CILIOPATHIES PANEL DG-5.0.0 (185 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
ACVR2B	100%	100%	100%	99.9%	99.5%	Heterotaxy, visceral, 4, autosomal, 613751
ADAMTS9	100%	100%	100%	99.8%	99%	
AHI1	98.7%	98.7%	100%	99.9%	99.4%	Joubert syndrome 3, 608629
ALMS1	100%	100%	100%	100%	99.7%	Alstrom syndrome, 203800
ANKS6	100%	99.9%	100%	100%	99.7%	Nephronophthisis 16, 615382
ARL13B	93.4%	93.4%	100%	100%	99.7%	Joubert syndrome 8, 612291
ARL3	86.8%	86.8%	100%	100%	99.5%	Retinitis pigmentosa 83, 618173;Joubert syndrome 35, 618161
ARL6	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
ARMC9	97.2%	94.8%	100%	100%	99.6%	Joubert syndrome 30, 617622
B9D1	100%	100%	100%	100%	99.4%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120
B9D2	100%	100%	100%	100%	99.8%	?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175
BBIP1	100%	100%	100%	100%	99.7%	Bardet-Biedl syndrome 18, 615995
BBS1	93.7%	93.7%	100%	99.9%	99.1%	Bardet-Biedl syndrome 1, 209900
BBS10	100%	100%	100%	100%	99.6%	Bardet-Biedl syndrome 10, 615987

BBS12	100%	100%	100%	100%	100%	Bardet-Biedl syndrome 12, 615989
BBS2	98%	98%	100%	100%	99.5%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100%	100%	100%	100%	99.6%	Bardet-Biedl syndrome 4, 615982
BBS5	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 5, 615983
BBS7	100%	100%	100%	100%	99.8%	Bardet-Biedl syndrome 7, 615984
BBS9	91.9%	91.9%	100%	100%	99.7%	Bardet-Biedl syndrome 9, 615986
C2CD3	96%	96%	100%	100%	99.7%	Orofaciodigital syndrome XIV, 615948
CBY1	100%	100%	100%	100%	99.6%	
CC2D2A	98.2%	98.2%	100%	100%	99.9%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CCDC28B	100%	100%	100%	100%	98.4%	{Bardet-Biedl syndrome 1, modifier of}, 209900
CCDC39	100%	100%	100%	100%	99.9%	Ciliary dyskinesia, primary, 14, 613807
CCDC40	100%	100%	100%	99.9%	98.9%	Ciliary dyskinesia, primary, 15, 613808
CCDC65	100%	100%	100%	100%	99.9%	
CCNO	100%	100%	100%	99.8%	99.5%	Ciliary dyskinesia, primary, 29, 615872
CENPF	98.6%	98.6%	100%	100%	99.7%	Stromme syndrome, 243605
CEP104	95.3%	95.1%	100%	99.9%	99.3%	Joubert syndrome 25, 616781;Intellectual developmental disorder, autosomal recessive 77, 619988

CEP120	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761
CEP164	100%	100%	100%	100%	99.5%	Nephronophthisis 15, 614845
CEP290	100%	100%	100%	100%	99.8%	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%	100%	100%	100%	99.9%	Joubert syndrome 15, 614464
CEP55	100%	100%	100%	100%	99.8%	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500
CEP83	100%	100%	100%	100%	99.6%	Nephronophthisis 18, 615862
CFAP251	100%	100%	100%	100%	99.6%	Spermatogenic failure 33, 618152
CFAP298	92.4%	87.5%	100%	100%	99.5%	Ciliary dyskinesia, primary, 26, 615500
CFAP300	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 38, 618063
CFAP410	100%	100%	100%	100%	99.4%	Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271
CFAP418	100%	100%	100%	100%	99.9%	Retinitis pigmentosa 64, 614500;Cone-rod dystrophy 16, 614500;Bardet-Biedl syndrome 21, 617406

