

# WES CILIOPATHIES DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt;10x</i>	<i>% covered &gt;20x</i>	<i>OMIM disease ID</i>
ACVR2B	124.3	98.6	95.1	613751
ADAMTS9	118.2	99.4	98.4	-
AHI1	132.9	99.4	97.4	608629
ALMS1	174.7	99.7	99.5	203800
ANKS6	91.7	94.2	89.7	615382
ARL13B	85.8	100.0	99.3	612291
ARL3	80.1	99.7	96.5	-
ARL6	112.4	99.1	98.4	613575;209900
ARMC4	108.1	92.4	89.9	615451
ARMC9	130.5	99.9	99.4	617622
B9D1	91.9	85.2	85.2	614209
B9D2	108.7	100.0	100.0	614175
BBIP1	142.6	95.7	87.4	615995
BBS1	160.2	100.0	100.0	209900
BBS10	151.0	100.0	99.9	209900
BBS12	167.8	100.0	100.0	209900
BBS2	148.8	99.4	98.0	209900
BBS4	108.1	99.9	98.9	209900
BBS5	95.0	98.4	94.7	209900
BBS7	151.4	99.0	96.5	209900
BBS9	104.9	92.0	89.0	209900
C2CD3	121.5	95.8	95.4	615948
C8orf37	135.1	99.7	99.6	617406;614500
CBY1	64.9	82.2	82.0	-
CC2D2A	109.3	98.3	96.6	612284;216360;612285

CCDC103	117.8	100.0	100.0	614679
CCDC114	138.5	100.0	99.8	615067
CCDC151	118.1	100.0	99.7	616037
CCDC28B	87.9	99.9	98.2	209900
CCDC39	88.8	99.0	96.2	613807
CCDC40	118.3	99.1	98.2	613808
CCDC65	83.9	97.0	92.5	615504
CCNO	102.8	100.0	99.0	615872
CENPF	112.9	99.4	96.9	616369
CEP104	113.2	99.9	98.0	616781
CEP120	140.6	99.9	99.6	616300
CEP164	95.5	99.8	98.2	614845
CEP290	86.2	96.2	90.8	611134;209900;611755;610189;610188
CEP41	77.1	98.8	93.4	614464
CEP55	110.9	100.0	99.8	236500
CEP83	107.1	99.0	96.6	615862
CFAP298	127.1	99.6	96.7	615500
CFAP300	88.0	99.2	98.3	618063
CFAP410	129.3	100.0	99.6	602271;617547
CFAP44	110.3	99.5	98.6	617593
CFAP53	136.1	99.3	96.6	614779
CFAP69	70.5	98.7	94.9	617959
CFC1	121.3	85.0	78.0	605376
CPLANE1	129.6	99.4	98.2	614615
CSPP1	116.4	99.7	98.1	615636
DCDC2	146.2	100.0	99.9	616217
DDX59	150.9	100.0	99.8	174300
DNAAF1	118.9	100.0	99.4	613193
DNAAF2	115.3	99.7	98.4	612518
DNAAF3	123.3	99.5	96.3	606763

DNAAF4	96.9	99.4	94.7	615482;127700
DNAAF5	103.3	85.6	78.8	614874
DNAH1	164.3	99.9	99.6	617577
DNAH11	126.3	99.8	98.8	611884
DNAH17	126.9	99.9	99.2	618643
DNAH5	119.0	99.9	98.9	608644
DNAH8	122.6	99.7	98.6	-
DNAI1	123.8	100.0	99.9	244400
DNAI2	184.4	98.2	95.8	612444
DNAJB13	123.0	100.0	99.6	610263
DNAL1	95.5	99.6	98.3	614017
DRC1	87.0	99.9	98.3	615294
DYNC2H1	107.7	98.6	95.2	613091
DYNC2LI1	97.3	99.6	98.4	617088
EVC	108.4	94.2	91.4	193530;225500
EVC2	124.2	98.0	96.2	225500
EXOC8	156.4	100.0	100.0	-
EXTL3	170.1	100.0	100.0	617425
FAM149B1	96.4	98.2	94.3	618763
FUZ	130.3	100.0	100.0	182940
GAS8	130.2	99.9	99.6	616726
GDF1	32.3	80.8	59.0	208530
GLIS2	143.2	100.0	99.9	611498
HYDIN	113.3	99.8	98.7	608647
HYLS1	145.3	100.0	100.0	236680
IFT122	131.2	99.9	99.2	218330
IFT140	120.4	99.9	99.2	266920
IFT172	94.8	99.6	98.6	615630
IFT27	119.6	100.0	100.0	615996
IFT43	116.4	100.0	100.0	614099

IFT52	130.6	100.0	99.9	617102
IFT80	68.7	97.2	85.7	611263
IFT81	91.9	92.9	89.6	617895
INPP5E	113.1	96.9	93.2	610156;213300
INTU	119.2	99.9	98.6	617925;617926
INVS	145.6	100.0	99.8	602088
IQCB1	96.5	92.8	82.8	609254
KCTD3	133.2	99.7	99.4	-
KIAA0556	122.8	100.0	99.6	616784
KIAA0586	126.1	97.1	92.0	616490
KIAA0753	120.5	99.9	98.9	617127
KIF14	124.3	99.2	96.8	616258
KIF7	105.7	93.6	91.9	200990;614120
LBR	102.2	97.9	91.0	215140;613471;169400
LCA5	136.1	99.6	97.9	604537
LRRC56	134.6	99.8	99.0	618254
LRRC6	156.4	99.3	96.9	614935
LZTFL1	112.9	99.7	99.4	209900
MAPKBP1	140.2	100.0	100.0	617271
MCIDAS	76.2	98.4	95.2	-
MKKS	204.9	100.0	100.0	209900;236700
MKS1	93.4	99.4	96.3	209900;249000
MMP21	101.4	99.8	99.2	616749
NCAPG2	123.2	99.8	99.0	618460
NEK1	120.2	99.5	98.2	263520
NEK8	152.3	100.0	99.8	615415;613824
NME5	186.4	99.7	99.7	-
NME8	95.0	98.9	94.2	610852
NODAL	154.7	100.0	100.0	270100
NPHP1	135.6	99.8	99.1	266900;609583;256100

NPHP3	122.6	99.6	98.5	208540;604387;267010
NPHP4	130.3	100.0	99.8	606996;606966
OCRL	107.2	99.4	97.6	300555;309000
OFD1	50.3	87.1	71.3	300804;300424;311200;300209
PDE6D	108.3	100.0	99.9	615665
PIBF1	64.2	99.1	95.0	617767
PIH1D3	73.1	97.9	86.0	300991
PIK3C2A	131.9	99.0	95.9	618440
PKD1	30.8	40.6	32.8	173900
PKD2	94.1	96.0	93.3	613095
PKHD1	134.4	100.0	99.6	263200
PMFBP1	107.5	99.8	98.5	618112
POC1A	119.6	100.0	100.0	614813
RPGRIP1L	129.9	96.5	95.3	216360;611560;611561
RSPH1	141.5	99.9	99.9	615481
RSPH3	125.2	99.6	98.4	616481
RSPH4A	141.5	98.2	95.4	612649
RSPH9	131.9	99.7	96.3	612650
SCLT1	91.0	95.4	89.6	-
SDCCAG8	115.0	99.8	99.8	613615
SPAG1	94.7	98.7	93.9	615505
SPATA7	122.1	99.6	98.2	604232
STK36	124.0	100.0	99.1	-
TBC1D32	96.8	98.7	96.4	-
TCTEX1D2	122.1	100.0	99.8	617405
TCTN1	102.5	96.8	92.8	614173
TCTN2	127.2	99.9	99.1	613885
TCTN3	118.9	100.0	100.0	614815;258860
TMEM107	141.9	100.0	100.0	617563;617562
TMEM138	87.3	99.8	93.1	614465

TMEM216	89.9	98.5	92.8	603194;608091
TMEM218	81.9	100.0	99.8	-
TMEM231	99.0	100.0	99.3	615397;614970
TMEM237	121.4	99.8	99.3	614424
TMEM260	122.7	98.7	95.4	617478
TMEM67	84.8	98.6	93.5	216360;607361;613550;209900;610688
TOGARAM1	127.1	99.6	97.5	-
TOPORS	183.9	100.0	100.0	-
TRAF3IP1	82.2	98.7	95.4	616629
TRIM32	119.5	100.0	99.9	254110;209900
TTBK2	119.7	99.9	98.3	604432
TTC21B	125.8	99.7	99.1	613819;613820
TTC25	78.6	100.0	99.7	617092
TTC26	143.7	99.8	97.9	-
TTC8	125.6	99.5	98.0	613464;209900
TULP1	128.6	99.8	98.2	600132;613843
VHL	131.2	95.5	90.6	263400;144700;171300;193300
WDPCP	101.6	98.0	94.1	209900
WDR19	128.4	99.8	98.6	614378;614376;614377
WDR34	119.3	100.0	99.8	615633
WDR35	147.5	99.6	98.4	614091;613610
WDR60	106.7	99.3	95.8	615503
WDR66	115.4	100.0	99.9	618152
XPNPEP3	113.1	100.0	100.0	613159
ZIC3	125.7	100.0	99.9	306955
ZMYND10	121.6	100.0	100.0	615444
ZNF423	189.1	100.0	100.0	614844

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.*

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