CILIOPATHIES GENE PANEL DG 2.16 (151 genes)

Releasedate: 07-06-2019

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ADAMTS9	115,7	99.5%	97.4%	No OMIM phenotype
AHI1	129,8	99.7%	98.3%	Joubert syndrome 3, 608629
ALMS1	172,8	100.0%	99.7%	Alstrom syndrome, 203800
ANKS6	94,2	98.3%	94.4%	Nephronophthisis 16, 615382
ARL13B	102,2	100.0%	99.4%	Joubert syndrome 8, 612291
ARL6	100,3	99.8%	98.2%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC4	107,2	94.4%	93.5%	Ciliary dyskinesia, primary, 23, 615451
ARMC9	124,8	100.0%	99.3%	Joubert syndrome 30, 617622
B9D1	103,7	92.2%	92.1%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	105,8	100.0%	100.0%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BBIP1	119,7	97.3%	90.3%	?Bardet-Biedl syndrome 18, 615995
BBS1	146,4	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	158,1	100.0%	99.9%	Bardet-Biedl syndrome 10, 615987
BBS12	187,1	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	150,7	99.9%	99.6%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	110,2	99.9%	99.2%	Bardet-Biedl syndrome 4, 615982
BBS5	98,5	98.0%	93.3%	Bardet-Biedl syndrome 5, 615983
BBS7	142,9	99.1%	96.5%	Bardet-Biedl syndrome 7, 615984
BBS9	113,6	98.6%	94.4%	Bardet-Biedl syndrome 9, 615986
C11orf70	NC	NC	NC	Ciliary dyskinesia, primary, 38, 618063
C21orf2	NC	NC	NC	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C21orf59	NC	NC	NC	Ciliary dyskinesia, primary, 26, 615500
C2CD3	116,9	95.8%	95.2%	Orofaciodigital syndrome XIV, 615948

C5orf42	NC	NC	NC	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8orf37	144,1	99.8%	99.4%	Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500 Retinitis pigmentosa 64, 614500
CC2D2A	111,7	99.0%	97.1%	COACH syndrome, 216360 Joubert syndrome 9, 612285 Meckel syndrome 6, 612284
CCDC103	116,2	100.0%	99.8%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	134,1	100.0%	99.8%	Ciliary dyskinesia, primary, 20, 615067
CCDC151	127,2	100.0%	100.0%	Ciliary dyskinesia, primary, 30, 616037
CCDC28B	84,3	100.0%	98.5%	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC39	86,7	99.4%	96.8%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	112	99.4%	98.4%	Ciliary dyskinesia, primary, 15, 613808
CCDC65	80,3	99.6%	97.1%	Ciliary dyskinesia, primary, 27, 615504
CCNO	130,9	100.0%	99.8%	Ciliary dyskinesia, primary, 29, 615872
CENPF	139,9	99.8%	98.7%	Stromme syndrome, 243605
CEP104	104	99.3%	97.5%	Joubert syndrome 25, 616781
CEP120	131,7	100.0%	99.4%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	89,3	99.8%	98.0%	Nephronophthisis 15, 614845
CEP290	82,6	97.3%	91.7%	?Bardet-Biedl syndrome 14, 615991 Joubert syndrome 5, 610188 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Senior-Loken syndrome 6, 610189
CEP41	78,2	98.9%	94.4%	Joubert syndrome 15, 614464
CEP55	124,5	100.0%	100.0%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	108,8	99.4%	96.6%	Nephronophthisis 18, 615862
CSPP1	119	100.0%	99.1%	Joubert syndrome 21, 615636
DCDC2	150	99.9%	99.8%	?Deafness, autosomal recessive 66, 610212 Nephronophthisis 19, 616217 Sclerosing cholangitis, neonatal, 617394
DDX59	141,5	100.0%	99.8%	Orofaciodigital syndrome V, 174300
DNAAF1	112,8	100.0%	99.5%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	150,1	99.9%	98.7%	Ciliary dyskinesia, primary, 10, 612518

DNAAF3	115	99.9%	98.5%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	94,1	99.9%	98.2%	Ciliary dyskinesia, primary, 25, 615482 {Dyslexia, susceptibility to, 1}, 127700
DNAAF5	107,6	95.4%	85.2%	Ciliary dyskinesia, primary, 18, 614874
DNAH1	158,1	100.0%	99.8%	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	131,3	99.8%	98.7%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	126	100.0%	99.6%	No OMIM phenotype ?Lung hypoplasia, polycystic kidneys and hypertrophy of the heart (Yates (2017) Genet Med 19,1171)
DNAH5	114,1	99.9%	99.1%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	115,4	100.0%	100.0%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	151,7	99.6%	96.6%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	116,3	100.0%	99.5%	Ciliary dyskinesia, primary, 34, 617091
DNAL1	104	99.3%	96.2%	Ciliary dyskinesia, primary, 16, 614017
DNHD1	148,3	100.0%	99.9%	No OMIM phenotype ?Diabetic retinopathy (Ung (2017) Vision Res epub) ?Global developmental delay (Anazi (2016) Mol Psychiatry epub,epub)
DRC1	91,2	100.0%	99.3%	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	102,2	98.8%	95.5%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	95,4	99.7%	97.0%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EVC	106,3	95.9%	92.4%	?Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EVC2	110,2	99.4%	96.3%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC8	152,6	100.0%	100.0%	No OMIM phenotype Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78)
EXTL3	184,1	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FUZ	128,5	100.0%	100.0%	{Neural tube defects, susceptibility to}, 182940
GAS8	127,3	100.0%	99.4%	Ciliary dyskinesia, primary, 33, 616726
GLIS2	129,6	100.0%	100.0%	Nephronophthisis 7, 611498
HYDIN	106,8	99.8%	98.9%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	156,6	100.0%	100.0%	Hydrolethalus syndrome, 236680
IFT122	120,5	99.9%	99.0%	Cranioectodermal dysplasia 1, 218330
IFT140	117,6	99.9%	99.2%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	94,5	100.0%	99.4%	Retinitis pigmentosa 71, 616394

				Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	115,8	100.0%	100.0%	?Bardet-Biedl syndrome 19, 615996
IFT43	112,4	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	121	100.0%	99.7%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	64,9	96.7%	84.7%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92,3	93.6%	89.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
INPP5E	116,8	100.0%	98.6%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INTU	115,4	99.8%	98.7%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	143,7	100.0%	99.9%	Nephronophthisis 2, infantile, 602088
IQCB1	93,3	91.6%	80.0%	Senior-Loken syndrome 5, 609254
KCTD3	129,3	99.9%	99.5%	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	126,6	100.0%	99.6%	Joubert syndrome 26, 616784
KIAA0586	117,7	97.0%	93.0%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	113,2	99.9%	98.7%	?Orofaciodigital syndrome XV, 617127
KIF14	116,6	99.6%	97.9%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF7	105,2	98.2%	93.5%	?Al-Gazali-Bakalinova syndrome, 607131 ?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
LBR	103	98.3%	91.5%	?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140 Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	139,9	99.8%	98.9%	Leber congenital amaurosis 5, 604537
LRRC56	130,7	100.0%	99.2%	Ciliary dyskinesia, primary, 39, 618254
LRRC6	139,3	99.8%	97.3%	Ciliary dyskinesia, primary, 19, 614935
LZTFL1	117	99.8%	99.2%	Bardet-Biedl syndrome 17, 615994
MAPKBP1	132,5	100.0%	100.0%	Nephronophthisis 20, 617271
MKKS	155,7	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231

				McKusick-Kaufman syndrome, 236700
MKS1	92,4	99.6%	97.8%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
NCAPG2	121,5	99.8%	98.2%	No OMIM phenotype
NEK1	115,9	99.7%	98.1%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK8	141,3	100.0%	99.9%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NME8	104,7	98.6%	93.8%	Ciliary dyskinesia, primary, 6, 610852
NPHP1	121,2	99.8%	98.5%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	121,4	99.8%	98.5%	Meckel syndrome 7, 267010 Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540
NPHP4	125,6	100.0%	99.7%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
OCRL	106,2	99.8%	98.3%	Dent disease 2, 300555 Lowe syndrome, 309000
OFD1	51,9	85.8%	70.8%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
PDE6D	114,7	100.0%	99.9%	?Joubert syndrome 22, 615665
PIBF1	74,3	99.2%	94.4%	Joubert syndrome 33, 617767
PIH1D3	74,7	98.4%	89.1%	Ciliary dyskinesia, primary, 36, X-linked, 300991
PKD1	35,9	43.0%	35.0%	Polycystic kidney disease 1, 173900
PKD2	102,3	98.7%	95.8%	Polycystic kidney disease 2, 613095
PKHD1	130,4	99.9%	99.4%	Polycystic kidney disease 4, with or without hepatic disease, 263200
POC1A	112,9	100.0%	100.0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RPGRIP1L	123,4	96.7%	95.4%	COACH syndrome, 216360 Joubert syndrome 7, 611560 Meckel syndrome 5, 611561
RSPH1	122,6	100.0%	99.9%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	139,5	99.9%	99.3%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	146,9	98.1%	95.3%	Ciliary dyskinesia, primary, 11, 612649

RSPH9	131,2	99.7%	97.1%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	90,3	95.8%	90.8%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36)
SDCCAG8	124,1	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SPAG1	101,6	99.1%	95.3%	Ciliary dyskinesia, primary, 28, 615505
SPATA7	122,7	99.4%	97.4%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232
TBC1D32	95,5	99.4%	96.5%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36)
TCTEX1D2	123,6	100.0%	99.4%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	94,8	95.6%	92.3%	Joubert syndrome 13, 614173
TCTN2	122,4	99.9%	99.0%	?Meckel syndrome 8, 613885 Joubert syndrome 24, 616654
TCTN3	116,3	100.0%	99.9%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TMEM107	148,7	100.0%	100.0%	?Joubert syndrome 29, 617562 Meckel syndrome 13, 617562 Orofaciodigital syndrome XVI, 617563
TMEM138	82,7	100.0%	99.2%	Joubert syndrome 16, 614465
TMEM216	88	99.7%	95.7%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	101,1	100.0%	99.3%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	117,7	100.0%	99.2%	Joubert syndrome 14, 614424
TMEM260	117,5	99.6%	97.6%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	83,1	99.1%	94.6%	?RHYNS syndrome, 602152 COACH syndrome, 216360 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991
TRAF3IP1	84,2	99.4%	97.1%	Senior-Loken syndrome 9, 616629
TRIM32	123	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TTBK2	108,4	99.9%	96.8%	Spinocerebellar ataxia 11, 604432
TTC21B	119,5	99.7%	98.8%	Nephronophthisis 12, 613820

				Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	93,7	100.0%	99.6%	Ciliary dyskinesia, primary, 35, 617092
TTC26	140,5	99.8%	99.2%	No OMIM phenotype
TTC8	115,2	99.8%	98.8%	?Retinitis pigmentosa 51, 613464 Bardet-Biedl syndrome 8, 615985
TULP1	117,9	100.0%	99.6%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
VHL	169,6	100.0%	98.3%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
WDPCP	106,7	97.8%	94.9%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	126,8	100.0%	99.2%	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR34	116,1	100.0%	100.0%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	141,8	99.7%	98.4%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR60	108,1	99.7%	98.1%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
XPNPEP3	99,9	100.0%	99.4%	Nephronophthisis-like nephropathy 1, 613159
ZMYND10	123	100.0%	100.0%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	192,9	100.0%	100.0%	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 : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors