

Heterotaxie PCD (Primaire ciliaire dyskinesie) panel		
versie	V1 (92 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ACTC1</i>	102540	Atrial septal defect 5, 612794 (3), Autosomal dominant; Cardiomyopathy, dilated, 1R, 613424 (3), Autosomal dominant; Cardiomyopathy, hypertrophic, 11, 612098 (3), Autosomal dominant; Left ventricular noncompaction 4, 613424 (3), Autosomal dominant
<i>ACVR2B</i>	602730	Heterotaxy, visceral, 4, autosomal, 613751 (3)
<i>AK7</i>	615364	No OMIM phenotype
<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>ANKS6</i>	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
<i>ARMC4</i>	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
<i>BBS10</i>	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
<i>BCL9L</i>	609004	No OMIM phenotype
<i>BCOR</i>	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
<i>BRAF</i>	164757	Adenocarcinoma of lung, somatic, 211980 (3); Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Colorectal cancer, somatic (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Melanoma, malignant, somatic (3); Non-small cell lung cancer, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant
<i>C21orf59</i>	615494	Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive
<i>CBL</i>	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
<i>CCDC103</i>	614677	Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive
<i>CCDC114</i>	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
<i>CCDC151</i>	615956	Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive
<i>CCDC39</i>	613798	Ciliary dyskinesia, primary, 14, 613807 (3)
<i>CCDC40</i>	613799	Ciliary dyskinesia, primary, 15, 613808 (3)
<i>CCDC65</i>	611088	Ciliary dyskinesia, primary, 27, 615504 (3), Autosomal recessive
<i>CCNO</i>	607752	Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive
<i>CENPF</i>	600236	Stromme syndrome, 243605 (3), Autosomal recessive
<i>CEP290</i>	610142	?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Meckel syndrome 4, 611134 (3), Autosomal recessive; Senior-Loken syndrome 6, 610189 (3), Autosomal recessive
<i>CFAP53 (CCDC11)</i>	614759	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive
<i>CFC1</i>	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant

<i>CHD7</i>	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
<i>CITED2</i>	602937	Atrial septal defect 8, 614433 (3), Autosomal dominant; Ventricular septal defect 2, 614431 (3), Autosomal dominant
<i>CRELD1</i>	607170	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3), Autosomal dominant; {Atrioventricular septal defect, susceptibility to, 2}, 606217 (3), Autosomal dominant
<i>DNAAF1</i>	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
<i>DNAAF2</i>	612517	Ciliary dyskinesia, primary, 10, 612518 (3)
<i>DNAAF3</i>	614566	Ciliary dyskinesia, primary, 2, 606763 (3), Autosomal recessive
<i>DNAAF4 (DYX1C1)</i>	608706	Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive; {Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant
<i>DNAAF5 (HEATR2)</i>	614864	Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive
<i>DNAH1</i>	603332	?Ciliary dyskinesia, primary, 37, 617577 (3), Autosomal recessive; Spermatogenic failure 18, 617576 (3), Autosomal recessive
<i>DNAH11</i>	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
<i>DNAH5</i>	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3)
<i>DNAH8</i>	603337	No OMIM phenotype
<i>DNAI1</i>	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
<i>DNAI2</i>	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3)
<i>DNAJB13</i>	610263	Ciliary dyskinesia, primary, 34, 617091 (3), Autosomal recessive
<i>DNAL1</i>	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
<i>DRC1</i>	615288	Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive
<i>ELN</i>	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
<i>FOXH1</i>	603621	No OMIM phenotype
<i>GAS8</i>	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
<i>GATA4</i>	600576	Atrial septal defect 2, 607941 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant
<i>GATA6</i>	601656	Atrial septal defect 9, 614475 (3), Autosomal dominant; Atrioventricular septal defect 5, 614474 (3), Autosomal dominant; Pancreatic agenesis and congenital heart defects, 600001 (3), Autosomal dominant; Persistent truncus arteriosus, 217095 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant

<i>GDF1</i>	602880	Double-outlet right ventricle, 217095 (3); Right atrial isomerism, 208530 (3), Autosomal recessive; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Transposition of great arteries, dextro-looped 3, 613854 (3), Autosomal dominant
<i>GJA1</i>	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratoderma variabilis et progressiva 3, 617525 (3); Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant
<i>GPC3</i>	300037	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive; Wilms tumor, somatic, 194070 (3)
<i>HYDIN</i>	610812	Ciliary dyskinesia, primary, 5, 608647 (3), Autosomal recessive
<i>INVS</i>	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
<i>JAG1</i>	601920	Alagille syndrome 1, 118450 (3), Autosomal dominant; ?Deafness, congenital heart defects, and posterior embryotoxon (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant
<i>LEFTY2</i>	601877	Left-right axis malformations (3)
<i>LRRC6</i>	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
<i>MAP2K1</i>	176872	Cardiofaciocutaneous syndrome 3, 615279 (3)
<i>MAP2K2</i>	601263	Cardiofaciocutaneous syndrome 4, 615280 (3)
<i>MCIDAS</i>	614086	No OMIM phenotype
<i>MED13L</i>	608771	Mental retardation and distinctive facial features with or without cardiac defects, 616789 (3), Autosomal dominant; Transposition of the great arteries, dextro-looped 1, 608808 (3), Autosomal dominant
<i>MEIS2</i>	601740	Cleft palate, cardiac defects, and mental retardation, 600987 (3), Autosomal dominant
<i>MKS1</i>	609883	Bardet-Biedl syndrome 13, 615990 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive; Meckel syndrome 1, 249000 (3), Autosomal recessive
<i>NAT10</i>	609221	No OMIM phenotype
<i>NEK8</i>	609799	?Nephronophthisis 9, 613824 (3); ?Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive
<i>NKX2-5</i>	600584	Atrial septal defect 7, with or without AV conduction defects, 108900 (3), Autosomal dominant; Conotruncal heart malformations, variable, 217095 (3); Hypoplastic left heart syndrome 2, 614435 (3), Autosomal dominant; Hypothyroidism, congenital nongoitrous, 5, 225250 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Ventricular septal defect 3, 614432 (3), Autosomal dominant

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<i>NKX2-6</i>	611770	Conotruncal heart malformations, 217095 (3); Persistent truncus arteriosus, 217095 (3)
<i>NME8</i>	607421	Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive
<i>NODAL</i>	601265	Heterotaxy, visceral, 5, 270100 (3), Autosomal dominant
<i>NOTCH1</i>	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
<i>NOTCH2</i>	600275	Alagille syndrome 2, 610205 (3), Autosomal dominant; Hajdu-Cheney syndrome, 102500 (3), Autosomal dominant
<i>NPHP3</i>	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive
<i>NR2F2</i>	107773	Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
<i>NSD1</i>	606681	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; Leukemia, acute myeloid, 601626 (1), Autosomal dominant; Sotos syndrome 1, 117550 (3), Autosomal dominant
<i>OFD1</i>	300170	Joubert syndrome 10, 300804 (3), X-linked recessive; Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
<i>PIH1D3</i>	300933	Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive
<i>PTPN11</i>	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3); Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant
<i>RAF1</i>	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3)
<i>RIT1</i>	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
<i>RPGR</i>	312610	Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3); Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3)
<i>RSPH1</i>	609314	Ciliary dyskinesia, primary, 24, 615481 (3), Autosomal recessive
<i>RSPH3</i>	615876	Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive
<i>RSPH4A</i>	612647	Ciliary dyskinesia, primary, 11, 612649 (3)
<i>RSPH9</i>	612648	Ciliary dyskinesia, primary, 12, 612650 (3)
<i>SHOC2</i>	602775	Noonan-like syndrome with loose anagen hair, 607721 (3), Autosomal dominant
<i>SHROOM3</i>	604570	No OMIM phenotype
<i>SMAD2</i>	601366	No OMIM phenotype
<i>SOS1</i>	182530	?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant; Noonan syndrome 4, 610733 (3), Autosomal dominant
<i>SPAG1</i>	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive

<i>TBX1</i>	602054	Conotruncal anomaly face syndrome, 217095 (3); DiGeorge syndrome, 188400 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Velocardiofacial syndrome, 192430 (3), Autosomal dominant
<i>TBX5</i>	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
<i>TTC25</i>	617095	Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive
<i>TTC8</i>	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
<i>ZIC3</i>	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
<i>ZMYND10</i>	607070	Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive
<i>ZNF423</i>	604557	Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant; Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.

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

OMIM release used for OMIM disease identifiers and descriptions: June 06, 2017

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.