

WES CRANIOFACIAL ANOMALIES DG 2.16

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ACP4	91.2	96.8	88.5	617297
ADAMTSL4	122.7	100.0	99.6	225100
ALX1	134.2	99.9	98.5	613456
ALX3	134.6	91.1	79.0	136760
ALX4	157.1	100.0	100.0	168500
AMBN	175.1	99.3	97.1	616270
AMELX	85.7	98.9	93.5	301200
AMER1	98.2	99.8	98.9	300373
AMTN	119.3	100.0	99.1	617607
ANKRD11	119.6	99.2	97.1	148050
ARHGAP29	138.7	99.7	98.8	8
AXIN2	124.2	100.0	99.9	608615
BCOR	102.7	98.8	95.3	300166
BMP2	163.4	100.0	100.0	7
BMP4	173.4	100.0	100.0	607932
C4orf26	206.1	100.0	100.0	614832
CDC45	138.9	99.6	98.1	617063;1
CDON	107.0	100.0	99.0	614226
CDSN	131.0	100.0	100.0	602593
CHD7	137.0	99.9	99.4	214800
COL11A1	96.6	97.9	94.0	604841;154780
COL11A2	111.6	100.0	99.4	277610;215150
COL2A1	112.2	100.0	99.7	108300
COL9A1	132.3	100.0	99.7	614135
COL9A2	95.2	99.9	98.8	600204;614284

COL9A3	107.8	99.6	96.8	120270
CTSK	86.3	100.0	99.8	265800
DHODH	98.8	100.0	100.0	263750
DISP1	164.3	99.9	99.7	2
DLX3	146.7	100.0	99.0	104510
DLX4	228.4	100.0	100.0	3
DSPP	79.0	98.4	93.8	125490
EDA	102.0	95.6	85.7	313500;305100
EDAR	126.6	100.0	100.0	129490;224900
EDARADD	89.8	99.7	98.3	129490;224900
EDN1	156.5	100.0	99.9	615706
EDNRA	150.7	100.0	99.8	-
EFNA4	152.5	100.0	100.0	601380
EFNB1	116.7	100.0	99.9	304110
EFTUD2	103.2	100.0	99.2	610536
EIF4A3	87.5	100.0	99.4	268305
ENAM	139.5	100.0	100.0	104500
ERF	146.8	100.0	99.2	600775
EYA1	120.2	99.9	99.8	113650
EZH2	130.0	99.4	97.6	277590
FAM20A	111.1	100.0	99.4	204690
FAM83H	120.2	100.0	99.9	130900
FGD1	86.7	98.4	93.0	305400
FGF10	120.5	100.0	99.6	149730
FGF3	139.5	100.0	100.0	610706
FGF8	130.0	97.9	86.8	4
FGF9	153.5	100.0	100.0	612961
FGFR1	122.6	100.0	99.6	123150
FGFR2	113.1	97.7	96.8	123500
FGFR3	138.5	100.0	99.6	101400

FOXC1	80.4	99.7	97.5	602482;601631
FOXE1	87.5	100.0	99.7	241850
GDF3	127.9	100.0	100.0	613702
GDF6	156.3	100.0	100.0	118100
GJA1	156.2	100.0	100.0	121014
GJB6	140.9	100.0	100.0	129500
GLI2	158.2	100.0	100.0	610829
GLI3	139.5	100.0	99.3	175700
GNAI3	88.3	98.9	94.5	602483
GPR68	165.4	99.9	99.0	617217
GRHL3	133.2	100.0	99.8	606713
GSC	133.8	100.0	98.9	602471
HOXA2	83.0	100.0	99.9	612290
HUWE1	79.3	99.1	94.3	300706
HYAL2	175.3	100.0	99.8	9
IFT122	120.5	99.9	99.0	218330
IFT43	112.4	100.0	100.0	614099
IFT88	94.7	99.6	97.4	10
IKBKG	60.1	88.1	78.8	300301;300291
IL11RA	131.5	100.0	99.6	614188
INTU	115.4	99.8	98.7	617926
IRF6	90.3	99.4	95.0	119300
ITGB6	127.5	96.7	95.0	616221
KAT6B	155.7	99.9	99.1	603736;606170
KDF1	110.3	100.0	99.9	617337
KDM1A	130.9	100.0	98.8	616728
KDM6A	97.7	95.3	87.8	300867
KLK4	164.3	100.0	100.0	204700
KMT2D	136.2	100.0	99.7	147920
KREMEN1	143.4	99.7	97.3	609898

LAMB3	116.9	100.0	99.4	104530
LRP2	139.2	100.0	99.9	222448
LRP6	136.8	99.9	99.2	5
LTBP3	147.5	100.0	99.6	613097
MASP1	131.1	100.0	99.3	257920
MED12	85.1	99.5	95.5	309520;300895;305450
MEGF8	144.0	100.0	99.5	614976
MEIS2	123.6	100.0	99.8	600987
MEOX1	105.0	99.9	97.4	214300
MID1	124.1	99.8	97.4	300000
MITF	141.1	100.0	99.8	193510
MMP20	90.8	99.8	97.6	612529
MSX1	143.3	99.9	98.6	189500
MSX2	101.1	100.0	100.0	168500
NAA10	105.0	100.0	98.8	300013
NECTIN1	134.0	100.0	99.9	225060
NFKBIA	134.6	95.3	89.4	612132
NIPBL	124.9	98.8	96.9	122470
NOG	233.6	100.0	100.0	186500
NSD1	147.0	100.0	99.8	117550
OFD1	51.9	85.8	70.8	311200
OTX2	127.4	100.0	99.3	610125
PAX3	106.9	100.0	99.7	193500
PAX6	116.5	100.0	99.8	602482
PAX7	131.1	100.0	100.0	268220
PAX9	236.1	99.8	99.6	604625
PGM1	128.8	100.0	99.8	614921
PITX2	164.8	100.0	99.5	180500
PLCB4	102.5	99.8	98.0	614669
POLR1C	98.3	98.9	94.9	248390

POLR1D	183.1	91.6	91.6	613717
PORCN	111.2	99.9	98.8	305600
PTCH1	110.2	99.9	98.4	109400
PTH1R	106.6	100.0	99.1	125350
RAB23	107.4	100.0	99.2	201000
RAD21	83.0	97.8	93.4	614701
RBM10	111.6	99.8	97.6	311900
RECQL4	159.9	100.0	99.8	603780
RIPK4	167.5	100.0	100.0	-
RUNX2	102.8	73.4	72.2	119600
SALL1	113.3	99.9	98.9	107480
SALL4	135.0	99.9	98.1	607323
SATB2	107.4	99.8	97.7	612313
SEMA3E	130.9	100.0	99.6	214800
SF3B4	75.5	99.9	98.3	154400
SH3BP2	139.3	91.9	91.4	118400
SHH	147.1	100.0	100.0	611638;147250
SIX1	131.1	99.9	98.7	608389
SIX3	206.0	100.0	99.9	157170
SKI	132.9	100.0	99.3	182212
SLC24A4	103.5	100.0	99.8	615887
SLC26A2	205.1	100.0	99.9	256050
SMAD6	180.5	98.8	89.1	6
SMC1A	87.8	99.9	97.8	300590
SMC3	84.0	96.0	89.7	610759
SMO	140.4	99.9	98.3	601707
SMOC2	88.7	77.0	75.7	125400
SNAI2	102.7	99.9	99.1	608890
SOX10	88.2	100.0	99.1	613266
SOX6	91.8	99.9	98.5	607257

SOX9	159.9	100.0	100.0	114290
SPECC1L	127.5	100.0	99.8	600251
SUMO1	20.0	62.3	46.0	613705
TBX1	101.2	93.0	86.9	192430
TBX22	105.0	99.4	94.8	303400
TCF12	137.7	99.9	99.9	615314
TCOF1	111.6	99.9	99.1	154500
TFAP2A	112.7	99.8	98.0	113620
TGFBR1	156.4	95.4	93.8	609192
TGFBR2	156.8	100.0	100.0	610168
TGIF1	141.2	100.0	100.0	142946
TP63	162.8	100.0	100.0	604292
TRAF6	75.0	96.3	85.8	602355
TSHZ1	147.6	98.9	98.7	607842
TSPEAR	139.3	100.0	99.8	618180
TWIST1	160.2	100.0	99.6	101400
UBB	37.6	100.0	93.3	119540
VAX1	95.1	99.5	95.7	614402
WDR19	126.8	100.0	99.2	614378
WDR35	141.8	99.7	98.4	613610
WDR72	123.8	96.8	96.1	613211
WNT10A	141.8	100.0	99.9	224750;257980
WNT10B	157.0	100.0	100.0	617073
ZEB2	140.1	99.7	98.4	235730
ZIC1	279.6	100.0	100.0	616602
ZIC2	165.6	97.5	95.4	609637

Gene symbols used follow HGCN 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