CILIOPATHIES GENE DG 2.17 (159 genes)

Releasedate: 06-12-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACVR2B	124.2	99.9%	98.3%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS9	119.6	99.5%	97.8%	No OMIM Disease ID
AHI1	125.5	99.9%	97.6%	Joubert syndrome 3, 608629
ALMS1	178.0	100.0%	99.8%	Alstrom syndrome, 203800
ANKS6	101.2	99.3%	96.5%	Nephronophthisis 16, 615382
ARL13B	98.7	100.0%	99.7%	Joubert syndrome 8, 612291
ARL6	91.8	99.9%	97.7%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARMC4	107.7	94.4%	93.5%	Ciliary dyskinesia, primary, 23, 615451
ARMC9	129.0	100.0%	99.4%	Joubert syndrome 30, 617622
B9D1	111.4	92.2%	92.2%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	115.7	100.0%	100.0%	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BBIP1	116.1	98.5%	91.4%	?Bardet-Biedl syndrome 18, 615995
BBS1	156.1	100.0%	100.0%	Bardet-Biedl syndrome 1, 209900
BBS10	156.7	100.0%	100.0%	Bardet-Biedl syndrome 10, 615987
BBS12	193.6	100.0%	100.0%	Bardet-Biedl syndrome 12, 615989
BBS2	153.3	100.0%	99.7%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	113.2	100.0%	98.4%	Bardet-Biedl syndrome 4, 615982
BBS5	94.9	98.4%	92.3%	Bardet-Biedl syndrome 5, 615983
BBS7	136.8	99.0%	95.3%	Bardet-Biedl syndrome 7, 615984
BBS9	113.2	98.8%	94.8%	Bardet-Biedl syndrome 9, 615986
C11orf70	83.8	99.8%	95.3%	Ciliary dyskinesia, primary, 38, 618063
C21orf2	146.9	100.0%	99.4%	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
C21orf59	144.1	99.3%	96.3%	Ciliary dyskinesia, primary, 26, 615500
C2CD3	121.4	95.8%	95.3%	Orofaciodigital syndrome XIV, 615948

C5orf42	122.3	99.7%	97.4%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8orf37	146.4	100.0%	99.9%	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
CC2D2A	112.6	99.0%	97.0%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CCDC103	126.7	100.0%	100.0%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	148.7	100.0%	99.9%	Ciliary dyskinesia, primary, 20, 615067
CCDC151	141.6	100.0%	100.0%	Ciliary dyskinesia, primary, 30, 616037
CCDC28B	92.3	100.0%	99.1%	No OMIM disease ID
CCDC39	82.4	99.6%	96.0%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	120.1	99.5%	98.5%	Ciliary dyskinesia, primary, 15, 613808
CCDC65	84.8	99.8%	97.8%	Ciliary dyskinesia, primary, 27, 615504
CCNO	151.7	100.0%	99.9%	Ciliary dyskinesia, primary, 29, 615872
CENPF	143.9	99.8%	98.7%	Stromme syndrome, 243605
CEP104	108.8	99.3%	97.8%	Joubert syndrome 25, 616781
CEP120	131.3	100.0%	99.6%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	96.7	99.9%	98.4%	Nephronophthisis 15, 614845
CEP290	77.6	96.9%	88.7%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	79.1	98.7%	94.4%	Joubert syndrome 15, 614464
CEP55	123.2	100.0%	100.0%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	103.3	99.8%	96.2%	Nephronophthisis 18, 615862
CFAP53	135.5	99.2%	97.0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFC1	136.1	91.1%	82.3%	Heterotaxy, visceral, 2, autosomal, 605376
CSPP1	117.4	100.0%	99.4%	Joubert syndrome 21, 615636
DCDC2	158.0	100.0%	99.9%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DDX59	143.3	100.0%	99.7%	Orofaciodigital syndrome V, 174300

DNAAF1	118.6	100.0%	99.7%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	169.3	99.9%	98.9%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	128.5	100.0%	99.2%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	92.2	99.9%	98.2%	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	119.1	98.6%	90.6%	Ciliary dyskinesia, primary, 18, 614874
DNAH1	170.9	100.0%	99.8%	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	132.3	99.9%	98.9%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	134.9	100.0%	99.7%	Spermatogenic failure 39, 618643
DNAH5	115.4	100.0%	99.3%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	120.5	100.0%	100.0%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	163.8	99.7%	98.0%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	124.0	100.0%	99.7%	Ciliary dyskinesia, primary, 34, 617091
DNAL1	104.2	99.8%	94.5%	Ciliary dyskinesia, primary, 16, 614017
DNHD1	161.4	100.0%	99.9%	No OMIM Disease ID
DRC1	96.8	100.0%	99.5%	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	98.0	98.9%	94.3%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	94.8	99.6%	97.1%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EVC	113.0	96.8%	92.1%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	115.9	99.6%	97.1%	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EXOC8	164.5	100.0%	100.0%	No OMIM Disease ID
EXTL3	200.7	100.0%	100.0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FUZ	143.2	100.0%	100.0%	No OMIM Disease ID
GAS8	134.7	100.0%	99.7%	Ciliary dyskinesia, primary, 33, 616726
GDF1	62.1	99.4%	91.2%	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GLIS2	148.5	100.0%	100.0%	Nephronophthisis 7, 611498
HYDIN	111.4	99.9%	99.2%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	160.4	100.0%	100.0%	Hydrolethalus syndrome, 236680
IFT122	126.6	100.0%	99.6%	Cranioectodermal dysplasia 1, 218330
IFT140	127.6	100.0%	99.6%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	98.4	100.0%	99.5%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	122.9	100.0%	99.9%	?Bardet-Biedl syndrome 19, 615996

IFT43	119.5	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT52	120.0	100.0%	99.7%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	61.7	95.6%	81.0%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	90.5	93.0%	88.0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
INPP5E	131.1	100.0%	99.3%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INTU	112.9	99.8%	98.0%	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	147.7	100.0%	100.0%	Nephronophthisis 2, infantile, 602088
IQCB1	90.6	91.0%	79.0%	Senior-Loken syndrome 5, 609254
KCTD3	128.1	100.0%	99.7%	No OMIM Disease ID
KIAA0556	135.2	100.0%	99.8%	Joubert syndrome 26, 616784
KIAA0586	115.1	97.3%	92.6%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	117.1	99.9%	98.9%	?Orofaciodigital syndrome XV, 617127
KIF14	112.4	99.8%	97.7%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF7	120.4	99.3%	96.6%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
LBR	104.4	97.4%	90.4%	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	137.1	99.9%	98.9%	Leber congenital amaurosis 5, 604537
LRRC56	148.8	100.0%	99.8%	Ciliary dyskinesia, primary, 39, 618254
LRRC6	138.2	99.5%	96.2%	Ciliary dyskinesia, primary, 19, 614935
LZTFL1	116.5	99.9%	99.2%	Bardet-Biedl syndrome 17, 615994
MAPKBP1	144.1	100.0%	100.0%	Nephronophthisis 20, 617271
MKKS	161.5	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	98.8	99.9%	98.5%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000

MMP21	103.7	100.0%	99.5%	Heterotaxy, visceral, 7, autosomal, 616749
NCAPG2	121.6	99.6%	97.6%	Khan-Khan-Katsanis syndrome, 618460
NEK1	111.2	99.9%	98.0%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	153.7	100.0%	99.9%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NME8	103.2	98.9%	92.4%	Ciliary dyskinesia, primary, 6, 610852
NODAL	156.2	100.0%	100.0%	Heterotaxy, visceral, 5, 270100
NPHP1	119.7	99.8%	97.8%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	121.6	99.7%	98.3%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	134.2	100.0%	99.8%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
OCRL	106.2	99.9%	98.6%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	52.3	85.5%	70.0%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
PDE6D	121.7	100.0%	100.0%	?Joubert syndrome 22, 615665
PIBF1	71.9	99.3%	92.7%	Joubert syndrome 33, 617767
PIH1D3	71.3	97.6%	85.8%	Ciliary dyskinesia, primary, 36, X-linked, 300991
PIK3C2A	126.2	99.1%	96.6%	Oculoskeletodental syndrome, 618440
PKD1	41.1	44.3%	36.9%	Polycystic kidney disease 1, 173900
PKD2	105.1	99.2%	96.8%	Polycystic kidney disease 2, 613095
PKHD1	132.5	100.0%	99.5%	Polycystic kidney disease 4, with or without hepatic disease, 263200
POC1A	120.3	100.0%	100.0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RPGRIP1L	124.2	96.8%	95.8%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RSPH1	127.7	100.0%	100.0%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	148.8	100.0%	99.3%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	155.2	98.7%	96.4%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	143.1	99.9%	98.0%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	84.0	95.8%	89.0%	No OMIM Disease ID

SDCCAG8	123.5	100.0%	99.7%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SPAG1	102.2	98.7%	93.2%	Ciliary dyskinesia, primary, 28, 615505
SPATA7	120.5	99.7%	97.1%	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
TBC1D32	89.9	99.3%	95.5%	No OMIM Disease ID
TCTEX1D2	126.1	99.9%	99.0%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	96.0	95.7%	92.6%	Joubert syndrome 13, 614173
TCTN2	127.0	100.0%	99.0%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	121.0	100.0%	100.0%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TMEM107	161.8	100.0%	100.0%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM138	87.8	100.0%	99.0%	Joubert syndrome 16, 614465
TMEM216	92.0	99.9%	96.9%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	112.1	100.0%	99.7%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	114.5	99.9%	98.8%	Joubert syndrome 14, 614424
TMEM260	117.1	99.9%	97.8%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	80.6	99.3%	93.5%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TRAF3IP1	87.5	99.1%	96.7%	Senior-Loken syndrome 9, 616629
TRIM32	132.8	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TTBK2	110.8	99.9%	98.1%	Spinocerebellar ataxia 11, 604432
TTC21B	115.1	99.9%	98.8%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	100.1	100.0%	99.7%	Ciliary dyskinesia, primary, 35, 617092
TTC26	138.2	99.9%	99.0%	No OMIM Disease ID
TTC8	116.8	99.7%	97.8%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464

TULP1	128.8	100.0%	99.7%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
VHL	182.8	100.0%	99.8%	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
WDPCP	105.7	97.1%	93.6%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	125.3	100.0%	99.4%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	129.6	100.0%	100.0%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	137.8	99.5%	98.3%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR60	111.7	99.8%	97.8%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
XPNPEP3	103.7	100.0%	99.9%	Nephronophthisis-like nephropathy 1, 613159
ZIC3	155.4	100.0%	99.8%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMYND10	132.9	100.0%	100.0%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	215.2	100.0%	100.0%	Nephronophthisis 14, 614844 Joubert syndrome 19, 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 : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors