CILIOPATHIES GENE PANEL DG 2.7/DG 2.8

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AHI1	151.9	98%	94%	Joubert syndrome-3, 608629
ALMS1	197.7	99%	99%	Alstrom syndrome, 203800
ANKS6	99.5	93%	89%	Nephronophthisis 16, 615382
ARL13B	100.4	99%	92%	Joubert syndrome 8, 612291
ARL6	99	97%	91%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	135.5	91%	89%	Ciliary dyskinesia, primary, 23, 615451
ATXN10	166.4	99%	96%	Spinocerebellar ataxia 10, 603516
B9D1	119.3	92%	91%	?Meckel syndrome 9, 614209
B9D2	123.3	100%	100%	Meckel syndrome 10, 614175
BBIP1	150.1	98%	92%	?Bardet-Biedl syndrome 18, 615995
BBS1	162.3	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	179.1	100%	99%	Bardet-Biedl syndrome 10, 615987
BBS12	225	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	210.1	100%	99%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	163.9	99%	97%	Bardet-Biedl syndrome 4, 615982
BBS5	122.1	95%	89%	Bardet-Biedl syndrome 5, 615983
BBS7	135.5	97%	92%	Bardet-Biedl syndrome 7, 615984
BBS9	124.1	96%	93%	Bardet-Biedl syndrome 9, 615986
C21orf59	160.1	99%	95%	Ciliary dyskinesia, primary, 26, 615500
C2CD3	163.2	95%	95%	?Orofaciodigital syndrome XIV, 615948
C5orf42	136.5	98%	94%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CC2D2A	137.5	98%	96%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284

CCDC103	114.9	99%	98%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	135.4	99%	99%	Ciliary dyskinesia, primary, 20, 615067
CCDC151	132	99%	98%	Ciliary dyskinesia, primary, 30, 616037
CCDC28B	90.5	99%	98%	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC39	90.9	96%	90%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	130.4	98%	97%	Ciliary dyskinesia, primary, 15, 613808
CCDC41	113.4	97%	89%	Nephronophthisis 18, 615862
CCDC65	109.3	99%	95%	Ciliary dyskinesia, primary, 27, 615504
CCNO	99	99%	96%	Ciliary dyskinesia, primary, 29, 615872
CENPF	151.1	99%	97%	Stromme syndrome, 243605
CEP104	141.5	99%	97%	Joubert syndrome 25, 616781
CEP120	145.3	99%	98%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	98.4	99%	96%	Nephronophthisis 15, 614845
CEP290	77.4	88%	77%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	96.7	97%	90%	Joubert syndrome 15, 614464
CSPP1	119.4	99%	96%	Joubert syndrome 21, 615636
DCDC2	160	99%	99%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DDX59	184.5	99%	98%	Orofaciodigital syndrome V, 174300
DNAAF1	128.5	100%	99%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	104.1	99%	95%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	97.1	98%	92%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	150.4	99%	98%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	144.4	99%	98%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	130.3	98%	97%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	163.5	97%	94%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	146.1	99%	97%	No OMIM phenotype
DNAL1	115.8	95%	83%	Ciliary dyskinesia, primary, 16, 614017
DNHD1	184.4	100%	99%	No OMIM phenotype
DRC1	105.8	99%	97%	Ciliary dyskinesia, primary, 21, 615294

DYNC2H1	102.9	95%	86%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYX1C1	88.3	97%	85%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EVC	117.3	93%	90%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	125.8	96%	92%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC8	169.7	100%	99%	No OMIM phenotype Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78)
GAS8	166.7	99%	99%	Ciliary dyskinesia, primary, 33, 616726
GLIS2	106.8	99%	97%	Nephronophthisis 7, 611498
HEATR2	119.8	90%	83%	Ciliary dyskinesia, primary, 18, 614874
HYDIN	151.7	99%	99%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	180.7	100%	100%	Hydrolethalus syndrome, 236680
IFT122	164.4	100%	99%	Cranioectodermal dysplasia 1, 218330
IFT140	124.5	99%	98%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	128.2	99%	99%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	131.5	99%	97%	?Bardet-Biedl syndrome 19, 615996
IFT43	130.6	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	66.9	84%	67%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
INPP5E	105.1	96%	91%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	176.4	100%	99%	Nephronophthisis 2, infantile, 602088
IQCB1	105.9	86%	75%	Senior-Loken syndrome 5, 609254
KIAA0586	126.6	97%	91%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIF14	129.2	97%	90%	?Meckel syndrome 12, 616258
KIF7	93.4	95%	88%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
LCA5	141.6	97%	95%	Leber congenital amaurosis 5, 604537
LRRC6	180.2	96%	91%	Ciliary dyskinesia, primary, 19, 614935

LZTFL1	136.5	98%	94%	Bardet-Biedl syndrome 17, 615994
MKKS	239.6	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	113.5	99%	98%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
NEK1	124	97%	93%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	187.6	100%	99%	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NME8	118.2	95%	89%	Ciliary dyskinesia, primary, 6, 610852
NPHP1	141	99%	97%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	128.7	98%	94%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	148	99%	99%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
OCRL	152.2	99%	97%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	56.1	84%	71%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
PDE6D	118.9	100%	99%	?Joubert syndrome 22, 615665
PKD1	28.2	42%	33%	Polycystic kidney disease, adult type I, 173900
PKD2	119.8	91%	87%	Polycystic kidney disease 2, 613095
PKHD1	173.7	99%	99%	Polycystic kidney and hepatic disease, 263200
POC1A	150.5	100%	99%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RPGRIP1L	153.6	95%	93%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RSPH1	181	100%	99%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	141.8	98%	96%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	157.3	97%	95%	Ciliary dyskinesia, primary, 11, 612649

RSPH9	150.8	99%	98%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	76.2	88%	79%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36)
SDCCAG8	136.3	99%	96%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SPAG1	93.2	95%	89%	Ciliary dyskinesia, primary, 28, 615505
SPATA7	136.3	97%	91%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
TBC1D32	87.1	96%	90%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36)
TCTN1	117.4	95%	92%	Joubert syndrome 13, 614173
TCTN2	163.8	98%	95%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	135.8	99%	99%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TMEM138	133.5	100%	99%	Joubert syndrome 16, 614465
TMEM216	159.2	100%	99%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	103.4	99%	98%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	115.6	99%	96%	Joubert syndrome 14, 614424
TMEM67	78.9	92%	83%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TRIM32	152.9	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TTBK2	169.4	100%	99%	Spinocerebellar ataxia 11, 604432
TTC21B	111.6	99%	96%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	107.8	100%	99%	Ciliary dyskinesia, primary, 35, 617092
TTC8	109.1	98%	93%	Bardet-Biedl syndrome 8, 615985

				?Retinitis pigmentosa 51, 613464
TULP1	108.1	98%	93%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
VHL	120.5	95%	88%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
WDPCP	121.7	92%	88%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	153.5	99%	97%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	112.3	98%	95%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	167.2	98%	96%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	120.8	98%	95%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
XPNPEP3	148.8	98%	97%	Nephronophthisis-like nephropathy 1, 613159
ZMYND10	155.6	99%	99%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	279.5	100%	100%	Joubert syndrome 19, 614844 Nephronophthisis 14, 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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 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