Heterotaxie PCD

Gene panel

Gene panel information

Gene panel	**Heterotaxie PCD**
Version	4
Total genes	168
Activation date	Thursday 24 april 2025
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ACTC1	98.57 %	102540	Left ventricular noncompaction 4, 613424 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 11, 612098 (3), Autosomal dominant; Atrial septal defect 5, 612794 (3), Autosomal dominant; Cardiomyopathy, dilated, 1R, 613424 (3), Autosomal dominant
ACVR2B	99.99 %	602730	Heterotaxy, visceral, 4, autosomal, 613751 (3)
AK7	99.93 %	615364	?Spermatogenic failure 27, 617965 (3), Autosomal recessive
ALMS1	99.9 %	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
ANKS6	100 %	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
ARL2BP	99.89 %	615407	Retinitis pigmentosa 82 with or without situs inversus, 615434 (3), Autosomal recessive
ARMC2	99.97 %	618424	Spermatogenic failure 38, 618433 (3), Autosomal recessive
BBS1	100 %	209901	Bardet-Biedl syndrome 1, 209900 (3), Digenic recessive, Autosomal recessive
BBS10	99.98 %	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
BBS2	99.9 %	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive
BCL9L	99.99 %	609004	*No OMIM phenotypes*
BCOR	99.97 %	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BRAF	99.78 %	164757	Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3)
BRWD1	99.88 %	617824	Ciliary dyskinesia, primary, 51, 620438 (3), Autosomal recessive
C1orf127	99.98 %	619700	*No OMIM phenotypes*
CBL	99.95 %	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant
CCDC103	99.68 %	614677	Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive
CCDC39	99.74 %	613798	Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive
CCDC40	100 %	613799	Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive
CCDC65	99.8 %	611088	Ciliary dyskinesia, primary, 27, 615504 (3), Autosomal recessive
CCNO	100 %	607752	Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive
CENPF	99.97 %	600236	Stromme syndrome, 243605 (3), Autosomal recessive
CEP164	99.99 %	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive

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CEP290	98.1 %	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
CFAP221	99.71 %	618704	*No OMIM phenotypes*
CFAP298	99.96 %	615494	Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive
CFAP300	99.47 %	618058	Ciliary dyskinesia, primary, 38, 618063 (3), Autosomal recessive
CFAP43	99.88 %	617558	Hydrocephalus, normal pressure, 1, 236690 (3), Autosomal dominant; Spermatogenic failure 19, 617592 (3), Autosomal recessive
CFAP45	99.97 %	605152	Heterotaxy, visceral, 11, autosomal, with male infertility, 619608 (3), Autosomal recessive
CFAP46	99.99 %	618543	*No OMIM phenotypes*
CFAP47	99.21 %	301057	Spermatogenic failure, X-linked 3, 301059 (3), X-linked recessive
CFAP52	99.92 %	609804	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607 (3), Autosomal recessive
CFAP53	99.95 %	614759	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive
CFAP54	99.07 %		*No OMIM phenotypes*
CFAP57	98.9 %	614259	Spermatogenic failure 95, 620917 (3), Autosomal recessive
CFAP58	99.96 %	619129	Spermatogenic failure 49, 619144 (3), Autosomal recessive
CFAP65	99.99 %	614270	Spermatogenic failure 40, 618664 (3), Autosomal recessive
CFAP74	99.98 %	620187	Ciliary dyskinesia, primary, 49, without situs inversus, 620197 (3), Autosomal recessive
CFC1	21.93 %	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant
CFTR	99.45 %	602421	Cystic fibrosis, 219700 (3), Autosomal recessive; Sweat chloride elevation without CF (3); Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; {Hypertrypsinemia, neonatal} (3)
CHD7	99.99 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CITED2	100 %	602937	Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant
CRELD1	99.99 %	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; Jeffries-Lakhani neurodevelopmental syndrome, 620771 (3), Autosomal recessive; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant
DAND5	100 %	609068	*No OMIM phenotypes*
DAW1	99.9 %	620279	Ciliary dyskinesia, primary, 52, 620570 (3), Autosomal recessive
DGAT2L6	99.97 %	300926	*No OMIM phenotypes*
DNAAF1	99.99 %	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
DNAAF11	99.8 %	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
DNAAF2	99.91 %	612517	Ciliary dyskinesia, primary, 10, 612518 (3), Autosomal recessive
DNAAF3	99.99 %	614566	Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive
DNAAF4	99.78 %	608706	{Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant; Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive
DNAAF5	99.99 %	614864	Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive
DNAAF6	98.73 %	300933	Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive

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DNAH1	99.98 %	603332	Spermatogenic failure 18, 617576 (3), Autosomal recessive; Ciliary dyskinesia, primary, 37, 617577 (3), Autosomal recessive
DNAH10	99.93 %	605884	Spermatogenic failure 56, 619515 (3), Autosomal recessive
DNAH11	99.93 %	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
DNAH12	99.36 %	603340	*No OMIM phenotypes*
DNAH14	99.64 %	603341	*No OMIM phenotypes*
DNAH17	99.98 %	610063	Spermatogenic failure 39, 618643 (3), Autosomal recessive
DNAH5	99.98 %	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3), Autosomal recessive
DNAH6	99.59 %	603336	*No OMIM phenotypes*
DNAH7	99.66 %	610061	Ciliary dyskinesia, primary, 50, 620356 (3), Autosomal recessive
DNAH8	99.84 %	603337	Spermatogenic failure 46, 619095 (3), Autosomal recessive
DNAH9	99.99 %	603330	Ciliary dyskinesia, primary, 40, 618300 (3), Autosomal recessive
DNAI1	99.92 %	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
DNAI2	99.86 %	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3), Autosomal recessive
DNAJB13	99.91 %	610263	Ciliary dyskinesia, primary, 34, 617091 (3), Autosomal recessive
DNAL1	99.79 %	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
DRC1	99.93 %	615288	Spermatogenic failure 80, 620222 (3), Autosomal recessive; Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive
DYNC1H1	99.99 %	600112	Charcot-Marie-Tooth disease, axonal, type 2O, 614228 (3), Autosomal dominant; Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 13, 614563 (3), Autosomal dominant
DYNLT2	99.95 %	186977	*No OMIM phenotypes*
EFCAB1	99.96 %	619564	Ciliary dyskinesia, primary, 53, 620642 (3), Autosomal recessive
ELN	99.86 %	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
EVC	99.95 %	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	99.97 %	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
FOXF1	99.99 %	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
FOXH1	100 %	603621	*No OMIM phenotypes*
FOXJ1	100 %	602291	Ciliary dyskinesia, primary, 43, 618699 (3), Autosomal dominant
GAS2L2	100 %	611398	?Ciliary dyskinesia, primary, 41, 618449 (3), Autosomal recessive
GAS8	100 %	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
GATA4	99.99 %	600576	Tetralogy of Fallot, 187500 (3), Autosomal dominant; Atrial septal defect 2, 607941 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GATA6	99.9 %	601656	Atrial septal defect 9, 614475 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
GDF1	100 %	602880	Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant; Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive
GJA1	100 %	121014	Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
GPC3	99.6 %	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
HAND1	99.99 %	602406	*No OMIM phenotypes*
HES7	100 %	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive
HYDIN	81.28 %	610812	Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive
IFT46	99.58 %	620506	*No OMIM phenotypes*
INVS	99.94 %	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
JAG1	100 %	601920	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2HH, 619574 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
KIF7	100 %	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
LEFTY1	99.98 %	603037	*No OMIM phenotypes*
LEFTY2	100 %	601877	*No OMIM phenotypes*
LMBRD1	99.67 %	612625	Methylmalonic aciduria and homocystinuria, cblF type, 277380 (3), Autosomal recessive
LMLN2	91.91 %	619703	Heterotaxy, visceral, 12, autosomal, 619702 (3), Autosomal recessive
LMNA	99.96 %	150330	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant
LRRC56	99.98 %	618227	Ciliary dyskinesia, primary, 39, 618254 (3), Autosomal recessive
LZTFL1	100 %	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
MAP2K1	99.98 %	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
MAP2K2	99.99 %	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
MCIDAS	100 %	614086	Ciliary dyskinesia, primary, 42, 618695 (3), Autosomal recessive
MED13L	99.99 %	608771	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MEGF8	99.9 %	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
MEIS2	99.97 %	601740	Cleft palate, cardiac defects, and impaired intellectual development, 600987 (3), Autosomal dominant
MKS1	99.92 %	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
MMP21	99.99 %	608416	Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive
MNS1	99.86 %	610766	Heterotaxy, visceral, 9, autosomal, with male infertility, 618948 (3), Autosomal recessive
MRE11	99.93 %	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
MYH6	100 %	160710	{Sick sinus syndrome 3}, 614090 (3); Atrial septal defect 3, 614089 (3); Cardiomyopathy, dilated, 1EE, 613252 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 14, 613251 (3), Autosomal dominant
NAT10	99.97 %	609221	*No OMIM phenotypes*
NEK10	96.92 %	618726	Ciliary dyskinesia, primary, 44, 618781 (3), Autosomal recessive
NEK3	99.96 %	604044	*No OMIM phenotypes*
NEK8	99.99 %	609799	Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; Polycystic kidney disease 8, 620903 (3); ?Nephronophthisis 9, 613824 (3)
NKX2-5	99.75 %	600584	Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Ventricular septal defect 3, 614432 (3), Autosomal dominant; Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant
NKX2-6	100 %	611770	Persistent truncus arteriosus, 217095 (3); Conotruncal heart malformations, 217095 (3)
NME5	99.81 %	603575	Ciliary dyskinesia, primary, 48, without situs inversus, 620032 (3), Autosomal recessive
NME8	99.83 %	607421	?Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive
NODAL	99.98 %	601265	Heterotaxy, visceral, 5, 270100 (3), Autosomal dominant
NOTCH1	99.98 %	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
NOTCH2	99.03 %	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
NPHP3	99.89 %	608002	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
NR2F2	100 %	107773	46XX sex reversal 5, 618901 (3), Autosomal dominant; Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
NSD1	99.98 %	606681	Sotos syndrome, 117550 (3), Autosomal dominant
ODAD1	96.04 %	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
ODAD2	98.19 %	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
ODAD3	99.96 %	615956	Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive
ODAD4	99.85 %	617095	Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive
OFD1	99.68 %	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

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PIK3CD	99.99 %	602839	Immunodeficiency 14A, autosomal dominant, 615513 (3), Autosomal dominant; Immunodeficiency 14B, autosomal recessive, 619281 (3), Autosomal recessive; ?Roifman-Chitayat syndrome, digenic, 613328 (3), Digenic recessive
PITRM1	99.89 %	618211	Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive
PKD1L1	99.84 %	609721	Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive
PKD2	99.91 %	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
PQBP1	99.99 %	300463	Renpenning syndrome, 309500 (3), X-linked recessive
PRRX1	99.56 %	167420	Agnathia-otocephaly complex, 202650 (3), Autosomal dominant, Autosomal recessive
PTPN11	99.98 %	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
RAF1	99.97 %	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3), Autosomal dominant
RIT1	99.78 %	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RPGR	94.45 %	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
RSPH1	99.87 %	609314	Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive
RSPH3	99.94 %	615876	Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive
RSPH4A	99.95 %	612647	Ciliary dyskinesia, primary, 11, 612649 (3), Autosomal recessive
RSPH9	99.99 %	612648	Ciliary dyskinesia, primary, 12, 612650 (3), Autosomal recessive
SERPINE2	99.97 %	177010	*No OMIM phenotypes*
SHOC2	99.96 %	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
SHROOM3	99.99 %	604570	*No OMIM phenotypes*
SMAD2	99.92 %	601366	Loeys-Dietz syndrome 6, 619656 (3), Autosomal dominant; Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657 (3), Autosomal dominant
SMAD6	100 %	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Radioulnar synostosis, nonsyndromic}, 179300 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
SOS1	99.68 %	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
SPAG1	99.78 %	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive
SPEF2	99.93 %	610172	Spermatogenic failure 43, 618751 (3), Autosomal recessive
STK36	99.98 %	607652	?Ciliary dyskinesia, primary, 46, 619436 (3), Autosomal recessive
TBX1	99.95 %	602054	Tetralogy of Fallot, 187500 (3), Autosomal dominant; DiGeorge syndrome, 188400 (3), Autosomal dominant; Conotruncal anomaly face syndrome, 217095 (3); Velocardiofacial syndrome, 192430 (3), Autosomal dominant
TBX5	99.98 %	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
TCTN2	99.99 %	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
TP73	100 %	601990	Ciliary dyskinesia, primary, 47, and lissencephaly, 619466 (3), Autosomal recessive
TTC12	99.97 %	610732	Ciliary dyskinesia, primary, 45, 618801 (3), Autosomal recessive
TTC8	99.67 %	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TUBB4B	100 %	602660	Leber congenital amaurosis with early-onset deafness, 617879 (3), Autosomal dominant
UBR1	99.93 %	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
WDR35	99.92 %	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
WDR47	95.08 %	615734	*No OMIM phenotypes*
WFDC2	100 %	617548	*No OMIM phenotypes*
ZFPM2	100 %	603693	Diaphragmatic hernia 3, 610187 (3); 46XY sex reversal 9, 616067 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
ZIC3	99.9 %	300265	Congenital heart defects, nonsyndromic, 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
ZMPSTE24	98.7 %	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy 1, 275210 (3), Autosomal recessive
ZMYND10	99.99 %	607070	Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive
ZNF423	98.94 %	604557	Nephronophthisis 14, 614844 (3), Autosomal dominant, Autosomal recessive; Joubert syndrome 19, 614844 (3), Autosomal dominant, Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

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.