WES CRANIOFACIAL ANOMALIES DG 3.1

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACP4	97.1	97.2	88.8	617297
ADAMTSL4	134.5	100.0	99.2	225100
ALX1	169.2	99.7	97.1	613456
ALX3	124.9	77.9	73.3	136760
ALX4	137.7	100.0	99.3	168500
AMBN	192.8	99.8	98.5	616270
AMELX	96.4	99.9	96.8	301200
AMER1	108.0	99.9	98.5	300373
AMTN	137.6	99.6	98.6	617607
ANKRD11	118.7	96.1	93.5	148050
ARHGAP29	168.5	99.5	98.0	8
AXIN2	140.7	100.0	99.9	608615
BCOR	117.1	99.6	97.4	300166
BMP2	178.0	100.0	100.0	-
BMP4	178.6	100.0	100.0	607932
CCBE1	81.2	99.8	98.8	No OMIM phenotype
CDC45	162.6	99.8	98.5	617063
CDON	132.5	100.0	99.6	614226
CDSN	146.4	100.0	100.0	602593
CHD7	158.7	100.0	99.5	214800
COL11A1	113.4	96.2	92.8	154780;604841
COL11A2	127.0	100.0	99.7	215150;277610
COL2A1	125.9	100.0	99.7	108300
COL9A1	159.8	100.0	99.2	614135
COL9A2	103.5	99.9	99.0	614284;600204

COL9A3	109.1	98.7	95.5	120270
COLEC11	178.1	100.0	100.0	No OMIM phenotype
CTSK	105.3	100.0	99.9	265800
CYP26B1	148.9	100.0	99.9	No OMIM phenotype
DHODH	112.7	100.0	100.0	263750
DISP1	190.7	100.0	99.9	2
DLX3	135.0	99.9	98.4	104510
DLX4	258.2	100.0	100.0	3
DSPP	67.8	96.8	86.1	125490
EDA	102.9	98.1	91.6	305100;313500
EDAR	148.4	100.0	99.9	129490;224900
EDARADD	101.9	99.9	98.8	129490;224900
EDN1	166.4	100.0	100.0	615706
EDNRA	196.2	100.0	100.0	616367
EFNA4	150.9	100.0	100.0	601380
EFNB1	142.1	100.0	100.0	304110
EFTUD2	123.9	100.0	99.8	610536
EIF4A3	106.3	100.0	99.5	268305
ENAM	166.6	100.0	100.0	104500
ERF	144.8	99.9	98.5	600775
ESCO2	139.5	98.7	95.2	268300
EYA1	145.7	99.9	99.7	113650
EZH2	171.4	100.0	99.5	277590
FAM20A	113.3	99.6	94.7	204690
FAM83H	76.6	84.9	81.5	130900
FGD1	91.2	97.3	92.8	305400
FGF10	155.3	100.0	99.8	149730
FGF3	110.3	99.8	95.1	610706
FGF8	127.7	98.2	88.9	4
FGF9	225.5	100.0	100.0	612961

FGFR1	137.7	100.0	99.9	123150
FGFR2	134.1	97.7	97.1	123500
FGFR3	122.6	99.8	97.7	101400
FLNA	138.5	100.0	99.9	No OMIM phenotype
FLNB	140.5	99.5	98.8	No OMIM phenotype
FOXC1	55.4	98.0	89.6	602482;601631
FOXE1	42.5	96.9	78.5	241850
GDF3	138.9	100.0	100.0	613702
GDF6	116.8	100.0	99.9	118100
GJA1	187.7	100.0	100.0	121014
GJB6	145.5	100.0	100.0	129500
GLI2	155.7	99.1	97.4	610829
GLI3	140.8	98.5	98.0	175700
GNAI3	108.8	99.3	95.2	602483
GNPTAB	167.2	100.0	99.9	No OMIM phenotype
GPR68	138.4	99.5	96.7	617217
GRHL3	156.5	100.0	100.0	606713
GSC	103.3	99.2	92.4	602471
HOXA2	86.8	100.0	99.9	612290
HUWE1	94.5	99.2	95.8	300706
HYAL2	175.3	100.0	100.0	9
IFT122	140.5	100.0	99.6	218330
IFT43	134.3	100.0	100.0	614099
IFT88	112.0	99.6	97.3	10
IKBKG	62.9	84.1	77.2	300291;300301
IL11RA	155.5	100.0	99.9	614188
IL6ST	112.4	96.4	90.3	No OMIM phenotype
IMPAD1	157.7	100.0	100.0	No OMIM phenotype
INTU	141.9	99.7	98.1	617926
IRF6	91.7	99.6	95.9	119300

