CILIOPATHIES GENE PANEL DG 3.2.0 (172 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ACVR2B	98,6	95,1	100	100	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS9	99,4	98,4	100	100	No OMIM disease ID
AHI1	99,4	97,4	100	100	Joubert syndrome 3, 608629
ALMS1	99,7	99,5	100	100	Alstrom syndrome, 203800
ANKS6	94,2	89,7	97	95	Nephronophthisis 16, 615382
ARL13B	100	99,3	100	100	Joubert syndrome 8, 612291
ARL3	99,7	96,5	100	100	Retinitis pigmentosa 83, 618173 Joubert syndrome 35, 618161
ARL6	99,1	98,4	100	100	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARMC9	99,9	99,4	100	100	Joubert syndrome 30, 617622
B9D1	85,2	85,2	95,8	94	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100	100	100	100	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BBIP1	95,7	87,4	100	100	?Bardet-Biedl syndrome 18, 615995
BBS1	100	100	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	100	99,9	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	100	100	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	99,4	98	100	100	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981
BBS4	99,9	98,9	100	99,9	Bardet-Biedl syndrome 4, 615982
BBS5	98,4	94,7	100	100	Bardet-Biedl syndrome 5, 615983
BBS7	99	96,5	100	99,9	Bardet-Biedl syndrome 7, 615984
BBS9	92	89	95,8	95,8	Bardet-Biedl syndrome 9, 615986

C2CD3	95,8	95,4	95,9	95,9	Orofaciodigital syndrome XIV, 615948
CBY1	82,2	82	100	100	No OMIM disease ID
CC2D2A	98,3	96,6	97,1	97	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCDC103	100	100	100	100	Ciliary dyskinesia, primary, 17, 614679
CCDC28B	99,9	98,2	100	100	No OMIM disease ID
CCDC39	99	96,2	100	99,9	Ciliary dyskinesia, primary, 14, 613807
CCDC40	99,1	98,2	100	100	Ciliary dyskinesia, primary, 15, 613808
CCDC65	97	92,5	100	100	Ciliary dyskinesia, primary, 27, 615504
CCNO	100	99	100	100	Ciliary dyskinesia, primary, 29, 615872
CENPF	99,4	96,9	100	100	Stromme syndrome, 243605
CEP104	99,9	98	100	100	Joubert syndrome 25, 616781
CEP120	99,9	99,6	100	100	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP164	99,8	98,2	100	100	Nephronophthisis 15, 614845
CEP290	96,2	90,8	100	99,9	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134
CEP41	98,8	93,4	100	100	Joubert syndrome 15, 614464
CEP55	100	99,8	100	100	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	99	96,6	100	99,9	Nephronophthisis 18, 615862
WDR66	100	99,9	100	100	Spermatogenic failure 33, 618152
CFAP298	99,6	96,7	100	100	Ciliary dyskinesia, primary, 26, 615500
CFAP300	99,2	98,3	100	99,9	Ciliary dyskinesia, primary, 38, 618063
CFAP410	100	99,6	100	100	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C8orf37	99,7	99,6	100	100	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 Bardet-Biedl syndrome 21, 617406

