MMP14, MMP2, MMP9, MNX1, MPDU1, MSX2, MTAP, MYCN, MYO7A, NAGLU, NBAS, NEK1, NEK8, NEU1, NF1, NFIX, NIN, NIPBL, NKX2-5, NKX3-2, NLRP3, NME8, NODAL, NOG, NOTCH1, NOTCH2, NPHP1, NPHP3, NPHP4, NPPC, NPR2, NSD1, NSDHL, OAT, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P4HB, PAPSS2, PAX3, PCDH15, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PHEX, PHF6, PHGDH, PIGV, PIK3CA, PIK3R1, PIN1, PITX1, PKD2, PKHD1, PLEKHM1, PLOD1, PLOD2, POC1A, POLR1C, POLR1D, POP1, POR, PPIB, PRKAR1A, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PTPRQ, PYCR1, RAB23, RAB3GAP2, RAD21, RASGRP2, RBM8A, RBPJ, RD3, RDH12, RECQL4, RFT1, RMRP, RNU4ATAC, ROR2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RSPH4A, RSPH9, RUNX2, SALL1, SALL4, SBDS, SCARF2, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SERPINF1, SERPINH1, SETD2, SF3B4, SGSH, SH3BP2, SH3PXD2B, SHH, SKI, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC1, SNX10, SOST, SOX9, SP7, SPATA7, SPECC1L, SULF1, SUMF1, TALDO1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCIRG1, TCOF1, TCTN1, TCTN2, TCTN3, TERT, TGFB1, TGFB2, TGFB1R1, TGFB2R2, THPO, TMCO1, TMEM138, TMEM165, TMEM216, TMEM231, TMEM237, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TNXB, TOPORS, TP63, TRAPPC2, TREM2, TRIM32, TRIP11, TRPS1, TRPV4, TSC1, TSC2, TTC21B, TTC8, TULP1, TWIST1, TWIST2, TYROBP, UMOD, USH1C, USH1G, USH2A, USP9X, VDR, VHL, WDPCP, WDR19, WDR34, WDR35, WDR60, WISP3, WNT10B, WNT3, WNT5A, WNT7A, WRN, XPNPEP3, XRCC4, XYLT1, XYLT2, YY1, ZBTB16, ZIC1, ZIC3, ZMPSTE24, ZNF423