

<b>CilioPathy panel</b>		
<b>versie</b>	v3 (120 genen)	Centrum voor Medische Genetica Gent
<b>Gene</b>	<b>OMIM gene ID</b>	<b>Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern</b>
<i>AHI1</i>	608894	Joubert syndrome 3, 608629 (3), Autosomal recessive
<i>ANKS6</i>	615370	Nephronophthisis 16, 615382 (3), Autosomal recessive
<i>ARL13B</i>	608922	Joubert syndrome 8, 612291 (3), Autosomal recessive
<i>ARL6</i>	608845	?Retinitis pigmentosa 55, 613575 (3); Bardet-Biedl syndrome 3, 600151 (3), Autosomal recessive; {Bardet-Biedl syndrome 1, modifier of}, 209900 (3), Autosomal recessive, Digenic recessive
<i>ARMC4</i>	615408	Ciliary dyskinesia, primary, 23, 615451 (3), Autosomal recessive
<i>B9D1</i>	614144	?Meckel syndrome 9, 614209 (3), Autosomal recessive; Joubert syndrome 27, 617120 (3), Autosomal recessive
<i>B9D2</i>	611951	Joubert syndrome 34, 614175 (3), Autosomal recessive; ?Meckel syndrome 10, 614175 (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>BBS12</i>	610683	Bardet-Biedl syndrome 12, 615989 (3), Autosomal recessive
<i>BBS2</i>	606151	Bardet-Biedl syndrome 2, 615981 (3), Autosomal recessive; Retinitis pigmentosa 74, 616562 (3), Autosomal recessive
<i>BBS4</i>	600374	Bardet-Biedl syndrome 4, 615982 (3), Autosomal recessive
<i>BBS5</i>	603650	Bardet-Biedl syndrome 5, 615983 (3), Autosomal recessive
<i>BBS7</i>	607590	Bardet-Biedl syndrome 7, 615984 (3), Autosomal recessive
<i>BBS9</i>	607968	Bardet-Biedl syndrome 9, 615986 (3), Autosomal recessive
<i>CC2D2A</i>	612013	Meckel syndrome 6, 612284 (3), Autosomal recessive; Joubert syndrome 9, 612285 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive
<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>CCDC172</i>	No OMIM gene	No OMIM phenotype
<i>CCDC32</i>	618941	No OMIM phenotype
<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>CCDC96</i>	No OMIM gene	No OMIM phenotype
<i>CCNO</i>	607752	Ciliary dyskinesia, primary, 29, 615872 (3), Autosomal recessive
<i>CEP104</i>	616690	Joubert syndrome 25, 616781 (3), Autosomal recessive
<i>CEP120</i>	613446	Joubert syndrome 31, 617761 (3), Autosomal recessive; Short-rib thoracic dysplasia 13 with or without polydactyly, 616300 (3), Autosomal recessive

<i>CEP164</i>	614848	Nephronophthisis 15, 614845 (3), Autosomal recessive ?Bardet-Biedl syndrome 14, 615991 (3), Autosomal recessive; Leber congenital amaurosis 10, 611755 (3); Senior-Loken syndrome 6, 610189 (3), Autosomal recessive; Meckel syndrome 4, 611134 (3), Autosomal recessive; Joubert syndrome 5, 610188 (3), Autosomal recessive
<i>CEP290</i>	610142	
<i>CEP295</i>	617728	No OMIM phenotype
<i>CEP41</i>	610523	Joubert syndrome 15, 614464 (3), Autosomal recessive
<i>CEP83</i>	615847	Nephronophthisis 18, 615862 (3), Autosomal recessive
<i>CFAP298</i>	615494	Ciliary dyskinesia, primary, 26, 615500 (3), Autosomal recessive
<i>CILK1 (ICK)</i>	612325	Endocrine-cerebroostedysplasia, 612651 (3), Autosomal recessive; {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924 (3), Autosomal dominant
<i>CPLANE1</i>	614571	Joubert syndrome 17, 614615 (3), Autosomal recessive; Orofaciodigital syndrome VI, 277170 (3), Autosomal recessive
<i>CSPP1</i>	611654	Joubert syndrome 21, 615636 (3), Autosomal recessive
<i>DCDC2</i>	605755	Sclerosing cholangitis, neonatal, 617394 (3), Autosomal recessive; Nephronophthisis 19, 616217 (3), Autosomal recessive; ?Deafness, autosomal recessive 66, 610212 (3), Autosomal recessive
<i>DEUP1</i>	617148	No OMIM phenotype
<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</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>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>DNAL1</i>	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
<i>DRC1</i>	615288	Ciliary dyskinesia, primary, 21, 615294 (3), Autosomal recessive
<i>DYNC2H1</i>	603297	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091 (3), Autosomal recessive, Digenic recessive
<i>DYNC2I1 (WDR60)</i>	615462	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503 (3), Autosomal recessive
<i>DYNC2I2 (WDR34)</i>	613363	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633 (3), Autosomal recessive

