

CILIOPATHIES PANEL DG-4.0.0 (184 GENES)

| <i>Gene</i> | <i>Twist X2 covered >10x</i> | <i>Twist X2 covered >20x</i> | <i>WGS covered >10x</i> | <i>WGS covered >20x</i> | <i>Associated Phenotype description and OMIM disease ID</i> |
|-------------|---------------------------------|---------------------------------|----------------------------|----------------------------|--|
| ACVR2B | 100.0% | 100.0% | 100.0% | 98.7% | Heterotaxy, visceral, 4, autosomal, 613751 |
| ADAMTS9 | 99.9% | 99.6% | 100.0% | 98.1% | |
| AHI1 | 98.7% | 98.7% | 100.0% | 98.2% | Joubert syndrome 3, 608629 |
| ALMS1 | 100.0% | 100.0% | 100.0% | 98.4% | Alstrom syndrome, 203800 |
| ANKS6 | 99.9% | 99.4% | 100.0% | 97.6% | Nephronophthisis 16, 615382 |
| ARL13B | 93.4% | 93.3% | 100.0% | 97.3% | Joubert syndrome 8, 612291 |
| ARL3 | 100.0% | 100.0% | 100.0% | 99.6% | Retinitis pigmentosa 83, 618173;Joubert syndrome 35, 618161 |
| ARL6 | 100.0% | 100.0% | 100.0% | 95.9% | Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151 |
| ARMC9 | 95.4% | 93.5% | 100.0% | 99.2% | Joubert syndrome 30, 617622 |
| B9D1 | 100.0% | 100.0% | 100.0% | 99.5% | ?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| B9D2 | 100.0% | 100.0% | 100.0% | 99.9% | ?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175 |
| BBIP1 | 100.0% | 100.0% | 100.0% | 98.9% | Bardet-Biedl syndrome 18, 615995 |
| BBS1 | 100.0% | 100.0% | 100.0% | 99.6% | Bardet-Biedl syndrome 1, 209900 |
| BBS10 | 100.0% | 100.0% | 100.0% | 98.7% | Bardet-Biedl syndrome 10, 615987 |
| BBS12 | 100.0% | 100.0% | 100.0% | 99.6% | Bardet-Biedl syndrome 12, 615989 |
| BBS2 | 98.0% | 98.0% | 100.0% | 98.8% | Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981 |
| BBS4 | 100.0% | 100.0% | 100.0% | 98.0% | Bardet-Biedl syndrome 4, 615982 |
| BBS5 | 100.0% | 100.0% | 100.0% | 98.9% | Bardet-Biedl syndrome 5, 615983 |
| BBS7 | 100.0% | 100.0% | 100.0% | 99.1% | Bardet-Biedl syndrome 7, 615984 |
| BBS9 | 95.8% | 95.8% | 100.0% | 97.9% | Bardet-Biedl syndrome 9, 615986 |
| C2CD3 | 96.0% | 96.0% | 100.0% | 98.8% | Orofaciodigital syndrome XIV, 615948 |
| CBY1 | 100.0% | 100.0% | 100.0% | 98.9% | |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| CC2D2A | 98.2% | 98.2% | 100.0% | 98.4% | COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285 |
| CCDC103 | 100.0% | 100.0% | 100.0% | 99.4% | Ciliary dyskinesia, primary, 17, 614679 |
| CCDC28B | 100.0% | 100.0% | 100.0% | 98.8% | {Bardet-Biedl syndrome 1, modifier of}, 209900 |
| CCDC39 | 100.0% | 100.0% | 100.0% | 96.4% | Ciliary dyskinesia, primary, 14, 613807 |
| CCDC40 | 100.0% | 100.0% | 100.0% | 99.0% | Ciliary dyskinesia, primary, 15, 613808 |
| CCDC65 | 100.0% | 100.0% | 100.0% | 97.8% | Ciliary dyskinesia, primary, 27, 615504 |
| CCNO | 100.0% | 100.0% | 100.0% | 98.9% | Ciliary dyskinesia, primary, 29, 615872 |
| CENPF | 100.0% | 100.0% | 100.0% | 97.8% | Stromme syndrome, 243605 |
| CEP104 | 100.0% | 100.0% | 100.0% | 98.1% | Joubert syndrome 25, 616781;Intellectual developmental disorder, autosomal recessive 77, 619988 |
| CEP120 | 100.0% | 100.0% | 100.0% | 98.9% | Short-rib thoracic dysplasia 13 with or without polydactyly, 616300;Joubert syndrome 31, 617761 |
| CEP164 | 100.0% | 100.0% | 100.0% | 98.2% | Nephronophthisis 15, 614845 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| CEP290 | 100.0% | 100.0% | 100.0% | 96.4% | 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% | 100.0% | 98.0% | Joubert syndrome 15, 614464 |
| CEP55 | 100.0% | 100.0% | 100.0% | 98.3% | Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 |
| CEP83 | 100.0% | 100.0% | 100.0% | 95.9% | Nephronophthisis 18, 615862 |
| CFAP251 | 100.0% | 100.0% | 100.0% | 98.1% | Spermatogenic failure 33, 618152 |
| CFAP298 | 100.0% | 100.0% | 100.0% | 97.7% | Ciliary dyskinesia, primary, 26, 615500 |
| CFAP300 | 100.0% | 100.0% | 100.0% | 97.0% | Ciliary dyskinesia, primary, 38, 618063 |
| CFAP410 | 100.0% | 100.0% | 100.0% | 99.0% | Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271 |
| CFAP418 | 100.0% | 100.0% | 100.0% | 98.5% | Retinitis pigmentosa 64, 614500;Cone-rod dystrophy 16, 614500;Bardet-Biedl syndrome 21, 617406 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| CFAP44 | 100.0% | 100.0% | 100.0% | 97.6% | Spermatogenic failure 20, 617593 |
| CFAP45 | 100.0% | 100.0% | 100.0% | 98.7% | Heterotaxy, visceral, 11, autosomal, with male infertility, 619608 |
| CFAP52 | 100.0% | 100.0% | 100.0% | 98.7% | Heterotaxy, visceral, 10, autosomal, with male infertility, 619607 |
| CFAP53 | 100.0% | 100.0% | 99.9% | 97.2% | Heterotaxy, visceral, 6, autosomal recessive, 614779 |
| CFAP69 | 100.0% | 100.0% | 100.0% | 97.6% | Spermatogenic failure 24, 617959 |
| CFC1 | 100.0% | 100.0% | 100.0% | 99.6% | Heterotaxy, visceral, 2, autosomal, 605376 |
| CPLANE1 | 100.0% | 100.0% | 100.0% | 98.0% | Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615 |
| CSPP1 | 96.9% | 96.9% | 100.0% | 97.9% | Joubert syndrome 21, 615636 |
| DCDC2 | 100.0% | 100.0% | 100.0% | 97.6% | Nephronophthisis 19, 616217;?Deafness, autosomal recessive 66, 610212;Sclerosing cholangitis, neonatal, 617394 |
| DDX59 | 100.0% | 100.0% | 100.0% | 98.1% | Orofaciodigital syndrome V, 174300 |
| DNAAF1 | 100.0% | 100.0% | 100.0% | 99.1% | Ciliary dyskinesia, primary, 13, 613193 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| DNAAF11 | 100.0% | 100.0% | 100.0% | 98.6% | Ciliary dyskinesia, primary, 19, 614935 |
| DNAAF2 | 100.0% | 100.0% | 100.0% | 96.4% | Ciliary dyskinesia, primary, 10, 612518 |
| DNAAF3 | 100.0% | 100.0% | 99.9% | 97.1% | Ciliary dyskinesia, primary, 2, 606763 |
| DNAAF4 | 100.0% | 100.0% | 100.0% | 96.1% | {Dyslexia, susceptibility to, 1}, 127700;Ciliary dyskinesia, primary, 25, 615482 |
| DNAAF5 | 100.0% | 99.9% | 99.9% | 96.3% | Ciliary dyskinesia, primary, 18, 614874 |
| DNAAF6 | 100.0% | 100.0% | 97.7% | 67.9% | Ciliary dyskinesia, primary, 36, X-linked, 300991 |
| DNAH1 | 100.0% | 100.0% | 100.0% | 99.2% | Spermatogenic failure 18, 617576;Ciliary dyskinesia, primary, 37, 617577 |
| DNAH11 | 100.0% | 100.0% | 100.0% | 98.3% | Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 |
| DNAH17 | 100.0% | 100.0% | 100.0% | 99.0% | Spermatogenic failure 39, 618643 |
| DNAH5 | 99.9% | 99.7% | 100.0% | 98.6% | Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 |
| DNAH8 | 100.0% | 99.7% | 100.0% | 97.8% | Spermatogenic failure 46, 619095 |
| DNAH9 | 100.0% | 100.0% | 100.0% | 98.8% | Ciliary dyskinesia, primary, 40, 618300 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| DNAI1 | 100.0% | 100.0% | 100.0% | 99.2% | Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 |
| DNAI2 | 100.0% | 100.0% | 100.0% | 97.9% | Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 |
| DNAJB13 | 100.0% | 100.0% | 100.0% | 98.3% | Ciliary dyskinesia, primary, 34, 617091 |
| DNAL1 | 100.0% | 100.0% | 100.0% | 98.0% | Ciliary dyskinesia, primary, 16, 614017 |
| DRC1 | 100.0% | 100.0% | 100.0% | 98.8% | Spermatogenic failure 80, 620222;Ciliary dyskinesia, primary, 21, 615294 |
| DYNC2H1 | 99.8% | 99.4% | 100.0% | 97.9% | Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 |
| DYNC2I1 | 100.0% | 100.0% | 100.0% | 98.0% | Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 |
| DYNC2I2 | 100.0% | 100.0% | 100.0% | 99.5% | Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 |
| DYNC2LI1 | 100.0% | 100.0% | 100.0% | 97.5% | Short-rib thoracic dysplasia 15 with polydactyly, 617088 |
| DYNLT2B | 100.0% | 100.0% | 100.0% | 94.3% | Short-rib thoracic dysplasia 17 with or without polydactyly, 617405 |
| EVC | 100.0% | 99.9% | 100.0% | 98.2% | Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| EVC2 | 100.0% | 100.0% | 100.0% | 98.5% | Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530 |
| EXOC8 | 100.0% | 100.0% | 100.0% | 97.6% | ?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076 |
| EXTL3 | 100.0% | 99.5% | 100.0% | 99.6% | Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 |
| FAM149B1 | 100.0% | 100.0% | 100.0% | 99.3% | Joubert syndrome 36, 618763 |
| FOXF1 | 100.0% | 100.0% | 100.0% | 92.4% | Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 |
| FOXJ1 | 100.0% | 100.0% | 100.0% | 97.2% | Ciliary dyskinesia, primary, 43, 618699 |
| FUZ | 100.0% | 100.0% | 100.0% | 98.6% | {Neural tube defects, susceptibility to}, 182940 |
| GAS8 | 100.0% | 100.0% | 100.0% | 99.5% | Ciliary dyskinesia, primary, 33, 616726 |
| GDF1 | 100.0% | 100.0% | 100.0% | 99.5% | Congenital heart defects, multiple types, 6, 613854;Right atrial isomerism (Ivemark), 208530 |
| GLIS2 | 100.0% | 100.0% | 100.0% | 99.6% | Nephronophthisis 7, 611498 |
| HYDIN | 100.0% | 100.0% | 100.0% | 98.3% | Ciliary dyskinesia, primary, 5, 608647 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| HYLS1 | 100.0% | 100.0% | 100.0% | 99.5% | Hydrolethalus syndrome, 236680 |
| IFT122 | 100.0% | 100.0% | 100.0% | 99.1% | Cranioectodermal dysplasia 1, 218330 |
| IFT140 | 100.0% | 100.0% | 100.0% | 99.1% | Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781 |
| IFT172 | 100.0% | 100.0% | 100.0% | 99.0% | Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 |
| IFT27 | 100.0% | 100.0% | 100.0% | 99.2% | Bardet-Biedl syndrome 19, 615996 |
| IFT43 | 100.0% | 100.0% | 100.0% | 98.7% | ?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866 |
| IFT52 | 100.0% | 100.0% | 100.0% | 98.2% | Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 |
| IFT56 | 100.0% | 100.0% | 100.0% | 98.5% | Biliary, renal, neurologic, and skeletal syndrome, 619534 |

| | | | | | |
|----------|--------|--------|--------|-------|---|
| IFT74 | 100.0% | 100.0% | 100.0% | 97.5% | Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582 |
| IFT80 | 100.0% | 100.0% | 100.0% | 98.2% | Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 |
| IFT81 | 94.9% | 94.9% | 100.0% | 98.4% | Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 |
| INPP5E | 100.0% | 100.0% | 100.0% | 97.0% | Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156;Joubert syndrome 1, 213300 |
| INTU | 100.0% | 100.0% | 100.0% | 97.2% | ?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925 |
| INVS | 100.0% | 100.0% | 100.0% | 99.2% | Nephronophthisis 2, infantile, 602088 |
| IQCB1 | 100.0% | 100.0% | 100.0% | 97.9% | Senior-Loken syndrome 5, 609254 |
| KATNIP | 100.0% | 100.0% | 100.0% | 99.2% | Joubert syndrome 26, 616784 |
| KCTD3 | 100.0% | 100.0% | 100.0% | 95.5% | |
| KIAA0586 | 95.6% | 95.5% | 100.0% | 98.0% | Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490 |

| | | | | | |
|----------|--------|--------|--------|-------|---|
| KIAA0753 | 100.0% | 100.0% | 100.0% | 98.8% | ?Orofaciodigital syndrome XV, 617127;?Joubert syndrome 38, 619476;Short-rib thoracic dysplasia 21 without polydactyly, 619479 |
| KIF14 | 100.0% | 100.0% | 100.0% | 98.4% | Microcephaly 20, primary, autosomal recessive, 617914;?Meckel syndrome 12, 616258 |
| KIF7 | 100.0% | 99.9% | 100.0% | 98.4% | Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131 |
| LBR | 100.0% | 100.0% | 100.0% | 97.9% | 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% | 100.0% | 97.8% | Leber congenital amaurosis 5, 604537 |
| LRRC56 | 100.0% | 100.0% | 100.0% | 99.3% | Ciliary dyskinesia, primary, 39, 618254 |
| LZTFL1 | 100.0% | 100.0% | 100.0% | 97.7% | Bardet-Biedl syndrome 17, 615994 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| MAPKBP1 | 100.0% | 100.0% | 100.0% | 99.2% | Nephronophthisis 20, 617271 |
| MCIDAS | 100.0% | 100.0% | 100.0% | 98.5% | Ciliary dyskinesia, primary, 42, 618695 |
| MKKS | 100.0% | 100.0% | 100.0% | 99.3% | McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231 |
| MKS1 | 99.0% | 99.0% | 100.0% | 98.9% | Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121 |
| MMP21 | 100.0% | 100.0% | 100.0% | 98.4% | Heterotaxy, visceral, 7, autosomal, 616749 |
| MNS1 | 100.0% | 100.0% | 100.0% | 97.1% | Heterotaxy, visceral, 9, autosomal, with male infertility, 618948 |
| NCAPG2 | 100.0% | 100.0% | 100.0% | 98.7% | Khan-Khan-Katsanis syndrome, 618460 |
| NEK1 | 100.0% | 100.0% | 100.0% | 98.2% | Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 |
| NEK10 | 100.0% | 99.8% | 100.0% | 98.5% | Ciliary dyskinesia, primary, 44, 618781 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| NEK8 | 100.0% | 100.0% | 100.0% | 99.5% | Renal-hepatic-pancreatic dysplasia 2, 615415;Polycystic kidney disease 8, 620903;?Nephronophthisis 9, 613824 |
| NME5 | 100.0% | 100.0% | 99.8% | 97.1% | Ciliary dyskinesia, primary, 48, without situs inversus, 620032 |
| NME8 | 99.9% | 99.5% | 100.0% | 98.0% | ?Ciliary dyskinesia, primary, 6, 610852 |
