

WES SHORT STATURE/SKELETAL DYSPLASIA DG 2.14

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ABCC9	157.9	99.9	99.2	239850
ACAN	121.6	91.6	85.0	165800
ACP5	196.2	100.0	99.9	607944
ACTB	129.0	99.1	94.2	243310
ACVR1	165.1	100.0	100.0	135100
ADAMTS10	107.8	99.9	98.7	277600
ADAMTS17	117.1	88.9	86.7	613195
ADAMTSL2	112.2	96.5	91.0	231050
AGA	130.2	100.0	100.0	208400
AGPS	51.7	96.8	84.8	600121
ALG12	156.2	100.0	100.0	607143
ALG3	132.9	100.0	100.0	601110
ALG9	124.3	100.0	99.6	608776
ALMS1	179.8	99.9	99.7	203800
ALPL	156.4	100.0	100.0	241500;146300;241510
AMER1	96.9	99.8	98.9	300373
AMMECR1	72.2	99.0	94.0	300990
ANKH	118.6	100.0	99.7	118600
ANKRD11	96.3	97.4	94.1	148050
ANO5	142.2	99.5	95.9	166260
ARSB	117.5	94.9	87.7	253200
ARSE	102.1	99.2	93.0	302950
B3GALT6	47.5	76.4	71.7	615349
B3GAT3	93.6	99.4	95.9	245600
B4GALT7	104.3	96.1	95.0	130070

BMP1	143.9	99.9	99.1	614856
BMP2	173.4	100.0	99.9	617877112600
BMPER	159.9	99.9	99.0	608022
BMPR1B	172.4	100.0	98.9	609441
BRAF	74.4	87.6	77.2	613706
BRF1	100.0	96.6	92.9	616202
BTK	116.2	100.0	99.6	307200
C21orf2	104.4	99.9	98.7	602271
CA2	140.7	100.0	99.3	259730
CANT1	142.1	100.0	99.8	251450
CBL	129.8	96.9	95.7	613563
CCDC8	111.9	100.0	100.0	614205
CDC42	97.0	96.7	89.5	616737
CDC45	160.7	99.4	97.5	617063
CDC6	165.4	99.8	98.3	613805
CDKN1C	21.1	68.1	51.8	614732
CDT1	99.3	96.8	93.8	613804
CEP120	129.7	99.8	98.1	616300
CHST3	91.6	100.0	97.5	143095
CLCN5	134.6	99.6	98.0	300009
CLCN7	129.7	99.5	98.2	166600
COG1	124.2	100.0	99.9	611209
COL10A1	88.4	98.6	93.9	156500
COL11A1	90.8	94.9	89.6	228520
COL11A2	92.2	99.9	98.3	614524
COL1A1	134.9	98.1	96.3	114000
COL1A2	101.8	96.7	93.6	225320
COL2A1	103.4	99.9	99.0	200610
COL9A1	121.2	99.5	96.9	614135
COL9A2	65.1	98.3	88.8	614284

COL9A3	65.5	95.6	86.7	600969
COLEC11	203.0	100.0	100.0	265050
COMP	121.4	93.6	92.4	132400
CREB3L1	113.8	99.4	97.0	616229
CREBBP	123.5	99.4	96.7	180849
CRTAP	110.4	99.8	97.3	610682
CSGALNACT 1	193.1	100.0	100.0	-
CTSA	134.1	100.0	99.4	256540
CTSK	105.0	100.0	99.9	265800
CUL7	149.6	99.8	97.9	273750
CYP26B1	178.1	100.0	99.9	614416
CYP27B1	137.1	100.0	99.1	264700
DDR2	155.0	100.0	99.9	271665
DHCR24	183.0	100.0	100.0	602398
DLL3	64.1	88.8	79.9	277300
DLX3	109.8	100.0	99.1	104510
DMP1	159.5	99.9	99.1	241520
DONSON	104.9	83.9	78.3	251230;617604
DPM1	131.1	91.7	86.7	608799
DVL1	113.9	97.8	94.1	616331
DYM	101.3	97.2	94.8	223800
DYNC2H1	90.5	96.6	87.0	613091
EBP	83.3	100.0	98.0	302960
EFL1	174.8	99.4	97.7	617941
EIF2AK3	147.1	95.1	91.3	226980
ENPP1	134.8	92.4	83.2	208000
EVC	110.4	93.2	89.8	225500
EVC2	119.3	96.4	94.3	225500
EXT1	105.4	99.9	98.5	133700
EXT2	163.5	99.9	99.1	133701

EXTL3	206.4	100.0	100.0	617425
FAM111A	292.2	100.0	99.9	602361
FAM20C	101.3	100.0	98.9	259775
FBN1	159.8	99.9	99.5	102370
FERMT3	122.4	100.0	98.9	612840
FGD1	85.7	92.7	86.5	305400
FGF23	106.0	99.9	97.8	193100
FGF8	111.4	90.2	79.7	612702
FGF9	165.0	100.0	100.0	612961
FGFR1	148.0	99.7	98.3	101600
FGFR2	140.1	97.4	96.4	101600
FGFR3	110.2	99.6	97.0	187601
FIG4	154.9	99.8	98.4	612577
FKBP10	158.6	96.9	92.8	259450
FLNA	138.1	100.0	99.5	305620
FLNB	149.9	99.8	99.2	108720
FN1	145.4	100.0	99.5	-
FUCA1	135.0	100.0	99.5	230000
FZD2	176.6	98.3	94.8	-
GALNS	93.2	99.0	95.6	253000
GALNT3	128.2	99.2	96.0	211900
GDF3	134.9	100.0	100.0	613702
GDF5	141.8	100.0	100.0	201250
GDF6	75.2	98.7	89.0	118100
GH1	175.2	100.0	100.0	262400
GHR	212.3	99.8	99.5	604271;262500
GHRHR	116.4	95.3	94.7	612781
GHSR	206.0	99.9	98.4	615925
GJA1	246.4	100.0	100.0	218400;104100;257850;186100;164200
GLB1	94.3	99.6	97.0	230600;230500;230650;253010

GLI2	138.5	99.4	97.4	615849;610829
GLI3	154.2	100.0	99.7	146510;174700;175700;174200;241800
GMNN	101.2	92.6	83.8	616835
GNAS	141.0	98.5	95.9	103580;219080;166350;612463;612462;174800;603233
GNPAT	133.6	99.4	96.4	222765
GNPTAB	167.7	98.3	97.4	252500;252600
GNPTG	151.6	96.1	89.7	252605
GNS	107.9	96.9	92.0	252940
GORAB	176.3	99.7	97.8	231070
GPC6	142.0	100.0	100.0	258315
GPR161	196.8	100.0	100.0	-
GPX4	119.2	85.2	76.6	250220
GUSB	116.1	92.2	89.4	253220
HDAC4	111.9	99.9	99.3	600430
HES7	29.1	64.9	42.6	613686
HESX1	57.6	99.2	92.6	182230
HGSNAT	101.0	86.4	85.7	252930
HMGA2	78.8	84.0	76.5	-
HOXA13	49.0	69.2	61.7	176305;14000
HPGD	88.0	100.0	98.5	119900;259100
HRAS	164.7	99.8	98.1	218040
HSPA9	91.6	91.1	85.9	182170
HSPG2	121.3	99.4	98.2	255800;224410
HYLS1	171.1	100.0	100.0	236680
IDH1	97.9	92.5	81.7	166000;614875;614569
IDH2	103.5	99.6	96.9	166000;613657;614569
IDS	111.3	99.6	98.3	309900
IDUA	123.0	88.1	80.0	607015;607014;607016
IFITM5	63.4	99.4	94.9	610967
IFT122	152.0	100.0	99.9	218330

IFT140	114.7	99.9	99.0	266920
IFT172	116.5	100.0	99.6	615630
IFT43	114.8	100.0	100.0	614099
IFT80	57.8	87.6	70.7	611263
IGF1	122.5	100.0	100.0	608747
IGF1R	144.0	100.0	99.8	270450
IGF2	100.0	100.0	100.0	616489
IGFALS	79.7	99.9	96.8	615961
IGSF1	84.8	99.5	96.6	300888
IHH	129.3	100.0	100.0	112500;607778
IKBKB	123.5	98.5	94.2	615592
IKBKG	52.5	84.6	73.2	300301;300291;308300
IL2RG	65.2	99.8	97.3	300400
IMPAD1	147.2	99.9	99.4	614078
INPPL1	123.6	96.7	93.7	258480
KIAA0753	123.9	99.9	98.7	-
KIF22	163.1	100.0	99.9	603546
KIF7	85.7	93.5	88.9	607131;200990;614120
KMT2A	152.5	99.3	98.6	605130
KRAS	64.7	99.9	98.7	609942
LBR	87.8	93.3	83.9	169400;215140
LEMD3	96.7	95.4	88.8	155950;166700
LFNG	91.6	85.4	83.3	609813
LHX3	84.7	94.0	80.6	221750
LHX4	144.9	100.0	99.8	262700
LIFR	123.4	97.2	92.1	601559
LMX1B	111.4	97.0	92.3	161200
LONP1	141.5	97.9	96.4	600373
LRP4	166.6	99.1	98.9	212780;614305
LRP5	189.8	98.2	97.9	601813;144750;607634;166710;601884;259770;607636

LRRK1	154.1	98.9	97.1	615198
LTBP2	104.6	99.6	97.1	614819
LTBP3	113.5	98.7	94.7	601216
LZTR1	134.0	100.0	99.4	616564
MAN2B1	122.3	99.1	96.2	248500
MANBA	119.9	99.7	97.2	248510
MAP2K1	92.3	99.8	95.6	615279
MAP2K2	107.9	97.6	89.2	615280
MAP3K7	114.6	99.7	98.1	157800;617137
MATN3	116.6	84.7	84.7	607078;140600;608728
MEOX1	76.8	96.6	91.2	214300
MESP2	81.5	93.1	87.9	608681
MGP	132.0	92.7	91.6	245150
MMP13	124.1	93.5	91.5	250400;602111
MMP14	152.1	100.0	99.5	277950
MMP2	164.4	100.0	100.0	259600
MMP9	125.4	96.2	91.8	613073
MTAP	109.2	93.6	86.5	112250
MYH3	110.4	99.9	98.6	178110;193700;601680
NAGLU	108.7	92.4	90.4	616491;252920
NANS	106.1	100.0	99.9	610442
NBAS	145.3	99.5	97.6	614800
NEK1	103.2	98.1	93.0	263520
NEK9	136.8	99.7	98.5	617022;614262
NEU1	148.1	99.4	97.1	256550
NIN	143.5	99.7	98.6	614851
NKX3-2	55.8	92.4	73.9	613330
NOTCH2	172.4	100.0	99.9	102500
NPPC	87.8	99.8	96.7	-
NPR2	164.8	100.0	100.0	615923;616255;602875

NRAS	188.4	100.0	100.0	613224
NXN	86.0	99.7	96.5	-
OBSL1	140.5	99.8	98.5	612921
OFD1	51.5	84.0	67.8	300209;300804;311200
ORC1	106.7	99.9	98.9	224690
ORC4	57.6	95.8	82.0	613800
ORC6	126.8	100.0	100.0	613803
OSTM1	80.7	90.8	88.4	259720
OTX2	154.8	100.0	99.8	610125;613986
P3H1	137.0	100.0	99.9	610915
P4HB	105.1	94.6	94.4	112240
PAM16	50.7	65.2	64.7	613320
PAPPA2	175.4	100.0	99.8	-
PAPSS2	108.5	99.7	98.5	612847
PCNT	117.6	98.9	96.0	210720
PCYT1A	113.5	98.3	94.7	608940
PDE4D	101.2	92.8	88.3	614613
PEX5	111.7	99.9	98.3	616716;214110;202370
PEX7	113.5	89.6	82.0	215100
PHEX	125.0	99.9	98.0	307800
PHGDH	115.6	100.0	99.8	256520
PIK3R1	129.3	99.7	97.3	269880
PITX1	144.6	91.4	86.8	119800;186550
PITX2	147.8	99.7	97.5	180500
PLEKHM1	141.1	100.0	99.9	611497
PLK4	145.5	99.5	96.3	616171
PLOD2	108.6	94.7	88.6	609220
PLS3	131.5	96.8	95.2	300910
POC1A	133.8	100.0	100.0	614813
POP1	114.2	100.0	99.7	617396

POU1F1	106.3	98.2	94.7	613038
PPIB	118.4	100.0	100.0	259440
PPP1CB	96.6	99.6	98.4	617506
PRKAR1A	90.7	99.1	93.9	101800
PROKR2	331.8	100.0	100.0	244200
PROP1	76.5	91.6	84.3	262600
PSAT1	53.2	91.4	75.8	616038
PTDSS1	127.2	100.0	100.0	151050
PTH1R	108.5	99.9	98.8	215045
PTPN11	103.1	97.9	92.5	163950
RAB33B	233.9	100.0	100.0	615222
RAF1	127.3	100.0	99.7	611553
RASGRP2	97.5	99.9	98.6	615888
RBM8A	106.6	100.0	99.4	274000
RBPJ	89.2	94.1	86.4	614814
RIPPLY2	63.5	99.0	83.8	616566
RIT1	165.6	100.0	100.0	615355
RMRP				607095
RNPC3	41.3	85.1	61.1	-
RNU4ATAC				210710
ROR2	165.9	99.4	98.0	268310
RPGRIP1L	126.2	96.4	93.9	216360
RPL10	85.9	99.1	93.1	300998
RRAS	116.3	89.4	81.6	-
RSPRY1	168.9	100.0	99.9	616723
RUNX2	106.4	72.3	72.2	119600
SBDS	212.3	100.0	99.9	260400
SCARF2	82.5	85.2	73.6	600920
SEC24D	136.9	99.9	98.6	616294
SERPINF1	101.8	100.0	99.4	613982

SERPINH1	183.7	100.0	99.9	613848
SGSH	129.0	95.1	93.6	252900
SH3PXD2B	140.2	100.0	99.8	249420
SHOC2	140.4	100.0	99.4	607721
SHOX	29.1	73.5	61.4	249700;127300;300582
SLC10A7	110.0	100.0	98.9	-
SLC17A5	119.6	96.8	92.9	269920
SLC25A24	115.8	98.6	96.3	-
SLC26A2	233.2	100.0	100.0	226900
SLC29A3	203.6	99.9	99.5	602782
SLC34A3	105.6	98.9	94.5	241530
SLC35D1	115.4	95.7	90.4	269250
SLC39A13	114.8	99.8	98.0	612350
SLCO2A1	110.5	100.0	99.6	614441
SLCO5A1	169.1	99.8	97.7	600383
SMAD4	125.5	100.0	100.0	139210
SMARCAL1	134.6	100.0	99.9	242900
SNRPB	75.2	99.8	97.4	117650
SNX10	118.9	96.2	96.1	615085
SOS1	94.3	96.7	90.3	610733
SOS2	97.1	98.5	92.8	616559
SOST	112.6	100.0	99.6	122860
SOX2	128.8	98.3	93.1	206900
SOX3	37.7	86.4	71.5	300123
SOX9	134.0	97.8	93.8	114290
SP7	159.1	99.9	99.3	613849
SPARC	161.0	100.0	100.0	616507
SPINK5	145.0	99.4	96.5	256500
SPR	166.5	98.9	90.0	612716
SPRED1	164.3	98.7	96.7	611431

SRCAP	153.9	99.8	99.1	136140
STAT3	119.5	99.9	99.0	615952
STAT5B	130.6	99.7	97.2	245590
SULF1	164.2	100.0	99.9	600383
SUMF1	103.3	98.6	91.1	272200
TAPT1	83.5	88.5	85.9	616897
TBCE	128.0	99.9	98.2	241410
TBX15	131.7	100.0	99.4	260660
TBX4	170.7	94.9	92.8	147891
TBX6	122.1	91.5	79.7	122600
TBXAS1	140.3	100.0	100.0	231095
TCIRG1	113.5	95.4	89.4	259700
TCTEX1D2	126.1	100.0	99.3	617405
TCTN2	144.2	99.5	97.0	616654
TCTN3	127.6	100.0	99.8	614815
TGFB1	86.9	99.7	95.1	131300
TMEM165	113.9	99.8	98.1	614727
TMEM216	111.9	100.0	98.7	603194
TMEM231	111.5	100.0	99.9	615397
TMEM38B	114.3	100.0	99.4	615066
TNFRSF11A	146.3	93.3	91.4	174810
TNFRSF11B	224.8	100.0	100.0	239000
TNFSF11	150.4	99.3	93.2	259710
TRAPPC2	85.3	87.0	66.7	313400
TRIP11	84.3	95.2	87.4	200600
TRPS1	175.0	100.0	99.8	190350
TRPV4	172.4	99.5	98.7	113500
TTC21B	100.7	99.7	97.6	613820
VDR	123.3	98.0	95.2	277440
WDR19	132.1	99.8	98.1	614377

WDR34	106.6	99.5	96.2	615633
WDR35	145.1	99.3	97.7	614091
WDR60	114.2	99.1	96.3	615503
WISP3	118.4	100.0	100.0	208230
WNT1	188.8	100.0	99.9	615220
WNT5A	155.7	100.0	100.0	180700
XRCC4	103.2	99.7	97.3	616541
XYLT1	132.5	90.4	87.1	615777
XYLT2	136.3	98.9	94.9	605822
ZBTB16	151.4	100.0	100.0	612447
ZMPSTE24	113.3	100.0	99.1	608612

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.

Median Coverage describes the average number of reads seen across 50 exomes.

% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.

% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.

Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors