Cataract

Gene panel

Gene panel information

Gene panel	**Cataract**
Version	4
Total genes	89
Activation date	Wednesday 18 december 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ABHD12	99.98 %	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674 (3), Autosomal recessive
ADAMTSL4	99.66 %	610113	Ectopia lentis et pupillae, 225200 (3), Autosomal recessive; Ectopia lentis, isolated, autosomal recessive, 225100 (3), Autosomal recessive
AGK	99.99 %	610345	Cataract 38, autosomal recessive, 614691 (3), Autosomal recessive; Sengers syndrome, 212350 (3), Autosomal recessive
ALDH18A1	99.96 %	138250	Spastic paraplegia 9A, autosomal dominant, 601162 (3), Autosomal dominant; Cutis laxa, autosomal recessive, type IIIA, 219150 (3), Autosomal recessive; Spastic paraplegia 9B, autosomal recessive, 616586 (3), Autosomal recessive; Cutis laxa, autosomal dominant 3, 616603 (3), Autosomal dominant
AQP5	99.88 %	600442	Palmoplantar keratoderma, Bothnian type, 600231 (3), Autosomal dominant
ATAD3A	99.62 %	612316	Harel-Yoon syndrome, 617183 (3), Autosomal dominant, Autosomal recessive; Pontocerebellar hypoplasia, hypotonia, and respiratory insufficiency syndrome, neonatal lethal, 618810 (3), Autosomal recessive
B3GLCT	99.9 %	610308	Peters-plus syndrome, 261540 (3), Autosomal recessive
BCOR	99.97 %	300485	Microphthalmia, syndromic 2, 300166 (3), X-linked dominant
BEST1	99.86 %	607854	Macular dystrophy, vitelliform, 2, 153700 (3), Autosomal dominant; ?Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma 2, 193220 (3), Autosomal dominant; Retinitis pigmentosa-50, 613194 (3); Retinitis pigmentosa, concentric, 613194 (3); Vitreoretinochoroidopathy, 193220 (3), Autosomal dominant; Bestrophinopathy, autosomal recessive, 611809 (3)
BFSP1	100 %	603307	Cataract 33, multiple types, 611391 (3), Autosomal dominant, Autosomal recessive
BFSP2	99.09 %	603212	Cataract 12, multiple types, 611597 (3), Autosomal dominant
CHMP4B	99.96 %	610897	Cataract 31, multiple types, 605387 (3), Autosomal dominant
COL11A1	90.72 %	120280	Fibrochondrogenesis 1, 228520 (3), Autosomal recessive; Stickler syndrome, type II, 604841 (3), Autosomal dominant; Marshall syndrome, 154780 (3), Autosomal dominant; Deafness, autosomal dominant 37, 618533 (3), Autosomal dominant; {Lumbar disc herniation, susceptibility to}, 603932 (3)
COL18A1	99.99 %	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant

Cataract

Gene panel

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
COL2A1	99.87 %	120140	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 (3), Autosomal dominant; Czech dysplasia, 609162 (3), Autosomal dominant; Achondrogenesis, type II or hypochondrogenesis, 200610 (3), Autosomal dominant; Spondyloperipheral dysplasia, 271700 (3), Autosomal dominant; SMED Strudwick type, 184250 (3), Autosomal dominant; ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 (3), Autosomal dominant; SED congenita, 183900 (3), Autosomal dominant; Kniest dysplasia, 156550 (3), Autosomal dominant; Stickler syndrome, type I, nonsyndromic ocular, 609508 (3), Autosomal dominant; Osteoarthritis with mild chondrodysplasia, 604864 (3), Autosomal dominant; Stickler syndrome, type I, 108300 (3), Autosomal dominant; Platyspondylic skeletal dysplasia, Torrance type, 151210 (3), Autosomal dominant; Spondyloepiphyseal dysplasia, Stanescu type, 616583 (3), Autosomal dominant; Avascular necrosis of the femoral head, 608805 (3), Autosomal dominant; Legg-Calve-Perthes disease, 150600 (3), Autosomal dominant
CRYAA	19.49 %	123580	Cataract 9, multiple types, 604219 (3), Autosomal dominant, Autosomal recessive
CRYAB	100 %	123590	Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related, 613869 (3), Autosomal recessive; Myopathy, myofibrillar, 2, 608810 (3), Autosomal dominant; Cataract 16, multiple types, 613763 (3), Autosomal dominant, Autosomal recessive; Cardiomyopathy, dilated, 1II, 615184 (3), Autosomal dominant
CRYBA1	99.99 %	123610	Cataract 10, multiple types, 600881 (3), Autosomal dominant
CRYBA2	100 %	600836	?Cataract 42, 115900 (3), Autosomal dominant
CRYBA4	100 %	123631	Cataract 23, 610425 (3), Autosomal dominant
CRYBB1	99.46 %	600929	Cataract 17, multiple types, 611544 (3), Autosomal dominant, Autosomal recessive
CRYBB2	99.94 %	123620	Cataract 3, multiple types, 601547 (3), Autosomal dominant
CRYBB3	99.99 %	123630	Cataract 22, 609741 (3), Autosomal dominant, Autosomal recessive
CRYGB	99.99 %	123670	Cataract 39, multiple types, autosomal dominant, 615188 (3), Autosomal dominant
CRYGC	100 %	123680	Cataract 2, multiple types, 604307 (3), Autosomal dominant
CRYGD	100 %	123690	Cataract 4, multiple types, 115700 (3), Autosomal dominant
CRYGS	100 %	123730	Cataract 20, multiple types, 116100 (3), Autosomal dominant
CTDP1	99.97 %	604927	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3), Autosomal recessive
CYP21A2	99.85 %	613815	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive
CYP27A1	100 %	606530	Cerebrotendinous xanthomatosis, 213700 (3), Autosomal recessive
CYP51A1	97.82 %	601637	*No OMIM phenotypes*
DNMBP	99.94 %	611282	Cataract 48, 618415 (3), Autosomal recessive
EPG5	99.95 %	615068	Vici syndrome, 242840 (3), Autosomal recessive
EPHA2	99.99 %	176946	Cataract 6, multiple types, 116600 (3), Autosomal dominant
EYA1	99.81 %	601653	Branchiootic syndrome 1, 602588 (3), Autosomal dominant; Branchiootorenal syndrome 1, with or without cataracts, 113650 (3), Autosomal dominant; Anterior segment anomalies with or without cataract, 602588 (3), Autosomal dominant; ?Otofaciocervical syndrome, 166780 (3), Autosomal dominant
FAM126A	99.81 %	610531	Leukodystrophy, hypomyelinating, 5, 610532 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FBN1	99.85 %	134797	Geleophysic dysplasia 2, 614185 (3), Autosomal dominant; Weill-Marchesani syndrome 2, dominant, 608328 (3), Autosomal dominant; Ectopia lentis, familial, 129600 (3), Autosomal dominant; MASS syndrome, 604308 (3), Autosomal dominant; Marfan lipodystrophy syndrome, 616914 (3), Autosomal dominant; Acromicric dysplasia, 102370 (3), Autosomal dominant; Marfan syndrome, 154700 (3), Autosomal dominant; Stiff skin syndrome, 184900 (3), Autosomal dominant
FOXE3	99.29 %	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	99.99 %	134790	Hyperferritinemia-cataract syndrome, 600886 (3), Autosomal dominant; L-ferritin deficiency, dominant and recessive, 615604 (3), Autosomal dominant, Autosomal recessive; Neurodegeneration with brain iron accumulation 3, 606159 (3), Autosomal dominant
FYCO1	100 %	607182	Cataract 18, autosomal recessive, 610019 (3), Autosomal recessive
GALK1	100 %	604313	Galactokinase deficiency with cataracts, 230200 (3), Autosomal recessive
GALT	100 %	606999	Galactosemia, 230400 (3), Autosomal recessive
GCNT2	100 %	600429	[Blood group, Ii], 110800 (3), Autosomal dominant; Adult i phenotype without cataract, 110800 (3), Autosomal dominant; Cataract 13 with adult i phenotype, 116700 (3), Autosomal recessive
GFER	100 %	600924	Myopathy, mitochondrial progressive, with congenital cataract and developmental delay, 613076 (3), Autosomal recessive
GJA1	100 %	121014	Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
GJA3	100 %	121015	Cataract 14, multiple types, 601885 (3), Autosomal dominant
GJA8	99.99 %	600897	Cataract 1, multiple types, 116200 (3), Autosomal dominant
HMX1	100 %	142992	Oculoauricular syndrome, 612109 (3), Autosomal recessive
HSF4	99.98 %	602438	Cataract 5, multiple types, 116800 (3), Autosomal dominant
INPP5K	99.94 %	607875	Muscular dystrophy, congenital, with cataracts and intellectual disability, 617404 (3), Autosomal recessive
INTS1	100 %	611345	Neurodevelopmental disorder with cataracts, poor growth, and dysmorphic facies, 618571 (3), Autosomal recessive
JAM3	100 %	606871	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730 (3), Autosomal recessive
LCAT	99.97 %	606967	Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive
LEMD2	100 %	616312	Marbach-Rustad progeroid syndrome, 619322 (3), Autosomal dominant; Cataract 46, juvenile-onset, 212500 (3), Autosomal recessive
LIM2	100 %	154045	Cataract 19, multiple types, 615277 (3), Autosomal dominant, Autosomal recessive
LONP1	99.99 %	605490	CODAS syndrome, 600373 (3), Autosomal recessive
LSS	99.98 %	600909	Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-intellectual disability syndrome 4, 618840 (3), Autosomal recessive
MAF	99.73 %	177075	Cataract 21, multiple types, 610202 (3), Autosomal dominant; Ayme-Gripp syndrome, 601088 (3), Autosomal dominant
MBTPS1	99.94 %	603355	?Spondyloepiphyseal dysplasia, Kondo-Fu type, 618392 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MED27	99.99 %	605044	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia, 619286 (3), Autosomal recessive
MIP	99.98 %	154050	Cataract 15, multiple types, 615274 (3), Autosomal dominant
MIR184	100 %	613146	EDICT syndrome, 614303 (3), Autosomal dominant
MYH9	99.95 %	160775	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss, 155100 (3), Autosomal dominant; Deafness, autosomal dominant 17, 603622 (3), Autosomal dominant
NDP	99.98 %	300658	Exudative vitreoretinopathy 2, X-linked, 305390 (3), X-linked recessive, X-linked dominant; Norrie disease, 310600 (3), X-linked recessive
NF2	100 %	607379	Meningioma, NF2-related, somatic, 607174 (3); Schwannomatosis, vestibular, 101000 (3), Autosomal dominant; Schwannomatosis, somatic, 101000 (3)
NHS	99.96 %	300457	Cataract 40, X-linked, 302200 (3), X-linked; Nance-Horan syndrome, 302350 (3), X-linked dominant
OCRL	99.89 %	300535	Dent disease 2, 300555 (3), X-linked recessive; Lowe syndrome, 309000 (3), X-linked recessive
OPA3	100 %	606580	3-methylglutaconic aciduria, type III, 258501 (3), Autosomal recessive; Optic atrophy 3 with cataract, 165300 (3), Autosomal dominant
P3H2	99.93 %	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive
PANK4	100 %	606162	?Cataract 49, 619593 (3), Autosomal dominant
PAX6	99.95 %	607108	Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; Microphthalmia/coloboma 12, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant
PIKFYVE	99.81 %	609414	Corneal fleck dystrophy, 121850 (3), Autosomal dominant
PITX3	100 %	602669	Cataract 11, multiple types, 610623 (3), Autosomal dominant, Autosomal recessive; Anterior segment dysgenesis 1, multiple subtypes, 107250 (3), Autosomal dominant; Cataract 11, syndromic, autosomal recessive, 610623 (3), Autosomal dominant, Autosomal recessive
PXDN	100 %	605158	Anterior segment dysgenesis 7, with sclerocornea, 269400 (3), Autosomal recessive
RNH1	99.99 %	173320	{Encephalopathy, acute, infection-induced, susceptibility to, 12}, 620461 (3), Autosomal recessive
RRAGA	100 %	612194	*No OMIM phenotypes*
SC5D	99.97 %	602286	Lathosterolosis, 607330 (3), Autosomal recessive
SIL1	99.95 %	608005	Marinesco-Sjogren syndrome, 248800 (3), Autosomal recessive
SIPA1L3	99.97 %	616655	?Cataract 45, 616851 (3), Autosomal recessive
SLC16A12	100 %	611910	Cataract 47, juvenile, with microcornea, 612018 (3), Autosomal dominant
SLC33A1	99.67 %	603690	Spastic paraplegia 42, autosomal dominant, 612539 (3), Autosomal dominant; Huppke-Brendel syndrome, 614482 (3), Autosomal recessive
TAPT1	99.64 %	612758	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897 (3), Autosomal recessive
TBC1D20	100 %	611663	Warburg micro syndrome 4, 615663 (3), Autosomal recessive
TDRD7	99.87 %	611258	Cataract 36, 613887 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TRPM3	99.89 %	608961	?Cataract 50 with or without glaucoma, 620253 (3), Autosomal dominant; Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures, 620224 (3), Autosomal dominant
UNC45B	100 %	611220	?Cataract 43, 616279 (3), Autosomal dominant; Myofibrillar myopathy 11, 619178 (3), Autosomal recessive
VIM	100 %	193060	Cataract 30, pulverulent, 116300 (3), Autosomal dominant
VSX2	99.99 %	142993	Microphthalmia, isolated 2, 610093 (3), Autosomal recessive; Microphthalmia with coloboma 3, 610092 (3), Autosomal recessive
WFS1	99.99 %	606201	Deafness, autosomal dominant 6/14/38, 600965 (3), Autosomal dominant; ?Cataract 41, 116400 (3), Autosomal dominant; Wolfram-like syndrome, autosomal dominant, 614296 (3), Autosomal dominant; {Diabetes mellitus, noninsulin-dependent, association with}, 125853 (3), Autosomal dominant; Wolfram syndrome 1, 222300 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2024-09-05**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

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

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.