CILIOPATHIES GENE PANEL DG 2.9 /DG 2.10 (127 genes)

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AHI1	176.9	99%	98%	Joubert syndrome-3, 608629
ALMS1	208.7	99%	99%	Alstrom syndrome, 203800
ANKS6	95.2	93%	90%	Nephronophthisis 16, 615382
ARL13B	117.5	99%	97%	Joubert syndrome 8, 612291
ARL6	121.4	100%	97%	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	135.4	91%	90%	Ciliary dyskinesia, primary, 23, 615451
ATXN10	158.1	99%	97%	Spinocerebellar ataxia 10, 603516
B9D1	127.2	92%	91%	?Meckel syndrome 9, 614209
B9D2	126.9	100%	100%	Meckel syndrome 10, 614175
BBIP1	170.9	99%	95%	?Bardet-Biedl syndrome 18, 615995
BBS1	178.6	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	195.4	100%	99%	Bardet-Biedl syndrome 10, 615987
BBS12	237	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	201.1	100%	99%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	153.5	99%	98%	Bardet-Biedl syndrome 4, 615982
BBS5	144	98%	96%	Bardet-Biedl syndrome 5, 615983
BBS7	156.1	99%	95%	Bardet-Biedl syndrome 7, 615984
BBS9	139.9	97%	95%	Bardet-Biedl syndrome 9, 615986
C21orf59	164.8	99%	96%	Ciliary dyskinesia, primary, 26, 615500
C2CD3	155.7	95%	95%	?Orofaciodigital syndrome XIV, 615948
C5orf42	154.1	99%	97%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CC2D2A	144.7	99%	97%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284

CCDC103	118.3	100%	99%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	132	100%	99%	Ciliary dyskinesia, primary, 20, 615067
CCDC151	134	100%	99%	Ciliary dyskinesia, primary, 30, 616037
CCDC28B	79.3	100%	98%	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC39	112.3	99%	96%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	135.7	99%	98%	Ciliary dyskinesia, primary, 15, 613808
CCDC41	126.7	99%	96%	Nephronophthisis 18, 615862
CCDC65	101.2	99%	96%	Ciliary dyskinesia, primary, 27, 615504
CCNO	101.5	99%	96%	Ciliary dyskinesia, primary, 29, 615872
CENPF	159.3	99%	98%	Stromme syndrome, 243605
CEP104	150.1	99%	97%	Joubert syndrome 25, 616781
CEP120	163	100%	99%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	100.3	99%	97%	Nephronophthisis 15, 614845
CEP290	92.8	95%	87%	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	98.2	98%	93%	Joubert syndrome 15, 614464
CSPP1	133.7	99%	98%	Joubert syndrome 21, 615636
DCDC2	180	100%	99%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DDX59	200.6	100%	99%	Orofaciodigital syndrome V, 174300
DNAAF1	125.5	100%	97%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	113.2	99%	97%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	95.4	98%	94%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	163.9	99%	99%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	159.6	100%	99%	No OMIM phenotype
DNAH5	151.6	99%	99%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	133.4	99%	98%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	166	96%	92%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	135.4	99%	97%	Ciliary dyskinesia, primary, 34, 617091
DNAL1	105.1	98%	92%	Ciliary dyskinesia, primary, 16, 614017
DNHD1	189	100%	100%	No OMIM phenotype

				?Global developmental delay (Anazi (2016) Mol Psychiatry epub,epub)
DRC1	108	99%	98%	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	110.6	98%	91%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYX1C1	101.6	99%	96%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EVC	126.1	94%	91%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	137.8	96%	94%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC8	181.3	100%	100%	No OMIM phenotype Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78)
EXTL3	221.3	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
GAS8	165.1	99%	99%	Ciliary dyskinesia, primary, 33, 616726
GLIS2	108.7	99%	97%	Nephronophthisis 7, 611498
HEATR2	123.9	90%	83%	Ciliary dyskinesia, primary, 18, 614874
HYDIN	143.3	99%	99%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	174.4	100%	100%	Hydrolethalus syndrome, 236680
IFT122	165.7	100%	99%	Cranioectodermal dysplasia 1, 218330
IFT140	124.5	99%	98%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	125.7	100%	99%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	133.2	100%	98%	?Bardet-Biedl syndrome 19, 615996
IFT43	128	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	79.6	92%	79%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
INPP5E	109	97%	92%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INVS	166	100%	100%	Nephronophthisis 2, infantile, 602088
IQCB1	122	92%	81%	Senior-Loken syndrome 5, 609254
KIAA0586	133.4	98%	94%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIF14	151.4	99%	95%	?Meckel syndrome 12, 616258
KIF7	95.3	95%	89%	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131

				?Hydrolethalus syndrome 2, 614120
LCA5	178	98%	96%	Leber congenital amaurosis 5, 604537
LRRC6	183.1	98%	95%	Ciliary dyskinesia, primary, 19, 614935
LZTFL1	142	99%	98%	Bardet-Biedl syndrome 17, 615994
MKKS	216.9	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	114.6	99%	99%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
NEK1	142.4	99%	96%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	192.1	100%	100%	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NME8	128.5	97%	93%	Ciliary dyskinesia, primary, 6, 610852
NPHP1	154.6	99%	98%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	146	99%	97%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	152.6	100%	99%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
OCRL	140.5	99%	98%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	59.2	87%	75%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
PDE6D	126.1	100%	99%	?Joubert syndrome 22, 615665
PKD1	30.1	41%	33%	Polycystic kidney disease, adult type I, 173900
PKD2	129.6	92%	88%	Polycystic kidney disease 2, 613095
PKHD1	176.8	100%	99%	Polycystic kidney and hepatic disease, 263200
POC1A	144.7	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RPGRIP1L	160.8	96%	95%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561

RSPH1	172.2	100%	100%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	145.1	99%	98%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	173	98%	96%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	156.2	99%	98%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	96.5	94%	85%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36)
SDCCAG8	155	99%	98%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SPAG1	103.9	97%	91%	Ciliary dyskinesia, primary, 28, 615505
SPATA7	149.2	98%	95%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
TBC1D32	100.8	98%	94%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36)
TCTN1	116.8	96%	93%	Joubert syndrome 13, 614173
TCTN2	166.2	99%	97%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	133.6	100%	99%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TMEM138	128	100%	99%	Joubert syndrome 16, 614465
TMEM216	147.1	99%	99%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	105.5	99%	98%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	126	99%	98%	Joubert syndrome 14, 614424
TMEM67	93.3	95%	89%	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TRIM32	147.3	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TTBK2	165.1	100%	99%	Spinocerebellar ataxia 11, 604432
TTC21B	132.4	99%	98%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819

TTC25	102.4	99%	99%	Ciliary dyskinesia, primary, 35, 617092
TTC26	173.1	100%	99%	No OMIM phenotype
TTC8	118.3	99%	98%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TULP1	116.2	98%	95%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
VHL	126	97%	90%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
WDPCP	137.3	95%	91%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	170	100%	99%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	122	99%	96%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	186.3	99%	98%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	131.2	99%	97%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
XPNPEP3	142.9	98%	97%	Nephronophthisis-like nephropathy 1, 613159
ZMYND10	161.7	100%	99%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	268.6	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 : April 14th, 2017
This list is accurate for panel version DG 2.9 and DG 2.10

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