CFAP44	100%	100%	100%	100%	99.8%	Spermatogenic failure 20, 617593
CFAP45	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608
CFAP52	100%	100%	100%	100%	99.7%	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607
CFAP53	100%	100%	100%	100%	99.6%	Heterotaxy, visceral, 6, autosomal recessive, 614779
CFAP69	100%	100%	100%	100%	99.7%	Spermatogenic failure 24, 617959
CFC1	100%	100%	100%	100%	99.8%	Heterotaxy, visceral, 2, autosomal, 605376
CPLANE1	100%	100%	100%	100%	99.7%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CSPP1	96.9%	96.9%	100%	99.9%	99.4%	Joubert syndrome 21, 615636
DCDC2	100%	100%	100%	100%	99.8%	Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394
DDX59	100%	100%	100%	100%	99.7%	Orofaciodigital syndrome V, 174300
DNAAF1	96.2%	96.1%	100%	100%	99.7%	Ciliary dyskinesia, primary, 13, 613193
DNAAF11	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 19, 614935
DNAAF19	100%	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 17, 614679
DNAAF2	100%	100%	100%	99.9%	99.6%	Ciliary dyskinesia, primary, 10, 612518
DNAAF3	100%	100%	100%	100%	98.8%	Ciliary dyskinesia, primary, 2, 606763

DNAAF4	100%	100%	100%	100%	99.9%	{Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482
DNAAF5	100%	100%	100%	99.9%	98.7%	Ciliary dyskinesia, primary, 18, 614874
DNAAF6	100%	100%	99.3%	91.6%	74.8%	Ciliary dyskinesia, primary, 36, X-linked, 300991
DNAH1	100%	100%	100%	100%	99.3%	Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577
DNAH11	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884
DNAH17	100%	100%	100%	100%	99.3%	Spermatogenic failure 39, 618643
DNAH5	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644
DNAH8	100%	100%	100%	100%	99.7%	Spermatogenic failure 46, 619095
DNAH9	100%	100%	100%	100%	99.5%	Ciliary dyskinesia, primary, 40, 618300
DNAI1	95.7%	95.7%	100%	100%	99.5%	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400
DNAI2	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444
DNAJB13	100%	100%	100%	100%	99.2%	Ciliary dyskinesia, primary, 34, 617091
DNAL1	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 16, 614017
DRC1	100%	100%	100%	100%	99.6%	Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294

DYNC2H1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2I1	95.7%	95.7%	100%	100%	99.7%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
DYNC2I2	100%	100%	100%	100%	99.4%	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633
DYNC2LI1	83%	81.4%	100%	100%	99.8%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
DYNLT2B	58.6%	58.6%	100%	100%	99.2%	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405
EVC	100%	100%	100%	100%	99.4%	Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530
EVC2	100%	100%	100%	100%	99.5%	Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530
EXOC8	100%	100%	100%	100%	99.5%	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076
EXTL3	100%	100%	100%	100%	99.5%	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425
FAM149B1	100%	100%	100%	100%	99.8%	Joubert syndrome 36, 618763
FOXF1	100%	100%	100%	99.8%	96%	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380

FOXJ1	100%	100%	100%	100%	99%	Ciliary dyskinesia, primary, 43, 618699
FUZ	94%	93.7%	100%	100%	99.3%	{Neural tube defects, susceptibility to}, 182940
GAS8	100%	100%	100%	99.9%	99.6%	
GDF1	100%	100%	100%	100%	99.2%	Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530
GLIS2	100%	100%	100%	100%	99.2%	Nephronophthisis 7, 611498
HYDIN	100%	99.9%	100%	99.9%	99.4%	Ciliary dyskinesia, primary, 5, 608647
HYLS1	100%	100%	100%	100%	99.9%	Hydrolethalus syndrome, 236680
IFT122	100%	100%	100%	100%	99.7%	Cranioectodermal dysplasia 1, 218330
IFT140	100%	100%	100%	100%	99.5%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180
IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	100%	98.5%	100%	100%	99.8%	Bardet-Biedl syndrome 19, 615996

IFT43	93.5%	93.5%	100%	100%	99.6%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT52	92.9%	92.9%	100%	100%	99.8%	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102
IFT56	100%	100%	100%	100%	99.8%	Biliary, renal, neurologic, and skeletal syndrome, 619534
IFT74	100%	100%	100%	100%	99.8%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IFT80	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IFT81	92.5%	92.5%	100%	100%	99.7%	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895
INPP5E	100%	100%	100%	99.8%	98.6%	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300
INTU	100%	100%	100%	100%	99.8%	?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925
INVS	100%	100%	100%	100%	99.6%	Nephronophthisis 2, infantile, 602088
IQCB1	95.8%	95.8%	100%	100%	99.9%	Senior-Loken syndrome 5, 609254

KATNIP	100%	100%	100%	100%	99.5%	Joubert syndrome 26, 616784
KCTD3	100%	100%	100%	99.9%	99.7%	
KIAA0586	95.6%	95.6%	100%	100%	99.8%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490
KIAA0753	100%	100%	100%	100%	99.7%	?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479
KIF14	97.6%	97.6%	100%	100%	99.8%	Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258
KIF7	100%	100%	100%	100%	99.1%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131
LBR	100%	100%	100%	100%	99.5%	Pelger-Huet anomaly, 169400;?Reynolds syndrome, 613471;Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019;Greenberg skeletal dysplasia, 215140
LCA5	100%	100%	100%	100%	99.9%	Leber congenital amaurosis 5, 604537
LRRC56	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 39, 618254
LZTFL1	100%	100%	100%	100%	99.9%	Bardet-Biedl syndrome 17, 615994

MAPKBP1	100%	100%	100%	100%	99.6%	Nephronophthisis 20, 617271
MCIDAS	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 42, 618695
MKKS	100%	100%	100%	100%	99.9%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKS1	99.5%	99%	100%	100%	99.5%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MMP21	100%	100%	100%	100%	99.6%	Heterotaxy, visceral, 7, autosomal, 616749
MNS1	100%	100%	100%	100%	99.9%	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948
NCAPG2	100%	100%	100%	100%	99.7%	Khan-Khan-Katsanis syndrome, 618460
NEK1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NEK10	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 44, 618781
NEK8	100%	100%	100%	100%	99.5%	Renal-hepatic-pancreatic dysplasia 2, 615415;Polycystic kidney disease 8, 620903;?Nephronophthisis 9, 613824
NME5	86.7%	86.6%	100%	100%	100%	Ciliary dyskinesia, primary, 48, without situs inversus, 620032
NME8	100%	100%	100%	100%	99.9%	?Ciliary dyskinesia, primary, 6, 610852

NODAL	100%	100%	100%	100%	99.5%	Heterotaxy, visceral, 5, autosomal, 270100
NPHP1	100%	100%	100%	99.9%	99.3%	Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900
NPHP3	100%	100%	100%	100%	99.8%	Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010
NPHP4	100%	100%	100%	99.9%	99.3%	Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966
OCRL	100%	100%	99.1%	89.6%	70.8%	Dent disease 2, 300555;Lowe syndrome, 309000
ODAD1	100%	100%	100%	100%	99.3%	Ciliary dyskinesia, primary, 20, 615067
ODAD2	96.1%	96.1%	100%	100%	99.7%	Ciliary dyskinesia, primary, 23, 615451
ODAD3	100%	100%	100%	99.9%	98.6%	Ciliary dyskinesia, primary, 30, 616037
ODAD4	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 35, 617092
OFD1	100%	100%	99.2%	90.8%	71.1%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
PDE6D	100%	100%	100%	100%	99.8%	Joubert syndrome 22, 615665
PIBF1	100%	100%	100%	100%	99.6%	Joubert syndrome 33, 617767
PIK3C2A	100%	100%	100%	100%	99.8%	Oculoskeletodental syndrome, 618440
PKD1	100%	99.9%	100%	99.9%	99.2%	Polycystic kidney disease 1, 173900

