Corneal Dystrophies

Precision Panel

Overview

Corneal Dystrophies (CD) are a group of genetic, commonly progressive, eye disorders in which abnormal material often accumulates in the clear outer layer of the eye (the cornea). The onset of symptoms varies between patients, from asymptomatic from significant vision impairment cases. This condition can either affect one or both eyes, and the intensity of symptoms in that case is not necessarily equal. The age of onset and specific symptoms vary among the different forms of corneal dystrophy. CD can be grouped by which layers of the cornea is affected:

1. Anterior/Superficial Coneal Dystrophies.
2. Stromal Corneal Dystrophies.
3. Posterior Corneal Dystrophies.

The mode of inheritance varies from autosomal dominant to autosomal recessive.

The Igenomix Corneal Dystrophies Precision Panel can be used to make an accurate and directed diagnosis leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Corneal Dystrophies Precision Panel is indicated for those patients with a clinical diagnosis or suspicion presenting with or without the following manifestations:

- Dry eyes
- Loss of vision.
- Sensitivity to light
- Pain in the eye
- Corneal erosions
- Blurred vision.

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of multidisciplinary treatment including a regular follow up in case of asymptomatic patients. Eye drops and ointments can be used in case of mild symptoms. Surgical treatment (corneal transplant) can be considered if the loss of vision is severe enough.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

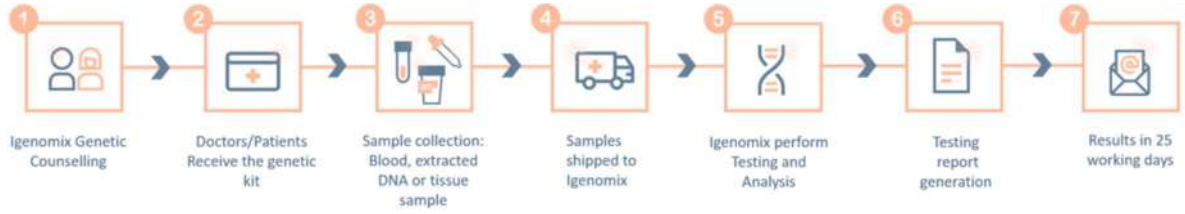
GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ADAMTS18	Microcornea, Myopic Chorioreti-L Atrophy, And Telecanthus	AR	100	14 of 14
AGBL1	Corneal Dystrophy, Fuchs Endothelial Dystrophy	AD	99.94	4 of 4
ARL2	Microcornea, Rod-Cone Dystrophy, Cataract, Posterior Staphyloma	AD	100	1 of 1
B3GALNT2	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	97.14	17 of 17
B3GALT6	Spondyloepimetaphyseal Dysplasia, Ehlers-Danlos Syndrome	AR	65.09	24 of 39
B3GLCT	Peters-Plus Syndrome	AR	99.96	-
B9D2	Meckel Syndrome	AR	84.81	4 of 5
BCOR	Microphthalmia, Oculofaciocardiodental Syndrome	X,XD,G	99.87	-
BEST1	Bestrophinopathy, Macular Dystrophy, Retinitis Pigmentosa, Vitreoretinochoroidopathy, Foveomacular Vitelliform Dystrophy, -Nophthalmos	AD,AR	94.35	342 of 344
BMP4	Microphthalmia	AD,MU,P	100	38 of 42
CDH11	Elsahy-Waters Syndrome, Branchioskeletogenital Syndrome	AR	99.95	10 of 10
CHN1	Duane Retraction Syndrome	AD	92.41	11 of 11
CHRDL1	Megalocornea, Meesmann Corneal Dystrophy	AR,X,XR,G	99.69	-
CHST5	Corneal Macular Dystrophy, Corneal Endothelial Dystrophy, Groenouw Corneal Dystrophy	-	99.97	-
CHST6	Corneal Dystrophy	AR	100	177 of 177
COL17A1	Epidermolysis Bullosa, Epithelial Recurrent Erosion Dystrophy	AD,AR	100	117 of 117
COL5A1	Ehlers-Danlos Syndrome	AD	99.08	191 of 195
COL8A2	Corneal Dystrophy, Fuchs Endothelial Dystrophy	AD	94.25	10 of 10
CRYAA	Cataract, Microcornea	AD,AR	100	26 of 26
CRYBA4	Cataract, Microcornea	AD	100	11 of 11
CRYBB1	Cataract	AD,AR	100	20 of 20
CRYBB2	Cataract, Microcornea	AD	100	28 of 28
CRYGC	Cataract, Microcornea	AD	100	31 of 31
CRYGD	Cataract, Microcornea	AD	99.98	28 of 28
CTCF	Intellectual Disability, Microcephaly	AD	96.6	39 of 41
CYP4V2	Bietti Crystalline Corneoreti-L Dystrophy, Fundus Dystrophy	AR	100	112 of 112
DAG1	Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eyeanomalies), Muscle-Eye-Brain Disease, Walker-Warburg Syndrome	AR	99.98	9 of 9
DCN	Corneal Dystrophy, Stromal Dystrophy	AD	97.89	5 of 5
DZANK1	Corneal Dystrophy	-	99.87	-

FBN1	Acromicric Dysplasia, Ectopia Lentis, Marfan Syndrome, Mass Syndrome, Weill-Marchesani Syndrome	AD	100	2836 of 2845
FMOD	Corneal Dystrophy	-	99.97	2 of 2
FOXE3	Aphakia, Peters Anomaly	AD,AR	81.19	25 of 31
GJA1	Oculodentodigital Dysplasia	AD,AR,MU,O	100	119 of 119
GJA8	Cataract	AD	99.2	72 of 73
GLA	Fabry Disease	X,XR,G	98	-
GNPTAB	Mucolipidosis	AR	100	279 of 280
GORAB	Geroderma Osteodysplastica	AR	96	17 of 18
GRHL2	Corneal Dystrophy	AD,AR	100	8 of 11
GSN	Amyloidosis, Corneal Dystrophy	AD	96.69	16 of 17
HMX1	Oculoauricular Syndrome	AR	85.58	2 of 2
JAG1	Alagille Syndrome	AD	99.98	640 of 641
KERA	Cornea Pla-	AR	99.93	17 of 17
KIF11	Lymphedema, Microcephaly, Chorioretinopathy	AD	99.78	82 of 89
KRAS	Cardiofaciocutaneous Syndrome, Oculoectodermal Syndrome	AD	100	38 of 38
KRT12	Corneal Dystrophy	AD	97.81	24 of 24
KRT3	Meesmann Corneal Dystrophy	AD	99.94	4 of 4
LCAT	Fish-Eye Disease	AR	90	110 of 110
LMX1B	-Il-Patella Syndrome	AD,AR	100	191 of 191
LOXHD1	Fuchs Corneal Dystrophy	AR	99.98	97 of 97
LTBP2	Megalocornea, Ectopia Lentis, Weill-Marchesani Syndrome	AR	99.98	34 of 34
LUM	Stromal Dystrophy, Macular Corneal Dystrophy, Cornea Pla-	-	99.83	1 of 1
MAB21L2	Microphthalmia	AD,AR	99.97	8 of 8
MAF	Cataract, Microcornea,	AD	75.14	23 of 23
MED25	Basel-Va-Gaite-Smirin-Yosef Syndrome	AR	100	5 of 5
MIR184	Edict Syndrome	AD	-	-
NHS	Cataract, -Nce-Horan Syndrome	X,XD,G	98.45	-
NIPBL	Cornelia De Lange Syndrome	AD	99.32	409 of 426
NLRP1	Corneal Intraepithelial Dyskeratosis, Palmoplantar Carcinoma	AD,AR,MU,P	99.37	15 of 15
NLRP3	Keratoendotheliitis Fugax Hereditaria	AD	100	152 of 152
OPN1LW	Cone-Rod Dystrophy, Color Blindness, Bornholm Eye Disease	X,XR,G	88	-
OPN1MW	Cone-Rod Dystrophy, Color Blindness, Bornholm Eye Disease	X,XR,G	41.73	-
OSMR	Amyloidosis	AD	100	14 of 14
OTX2	Microphthalmia	AD	100	56 of 58
OVOL2	Corneal Dystrophy	AD	99.87	0 of 3
PAX6	Aniridia, Coloboma Of Optic Nerve, Foveal Hypoplasia, Keratitis, Peters Anomaly, Wagr Syndrome	AD	100	460 of 485
PIKFYVE	Corneal Dystrophy, Yunis-Varon Syndrome,	AD	99.91	16 of 17
PITX2	Rieger Syndrome, Ring Dermoid Of Cornea, Peters Anomaly	AD	99.97	104 of 107
PITX3	Anterior Segment Mesenchymal Dysgenesis, Cataract	AD,AR	99.49	8 of 11
PLCB3	Spondylometaphyseal Dysplasia, Corneal Dystrophy	AR	100	1 of 1
PLK4	Microcephaly And Chorioretinopathy	AR	99.74	10 of 10
PRDM5	Brittle Cornea Syndrome, Axenfeld-Rieger Syndrome	AR	99.86	13 of 13
PRSS56	Microphthalmia, -Nophthalmos	AR	99.65	28 of 30
PXDN	Corneal Opacification	AR	100	14 of 14

RAB18	Warburg Micro Syndrome	AR	100	4 of 4
RAB3GAP1	Warburg Micro Syndrome, Cataract	AR	99.94	70 of 70
RAB3GAP2	Martsolf Syndrome, Warburg Micro Syndrome, Spastic Paraplegia, Cataract	AR	100	17 of 17
RBBP9	Corneal Dystrophy	-	100	-
RBP4	Microphthalmia, Coloboma, Reti-L Dystrophy	AD,AR	100	8 of 8
SLC16A12	Cataract, Microcornea	AD	99.95	18 of 18
SLC4A11	Corneal Dystrophy, Fuchs Endothelial Dystrophy	AD,AR	99.98	108 of 109
TACSTD2	Corneal Dystrophy	AR	97.66	31 of 32
TBC1D20	Warburg Micro Syndrome	AR	99.94	6 of 6
TCF4	Fuchs Corneal Dystrophy	AD	98.91	124 of 124
TEK	Glaucoma	AD,AR	100	35 of 35
TENM3	Microphthalmia, Coloboma	AR	99.94	6 of 6
TGFBI	Groenouw Corneal Dystrophy	AD	99.98	73 of 73
UBIAD1	Schnyder Corneal Dystrophy, Stromal Dystrophy	AD	92	32 of 32
VSX1	Keratoconus, Corneal Dystrophy	AD	94.56	25 of 25
ZEB1	Corneal Dystrophy	AD	89.95	63 of 65
ZNF133	Corneal Dystrophy	-	99.83	3 of 3
ZNF469	Brittle Cornea Syndrome	AR	99.91	79 of 79

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology



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