<i>EVC</i>	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EVC2</i>	607261	Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant; Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive
<i>FAM166B</i>	No OMIM gene	No OMIM phenotype
<i>GAS8</i>	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
<i>GLIS2</i>	608539	Nephronophthisis 7, 611498 (3)
<i>IFT122</i>	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
<i>IFT140</i>	614620	Retinitis pigmentosa 80, 617781 (3), Autosomal recessive; Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 (3), Autosomal recessive
<i>IFT172</i>	607386	Retinitis pigmentosa 71, 616394 (3), Autosomal recessive; Short-rib thoracic dysplasia 10 with or without polydactyly, 615630 (3), Autosomal recessive
<i>IFT80</i>	611177	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263 (3), Autosomal recessive
<i>INPP5E</i>	613037	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156 (3), Autosomal recessive; Joubert syndrome 1, 213300 (3), Autosomal recessive
<i>INVS</i>	243305	Nephronophthisis 2, infantile, 602088 (3), Autosomal recessive
<i>IQCB1</i>	609237	Senior-Loken syndrome 5, 609254 (3), Autosomal recessive
<i>KATNIP</i> ( <i>KIAA0556</i> )	616650	Joubert syndrome 26, 616784 (3), Autosomal recessive
<i>KIAA0586</i>	610178	Joubert syndrome 23, 616490 (3), Autosomal recessive; Short-rib thoracic dysplasia 14 with polydactyly, 616546 (3), Autosomal recessive
<i>KIF14</i>	611279	Microcephaly 20, primary, autosomal recessive, 617914 (3), Autosomal recessive; ?Meckel syndrome 12, 616258 (3), Autosomal recessive
<i>KIF3B</i>	603754	Retinitis pigmentosa 89, 618955 (3), Autosomal dominant
<i>KIF7</i>	611254	?Hydrolethalus syndrome 2, 614120 (3), Autosomal recessive; Acrocallosal syndrome, 200990 (3), Autosomal recessive; Joubert syndrome 12, 200990 (3), Autosomal recessive; ?Al-Gazali-Bakalinova syndrome, 607131 (3), Autosomal recessive
<i>LRRC34</i>	No OMIM gene	No OMIM phenotype
<i>LRRC6</i>	614930	Ciliary dyskinesia, primary, 19, 614935 (3), Autosomal recessive
<i>MCIDAS</i>	614086	Ciliary dyskinesia, primary, 42, 618695 (3), Autosomal recessive
<i>MKKS</i>	604896	Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive; McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive
<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>MRE11</i>	600814	Ataxia-telangiectasia-like disorder 1, 604391 (3), Autosomal recessive

