CILIOPATHIES GENE PANEL DG 2.11 (140 genes)

Gene	Median	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AHI1	139.1	99	95	Joubert syndrome-3, 608629
ALMS1	179.4	99	99	Alstrom syndrome, 203800
ANKS6	91.8	92	88	Nephronophthisis 16, 615382
ARL13B	97.3	98	92	Joubert syndrome 8, 612291
ARL6	85.2	99	95	Bardet-Biedl syndrome 3, 600151 ?Retinitis pigmentosa 55, 613575 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	121.6	91	89	Ciliary dyskinesia, primary, 23, 615451
ARMC9	134.8	99	98	Joubert syndrome 30, 617622
B9D1	115.4	92	91	?Meckel syndrome 9, 614209
B9D2	111	100	100	Meckel syndrome 10, 614175
BBIP1	131.9	99	94	?Bardet-Biedl syndrome 18, 615995
BBS1	149.1	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	172.3	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	208.7	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	181.9	100	99	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	135.2	99	97	Bardet-Biedl syndrome 4, 615982
BBS5	110.5	97	90	Bardet-Biedl syndrome 5, 615983
BBS7	120.5	98	91	Bardet-Biedl syndrome 7, 615984
BBS9	112.5	96	93	Bardet-Biedl syndrome 9, 615986
C21orf2	104.8	99	98	No OMIM phenotype Retinal dystrophy,early-onset with macular staphyloma (Khan (2015) Br J Ophtalmol 99,1725) Cone-rod dystrophy (Abu-Safieh (2013) Genome Res 23,236) Jeune syndrome (Wheway (2015) Nat Cell Biol 17,1074)
C21orf59	145.8	98	94	Ciliary dyskinesia, primary, 26, 615500
C2CD3	143.1	95	95	?Orofaciodigital syndrome XIV, 615948
C5orf42	122.4	98	95	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170

C8orf37	126.4	100	99	Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
CC2D2A	127.4	99	97	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC103	117	100	99	Ciliary dyskinesia, primary, 17, 614679
CCDC114	120.7	100	99	Ciliary dyskinesia, primary, 20, 615067
CCDC151	113.2	100	99	Ciliary dyskinesia, primary, 30, 616037
CCDC28B	83.7	100	98	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC39	76.2	96	89	Ciliary dyskinesia, primary, 14, 613807
CCDC40	126.6	98	97	Ciliary dyskinesia, primary, 15, 613808
CCDC41	96.9	98	89	Nephronophthisis 18, 615862
CCDC65	105.9	99	97	Ciliary dyskinesia, primary, 27, 615504
CCNO	103	99	95	Ciliary dyskinesia, primary, 29, 615872
CENPF	139.7	99	97	Stromme syndrome, 243605
CEP104	120.1	99	97	Joubert syndrome 25, 616781
CEP120	128.1	99	98	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	94.4	99	97	Nephronophthisis 15, 614845
CEP290	66	88	76	Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991
CEP41	83.6	97	89	Joubert syndrome 15, 614464
CEP55	129.5	100	99	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly ,236500
CSPP1	111.9	99	97	Joubert syndrome 21, 615636
DCDC2	150.6	99	99	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DDX59	149.8	99	97	Orofaciodigital syndrome V, 174300
DNAAF1	115.8	100	99	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	105.2	99	97	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	91.8	97	90	Ciliary dyskinesia, primary, 2, 606763
DNAH11	134	99	98	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884

DNAH17	136.7	100	99	No OMIM phenotype
DNAH5	123.8	99	98	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	127.9	98	97	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	156.6	98	95	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	138.6	100	97	Ciliary dyskinesia, primary, 34, 617091
DNAL1	98.8	95	84	Ciliary dyskinesia, primary, 16, 614017
DNHD1	173.3	100	99	No OMIM phenotype ?Global developmental delay (Anazi (2016) Mol Psychiatry epub,epub)
DRC1	97.2	99	98	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	90.4	96	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYX1C1	79.6	96	84	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
EVC	110.7	93	89	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	119	96	94	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC8	174.3	100	100	No OMIM phenotype Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78)
EXTL3	206.6	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FUZ	116.2	100	100	Neural tube defects, 182940
GAS8	150.8	99	99	Ciliary dyskinesia, primary, 33, 616726
GLIS2	109.2	100	98	Nephronophthisis 7, 611498
HEATR2	106.5	85	79	Ciliary dyskinesia, primary, 18, 614874
HYDIN	133.1	99	99	Ciliary dyskinesia, primary, 5, 608647
HYLS1	171.2	100	100	Hydrolethalus syndrome, 236680
IFT122	152.3	100	99	Cranioectodermal dysplasia 1, 218330
IFT140	115	100	99	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	116.5	100	99	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	131.8	100	99	?Bardet-Biedl syndrome 19, 615996
IFT43	114.8	100	100	Cranioectodermal dysplasia 3, 614099
IFT52	123.6	100	99	No OMIM phenotype
IFT80	57.6	87	70	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263

INPP5E	89.3	95	90	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INTU	123.8	99	96	
INVS	159.6	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92.2	89	75	Senior-Loken syndrome 5, 609254
KIAA0556	134.6	99	99	?Joubert syndrome 26, 616784
KIAA0586	114.7	98	92	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	123.9	99	98	?Orofaciodigital syndrome XV, 617127
KIF14	111.3	98	89	?Meckel syndrome 12, 616258
KIF7	85.8	93	88	Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120
LBR	87.9	93	83	Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471
LCA5	127.6	97	95	Leber congenital amaurosis 5, 604537
LRRC6	138	94	91	Ciliary dyskinesia, primary, 19, 614935
LZTFL1	108.7	99	95	Bardet-Biedl syndrome 17, 615994
MAPKBP1	144.3	100	100	No OMIM phenotype
MKKS	225.1	89	89	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	115.6	99	98	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000
NEK1	103	98	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	171.6	100	99	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NME8	105.7	97	91	Ciliary dyskinesia, primary, 6, 610852
NPHP1	117.6	98	96	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	114.3	99	95	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387

				Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	137.3	99	99	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
OCRL	123	98	96	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	53	84	68	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
PDE6D	106.1	100	99	?Joubert syndrome 22, 615665
PIH1D3	70.9	94	80	Ciliary dyskinesia, primary, 36, X-linked , 300991
PKD1	28.1	42	34	Polycystic kidney disease, adult type I, 173900
PKD2	110.7	89	84	Polycystic kidney disease 2, 613095
PKHD1	155	100	99	Polycystic kidney and hepatic disease, 263200
POC1A	134.1	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RPGRIP1L	126.2	96	93	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RSPH1	146.2	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	132.6	99	97	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	139.4	98	96	Ciliary dyskinesia, primary, 11, 612649
RSPH9	127.6	100	99	Ciliary dyskinesia, primary, 12, 612650
SCLT1	69.1	90	80	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36)
SDCCAG8	124.4	99	97	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SPAG1	87.3	96	88	Ciliary dyskinesia, primary, 28, 615505
SPATA7	119.4	97	90	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
TBC1D32	81.2	96	91	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36)
TCTN1	100.5	96	94	Joubert syndrome 13, 614173

TCTN2	144.2	99	97	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	127.5	100	99	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TMEM107	164.2	100	100	?Joubert syndrome 29 , 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM138	100.3	100	99	Joubert syndrome 16, 614465
TMEM216	112	100	98	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	111.8	100	99	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100.6	99	98	Joubert syndrome 14, 614424
TMEM67	72.9	93	83	COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TRAF3IP1	90.2	96	92	Senior-Loken syndrome 9, 616629
TRIM32	141.3	100	100	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TTBK2	148.5	100	99	Spinocerebellar ataxia 11, 604432
TTC21B	100.6	99	97	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	103.6	100	99	Ciliary dyskinesia, primary, 35, 617092
TTC26	134.4	99	98	No OMIM phenotype
TTC8	106.8	97	92	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TULP1	98	96	91	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
VHL	119.6	92	85	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700

				von Hippel-Lindau syndrome, 193300
WDPCP	107.3	93	88	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	132	99	98	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	106.7	99	96	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	145.1	99	97	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	114.2	99	96	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
XPNPEP3	134.8	98	97	Nephronophthisis-like nephropathy 1, 613159
ZMYND10	136.7	100	100	Ciliary dyskinesia, primary, 22, 615444
ZNF423	251.1	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 2.11
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors