

# WES CRANIOFACIAL ANOMALIES DG 3.3

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ACP4	212.3	100.0	100.0	617297
ACTG1	262.0	100.0	100.0	No OMIM phenotype
ADAMTSL4	186.5	100.0	100.0	225100
ALX1	179.4	100.0	100.0	613456
ALX3	167.5	100.0	100.0	136760
ALX4	200.3	100.0	100.0	168500
AMBN	171.2	100.0	100.0	616270
AMELX	214.2	100.0	100.0	301200
AMER1	236.7	100.0	100.0	300373
AMTN	164.1	100.0	100.0	617607
ANKRD11	185.4	100.0	100.0	148050
ARHGAP29	154.1	100.0	100.0	-
AXIN2	235.9	100.0	100.0	608615
BCOR	213.5	100.0	100.0	300166
BMP2	190.7	100.0	100.0	-
BMP4	208.6	100.0	100.0	607932
CCBE1	158.4	100.0	100.0	No OMIM phenotype
CDC45	161.2	100.0	100.0	617063
CDON	157.4	100.0	100.0	614226
CDSN	345.7	100.0	100.0	602593
CHD7	180.1	100.0	100.0	214800
COL11A1	143.0	100.0	100.0	154780;604841
COL11A2	264.7	100.0	100.0	215150;277610
COL2A1	167.0	100.0	100.0	108300
COL9A1	155.8	100.0	100.0	614135

COL9A2	174.2	100.0	100.0	614284;600204
COL9A3	188.4	100.0	100.0	120270
COLEC11	203.2	100.0	100.0	No OMIM phenotype
CTSK	170.6	100.0	100.0	265800
CYP26B1	215.6	100.0	100.0	No OMIM phenotype
DHODH	164.7	100.0	100.0	263750
DISP1	182.2	100.0	100.0	-
DLX3	200.8	100.0	100.0	104510
DLX4	205.2	100.0	100.0	616788
DSPP	284.1	100.0	100.0	125490
EDA	194.5	100.0	100.0	305100;313500
EDAR	175.0	100.0	100.0	129490;224900
EDARADD	172.2	100.0	100.0	129490;224900
EDN1	134.7	100.0	100.0	615706
EDNRA	173.8	100.0	100.0	616367
EFNA4	192.6	100.0	100.0	601380
EFNB1	192.2	100.0	100.0	304110
EFTUD2	141.6	100.0	100.0	610536
EIF4A3	154.1	100.0	100.0	268305
ENAM	177.9	100.0	100.0	104500
ERF	211.9	100.0	100.0	600775
ESCO2	137.2	100.0	100.0	268300
EYA1	155.5	100.0	100.0	113650
EZH2	179.2	100.0	100.0	277590
FAM20A	163.0	100.0	100.0	204690
FAM83H	278.1	100.0	100.0	130900
FGD1	194.7	100.0	100.0	305400
FGF10	167.2	100.0	100.0	149730
FGF3	240.5	100.0	100.0	610706
FGF8	183.9	100.0	100.0	612702

FGF9	152.5	100.0	100.0	612961
FGFR1	220.5	100.0	100.0	123150
FGFR2	200.5	100.0	100.0	123500
FGFR3	257.6	100.0	100.0	101400
FLNA	204.7	100.0	100.0	No OMIM phenotype
FLNB	168.1	100.0	100.0	No OMIM phenotype
FOXC1	210.5	100.0	100.0	602482;601631
FOXE1	185.4	100.0	100.0	241850
GDF3	191.3	100.0	100.0	613702
GDF6	171.8	100.0	100.0	118100
GJA1	204.2	100.0	100.0	121014
GJB6	181.1	100.0	100.0	129500
GLI2	218.7	100.0	100.0	610829
GLI3	193.7	100.0	100.0	175700
GNAI3	140.1	100.0	100.0	602483
GNPTAB	163.6	100.0	100.0	No OMIM phenotype
GPR68	231.4	100.0	100.0	617217
GRHL3	155.1	100.0	100.0	606713
GSC	158.9	100.0	100.0	602471
HOXA2	189.0	100.0	100.0	612290
HUWE1	160.2	100.0	100.0	300706
HYAL2	195.8	100.0	100.0	-
IFT122	157.2	100.0	100.0	218330
IFT43	145.7	100.0	100.0	614099
IFT88	131.6	100.0	100.0	-
IKBKG	179.9	100.0	100.0	300291;300301
IL11RA	175.2	100.0	100.0	614188
IL6ST	168.1	100.0	100.0	No OMIM phenotype
IMPAD1	190.4	100.0	100.0	No OMIM phenotype
INTU	145.8	100.0	100.0	617926

IRF6	179.6	100.0	100.0	119300
ITGB6	153.1	100.0	100.0	616221
KAT6B	217.3	100.0	100.0	606170;603736
KDF1	188.7	100.0	100.0	617337
KDM1A	146.7	100.0	100.0	616728
KDM6A	172.8	100.0	100.0	300867
KLK4	169.6	100.0	100.0	204700
KMT2D	223.4	100.0	100.0	147920
KREMEN1	161.7	100.0	100.0	609898
LAMB3	183.1	100.0	100.0	104530
LRP2	164.3	100.0	100.0	222448
LRP6	171.4	100.0	100.0	616724
LTBP3	216.1	100.0	100.0	613097
MASP1	181.0	100.0	100.0	257920
MED12	186.8	100.0	100.0	300895;305450;309520
MEGF8	200.9	100.0	100.0	614976
MEIS2	178.4	100.0	100.0	600987
MEOX1	197.3	100.0	100.0	214300
MID1	183.7	100.0	100.0	300000
MITF	212.8	100.0	100.0	193510
MMP20	146.6	100.0	100.0	612529
MN1	278.5	100.0	100.0	No OMIM phenotype
MSX1	227.3	100.0	100.0	189500
MSX2	195.8	100.0	100.0	168500
NAA10	179.0	100.0	100.0	300013
NECTIN1	189.3	100.0	100.0	225060
NFKBIA	204.8	100.0	100.0	612132
NIPBL	150.3	100.0	100.0	122470
NOG	216.5	100.0	100.0	186500
NSD1	211.8	100.0	100.0	117550

ODAPH	167.8	100.0	100.0	614832
OFD1	128.6	100.0	100.0	311200
OTX2	197.9	100.0	100.0	610125
P4HB	158.4	100.0	100.0	No OMIM phenotype
PAX3	208.8	100.0	100.0	193500
PAX6	193.9	100.0	100.0	602482
PAX7	237.8	100.0	100.0	268220
PAX9	217.3	100.0	100.0	604625
PGM1	151.5	94.2	94.2	614921
PITX2	179.6	100.0	100.0	180500
PLCB4	145.9	100.0	100.0	614669
POLR1C	131.1	83.2	82.8	248390
POLR1D	164.7	100.0	100.0	613717
POR	197.4	100.0	100.0	No OMIM phenotype
PORCN	171.6	100.0	100.0	305600
PTCH1	222.9	100.0	100.0	109400
PTH1R	185.4	100.0	100.0	125350
RAB23	136.4	100.0	100.0	201000
RAD21	171.5	100.0	100.0	614701
RBM10	196.1	100.0	100.0	311900
RECQL4	280.9	100.0	100.0	603780
RIPK4	225.5	100.0	100.0	263650
RUNX2	197.8	100.0	100.0	119600
SALL1	207.4	100.0	100.0	107480
SALL4	193.7	100.0	100.0	607323
SATB2	184.8	100.0	100.0	612313
SCARF2	202.8	100.0	100.0	No OMIM phenotype
SEC24D	157.4	100.0	100.0	No OMIM phenotype
SEMA3E	153.5	100.0	100.0	214800
SF3B2	180.8	100.0	100.0	34344887

SF3B4	177.5	100.0	100.0	154400
SH3BP2	163.2	99.6	98.8	118400
SHH	212.2	100.0	100.0	147250;611638
SIX1	199.4	100.0	100.0	608389
SIX3	224.2	100.0	100.0	157170
SIX5	211.5	100.0	100.0	No OMIM phenotype
SKI	168.6	100.0	100.0	182212
SLC24A4	157.7	100.0	100.0	615887
SLC26A2	178.4	100.0	100.0	256050
SMAD6	186.5	100.0	100.0	617439
SMC1A	161.6	100.0	100.0	300590
SMC3	136.3	100.0	100.0	610759
SMO	224.2	100.0	100.0	601707
SMOC2	164.6	100.0	100.0	125400
SNAI2	181.9	100.0	100.0	608890
SOX10	241.5	100.0	100.0	613266
SOX6	148.0	100.0	100.0	607257
SOX9	253.7	100.0	100.0	114290
SPECC1L	157.0	97.8	96.1	600251
SUMO1	101.7	69.4	69.4	613705
TBX1	144.8	97.3	94.9	192430
TBX22	202.4	100.0	100.0	303400
TCF12	166.1	100.0	100.0	615314
TCOF1	175.6	100.0	100.0	154500
TFAP2A	180.3	100.0	100.0	113620
TGFBR1	163.4	100.0	99.9	609192
TGFBR2	228.9	100.0	100.0	610168
TGIF1	200.5	100.0	100.0	142946
TLK2	141.8	100.0	100.0	No OMIM phenotype
TP63	185.9	100.0	100.0	604292

TRAF6	171.4	100.0	100.0	602355
TSHZ1	220.5	100.0	100.0	607842
TSPEAR	168.6	100.0	100.0	618180
TWIST1	176.0	100.0	99.9	101400
UBB	268.0	100.0	100.0	119540
VAX1	147.7	99.9	99.2	614402
WDR19	143.8	100.0	100.0	614378
WDR35	146.2	100.0	100.0	613610
WDR72	155.0	96.9	96.9	613211
WNT10A	201.8	100.0	100.0	224750;257980
WNT10B	221.4	100.0	100.0	617073
ZEB2	169.1	97.4	97.4	235730
ZIC1	250.2	100.0	100.0	616602
ZIC2	207.5	100.0	99.8	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.*

*TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.*

*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 no value for coverage are non protein coding genes.*

*Non protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*

*OMIM release used for OMIM disease identifiers and descriptions: September 1st, 2021.*

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