Myopia

Gene panel

Gene panel information

Gene panel	**Myopia**
Version	2
Total genes	39
Activation date	Monday 22 may 2023
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
ADAMTS18	99.99 %	607512	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458 (3), Autosomal recessive
AGRN	99.99 %	103320	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120 (3), Autosomal recessive
ARR3	99.94 %	301770	Myopia 26, X-linked, female-limited, 301010 (3), X-linked
BSG	100 %	109480	[Blood group, OK], 111380 (3)
CACNA1F	99.94 %	300110	Cone-rod dystrophy, X-linked, 3, 300476 (3), X-linked recessive; Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071 (3), X-linked; Aland Island eye disease, 300600 (3), X-linked
CNGB3	99.96 %	605080	Achromatopsia 3, 262300 (3), Autosomal recessive
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
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
COL9A1	99.91 %	120210	Stickler syndrome, type IV, 614134 (3), Autosomal recessive; ?Epiphyseal dysplasia, multiple, 6, 614135 (3), Autosomal dominant
COL9A2	98.76 %	120260	Epiphyseal dysplasia, multiple, 2, 600204 (3), Autosomal dominant; ?Stickler syndrome, type V, 614284 (3), Autosomal recessive
COL9A3	99.99 %	120270	{Intervertebral disc disease, susceptibility to}, 603932 (3); Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 (3), Autosomal dominant; Stickler syndrome, type VI, 620022 (3), Autosomal recessive
CPSF1	99.99 %	606027	Myopia 27, 618827 (3), Autosomal dominant
CTSH	99.99 %	116820	*No OMIM phenotypes*

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DZIP1	99.97 %	608671	Spermatogenic failure 47, 619102 (3), Autosomal recessive; ?Mitral valve prolapse 3, 610840 (3), Autosomal dominant
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
GPR179	99.99 %	614515	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565 (3), Autosomal recessive
GRM6	100 %	604096	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270 (3), Autosomal recessive
GZF1	100 %	613842	Joint laxity, short stature, and myopia, 617662 (3), Autosomal recessive
LOXL3	99.95 %	607163	Myopia 28, autosomal recessive, 619781 (3), Autosomal recessive
LRIT3	100 %	615004	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058 (3), Autosomal recessive
LRP2	99.86 %	600073	Donnai-Barrow syndrome, 222448 (3), Autosomal recessive
LRPAP1	99.99 %	104225	Myopia 23, autosomal recessive, 615431 (3), Autosomal recessive
NDUFAF7	99.98 %	615898	*No OMIM phenotypes*
NYX	100 %	300278	Night blindness, congenital stationary (complete), 1A, X-linked, 310500 (3), X-linked recessive
OPN1LW	71.75 %	300822	Blue cone monochromacy, 303700 (3), X-linked recessive; Colorblindness, protan, 303900 (3), X-linked
OPN1MW	30.23 %	300821	Colorblindness, deutan, 303800 (3), X-linked; Blue cone monochromacy, 303700 (3), X-linked recessive
P3H2	99.93 %	610341	Myopia, high, with cataract and vitreoretinal degeneration, 614292 (3), Autosomal recessive
P4HA2	100 %	600608	Myopia 25, autosomal dominant, 617238 (3), Autosomal dominant
PRIMPOL	99.86 %	615421	Myopia 22, autosomal dominant, 615420 (3), Autosomal dominant
RAB28	99.97 %	612994	Cone-rod dystrophy 18, 615374 (3), Autosomal recessive
RPGR	94.45 %	312610	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (3), X-linked; Cone-rod dystrophy, X-linked, 1, 304020 (3), X-linked recessive; Retinitis pigmentosa 3, 300029 (3), X-linked; Macular degeneration, X-linked atrophic, 300834 (3), X-linked recessive
SCO2	100 %	604272	Myopia 6, 608908 (3), Autosomal dominant; Mitochondrial complex IV deficiency, nuclear type 2, 604377 (3), Autosomal recessive
SLC39A5	99.99 %	608730	Myopia 24, autosomal dominant, 615946 (3), Autosomal dominant
SLITRK6	100 %	609681	Deafness and myopia, 221200 (3), Autosomal recessive
TNFRSF21	99.97 %	605732	*No OMIM phenotypes*
TRPM1	99.96 %	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3)
XYLT1	99.98 %	608124	Desbuquois dysplasia 2, 615777 (3), Autosomal recessive; {Pseudoxanthoma elasticum, modifier of severity of}, 264800 (3), Autosomal recessive
ZNF644	99.37 %	614159	Myopia 21, autosomal dominant, 614167 (3), Autosomal dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

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.

* Exome panels: >=20x, HyperCap panels: >=30x