

| 2.7 Displasie scheletriche |
|----------------------------|
| ABL1                       |
| ACAN                       |
| ACP5                       |
| ACVR1                      |
| ACVR2B                     |
| ADAMTS10                   |
| ADAMTS17                   |
| ADAMTSL2                   |
| ADGRV1                     |
| ADI1                       |
| AFF3                       |
| AGA                        |
| AGPS                       |
| AHI1                       |
| AIPL1                      |
| AKT1                       |
| ALG12                      |
| ALG3                       |
| ALG9                       |
| ALPL                       |
| ALX1                       |
| ALX3                       |
| ALX4                       |
| AMER1                      |
| ANAPC1                     |
| ANKH                       |
| ANKRD11                    |
| ANO5                       |
| ANTXR2                     |
| ARCN1                      |
| ARHGAP31                   |
| ARID1A                     |
| ARID1B                     |
| ARL13B                     |
| ARL6                       |
| ARSB                       |
| ARSK                       |
| ARSL                       |
| ASXL1                      |
| ASXL2                      |
| ATP6V0A2                   |
| ATP7A                      |

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| ATXN10  |
| AXIN1   |
| B3GALT6 |
| B3GAT3  |
| B3GLCT  |
| B4GALT7 |
| B9D1    |
| B9D2    |
| BANF1   |
| BBS1    |
| BBS10   |
| BBS12   |
| BBS2    |
| BBS4    |
| BBS5    |
| BBS7    |
| BBS9    |
| BHLHA9  |
| BMP1    |
| BMP2    |
| BMPER   |
| BMPR1B  |
| BPNT2   |
| C2CD3   |
| CA2     |
| CANT1   |
| CASR    |
| CC2D2A  |
| CCDC28B |
| CCDC39  |
| CCDC40  |
| CCDC8   |
| CCN6    |
| CCNQ    |
| CD96    |
| CDC45   |
| CDC6    |
| CDH23   |
| CDH3    |
| CDKN1C  |
| CDT1    |
| CEP120  |
| CEP164  |

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| CEP290     |
| CEP41      |
| CFAP410    |
| CFTR       |
| CHST14     |
| CHST3      |
| CHSY1      |
| CILK1      |
| CKAP2L     |
| CLCN5      |
| CLCN7      |
| CLRN1      |
| COG1       |
| COG4       |
| COL10A1    |
| COL11A1    |
| COL11A2    |
| COL12A1    |
| COL1A1     |
| COL1A2     |
| COL2A1     |
| COL5A1     |
| COL9A1     |
| COL9A2     |
| COL9A3     |
| COLEC10    |
| COLEC11    |
| COMP       |
| COPB2      |
| CPLANE1    |
| CRB1       |
| CREB3L1    |
| CREBBP     |
| CRELD1     |
| CRTAP      |
| CRX        |
| CSGALNACT1 |
| CSNK1G1    |
| CSP1       |
| CTSA       |
| CTSC       |
| CTSK       |
| CUL7       |

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| CYP26B1  |
| CYP27B1  |
| CYP2R1   |
| DACT1    |
| DCC      |
| DDR2     |
| DDRGK1   |
| DHCR24   |
| DHCR7    |
| DHODH    |
| DIS3L2   |
| DLL3     |
| DLL4     |
| DLX3     |
| DLX5     |
| DLX6     |
| DMP1     |
| DNAAF1   |
| DNAAF2   |
| DNAAF3   |
| DNAH11   |
| DNAH5    |
| DNAI1    |
| DNAI2    |
| DNAL1    |
| DNMT3A   |
| DOCK6    |
| DOLPP1   |
| DPAGT1   |
| DPM1     |
| DPM2     |
| DPM3     |
| DROSHA   |
| DSPP     |
| DVL1     |
| DVL2     |
| DVL3     |
| DYM      |
| DYNC2H1  |
| DYNC2I1  |
| DYNC2I2  |
| DYNC2LI1 |
| DYNLT2B  |

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| EBP     |
| EED     |
| EFNB1   |
| EFTUD2  |
| EIF2AK3 |
| EN1     |
| ENPP1   |
| EOGT    |
| EP300   |
| ERF     |
| ERI1    |
| ESCO2   |
| ESR1    |
| ETF1    |
| EVC     |
| EVC2    |
| EXT1    |
| EXT2    |
| EXTL3   |
| EZH2    |
| FAM111A |
| FAM20C  |
| FBLIM1  |
| FBLN1   |
| FBN1    |
| FBN2    |
| FBXW11  |
| FBXW4   |
| FERMT3  |
| FGF10   |
| FGF16   |
| FGF23   |
| FGF8    |
| FGF9    |
| FGFR1   |
| FGFR2   |
| FGFR3   |
| FIG4    |
| FKBP10  |
| FLNA    |
| FLNB    |
| FMN1    |
| FN1     |

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| FOXC1   |
| FOXH1   |
| FUCA1   |
| FZD2    |
| GALNS   |
| GALNT3  |
| GDF1    |
| GDF3    |
| GDF5    |
| GDF6    |
| GHR     |
| GJA1    |
| GLB1    |
| GLI3    |
| GLIS2   |
| GNAS    |
| GNPAT   |
| GNPNAT1 |
| GNPTAB  |
| GNPTG   |
| GNS     |
| GORAB   |
| GPC6    |
| GPX4    |
| GREM1   |
| GSC     |
| GUCY2D  |
| GUSB    |
| GZF1    |
| HDAC4   |
| HDAC5   |
| HDAC8   |
| HEATR3  |
| HES7    |
| HGSNAT  |
| HHAT    |
| HNRNPK  |
| HOXA11  |
| HOXA13  |
| HOXD11  |
| HOXD13  |
| HPGD    |
| HS2ST1  |

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| HSPG2    |
| HYLS1    |
| IDH1     |
| IDH2     |
| IDS      |
| IDUA     |
| IFIH1    |
| IFITM5   |
| IFT122   |
| IFT140   |
| IFT172   |
| IFT43    |
| IFT52    |
| IFT80    |
| IFT81    |
| IFT88    |
| IGF1R    |
| IHH      |
| IKBKG    |
| IL11RA   |
| IL1RN    |
| IMPDH1   |
| INPPL1   |
| INVS     |
| IQCB1    |
| KAT6A    |
| KAT6B    |
| KCNJ13   |
| KDEL2    |
| KIAA0753 |
| KIAA1217 |
| KIF22    |
| KIF24    |
| KIF5B    |
| KIF7     |
| KMT2D    |
| LBR      |
| LCA5     |
| LEFTY2   |
| LEMD3    |
| LFNG     |
| LIFR     |
| LMBR1    |

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| LMNA    |
| LMX1B   |
| LONP1   |
| LOXL3   |
| LPIN2   |
| LRAT    |
| LRP4    |
| LRP5    |
| LRP6    |
| LRRK1   |
| LTBP1   |
| LTBP2   |
| LTBP3   |
| MAFB    |
| MAN2B1  |
| MAN2C1  |
| MANBA   |
| MAP3K7  |
| MASP1   |
| MATN3   |
| MBTPS1  |
| MBTPS2  |
| MCM5    |
| MEGF8   |
| MEOX1   |
| MESD    |
| MESP2   |
| MGP     |
| MIA3    |
| MIR17HG |
| MKKS    |
| MKS1    |
| MMP13   |
| MMP14   |
| MMP2    |
| MMP9    |
| MNX1    |
| MPDU1   |
| MSX2    |
| MTAP    |
| MTX2    |
| MYCN    |
| MYH3    |



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| MYO18B |
| MYO1H  |
| MYO7A  |
| NAGLU  |
| NANS   |
| NBAS   |
| NEK1   |
| NEK8   |
| NEPRO  |
| NEU1   |
| NF1    |
| NFIX   |
| NIN    |
| NIPBL  |
| NKX2-5 |
| NKX3-2 |
| NLRP3  |
| NME8   |
| NMNAT1 |
| NODAL  |
| NOG    |
| NOTCH1 |
| NOTCH2 |
| NPHP1  |
| NPHP3  |
| NPHP4  |
| NPPC   |
| NPR2   |
| NPR3   |
| NRCAM  |
| NSD1   |
| NSDHL  |
| NXN    |
| OAT    |
| OBSL1  |
| OFD1   |
| ORC1   |
| ORC4   |
| ORC6   |
| OSTM1  |
| P3H1   |
| P4HB   |
| PAM16  |

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| PAPSS2  |
| PAX3    |
| PCARE   |
| PCDH15  |
| 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    |
| PRKAR1A |

