

# WES CRANIOFACIAL ANOMALIES DG 2.14

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ADAMTSL4	90.6	99.9	98.8	225100
ALX1	153.2	99.9	98.4	613456
ALX3	102.7	73.3	70.9	136760
ALX4	132.7	98.4	92.5	168500
AMELX	98.2	99.0	95.0	301200
ANKRD11	96.3	97.4	94.1	148050
ARHGAP29	136.4	98.9	95.0	8
AXIN2	114.5	99.7	98.9	608615
BCOR	109.7	99.3	96.8	300166
BMP2	173.4	100.0	99.9	7
BMP4	151.7	100.0	99.9	607932
C4orf26	197.6	100.0	100.0	614832
CDC45	160.7	99.4	97.5	1
CDON	143.8	100.0	99.6	614226
CDSN	119.3	100.0	99.5	602593
CHD7	150.7	99.9	98.9	214800
COL11A1	90.8	94.9	89.6	604841;154780
COL11A2	92.2	99.9	98.3	277610;215150
COL2A1	103.4	99.9	99.0	108300
COL9A1	121.2	99.5	96.9	614135
COL9A2	65.1	98.3	88.8	600204;614284
COL9A3	65.5	95.6	86.7	120270
CTSK	105.0	100.0	99.9	265800
DHODH	92.2	100.0	99.9	263750
DISP1	211.4	99.9	99.1	2

DLX3	109.8	100.0	99.1	104510
DLX4	162.4	100.0	100.0	3
DSPP	155.7	99.9	99.3	125490
EDA	88.5	85.7	77.3	313500;305100
EDAR	138.6	100.0	99.6	129490;224900
EDARADD	99.1	99.3	93.3	129490;224900
EDN1	145.5	100.0	100.0	615706
EDNRA	218.9	100.0	99.7	-
EFNA4	106.6	100.0	100.0	601380
EFNB1	118.5	100.0	99.9	304110
EFTUD2	124.2	100.0	99.4	610536
EIF4A3	106.8	100.0	99.9	268305
ENAM	148.9	100.0	99.9	104500
ERF	107.9	99.9	97.7	600775
EYA1	144.2	100.0	99.7	113650
EZH2	139.5	99.8	97.6	277590
FAM83H	76.7	94.9	87.7	130900
FGD1	85.7	92.7	86.5	305400
FGF10	142.2	100.0	100.0	149730
FGF3	73.9	92.0	75.7	610706
FGF8	111.4	90.2	79.7	4
FGFR1	148.0	99.7	98.3	123150
FGFR2	140.1	97.4	96.4	123500
FGFR3	110.2	99.6	97.0	101400
FOXC1	32.7	86.0	68.5	602482;601631
FOXE1	29.3	72.3	56.2	241850
GDF3	134.9	100.0	100.0	613702
GDF6	75.2	98.7	89.0	118100
GJA1	246.4	100.0	100.0	121014
GJB6	185.4	100.0	100.0	129500

GLI2	138.5	99.4	97.4	610829
GLI3	154.2	100.0	99.7	175700
GNAI3	110.6	99.0	92.0	602483
GRHL3	140.7	100.0	99.9	606713
GSC	85.4	86.9	74.5	602471
HOXA2	74.1	99.5	95.6	612290
HUWE1	98.4	99.2	97.0	300706
HYAL2	223.2	100.0	100.0	9
IFT122	152.0	100.0	99.9	218330
IFT43	114.8	100.0	100.0	614099
IFT88	78.1	98.1	90.7	10
IKBKG	52.5	84.6	73.2	300301;300291
IL11RA	139.9	100.0	99.5	614188
INTU	122.0	99.7	96.6	617926
IRF6	113.7	99.9	97.9	119300
KAT6B	192.3	99.6	98.5	603736;606170
KDM1A	129.7	96.2	93.4	616728
KDM6A	109.0	93.2	84.3	300867
KLK4	185.1	100.0	98.8	204700
KMT2D	142.1	99.9	99.0	147920
KREMEN1	149.1	94.7	93.6	609898
LRP2	176.3	100.0	99.8	222448
LRP6	169.3	100.0	99.7	5
LTBP3	113.5	98.7	94.7	613097
MASP1	148.6	100.0	99.6	257920
MED12	105.7	98.0	94.8	309520;300895;305450
MEGF8	127.6	99.9	98.6	614976
MEOX1	76.8	96.6	91.2	214300
MID1	164.6	99.8	98.4	300000
MITF	155.5	100.0	99.9	193510

MMP20	100.5	100.0	98.6	612529
MSX1	75.2	95.4	87.5	189500
MSX2	94.2	98.0	85.5	168500
NAA10	102.4	98.7	96.7	300013
NECTIN1	145.4	100.0	100.0	225060
NFKBIA	116.3	98.5	93.8	612132
NIPBL	116.1	96.5	94.5	122470
NOG	191.9	100.0	100.0	186500
NSD1	155.2	100.0	99.9	117550
OFD1	51.5	84.0	67.8	311200
OTX2	154.8	100.0	99.8	610125
PAX3	118.5	100.0	100.0	193500
PAX6	119.9	100.0	99.9	602482
PAX7	117.8	100.0	100.0	268220
PAX9	238.8	99.6	99.3	604625
PITX2	147.8	99.7	97.5	180500
PLCB4	126.1	99.2	95.7	614669
POLR1C	117.0	99.7	96.1	248390
POLR1D	176.2	91.6	91.6	613717
PORCN	117.7	100.0	99.3	305600
PTCH1	114.6	98.4	95.9	109400
PTH1R	108.5	99.9	98.8	125350
RAB23	110.3	99.7	98.0	201000
RAD21	78.5	98.8	94.7	614701
RECQL4	149.6	99.2	96.5	603780
RIPK4	163.3	100.0	99.6	-
RUNX2	106.4	72.3	72.2	119600
SALL1	138.5	99.3	98.4	107480
SALL4	147.5	97.6	96.3	607323
SATB2	110.5	98.5	93.4	612313

SEMA3E	142.6	99.9	99.0	214800
SF3B4	89.5	99.8	97.7	154400
SH3BP2	110.9	91.4	91.4	118400
SHH	117.5	99.0	94.0	611638;147250
SIX1	117.3	99.7	97.6	608389
SIX3	145.3	100.0	98.9	157170
SKI	85.3	96.4	90.8	182212
SMAD6	100.5	80.0	72.0	6
SMC1A	99.4	99.9	98.8	300590
SMC3	81.4	93.8	87.6	610759
SMOC2	91.5	75.4	72.6	125400
SNAI2	129.8	100.0	99.8	608890
SOX10	65.8	98.2	91.3	613266
SOX6	102.7	99.7	97.9	607257
SPECC1L	157.9	100.0	100.0	600251
SUMO1	17.6	58.8	37.0	613705
TBX22	121.8	99.2	96.3	303400
TCF12	150.3	100.0	99.8	615314
TCOF1	98.6	99.5	97.3	154500
TFAP2A	109.3	100.0	99.3	113620
TGFBR1	173.4	93.7	93.6	609192
TGFBR2	193.5	100.0	99.9	610168
TGIF1	138.3	100.0	100.0	142946
TP63	206.3	100.0	100.0	604292
TRAF6	106.9	96.1	87.1	602355
TSHZ1	166.6	98.8	98.5	607842
TWIST1	134.4	96.6	87.2	101400
UBB	61.9	100.0	99.7	119540
VAX1	52.2	88.4	78.0	614402
WDR19	132.1	99.8	98.1	614378

WDR35	145.1	99.3	97.7	613610
WDR72	132.2	96.5	95.4	613211
WNT10A	114.0	100.0	99.1	224750;257980
ZEB2	157.0	99.8	98.8	235730
ZIC1	231.1	100.0	100.0	616602
ZIC2	122.5	90.5	78.9	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*