| NODAL | 100.0% | 100.0% | 100.0% | 99.1% | Heterotaxy, visceral, 5, 270100 |
| NPHP1 | 100.0% | 100.0% | 100.0% | 98.8% | Joubert syndrome 4, 609583;Nephronophthisis 1, juvenile, 256100;Senior-Loken syndrome-1, 266900 |
| NPHP3 | 100.0% | 100.0% | 100.0% | 98.3% | Nephronophthisis 3, 604387;Renal-hepatic-pancreatic dysplasia 1, 208540;Meckel syndrome 7, 267010 |
| NPHP4 | 100.0% | 100.0% | 100.0% | 99.5% | Senior-Loken syndrome 4, 606996;Nephronophthisis 4, 606966 |
| OCRL | 100.0% | 100.0% | 97.8% | 69.7% | Dent disease 2, 300555;Lowe syndrome, 309000 |
| ODAD1 | 100.0% | 100.0% | 100.0% | 98.9% | Ciliary dyskinesia, primary, 20, 615067 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| ODAD2 | 95.9% | 95.6% | 100.0% | 98.0% | Ciliary dyskinesia, primary, 23, 615451 |
| ODAD3 | 100.0% | 100.0% | 100.0% | 99.3% | Ciliary dyskinesia, primary, 30, 616037 |
| ODAD4 | 100.0% | 100.0% | 100.0% | 98.5% | Ciliary dyskinesia, primary, 35, 617092 |
| OFD1 | 100.0% | 100.0% | 96.1% | 66.2% | Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804 |
| PDE6D | 100.0% | 100.0% | 100.0% | 97.0% | Joubert syndrome 22, 615665 |
| PIBF1 | 100.0% | 100.0% | 100.0% | 95.0% | Joubert syndrome 33, 617767 |
| PIK3C2A | 100.0% | 100.0% | 100.0% | 98.0% | Oculoskeletodental syndrome, 618440 |
| PKD1 | 99.9% | 99.7% | 100.0% | 98.3% | Polycystic kidney disease 1, 173900 |
| PKD1L1 | 100.0% | 100.0% | 100.0% | 98.8% | Heterotaxy, visceral, 8, autosomal, 617205 |
| PKD2 | 100.0% | 100.0% | 99.8% | 93.4% | Polycystic kidney disease 2, 613095 |
| PKHD1 | 100.0% | 100.0% | 100.0% | 98.8% | Polycystic kidney disease 4, with or without hepatic disease, 263200 |
| PMFBP1 | 100.0% | 100.0% | 100.0% | 98.3% | Spermatogenic failure 31, 618112 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| POC1A | 100.0% | 100.0% | 100.0% | 99.6% | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 |
| RPGRIP1L | 100.0% | 100.0% | 100.0% | 97.4% | Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113 |
| RSPH1 | 100.0% | 100.0% | 100.0% | 98.4% | Ciliary dyskinesia, primary, 24, 615481 |
| RSPH3 | 100.0% | 100.0% | 100.0% | 98.9% | Ciliary dyskinesia, primary, 32, 616481 |
| RSPH4A | 100.0% | 100.0% | 100.0% | 96.7% | Ciliary dyskinesia, primary, 11, 612649 |
| RSPH9 | 100.0% | 100.0% | 100.0% | 98.7% | Ciliary dyskinesia, primary, 12, 612650 |
| SCLT1 | 95.2% | 95.2% | 100.0% | 97.2% | |
| SDCCAG8 | 100.0% | 100.0% | 100.0% | 97.9% | Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993 |
| SPAG1 | 100.0% | 100.0% | 100.0% | 96.4% | Ciliary dyskinesia, primary, 28, 615505 |
| SPATA7 | 100.0% | 100.0% | 100.0% | 97.6% | Leber congenital amaurosis 3, 604232;Retinitis pigmentosa 94, variable age at onset, autosomal recessive, 604232 |
| STK36 | 100.0% | 100.0% | 100.0% | 98.8% | ?Ciliary dyskinesia, primary, 46, 619436 |
| TBC1D32 | 100.0% | 100.0% | 100.0% | 98.0% | |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| TCTN1 | 97.8% | 96.4% | 100.0% | 97.5% | Joubert syndrome 13, 614173 |
| TCTN2 | 98.5% | 98.5% | 100.0% | 99.1% | Joubert syndrome 24, 616654;?Meckel syndrome 8, 613885 |
| TCTN3 | 100.0% | 100.0% | 100.0% | 98.9% | Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860 |
| TMEM107 | 100.0% | 100.0% | 100.0% | 98.7% | Orofaciodigital syndrome XVI, 617563;Meckel syndrome 13, 617562;?Joubert syndrome 29, 617562 |
| TMEM138 | 100.0% | 96.8% | 100.0% | 98.7% | Joubert syndrome 16, 614465 |
| TMEM216 | 100.0% | 100.0% | 100.0% | 98.6% | Joubert syndrome 2, 608091;Meckel syndrome 2, 603194 |
| TMEM218 | 100.0% | 100.0% | 100.0% | 98.9% | Joubert syndrome 39, 619562 |
| TMEM231 | 93.2% | 93.2% | 100.0% | 99.5% | Joubert syndrome 20, 614970;Meckel syndrome 11, 615397 |
| TMEM237 | 98.2% | 98.2% | 99.9% | 97.8% | Joubert syndrome 14, 614424 |
| TMEM260 | 100.0% | 100.0% | 100.0% | 97.9% | Structural heart defects and renal anomalies syndrome, 617478 |

| | | | | | |
|----------|--------|--------|--------|-------|---|
| TMEM67 | 96.1% | 96.1% | 100.0% | 95.5% | 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 | 100.0% | 100.0% | 100.0% | 97.8% | Joubert syndrome 37, 619185 |
| TOPORS | 100.0% | 100.0% | 100.0% | 97.9% | Retinitis pigmentosa 31, 609923 |
| TP73 | 100.0% | 100.0% | 100.0% | 99.7% | Ciliary dyskinesia, primary, 47, and lissencephaly, 619466 |
| TRAF3IP1 | 100.0% | 100.0% | 100.0% | 96.4% | Senior-Loken syndrome 9, 616629 |
| TRIM32 | 100.0% | 100.0% | 100.0% | 99.9% | ?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 |
| TTBK2 | 100.0% | 100.0% | 100.0% | 98.8% | Spinocerebellar ataxia 11, 604432 |
| TTC12 | 100.0% | 100.0% | 100.0% | 99.3% | Ciliary dyskinesia, primary, 45, 618801 |
| TTC21B | 98.2% | 97.6% | 100.0% | 98.6% | Short-rib thoracic dysplasia 4 with or without polydactyly, 613819;Nephronophthisis 12, 613820 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| TTC8 | 100.0% | 99.9% | 100.0% | 97.9% | Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464 |
| TULP1 | 100.0% | 100.0% | 100.0% | 98.3% | Leber congenital amaurosis 15, 613843;Retinitis pigmentosa 14, 600132 |
| TXNDC15 | 100.0% | 100.0% | 100.0% | 99.4% | Meckel syndrome 14, 619879 |
| VHL | 88.0% | 87.9% | 100.0% | 99.3% | Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300;Hemangioblastoma, cerebellar, somatic, |
| WDPCP | 97.5% | 97.3% | 100.0% | 98.7% | Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 |
| WDR19 | 100.0% | 100.0% | 99.9% | 97.7% | Nephronophthisis 13, 614377;Craniectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| WDR35 | 100.0% | 100.0% | 100.0% | 98.9% | Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610 |
| XPNPEP3 | 100.0% | 100.0% | 100.0% | 99.2% | Nephronophthisis-like nephropathy 1, 613159 |
| ZIC3 | 100.0% | 100.0% | 97.4% | 68.8% | Congenital heart defects, nonsyndromic, 1, X-linked, 306955;Heterotaxy, visceral, 1, X-linked, 306955;VACTERL association, X-linked, 314390 |
| ZMYND10 | 100.0% | 100.0% | 100.0% | 99.8% | Ciliary dyskinesia, primary, 22, 615444 |
| ZNF423 | 100.0% | 100.0% | 100.0% | 99.2% | Nephronophthisis 14, 614844;Joubert syndrome 19, 614844 |

Gene symbols used follow HGNC 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.

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.

srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.

This list is accurate for panel version DG 4.0.0