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| PRKG2    |
| PRMT7    |
| PSAT1    |
| PSMC3    |
| PSPH     |
| PTDSS1   |
| PTH1R    |
| PTHLH    |
| PTPN11   |
| PTPRQ    |
| PUF60    |
| PYCR1    |
| RAB23    |
| RAB33B   |
| RAB3GAP2 |
| RAD21    |
| RASGRP2  |
| RBM8A    |
| RBPJ     |
| RD3      |
| RDH12    |
| RECQL4   |
| RFT1     |
| RINT1    |
| RIPPLY2  |
| RMRP     |
| RNU4ATAC |
| ROR2     |
| RPE65    |
| RPGR     |
| RPGRIP1  |
| RPGRIP1L |
| RPL13    |
| RSPH4A   |
| RSPH9    |
| RUNX2    |
| SALL1    |
| SALL4    |
| SBDS     |
| SCARF2   |
| SCNN1A   |
| SCNN1B   |
| SCNN1G   |

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| SCUBE3   |
| SDCCAG8  |
| SEC24D   |
| SEM1     |
| SERPINF1 |
| SERPINH1 |
| SETD2    |
| SETD5    |
| SF3B4    |
| SFRP4    |
| SGMS2    |
| SGSH     |
| SH3BP2   |
| SH3PXD2B |
| SHH      |
| SHOX     |
| SKI      |
| SLC10A7  |
| SLC17A5  |
| SLC26A2  |
| SLC29A3  |
| SLC34A1  |
| SLC34A3  |
| SLC35B2  |
| SLC35C1  |
| SLC35D1  |
| SLC39A13 |
| SLCO2A1  |
| SLCO5A1  |
| SMAD3    |
| SMAD4    |
| SMAD6    |
| SMARCA2  |
| SMARCA4  |
| SMARCAL1 |
| SMARCB1  |
| SMARCE1  |
| SMC1A    |
| SMC3     |
| SMOC1    |
| SNRPB    |
| SNX10    |
| SOST     |

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| SOX11   |
| SOX9    |
| SP7     |
| SPARC   |
| SPATA7  |
| SPECC1L |
| STT3A   |
| SUCO    |
| SULF1   |
| SUMF1   |
| TALDO1  |
| TAPT1   |
| TBCE    |
| TBX15   |
| TBX3    |
| TBX4    |
| TBX5    |
| TBX6    |
| TBXAS1  |
| TCF12   |
| TCIRG1  |
| TCOF1   |
| TCTN1   |
| TCTN2   |
| TCTN3   |
| TDP2    |
| TENT5A  |
| TERT    |
| TGDS    |
| TGFB1   |
| TGFB2   |
| TGFBR1  |
| TGFBR2  |
| THPO    |
| TMCO1   |
| TMEM138 |
| TMEM165 |
| TMEM216 |
| TMEM231 |
| TMEM237 |
| TMEM251 |
| TMEM38B |
| TMEM67  |

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| TNFRSF11A |
| TNFRSF11B |
| TNFSF11   |
| TNXB      |
| TONSL     |
| TOPORS    |
| TP53      |
| TP63      |
| TRAPPC2   |
| TREM2     |
| TRIM32    |
| TRIP11    |
| TRMT10A   |
| TRPS1     |
| TRPV4     |
| TRPV6     |
| TSC1      |
| TSC2      |
| TTC21B    |
| TTC8      |
| TULP1     |
| TWIST1    |
| TWIST2    |
| TYROBP    |
| UBA2      |
| UFSP2     |
| UMOD      |
| UNC45A    |
| USH1C     |
| USH1G     |
| USH2A     |
| USP9X     |
| VAC14     |
| VDR       |
| VHL       |
| VPS35L    |
| WBP11     |
| WDPCP     |
| WDR19     |
| WDR35     |
| WHRN      |
| WNT1      |
| WNT10B    |

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| WNT3     |
| WNT5A    |
| WNT7A    |
| WRN      |
| XPNPEP3  |
| XRCC4    |
| XYLT1    |
| XYLT2    |
| YY1      |
| ZBTB16   |
| ZIC1     |
| ZIC3     |
| ZMPSTE24 |
| ZNF423   |
| ZNF687   |
| ZSWIM6   |
| ABCC9    |