CILIOPATHIES GENE PANEL DG 2.18 (163 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ACVR2B	98,30%	95,00%	100%	100%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS9	99,50%	98,70%	100%	100%	No OMIM disease ID
AHI1	99,70%	97,90%	100%	100%	Joubert syndrome 3, 608629
ALMS1	99,80%	99,50%	100%	100%	Alstrom syndrome, 203800
ANKS6	93,80%	89,50%	97,90%	95,80%	Nephronophthisis 16, 615382
ARL13B	100%	99,20%	100%	100%	Joubert syndrome 8, 612291
ARL6	99,90%	98,60%	100%	100%	?Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARMC4	95,60%	93,50%	100%	100%	Ciliary dyskinesia, primary, 23, 615451
ARMC9	100%	99,80%	100%	100%	Joubert syndrome 30, 617622
B9D1	92,20%	92,00%	100%	100%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100%	100%	100%	100%	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BBIP1	98,60%	92,40%	100%	100%	?Bardet-Biedl syndrome 18, 615995
BBS1	100%	100%	100%	100%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	99,80%	100%	100%	Bardet-Biedl syndrome 10, 615987
BBS12	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	100%	99,50%	100%	100%	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,90%	99,30%	100%	100%	Bardet-Biedl syndrome 4, 615982
BBS5	99,00%	93,90%	100%	100%	Bardet-Biedl syndrome 5, 615983
BBS7	98,70%	95,50%	100%	100%	Bardet-Biedl syndrome 7, 615984
BBS9	99,70%	97,60%	100%	100%	Bardet-Biedl syndrome 9, 615986
C11orf70	99,30%	95,90%	100%	100%	Ciliary dyskinesia, primary, 38, 618063
C21orf2	100%	99,30%	100%	100%	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
C21orf59	100%	99,70%	100%	100%	Ciliary dyskinesia, primary, 26, 615500
C2CD3	95,80%	95,60%	95,90%	95,90%	Orofaciodigital syndrome XIV, 615948

C5orf42	99,70%	98,40%	100%	100%	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
C8orf37	100%	99,40%	100%	100%	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
CC2D2A	99,70%	97,70%	98,20%	98,20%	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome, 216360
CCDC103	100%	100%	100%	100%	Ciliary dyskinesia, primary, 17, 614679
CCDC114	100%	100%	100%	100%	Ciliary dyskinesia, primary, 20, 615067
CCDC151	100%	99,70%	100%	100%	Ciliary dyskinesia, primary, 30, 616037
CCDC28B	100%	99,70%	100%	100%	No OMIM disease ID
CCDC39	99,50%	96,50%	100%	100%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	99,10%	98,10%	100%	100%	Ciliary dyskinesia, primary, 15, 613808
CCDC65	99,60%	97,10%	100%	100%	Ciliary dyskinesia, primary, 27, 615504
CCNO	100%	99,20%	100%	100%	Ciliary dyskinesia, primary, 29, 615872
CENPF	99,80%	98,50%	100%	100%	Stromme syndrome, 243605
CEP104	100%	99,20%	100%	100%	Joubert syndrome 25, 616781
CEP120	100%	99,50%	100%	100%	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	99,90%	98,30%	100%	100%	Nephronophthisis 15, 614845
CEP290	96,10%	90,00%	100%	100%	?Bardet-Biedl syndrome 14, 615991 Leber congenital amaurosis 10, 611755 Senior-Loken syndrome 6, 610189 Meckel syndrome 4, 611134 Joubert syndrome 5, 610188
CEP41	99,80%	97,40%	100%	100%	Joubert syndrome 15, 614464
CEP55	100%	99,80%	100%	100%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	99,80%	97,40%	100%	100%	Nephronophthisis 18, 615862
CFAP44	99,80%	98,90%	100%	100%	?Spermatogenic failure 20, 617593
CFAP53	99,60%	97,40%	100%	100%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP69	98,70%	93,50%	100%	100%	Spermatogenic failure 24, 617959
CFC1	84,20%	74,10%	100%	100%	Heterotaxy, visceral, 2, autosomal, 605376
CSPP1	99,80%	98,70%	100%	100%	Joubert syndrome 21, 615636
DCDC2	100%	99,90%	100%	100%	Sclerosing cholangitis, neonatal, 617394 Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212
DDX59	100%	100%	100%	100%	Orofaciodigital syndrome V, 174300

