

# WES CRANIOFACIAL ANOMALIES DG 3.5

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ACP4	127.7	100.0	100.0	617297
ACTG1	221.9	100.0	100.0	No OMIM phenotype
ADAMTSL4	119.0	100.0	100.0	225100
ALX1	144.1	100.0	100.0	613456
ALX3	118.2	100.0	100.0	136760
ALX4	126.2	100.0	100.0	168500
AMBN	146.6	100.0	99.5	616270
AMELX	147.0	100.0	100.0	301200
AMER1	95.9	100.0	100.0	300373
AMTN	130.9	100.0	99.5	617607
ANKRD11	122.6	100.0	100.0	148050
ARHGAP29	154.9	100.0	100.0	-
AXIN2	137.9	100.0	100.0	608615
BCOR	94.2	100.0	99.8	300166
BMP2	144.6	100.0	100.0	-
BMP4	129.1	100.0	100.0	607932
CCBE1	126.1	100.0	100.0	No OMIM phenotype
CDC45	134.4	100.0	100.0	617063
CDON	135.3	100.0	100.0	614226
CDSN	121.5	100.0	100.0	602593
CHD7	133.9	100.0	100.0	214800
COL11A1	152.4	100.0	100.0	154780;604841
COL11A2	121.0	100.0	100.0	215150;277610
COL2A1	126.1	100.0	100.0	108300
COL9A1	143.6	100.0	100.0	614135

COL9A2	130.7	100.0	100.0	614284;600204
COL9A3	141.1	100.0	100.0	120270
COLEC11	125.4	100.0	100.0	No OMIM phenotype
CTSK	130.5	100.0	100.0	265800
CYP26B1	124.4	100.0	100.0	No OMIM phenotype
DHODH	124.0	100.0	100.0	263750
DISP1	142.8	100.0	100.0	-
DLX3	127.7	100.0	100.0	104510
DLX4	121.8	100.0	100.0	616788
DSPP	249.2	100.0	100.0	125490
EDA	108.9	100.0	99.6	305100;313500
EDAR	131.8	100.0	100.0	129490;224900
EDARADD	140.1	100.0	100.0	129490;224900
EDN1	135.5	100.0	100.0	615706
EDNRA	137.2	100.0	100.0	616367
EFNA4	135.4	100.0	100.0	601380
EFNB1	100.5	100.0	99.9	304110
EFTUD2	123.3	100.0	100.0	610536
EIF4A3	130.7	100.0	100.0	268305
ENAM	146.6	100.0	100.0	104500
ERF	134.6	100.0	100.0	600775
ESCO2	145.6	100.0	100.0	268300
EYA1	145.7	100.0	100.0	113650
EZH2	139.3	100.0	100.0	277590
FAM20A	128.9	100.0	100.0	204690
FAM83H	142.8	100.0	100.0	130900
FGD1	101.6	99.9	99.5	305400
FGF10	159.0	99.9	99.3	149730
FGF3	124.1	100.0	100.0	610706
FGF8	128.5	100.0	100.0	612702

FGF9	152.8	100.0	100.0	612961
FGFR1	125.7	100.0	100.0	123150
FGFR2	138.1	100.0	100.0	123500
FGFR3	140.9	100.0	100.0	101400
FLNA	105.8	100.0	99.9	No OMIM phenotype
FLNB	121.6	100.0	100.0	No OMIM phenotype
FOXC1	156.9	100.0	100.0	602482;601631
FOXE1	151.1	100.0	100.0	241850
GDF3	113.0	100.0	100.0	613702
GDF6	158.5	100.0	100.0	118100
GJA1	163.4	100.0	100.0	121014
GJB6	137.1	100.0	100.0	129500
GLI2	128.9	100.0	100.0	610829
GLI3	127.4	100.0	100.0	175700
GNAI3	141.0	100.0	100.0	602483
GNPTAB	150.9	100.0	100.0	No OMIM phenotype
GPR68	137.5	100.0	100.0	617217
GRHL3	119.5	100.0	100.0	606713
GSC	115.9	100.0	100.0	602471
HOXA2	144.2	100.0	100.0	612290
HUWE1	93.6	100.0	99.8	300706
HYAL2	118.5	100.0	100.0	-
IFT122	133.2	100.0	100.0	218330
IFT43	133.8	100.0	100.0	614099
IFT88	154.4	100.0	100.0	-
IKBKG	91.0	99.9	98.4	300291;300301
IL11RA	134.1	100.0	100.0	614188
IL6ST	157.5	100.0	100.0	No OMIM phenotype
IMPAD1	150.5	100.0	100.0	No OMIM phenotype
INTU	144.6	100.0	100.0	617926

IRF6	126.8	100.0	100.0	119300
ITGB6	138.4	100.0	100.0	616221
KAT6B	137.4	100.0	100.0	606170;603736
KDF1	108.4	100.0	100.0	617337
KDM1A	139.5	100.0	100.0	616728
KDM6A	117.1	100.0	99.9	300867
KLK4	130.8	100.0	100.0	204700
KMT2D	124.8	100.0	100.0	147920
KREMEN1	117.4	100.0	100.0	609898
LAMB3	125.9	100.0	100.0	104530
LRP2	137.5	100.0	100.0	222448
LRP6	131.6	100.0	100.0	616724
LTBP3	152.1	100.0	100.0	613097
MASP1	123.9	100.0	100.0	257920
MED12	99.8	100.0	99.8	300895;305450;309520
MEGF8	128.9	100.0	100.0	614976
MEIS2	134.6	100.0	100.0	600987
MEOX1	118.8	100.0	100.0	214300
MID1	104.6	99.6	99.1	300000
MITF	137.1	99.9	99.7	193510
MMP20	131.3	100.0	100.0	612529
MN1	149.0	100.0	100.0	No OMIM phenotype
MSX1	126.3	100.0	100.0	189500
MSX2	157.8	100.0	100.0	168500
NAA10	101.9	100.0	100.0	300013
NECTIN1	121.6	100.0	100.0	225060
NFKBIA	136.3	100.0	100.0	612132
NIPBL	151.6	100.0	100.0	122470
NOG	118.4	100.0	100.0	186500
NSD1	126.6	100.0	100.0	117550

ODAPH	154.5	100.0	100.0	614832
OFD1	111.2	100.0	100.0	311200
OTX2	145.7	100.0	100.0	610125
P4HB	121.9	100.0	100.0	No OMIM phenotype
PAX3	127.0	100.0	99.8	193500
PAX6	138.8	100.0	100.0	602482
PAX7	130.1	100.0	100.0	268220
PAX9	152.7	100.0	100.0	604625
PGM1	127.5	94.0	94.0	614921
PITX2	113.7	100.0	100.0	180500
PLCB4	151.7	100.0	99.9	614669
POLR1C	117.4	83.3	83.2	248390
POLR1D	139.2	100.0	100.0	613717
POR	138.4	100.0	100.0	No OMIM phenotype
PORCN	88.6	100.0	99.8	305600
PRRX1	138.4	100.0	100.0	202650
PTCH1	125.9	100.0	100.0	109400
PTH1R	130.8	100.0	100.0	125350
RAB23	150.9	100.0	100.0	201000
RAD21	145.4	100.0	100.0	614701
RBM10	100.1	100.0	99.9	311900
RECQL4	130.2	100.0	100.0	603780
RIPK4	136.5	100.0	100.0	263650
RUNX2	155.9	100.0	100.0	119600
SALL1	124.6	100.0	100.0	107480
SALL4	116.7	100.0	100.0	607323
SATB2	134.0	100.0	99.7	612313
SCARF2	140.3	100.0	100.0	No OMIM phenotype
SEC24D	136.0	100.0	99.9	No OMIM phenotype
SEMA3E	144.5	100.0	100.0	214800

SF3B2	121.9	100.0	100.0	-
SF3B4	156.0	100.0	100.0	154400
SH3BP2	112.5	99.9	99.4	118400
SHH	145.6	100.0	100.0	147250;611638
SIX1	122.9	100.0	100.0	608389
SIX3	134.2	100.0	100.0	157170
SIX5	121.0	100.0	100.0	No OMIM phenotype
SKI	122.2	100.0	99.9	182212
SLC24A4	128.4	100.0	100.0	615887
SLC26A2	155.7	100.0	100.0	256050
SMAD6	139.0	100.0	100.0	617439
SMC1A	96.9	100.0	99.8	300590
SMC3	146.3	100.0	100.0	610759
SMO	120.0	100.0	100.0	601707
SMOC2	129.9	100.0	100.0	125400
SNAI2	150.3	100.0	100.0	608890
SOX10	142.9	100.0	100.0	613266
SOX6	131.8	99.8	99.3	607257
SOX9	138.4	100.0	100.0	114290
SPECC1L	129.9	100.0	100.0	600251
SUMO1	107.1	71.0	71.0	613705
TBX1	117.8	97.7	95.5	192430
TBX22	109.8	99.4	98.1	303400
TCF12	138.9	100.0	100.0	615314
TCOF1	124.5	100.0	100.0	154500
TFAP2A	131.0	100.0	100.0	113620
TGFBR1	143.0	100.0	100.0	609192
TGFBR2	135.7	100.0	100.0	610168
TGIF1	131.1	100.0	100.0	142946
TLK2	128.3	100.0	100.0	No OMIM phenotype

TP63	125.4	100.0	99.9	604292
TRAF6	137.1	100.0	100.0	602355
TSHZ1	115.8	100.0	100.0	607842
TSPEAR	124.2	100.0	100.0	618180
TWIST1	149.1	100.0	100.0	101400
UBB	222.5	100.0	100.0	119540
VAX1	107.9	99.9	99.1	614402
WDR19	140.1	100.0	100.0	614378
WDR35	142.1	100.0	100.0	613610
WDR72	143.8	96.8	96.8	613211
WNT10A	145.6	100.0	100.0	224750;257980
WNT10B	124.7	100.0	100.0	617073
ZEB2	131.4	96.8	96.7	235730
ZIC1	141.1	100.0	100.0	616602
ZIC2	143.5	100.0	99.9	609637

*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.*

*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*