<i>NEK1</i>	604588	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant; Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive
<i>NEK8</i>	609799	?Nephronophthisis 9, 613824 (3); Renal-hepatic-pancreatic dysplasia 2, 615415 (3), Autosomal recessive
<i>NME5</i>	603575	No OMIM phenotype
<i>NME8</i>	607421	Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive
<i>NPHP1</i>	607100	Nephronophthisis 1, juvenile, 256100 (3), Autosomal recessive; Senior-Loken syndrome-1, 266900 (3), Autosomal recessive; Joubert syndrome 4, 609583 (3), Autosomal recessive
<i>NPHP3</i>	608002	Meckel syndrome 7, 267010 (3), Autosomal recessive; Renal-hepatic-pancreatic dysplasia 1, 208540 (3), Autosomal recessive; Nephronophthisis 3, 604387 (3), Autosomal recessive
<i>NPHP4</i>	607215	Nephronophthisis 4, 606966 (3), Autosomal recessive; Senior-Loken syndrome 4, 606996 (3), Autosomal recessive
<i>OFD1</i>	300170	Orofaciodigital syndrome I, 311200 (3), X-linked dominant; ?Retinitis pigmentosa 23, 300424 (3), X-linked recessive; Joubert syndrome 10, 300804 (3), X-linked recessive; Simpson-Golabi-Behmel syndrome, type 2, 300209 (3), X-linked recessive
<i>PDE6D</i>	602676	?Joubert syndrome 22, 615665 (3), Autosomal recessive
<i>PKD2</i>	173910	Polycystic kidney disease 2, 613095 (3), Autosomal dominant
<i>PKHD1</i>	606702	Polycystic kidney disease 4, with or without hepatic disease, 263200 (3), Autosomal recessive
<i>RAB28</i>	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
<i>RPGR</i>	312610	Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked; Retinitis pigmentosa 3, 300029 (3); Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3); Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
<i>RPGRIP1L</i>	610937	COACH syndrome, 216360 (3), Autosomal recessive; Meckel syndrome 5, 611561 (3), Autosomal recessive; Joubert syndrome 7, 611560 (3), Autosomal 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>SCLT1</i>	611399	No OMIM phenotype
<i>SDCCAG8</i>	613524	Bardet-Biedl syndrome 16, 615993 (3), Autosomal recessive; Senior-Loken syndrome 7, 613615 (3)
<i>SPAG1</i>	603395	Ciliary dyskinesia, primary, 28, 615505 (3), Autosomal recessive
<i>TCTN1</i>	609863	Joubert syndrome 13, 614173 (3), Autosomal recessive
<i>TCTN2</i>	613846	Joubert syndrome 24, 616654 (3), Autosomal recessive; ?Meckel syndrome 8, 613885 (3), Autosomal recessive
<i>TCTN3</i>	613847	Orofaciodigital syndrome IV, 258860 (3), Autosomal recessive; Joubert syndrome 18, 614815 (3), Autosomal recessive

<i>TMEM138</i>	614459	Joubert syndrome 16, 614465 (3), Autosomal recessive
<i>TMEM17</i>	614950	No OMIM phenotype
<i>TMEM216</i>	613277	Meckel syndrome 2, 603194 (3), Autosomal recessive; Joubert syndrome 2, 608091 (3), Autosomal recessive
<i>TMEM231</i>	614949	Meckel syndrome 11, 615397 (3), Autosomal recessive; Joubert syndrome 20, 614970 (3), Autosomal recessive
<i>TMEM237</i>	614423	Joubert syndrome 14, 614424 (3), Autosomal recessive
<i>TMEM67</i>	609884	Meckel syndrome 3, 607361 (3), Autosomal recessive; ?RHYSN syndrome, 602152 (3), Autosomal recessive; Nephronophthisis 11, 613550 (3), Autosomal recessive; {Bardet-Biedl syndrome 14, modifier of}, 615991 (3), Autosomal recessive; COACH syndrome, 216360 (3), Autosomal recessive; Joubert syndrome 6, 610688 (3), Autosomal recessive
<i>TRIM32</i>	602290	?Bardet-Biedl syndrome 11, 615988 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 (3), Autosomal recessive
<i>TTC21B</i>	612014	Nephronophthisis 12, 613820 (3), Autosomal recessive, Autosomal dominant; Short-rib thoracic dysplasia 4 with or without polydactyly, 613819 (3), Autosomal recessive
<i>TTC23</i>	No OMIM gene	No OMIM phenotype
<i>TTC6</i>	No OMIM gene	No OMIM phenotype
<i>TTC8</i>	608132	Bardet-Biedl syndrome 8, 615985 (3), Autosomal recessive; ?Retinitis pigmentosa 51, 613464 (3), Autosomal recessive
<i>TUBGCP4</i>	609610	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335 (3), Autosomal recessive
<i>WDPCP</i>	613580	?Bardet-Biedl syndrome 15, 615992 (3), Autosomal recessive; ?Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085 (3), Autosomal recessive
<i>WDR19</i>	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (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>XPNPEP3</i>	613553	Nephronophthisis-like nephropathy 1, 613159 (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

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: Sep 01, 2020

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.