CILIOPATHIES GENE PANEL DG 3.4.0 (174 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACVR2B	100,0%	100,0%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS9	100,0%	100,0%	No OMIM Disease ID
AHI1	100,0%	100,0%	Joubert syndrome 3, 608629
ALMS1	100,0%	100,0%	Alstrom syndrome, 203800
ANKS6	100,0%	99,9%	Nephronophthisis 16, 615382
ARL13B	100,0%	100,0%	Joubert syndrome 8, 612291
ARL3	100,0%	100,0%	Retinitis pigmentosa 83, 618173 Joubert syndrome 35, 618161
ARL6	100,0%	100,0%	Retinitis pigmentosa 55, 613575 Bardet-Biedl syndrome 3, 600151
ARMC9	100,0%	100,0%	Joubert syndrome 30, 617622
B9D1	96,6%	94,1%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
B9D2	100,0%	100,0%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BBIP1	100,0%	100,0%	?Bardet-Biedl syndrome 18, 615995
BBS1	100,0%	100,0%	Bardet-Biedl syndrome 1, 209900
BBS10	100,0%	100,0%	Bardet-Biedl syndrome 10, 615987
BBS12	100,0%	100,0%	Bardet-Biedl syndrome 12, 615989
BBS2	100,0%	100,0%	Retinitis pigmentosa 74, 616562 Bardet-Biedl syndrome 2, 615981
BBS4	100,0%	100,0%	Bardet-Biedl syndrome 4, 615982
BBS5	100,0%	100,0%	Bardet-Biedl syndrome 5, 615983
BBS7	100,0%	100,0%	Bardet-Biedl syndrome 7, 615984
BBS9	95,8%	95,8%	Bardet-Biedl syndrome 9, 615986
C2CD3	95,9%	95,9%	Orofaciodigital syndrome XIV, 615948
CBY1	100,0%	100,0%	No OMIM Disease ID

CC2D2A	97,1%	97,1%	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CCDC103	100,0%	100,0%	Ciliary dyskinesia, primary, 17, 614679
CCDC28B	100,0%	100,0%	No OMIM Disease ID
CCDC39	100,0%	100,0%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100,0%	100,0%	Ciliary dyskinesia, primary, 15, 613808
CCDC65	100,0%	100,0%	Ciliary dyskinesia, primary, 27, 615504
CCNO	100,0%	100,0%	Ciliary dyskinesia, primary, 29, 615872
CENPF	100,0%	100,0%	Stromme syndrome, 243605
CEP104	100,0%	100,0%	Joubert syndrome 25, 616781
CEP120	100,0%	100,0%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 Joubert syndrome 31, 617761
CEP164	100,0%	100,0%	Nephronophthisis 15, 614845
CEP290	100,0%	100,0%	Leber congenital amaurosis 10, 611755 Joubert syndrome 5, 610188 Senior-Loken syndrome 6, 610189 ?Bardet-Biedl syndrome 14, 615991 Meckel syndrome 4, 611134
CEP41	100,0%	100,0%	Joubert syndrome 15, 614464
CEP55	100,0%	100,0%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	100,0%	100,0%	Nephronophthisis 18, 615862
WDR66	100,0%	100,0%	Spermatogenic failure 33, 618152
CFAP298	100,0%	100,0%	Ciliary dyskinesia, primary, 26, 615500
CFAP300	100,0%	100,0%	Ciliary dyskinesia, primary, 38, 618063
CFAP410	100,0%	100,0%	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
C8orf37	100,0%	100,0%	Retinitis pigmentosa 64, 614500 Cone-rod dystrophy 16, 614500 Bardet-Biedl syndrome 21, 617406
CFAP44	100,0%	100,0%	?Spermatogenic failure 20, 617593
CFAP53	100,0%	100,0%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP69	100,0%	100,0%	Spermatogenic failure 24, 617959
CFC1	100,0%	100,0%	Heterotaxy, visceral, 2, autosomal, 605376
CPLANE1	100,0%	100,0%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CSPP1	100,0%	100,0%	Joubert syndrome 21, 615636

DCDC2	100,0%	100,0%	Nephronophthisis 19, 616217 ?Deafness, autosomal recessive 66, 610212 Sclerosing cholangitis, neonatal, 617394
DDX59	100,0%	100,0%	Orofaciodigital syndrome V, 174300
DNAAF1	100,0%	100,0%	Ciliary dyskinesia, primary, 13, 613193
LRRC6	100,0%	100,0%	Ciliary dyskinesia, primary, 19, 614935
DNAAF2	100,0%	100,0%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	100,0%	100,0%	Ciliary dyskinesia, primary, 2, 606763
DNAAF4	100,0%	100,0%	Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100,0%	100,0%	Ciliary dyskinesia, primary, 18, 614874
PIH1D3	100,0%	100,0%	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100,0%	100,0%	Spermatogenic failure 18, 617576 ?Ciliary dyskinesia, primary, 37, 617577
DNAH11	100,0%	100,0%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	100,0%	100,0%	Spermatogenic failure 39, 618643
DNAH5	100,0%	100,0%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH8	100,0%	100,0%	Spermatogenic failure 46, 619095
DNAI1	100,0%	100,0%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100,0%	100,0%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100,0%	100,0%	Ciliary dyskinesia, primary, 34, 617091
DNAL1	100,0%	100,0%	Ciliary dyskinesia, primary, 16, 614017
DRC1	100,0%	100,0%	Ciliary dyskinesia, primary, 21, 615294
DYNC2H1	100,0%	100,0%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	100,0%	100,0%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WDR34	100,0%	100,0%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	100,0%	100,0%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
TCTEX1D2	100,0%	100,0%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
EVC	100,0%	99,8%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	100,0%	100,0%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXOC8	100,0%	100,0%	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076
EXTL3	100,0%	100,0%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FAM149B1	100,0%	100,0%	Joubert syndrome 36, 618763
FOXF1	100,0%	100,0%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380
FUZ	100,0%	100,0%	No OMIM Disease ID
GAS8	100,0%	100,0%	Ciliary dyskinesia, primary, 33, 616726

GDF1	100,0%	100,0%	Congenital heart defects, multiple types, 6, 613854 Right atrial isomerism (Ivemark), 208530
GLIS2	100,0%	100,0%	Nephronophthisis 7, 611498
HYDIN	100,0%	100,0%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100,0%	100,0%	Hydrolethalus syndrome, 236680
IFT122	100,0%	100,0%	Cranioectodermal dysplasia 1, 218330
IFT140	100,0%	100,0%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	100,0%	100,0%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100,0%	100,0%	Bardet-Biedl syndrome 19, 615996
IFT43	100,0%	100,0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	100,0%	100,0%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT74	100,0%	100,0%	Spermatogenic failure 58, 619585 Joubert syndrome 40, 619582 ?Bardet-Biedl syndrome 22, 617119
IFT80	100,0%	100,0%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	95,0%	95,0%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
INPP5E	100,0%	100,0%	Joubert syndrome 1, 213300 Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156
INTU	100,0%	100,0%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100,0%	100,0%	Nephronophthisis 2, infantile, 602088
IQCB1	100,0%	100,0%	Senior-Loken syndrome 5, 609254
KIAA0556	100,0%	100,0%	Joubert syndrome 26, 616784
KCTD3	100,0%	100,0%	No OMIM Disease ID
KIAA0586	95,8%	95,8%	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIAA0753	100,0%	100,0%	?Orofaciodigital syndrome XV, 617127 ?Joubert syndrome 38, 619476 Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIF14	100,0%	100,0%	Microcephaly 20, primary, autosomal recessive, 617914 ?Meckel syndrome 12, 616258

KIF7	100,0%	100,0%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
LBR	100,0%	100,0%	Pelger-Huet anomaly, 169400 ?Reynolds syndrome, 613471 Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019 Greenberg skeletal dysplasia, 215140
LCA5	100,0%	100,0%	Leber congenital amaurosis 5, 604537
LRRC56	100,0%	100,0%	Ciliary dyskinesia, primary, 39, 618254
LZTFL1	100,0%	100,0%	Bardet-Biedl syndrome 17, 615994
MAPKBP1	100,0%	100,0%	Nephronophthisis 20, 617271
MCIDAS	100,0%	100,0%	Ciliary dyskinesia, primary, 42, 618695
MKKS	100,0%	100,0%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKS1	100,0%	100,0%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MMP21	100,0%	100,0%	Heterotaxy, visceral, 7, autosomal, 616749
NCAPG2	100,0%	100,0%	Khan-Khan-Katsanis syndrome, 618460
NEK1	100,0%	100,0%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NEK8	100,0%	100,0%	Renal-hepatic-pancreatic dysplasia 2, 615415 ?Nephronophthisis 9, 613824
NME5	100,0%	100,0%	No OMIM Disease ID
NME8	100,0%	100,0%	Ciliary dyskinesia, primary, 6, 610852
NODAL	100,0%	100,0%	Heterotaxy, visceral, 5, 270100
NPHP1	100,0%	100,0%	Joubert syndrome 4, 609583 Nephronophthisis 1, juvenile, 256100 Senior-Loken syndrome-1, 266900
NPHP3	100,0%	100,0%	Nephronophthisis 3, 604387 Renal-hepatic-pancreatic dysplasia 1, 208540 Meckel syndrome 7, 267010
NPHP4	100,0%	100,0%	Senior-Loken syndrome 4, 606996 Nephronophthisis 4, 606966
OCRL	100,0%	100,0%	Dent disease 2, 300555 Lowe syndrome, 309000
CCDC114	100,0%	100,0%	Ciliary dyskinesia, primary, 20, 615067