CFAP44	99,5	98,6	100	100	?Spermatogenic failure 20, 617593
CFAP53	99,3	96,6	100	100	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP69	98,7	94,9	100	100	Spermatogenic failure 24, 617959
CFC1	85	78	100	100	Heterotaxy, visceral, 2, autosomal, 605376
CPLANE1	99,4	98,2	100	100	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CSPP1	99,7	98,1	100	100	Joubert syndrome 21, 615636
DCDC2	100	99,9	100	100	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394
DDX59	100	99,8	100	100	Orofaciodigital syndrome V, 174300
DNAAF1	100	99,4	100	100	Ciliary dyskinesia, primary, 13, 613193
LRRC6	99,3	96,9	100	100	Ciliary dyskinesia, primary, 19, 614935
DNAAF2	99,7	98,4	100	100	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	99,5	96,3	100	100	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	99,4	94,7	100	99,7	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	85,6	78,8	98,8	96,9	Ciliary dyskinesia, primary, 18, 614874
PIH1D3	97,9	86	99,9	99,6	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	99,9	99,6	100	100	Spermatogenic failure 18, 617576 ?Ciliary dyskinesia, primary, 37, 617577
DNAH11	99,8	98,8	100	100	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	99,9	99,2	100	100	Spermatogenic failure 39, 618643
DNAH5	99,9	98,9	100	100	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH8	99,7	98,6	100	99,9	Spermatogenic failure 46, 619095
DNAI1	100	99,9	100	100	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	98,2	95,8	100	100	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100	99,6	100	100	Ciliary dyskinesia, primary, 34, 617091
DNAL1	99,6	98,3	100	99,2	Ciliary dyskinesia, primary, 16, 614017
DRC1	99,9	98,3	100	100	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	98,6	95,2	100	99,8	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091

WDR60	99,3	95,8	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR34	100	99,8	100	100	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	99,6	98,4	100	100	Short-rib thoracic dysplasia 15 with polydactyly, 617088
TCTEX1D2	100	99,8	100	100	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
EVC	94,2	91,4	97,5	95,1	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	98	96,2	100	100	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC8	100	100	100	100	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076
EXTL3	100	100	100	100	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FAM149B1	98,2	94,3	100	100	Joubert syndrome 36, 618763
FUZ	100	100	100	100	No OMIM disease ID
GAS8	99,9	99,6	100	100	Ciliary dyskinesia, primary, 33, 616726
GDF1	80,8	59	98,5	92	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GLIS2	100	99,9	100	100	Nephronophthisis 7, 611498
HYDIN	99,8	98,7	100	100	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
IFT122	99,9	99,2	100	100	Cranioectodermal dysplasia 1, 218330
IFT140	99,9	99,2	100	100	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	99,6	98,6	100	100	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 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 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	100	99,9	100	99,9	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	97,2	85,7	100	99,9	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92,9	89,6	94,9	94,6	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
INPP5E	96,9	93,2	100	100	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156

INTU	99,9	98,6	100	100	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100	99,8	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	92,8	82,8	100	100	Senior-Loken syndrome 5, 609254
KIAA0556	100	99,6	100	100	Joubert syndrome 26, 616784
KCTD3	99,7	99,4	100	100	No OMIM disease ID
KIAA0586	97,1	92	95,8	95,7	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIAA0753	99,9	98,9	100	100	?Orofaciodigital syndrome XV, 617127 ?Joubert syndrome 38, 619476 Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIF14	99,2	96,8	100	99,9	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF7	93,6	91,9	99,7	98,6	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
LBR	97,9	91	100	100	Pelger-Huet anomaly, 169400 Pelger-Huet anomaly with mild skeletal anomalies, 618019 ?Reynolds syndrome, 613471 Greenberg skeletal dysplasia, 215140
LCA5	99,6	97,9	100	100	Leber congenital amaurosis 5, 604537
LRRC56	99,8	99	100	100	Ciliary dyskinesia, primary, 39, 618254
LZTFL1	99,7	99,4	100	99,9	Bardet-Biedl syndrome 17, 615994
MAPKBP1	100	100	100	100	Nephronophthisis 20, 617271
MCIDAS	98,4	95,2	100	100	Ciliary dyskinesia, primary, 42, 618695
MKKS	100	100	100	100	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKS1	99,4	96,3	100	100	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MMP21	99,8	99,2	100	100	Heterotaxy, visceral, 7, autosomal, 616749
NCAPG2	99,8	99	100	100	Khan-Khan-Katsanis syndrome, 618460
NEK1	99,5	98,2	100	99,9	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520

