Ciliopathy

Gene panel

Gene panel information

Gene panel	**Ciliopathy**
Version	5
Total genes	208
Activation date	Thursday 03 november 2022
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ACVR2B	99.79 %	602730	Heterotaxy, visceral, 4, autosomal, 613751 (3)
ADAMTS9	99.64 %	605421	*No OMIM phenotypes*
AHI1	99.8 %	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
ALMS1	99.82 %	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ANKS6	99.91 %	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
ARL13B	99.78 %	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
ARL3	99.94 %	604695	Retinitis pigmentosa 83, 618173 (3), Autosomal dominant; Joubert syndrome 35, 618161 (3), Autosomal recessive
ARL6	99.89 %	608845	Retinitis pigmentosa 55, 613575 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive; Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive
ARMC9	99.29 %	617612	Joubert syndrome 30, 617622 (3), Autosomal recessive
B9D1	99.49 %	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
B9D2	99.25 %	611951	?Meckel syndrome 10, 614175 (3), Autosomal recessive; Joubert syndrome 34, 614175 (3), Autosomal recessive
BBIP1	99.91 %	613605	Bardet-Biedl syndrome 18, 615995 (3), Autosomal recessive
BBS1	99.96 %	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
BBS10	99.8 %	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS12	99.98 %	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
BBS2	99.4 %	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive
BBS4	99.38 %	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
BBS5	98.81 %	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
BBS7	99.54 %	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
BBS9	99.75 %	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
C2CD3	99.73 %	615944	Orofaciodigital syndrome XIV, 615948 (3), Autosomal recessive
CBY1	99.98 %	607757	*No OMIM phenotypes*
CC2D2A	99.85 %	612013	COACH syndrome 2, 619111 (3), Autosomal recessive; Retinitis pigmentosa 93, 619845 (3), Autosomal recessive; Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive
CCDC103	99.55 %	614677	Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive
CCDC172	99.76 %		*No OMIM phenotypes*
CCDC28B	99.56 %	610162	{Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive
CCDC32	99.81 %	618941	Cardiofacioneurodevelopmental syndrome, 619123 (3), Autosomal recessive

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CCDC39	99.76 %	613798	Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive
CCDC40	99.94 %	613799	Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive
CCDC65	99.63 %	611088	Ciliary dyskinesia, primary, 27, 615504 (3), Autosomal recessive
CCDC96	99.96 %	619347	*No OMIM phenotypes*
CCNO	99.94 %	607752	Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive
CENPF	99.88 %	600236	Stromme syndrome, 243605 (3), Autosomal recessive
CEP104	99.96 %	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive; Intellectual developmental disorder, autosomal recessive 77, 619988 (3), Autosomal recessive
CEP120	99.85 %	613446	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive; Joubert syndrome 31, 617761 (3), Autosomal recessive
CEP164	99.9 %	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
CEP290	98.33 %	610142	Leber congenital amaurosis 10, 611755 (3); Joubert syndrome 5, 610188 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive
CEP295	99.93 %	617728	Seckel syndrome 11, 620767 (3), Autosomal recessive
CEP41	99.97 %	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
CEP55	99.77 %	610000	Multinucleated neurons, anhydramnios, renal dysplasia, cerebellar hypoplasia, and hydranencephaly, 236500 (3), Autosomal recessive
CEP83	98.61 %	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
CFAP251	99.95 %	618146	Spermatogenic failure 33, 618152 (3), Autosomal recessive
CFAP298	99.7 %	615494	Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive
CFAP300	99.81 %	618058	Ciliary dyskinesia, primary, 38, 618063 (3), Autosomal recessive
CFAP410	99.95 %	603191	Retinal dystrophy with macular staphyloma, 617547 (3), Autosomal recessive; Spondylometaphyseal dysplasia, axial, 602271 (3), Autosomal recessive
CFAP418	99.92 %	614477	Retinitis pigmentosa 64, 614500 (3), Autosomal recessive; Cone-rod dystrophy 16, 614500 (3), Autosomal recessive; Bardet-Biedl syndrome 21, 617406 (3), Autosomal recessive
CFAP44	99.69 %	617559	Spermatogenic failure 20, 617593 (3), Autosomal recessive
CFAP53	99.91 %	614759	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive
CFAP69	99.21 %	617949	Spermatogenic failure 24, 617959 (3), Autosomal recessive
CFC1	20.68 %	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant
CILK1	99.45 %	612325	Cranioectodermal dysplasia 6, 621337 (3), Autosomal recessive; {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant; Endocrine-cerebroosteodysplasia, 612651 (3), Autosomal recessive
CPLANE1	99.74 %	614571	Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive; Joubert syndrome 17, 614615 (3), Autosomal recessive
CRB2	99.65 %	609720	Focal segmental glomerulosclerosis 9, 616220 (3), Autosomal recessive; Ventriculomegaly with cystic kidney disease, 219730 (3), Autosomal recessive
CSPP1	99.85 %	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
DCDC2	99.89 %	605755	Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive; Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive
DDX59	99.38 %	615464	Orofaciodigital syndrome V, 174300 (3), Autosomal recessive
DEUP1	99.85 %	617148	*No OMIM phenotypes*
DHCR7	99.89 %	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DLG5	99.56 %	604090	Yuksel-Vogel-Bauser syndrome, 620703 (3), Autosomal recessive
DNAAF1	99.66 %	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive

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DNAAF11	99.73 %	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
DNAAF2	99.89 %	612517	Ciliary dyskinesia, primary, 10, 612518 (3), Autosomal recessive
DNAAF3	99.83 %	614566	Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive
DNAAF4	99.72 %	608706	{Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant; Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive
DNAAF5	99.79 %	614864	Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive
DNAAF6	99.34 %	300933	Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive
DNAH1	99.67 %	603332	Spermatogenic failure 18, 617576 (3), Autosomal recessive; Ciliary dyskinesia, primary, 37, 617577 (3), Autosomal recessive
DNAH11	99.87 %	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
DNAH5	99.89 %	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3), Autosomal recessive
DNAH8	99.69 %	603337	Spermatogenic failure 46, 619095 (3), Autosomal recessive
DNAI1	99.56 %	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
DNAI2	99.82 %	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3), Autosomal recessive
DNAJB13	99.46 %	610263	Ciliary dyskinesia, primary, 34, 617091 (3), Autosomal recessive
DNAL1	99.88 %	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
DRC1	99.73 %	615288	Spermatogenic failure 80, 620222 (3), Autosomal recessive; Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive
DYNC2H1	99.74 %	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive
DYNC2I1	99.94 %	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
DYNC2I2	99.68 %	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive
DYNC2LI1	99.86 %	617083	Short-rib thoracic dysplasia 15 with polydactyly, 617088 (3), Autosomal recessive
DYNLT2B	99.98 %	617353	Short-rib thoracic dysplasia 17 with or without polydactyly, 617405 (3), Autosomal recessive
EVC	99.73 %	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	99.79 %	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EXOC3L2	99.81 %	616927	Brain malformation renal syndrome, 620943 (3), Autosomal recessive
EXOC6B	99.39 %	607880	Spondyloepimetaphyseal dysplasia with joint laxity, type 3, 618395 (3), Autosomal recessive
EXOC8	100 %	615283	?Neurodevelopmental disorder with microcephaly, seizures, and brain atrophy, 619076 (3), Autosomal recessive
EXTL3	99.94 %	605744	Immunoskeletal dysplasia with neurodevelopmental abnormalities, 617425 (3), Autosomal recessive
FAM149B1	98.11 %	618413	Joubert syndrome 36, 618763 (3), Autosomal recessive
FAM166B	99.9 %		*No OMIM phenotypes*
FOXF1	99.94 %	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
FUZ	99.56 %	610622	{Neural tube defects, susceptibility to}, 182940 (3), Autosomal dominant
GAS2L2	99.91 %	611398	?Ciliary dyskinesia, primary, 41, 618449 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GAS8	99.95 %	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
GLI3	99.97 %	165240	Greig cephalopolysyndactyly syndrome, 175700 (3), Autosomal dominant; Polydactyly, postaxial, types A1 and B, 174200 (3), Autosomal dominant; Pallister-Hall syndrome, 146510 (3), Autosomal dominant; Polydactyly, preaxial, type IV, 174700 (3), Autosomal dominant
GLIS2	99.95 %	608539	Nephronophthisis 7, 611498 (3), Autosomal recessive
HNF1B	99.77 %	189907	Type 2 diabetes mellitus, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3)
HYDIN	76.66 %	610812	Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive
HYLS1	100 %	610693	Hydrolethalus syndrome, 236680 (3), Autosomal recessive
IFT122	99.49 %	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT140	99.77 %	614620	{Polycystic kidney disease 9, susceptibility to}, 621164 (3), Autosomal dominant; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive; Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Cranioectodermal dysplasia 5, 621180 (3), Autosomal recessive
IFT172	99.83 %	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Bardet-Biedl syndrome 20, 619471 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
IFT27	99.94 %	615870	Bardet-Biedl syndrome 19, 615996 (3), Autosomal recessive
IFT43	99.83 %	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive
IFT52	99.81 %	617094	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102 (3), Autosomal recessive
IFT74	99.71 %	608040	Bardet-Biedl syndrome 22, 617119 (3), Autosomal recessive; Spermatogenic failure 58, 619585 (3), Autosomal recessive; Joubert syndrome 40, 619582 (3), Autosomal recessive
IFT80	99.71 %	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
IFT81	94.88 %	605489	Short-rib thoracic dysplasia 19 with or without polydactyly, 617895 (3), Autosomal recessive
INPP5E	99.35 %	613037	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
INTS13	98.85 %	615079	*No OMIM phenotypes*
INTU	99.87 %	610621	?Orofaciodigital syndrome XVII, 617926 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 20 with polydactyly, 617925 (3), Autosomal recessive
INVS	99.93 %	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
IQCB1	99.56 %	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
IQCE	99.87 %	617631	Polydactyly, postaxial, type A7, 617642 (3), Autosomal recessive
KATNIP	99.1 %	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
KCTD3	99.38 %	613272	*No OMIM phenotypes*
KIAA0586	99.33 %	610178	Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive; Joubert syndrome 23, 616490 (3), Autosomal recessive
KIAA0753	99.95 %	617112	?Orofaciodigital syndrome XV, 617127 (3), Autosomal recessive; ?Joubert syndrome 38, 619476 (3), Autosomal recessive; Short-rib thoracic dysplasia 21 without polydactyly, 619479 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KIF14	98.75 %	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive
KIF3B	99.98 %	603754	Retinitis pigmentosa 89, 618955 (3), Autosomal dominant
KIF7	99.84 %	611254	Joubert syndrome 12, 200990 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; ?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
LBR	99.84 %	600024	Pelger-Huet anomaly, 169400 (3), Autosomal dominant; ?Reynolds syndrome, 613471 (3), Autosomal dominant; Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019 (3), Autosomal recessive; Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive
LCA5	99.89 %	611408	Leber congenital amaurosis 5, 604537 (3), Autosomal recessive
LRRC34	99.35 %	619037	*No OMIM phenotypes*
LRRC45	99.77 %	621312	*No OMIM phenotypes*
LRRC56	99.77 %	618227	Ciliary dyskinesia, primary, 39, 618254 (3), Autosomal recessive
LZTFL1	99.98 %	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
MAPKBP1	99.77 %	616786	Nephronophthisis 20, 617271 (3), Autosomal recessive
MCIDAS	99.93 %	614086	Ciliary dyskinesia, primary, 42, 618695 (3), Autosomal recessive
MKKS	99.99 %	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
MKS1	99.62 %	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
MMP21	99.82 %	608416	Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive
MRE11	99.93 %	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
NCAPG2	99.83 %	608532	Khan-Khan-Katsanis syndrome, 618460 (3), Autosomal recessive
NEK1	99.87 %	604588	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive; ?Orofaciodigital syndrome II, 252100 (3), Autosomal recessive; {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant
NEK8	99.8 %	609799	Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; Polycystic kidney disease 8, 620903 (3), Autosomal dominant; ?Nephronophthisis 9, 613824 (3)
NEK9	99.84 %	609798	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 (3), Autosomal recessive; Nevus comedonicus, somatic, 617025 (3); Lethal congenital contracture syndrome 10, 617022 (3), Autosomal recessive
NME5	99.69 %	603575	Ciliary dyskinesia, primary, 48, without situs inversus, 620032 (3), Autosomal recessive
NME8	99.8 %	607421	?Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive
NPHP1	97.52 %	607100	Joubert syndrome 4, 609583 (3), Autosomal recessive; Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive
NPHP3	99.49 %	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
NPHP4	99.89 %	607215	Senior-Loken syndrome 4, 606996 (3), Autosomal recessive; Nephronophthisis 4, 606966 (3), Autosomal recessive
OCRL	99.78 %	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive

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ODAD1	99.76 %	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
ODAD2	98.01 %	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
ODAD3	99.65 %	615956	Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive
ODAD4	99.5 %	617095	Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive
OFD1	99.71 %	300170	Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; Joubert syndrome 10, 300804 (3), X-linked recessive
PDE6D	99.44 %	602676	Joubert syndrome 22, 615665 (3), Autosomal recessive
PIBF1	99.91 %	607532	Joubert syndrome 33, 617767 (3), Autosomal recessive
PIK3C2A	99.81 %	603601	Oculoskeletodental syndrome, 618440 (3), Autosomal recessive
PKD1	99.48 %	601313	Polycystic kidney disease 1, 173900 (3), Autosomal dominant
PKD2	99.79 %	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
PKHD1	99.85 %	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
PMFBP1	99.58 %	618085	Spermatogenic failure 31, 618112 (3), Autosomal recessive
PMM2	99.59 %	601785	Congenital disorder of glycosylation, type Ia, 212065 (3), Autosomal recessive
POC1A	99.49 %	614783	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 (3), Autosomal recessive
POC1B	98.54 %	614784	Cone-rod dystrophy 20, 615973 (3), Autosomal recessive
RAB28	99.88 %	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
RPGR	99.36 %	312610	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3), X-linked; Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
RPGRIP1L	96.51 %	610937	Joubert syndrome 7, 611560 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; ?COACH syndrome 3, 619113 (3), Autosomal recessive
RSPH1	98.95 %	609314	Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive
RSPH3	99.85 %	615876	Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive
RSPH4A	99.84 %	612647	Ciliary dyskinesia, primary, 11, 612649 (3), Autosomal recessive
RSPH9	99.64 %	612648	Ciliary dyskinesia, primary, 12, 612650 (3), Autosomal recessive
SBDS	99.32 %	607444	{Aplastic anemia, susceptibility to}, 609135 (3); Shwachman-Diamond syndrome 1, 260400 (3), Autosomal recessive
SCLT1	98.43 %	611399	*No OMIM phenotypes*
SCNM1	98.8 %	608095	Orofaciodigital syndrome XIX, 620107 (3), Autosomal recessive
SDCCAG8	100 %	613524	Senior-Loken syndrome 7, 613615 (3), Autosomal recessive; Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive
SLC30A7	96.67 %	611149	Ziegler-Huang syndrome, 620501 (3), Autosomal recessive
SPAG1	99.63 %	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive
STK36	99.63 %	607652	?Ciliary dyskinesia, primary, 46, 619436 (3), Autosomal recessive
SUFU	99.9 %	607035	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Basal cell nevus syndrome 2, 620343 (3); {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant
TBC1D32	99.75 %	615867	Orofaciodigital syndrome IX, 258865 (3), Autosomal recessive; Alsahan-Harris syndrome, 621307 (3), Autosomal recessive; Retinitis pigmentosa 100, 621280 (3), Autosomal recessive

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TCTN1	99.84 %	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
TCTN2	99.95 %	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TCTN3	99.84 %	613847	Joubert syndrome 18, 614815 (3), Autosomal recessive; Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive
TMEM107	99.96 %	616183	Orofaciodigital syndrome XVI, 617563 (3), Autosomal recessive; Meckel syndrome 13, 617562 (3), Autosomal recessive; ?Joubert syndrome 29, 617562 (3), Autosomal recessive
TMEM138	99.94 %	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
TMEM17	99.16 %	614950	*No OMIM phenotypes*
TMEM216	99.5 %	613277	Joubert syndrome 2, 608091 (3), Autosomal recessive; Retinitis pigmentosa 98, 620996 (3), Autosomal recessive; Meckel syndrome 2, 603194 (3), Autosomal recessive
TMEM218	99.92 %	619285	Joubert syndrome 39, 619562 (3), Autosomal recessive
TMEM231	93.22 %	614949	Joubert syndrome 20, 614970 (3), Autosomal recessive; Meckel syndrome 11, 615397 (3), Autosomal recessive
TMEM237	99.12 %	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
TMEM260	99.88 %	617449	Structural heart defects and renal anomalies syndrome, 617478 (3), Autosomal recessive
TMEM67	99.79 %	609884	Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive; Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYNS syndrome, 602152 (3), Autosomal recessive; COACH syndrome 1, 216360 (3), Autosomal recessive
TOGARAM1	99.92 %	617618	Joubert syndrome 37, 619185 (3), Autosomal recessive
TOPORS	99.88 %	609507	Retinitis pigmentosa 31, 609923 (3), Autosomal dominant
TRAF3IP1	99.94 %	607380	Senior-Loken syndrome 9, 616629 (3), Autosomal recessive
TRIM32	99.99 %	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
TTC21B	99.13 %	612014	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive; Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant
TTC23	99.78 %		*No OMIM phenotypes*
TTC26	99.82 %	617453	Biliary, renal, neurologic, and skeletal syndrome, 619534 (3), Autosomal recessive
TTC6	99.77 %	620954	*No OMIM phenotypes*
TTC8	99.76 %	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
TUBGCP4	99.8 %	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
TULP1	99.84 %	602280	Leber congenital amaurosis 15, 613843 (3), Autosomal recessive; Retinitis pigmentosa 14, 600132 (3), Autosomal recessive
TULP3	99.37 %	604730	Hepatorenocardiac degenerative fibrosis, 619902 (3), Autosomal recessive
TXNDC15	99.86 %	617778	Meckel syndrome 14, 619879 (3), Autosomal recessive
VHL	99.98 %	608537	Hemangioblastoma, cerebellar, somatic (3); Erythrocytosis, familial, 2, 263400 (3), Autosomal recessive; von Hippel-Lindau syndrome, 193300 (3), Autosomal dominant; Renal cell carcinoma, somatic, 144700 (3); Pheochromocytoma, 171300 (3), Autosomal dominant
VPS13B	99.81 %	607817	Cohen syndrome, 216550 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
WDPCP	99.79 %	613580	Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
WDR19	99.62 %	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive
WDR35	99.87 %	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
XPNPEP3	99.89 %	613553	Nephronophthisis-like nephropathy 1, 613159 (3), Autosomal recessive
ZFYVE19	99.85 %	619635	Cholestasis, progressive familial intrahepatic, 9, 619849 (3), Autosomal recessive
ZIC3	99.87 %	300265	Congenital heart defects, nonsyndromic, multiple types, 1, X-linked, 306955 (3), X-linked recessive; Heterotaxy, visceral, 1, X-linked, 306955 (3), X-linked recessive; VACTERL association, X-linked, 314390 (3), X-linked recessive
ZMYND10	99.88 %	607070	Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive
ZNF423	99.92 %	604557	Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant; Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant
ZSWIM6	99.51 %	615951	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 (3), Autosomal dominant; Acromelic frontonasal dysostosis, 603671 (3), Autosomal dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.