

WES OROFACIAL CLEFTING DG 3.5

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ACTB	238.4	100.0	100.0	243310
ACTG1	221.9	100.0	100.0	614583
ALX1	144.1	100.0	100.0	613456
ALX3	118.2	100.0	100.0	136760
AMER1	95.9	100.0	100.0	300373
AMMECR1	100.1	100.0	99.8	300990
ANKRD11	122.6	100.0	100.0	148050
ARHGAP29	154.9	100.0	100.0	-
ARHGAP31	131.2	100.0	100.0	100300
ASXL1	133.0	100.0	100.0	605039
B3GALT6	120.2	99.9	98.0	615349
B3GLCT	138.8	100.0	100.0	261540
B4GALT7	120.5	100.0	100.0	130070
B9D2	103.6	100.0	100.0	614175
BCOR	94.2	100.0	99.8	300166
BMP2	144.6	100.0	100.0	617877
BMPER	132.9	100.0	100.0	608022
C2CD3	127.8	96.0	96.0	615948
CC2D2A	138.0	98.2	98.2	612284
CDC45	134.4	100.0	100.0	617063
CDH1	125.2	98.7	98.7	137215
CDKN1C	165.2	100.0	100.0	614732
CHD7	133.9	100.0	100.0	214800
CHRNA	123.2	100.0	100.0	253290;265000
CHST14	124.8	100.0	100.0	601776

CILK1	126.0	100.0	100.0	612651
COL11A1	152.4	100.0	100.0	604841;154780
COL11A2	121.0	100.0	100.0	184840;215150
COL2A1	126.1	100.0	100.0	108300
COL9A1	143.6	100.0	100.0	614134
COLEC10	139.7	100.0	100.0	248340
COLEC11	125.4	100.0	100.0	265050
CPLANE1	151.7	100.0	100.0	277170
CTCF	122.6	100.0	100.0	615502
CTNND1	121.6	100.0	100.0	617681
DDX3X	113.9	99.1	98.3	300958
DDX59	139.5	100.0	100.0	174300
DHCR7	129.7	100.0	100.0	270400
DHODH	124.0	100.0	100.0	263750
DLL4	125.2	100.0	100.0	616589
DOCK6	123.3	100.0	100.0	614219
DVL1	138.3	100.0	100.0	616331
DVL3	136.2	100.0	100.0	616894
DYNC2H1	152.6	99.8	99.4	613091
DYNC2LI1	152.9	100.0	100.0	617088
EBP	99.4	100.0	100.0	302960
EDN1	135.5	100.0	100.0	615706
EDNRA	137.2	100.0	100.0	616367
EFNB1	100.5	100.0	99.9	304110
EFTUD2	123.3	100.0	100.0	610536
EIF2S3	114.0	100.0	100.0	300148
EIF4A3	130.7	100.0	100.0	268305
EOGT	124.7	98.1	94.0	615297
EPG5	129.6	100.0	100.0	242840
ESCO2	145.6	100.0	100.0	268300

EYA1	145.7	100.0	100.0	113650
FAM20C	131.1	100.0	100.0	259775
FGD1	101.6	99.9	99.5	305400
FGF8	128.5	100.0	100.0	612702
FGFR1	125.7	100.0	100.0	615465
FGFR2	138.1	100.0	100.0	101400
FLNA	105.8	100.0	99.9	311300
FLNB	121.6	100.0	100.0	150250
FOXC2	160.7	100.0	100.0	153400
FOXE1	151.1	100.0	100.0	241850
FRAS1	126.6	100.0	99.9	219000
FTO	138.7	94.5	94.5	612938
GDF6	158.5	100.0	100.0	118100
GJA1	163.4	100.0	100.0	164200
GLI2	128.9	100.0	100.0	610829;615849
GLI3	127.4	100.0	100.0	146510
GNAI3	141.0	100.0	100.0	602483
GNB1	122.7	100.0	100.0	616973
GPC3	107.5	99.6	98.9	312870
GRHL3	119.5	100.0	100.0	606713
HDAC8	105.5	97.6	97.2	300882
HYLS1	148.1	100.0	100.0	236680
IFT140	135.5	100.0	100.0	266920
IFT172	131.1	100.0	100.0	615630
IFT57	145.2	100.0	100.0	617927
IFT80	154.1	100.0	100.0	611263
IMPAD1	150.5	100.0	100.0	614078
INTU	144.6	100.0	100.0	617926
IRF6	126.8	100.0	100.0	119500;119300
KANSL1	127.7	100.0	100.0	610443

KAT6A	131.8	100.0	100.0	616268
KCNJ2	153.4	100.0	100.0	170390
KCNK9	115.6	100.0	100.0	612292
KDM6A	117.1	100.0	99.9	300867
KIAA0586	134.8	95.6	95.5	616546
KIF7	126.6	100.0	99.9	200990
KIFBP	144.8	95.6	95.6	609460
KMT2D	124.8	100.0	100.0	147920
MAP3K7	151.7	100.0	100.0	617137
MAPRE2	150.1	100.0	100.0	616734
MASP1	123.9	100.0	100.0	257920
MBTPS2	116.7	100.0	100.0	308205
MED25	135.7	100.0	100.0	616449
MEIS2	134.6	100.0	100.0	600987
MID1	104.6	99.6	99.1	300000
MKS1	148.5	100.0	100.0	249000
MSX1	126.3	100.0	100.0	608874;106600
MYMK	108.3	100.0	100.0	254940
NECTIN1	121.6	100.0	100.0	225060
NEDD4L	139.4	100.0	100.0	617201
NEK1	151.1	100.0	100.0	263520
NIPBL	151.6	100.0	100.0	122470
NOTCH1	130.3	100.0	100.0	616028
OFD1	111.2	100.0	100.0	311200
ORC1	127.7	100.0	100.0	224690
PAX3	127.0	100.0	99.8	193500
PGM1	127.5	94.0	94.0	614921
PHF8	96.2	100.0	99.9	300263
PHGDH	136.5	100.0	100.0	256520
PIEZO2	133.8	100.0	100.0	114300

PIGN	152.5	100.0	99.9	614080
PIGV	128.7	100.0	100.0	239300
PLCB4	151.7	100.0	99.9	614669
POLR1A	127.2	100.0	100.0	616462
POLR1C	117.4	83.3	83.2	248390
POLR1D	139.2	100.0	100.0	613717
POMT1	132.0	100.0	100.0	236670
PORCN	88.6	100.0	99.8	305600
PQBP1	97.4	100.0	100.0	309500
PROKR2	155.8	100.0	100.0	244200
PRRX1	138.4	100.0	100.0	202650
PTCH1	125.9	100.0	100.0	610828
PTCH2	122.3	100.0	100.0	109400
RBM10	100.1	100.0	99.9	311900
RIPK4	136.5	100.0	100.0	263650
ROR2	125.6	100.0	100.0	268310
RPGRIP1L	147.0	100.0	100.0	611561
RPL11	124.1	100.0	100.0	612562
RPL26	171.5	100.0	100.0	614900
RPL5	155.9	100.0	100.0	612561
RPS19	124.1	100.0	100.0	105650
RPS26	74.6	100.0	98.8	613309
RPS28	138.2	100.0	100.0	606164
RUNX2	155.9	100.0	100.0	119600
SALL4	116.7	100.0	100.0	-
SATB2	134.0	100.0	99.7	612313
SCARF2	140.3	100.0	100.0	600920
SEC23A	137.5	100.0	100.0	607812
SEMA3E	144.5	100.0	100.0	214800
SEPTIN9	119.1	100.0	100.0	162100

SF3B4	156.0	100.0	100.0	154400
SHH	145.6	100.0	100.0	142945
SIX1	122.9	100.0	100.0	608389
SIX3	134.2	100.0	100.0	157170
SIX5	121.0	100.0	100.0	610896
SKI	122.2	100.0	99.9	182212
SLC10A7	142.8	100.0	100.0	618363
SLC26A2	155.7	100.0	100.0	222600
SMAD3	141.0	100.0	100.0	613795
SMAD4	142.1	100.0	100.0	139210
SMC1A	96.9	100.0	99.8	300590
SMC3	146.3	100.0	100.0	610759
SMCHD1	149.9	100.0	100.0	603457
SMS	103.0	100.0	99.4	309583
SNRPB	127.2	100.0	100.0	117650
SON	139.7	100.0	100.0	617140
SOX9	138.4	100.0	100.0	114290
SPECC1L	129.9	100.0	100.0	145410
STAC3	124.2	100.0	100.0	255995
STAMBP	137.1	100.0	100.0	614261
TAPT1	146.9	100.0	100.0	616897
TBX1	117.8	97.7	95.5	192430;188400
TBX15	127.5	100.0	99.4	260660
TBX2	119.3	100.0	99.6	618223
TBX22	109.8	99.4	98.1	303400
TCOF1	124.5	100.0	100.0	154500
TCTN3	141.7	100.0	100.0	258860
TFAP2A	131.0	100.0	100.0	113620
TGDS	148.4	100.0	100.0	616145
TGFB3	136.1	100.0	100.0	615582

TGFBR1	143.0	100.0	100.0	609192
TGFBR2	135.7	100.0	100.0	610168
TGIF1	131.1	100.0	100.0	142946
TMCO1	126.4	88.0	87.7	213980
TMEM216	123.1	100.0	100.0	603194
TP63	125.4	100.0	99.9	603543;106260;618149;129400;103285;604292
TRIM37	137.1	98.3	98.3	253250
TUBB	153.2	99.6	98.8	156610
TWIST1	149.1	100.0	100.0	101400;617746
TXNL4A	153.5	100.0	100.0	608572
USP9X	118.5	100.0	99.8	300968
WASHC5	137.3	100.0	100.0	220210
WDR35	142.1	100.0	100.0	613610;614091
WNT4	109.9	100.0	99.8	611812
WNT5A	134.5	100.0	100.0	180700
XYLT1	119.8	100.0	99.8	615777
ZEB2	131.4	96.8	96.7	235730
ZIC2	143.5	100.0	99.9	609637
ZIC3	118.4	100.0	100.0	314390
ZMPSTE24	152.4	100.0	100.0	275210
ZSWIM6	120.5	97.5	95.9	603671

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