

Supplementary Table 2. Lists of including genes in a skeletal dysplasia targeted panel

	GCG-UP v1 (essential & addendum gene)
Probe name	Celemics G-mendeliome probe
No. of genes	433
Lists of genes	<p><i>ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AGA, AGPS, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARHGAP31, ARL6, ARSB, ARSL, ASXL1, ASXL2, ATP6V0A2, ATP7A, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, C2CD3, CA2, CANT1, CASR, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDH3, CDKN1C, CDT1, CEP120, CEP290, CFAP410, CHST14, CHST3, CHSY1, CILK1, CLCN5, CLCN7, COG1, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, CREB3L1, CREBBP, CRTAP, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DHCR24, DHCR7, DHODH, DIS3L2, DLL3, DLL4, DLX3, DLX5, DMP1, DNMT3A, DOCK6, DPAGT1, DPM1, DSPP, DVL1, DVL3, DYM, DYNC2H1, DYNC211, DYNC212, DYNC2L1, DYNLT2B, EBP, EED, EFTUD2, EIF2AK3, ENPP1, EOGT, ERF, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FBN1, FBN2, FERMT3, FGF10, FGF16, FGF23, FGFRI1, FGFRI2, FGFRI3, FIG4, FKBP10, FLNA, FLNB, FNI, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GJA1, GLB1, GLI3, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GSC, GUSB, HDAC8, HES7, HGSNAT, HOXA13, HOXD13, HPGD, HSPG2, IDH1, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IHH, IKBKG, IL11RA, IL1RN, INPPL1, KAT6B, KIAA0753, KIF22, KIF7, KMT2D, LBR, LEMD3, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LTBP3, MAFB, MAN2B1, MAP3K7, MASP1, MATN3, MEGF8, MEOX1, MESP2, MGP, MKKS, MKS1, MMP13, MMP2, MNX1, MPDU1, MSX2, MYCN, NAGLU, NANS, NBAS, NEK1, NEU1, NFI1, NFIX, NIPBL, NKX32, NLRP3, NOG, NOTCH1, NOTCH2, NPR2, NSD1, NSDHL, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PITX1, PLOD2, PLS3, POC1A, POLR1A, POLR1C, POLR1D, POP1, POR, PPIB, PRKAR1A, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RASGRP2, RBM8A, RBPJ, RECQL4, RFT1, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RUNX2, SALL1, SALL4, SBDS, SCARF2, SEC24D, SERPINF1, SERPINH1, SETD2, SF3B4, SFRP4, SGSH, SH3BP2, SH3PXD2B, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMARCA1, SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFB3, TMCO1, TMEM165, TMEM216, TMEM231, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TWIST1, TYROBP, VDR, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZMPSTE24, ZSWIM6, ABL1, ANAPC1, ARCN1, B9D1, COG4, CSGALNACT1, DCC, EFN1, EN1, FBLN1, GNPAT1, GPX4, GZF1, HDAC4, HHAT, HNRNP, HS2ST1, IGF1R, KAT6A, KDELR2, KIAA1217, LTBP1, MANBA, MBTPS1, MIR17HG, MMP9, MTX2, MYO18B, NEPRO, NMNAT1, NPR3, NXN, PAM16, PFN1, PISD, PKDCC, PLEKHM1, POLR1B, PRKG2, RAD21, RINT1, SCUBE3, SMAD6, TCF1, TGFB1, TMEM251, TONSL, TWIST2, UBA2, UFSP2, VPS35L, WBP11, ZIC1,</i></p>