

## CILIOPATHIES GENE PANEL DG 2.4.x

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated Phenotype description and OMIM ID</i>
AHI1	120.1	100%	99%	Joubert syndrome-3, 608629
ALMS1	206.5	98%	98%	Alstrom syndrome, 203800
ANKS6	66.6	97%	85%	Nephronophthisis 16, 615382
ARL13B	132.1	100%	98%	Joubert syndrome 8, 612291
ARL6	155.8	100%	100%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ARMC4	100.3	89%	86%	Ciliary dyskinesia, primary, 23, 615451
ATXN10	127.5	100%	100%	Spinocerebellar ataxia 10, 603516
B9D1	80.4	91%	86%	Meckel syndrome 9, 614209
B9D2	55.6	100%	97%	Meckel syndrome 10, 614175
BBIP1	112.5	92%	85%	?Bardet-Biedl syndrome 18, 615995
BBS1	126.1	99%	99%	Bardet-Biedl syndrome 1, 209900
BBS10	135.2	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	160	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	118.5	100%	100%	Bardet-Biedl syndrome 2, 209900
BBS4	99	98%	92%	Bardet-Biedl syndrome 4, 209900
BBS5	135.1	100%	100%	Bardet-Biedl syndrome 5, 209900
BBS7	130.4	100%	99%	Bardet-Biedl syndrome 7, 209900
BBS9	132.4	100%	98%	Bardet-Biedl syndrome 9, 209900
C21orf59	101	100%	98%	Ciliary dyskinesia, primary, 26, 615500
C2CD3	117.9	95%	95%	?Orofaciodigital syndrome XIV
C2orf71	129.3	99%	94%	Retinitis pigmentosa 54, 613428
C5orf42	133.5	100%	100%	Joubert syndrome 17, 614615
C8orf37	91.8	100%	100%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500

CC2D2A	101.8	98%	97%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CCDC103	113.2	100%	100%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	86.8	100%	97%	Ciliary dyskinesia, primary, 20, 615067
CCDC151	119.9	95%	95%	Ciliary dyskinesia, primary, 30, 616037
CCDC28B	78.3	100%	100%	{Bardet-Biedl syndrome, modifier of}, 209900
CCDC39	111.5	100%	100%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	92.9	98%	95%	Ciliary dyskinesia, primary, 15, 613808
CCDC41	127.7	100%	99%	Nephronophthisis 18, 615862
CCDC65	86.4	100%	98%	Ciliary dyskinesia, primary, 27, 615504
CCNO	81.4	99%	97%	Ciliary dyskinesia, primary, 29, 615872
CDH23	102.2	99%	97%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CENPF	140.2	100%	99%	Ciliary dyskinesia, primary, 31, 616369
CEP120	124.7	100%	100%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	87	99%	94%	Nephronophthisis 15, 614845
CEP290	104.1	100%	98%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	92.3	100%	100%	Joubert syndrome 15, 614464
CLRN1	167	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome type 3A, 27690
CSPP1	126.7	100%	100%	Joubert syndrome 21, 615636
DCDC2	168.2	100%	99%	?Deafness, autosomal recessive 66, 610212 Nephronophthisis19, 616217
DDX59	157.8	100%	100%	Orofaciodigital syndrome V, 174300
DFNB31	102	100%	98%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DNAAF1	139.8	99%	98%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	123.2	100%	100%	Ciliary dyskinesia, primary, 10, 612518

DNAAF3	71.8	96%	77%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	123.9	100%	99%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	101.1	99%	98%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	134.8	100%	100%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	111.1	96%	91%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAL1	130	100%	100%	Ciliary dyskinesia, primary, 16, 614017
DRC1	79.7	99%	97%	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	119.8	99%	99%	Asphyxiating thoracic dystrophy 3, 613091 Short rib-polydactyly syndrome, type III, 263510 Short rib-polydactyly syndrome, type IIB, 615087
DYX1C1	87.8	100%	100%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EVC	85.2	92%	89%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	109.7	96%	93%	Ellis-van Creveld syndrome, 225500
EXOC8	173.1	100%	100%	No OMIM phenotype Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78)
FAM161A	154	100%	100%	Retinitis pigmentosa 28, 606068
FLCN	127.6	100%	99%	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500
GLIS2	103.9	100%	100%	Nephronophthisis 7, 611498
GPR98	118.4	99%	98%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
HEATR2	72.2	86%	73%	Ciliary dyskinesia, primary, 18, 614874
HYDIN	88.9	88%	86%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	176.9	100%	100%	Hydroletharus syndrome, 236680
IFT122	87.9	95%	95%	Cranioectodermal dysplasia 1, 218330
IFT140	93.6	99%	91%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	101.8	100%	96%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 Retinitis pigmentosa 71, 616394
IFT27	108.8	83%	82%	Bardet-Biedl syndrome 19, 615996

IFT43	88	100%	94%	Cranioectodermal dysplasia 3, 614099
IFT80	86.3	99%	95%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
INPP5E	81.6	99%	97%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INVS	123.3	100%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	93.6	97%	88%	Senior-Loken syndrome 5, 609254
KIAA0586	117.5	98%	96%	Joubert syndrome 23,
KIF14	121.3	100%	100%	?Meckel syndrome 12, 616258
KIF7	80.4	94%	89%	Hydrolethalmus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
LCA5	148	100%	98%	Leber congenital amaurosis 5, 604537
LRR6	129	100%	100%	Ciliary dyskinesia, primary, 19, 614935
LZTFL1	92.8	100%	100%	Bardet-Biedl syndrome 17, 615994
MAK	83.7	95%	91%	Retinitis pigmentosa 62, 614181
MKKS	138.3	89%	89%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MKS1	113	100%	98%	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 209900
MYO7A	86.9	97%	92%	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317
NEK1	129.3	100%	99%	Short rib-polydactyly syndrome, type IIA, 263520
NEK8	121.9	100%	100%	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NME8	108.4	100%	100%	Ciliary dyskinesia, primary, 6, 610852
NPHP1	121.6	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	117.4	100%	100%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	91.5	99%	96%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996

OCRL	65.3	97%	95%	Low syndrome, 309000 Dent disease 2, 300555
OFD1	39.7	87%	78%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
PCDH15	134.7	99%	99%	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067
PDE6D	98	100%	100%	?Joubert syndrome 22, 615665
PDZD7	80.8	95%	84%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PKD1	14	20%	18%	Polycystic kidney disease, adult type I, 173900
PKD2	102.3	99%	93%	Polycystic kidney disease 2, 613095
PKHD1	109.5	99%	98%	Polycystic kidney and hepatic disease, 263200
POC1A	113	100%	93%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
PTPRQ	116.3	94%	93%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RP1	189.1	100%	100%	Retinitis pigmentosa 1, 180100 {Hypertriglyceridemia, susceptibility to}, 145750
RP2	63	100%	100%	Retinitis pigmentosa 2, 312600
RPGR	78.1	84%	78%	Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834 Cone-rod dystrophy, X-linked, 1, 304020
RPGRIP1	128.5	100%	98%	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194
RPGRIP1L	109.1	98%	96%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RSPH1	114.7	100%	100%	Ciliary dyskinesia, primary, 24, 615481
RSPH4A	149.3	100%	100%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	102.5	100%	99%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	117.2	95%	95%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36)
SDCCAG8	104.6	100%	99%	Senior-Loken syndrome 7, 613615

SPAG1	128.5	100%	96%	Ciliary dyskinesia, primary, 28, 615505
SPATA7	136.5	100%	97%	Leber congenital amaurosis 3, 604232
TBC1D32	118.2	100%	100%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36)
TCTN1	105.4	95%	95%	Joubert syndrome 13, 614173
TCTN2	97.2	100%	99%	Meckel syndrome 8, 613885
TCTN3	109	100%	98%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TMEM138	99.3	100%	100%	Joubert syndrome 16, 614465
TMEM216	73.2	100%	95%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	75.3	96%	86%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397
TMEM237	98.7	100%	97%	Joubert syndrome 14, 614424
TMEM67	121.8	100%	98%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TOPORS	165.3	100%	100%	Retinitis pigmentosa 31, 609923
TRIM32	125.9	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TTBK2	129.7	100%	100%	Spinocerebellar ataxia 11, 604432
TTC21B	114.3	99%	98%	Nephronophthisis 12, 613820 Asphyxiating thoracic dystrophy 4, 613819
TTC8	109.6	100%	100%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TULP1	94.7	100%	96%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
USH1C	77.6	99%	96%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	114	99%	91%	Usher syndrome, type 1G, 606943
USH2A	123.3	100%	99%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809

VHL	126.7	100%	100%	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400
WDPCP	96.9	98%	92%	?Bardet-Biedl syndrome 15, 615992
WDR19	130.6	100%	100%	Asphyxiating thoracic dystrophy 5, 614376 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378
WDR34	106.9	100%	99%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	120.2	100%	99%	Cranioectodermal dysplasia 2, 613610 Short rib-polydactyly syndrome, type V, 614091
WDR60	108.4	99%	98%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
XPNPEP3	126.9	97%	95%	Nephronophthisis-like nephropathy 1, 613159
ZMYND10	97	100%	94%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	144.8	100%	99%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844

Gene symbols used follow HGCN 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

OMIM release used for OMIM disease identifiers and descriptions : November 15<sup>th</sup>, 2015

This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

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