Cataract panel (v2)

versie	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
ABHD12	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
ADAMTSL4	610113	Ectopia lentis et pupillae, 225200 (3), Autosomal recessive; Ectopia lentis, isolated, autosomal recessive, 225100 (3), Autosomal recessive
AGK	610345	Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive; Sengers syndrome, 212350 (3), Autosomal recessive
BCOR	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BEST1	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); Vitreoretinochoroidopathy, 193220 (3), Autosomal dominant
BFSP1	603307	Cataract 33, multiple types, 611391 (3), Autosomal recessive, Autosomal dominant
BFSP2	603212	Cataract 12, multiple types, 611597 (3), Autosomal dominant
CHMP4B	610897	Cataract 31, multiple types, 605387 (3), Autosomal dominant
COL18A1	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive
COL2A1	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 sydrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Vitreoretinopathy with phalangeal epiphyseal dysplasia (3)

CRYAA	123580	Cataract 9, multiple types, 604219 (3), Autosomal recessive, Autosomal dominant
CRYAB	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
CRYBA1	123610	Cataract 10, multiple types, 600881 (3), Autosomal dominant
CRYBA2	600836	?Cataract 42, 115900 (3), Autosomal dominant
CRYBA4	123631	Cataract 23, 610425 (3)
CRYBB1	600929	Cataract 17, multiple types, 611544 (3), Autosomal recessive, Autosomal dominant
CRYBB2	123620	Cataract 3, multiple types, 601547 (3), Autosomal dominant
CRYBB3	123630	Cataract 22, 609741 (3), Autosomal recessive, Autosomal dominant
CRYGB	123670	Cataract 39, multiple types, autosomal dominant, 615188 (3), Autosomal dominant
CRYGC	123680	Cataract 2, multiple types, 604307 (3), Autosomal dominant
CRYGD	123690	Cataract 4, multiple types, 115700 (3), Autosomal dominant
CRYGS	123730	Cataract 20, multiple types, 116100 (3), Autosomal dominant
CTDP1	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
CYP27A1	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
EPG5	615068	Vici syndrome, 242840 (3), Autosomal recessive
EPHA2	176946	Cataract 6, multiple types, 116600 (3), Autosomal dominant
EYA1	601653	Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant
FAM126A	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive
FBN1	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
FOXE3	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)

FTL	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
FYCO1	607182	Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive
FZD4	604579	Exudative vitreoretinopathy 1, 133780 (3), Autosomal dominant; Retinopathy of prematurity, 133780 (3), Autosomal dominant
GALK1	604313	Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive
GALT	606999	Galactosemia, 230400 (3), Autosomal recessive
GCNT2	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
GJA1	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratodermia 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
GJA3	121015	Cataract 14, multiple types, 601885 (3), Autosomal dominant
GJA8	600897	Cataract 1, multiple types, 116200 (3), Autosomal dominant
HSF4	602438	Cataract 5, multiple types, 116800 (3), Autosomal dominant
INTS1	611345	No OMIM phenotype
LEMD2	616312	Cataract 46, juvenile-onset, 212500 (3), Autosomal recessive
LIM2	154045	Cataract 19, multiple types, 615277 (3), Autosomal recessive
LSS	600909	Cataract 44, 616509 (3), Autosomal recessive
MAF	177075	Ayme-Gripp syndrome, 601088 (3), Autosomal dominant; Cataract 21, multiple types, 610202 (3), Autosomal dominant
MIP	154050	Cataract 15, multiple types, 615274 (3), Autosomal dominant
MYH9	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
NF2	607379	Meningioma, NF2-related, somatic, 607174 (3); Neurofibromatosis, type 2, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 162091 (3)
NHS	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
OCRL	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive

OPA3	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
P3H2	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive
PANK4	606162	No OMIM phenotype
PAX6	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
PITX3	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
RRAGA	612194	No OMIM phenotype
SIL1	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SIPA1L3	616655	?Cataract 45, 616851 (3), Autosomal recessive
SLC16A12	611910	Cataract 47, juvenile, with microcornea, 612018 (3), Autosomal dominant
SLC33A1	603690	Congenital cataracts, hearing loss, and neurodegeneration, 614482 (3), Autosomal recessive; Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant
TDRD7	611258	Cataract 36, 613887 (3), Autosomal recessive
UNC45B	611220	?Cataract 43, 616279 (3), Autosomal dominant
VIM	193060	Cataract 30, pulverulent, 116300 (3), Autosomal dominant
VSX2	142993	Microphthalmia with coloboma 3, 610092 (3); Microphthalmia, isolated 2, 610093 (3)
WFS1	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.