CILIOPATHIES GENE PANEL DG 2.14 (146 genes)

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AHI1	139.3	99.2	95.1	Joubert syndrome 3, 608629
ALMS1	179.8	99.9	99.7	Alstrom syndrome, 203800
ANKS6	91.8	92.8	88.6	Nephronophthisis 16, 615382
ARL13B	97.3	98.9	92.8	Joubert syndrome 8, 612291
ARL6	85.2	99.8	95.3	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	129.1	94.4	93.4	Ciliary dyskinesia, primary, 23, 615451
ARMC9	127.3	99.8	98.5	Joubert syndrome 30, 617622
B9D1	115.3	92.1	91.4	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	110.9	100	100	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BBIP1	132	99.4	94.8	?Bardet-Biedl syndrome 18, 615995
BBS1	148.9	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	172.6	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	208.6	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	181.8	100	99.8	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	135.9	99.7	97.3	Bardet-Biedl syndrome 4, 615982
BBS5	81.1	95.8	84.1	Bardet-Biedl syndrome 5, 615983
BBS7	120.7	98.1	91.7	Bardet-Biedl syndrome 7, 615984
BBS9	112.9	96	93.8	Bardet-Biedl syndrome 9, 615986
C11orf70	60.7	92.3	82.2	Ciliary dyskinesia, primary, 38, 618063
C21orf2	104.4	99.9	98.7	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C21orf59	145.8	98.7	94.6	Ciliary dyskinesia, primary, 26, 615500
C2CD3	143.1	95.8	95.6	?Orofaciodigital syndrome XIV, 615948
C5orf42	122.8	98.6	95.5	Joubert syndrome 17, 614615

				Orofaciodigital syndrome VI, 277170
C8orf37	126.4	100	99	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
CC2D2A	127.4	99.5	97.1	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC103	116.8	100	99	Ciliary dyskinesia, primary, 17, 614679
CCDC114	120.7	100	99.6	Ciliary dyskinesia, primary, 20, 615067
CCDC151	116.2	100	99.7	Ciliary dyskinesia, primary, 30, 616037
CCDC28B	83.7	100	98.6	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC39	74.3	96.6	88.9	Ciliary dyskinesia, primary, 14, 613807
CCDC40	126.5	98.9	97.8	Ciliary dyskinesia, primary, 15, 613808
CCDC65	105.9	99.7	97.6	Ciliary dyskinesia, primary, 27, 615504
CCNO	103.1	99	95.6	Ciliary dyskinesia, primary, 29, 615872
CENPF	139.5	99.5	97.6	Stromme syndrome, 243605
CEP104	119.9	99	97.9	Joubert syndrome 25, 616781
CEP120	129.7	99.8	98.1	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	94.2	99.9	98	Nephronophthisis 15, 614845
CEP290	66.1	88.4	76.7	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	83.5	97.7	89.6	Joubert syndrome 15, 614464
CEP55	129.5	100	99.9	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	96.7	98.3	89.1	Nephronophthisis 18, 615862
CSPP1	112	99.8	97.8	Joubert syndrome 21, 615636
DCDC2	150.5	99.9	99.6	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DDX59	151.7	99.7	97.6	Orofaciodigital syndrome V, 174300

DNAAF1	115.8	100	99.7	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	105.1	99.7	96.9	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	91.8	97.7	90.6	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	79.6	96.3	84.1	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
DNAAF5	107.9	84.5	78.2	Ciliary dyskinesia, primary, 18, 614874
DNAH11	134	99.8	98.4	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	136.9	100	99.7	No OMIM phenotype ?Lung hypoplasia, polycystic kidneys and hypertrophy of the heart (Yates (2017) Genet Med 19,1171)
DNAH5	123.8	99.7	98.5	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	135.3	100	100	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	156.6	98.4	95.5	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	138.3	100	97.3	Ciliary dyskinesia, primary, 34, 617091
DNAL1	99	95.7	84.5	Ciliary dyskinesia, primary, 16, 614017
DNHD1	173.1	100	99.9	No OMIM phenotype ?Diabetic retinopathy (Ung (2017) Vision Res epub) ?Global developmental delay (Anazi (2016) Mol Psychiatry epub,epub)
DRC1	97	99.9	98.6	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	90.5	96.6	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	95.1	99.3	96	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EVC	110.4	93.2	89.8	?Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EVC2	119.3	96.4	94.3	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC8	174.4	100	100	No OMIM phenotype Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78)
EXTL3	206.4	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FUZ	118.8	100	100	Neural tube defects, 182940
GAS8	150.7	99.8	99.4	Ciliary dyskinesia, primary, 33, 616726
GLIS2	109	99.9	98.2	Nephronophthisis 7, 611498
HYDIN	133.6	99.9	99.5	Ciliary dyskinesia, primary, 5, 608647
HYLS1	171.1	100	100	Hydrolethalus syndrome, 236680
IFT122	152	100	99.9	Cranioectodermal dysplasia 1, 218330
IFT140	114.7	99.9	99	Retinitis pigmentosa 80, 617781

				Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99.6	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	131.7	100	99.6	?Bardet-Biedl syndrome 19, 615996
IFT43	114.8	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	123.6	100	99.3	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	57.8	87.6	70.7	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92.9	88.3	81.2	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
INPP5E	89.1	95.8	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INTU	122	99.7	96.6	?Orofaciodigital syndrome XVII, 617926 ?Short-rib throacic dysplasia 20 with polydactyly, 617925
INVS	160.5	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92.2	89.3	75.4	Senior-Loken syndrome 5, 609254
KCTD3	137.8	99.7	97.9	No OMIM phenotype Ciliopathy and Joubert syndrome (Alfares (2017) Mol Genet Metab 121,91) Severe psychomotor retardation, seizure and cerebellar hypoplasia (Alazami (2015) Cell Rep 10,148)
KIAA0556	134.4	99.9	99.4	Joubert syndrome 26, 616784
KIAA0586	114.7	98.2	92.7	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	123.9	99.9	98.7	?Orofaciodigital syndrome XV, 617127
KIF14	111.3	98.1	89.9	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF7	85.7	93.5	88.9	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
LBR	87.8	93.3	83.9	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	127.8	97.3	95.7	Leber congenital amaurosis 5, 604537

LRRC6	137.9	94.7	91	Ciliary dyskinesia, primary, 19, 614935
LZTFL1	109.1	99.1	95.3	Bardet-Biedl syndrome 17, 615994
MAPKBP1	144.1	100	100	Nephronophthisis 20, 617271
MKKS	208.5	83.2	83.1	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	114.5	99.9	98.5	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
NEK1	103.2	98.1	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK8	171.4	100	99.9	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NME8	105.6	97.5	91	Ciliary dyskinesia, primary, 6, 610852
NPHP1	117.6	98.8	96.4	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	115.6	99.4	96.1	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	136.7	99.9	99.3	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
OCRL	122.2	98.8	96.3	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	51.5	84	67.8	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
PDE6D	106.1	100	99.9	?Joubert syndrome 22, 615665
PIBF1	66	96.6	82.4	Joubert syndrome 33, 617767
PIH1D3	70.5	94.3	79.6	Ciliary dyskinesia, primary, 36, X-linked, 300991
PKD1	28.1	42.6	34.5	Polycystic kidney disease 1, 173900
PKD2	110.6	89.3	84.2	Polycystic kidney disease 2, 613095
PKHD1	154.9	100	99.7	Polycystic kidney disease 4, with or without hepatic disease, 263200
POC1A	133.8	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813

RPGRIP1L	126.2	96.4	93.9	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RSPH1	146.1	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	132.5	99.7	97.5	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	139	98.3	96.5	Ciliary dyskinesia, primary, 11, 612649
RSPH9	127.4	100	99.6	Ciliary dyskinesia, primary, 12, 612650
SCLT1	69.2	90.5	80	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36)
SDCCAG8	123.9	99.8	97.4	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SPAG1	87.3	96.3	88.2	Ciliary dyskinesia, primary, 28, 615505
SPATA7	119.6	97.8	90.8	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
TBC1D32	81.2	96.6	91.2	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36)
TCTEX1D2	126.1	100	99.3	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	98.8	95.7	92.8	Joubert syndrome 13, 614173
TCTN2	144.2	99.5	97	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	127.6	100	99.8	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TMEM107	163.8	100	100	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM138	100.2	100	99.5	Joubert syndrome 16, 614465
TMEM216	111.9	100	98.7	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	111.5	100	99.9	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100.7	99.8	98.3	Joubert syndrome 14, 614424
TMEM67	72.9	93.3	83.4	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361

				Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TRAF3IP1	90.3	96.3	92.8	Senior-Loken syndrome 9, 616629
TRIM32	141.2	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, type 2H, 254110
TTBK2	123.2	100	98.9	Spinocerebellar ataxia 11, 604432
TTC21B	100.7	99.7	97.6	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	103.4	100	99.5	Ciliary dyskinesia, primary, 35, 617092
TTC26	134.3	99.8	98.3	No OMIM phenotype
TTC8	106.9	97.9	92	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TULP1	97.8	96.8	91.7	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
VHL	119.7	92.6	85.3	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
WDPCP	107.3	93.9	88.9	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	132.1	99.8	98.1	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR34	106.6	99.5	96.2	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	145.1	99.3	97.7	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	114.2	99.1	96.3	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
XPNPEP3	134	100	99.2	Nephronophthisis-like nephropathy 1, 613159
ZMYND10	136.5	100	100	Ciliary dyskinesia, primary, 22, 615444
ZNF423	250.8	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 : September 11th, 2018.
This list is accurate for panel version DG 2.14
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors