

WES OROFACIAL CLEFTING DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
ACTB	80.2	99.9	97.2	243310
ACTG1	128.6	100.0	100.0	614583
ALX1	131.8	99.6	95.2	613456
ALX3	124.2	80.2	72.8	136760
AMER1	96.7	99.6	96.6	300373
AMMECR1	86.2	99.9	98.4	300990
ANKRD11	118.2	97.0	94.0	148050
ARHGAP29	144.0	98.9	97.6	-
ARHGAP31	143.3	99.7	98.2	100300
ASXL1	128.2	99.8	98.9	605039
B3GALT6	53.2	77.0	73.0	615349
B3GLCT	100.4	99.7	98.2	261540
B4GALT7	124.9	99.7	96.8	130070
B9D2	108.7	100.0	100.0	614175
BCOR	99.9	99.2	95.8	300166
BMP2	167.5	100.0	100.0	617877
BMPER	129.9	100.0	99.6	608022
C2CD3	121.5	95.8	95.4	615948
CC2D2A	109.3	98.3	96.6	612284
CDC45	144.4	99.8	98.5	617063
CDH1	108.4	99.2	98.6	137215
CDKN1C	71.4	89.9	81.6	614732
CHD7	137.1	100.0	99.2	214800
CHRNA1	151.9	100.0	100.0	253290;265000
CHST14	149.1	99.9	98.8	601776

CILK1	111.2	99.6	98.0	612651
COL11A1	100.9	96.0	92.7	604841;154780
COL11A2	126.7	100.0	99.6	184840;215150
COL2A1	122.0	100.0	99.8	108300
COL9A1	129.1	99.9	98.6	614134
COLEC10	128.6	100.0	99.9	248340
COLEC11	169.1	100.0	100.0	265050
CPLANE1	129.6	99.4	98.2	277170
CTCF	126.8	99.7	98.4	615502
CTNND1	132.3	100.0	99.8	617681
DDX3X	66.1	81.1	78.6	300958
DDX59	150.9	100.0	99.8	174300
DHCR7	147.5	100.0	100.0	270400
DHODH	106.1	100.0	99.9	263750
DLL4	164.9	100.0	99.4	616589
DOCK6	124.2	99.4	98.7	614219
DVL1	147.0	97.2	95.1	616331
DVL3	190.2	100.0	100.0	616894
DYNC2H1	107.7	98.6	95.2	613091
DYNC2LI1	97.3	99.6	98.4	617088
EBP	67.1	99.5	94.3	302960
EDN1	135.4	100.0	99.4	615706
EDNRA	166.2	99.8	99.8	616367
EFNB1	136.4	100.0	100.0	304110
EFTUD2	111.1	100.0	99.2	610536
EIF2S3	82.9	95.0	86.8	300148
EIF4A3	87.6	100.0	99.2	268305
EOGT	108.0	79.3	77.8	615297
EPG5	113.6	99.2	97.8	242840
ESCO2	114.4	98.5	94.6	268300

EYA1	120.9	99.9	99.5	113650
FAM20C	135.5	100.0	100.0	259775
FGD1	83.8	97.2	91.2	305400
FGF8	123.7	97.1	87.2	612702
FGFR1	128.6	100.0	99.3	615465
FGFR2	113.3	97.6	97.0	101400
FLNA	143.7	100.0	99.9	311300
FLNB	130.4	99.4	98.7	150250
FOXC2	94.3	100.0	98.1	153400
FOXE1	42.9	97.9	82.2	241850
FRAS1	124.8	100.0	99.2	219000
FTO	100.1	83.8	83.7	612938
GDF6	113.8	100.0	100.0	118100
GJA1	151.1	100.0	100.0	164200
GLI2	163.8	99.8	98.6	610829;615849
GLI3	131.0	98.5	97.7	146510
GNAI3	102.5	98.4	93.2	602483
GNB1	161.1	100.0	100.0	616973
GPC3	77.3	98.8	92.9	312870
GRHL3	151.8	100.0	99.9	606713
HDAC8	90.2	85.7	83.7	300882
HYLS1	145.3	100.0	100.0	236680
IFT140	120.4	99.9	99.2	266920
IFT172	94.8	99.6	98.6	615630
IFT57	126.6	99.9	99.0	617927
IFT80	68.7	97.2	85.7	611263
IMPAD1	147.0	100.0	99.9	614078
INTU	119.2	99.9	98.6	617926
IRF6	85.2	99.4	93.0	119500;119300
KANSL1	143.1	99.8	98.2	610443

KAT6A	157.2	100.0	99.2	616268
KCNJ2	149.7	100.0	100.0	170390
KCNK9	170.6	97.3	97.3	612292
KDM6A	98.7	94.2	85.9	300867
KIAA0586	126.1	97.1	92.0	616546
KIF7	105.7	93.6	91.9	200990
KIFBP	144.9	96.1	96.0	609460
KMT2D	133.9	99.9	99.0	147920
MAP3K7	118.3	99.8	99.6	617137
MAPRE2	153.4	100.0	98.5	616734
MASP1	141.9	100.0	99.6	257920
MBTPS2	116.3	99.9	98.5	308205
MED25	148.5	100.0	99.9	616449
MEIS2	122.2	100.0	99.7	600987
MID1	137.3	99.6	97.7	300000
MKS1	93.4	99.4	96.3	249000
MSX1	120.3	97.7	92.6	608874;106600
MYMK	154.0	100.0	100.0	254940
NECTIN1	131.2	100.0	99.7	225060
NEDD4L	98.2	71.9	71.7	617201
NEK1	120.2	99.5	98.2	263520
NIPBL	119.2	98.4	96.3	122470
NOTCH1	128.4	99.3	97.9	616028
OFD1	50.3	87.1	71.3	311200
ORC1	92.5	99.9	97.9	224690
PAX3	97.0	100.0	99.8	193500
PGM1	124.5	94.2	94.1	614921
PHF8	81.8	98.9	94.4	300263
PHGDH	111.5	99.9	98.2	256520
PIEZO2	106.7	99.8	99.2	114300

PIGN	96.7	93.1	89.6	614080
PIGV	122.2	100.0	100.0	239300
PLCB4	99.9	99.8	98.7	614669
POLR1A	111.2	99.9	98.8	616462
POLR1C	83.8	89.6	84.8	248390
POLR1D	172.9	91.6	91.6	613717
POMT1	131.1	99.5	97.3	236670
PORCN	110.8	100.0	99.1	305600
PQBP1	138.7	100.0	99.3	309500
PROKR2	228.0	100.0	100.0	244200
PRRX1	83.1	100.0	99.4	202650
PTCH1	110.2	99.3	96.6	610828
PTCH2	115.8	99.9	98.4	109400
RBM10	119.3	99.8	97.3	311900
RIPK4	151.7	100.0	99.9	263650
ROR2	172.6	100.0	99.4	268310
RPGRIP1L	129.9	96.5	95.3	611561
RPL11	89.1	99.9	97.9	612562
RPL26	36.0	94.2	75.5	614900
RPL5	32.6	81.9	59.7	612561
RPS19	95.1	100.0	99.9	105650
RPS26	89.3	93.2	81.2	613309
RPS28	47.1	99.7	86.3	606164
RUNX2	105.6	72.2	72.2	119600
SALL4	137.8	99.1	96.4	-
SATB2	102.1	99.5	96.5	612313
SCARF2	76.3	97.4	88.9	600920
SEC23A	137.9	99.7	97.0	607812
SEMA3E	137.7	99.1	98.9	214800
SEPTIN9	151.1	100.0	99.5	162100

SF3B4	59.4	99.8	94.1	154400
SHH	125.7	100.0	100.0	142945
SIX1	122.0	100.0	99.7	608389
SIX3	164.7	99.3	96.9	157170
SIX5	66.7	96.9	90.1	610896
SKI	110.2	99.7	97.1	182212
SLC10A7	107.8	99.5	98.1	618363
SLC26A2	199.3	100.0	100.0	222600
SMAD3	132.6	99.9	98.4	613795
SMAD4	113.8	99.9	99.9	139210
SMC1A	89.7	99.6	97.1	300590
SMC3	79.3	94.5	89.0	610759
SMCHD1	101.6	99.3	96.4	603457
SMS	66.0	87.9	72.1	309583
SNRPB	79.3	100.0	98.6	117650
SON	123.9	97.6	92.6	617140
SOX9	171.5	100.0	99.9	114290
SPECC1L	118.1	96.0	95.0	145410
STAC3	118.7	100.0	100.0	255995
STAMBP	94.2	99.4	96.4	614261
TAPT1	90.1	93.0	87.1	616897
TBX1	91.3	87.4	77.6	192430;188400
TBX15	102.0	100.0	99.7	260660
TBX2	130.6	99.9	97.8	618223
TBX22	116.2	98.4	93.8	303400
TCOF1	120.8	99.7	98.7	154500
TCTN3	118.9	100.0	100.0	258860
TFAP2A	101.7	98.1	92.1	113620
TGDS	95.5	99.4	95.9	616145
TGFB3	139.0	100.0	100.0	615582

TGFBR1	156.6	93.6	93.6	609192
TGFBR2	159.9	100.0	99.9	610168
TGIF1	133.3	100.0	100.0	142946
TMCO1	73.6	87.8	87.0	213980
TMEM216	89.9	98.5	92.8	603194
TP63	170.6	100.0	100.0	603543;106260;618149;129400;103285;604292
TRIM37	123.0	98.3	97.1	253250
TUBB	116.1	96.8	93.7	156610
TWIST1	102.3	100.0	99.4	101400;617746
TXNL4A	112.7	99.3	99.1	608572
USP9X	97.2	98.1	91.7	300968
WASHC5	139.3	99.8	99.7	220210
WDR35	147.5	99.6	98.4	613610;614091
WNT4	228.0	97.8	93.6	611812
WNT5A	123.1	100.0	100.0	180700
XYLT1	134.5	97.8	91.1	615777
ZEB2	137.0	99.7	98.5	235730
ZIC2	104.5	100.0	99.3	609637
ZIC3	125.7	100.0	99.9	314390
ZMPSTE24	134.5	99.6	99.4	275210
ZSWIM6	118.8	95.1	91.6	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.