# CILIOPATHIES GENE PANEL DG 2.3.x

| *Gene* | *Median coverage* | *% covered > 10x* | *% covered > 20x* | *Associated Phenotype description and OMIM ID* |
|---|---|---|---|---|
| AHI1 | 105,6 | 100,00% | 98,60% | Joubert syndrome-3, 608629 |
| ALMS1 | 189 | 98,40% | 98,10% | Alstrom syndrome, 203800 |
| ANKS6 | 56,3 | 94,50% | 80,60% | Nephronophthisis 16, 615382 |
| ARL13B | 121,8 | 100,00% | 95,50% | Joubert syndrome 8, 612291 |
| ARL6 | 155,1 | 100,00% | 100,00% | Bardet-Biedl syndrome 3, 209900<br>{Bardet-Biedl syndrome 1, modifier of}, 209900<br>Retinitis pigmentosa 55, 613575 |
| ARMC4 | 88,6 | 88,40% | 85,30% | Ciliary dyskinesia, primary, 23, 615451 |
| ATXN10 | 110 | 100,00% | 99,90% | Spinocerebellar ataxia 10, 603516 |
| B9D1 | 72,5 | 92,20% | 86,50% | Meckel syndrome 9, 614209 |
| B9D2 | 49,5 | 100,00% | 100,00% | Meckel syndrome 10, 614175 |
| BBIP1 | 111 | 82,10% | 77,10% | ?Bardet-Biedl syndrome 18, 615995 |
| BBS1 | 116,1 | 99,80% | 98,40% | Bardet-Biedl syndrome 1, 209900 |
| BBS10 | 115,8 | 100,00% | 100,00% | Bardet-Biedl syndrome 10, 209900 |
| BBS12 | 132,3 | 100,00% | 100,00% | Bardet-Biedl syndrome 12, 209900 |
| BBS2 | 114,4 | 100,00% | 100,00% | Bardet-Biedl syndrome 2, 209900 |
| BBS4 | 98,5 | 96,10% | 93,40% | Bardet-Biedl syndrome 4, 209900 |
| BBS5 | 137,8 | 100,00% | 100,00% | Bardet-Biedl syndrome 5, 209900 |
| BBS7 | 119,1 | 100,00% | 98,90% | Bardet-Biedl syndrome 7, 209900 |
| BBS9 | 122,1 | 100,00% | 98,60% | Bardet-Biedl syndrome 9, 209900 |
| C21orf59 | 95,8 | 99,60% | 95,40% | Ciliary dyskinesia, primary, 26, 615500 |
| C2CD3 | 106,6 | 95,90% | 94,90% | ?Orofaciodigital syndrome XIV |
| C2orf71 | 105,4 | 99,70% | 94,90% | Retinitis pigmentosa 54, 613428 |
| C5orf42 | 120,5 | 100,00% | 100,00% | Joubert syndrome 17, 614615 |
| C8orf37 | 93,6 | 100,00% | 100,00% | Retinitis pigmentosa 64, 614500<br>Cone-rod dystrophy 16, 614500 |

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| CC2D2A | 94,8 | 98,20% | 97,70% | Joubert syndrome 9, 612285<br>Meckel syndrome 6, 612284<br>COACH syndrome, 216360 |
| CCDC103 | 106,7 | 100,00% | 100,00% | Ciliary dyskinesia, primary, 17, 614679 |
| CCDC114 | 69,7 | 100,00% | 96,30% | Ciliary dyskinesia, primary, 20, 615067 |
| CCDC151 | 99,7 | 93,30% | 91,50% | Ciliary dyskinesia, primary, 30, 616037 |
| CCDC28B | 80 | 100,00% | 100,00% | {Bardet-Biedl syndrome, modifier of}, 209900 |
| CCDC39 | 106 | 100,00% | 99,50% | Ciliary dyskinesia, primary, 14, 613807 |
| CCDC40 | 83,5 | 97,50% | 94,00% | Ciliary dyskinesia, primary, 15, 613808 |
| CCDC41 | 123,6 | 100,00% | 100,00% | Nephronophthisis 18, 615862 |
| CCDC65 | 72,6 | 98,60% | 96,50% | Ciliary dyskinesia, primary, 27, 615504 |
| CCNO | 82,7 | 98,60% | 96,00% | Ciliary diskinesia, primary, 29, 615872 |
| CDH23 | 93,4 | 99,20% | 95,80% | Usher syndrome, type 1D, 601067<br>Deafness, autosomal recessive 12, 601386<br>Usher syndrome, type 1D/F digenic, 601067 |
| CENPF | 124,6 | 99,60% | 99,20% | Ciliary dyskinesia, primary, 31, 616369 |
| CEP120 | 121 | 100,00% | 98,20% | Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 |
| CEP164 | 72,4 | 97,00% | 90,10% | Nephronophthisis 15, 614845 |
| CEP290 | 95,3 | 100,00% | 98,90% | Joubert syndrome 5, 610188<br>Senior-Loken syndrome 6, 610189<br>Leber congenital amaurosis 10, 611755<br>Meckel syndrome 4, 611134<br>Bardet-Biedl syndrome 14, 209900 |
| CEP41 | 89,6 | 100,00% | 100,00% | Joubert syndrome 15, 614464 |
| CLRN1 | 145,2 | 100,00% | 100,00% | Retinitis pigmentosa 61, 614180<br>Usher syndrome type 3A, 276902<br>Retinitis pigmentosa 61, 614180 |
| CSPP1 | 119,1 | 100,00% | 100,00% | Joubert syndrome 21, 615636 |
| DCDC2 | 159,6 | 100,00% | 99,60% | ?Deafness, autosomal recessive 66, 610212<br>Nephronophthisis19, 616217 |
| DDX59 | 138,8 | 100,00% | 100,00% | Orofaciodigital syndrome V, 174300 |
| DFNB31 | 90,4 | 99,70% | 98,20% | Deafness, autosomal recessive 31, 607084<br>Usher syndrome, type 2D, 611383 |
| DNAAF1 | 113,4 | 99,20% | 96,00% | Ciliary dyskinesia, primary, 13, 613193 |

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| DNAAF2 | 114,8 | 100,00% | 100,00% | Ciliary dyskinesia, primary, 10, 612518 |
| DNAAF3 | 66,6 | 88,90% | 77,40% | Ciliary dyskinesia, primary, 2, 606763 |
| DNAH11 | 113,5 | 100,00% | 99,20% | Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 |
| DNAH5 | 92,2 | 100,00% | 98,50% | Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 |
| DNAI1 | 121,6 | 100,00% | 99,80% | Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 |
| DNAI2 | 98,6 | 97,20% | 94,70% | Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 |
| DNAL1 | 135,4 | 100,00% | 100,00% | Ciliary dyskinesia, primary, 16, 614017 |
| DRC1 | 74,1 | 100,00% | 97,50% | Ciliary dyskinesia, primary, 21, 615294 |
| DYNC2H1 | 113,2 | 99,40% | 98,90% | Asphyxiating thoracic dystrophy 3, 613091<br>Short rib-polydactyly syndrome, type III, 263510<br>Short rib-polydactyly syndrome, type IIB, 615087 |
| DYX1C1 | 85,8 | 100,00% | 97,90% | Ciliary dyskinesia, primary, 25, 615482<br>{Dyslexia, susceptibility to, 1}, 127700 |
| EVC | 71 | 91,30% | 87,60% | Ellis-van Creveld syndrome, 225500<br>Weyers acrodental dysostosis, 193530 |
| EVC2 | 98,4 | 93,70% | 92,20% | Ellis-van Creveld syndrome, 225500 |
| EXOC8 | 162,2 | 100,00% | 100,00% | No OMIM phenotype<br>Joubert syndrome (Dixon-Salazar (2012) Sci Transl Med 4, 138ra78) |
| FAM161A | 138,9 | 100,00% | 100,00% | Retinitis pigmentosa 28, 606068 |
| FLCN | 102,5 | 99,90% | 97,20% | Birt-Hogg-Dube syndrome, 135150<br>Pneumothorax, primary spontaneous, 173600<br>Renal carcinoma, chromophobe, somatic, 144700<br>Colorectal cancer, somatic, 114500 |
| GLIS2 | 88,3 | 100,00% | 97,00% | Nephronophthisis 7, 611498 |
| GPR98 | 112 | 99,40% | 98,00% | Febrile seizures, familial, 4, 604352<br>Usher syndrome, type 2C, 605472<br>Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472 |
| HEATR2 | 65,5 | 81,10% | 72,90% | Ciliary dyskinesia, primary, 18, 614874 |
| HYDIN | 90,6 | 88,20% | 85,50% | Ciliary dyskinesia, primary, 5, 608647 |
| HYLS1 | 153,5 | 100,00% | 100,00% | Hydrolethalus syndrome, 236680 |
| IFT122 | 77,6 | 96,10% | 92,00% | Cranioectodermal dysplasia 1, 218330 |
| IFT140 | 80,2 | 99,10% | 94,20% | Short-rib thoracic dysplasia 9 with of without polydactyly, 266920 |
| IFT172 | 92,7 | 99,90% | 96,60% | Short-rib thoracic dysplasia 10 with or without polydactyly, 615630<br>Retinitis pigmentosa 71, 616394 |

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| IFT27 | 90,1 | 82,90% | 82,40% | Bardet-Biedl syndrome 19, 615996 |
| IFT43 | 88,9 | 100,00% | 100,00% | Cranioectodermal dysplasia 3, 614099 |
| IFT80 | 81 | 99,30% | 92,60% | Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 |
| INPP5E | 75,8 | 99,80% | 96,60% | Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156<br>Joubert syndrome 1, 213300 |
| INVS | 114,4 | 99,10% | 97,00% | Nephronophthisis 2, infantile, 602088 |
| IQCB1 | 93 | 100,00% | 93,30% | Senior-Loken syndrome 5, 609254 |
| KIAA0586 | 107,3 | 98,20% | 96,50% | No OMIM phenotype |
| KIF14 | 112 | 100,00% | 99,50% | ?Meckel syndrome 12, 616258 |
| KIF7 | 68,8 | 89,20% | 84,80% | Hydrolethalus syndrome 2, 614120<br>Acrocallosal syndrome, 200990<br>Joubert syndrome 12, 200990 |
| LCA5 | 148,7 | 100,00% | 98,10% | Leber congenital amaurosis 5, 604537 |
| LRRC6 | 111 | 100,00% | 100,00% | Ciliary dyskinesia, primary, 19, 614935 |
| LZTFL1 | 85 | 100,00% | 98,70% | Bardet-Biedl syndrome 17, 615994 |
| MAK | 81,3 | 95,20% | 93,30% | Retinitis pigmentosa 62, 614181 |
| MKKS | 128,9 | 89,90% | 89,90% | McKusick-Kaufman syndrome, 236700<br>Bardet-Biedl syndrome 6, 209900 |
| MKS1 | 102,4 | 98,80% | 95,20% | Meckel syndrome 1, 249000<br>Bardet-Biedl syndrome 13, 209900 |
| MYO7A | 75,7 | 95,60% | 89,60% | Usher syndrome, type 1B, 276900<br>Deafness, autosomal recessive 2, 600060<br>Deafness, autosomal dominant 11, 601317 |
| NEK1 | 119,4 | 99,90% | 99,10% | Short rib-polydactyly syndrome, type IIA, 263520 |
| NEK8 | 104,7 | 100,00% | 100,00% | ?Nephronophthisis 9, 613824<br>?Renal-hepatic-pancreatic dysplasia 2, 615415 |
| NME8 | 103,9 | 100,00% | 100,00% | Ciliary dyskinesia, primary, 6, 610852 |
| NPHP1 | 118,9 | 100,00% | 99,50% | Nephronophthisis 1, juvenile, 256100<br>Senior-Loken syndrome-1, 266900<br>Joubert syndrome 4, 609583 |
| NPHP3 | 105 | 100,00% | 99,70% | Nephronophthisis 3, 604387<br>Renal-hepatic-pancreatic dysplasia 1, 208540<br>Meckel syndrome 7, 267010 |

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| NPHP4 | 87,7 | 98,70% | 93,90% | Nephronophthisis 4, 606966<br>Senior-Loken syndrome 4, 606996 |
| OCRL | 59,7 | 98,60% | 96,10% | Lowe syndrome, 309000<br>Dent disease 2, 300555 |
| OFD1 | 36,6 | 89,40% | 76,60% | Oral-facial-digital syndrome 1, 311200<br>Simpson-Golabi-Behmel syndrome, type 2, 300209<br>Joubert syndrome 10, 300804 |
| PCDH15 | 127 | 99,80% | 99,70% | Usher syndrome, type 1F, 602083<br>Deafness, autosomal recessive 23, 609533<br>Usher syndrome, type 1D/F digenic, 601067 |
| PDE6D | 97,4 | 100,00% | 100,00% | ?Joubert syndrome 22, 615665 |
| PDZD7 | 71,2 | 95,40% | 89,60% | {Retinal disease in Usher syndrome type IIA, modifier of}, 276901<br>Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472 |
| PKD1 | 13,7 | 20,00% | 18,50% | Polycystic kidney disease, adult type I, 173900 |
| PKD2 | 97,1 | 95,50% | 89,70% | Polycystic kidney disease 2, 613095 |
| PKHD1 | 97,1 | 98,90% | 96,50% | Polycystic kidney and hepatic disease, 263200 |
| POC1A | 96 | 98,50% | 96,00% | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 |
| PTPRQ | 106,5 | 94,90% | 93,50% | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 |
| RP1 | 169,5 | 100,00% | 100,00% | Retinitis pigmentosa 1, 180100<br>{Hypertriglyceridemia, susceptibility to}, 145750 |
| RP2 | 64,5 | 100,00% | 99,80% | Retinitis pigmentosa 2, 312600 |
| RPGR | 73,2 | 85,40% | 81,20% | Retinitis pigmentosa 3, 300029<br>Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455<br>Macular degeneration, X-linked atrophic, 300834<br>Cone-rod dystrophy, X-linked, 1, 304020 |
| RPGRIP1 | 113,3 | 100,00% | 97,80% | Leber congenital amaurosis 6, 613826<br>Cone-rod dystrophy 13, 608194 |
| RPGRIP1L | 99,8 | 96,70% | 96,70% | Joubert syndrome 7, 611560<br>Meckel syndrome 5, 611561<br>COACH syndrome, 216360 |
| RSPH1 | 110,1 | 100,00% | 99,70% | Ciliary dyskinesia, primary, 24, 615481 |
| RSPH4A | 139,2 | 100,00% | 100,00% | Ciliary dyskinesia, primary, 11, 612649 |
| RSPH9 | 75,7 | 100,00% | 94,60% | Ciliary dyskinesia, primary, 12, 612650 |

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| SCLT1 | 117,2 | 94,90% | 93,80% | No OMIM phenotype<br>Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35,36) |
| SDCCAG8 | 100,8 | 100,00% | 99,60% | Senior-Loken syndrome 7, 613615 |
| SPAG1 | 122 | 100,00% | 98,60% | Ciliary dyskinesia, primary, 28, 615505 |
| SPATA7 | 125,3 | 100,00% | 99,00% | Leber congenital amaurosis 3, 604232<br>Retinitis pigmentosa, juvenile, autosomal recessive, 604232 |
| TBC1D32 | 110,4 | 100,00% | 100,00% | No OMIM phenotype<br>Oro-facio-digital syndrome type IX (Adly (2014) Hum Mutat 35, 36) |
| TCTN1 | 100 | 95,70% | 94,90% | Joubert syndrome 13, 614173 |
| TCTN2 | 85,8 | 100,00% | 97,50% | Meckel syndrome 8, 613885 |
| TCTN3 | 98,3 | 100,00% | 99,30% | Orofaciodigital syndrome IV, 258860<br>Joubert syndrome 18, 614815 |
| TMEM138 | 92,6 | 100,00% | 100,00% | Joubert syndrome 16, 614465 |
| TMEM216 | 65,6 | 100,00% | 86,40% | Joubert syndrome 2, 608091<br>Meckel syndrome 2, 603194 |
| TMEM231 | 59,6 | 97,00% | 87,60% | Joubert syndrome 20, 614970<br>Meckel syndrome, type 11, 615397 |
| TMEM237 | 87,8 | 100,00% | 92,50% | Joubert syndrome 14, 614424 |
| TMEM67 | 115,7 | 100,00% | 99,80% | Meckel syndrome 3, 607361<br>Joubert syndrome 6, 610688<br>{Bardet-Biedl syndrome 14, modifier of}, 209900<br>COACH syndrome, 216360<br>Nephronophthisis 11, 613550 |
| TOPORS | 149,8 | 100,00% | 100,00% | Retinitis pigmentosa 31, 609923 |
| TRIM32 | 106,2 | 100,00% | 100,00% | Muscular dystrophy, limb-girdle, type 2H, 254110<br>Bardet-Biedl syndrome 11, 209900 |
| TTBK2 | 116,7 | 100,00% | 99,50% | Spinocerebellar ataxia 11, 604432 |
| TTC21B | 108,1 | 98,90% | 98,00% | Nephronophthisis 12, 613820<br>Asphyxiating thoracic dystrophy 4, 613819 |
| TTC8 | 103,9 | 100,00% | 100,00% | Bardet-Biedl syndrome 8, 209900<br>Retinitis pigmentosa 51, 613464 |
| TULP1 | 86,5 | 97,50% | 91,40% | Retinitis pigmentosa 14, 600132<br>Leber congenital amaurosis 15, 613843 |

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| USH1C | 71,5 | 98,40% | 93,00% | Acadian and Samaritan variety Usher syndrome, type 1C, 276904<br>Deafness, autosomal recessive 18A, 602092 |
| USH1G | 94,2 | 93,00% | 86,70% | Usher syndrome, type 1G, 606943 |
| USH2A | 113,1 | 100,00% | 99,30% | Usher syndrome, type 2A, 276901<br>Retinitis pigmentosa 39, 613809 |
| VHL | 111 | 100,00% | 100,00% | von Hippel-Lindau syndrome, 193300<br>Renal cell carcinoma, somatic, 144700<br>Pheochromocytoma, 171300<br>Hemangioblastoma, cerebellar, somatic<br>Erythrocytosis, familial, 2, 263400 |
| WDPCP | 86,8 | 97,80% | 95,60% | ?Bardet-Biedl syndrome 15, 615992 |
| WDR19 | 122,7 | 100,00% | 100,00% | Asphyxiating thoracic dystrophy 5, 614376<br>Nephronophthisis 13, 614377<br>Cranioectodermal dysplasia 4, 614378 |
| WDR34 | 87,2 | 100,00% | 97,60% | Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 |
| WDR35 | 112,4 | 100,00% | 97,80% | Cranioectodermal dysplasia 2, 613610<br>Short rib-polydactyly syndrome, type V, 614091 |
| WDR60 | 106,8 | 99,50% | 98,60% | Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 |
| XPNPEP3 | 115,8 | 97,30% | 94,90% | Nephronophthisis-like nephropathy 1, 613159 |
| ZMYND10 | 84,4 | 100,00% | 96,20% | Ciliary dyskinesia, primary, 22, 615444 |
| ZNF423 | 123,1 | 100,00% | 99,60% | Nephronophthisis 14, 614844<br>Joubert syndrome 19, 614844 |

*Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014*
*Median Coverage describes the average number of reads seen across 50 exomes*
*% 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*
*OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015*
*This list is accurate for all panel versions starting with DG 2.3. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)*

*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*