NEK8	100	99,8	100	100	Renal-hepatic-pancreatic dysplasia 2, 615415 ?Nephronophthisis 9, 613824
NME5	99,7	99,7	100	100	No OMIM disease ID
NME8	98,9	94,2	100	100	Ciliary dyskinesia, primary, 6, 610852
NODAL	100	100	100	100	Heterotaxy, visceral, 5, 270100
NPHP1	99,8	99,1	100	100	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	99,6	98,5	100	99,9	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	100	99,8	100	100	Senior-Loken syndrome 4, 606996 Nephronophthisis 4, 606966
OCRL	99,4	97,6	100	99,9	Dent disease 2, 300555 Lowe syndrome, 309000
CCDC114	100	99,8	100	100	Ciliary dyskinesia, primary, 20, 615067
ARMC4	92,4	89,9	96,3	96,2	Ciliary dyskinesia, primary, 23, 615451
CCDC151	100	99,7	100	100	Ciliary dyskinesia, primary, 30, 616037
TTC25	100	99,7	100	100	Ciliary dyskinesia, primary, 35, 617092
OFD1	87,1	71,3	100	99,8	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
PDE6D	100	99,9	100	100	Joubert syndrome 22, 615665
PIBF1	99,1	95	100	99,9	Joubert syndrome 33, 617767
PIK3C2A	99	95,9	100	100	Oculoskeletodental syndrome, 618440
PKD1	40,6	32,8	99,3	99	Polycystic kidney disease 1, 173900
PKD2	96	93,3	99,6	97,9	Polycystic kidney disease 2, 613095
PKHD1	100	99,6	100	100	Polycystic kidney disease 4, with or without hepatic disease, 263200
PMFBP1	99,8	98,5	100	100	Spermatogenic failure 31, 618112
POC1A	100	100	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813

RPGRIP1L	96,5	95,3	100	99,4	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RSPH1	99,9	99,9	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	99,6	98,4	100	99,9	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	98,2	95,4	100	100	Ciliary dyskinesia, primary, 11, 612649
RSPH9	99,7	96,3	100	100	Ciliary dyskinesia, primary, 12, 612650
SCLT1	95,4	89,6	95,1	95	No OMIM disease ID
SDCCAG8	99,8	99,8	100	100	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993
SPAG1	98,7	93,9	99,6	97,9	Ciliary dyskinesia, primary, 28, 615505
SPATA7	99,6	98,2	100	100	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
STK36	100	99,1	100	100	?Ciliary dyskinesia, primary, 46, 619436
TBC1D32	98,7	96,4	100	99,9	No OMIM disease ID
TCTN1	96,8	92,8	94,7	94,7	Joubert syndrome 13, 614173
TCTN2	99,9	99,1	100	100	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100	100	100	100	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TMEM107	100	100	100	100	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM138	99,8	93,1	100	100	Joubert syndrome 16, 614465
TMEM216	98,5	92,8	100	100	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM218	100	99,8	100	100	No OMIM disease ID
TMEM231	100	99,3	100	100	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	99,8	99,3	100	100	Joubert syndrome 14, 614424
TMEM260	98,7	95,4	100	100	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	98,6	93,5	100	99,6	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361

					?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TOGARAM1	99,6	97,5	100	99,9	Joubert syndrome 37, 619185
TOPORS	100	100	100	100	Retinitis pigmentosa 31, 609923
TRAF3IP1	98,7	95,4	100	100	Senior-Loken syndrome 9, 616629
TRIM32	100	99,9	100	100	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TTBK2	99,9	98,3	100	100	Spinocerebellar ataxia 11, 604432
TTC21B	99,7	99,1	100	99,9	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 Nephronophthisis 12, 613820
TTC26	99,8	97,9	100	100	No OMIM disease ID
TTC8	99,5	98	100	100	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TULP1	99,8	98,2	100	100	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
VHL	95,5	90,6	100	100	Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic,
WDPCP	98	94,1	98,1	98	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	99,8	98,6	100	99,9	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Cranioectodermal dysplasia 4, 614378
WDR35	99,6	98,4	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
XPNPEP3	100	100	100	100	Nephronophthisis-like nephropathy 1, 613159
ZIC3	100	99,9	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.
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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-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors