CILIOPATHIES GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
AHI1	104,3	100%	98%	Joubert syndrome-3, 608629
ALMS1	189,2	98%	98%	Alstrom syndrome, 203800
ANKS6	55,8	94%	80%	Nephronophthisis 16, 615382
ARL13B	126,4	100%	95%	Joubert syndrome 8, 612291
ARL6	153,9	100%	100%	Bardet-Biedl syndrome 3, 209900 {Bardet-Biedl syndrome 1, modifier of}, 209900 Retinitis pigmentosa 55, 613575
ARMC4	88,5	88%	85%	Ciliary dyskinesia, primary, 23, 615451
ATXN10	113,1	100%	99%	Spinocerebellar ataxia 10, 603516
B9D1	85,7	100%	93%	Meckel syndrome 9, 614209
B9D2	49,5	100%	100%	Meckel syndrome 10, 614175
BBS1	116,9	100%	98%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
BBS1	116,9	100%	98%	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 209900
BBS1	116,9	100%	98%	Bardet-Biedl syndrome 1, 209900
BBS10	115,8	100%	100%	Bardet-Biedl syndrome 10, 209900
BBS12	132,2	100%	100%	Bardet-Biedl syndrome 12, 209900
BBS2	114,4	100%	100%	Bardet-Biedl syndrome 2, 209900

BBS4	93,2	92%	86%	Bardet-Biedl syndrome 4, 209900
BBS5	147,4	100%	100%	Bardet-Biedl syndrome 5, 209900
BBS7	119,5	100%	98%	Bardet-Biedl syndrome 7, 209900
BBS9	124,7	100%	100%	Bardet-Biedl syndrome 9, 209900
C2orf71	105,4	99%	94%	Retinitis pigmentosa 54, 613428
C5orf42	121	100%	100%	Joubert syndrome 17, 614615
C8orf37	93,6	100%	100%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 -3
CC2D2A	94,8	98%	97%	Joubert syndrome 9, 612285 Meckel syndrome 6, 612284 COACH syndrome, 216360
CCDC103	104,9	100%	100%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	69,7	100%	96%	Ciliary dyskinesia, primary, 20, 615067
CCDC28B	72,7	100%	100%	{Bardet-Biedl syndrome, modifier of}, 209900
CCDC39	101,1	100%	99%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	86,1	97%	93%	Ciliary dyskinesia, primary, 15, 613808
CCDC65	73,8	98%	96%	Ciliary dyskinesia, primary, 27, 615504
CDH23	93,6	99%	97%	Usher syndrome, type 1D, 601067 Deafness, autosomal recessive 12, 601386 Usher syndrome, type 1D/F digenic, 601067
CEP164	74,7	97%	90%	Nephronophthisis 15, 614845

CEP290	95,3	100%	98%	Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 Leber congenital amaurosis 10, 611755 Meckel syndrome 4, 611134 Bardet-Biedl syndrome 14, 209900
CEP41	91	100%	100%	Joubert syndrome 15, 614464
CLRN1	153,9	100%	100%	?Usher syndrome, type 3A, 276902 Retinitis pigmentosa 61, 614180 -3
DFNB31	80,6	99%	98%	Deafness, autosomal recessive 31, 607084 Usher syndrome, type 2D, 611383
DNAAF3	66,6	88%	77%	Ciliary dyskinesia, primary, 2, 606763
DNAH11	113,5	100%	99%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH5	92,2	100%	98%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	120,1	100%	99%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	104,2	97%	94%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAL1	131	100%	100%	Ciliary dyskinesia, primary, 16, 614017
DRC1	74,1	100%	97%	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	114	99%	99%	Asphyxiating thoracic dystrophy 3, 613091 Short rib-polydactyly syndrome, type III, 263510 Short rib-polydactyly syndrome, type IIB, 615087
DYX1C1	88,6	100%	98%	{Dyslexia, susceptibility to, 1}, 127700
EVC	71,3	91%	87%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	97,4	93%	92%	Ellis-van Creveld syndrome, 225500
EXOC8	162,2	100%	100%	No OMIM phenotype Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78)

FAM161A	137,3	100%	100%	Retinitis pigmentosa 28, 606068
FLCN	98,7	99%	96%	Birt-Hogg-Dube syndrome, 135150 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Colorectal cancer, somatic, 114500
GLIS2	88,3	100%	97%	Nephronophthisis 7, 611498
GPR98	112	99%	98%	Febrile seizures, familial, 4, 604352 Usher syndrome, type 2C, 605472 Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472
HEATR2	67,2	82%	74%	Ciliary dyskinesia, primary, 18, 614874
HYDIN	90,8	88%	85%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	153,3	100%	100%	Hydrolethalus syndrome, 236680
IFT122	76,7	96%	92%	Cranioectodermal dysplasia 1, 218330
IFT140	80,2	99%	94%	Mainzer-Saldino syndrome, 266920
IFT43	83	100%	100%	Cranioectodermal dysplasia 3, 614099
IFT80	81	99%	92%	Asphyxiating thoracic dystrophy 2, 611263
INPP5E	75,8	99%	96%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INVS	120,4	99%	98%	Nephronophthisis 2, infantile, 602088
IQCB1	92,3	100%	93%	Senior-Loken syndrome 5, 609254
KIF7	68,8	89%	84%	Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990
LCA5	155	100%	98%	Leber congenital amaurosis 5, 604537
LRRC6	110,7	100%	100%	Ciliary dyskinesia, primary, 19, 614935

LZTFL1	85	100%	98%	Bardet-Biedl syndrome 17, 615994
MAK	80,4	96%	94%	Retinitis pigmentosa 62, 614181
MKKS	143,4	100%	100%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 209900
MKS1	101,9	98%	94%	Meckel syndrome 1, 249000 Bardet-Biedl syndrome 13, 209900
MYO7A	75,7	95%	89%	Usher syndrome, type 1B, 276900 Deafness, autosomal recessive 2, 600060 Deafness, autosomal dominant 11, 601317
NEK1	119,3	99%	99%	Short rib-polydactyly syndrome, type IIA, 263520
NEK8	104,7	100%	100%	?Nephronophthisis 9, 613824 ?Renal-hepatic-pancreatic dysplasia 2, 615415
NME8	103,9	100%	100%	Ciliary dyskinesia, primary, 6, 610852
NPHP1	114,1	100%	99%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	106,7	100%	99%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	87,7	98%	93%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
OCRL	59,5	98%	96%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	36,4	89%	76%	Oral-facial-digital syndrome 1, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 Joubert syndrome 10, 300804
PCDH15	126,7	100%	99%	Usher syndrome, type 1F, 602083 Deafness, autosomal recessive 23, 609533 Usher syndrome, type 1D/F digenic, 601067
PDZD7	76,5	97%	91%	{Retinal disease in Usher syndrome type IIA, modifier of}, 276901 Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472

PKD1	13,7	20%	18%	Polycystic kidney disease, adult type I, 173900
PKD2	97,1	95%	89%	Polycystic kidney disease 2, 613095
PKHD1	97	99%	96%	Polycystic kidney and hepatic disease, 263200
POC1A	94,7	97%	94%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
PTPRQ	113,8	100%	99%	Deafness, autosomal recessive 84A, 613391
RP1	169,5	100%	100%	Retinitis pigmentosa 1, 180100 {Hypertriglyceridemia, susceptibility to}, 145750
RP2	64,5	100%	99%	Retinitis pigmentosa 2, 312600
RPGR	45,8	83%	78%	Retinitis pigmentosa 3, 300029 Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 Macular degeneration, X-linked atrophic, 300834 Cone-rod dystrophy, X-linked, 1, 304020
RPGRIP1	114,3	100%	97%	Leber congenital amaurosis 6, 613826 Cone-rod dystrophy 13, 608194
RPGRIP1L	99,7	96%	96%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 COACH syndrome, 216360
RSPH1	109,1	100%	99%	Ciliary dyskinesia, primary, 24, 615481
RSPH4A	134,6	100%	100%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	74,6	100%	95%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	123,5	100%	98%	No OMIM phenotype Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36)
SDCCAG8	101,3	100%	99%	Senior-Loken syndrome 7, 613615
SPAG1	115,8	100%	98%	No OMIM phenotype Primary ciliary dyskinesia (Knowles (2013) Am J Hum Genet 93, 711)
SPATA7	133,7	100%	99%	Leber congenital amaurosis 3, 604232 Retinitis pigmentosa, juvenile, autosomal recessive, 604232

TCTN2	85,8	100%	97%	Meckel syndrome 8, 613885
TCTN3	101,9	100%	99%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TMEM138	92,6	100%	100%	Joubert syndrome 16, 614465
TMEM216	66	100%	86%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM231	64,4	97%	89%	Joubert syndrome 20, 614970 Meckel syndrome, type 11, 615397 -3
TMEM237	87,6	100%	92%	Joubert syndrome 14, 614424
TMEM67	115,2	100%	99%	Meckel syndrome 3, 607361 Joubert syndrome 6, 610688 {Bardet-Biedl syndrome 14, modifier of}, 209900 COACH syndrome, 216360 Nephronophthisis 11, 613550
TOPORS	149,7	100%	100%	Retinitis pigmentosa 31, 609923
TRIM32	106,2	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 Bardet-Biedl syndrome 11, 209900
TTBK2	117	100%	99%	Spinocerebellar ataxia 11, 604432
TTC21B	108,1	98%	98%	Nephronophthisis 12, 613820 Asphyxiating thoracic dystrophy 4, 613819
TTC8	100,5	100%	100%	Bardet-Biedl syndrome 8, 209900 Retinitis pigmentosa 51, 613464
TULP1	87,4	97%	91%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
USH1C	73,2	98%	93%	Acadian and Samaritan variety Usher syndrome, type 1C, 276904 Deafness, autosomal recessive 18A, 602092
USH1G	94,2	93%	86%	Usher syndrome, type 1G, 606943
USH2A	112,8	100%	99%	Usher syndrome, type 2A, 276901 Retinitis pigmentosa 39, 613809 -3

VHL	115,6	100%	100%	von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic Erythrocytosis, familial, 2, 263400
WDR19	122,7	100%	100%	Asphyxiating thoracic dystrophy 5, 614376 Nephronophthisis 13, 614377 Cranioectodermal dysplasia 4, 614378
WDR34	87,2	100%	97%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	112,7	100%	97%	Cranioectodermal dysplasia 2, 613610 Short rib-polydactyly syndrome, type V, 614091
WDR60	106,8	99%	98%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
XPNPEP3	116,2	100%	97%	Nephronophthisis-like nephropathy 1, 613159
ZMYND10	88	100%	97%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	123,1	100%	99%	Nephronophthisis 14, 614844 Joubert syndrome 19, 614844

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
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
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors