

Corneale dystrofie panel		
versie	V1 (31 genen)	Centrum voor Medische Genetica Gent
Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>AGBL1</i>	615496	Corneal dystrophy, Fuchs endothelial, 8, 615523 (3), Autosomal dominant
<i>CHRD1</i>	300350	Megalocornea 1, X-linked, 309300 (3), X-linked recessive
<i>CHST6</i>	605294	Macular corneal dystrophy, 217800 (3), Autosomal recessive
<i>COL17A1</i>	113811	Epidermolysis bullosa, junctional, localisata variant, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant
<i>COL5A1</i>	120215	Ehlers-Danlos syndrome, classic type, 1, 130000 (3), Autosomal dominant
<i>COL8A2</i>	120252	Corneal dystrophy, Fuchs endothelial, 1, 136800 (3), Autosomal dominant; Corneal dystrophy, posterior polymorphous 2, 609140 (3), Autosomal dominant
<i>CYP4V2</i>	608614	Bietti crystalline corneoretinal dystrophy, 210370 (3), Autosomal recessive
<i>DCN</i>	125255	Corneal dystrophy, congenital stromal, 610048 (3), Autosomal dominant
<i>GRHL2</i>	608576	Corneal dystrophy, posterior polymorphous, 4, 618031 (3), Autosomal dominant; Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive
<i>GSN</i>	137350	Amyloidosis, Finnish type, 105120 (3), Autosomal dominant
<i>KERA</i>	603288	Cornea plana 2, autosomal recessive, 217300 (3), Autosomal recessive
<i>KRT12</i>	601687	Meesmann corneal dystrophy, 122100 (3), Autosomal dominant
<i>KRT3</i>	148043	Meesmann corneal dystrophy, 122100 (3), Autosomal dominant
<i>LCAT</i>	606967	Fish-eye disease, 136120 (3), Autosomal recessive; Norum disease, 245900 (3), Autosomal recessive
<i>LOXHD1</i>	613072	Deafness, autosomal recessive 77, 613079 (3), Autosomal recessive
<i>NLRP1</i>	606636	Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant; {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3)

<i>NLRP3</i>	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
<i>OVOL2</i>	616441	Corneal dystrophy, posterior polymorphous, 1, 122000 (3), Autosomal dominant
<i>PAX6</i>	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
<i>PIKFYVE</i>	609414	Corneal fleck dystrophy, 121850 (3), Autosomal dominant
<i>PITX2</i>	601542	Anterior segment dysgenesis 4, 137600 (3), Autosomal dominant; Axenfeld-Rieger syndrome, type 1, 180500 (3), Autosomal dominant; Ring dermoid of cornea, 180550 (3), Autosomal dominant
<i>PRDM5</i>	614161	Brittle cornea syndrome 2, 614170 (3), Autosomal recessive
<i>SLC4A11</i>	610206	Corneal dystrophy, Fuchs endothelial, 4, 613268 (3); Corneal endothelial dystrophy and perceptive deafness, 217400 (3), Autosomal recessive; Corneal endothelial dystrophy, autosomal recessive, 217700 (3), Autosomal recessive
<i>SOD1</i>	147450	Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant; Spastic tetraplegia and axial hypotonia, progressive, 618598 (3)
<i>TACSTD2</i>	137290	Corneal dystrophy, gelatinous drop-like, 204870 (3), Autosomal recessive
<i>TCF4</i>	602272	Corneal dystrophy, Fuchs endothelial, 3, 613267 (3), Autosomal dominant; Pitt-Hopkins syndrome, 610954 (3), Autosomal dominant
<i>TGFBI</i>	601692	Corneal dystrophy, Avellino type, 607541 (3), Autosomal dominant; Corneal dystrophy, Groenouw type I, 121900 (3), Autosomal dominant; Corneal dystrophy, Reis-Bucklers type, 608470 (3); Corneal dystrophy, Thiel-Behnke type, 602082 (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
<i>UBIAD1</i>	611632	Corneal dystrophy, Schnyder type, 121800 (3), Autosomal dominant

<i>VSX1</i>	605020	?Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195 (3); Keratoconus 1, 148300 (3), Autosomal dominant
<i>ZEB1</i>	189909	Corneal dystrophy, Fuchs endothelial, 6, 613270 (3); Corneal dystrophy, posterior polymorphous, 3, 609141 (3)
<i>ZNF469</i>	612078	Brittle cornea syndrome 1, 229200 (3), Autosomal recessive

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: Sept 30, 2019

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.