DNAAF1	100%	99,80%	100%	100%	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	99,90%	98,90%	100%	100%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	99,50%	96,10%	100%	100%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	99,80%	97,00%	100%	100%	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	84,60%	78,60%	99,10%	97,50%	Ciliary dyskinesia, primary, 18, 614874
DNAH1	100%	99,70%	100%	100%	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	99,90%	99,00%	100%	100%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	100%	99,60%	100%	99,90%	Spermatogenic failure 39, 618643
DNAH5	99,90%	99,30%	100%	100%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAI1	100%	100%	100%	100%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	98,60%	96,20%	100%	100%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100%	100%	100%	100%	Ciliary dyskinesia, primary, 34, 617091
DNAL1	99,00%	96,80%	100%	100%	Ciliary dyskinesia, primary, 16, 614017
DRC1	100%	99,50%	100%	100%	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	98,80%	95,50%	100%	100%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	99,70%	97,60%	100%	100%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EVC	93,90%	88,60%	96,90%	94,80%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	97,70%	96,10%	100%	100%	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EXOC8	100%	100%	100%	100%	No OMIM disease ID
EXTL3	100%	100%	100%	100%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FUZ	100%	100%	100%	100%	No OMIM disease ID
GAS8	99,90%	99,30%	100%	100%	Ciliary dyskinesia, primary, 33, 616726
GDF1	73,90%	54,00%	98,70%	92,00%	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GLIS2	100%	99,80%	100%	100%	Nephronophthisis 7, 611498
HYDIN	99,90%	99,30%	100%	100%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100%	100%	100%	100%	Hydrolethalus syndrome, 236680
IFT122	100%	99,60%	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT140	99,80%	98,80%	100%	100%	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,90%	99,10%	100%	100%	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100%	100%	100%	100%	?Bardet-Biedl syndrome 19, 615996

IFT43	100%	100%	100%	100%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT52	100%	99,90%	100%	100%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	97,60%	88,20%	100%	100%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	93,50%	90,10%	95,00%	94,90%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
INPP5E	97,10%	92,70%	100%	100%	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INTU	99,70%	98,10%	100%	100%	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	100%	100%	100%	100%	Nephronophthisis 2, infantile, 602088
IQCB1	93,90%	85,00%	100%	100%	Senior-Loken syndrome 5, 609254
KCTD3	100%	99,70%	100%	100%	No OMIM disease ID
KIAA0556	100%	99,90%	100%	100%	Joubert syndrome 26, 616784
KIAA0586	97,30%	93,10%	95,80%	95,80%	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	100%	99,30%	100%	100%	?Orofaciodigital syndrome XV, 617127
KIF14	99,60%	97,70%	100%	100%	?Meckel syndrome 12, 616258 Microcephaly 20, primary, autosomal recessive, 617914
KIF7	93,60%	90,60%	99,10%	97,80%	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
LBR	99,40%	94,50%	100%	100%	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	99,90%	99,20%	100%	100%	Leber congenital amaurosis 5, 604537
LRRC56	100%	99,00%	100%	100%	Ciliary dyskinesia, primary, 39, 618254
LRRC6	99,20%	96,30%	100%	100%	Ciliary dyskinesia, primary, 19, 614935
LZTFL1	99,90%	99,20%	100%	100%	Bardet-Biedl syndrome 17, 615994
MAPKBP1	100%	100%	100%	100%	Nephronophthisis 20, 617271
MKKS	83,20%	83,20%	90,70%	90,70%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	99,80%	97,90%	100%	100%	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MMP21	99,90%	98,80%	100%	100%	Heterotaxy, visceral, 7, autosomal, 616749

NCAPG2	99,90%	99,20%	100%	100%	Khan-Khan-Katsanis syndrome, 618460
NEK1	99,80%	98,00%	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	100%	99,90%	100%	100%	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NME5	100%	100%	100%	100%	No OMIM disease ID
NME8	99,20%	95,30%	100%	100%	Ciliary dyskinesia, primary, 6, 610852
NODAL	100%	100%	100%	100%	Heterotaxy, visceral, 5, 270100
NPHP1	100%	99,00%	100%	100%	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	99,70%	98,40%	100%	100%	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	100%	99,80%	100%	100%	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
OCRL	99,90%	98,60%	100%	99,90%	Lowe syndrome, 309000 Dent disease 2, 300555
OFD1	88,00%	73,70%	100%	99,90%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
PDE6D	100%	100%	100%	100%	?Joubert syndrome 22, 615665
PIBF1	99,50%	96,20%	100%	100%	Joubert syndrome 33, 617767
PIH1D3	99,10%	92,50%	100%	100%	Ciliary dyskinesia, primary, 36, X-linked, 300991
PIK3C2A	99,20%	96,90%	100%	100%	Oculoskeletodental syndrome, 618440
PKD1	39,20%	30,00%	99,20%	98,90%	Polycystic kidney disease 1, 173900
PKD2	95,50%	91,10%	99,30%	97,70%	Polycystic kidney disease 2, 613095
PKHD1	100%	99,60%	100%	100%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PMFBP1	99,90%	99,30%	100%	100%	Spermatogenic failure 31, 618112
POC1A	100%	100%	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RPGRIP1L	96,70%	95,70%	100%	99,50%	COACH syndrome, 216360 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RSPH1	100%	100%	100%	100%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	99,90%	98,80%	100%	100%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	98,10%	95,60%	100%	100%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	99,90%	97,90%	100%	100%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	96,00%	90,90%	95,10%	95,10%	No OMIM disease ID

SDCCAG8	100%	99,90%	100%	100%	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SPAG1	99,30%	95,80%	99,90%	98,60%	Ciliary dyskinesia, primary, 28, 615505
SPATA7	99,80%	98,20%	100%	100%	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
TBC1D32	99,00%	95,80%	100%	100%	No OMIM disease ID
TCTEX1D2	100%	100%	100%	100%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
TCTN1	96,70%	93,00%	94,70%	94,70%	Joubert syndrome 13, 614173
TCTN2	100%	99,50%	100%	100%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TMEM107	100%	100%	100%	100%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM138	100%	99,10%	100%	100%	Joubert syndrome 16, 614465
TMEM216	99,90%	98,10%	100%	100%	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM231	100%	99,60%	100%	100%	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	100%	99,90%	100%	100%	Joubert syndrome 14, 614424
TMEM260	97,50%	93,40%	100%	100%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	99,50%	95,00%	100%	99,90%	Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 COACH syndrome, 216360 Joubert syndrome 6, 610688
TRAF3IP1	99,60%	97,60%	100%	100%	Senior-Loken syndrome 9, 616629
TRIM32	100%	100%	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TTBK2	99,80%	97,60%	100%	100%	Spinocerebellar ataxia 11, 604432
TTC21B	99,90%	99,30%	100%	100%	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC25	100%	100%	100%	100%	Ciliary dyskinesia, primary, 35, 617092
TTC26	99,90%	98,80%	100%	100%	No OMIM disease ID
TTC8	99,60%	98,10%	100%	100%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464

TULP1	100%	99,50%	100%	100%	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
VHL	96,30%	91,40%	100%	100%	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
WDPCP	98,20%	94,40%	98,10%	98,10%	?Bardet-Biedl syndrome 15, 615992 ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	100%	99,40%	100%	100%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR34	100%	99,60%	100%	100%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
WDR35	99,80%	98,90%	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR60	99,50%	97,00%	100%	100%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR66	100%	100%	100%	100%	Spermatogenic failure 33, 618152
XPNPEP3	100%	100%	100%	100%	Nephronophthisis-like nephropathy 1, 613159
ZIC3	100%	99,90%	100%	100%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMYND10	100%	100%	100%	100%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	100%	100%	100%	100%	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors