CILIOPATHIES GENE PANEL DG 2.5/2.6

Gene name	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AHI1	133	99%	93%	Joubert syndrome-3, 608629
ALMS1	167.8	99%	99%	Alstrom syndrome, 203800
ANKS6	77.2	93%	88%	Nephronophthisis 16, 615382
ARL13B	78.8	99%	81%	Joubert syndrome 8, 612291
ARL6	94.9	99%	96%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ARMC4	123.8	90%	89%	Ciliary dyskinesia, primary, 23, 615451
ATXN10	152.3	96%	95%	Spinocerebellar ataxia 10, 603516
B9D1	101.1	92%	91%	Meckel syndrome 9, 614209
B9D2	110.8	100%	100%	Meckel syndrome 10, 614175
BBIP1	121	99%	87%	?Bardet-Biedl syndrome 18, 615995
BBS1	133.8	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	160.2	100%	99%	Bardet-Biedl syndrome 10, 209900
BBS12	196.3	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	173	100%	98%	Bardet-Biedl syndrome 2, 209900
BBS4	136.1	99%	95%	Bardet-Biedl syndrome 4, 209900
BBS5	103.3	97%	91%	Bardet-Biedl syndrome 5, 209900
BBS7	121.3	96%	90%	Bardet-Biedl syndrome 7, 209900
BBS9	105.1	94%	93%	Bardet-Biedl syndrome 9, 209900
C21orf59	152.5	100%	91%	Ciliary dyskinesia, primary, 26, 615500
C2CD3	140.4	95%	95%	?Orofaciodigital syndrome XIV
C2orf71	116.9	100%	99%	Retinitis pigmentosa 54, 613428
C5orf42	119.9	95%	91%	Joubert syndrome 17, 614615
C8orf37	116.7	100%	99%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500

CC2D2A	121.8	99%	94%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CCDC103	96.6	100%	98%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	105.7	100%	99%	Ciliary dyskinesia, primary, 20, 615067
CCDC151	111.8	100%	95%	Ciliary dyskinesia, primary, 30, 616037
CCDC28B	68.2	100%	99%	{Bardet-Biedl syndrome, modifier of}, 209900
CCDC39	81.5	95%	90%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	106.5	99%	97%	Ciliary dyskinesia, primary, 15, 613808
CCDC41	103.8	99%	96%	Nephronophthisis 18, 615862
CCDC65	89.6	98%	87%	Ciliary dyskinesia, primary, 27, 615504
CCNO	75.7	100%	95%	Ciliary diskinesia, primary, 29, 615872
CDH23	183	100%	99%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CENPF	131.3	99%	97%	Ciliary dyskinesia, primary, 31, 616369
CEP120	130.9	100%	97%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	75.2	99%	94%	Nephronophthisis 15, 614845
CEP290	69.5	89%	76%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	83.4	97%	91%	Joubert syndrome 15, 614464
CLRN1	141.9	100%	100%	Retinitis pigmentosa 61, 614180 Usher syndrome type 3A, 276902 Retinitis pigmentosa 61, 614180
CSPP1	99.3	99%	92%	Joubert syndrome 21, 615636
DCDC2	132	100%	100%	?Deafness, autosomal recessive 66, 610212 Nephronophthisis19, 616217
DDX59	157.1	100%	98%	Orofaciodigital syndrome V, 174300
DFNB31	102.8	100%	97%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DNAAF1	102.1	100%	98%	Ciliary dyskinesia, primary, 13, 613193

DNAAF2	82.9	94%	90%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	82.3	97%	89%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	130.3	99%	97%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	123.9	99%	96%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	107.4	98%	96%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	138.5	95%	93%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAL1	103.6	93%	79%	Ciliary dyskinesia, primary, 16, 614017
DRC1	91.7	100%	98%	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	91.6	95%	82%	Asphyxiating thoracic dystrophy 3, 613091 Short rib-polydactyly syndrome, type III, 263510 Short rib-polydactyly syndrome, type IIB, 615087
DYX1C1	78	96%	84%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EVC	95.9	94%	89%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	105.5	95%	91%	Ellis-van Creveld syndrome, 225500
EXOC8	140.3	100%	100%	No OMIM disease Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78)
FAM161A	110.4	99%	93%	Retinitis pigmentosa 28, 606068
FLCN	142.8	100%	98%	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500
GLIS2	75.5	99%	90%	Nephronophthisis 7, 611498
GPR98	145.8	99%	94%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
HEATR2	107.9	84%	77%	Ciliary dyskinesia, primary, 18, 614874
HYDIN	130.8	99%	99%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	148.9	100%	100%	Hydrolethalus syndrome, 236680
IFT122	145.6	100%	99%	Cranioectodermal dysplasia 1, 218330
IFT140	105.2	99%	96%	Short-rib thoracic dysplasia 9 with of without polydactyly, 266920
IFT172	108.6	99%	97%	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 Retinitis pigmentosa 71, 616394

IFT27	106.4	100%	99%	Bardet-Biedl syndrome 19, 615996
IFT43	110.9	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	55.9	79%	58%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
INPP5E	83.9	96%	88%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INVS	153.4	100%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	96.9	78%	74%	Senior-Loken syndrome 5, 609254
KIAA0586	110.4	96%	92%	Joubert syndrome 23,
KIF14	110.8	99%	89%	?Meckel syndrome 12, 616258
KIF7	70	91%	84%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
LCA5	130	95%	94%	Leber congenital amaurosis 5, 604537
LRRC6	163.6	96%	92%	Ciliary dyskinesia, primary, 19, 614935
LZTFL1	121.2	98%	90%	Bardet-Biedl syndrome 17, 615994
MAK	146.6	94%	93%	Retinitis pigmentosa 62, 614181
MKKS	222.1	89%	89%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MKS1	95.9	99%	96%	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 209900
MYO7A	127	98%	95%	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317
NEK1	113.4	98%	91%	Short rib-polydactyly syndrome, type IIA, 263520
NEK8	158	100%	100%	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NME8	107.5	97%	89%	Ciliary dyskinesia, primary, 6, 610852
NPHP1	122	99%	94%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	111.4	98%	91%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	127.5	99%	97%	Nephronophthisis 4, 606966

				Senior-Loken syndrome 4, 606996
OCRL	87	97%	88%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	29.6	74%	53%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
PCDH15	164.2	99%	98%	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067
PDE6D	93.9	100%	100%	?Joubert syndrome 22, 615665
PDZD7	77.7	100%	89%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472
PKD1	23.2	39%	30%	Polycystic kidney disease, adult type I, 173900
PKD2	95.3	87%	85%	Polycystic kidney disease 2, 613095
PKHD1	154	100%	99%	Polycystic kidney and hepatic disease, 263200
POC1A	132.6	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
PTPRQ	105	92%	87%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RP1	123.7	99%	96%	Retinitis pigmentosa 1, 180100 {Hypertriglyceridemia, susceptibility to}, 145750
RP2	117.2	100%	98%	Retinitis pigmentosa 2, 312600
RPGR	59.2	81%	66%	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	145.2	100%	99%	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194
RPGRIP1L	133.2	95%	93%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RSPH1	151.3	100%	100%	Ciliary dyskinesia, primary, 24, 615481
RSPH4A	128.9	98%	96%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	123.6	100%	97%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	61.1	83%	78%	No OMIM disease

				Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36)
SDCCAG8	116.7	100%	96%	Senior-Loken syndrome 7, 613615
SPAG1	71.3	91%	82%	Ciliary dyskinesia, primary, 28, 615505
SPATA7	128	96%	86%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
TBC1D32	80.1	97%	91%	No OMIM disease Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36)
TCTN1	98.8	94%	92%	Joubert syndrome 13, 614173
TCTN2	143.3	98%	95%	Meckel syndrome 8, 613885
TCTN3	116.3	99%	98%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TMEM138	102.7	99%	97%	Joubert syndrome 16, 614465
TMEM216	129.4	100%	97%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	86.1	99%	95%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397
TMEM237	96.1	99%	97%	Joubert syndrome 14, 614424
TMEM67	70.3	91%	84%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TOPORS	234.8	100%	100%	Retinitis pigmentosa 31, 609923
TRIM32	134.2	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TTBK2	144.9	100%	99%	Spinocerebellar ataxia 11, 604432
TTC21B	94.2	98%	94%	Nephronophthisis 12, 613820 Asphyxiating thoracic dystrophy 4, 613819
TTC8	88.4	98%	88%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TULP1	88.6	98%	93%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
USH1C	99.1	98%	95%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092

USH1G	153.6	95%	93%	Usher syndrome, type 1G, 606943
USH2A	156.8	99%	98%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809
VHL	91.1	98%	75%	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400
WDPCP	107.1	94%	93%	?Bardet-Biedl syndrome 15, 615992
WDR19	133.5	100%	98%	Asphyxiating thoracic dystrophy 5, 614376 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378
WDR34	91.1	94%	89%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	143.9	97%	94%	Cranioectodermal dysplasia 2, 613610 Short rib-polydactyly syndrome, type V, 614091
WDR60	103.3	98%	94%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
XPNPEP3	128.2	97%	97%	Nephronophthisis-like nephropathy 1, 613159
ZMYND10	135	100%	100%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	226.6	100%	100%	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
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 : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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