

WES CILIOPATHIES DG 2.14

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AHI1	139.3	99.2	95.1	608629
ALMS1	179.8	99.9	99.7	203800
ANKS6	91.8	92.8	88.6	615382
ARL13B	97.3	98.9	92.8	612291
ARL6	85.2	99.8	95.3	613575;209900
ARMC4	129.1	94.4	93.4	615451
ARMC9	127.3	99.8	98.5	617622
B9D1	115.3	92.1	91.4	614209
B9D2	110.9	100.0	100.0	614175
BBIP1	132.0	99.4	94.8	615995
BBS1	148.9	100.0	100.0	209900
BBS10	172.6	100.0	100.0	209900
BBS12	208.6	100.0	100.0	209900
BBS2	181.8	100.0	99.8	209900
BBS4	135.9	99.7	97.3	209900
BBS5	81.1	95.8	84.1	209900
BBS7	120.7	98.1	91.7	209900
BBS9	112.9	96.0	93.8	209900
C11orf70	60.7	92.3	82.2	618063
C21orf2	104.4	99.9	98.7	602271;617547
C21orf59	145.8	98.7	94.6	615500
C2CD3	143.1	95.8	95.6	615948
C5orf42	122.8	98.6	95.5	614615
C8orf37	126.4	100.0	99.0	614500;617406
CC2D2A	127.4	99.5	97.1	612285;216360;612284

CCDC103	116.8	100.0	99.0	614679
CCDC114	120.7	100.0	99.6	615067
CCDC151	116.2	100.0	99.7	616037
CCDC28B	83.7	100.0	98.6	209900
CCDC39	74.3	96.6	88.9	613807
CCDC40	126.5	98.9	97.8	613808
CCDC65	105.9	99.7	97.6	615504
CCNO	103.1	99.0	95.6	615872
CENPF	139.5	99.5	97.6	616369
CEP104	119.9	99.0	97.9	616781
CEP120	129.7	99.8	98.1	616300
CEP164	94.2	99.9	98.0	614845
CEP290	66.1	88.4	76.7	610188;610189;611134;611755;209900
CEP41	83.5	97.7	89.6	614464
CEP55	129.5	100.0	99.9	236500
CEP83	96.7	98.3	89.1	615862
CSPP1	112.0	99.8	97.8	615636
DCDC2	150.5	99.9	99.6	616217
DDX59	151.7	99.7	97.6	174300
DNAAF1	115.8	100.0	99.7	613193
DNAAF2	105.1	99.7	96.9	612518
DNAAF3	91.8	97.7	90.6	606763
DNAAF4	79.6	96.3	84.1	615482;127700
DNAAF5	107.9	84.5	78.2	614874
DNAH11	134.0	99.8	98.4	611884
DNAH17	136.9	100.0	99.7	4
DNAH5	123.8	99.7	98.5	608644
DNAI1	135.3	100.0	100.0	244400
DNAI2	156.6	98.4	95.5	612444
DNAJB13	138.3	100.0	97.3	610263

DNAL1	99.0	95.7	84.5	614017
DNHD1	173.1	100.0	99.9	5
DRC1	97.0	99.9	98.6	615294
DYNC2H1	90.5	96.6	87.0	613091
DYNC2LI1	95.1	99.3	96.0	617088
EVC	110.4	93.2	89.8	225500;193530
EVC2	119.3	96.4	94.3	225500
EXOC8	174.4	100.0	100.0	1
EXTL3	206.4	100.0	100.0	6
FUZ	118.8	100.0	100.0	182940
GAS8	150.7	99.8	99.4	616726
GLIS2	109.0	99.9	98.2	611498
HYDIN	133.6	99.9	99.5	608647
HYLS1	171.1	100.0	100.0	236680
IFT122	152.0	100.0	99.9	218330
IFT140	114.7	99.9	99.0	266920
IFT172	116.5	100.0	99.6	615630
IFT27	131.7	100.0	99.6	615996
IFT43	114.8	100.0	100.0	614099
IFT52	123.6	100.0	99.3	617102
IFT80	57.8	87.6	70.7	611263
IFT81	92.9	88.3	81.2	617895
INPP5E	89.1	95.8	90.0	610156;213300
INTU	122.0	99.7	96.6	-
INVS	160.5	100.0	100.0	602088
IQCB1	92.2	89.3	75.4	609254
KCTD3	137.8	99.7	97.9	-
KIAA0556	134.4	99.9	99.4	616784
KIAA0586	114.7	98.2	92.7	616490
KIAA0753	123.9	99.9	98.7	617127

KIF14	111.3	98.1	89.9	616258
KIF7	85.7	93.5	88.9	200990;614120
LBR	87.8	93.3	83.9	169400;613471;215140
LCA5	127.8	97.3	95.7	604537
LRRC6	137.9	94.7	91.0	614935
LZTFL1	109.1	99.1	95.3	209900
MAPKBP1	144.1	100.0	100.0	617271
MKKS	208.5	83.2	83.1	236700;209900
MKS1	114.5	99.9	98.5	249000;209900
NEK1	103.2	98.1	93.0	263520
NEK8	171.4	100.0	99.9	613824;615415
NME8	105.6	97.5	91.0	610852
NPHP1	117.6	98.8	96.4	266900;609583;256100
NPHP3	115.6	99.4	96.1	208540;267010;604387
NPHP4	136.7	99.9	99.3	606996;606966
OCRL	122.2	98.8	96.3	300555;309000
OFD1	51.5	84.0	67.8	300424;300209;300804;311200
PDE6D	106.1	100.0	99.9	615665
PIBF1	66.0	96.6	82.4	617767
PIH1D3	70.5	94.3	79.6	300991
PKD1	28.1	42.6	34.5	173900
PKD2	110.6	89.3	84.2	613095
PKHD1	154.9	100.0	99.7	263200
POC1A	133.8	100.0	100.0	614813
RPGRIP1L	126.2	96.4	93.9	611561;611560;216360
RSPH1	146.1	100.0	100.0	615481
RSPH3	132.5	99.7	97.5	616481
RSPH4A	139.0	98.3	96.5	612649
RSPH9	127.4	100.0	99.6	612650
SCLT1	69.2	90.5	80.0	2

SDCCAG8	123.9	99.8	97.4	613615
SPAG1	87.3	96.3	88.2	615505
SPATA7	119.6	97.8	90.8	604232
TBC1D32	81.2	96.6	91.2	3
TCTEX1D2	126.1	100.0	99.3	617405
TCTN1	98.8	95.7	92.8	614173
TCTN2	144.2	99.5	97.0	613885
TCTN3	127.6	100.0	99.8	614815;258860
TMEM107	163.8	100.0	100.0	617562;617563
TMEM138	100.2	100.0	99.5	614465
TMEM216	111.9	100.0	98.7	603194;608091
TMEM231	111.5	100.0	99.9	615397;614970
TMEM237	100.7	99.8	98.3	614424
TMEM67	72.9	93.3	83.4	607361;216360;613550;610688;209900
TRAF3IP1	90.3	96.3	92.8	616629
TRIM32	141.2	100.0	100.0	254110;209900
TTBK2	123.2	100.0	98.9	604432
TTC21B	100.7	99.7	97.6	613819;613820
TTC25	103.4	100.0	99.5	617092
TTC26	134.3	99.8	98.3	7
TTC8	106.9	97.9	92.0	613464;209900
TULP1	97.8	96.8	91.7	600132;613843
VHL	119.7	92.6	85.3	193300;171300;144700;263400
WDPCP	107.3	93.9	88.9	209900
WDR19	132.1	99.8	98.1	614376;614377;614378
WDR34	106.6	99.5	96.2	615633
WDR35	145.1	99.3	97.7	613610;614091
WDR60	114.2	99.1	96.3	615503
XPNPEP3	134.0	100.0	99.2	613159
ZMYND10	136.5	100.0	100.0	615444

ZNF423	250.8	100.0	100.0	614844
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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.

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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.

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