

<b>Cataract panel (v2)</b>		
----------------------------	--	--

<b>versie</b>	2019-09-03 (65 genen)	Centrum voor Medische Genetica Gent
---------------	--------------------------	-------------------------------------

GitHub commit: 7093fc1bdca5578a08e987e2ccea0dd0eecec9e3

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>ABHD12</i>	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
<i>ADAMTSL4</i>	610113	Ectopia lentis et pupillae, 225200 (3), Autosomal recessive; Ectopia lentis, isolated, autosomal recessive, 225100 (3), Autosomal recessive
<i>AGK</i>	610345	Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive; Sengers syndrome, 212350 (3), Autosomal recessive
<i>BCOR</i>	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
<i>BEST1</i>	607854	Bestrophinopathy, autosomal recessive, 611809 (3); Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220 (3), Autosomal dominant; Retinitis pigmentosa, concentric, 613194 (3); Retinitis pigmentosa-50, 613194 (3); Vitreoretinopathopathy, 193220 (3), Autosomal dominant
<i>BFSP1</i>	603307	Cataract 33, multiple types, 611391 (3), Autosomal recessive, Autosomal dominant
<i>BFSP2</i>	603212	Cataract 12, multiple types, 611597 (3), Autosomal dominant
<i>CHMP4B</i>	610897	Cataract 31, multiple types, 605387 (3), Autosomal dominant
<i>COL18A1</i>	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive
<i>COL2A1</i>	120140	Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Vitreoretinopathy with phalangeal epiphyseal dysplasia (3)

<i>CRYAA</i>	123580	Cataract 9, multiple types, 604219 (3), Autosomal recessive, Autosomal dominant
<i>CRYAB</i>	123590	Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal recessive, Autosomal dominant; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive
<i>CRYBA1</i>	123610	Cataract 10, multiple types, 600881 (3), Autosomal dominant
<i>CRYBA2</i>	600836	?Cataract 42, 115900 (3), Autosomal dominant
<i>CRYBA4</i>	123631	Cataract 23, 610425 (3)
<i>CRYBB1</i>	600929	Cataract 17, multiple types, 611544 (3), Autosomal recessive, Autosomal dominant
<i>CRYBB2</i>	123620	Cataract 3, multiple types, 601547 (3), Autosomal dominant
<i>CRYBB3</i>	123630	Cataract 22, 609741 (3), Autosomal recessive, Autosomal dominant
<i>CRYGB</i>	123670	Cataract 39, multiple types, autosomal dominant, 615188 (3), Autosomal dominant
<i>CRYGC</i>	123680	Cataract 2, multiple types, 604307 (3), Autosomal dominant
<i>CRYGD</i>	123690	Cataract 4, multiple types, 115700 (3), Autosomal dominant
<i>CRYGS</i>	123730	Cataract 20, multiple types, 116100 (3), Autosomal dominant
<i>CTDP1</i>	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
<i>CYP27A1</i>	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
<i>EPG5</i>	615068	Vici syndrome, 242840 (3), Autosomal recessive
<i>EPHA2</i>	176946	Cataract 6, multiple types, 116600 (3), Autosomal dominant
<i>EYA1</i>	601653	Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; Branchiootoc syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant
<i>FAM126A</i>	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
<i>FBN1</i>	134797	Acromicric dysplasia, 102370 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; MASS syndrome, 604308 (3); Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant
<i>FOXE3</i>	601094	Anterior segment dysgenesis 2, multiple subtypes, 610256 (3), Autosomal recessive; {Aortic aneurysm, familial thoracic 11, susceptibility to}, 617349 (3), Autosomal dominant; Cataract 34, multiple types, 612968 (3)

<i>FTL</i>	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal recessive, Autosomal dominant; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant
<i>FYCO1</i>	607182	Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive
<i>FZD4</i>	604579	Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant; Retinopathy of prematurity, 133780 (3), Autosomal dominant
<i>GALK1</i>	604313	Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive
<i>GALT</i>	606999	Galactosemia, 230400 (3), Autosomal recessive
<i>GCNT2</i>	600429	Adult i phenotype without cataract, 110800 (3), Autosomal dominant; [Blood group, Ii], 110800 (3), Autosomal dominant; Cataract 13 with adult i phenotype, 116700 (3), Autosomal recessive
<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), Autosomal dominant; 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>GJA3</i>	121015	Cataract 14, multiple types, 601885 (3), Autosomal dominant
<i>GJA8</i>	600897	Cataract 1, multiple types, 116200 (3), Autosomal dominant
<i>HSF4</i>	602438	Cataract 5, multiple types, 116800 (3), Autosomal dominant
<i>INTS1</i>	611345	No OMIM phenotype
<i>LEMD2</i>	616312	Cataract 46, juvenile-onset, 212500 (3), Autosomal recessive
<i>LIM2</i>	154045	Cataract 19, multiple types, 615277 (3), Autosomal recessive
<i>LSS</i>	600909	Cataract 44, 616509 (3), Autosomal recessive
<i>MAF</i>	177075	Ayme-Gripp syndrome, 601088 (3), Autosomal dominant; Cataract 21, multiple types, 610202 (3), Autosomal dominant
<i>MIP</i>	154050	Cataract 15, multiple types, 615274 (3), Autosomal dominant
<i>MYH9</i>	160775	Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant; Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 (3), Autosomal dominant
<i>NF2</i>	607379	Meningioma, NF2-related, somatic, 607174 (3); Neurofibromatosis, type 2, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 162091 (3)
<i>NHS</i>	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
<i>OCRL</i>	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive

<i>OPA3</i>	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
<i>P3H2</i>	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive
<i>PANK4</i>	606162	No OMIM phenotype
<i>PAX6</i>	607108	Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3); Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Optic nerve hypoplasia, 165550 (3), Autosomal dominant
<i>PITX3</i>	602669	Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, multiple types, 610623 (3), Autosomal dominant; Cataract 11, syndromic, 610623 (3), Autosomal dominant
<i>RRAGA</i>	612194	No OMIM phenotype
<i>SIL1</i>	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
<i>SIPA1L3</i>	616655	?Cataract 45, 616851 (3), Autosomal recessive
<i>SLC16A12</i>	611910	Cataract 47, juvenile, with microcornea, 612018 (3), Autosomal dominant
<i>SLC33A1</i>	603690	Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive; Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant
<i>TDRD7</i>	611258	Cataract 36, 613887 (3), Autosomal recessive
<i>UNC45B</i>	611220	?Cataract 43, 616279 (3), Autosomal dominant
<i>VIM</i>	193060	Cataract 30, pulverulent, 116300 (3), Autosomal dominant
<i>VSX2</i>	142993	Microphthalmia with coloboma 3, 610092 (3); Microphthalmia, isolated 2, 610093 (3)
<i>WFS1</i>	606201	?Cataract 41, 116400 (3), Autosomal dominant; Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive; Wolfram-like syndrome, autosomal dominant, 614296 (3), 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: July 04, 2018

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.