PKD1L1	100%	100%	100%	100%	99.6%	Heterotaxy, visceral, 8, autosomal, 617205
PKD2	100%	100%	100%	100%	99.2%	Polycystic kidney disease 2, 613095
PKHD1	100%	100%	100%	100%	99.7%	Polycystic kidney disease 4, with or without hepatic disease, 263200
PMFBP1	100%	100%	100%	100%	99.4%	Spermatogenic failure 31, 618112
POC1A	100%	100%	100%	100%	99.4%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
RPGRIP1L	100%	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RSPH1	100%	100%	100%	99.8%	99.3%	Ciliary dyskinesia, primary, 24, 615481
RSPH3	100%	100%	100%	100%	99.7%	Ciliary dyskinesia, primary, 32, 616481
RSPH4A	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 11, 612649
RSPH9	100%	100%	100%	100%	99.4%	Ciliary dyskinesia, primary, 12, 612650
SCLT1	95.2%	95.2%	100%	100%	99.7%	
SDCCAG8	100%	100%	100%	100%	99.7%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993
SPAG1	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 28, 615505
SPATA7	100%	100%	100%	100%	99.8%	Leber congenital amaurosis 3, 604232;Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232

STK36	100%	100%	100%	100%	99.4%	?Ciliary dyskinesia, primary, 46, 619436
SUFU	100%	100%	100%	100%	99.3%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
TBC1D32	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome IX, 258865;Alsahan-Harris syndrome, 621307;Retinitis pigmentosa 100, 621280
TCTN1	93.6%	92.2%	100%	100%	99.5%	Joubert syndrome 13, 614173
TCTN2	98.5%	98.5%	100%	100%	99.5%	Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885
TCTN3	100%	100%	100%	100%	99.9%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TMEM107	100%	100%	100%	100%	99.1%	Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562
TMEM138	100%	100%	100%	100%	99.7%	Joubert syndrome 16, 614465
TMEM216	100%	100%	100%	100%	99.9%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194
TMEM218	100%	100%	100%	99.9%	99.3%	Joubert syndrome 39, 619562
TMEM231	93.5%	93.5%	100%	100%	99.4%	Joubert syndrome 20, 614970;Meckel syndrome 11, 615397

TMEM237	98.2%	98.2%	100%	100%	99.8%	Joubert syndrome 14, 614424
TMEM260	94.8%	94.8%	100%	100%	100%	Structural heart defects and renal anomalies syndrome, 617478
TMEM67	96.1%	96.1%	100%	100%	99.8%	Nephronophthisis 11, 613550;{Bardet-Biedl syndrome 14, modifier of}, 615991;Joubert syndrome 6, 610688;Meckel syndrome 3, 607361;?RHYNS syndrome, 602152;COACH syndrome 1, 216360
TOGARAM1	100%	100%	100%	100%	99.7%	Joubert syndrome 37, 619185
TOPORS	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 31, 609923
TP73	100%	100%	100%	99.9%	99.1%	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466
TRAF3IP1	100%	100%	100%	100%	99.2%	Senior-Loken syndrome 9, 616629
TRIM32	100%	100%	100%	100%	99.2%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TTBK2	100%	100%	100%	100%	99.7%	Spinocerebellar ataxia 11, 604432
TTC12	100%	100%	100%	100%	99.8%	Ciliary dyskinesia, primary, 45, 618801
TTC21B	98.4%	98.4%	100%	100%	99.8%	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820
TTC8	91.4%	91.4%	100%	100%	99.8%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464

TULP1	100%	100%	100%	99.9%	99.3%	Leber congenital amaurosis 15, 613843;Retinitis pigmentosa 14, 600132
TXNDC15	100%	99.2%	100%	99.9%	99.5%	Meckel syndrome 14, 619879
VHL	87.7%	87.7%	100%	100%	99.4%	Hemangioblastoma, cerebellar, somatic;Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300
WDPCP	92.1%	92.1%	100%	100%	99.8%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR19	100%	100%	100%	100%	99.8%	Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867
WDR35	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
XPNPEP3	100%	100%	100%	100%	99.5%	Nephronophthisis-like nephropathy 1, 613159

ZIC3	100%	100%	98.6%	84.4%	64.3%	Congenital heart defects, nonsyndromic, multiple types, 1, X-linked, 306955;Heterotaxy, visceral, 1, X-linked, 306955;VACTERL association, X-linked, 314390
ZMYND10	100%	100%	100%	100%	99.6%	Ciliary dyskinesia, primary, 22, 615444
ZNF423	100%	100%	100%	100%	99.5%	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.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors