

Heterotaxie PCD (Primaire ciliaire dyskinesie) panel		
versie	v2 (125 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	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
<i>ACVR2B</i>	602730	Heterotaxy, visceral, 4, autosomal, 613751 (3)
<i>AK7</i>	615364	?Spermatogenic failure 27, 617965 (3), Autosomal recessive
<i>ALMS1</i>	606844	Alstrom syndrome, 203800 (3), Autosomal recessive
<i>ANKS6</i>	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
<i>BBS1</i>	209901	Bardet-Biedl syndrome 1, 209900 (3), Autosomal recessive, Digenic recessive
<i>BBS10</i>	610148	Bardet-Biedl syndrome 10, 615987 (3), Autosomal recessive
<i>BBS2</i>	606151	Retinitis pigmentosa 74, 616562 (3), Autosomal recessive; Bardet-Biedl syndrome 2, 615981 (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	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); Non-small cell lung cancer, somatic, 211980 (3)
<i>CBL</i>	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant
<i>CCDC103</i>	614677	Ciliary dyskinesia, primary, 17, 614679 (3), Autosomal recessive
<i>CCDC39</i>	613798	Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive
<i>CCDC40</i>	613799	Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive
<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>CEP164</i>	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive
<i>CEP290</i>	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
<i>CFAP298</i>	615494	Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive

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<i>CFAP300</i>	618058	Ciliary dyskinesia, primary, 38, 618063 (3), Autosomal recessive
<i>CFAP52</i>	609804	Heterotaxy, visceral, 10, autosomal, with male infertility, 619607 (3), Autosomal recessive
<i>CFAP53</i>	614759	Heterotaxy, visceral, 6, autosomal recessive, 614779 (3), Autosomal recessive
<i>CFC1</i>	605194	Heterotaxy, visceral, 2, autosomal, 605376 (3), Autosomal dominant
<i>CFTR</i>	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)
<i>CHD7</i>	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (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>DNAAF11</i>	614930	Ciliary dyskinesia, primary, 19, 614935 (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</i>	608706	{Dyslexia, susceptibility to, 1}, 127700 (3), Autosomal dominant; Ciliary dyskinesia, primary, 25, 615482 (3), Autosomal recessive
<i>DNAAF5</i>	614864	Ciliary dyskinesia, primary, 18, 614874 (3), Autosomal recessive
<i>DNAAF6</i>	300933	Ciliary dyskinesia, primary, 36, X-linked, 300991 (3), X-linked recessive
<i>DNAH1</i>	603332	Spermatogenic failure 18, 617576 (3), Autosomal recessive; ?Ciliary dyskinesia, primary, 37, 617577 (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	Spermatogenic failure 46, 619095 (3), Autosomal recessive
<i>DNAH9</i>	603330	Ciliary dyskinesia, primary, 40, 618300 (3), Autosomal recessive
<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), Autosomal recessive
<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

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<i>ELN</i>	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
<i>EVC</i>	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EVC2</i>	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>FOXF1</i>	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
<i>FOXH1</i>	603621	No OMIM phenotype
<i>GAS2L2</i>	611398	?Ciliary dyskinesia, primary, 41, 618449 (3), Autosomal recessive
<i>GAS8</i>	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
<i>GATA4</i>	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
<i>GATA6</i>	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
<i>GDF1</i>	602880	Congenital heart defects, multiple types, 6, 613854 (3), Autosomal dominant; Right atrial isomerism (Ivemark), 208530 (3), Autosomal recessive
<i>GJA1</i>	121014	Erythrokeratoderma variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Atrioventricular septal defect 3, 600309 (3), Autosomal dominant
<i>GPC3</i>	300037	Wilms tumor, somatic, 194070 (3); Simpson-Golabi-Behmel syndrome, type 1, 312870 (3), X-linked recessive
<i>HAND1</i>	602406	No OMIM phenotype
<i>HES7</i>	608059	Spondylocostal dysostosis 4, autosomal recessive, 613686 (3), Autosomal recessive
<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	?Deafness, congenital heart defects, and posterior embryotoxon, 617992 (3), Autosomal dominant; Alagille syndrome 1, 118450 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant

<i>KIF7</i>	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
<i>LEFTY2</i>	601877	No OMIM phenotype
<i>LMNA</i>	150330	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; 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
<i>LRRC56</i>	618227	Ciliary dyskinesia, primary, 39, 618254 (3), Autosomal recessive
<i>LZTFL1</i>	606568	Bardet-Biedl syndrome 17, 615994 (3), Autosomal recessive
<i>MAP2K1</i>	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
<i>MAP2K2</i>	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
<i>MCIDAS</i>	614086	Ciliary dyskinesia, primary, 42, 618695 (3), Autosomal recessive
<i>MED13L</i>	608771	Impaired intellectual development 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>MEGF8</i>	604267	Carpenter syndrome 2, 614976 (3), Autosomal recessive
<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; Meckel syndrome 1, 249000 (3), Autosomal recessive; Joubert syndrome 28, 617121 (3), Autosomal recessive
<i>MMP21</i>	608416	Heterotaxy, visceral, 7, autosomal, 616749 (3), Autosomal recessive
<i>MRE11</i>	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive
<i>MYH6</i>	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
<i>NAT10</i>	609221	No OMIM phenotype
<i>NEK8</i>	609799	Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive; ?Nephronophthisis 9, 613824 (3)

<i>NKX2-5</i>	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
<i>NKX2-6</i>	611770	Persistent truncus arteriosus, 217095 (3); Conotruncal heart malformations, 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	Nephronophthisis 3, 604387 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Meckel syndrome 7, 267010 (3), Autosomal recessive
<i>NR2F2</i>	107773	46,XX sex reversal 5, 618901 (3), Autosomal dominant; Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
<i>NSD1</i>	606681	Sotos syndrome 1, 117550 (3), Autosomal dominant
<i>ODAD1</i>	615038	Ciliary dyskinesia, primary, 20, 615067 (3), Autosomal recessive
<i>ODAD2</i>	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
<i>ODAD3</i>	615956	Ciliary dyskinesia, primary, 30, 616037 (3), Autosomal recessive
<i>ODAD4</i>	617095	Ciliary dyskinesia, primary, 35, 617092 (3), Autosomal recessive
<i>OFD1</i>	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
<i>PITRM1</i>	618211	Spinocerebellar ataxia, autosomal recessive 30, 619405 (3), Autosomal recessive
<i>PKD1L1</i>	609721	Heterotaxy, visceral, 8, autosomal, 617205 (3), Autosomal recessive
<i>PKD2</i>	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
<i>PQBP1</i>	300463	Renpenning syndrome, 309500 (3), X-linked recessive
<i>PRRX1</i>	167420	Agnathia-otocephaly complex, 202650 (3), Autosomal recessive, Autosomal dominant
<i>PTPN11</i>	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)
<i>RAF1</i>	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3)
<i>RIT1</i>	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant

<i>RPGR</i>	312610	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3); Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Retinitis pigmentosa 3, 300029 (3); Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
<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 syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
<i>SHROOM3</i>	604570	No OMIM phenotype
<i>SMAD2</i>	601366	Congenital heart defects, multiple types, 8, with or without heterotaxy, 619657 (3), Autosomal dominant
<i>SMAD6</i>	602931	Aortic valve disease 2, 614823 (3), Autosomal dominant; {Radioulnar synostosis, nonsyndromic}, 179300 (3), Autosomal dominant; {Craniosynostosis 7, susceptibility to}, 617439 (3), Autosomal dominant
<i>SOS1</i>	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; ?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
<i>SPAG1</i>	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive
<i>STK36</i>	607652	?Ciliary dyskinesia, primary, 46, 619436 (3), Autosomal recessive
<i>TBX1</i>	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
<i>TBX5</i>	601620	Holt-Oram syndrome, 142900 (3), Autosomal dominant
<i>TCTN2</i>	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
<i>TTC8</i>	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
<i>UBR1</i>	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
<i>WDR35</i>	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
<i>ZFPM2</i>	603693	Diaphragmatic hernia 3, 610187 (3); 46XY sex reversal 9, 616067 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
<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>ZMPSTE24</i>	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
<i>ZMYND10</i>	607070	Ciliary dyskinesia, primary, 22, 615444 (3), Autosomal recessive

<i>ZNF423</i>	604557	Nephronophthisis 14, 614844 (3), Autosomal recessive, Autosomal dominant; Joubert syndrome 19, 614844 (3), Autosomal recessive, Autosomal dominant
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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.