# Corneal dystrophy

Gene panel

## Gene panel information

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| **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** | 99.99 % | 615496 | Corneal dystrophy, Fuchs endothelial, 8, 615523 (3), Autosomal dominant |
| **CHRDL1** | 99.94 % | 300350 | Megalocornea 1, X-linked, 309300 (3), X-linked recessive |
| **CHST6** | 100 % | 605294 | Macular corneal dystrophy, 217800 (3), Autosomal recessive |
| **COL17A1** | 99.98 % | 113811 | Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant; Epidermolysis bullosa, junctional 4, intermediate, 619787 (3), Autosomal recessive |
| **COL3A1** | 99.87 % | 120180 | Ehlers-Danlos syndrome, vascular type, 130050 (3), Autosomal dominant; Polymicrogyria with or without vascular-type EDS, 618343 (3), Autosomal recessive |
| **COL5A1** | 99.99 % | 120215 | Ehlers-Danlos syndrome, classic type, 1, 130000 (3), Autosomal dominant; Fibromuscular dysplasia, multifocal, 619329 (3), Autosomal dominant |
| **COL8A2** | 99.94 % | 120252 | Corneal dystrophy, posterior polymorphous 2, 609140 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 1, 136800 (3), Autosomal dominant |
| **CYP4V2** | 99.98 % | 608614 | Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive |
| **DCN** | 99.42 % | 125255 | Corneal dystrophy, congenital stromal, 610048 (3), Autosomal dominant |
| **GJA8** | 99.99 % | 600897 | Cataract 1, multiple types, 116200 (3), Autosomal dominant |
| **GRHL2** | 100 % | 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** | 99.93 % | 137350 | Amyloidosis, Finnish type, 105120 (3), Autosomal dominant |
| **KERA** | 99.95 % | 603288 | Cornea plana 2, autosomal recessive, 217300 (3), Autosomal recessive |
| **KRT12** | 99.92 % | 601687 | Meesmann corneal dystrophy 1, 122100 (3), Autosomal dominant |
| **KRT3** | 99.89 % | 148043 | Meesmann corneal dystrophy 2, 618767 (3), Autosomal dominant |
| **LCAT** | 99.97 % | 606967 | Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive |
| **LOXHD1** | 99.99 % | 613072 | Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive |
| **MCOLN1** | 100 % | 605248 | Lisch epithelial corneal dystrophy, 620763 (3), Autosomal dominant; Mucolipidosis IV, 252650 (3), Autosomal recessive |
| **MIR184** | 100 % | 613146 | EDICT syndrome, 614303 (3), Autosomal dominant |
| **NLRP1** | 95.26 % | 606636 | {Vitiligo-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 dominant, Autosomal recessive; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant |
| **NLRP3** | 100 % | 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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| Gene | % at least 20 x covered* | OMIM gene id | OMIM Phenotypes |
|---|---|---|---|
| **OVOL2** | 100 % | 616441 | Corneal dystrophy, posterior polymorphous, 1, 122000 (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 |
| **PITX2** | 99.98 % | 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** | 100 % | 600230 | Spondylometaphyseal dysplasia with corneal dystrophy, 618961 (3), Autosomal recessive |
| **PRDM5** | 99.76 % | 614161 | Brittle cornea syndrome 2, 614170 (3), Autosomal recessive |
| **PRDX3** | 99.94 % | 604769 | Spinocerebellar ataxia, autosomal recessive 32, 619862 (3), Autosomal recessive; Corneal dystrophy, punctiform and polychromatic pre-Descemet, 619871 (3), Autosomal dominant |
| **SLC4A11** | 100 % | 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** | 99.97 % | 603345 | Proximal renal tubular acidosis-ocular anomaly syndrome, 604278 (3), Autosomal recessive |
| **STS** | 99.81 % | 300747 | Ichthyosis, X-linked, 308100 (3), X-linked recessive |
| **TACSTD2** | 100 % | 137290 | Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive |
| **TGFBI** | 99.89 % | 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** | 99.94 % | 617878 | Keratoconus 9, 617928 (3), Autosomal dominant |
| **UBIAD1** | 99.98 % | 611632 | Corneal dystrophy, Schnyder type, 121800 (3), Autosomal dominant |
| **ZEB1** | 100 % | 189909 | Corneal dystrophy, posterior polymorphous, 3, 609141 (3), Autosomal dominant; Corneal dystrophy, Fuchs endothelial, 6, 613270 (3), Autosomal dominant |
| **ZNF469** | 100 % | 612078 | Brittle cornea syndrome 1, 229200 (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.