

Corneal dystrophy

Gene panel

Gene panel information

Gene panel	Corneal dystrophy
Version	4
Total genes	37
Activation date	Tuesday 24 september 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AGBL1	99.92 %	615496	Corneal dystrophy, Fuchs endothelial, 8, 615523 (3), Autosomal dominant
CHRD1	99.67 %	300350	Megalocornea 1, X-linked, 309300 (3), X-linked recessive
CHST6	99.94 %	605294	Macular corneal dystrophy, 217800 (3), Autosomal recessive
COL17A1	99.88 %	113811	Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant; Epidermolysis bullosa, junctional 4, intermediate, 619787 (3), Autosomal recessive
COL3A1	99.79 %	120180	Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant; Polymicrogyria with or without vascular-type EDS, 618343 (3), Autosomal recessive
COL5A1	99.82 %	120215	Ehlers-Danlos syndrome, classic type, 1, 130000 (3), Autosomal dominant; Fibromuscular dysplasia, multifocal, 619329 (3), Autosomal dominant
COL8A2	98.99 %	120252	Corneal dystrophy, posterior polymorphous 2, 609140 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 1, 136800 (3), Autosomal dominant
CYP4V2	99.91 %	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
DCN	99.41 %	125255	Corneal dystrophy, congenital stromal, 610048 (3), Autosomal dominant
GJA8	99.95 %	600897	Cataract 1, multiple types, 116200 (3), Autosomal dominant
GRHL2	99.68 %	608576	Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive; Corneal dystrophy, posterior polymorphous, 4, 618031 (3), Autosomal dominant
GSN	99.67 %	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
KERA	99.43 %	603288	Cornea plana 2, autosomal recessive, 217300 (3), Autosomal recessive
KRT12	99.69 %	601687	Meesmann corneal dystrophy 1, 122100 (3), Autosomal dominant
KRT3	98.91 %	148043	Meesmann corneal dystrophy 2, 618767 (3), Autosomal dominant
LCAT	99.91 %	606967	Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive
LOXHD1	99.89 %	613072	Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive
MCOLN1	99.64 %	605248	Lisch epithelial corneal dystrophy, 620763 (3), Autosomal dominant; Mucopolysaccharidosis IV, 252650 (3), Autosomal recessive
MIR184	100 %	613146	EDICT syndrome, 614303 (3), Autosomal dominant
NLRP1	99.92 %	606636	{Viteligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3); ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 (3), Autosomal recessive; Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant
NLRP3	99.95 %	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant

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OVOL2	98.76 %	616441	Corneal dystrophy, posterior polymorphous, 1, 122000 (3), Autosomal dominant
PAX6	99.91 %	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	99.5 %	609414	Corneal fleck dystrophy, 121850 (3), Autosomal dominant
PITX2	99.85 %	601542	Ring dermoid of cornea, 180550 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant
PLCB3	99.82 %	600230	Spondylometaphyseal dysplasia with corneal dystrophy, 618961 (3), Autosomal recessive
PRDM5	99.75 %	614161	Brittle cornea syndrome 2, 614170 (3), Autosomal recessive
PRDX3	99.97 %	604769	Spinocerebellar ataxia, autosomal recessive 32, 619862 (3), Autosomal recessive; Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871 (3), Autosomal dominant
SLC4A11	99.86 %	610206	Corneal endothelial dystrophy, autosomal recessive, 217700 (3), Autosomal recessive; Corneal dystrophy, Fuchs endothelial, 4, 613268 (3); Corneal endothelial dystrophy and perceptive deafness, 217400 (3), Autosomal recessive
SLC4A4	99.83 %	603345	Proximal renal tubular acidosis-ocular anomaly syndrome, 604278 (3), Autosomal recessive
STS	99.8 %	300747	Ichthyosis, X-linked, 308100 (3), X-linked recessive
TACSTD2	99.94 %	137290	Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive
TGFBI	99.79 %	601692	Corneal dystrophy, Avellino type, 607541 (3), Autosomal dominant; Corneal dystrophy, Reis-Bucklers type, 608470 (3), Autosomal dominant; Corneal dystrophy, Thiel-Behnke type, 602082 (3), Autosomal dominant; Corneal dystrophy, Groenouw type I, 121900 (3), Autosomal dominant; Corneal dystrophy, epithelial basement membrane, 121820 (3), Autosomal dominant; Corneal dystrophy, lattice type I, 122200 (3), Autosomal dominant; Corneal dystrophy, lattice type IIIA, 608471 (3), Autosomal dominant
TUBA3D	99.82 %	617878	Keratoconus 9, 617928 (3), Autosomal dominant
UBIAD1	99.89 %	611632	Corneal dystrophy, Schnyder type, 121800 (3), Autosomal dominant
ZEB1	99.95 %	189909	Corneal dystrophy, posterior polymorphous, 3, 609141 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 6, 613270 (3), Autosomal dominant
ZNF469	99.95 %	612078	Brittle cornea syndrome 1, 229200 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

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.