CILIOPATHIES GENE PANEL DG 3.00 (170 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
ACVR2B	98,3	95	100	100	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS9	99,5	98,7	100	100	No OMIM disease ID
AHI1	99,7	97,9	100	100	Joubert syndrome 3, 608629
ALMS1	99,8	99,5	100	100	Alstrom syndrome, 203800
ANKS6	93,8	89,5	97,9	95,8	Nephronophthisis 16, 615382
ARL13B	100	99,2	100	100	Joubert syndrome 8, 612291
ARL3	100	98,4	100	100	Joubert syndrome 35, 618161 Retinitis pigmentosa 83, 618173
ARL6	99,9	98,6	100	100	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151 {Bardet-Biedl syndrome 1, modifier of}, 209900
ARMC9	100	99,8	100	100	Joubert syndrome 30, 617622
B9D1	85,2	85,1	94,2	93,9	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100	100	100	100	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BBIP1	98,6	92,4	100	100	?Bardet-Biedl syndrome 18, 615995
BBS1	100	100	100	100	Bardet-Biedl syndrome 1, 209900
BBS10	100	99,8	100	100	Bardet-Biedl syndrome 10, 615987
BBS12	100	100	100	100	Bardet-Biedl syndrome 12, 615989
BBS2	100	99,5	100	100	Bardet-Biedl syndrome 2, 615981 Retinitis pigmentosa 74, 616562
BBS4	99,9	99,3	100	100	Bardet-Biedl syndrome 4, 615982

BBS5	99	93,9	100	100	Bardet-Biedl syndrome 5, 615983
BBS7	98,7	95,5	100	100	Bardet-Biedl syndrome 7, 615984
BBS9	92,3	90,4	95,8	95,8	Bardet-Biedl syndrome 9, 615986
C2CD3	95,8	95,6	95,9	95,9	Orofaciodigital syndrome XIV, 615948
C8orf37	100	99,4	100	100	Retinitis pigmentosa 64, 614500 Bardet-Biedl syndrome 21, 617406 Cone-rod dystrophy 16, 614500
CC2D2A	98,5	96,5	97,1	97,1	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome 2, 619111
CCDC103	100	100	100	100	Ciliary dyskinesia, primary, 17, 614679
CCDC28B	100	99,7	100	100	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC39	99,5	96,5	100	100	Ciliary dyskinesia, primary, 14, 613807
CCDC40	99,1	98,1	100	100	Ciliary dyskinesia, primary, 15, 613808
CCDC65	99,6	97,1	100	100	Ciliary dyskinesia, primary, 27, 615504
CCNO	100	99,2	100	100	Ciliary dyskinesia, primary, 29, 615872
CENPF	99,8	98,5	100	100	Stromme syndrome, 243605
CEP104	100	99,2	100	100	Joubert syndrome 25, 616781
CEP120	100	99,5	100	100	Joubert syndrome 31, 617761 Short-rib thoracic dysplasia 13 with or without polydactyly, 616300
CEP164	99,9	98,3	100	100	Nephronophthisis 15, 614845
CEP290	96,1	90	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	99,8	97,4	100	100	Joubert syndrome 15, 614464
CEP55	100	99,8	100	100	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	99,8	97,4	100	100	Nephronophthisis 18, 615862

WDR66	100	100	100	100	Spermatogenic failure 33, 618152
CFAP298	100	99,7	100	100	Ciliary dyskinesia, primary, 26, 615500
CFAP300	99,3	95,9	100	100	Ciliary dyskinesia, primary, 38, 618063
CFAP410	100	99,3	100	100	Spondylometaphyseal dysplasia, axial, 602271 Retinal dystrophy with macular staphyloma, 617547
CFAP44	99,8	98,9	100	100	?Spermatogenic failure 20, 617593
CFAP53	99,6	97,4	100	100	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP69	98,7	93,5	100	100	Spermatogenic failure 24, 617959
CFC1	84,2	74,1	100	100	Heterotaxy, visceral, 2, autosomal, 605376
CPLANE1	99,7	98,4	100	100	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CSPP1	99,8	98,7	100	100	Joubert syndrome 21, 615636
DCDC2	100	99,9	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	99,8	100	100	Ciliary dyskinesia, primary, 13, 613193
DNAAF2	99,9	98,9	100	100	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	99,5	96,1	100	100	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	99,8	97	100	100	{Dyslexia, susceptibility to, 1}, 127700 Ciliary dyskinesia, primary, 25, 615482
DNAAF5	84,6	78,6	99,1	97,5	Ciliary dyskinesia, primary, 18, 614874
PIH1D3	99,1	92,5	100	100	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100	99,7	100	100	?Ciliary dyskinesia, primary, 37, 617577 Spermatogenic failure 18, 617576
DNAH11	99,9	99	100	100	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	100	99,6	100	99,9	Spermatogenic failure 39, 618643
DNAH5	99,9	99,3	100	100	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644

DNAH8	99,9	99	100	100	Spermatogenic failure 46, 619095
DNAI1	100	100	100	100	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	98,6	96,2	100	100	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100	100	100	100	Ciliary dyskinesia, primary, 34, 617091
DNAL1	99	96,8	100	100	Ciliary dyskinesia, primary, 16, 614017
DRC1	100	99,5	100	100	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	98,8	95,5	100	100	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	99,5	97	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR34	100	99,6	100	100	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	99,7	97,6	100	100	Short-rib thoracic dysplasia 15 with polydactyly, 617088
TCTEX1D2	100	100	100	100	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
EVC	93,9	88,6	96,9	94,8	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	97,7	96,1	100	100	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
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	99,5	95,4	100	100	Joubert syndrome 36, 618763
FUZ	100	100	100	100	{Neural tube defects, susceptibility to}, 182940
GAS8	99,9	99,3	100	100	Ciliary dyskinesia, primary, 33, 616726
GDF1	73,9	54	98,7	92	Right atrial isomerism (Ivemark), 208530 Congenital heart defects, multiple types, 6, 613854
GLIS2	100	99,8	100	100	Nephronophthisis 7, 611498
HYDIN	99,9	99,3	100	100	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
IFT122	100	99,6	100	100	Cranioectodermal dysplasia 1, 218330

IFT140	99,8	98,8	100	100	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,9	99,1	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	99,9	100	100	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT80	97,6	88,2	100	100	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	93,5	90,1	95	94,9	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
INPP5E	97,1	92,7	100	100	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 Joubert syndrome 1, 213300
INTU	99,7	98,1	100	100	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
INVS	100	100	100	100	Nephronophthisis 2, infantile, 602088
IQCB1	93,9	85	100	100	Senior-Loken syndrome 5, 609254
KIAA0556	100	99,9	100	100	Joubert syndrome 26, 616784
KCTD3	100	99,7	100	100	No OMIM disease ID
KIAA0586	97,3	93,1	95,8	95,8	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIAA0753	100	99,3	100	100	?Orofaciodigital syndrome XV, 617127
KIF14	99,6	97,7	100	100	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258
KIF7	93,6	90,6	99,1	97,8	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
LBR	99,4	94,5	100	100	Pelger-Huet anomaly, 169400 Greenberg skeletal dysplasia, 215140 ?Reynolds syndrome, 613471 Pelger-Huet anomaly with mild skeletal anomalies, 618019
LCA5	99,9	99,2	100	100	Leber congenital amaurosis 5, 604537

LRRC56	100	99	100	100	Ciliary dyskinesia, primary, 39, 618254
LRRC6	99,2	96,3	100	100	Ciliary dyskinesia, primary, 19, 614935
LZTFL1	99,9	99,2	100	100	Bardet-Biedl syndrome 17, 615994
MAPKBP1	100	100	100	100	Nephronophthisis 20, 617271
MCIDAS	99,3	96,2	100	100	Ciliary dyskinesia, primary, 42, 618695
MKKS	100	100	100	100	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKS1	99,8	97,9	100	100	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MMP21	99,9	98,8	100	100	Heterotaxy, visceral, 7, autosomal, 616749
NCAPG2	99,9	99,2	100	100	Khan-Khan-Katsanis syndrome, 618460
NEK1	99,8	98	100	100	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	100	99,9	100	100	?Nephronophthisis 9, 613824 Renal-hepatic-pancreatic dysplasia 2, 615415
NME5	100	100	100	100	No OMIM disease ID
NME8	99,2	95,3	100	100	Ciliary dyskinesia, primary, 6, 610852
NODAL	100	100	100	100	Heterotaxy, visceral, 5, 270100
NPHP1	100	99	100	100	Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900 Joubert syndrome 4, 609583
NPHP3	99,7	98,4	100	100	Meckel syndrome 7, 267010 Renal-hepatic-pancreatic dysplasia 1, 208540 Nephronophthisis 3, 604387
NPHP4	100	99,8	100	100	Nephronophthisis 4, 606966 Senior-Loken syndrome 4, 606996
OCRL	99,9	98,6	100	99,9	Lowe syndrome, 309000 Dent disease 2, 300555
CCDC114	100	100	100	100	Ciliary dyskinesia, primary, 20, 615067
ARMC4	92,1	90	96,3	96,3	Ciliary dyskinesia, primary, 23, 615451

CCDC151	100	99,7	100	100	Ciliary dyskinesia, primary, 30, 616037
TTC25	100	100	100	100	Ciliary dyskinesia, primary, 35, 617092
OFD1	88	73,7	100	99,9	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	99,5	96,2	100	100	Joubert syndrome 33, 617767
PIK3C2A	99,2	96,9	100	100	Oculoskeletodental syndrome, 618440
PKD1	39,2	30	99,2	98,9	Polycystic kidney disease 1, 173900
PKD2	95,5	91,1	99,3	97,7	Polycystic kidney disease 2, 613095
PKHD1	100	99,6	100	100	Polycystic kidney disease 4, with or without hepatic disease, 263200
PMFBP1	99,9	99,3	100	100	Spermatogenic failure 31, 618112
POC1A	100	100	100	100	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RPGRIP1L	96,7	95,7	100	99,5	?COACH syndrome 3, 619113 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RSPH1	100	100	100	100	Ciliary dyskinesia, primary, 24, 615481
RSPH3	99,9	98,8	100	100	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	98,1	95,6	100	100	Ciliary dyskinesia, primary, 11, 612649
RSPH9	99,9	97,9	100	100	Ciliary dyskinesia, primary, 12, 612650
SCLT1	96	90,9	95,1	95,1	No OMIM disease ID
SDCCAG8	100	99,9	100	100	Bardet-Biedl syndrome 16, 615993 Senior-Loken syndrome 7, 613615
SPAG1	99,3	95,8	99,9	98,6	Ciliary dyskinesia, primary, 28, 615505
SPATA7	99,8	98,2	100	100	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
STK36	100	99,1	100	100	No OMIM disease ID
TBC1D32	99	95,8	100	100	No OMIM disease ID

TCTN1	96,7	93	94,7	94,7	Joubert syndrome 13, 614173
TCTN2	100	99,5	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	99,1	100	100	Joubert syndrome 16, 614465
TMEM216	99,9	98,1	100	100	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
TMEM218	100	99,9	100	100	No OMIM disease ID
TMEM231	100	99,6	100	100	Meckel syndrome 11, 615397 Joubert syndrome 20, 614970
TMEM237	100	99,9	100	100	Joubert syndrome 14, 614424
TMEM260	97,5	93,4	100	100	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	99,5	95	100	99,9	Meckel syndrome 3, 607361 COACH syndrome 1, 216360 ?RHYNS syndrome, 602152 Nephronophthisis 11, 613550 {Bardet-Biedl syndrome 14, modifier of}, 615991 Joubert syndrome 6, 610688
TOGARAM1	99,6	98,1	100	100	No OMIM disease ID
TRAF3IP1	99,6	97,6	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	99,8	97,6	100	100	Spinocerebellar ataxia 11, 604432
TTC21B	99,9	99,3	100	100	Nephronophthisis 12, 613820 Short-rib thoracic dysplasia 4 with or without polydactyly, 613819
TTC26	99,9	98,8	100	100	No OMIM disease ID
TTC8	99,6	98,1	100	100	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464

TULP1	100	99,5	100	100	Retinitis pigmentosa 14, 600132 Leber congenital amaurosis 15, 613843
VHL	96,3	91,4	100	100	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
WDPCP	98,2	94,4	98,1	98,1	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	100	99,4	100	100	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	99,8	98,9	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.
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 : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors