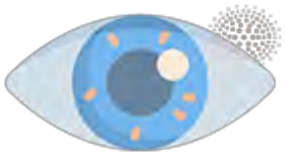
Congenital Cataract

Precision Panel



Overview

A cataract is a loss of lens transparency. The crystalline lens plays a crucial role in the refractive vision by facilitating variable fine focusing of light onto the retina. Congenital cataracts are usually diagnosed at birth, failure to do so can result in permanent vision loss. They are the most common cause of visual impairment and blindness in children worldwide. Genetic, metabolic, traumatic and infectious factors can all lead to childhood cataracts. However, about one quarter of congenital cataracts are associated to genetic defects, are usually bilateral and quite heterogeneous. Congenital cataracts can be inherited in an autosomal dominant or recessive pattern as well as X-linked.

The Igenomix Congenital Cataracts Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of blindness ultimately 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 Congenital Cataracts Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Lenticular opacity
- Irregular red reflex
- White reflex
- Retinal detachment

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 treatment with a multidisciplinary team in the form of medical therapy to prevent the progression to amblyopia, cataract surgery and dietary restrictions in the cause of galactosemia.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AGK	Congenital Cataract-Hypertrophic Cardiomyopathy-Mitochondrial Myopathy Syndrome	AR	99.98	33 of 33
BCOR	Syndromic Micropthalmia, Oculofaciocardiodental Syndrome	X,XD,G	99.87	NA of NA
BFSP1	Cataract, Multiple Types	AD,AR	97.54	7 of 8
BFSP2	Autosomal Dominant Cataract, Multiple Types	AD	100	7 of 7
CAV1	Lipodystrophy With Congenital Cataracts And Neurodegeneration , Lipodystrophy, Berardinelli-Seip Congenital Lipodystrophy, Diffuse Cutaneous Systemic Sclerosis, Limited Cutaneous Systemic Sclerosis	AD,AR	100	18 of 18
CHMP4B	Cataract, Multiple Types	AD	99.72	3 of 3
CRYAA	Cataract, Multiple Types, Cataract-Microcornea Syndrome	AD,AR	100	26 of 26
CRYAB	Alpha-b Crystallinopathy, Posterior Polar Cataract, Congenital Lamellar Cataract, Familial Isolated Dilated Cardiomyopathy	AD,AR	100	30 of 30
CRYBA1	Congenital Zonular Cataract With Sutural Opacities	AD	100	14 of 14
CRYBA2	Floriform Cataracts	AD	100	2 of 2
CRYBA4	Cataract-Microcornea Syndrome	AD	100	11 of 11
CRYBB1	Autosomal Recessive Congenital Nuclear Cataract, Cataract-Microcornea Syndrome	AD,AR	100	20 of 20
CRYBB2	Congenital Cataract Cerulean Type, Cataract-Microcornea Syndrome	AD	100	28 of 28
CRYBB3	Autosomal Recessive Congenital Nuclear Cataract	AD,AR	100	7 of 7
CRYGB	Cataract Multiple Types	AD	99.57	2 of 2
CRYGC	Coppock-Like Cataract, Cataract-Microcornea Syndrome	AD	100	31 of 31
CRYGD	Crystalline Aculeiform Cataract, Cataract-Microcornea Syndrome	AD	99.98	28 of 28
CRYGS	Membranous Cataract	AD	100	9 of 9
CTDP1	Congenital Cataracts, Facial Dysmorphism, And Neuropathy	AR	97.52	0 of 1
EPHA2	Posterior Polar Cataract, Congenital Total Cataract	AD	100	24 of 24
EYA1	Branchiootic Syndrome, Branchiootorenal Syndrome, Otofaciocervical Syndrome, Bor Syndrome	AD	100	197 of 199
FAM126A	Hypomyelination And Congenital Cataract	AR	100	11 of 12
FOXC1	Axenfeld-Rieger Syndrome Type 3, Iridogoniodysgenesis Type 1, Isolated Aniridia, Peters Anomaly	AD	88.98	94 of 100
FOXE3	Familial Thoracic Aortic Aneurysm, Anterior Segment Developmental Anomaly, Congenital Primary Aphakia, Peters Anomaly	AD,AR	81.19	25 of 31
FTL	Hyperferritinemia-Cataract Syndrome	AD,AR	100	21 of 63
FYCO1	Autosomal Recessive Congenital Cataract	AR	99.98	20 of 20
GALK1	Galactokinase Deficiency	AR	97.92	45 of 45
GCNT2	Cataract With Adult I Phenotype	AD,AR	97.19	9 of 10
GFER	Mitochondrial Progressive Myopathy, With Congenital Cataract, Hearing Loss, And Developmental Delay	AR	99.89	6 of 6
GJA3	Zonular Pulverulent Cataract	AD	95.63	35 of 45
GJA8	Zonular Pulverulent Cataract, Cataract-Microcornea Syndrome	AD	99.2	72 of 73
HSF4	Lamellar Cataract	AD	100	26 of 26
IARS2	Cataracts, Growth Hormone Deficiency, Sensory Neuropathy, Sensorineural Hearing Loss, And Skeletal Dysplasia	AR	99.95	11 of 11
INPP5K	Muscular Dystrophy, Congenital, With Cataracts And Intellectual Disability, Marinesco-Sjogren Syndrome	AR	92	10 of 10
LEMD2	Cataract, Congenital Or Juvenile Cataract	AR	93.48	3 of 3
LIM2	Cataract, Multiple Types	AR	100	4 of 4
LSS	Alopecia-Mental Retardation Syndrome, Cataract	AR	100	22 of 22
MAF	Ayme-Gripp Syndrome, Cataract Multiple Types, Cataract-Microcornea Syndrome	AD	75.14	23 of 23
MED25	Basel-Vanagaite-Smirin-Yosef Syndrome, Congenital Cataract-Microcephaly-Nevus Flammeus Simplex-Severe Intellectual Disability Syndrome	AR	100	5 of 5
MIP	Cataract Multiple Types	AD	100	29 of 29
MIR184	Edict Syndrome	AD	na	na
MIR204	Retinal Dystrophy And Iris Coloboma With Or Without Congenital Cataract	AD	na	na
MSMO1	Microcephaly, Congenital Cataract, And Psoriasiform Dermatitis	AR	99.78	4 of 4
NHS	Congenital Total Cataract With Posterior Sutural Opacities In Heterozygotes, Nance-Horan Syndrome	X,XD,G	98.45	NA of NA
OCRL	Dent Disease, Lowe Oculocerebrorenal Syndrome	X,XR,G	100	NA of NA
P3H2	Myopia, High, With Cataract And Vitreoretinal Degeneration	AR	99.81	NA of NA

PANK4	Neurodegeneration With Brain Iron Accumulation, Choreoacanthocytosis	-	99.6	1 of 1
PAX6	Aniridia, Anterior Segment Dysgenesis, Multiple Subtypes, Coloboma Of Optic Nerve, Bilateral Congenital, Ocular Coloboma, Foveal Hypoplasia And Presenile Cataract Syndrome, Hereditary Keratitis, Optic Nerve Hypoplasia, Bilateral Optic Nerve Aplasia, Wilms Tumor, Aniridia, Genitourinary Anomalies, And Mental Retardation Syndrome, Aniridia-Cerebellar Ataxia-Intellectual Disability Syndrome	AD	100	460 of 485
PITX2	Iridogoniodysgenesis Type 2, Rieger Syndrome Type 1, Ring Dermoid Of Cornea, Axenfeld-Rieger Syndrome, Peters Anomaly	AD	99.97	104 of 107
PITX3	Anterior Segment Mesenchymal Dysgenesis , Cataract Multiple Types	AD,AR	99.49	8 of 11
SIL1	Marinesco-Sjogren Syndrome	AR	100	47 of 48
SIPA1L3	Cataract	AR	96.17	3 of 3
SLC16A12	Juvenile Cataract, With Microcornea And Glucosuria	AD	99.95	18 of 18
SLC25A4	Progressive External Ophthalmoplegia With Mitochondrial DNA Deletions, Congenital Cataract-Hypertrophic Cardiomyopathy-Mitochondrial Myopathy Syndrome	AD,AR	99.84	16 of 16
SLC33A1	Congenital Cataracts, Hearing Loss, And Neurodegeneration	AD,AR	99.44	9 of 9
TDRD7	Cataract, Autosomal Recessive Congenital Cataract	AR	99.98	6 of 6
TKFC	Congenital Cataract-Hypertrophic Cardiomyopathy-Mitochondrial Myopathy Syndrome	AR	99.91	NA of NA
UNC45B	Cataract	AD	99.72	6 of 6
VIM	Cataract Multiple Types	AD	100	4 of 4
VSX2	Microphthalmia With Coloboma, Microphthalmia	AR	100	13 of 13
WFS1	Nuclear Total Cataract, Wolfram Syndrome	AD,AR	99.97	390 of 395

*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

Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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