ITGB6	158.3	97.2	95.8	616221
KAT6B	171.3	99.6	98.3	606170;603736
KDF1	111.2	100.0	99.8	617337
KDM1A	149.1	98.2	95.3	616728
KDM6A	114.8	96.1	88.7	300867
KLK4	182.0	100.0	100.0	204700
KMT2D	142.1	100.0	99.4	147920
KREMEN1	162.3	97.7	94.4	609898
LAMB3	124.1	100.0	99.6	104530
LRP2	171.0	100.0	99.9	222448
LRP6	171.7	100.0	99.9	5
LTBP3	132.5	99.6	98.1	613097
MASP1	146.6	100.0	99.9	257920
MED12	101.2	99.8	96.7	300895;305450;309520
MEGF8	138.3	99.9	99.0	614976
MEIS2	149.4	100.0	100.0	600987
MEOX1	113.3	100.0	98.9	214300
MID1	159.2	99.8	98.7	300000
MITF	160.6	100.0	99.9	193510
MMP20	118.4	100.0	100.0	612529
MN1	108.4	100.0	99.3	No OMIM phenotype
MSX1	96.5	96.9	89.3	189500
MSX2	90.4	100.0	99.4	168500
NAA10	116.8	99.7	98.5	300013
NECTIN1	135.3	100.0	99.9	225060
NFKBIA	132.6	95.2	88.0	612132
NIPBL	142.8	98.9	97.0	122470
NOG	175.6	100.0	100.0	186500
NSD1	175.5	100.0	99.9	117550
ODAPH	279.7	100.0	100.0	614832

OFD1	61.2	88.0	73.7	311200
OTX2	152.3	100.0	99.7	610125
P4HB	119.0	94.6	94.0	No OMIM phenotype
PAX3	114.5	100.0	99.9	193500
PAX6	141.4	100.0	100.0	602482
PAX7	135.1	100.0	100.0	268220
PAX9	210.3	99.7	99.6	604625
PGM1	147.7	94.2	94.2	614921
PITX2	158.5	99.9	97.7	180500
PLCB4	117.7	99.9	98.8	614669
POLR1C	102.8	90.5	87.0	248390
POLR1D	210.1	91.6	91.6	613717
POR	175.0	99.8	98.6	No OMIM phenotype
PORCN	121.0	100.0	99.1	305600
PTCH1	127.2	99.2	97.6	109400
PTH1R	104.5	100.0	98.7	125350
RAB23	130.8	100.0	99.5	201000
RAD21	103.0	99.2	96.6	614701
RBM10	121.7	99.5	97.1	311900
RECQL4	149.8	99.8	98.1	603780
RIPK4	144.6	100.0	99.9	263650
RUNX2	113.5	72.2	72.2	119600
SALL1	133.4	99.9	99.0	107480
SALL4	136.9	98.6	96.7	607323
SATB2	126.5	99.7	97.4	612313
SCARF2	74.5	95.4	86.2	No OMIM phenotype
SEC24D	160.3	100.0	99.7	No OMIM phenotype
SEMA3E	158.1	99.2	98.9	214800
SF3B4	69.4	99.9	97.3	154400
SH3BP2	152.8	91.4	91.2	118400

SHH	128.7	100.0	99.5	147250;611638
SIX1	137.0	100.0	99.2	608389
SIX3	154.3	99.9	98.6	157170
SKI	101.8	99.3	94.9	182212
SLC24A4	123.9	100.0	99.8	615887
SLC26A2	234.0	100.0	100.0	256050
SMAD6	149.8	90.9	81.0	6
SMC1A	102.0	100.0	98.7	300590
SMC3	91.6	95.2	91.0	610759
SMO	139.7	97.8	94.7	601707
SMOC2	101.1	76.8	76.6	125400
SNAI2	115.6	100.0	99.1	608890
SOX10	70.3	99.9	97.9	613266
SOX6	123.0	99.9	99.4	607257
SOX9	161.0	100.0	98.6	114290
SPECC1L	135.8	96.0	95.7	600251
SUMO1	23.7	67.2	49.9	613705
TBX1	89.7	87.0	77.5	192430
TBX22	142.6	99.2	95.7	303400
TCF12	155.4	100.0	99.9	615314
TCOF1	123.4	99.7	98.6	154500
TFAP2A	119.1	99.4	94.3	113620
TGFBR1	181.8	93.7	93.6	609192
TGFBR2	171.6	100.0	100.0	610168
TGIF1	160.4	100.0	100.0	142946
TLK2	108.4	99.1	95.1	No OMIM phenotype
TP63	188.5	100.0	100.0	604292
TRAF6	92.6	97.1	88.9	602355
TSHZ1	155.0	98.8	98.8	607842
TSPEAR	160.3	100.0	99.2	618180

TWIST1	96.4	100.0	98.9	101400
UBB	49.1	100.0	99.4	119540
VAX1	97.2	97.5	91.5	614402
WDR19	153.1	100.0	99.4	614378
WDR35	172.4	99.8	98.9	613610
WDR72	151.9	96.8	96.4	613211
WNT10A	118.7	100.0	99.4	224750;257980
WNT10B	144.0	100.0	99.4	617073
ZEB2	158.7	99.9	99.1	235730
ZIC1	219.9	100.0	100.0	616602
ZIC2	110.1	100.0	98.7	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 10x 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