ARMC4	96,3%	96,3%	Ciliary dyskinesia, primary, 23, 615451
CCDC151	100,0%	100,0%	Ciliary dyskinesia, primary, 30, 616037
TTC25	100,0%	100,0%	Ciliary dyskinesia, primary, 35, 617092
OFD1	100,0%	100,0%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
PDE6D	100,0%	100,0%	Joubert syndrome 22, 615665
PIBF1	100,0%	100,0%	Joubert syndrome 33, 617767
PIK3C2A	100,0%	100,0%	Oculoskeletodental syndrome, 618440
PKD1	99,9%	99,7%	Polycystic kidney disease 1, 173900
PKD2	100,0%	100,0%	Polycystic kidney disease 2, 613095
PKHD1	100,0%	100,0%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PMFBP1	100,0%	100,0%	Spermatogenic failure 31, 618112
POC1A	100,0%	100,0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RPGRIP1L	100,0%	99,8%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RSPH1	100,0%	100,0%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	100,0%	100,0%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	100,0%	100,0%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	100,0%	100,0%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	95,1%	95,1%	No OMIM Disease ID
SDCCAG8	100,0%	100,0%	Senior-Loken syndrome 7, 613615 Bardet-Biedl syndrome 16, 615993
SPAG1	100,0%	100,0%	Ciliary dyskinesia, primary, 28, 615505
SPATA7	100,0%	100,0%	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 Leber congenital amaurosis 3, 604232
STK36	100,0%	100,0%	?Ciliary dyskinesia, primary, 46, 619436
TBC1D32	100,0%	100,0%	No OMIM Disease ID
TCTN1	95,5%	94,7%	Joubert syndrome 13, 614173
TCTN2	100,0%	100,0%	Joubert syndrome 24, 616654 ?Meckel syndrome 8, 613885
TCTN3	100,0%	100,0%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860

TMEM107	100,0%	100,0%	Orofaciodigital syndrome XVI, 617563 Meckel syndrome 13, 617562 ?Joubert syndrome 29, 617562
TMEM138	100,0%	100,0%	Joubert syndrome 16, 614465
TMEM216	100,0%	100,0%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TMEM218	100,0%	100,0%	Joubert syndrome 39, 619562
TMEM231	100,0%	100,0%	Joubert syndrome 20, 614970 Meckel syndrome 11, 615397
TMEM237	100,0%	100,0%	Joubert syndrome 14, 614424
TMEM260	100,0%	100,0%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	100,0%	100,0%	Nephronophthisis 11, 613550 Joubert syndrome 6, 610688 Meckel syndrome 3, 607361 ?RHYNS syndrome, 602152 COACH syndrome 1, 216360
TOGARAM1	100,0%	100,0%	Joubert syndrome 37, 619185
TOPORS	100,0%	100,0%	Retinitis pigmentosa 31, 609923
TRAF3IP1	100,0%	100,0%	Senior-Loken syndrome 9, 616629
TRIM32	100,0%	100,0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TTBK2	100,0%	100,0%	Spinocerebellar ataxia 11, 604432
TTC21B	100,0%	100,0%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 Nephronophthisis 12, 613820
TTC26	100,0%	100,0%	Biliary, renal, neurologic, and skeletal syndrome, 619534
TTC8	100,0%	100,0%	Bardet-Biedl syndrome 8, 615985 ?Retinitis pigmentosa 51, 613464
TULP1	100,0%	100,0%	Leber congenital amaurosis 15, 613843 Retinitis pigmentosa 14, 600132
VHL	100,0%	100,0%	Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic,
WDPCP	98,1%	98,1%	?Bardet-Biedl syndrome 15, 615992 Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	100,0%	100,0%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307

			?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Cranioectodermal dysplasia 4, 614378
WDR35	100,0%	100,0%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
XPNPEP3	100,0%	100,0%	Nephronophthisis-like nephropathy 1, 613159
ZIC3	100,0%	100,0%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMYND10	100,0%	100,0%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	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.
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TWIST is the chemistry used for 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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors