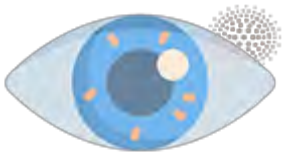
Leber Congenital Amaurosis

Precision Panel



Overview

Leber Congenital Amaurosis (LCA) belongs to the spectrum of early-onset retinal dystrophies. It is a group of monogenic inherited retinal degenerations that show early onset and severe visual dysfunction. Although it is a genetically and phenotypically heterogeneous group of disease it presents in the first few years of life, most often before the age of 1 year and is characterized by wandering nystagmus and reduced vision from birth. Patients may have normal intelligence, but data has demonstrated that as many as 20% develop intellectual disability. The genetic causes of these diseases are due to mutations in genes that play a role in the development and function of the retina and is generally inherited in an autosomal recessive pattern.

The Igenomix Leber Congenital Amaurosis 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 Leber Congenital Amaurosis Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Nystagmus
- Amaurotic pupil response
- Visual loss
- Photophobia
- Cone-shaped cornea
- Strabismus
- Cataract
- Abnormal retinal pigment

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 novel gene therapy and medical care.

- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Detect novel disease-causing genes and novel variant in disease-causing genes.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AIPL1	Leber Congenital Amaurosis, Retinitis Pigmentosa, Cone Rod Dystrophy	AD,AR,X,XR,G	89	82 of 82
CEP290	Bardet-Biedl Syndrome, Joubert Syndrome, Leber Congenital Amaurosis, Meckel Syndrome, Senior-Loken Syndrome, Oculorenal Defect	AR	96.47	293 of 327
CRB1	Leber Congenital Amaurosis, Pigmented Paravenous Chorioretinal Atrophy, Retinitis Pigmentosa, Nanophthalmos	AD,AR,X,G	99.84	365 of 371
CRX	Cone-Rod Dystrophy, Leber Congenital Amaurosis, Retinitis Pigmentosa	AD,AR,X,XR,G	99.91	117 of 117
GDF6	Klippel-Feil Syndrome, Leber Congenital Amaurosis, Microphthalmia, Multiple Synostoses Syndrome	AD,AR,MU,D	98.58	19 of 19
GUCY2D	Choroidal Dystrophy, Leber Congenital Amaurosis, Night Blindness, Cone Rod Dystrophy	AD,AR	99.98	248 of 248
IFT140	Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia, Polydactyly, Jeune Syndrome, Leber Congenital Amaurosis	AR	99.97	81 of 81
IMPDH1	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD	99.98	29 of 29
IQCB1	Senior-Loken Syndrome, Leber Congenital Amaurosis	AR	99.98	43 of 43
KCNJ13	Leber Congenital Amaurosis, Vitreoretinal Degeneration	AD,AR	99.64	11 of 11
LCA5	Leber Congenital Amaurosis	AR	99.67	51 of 52
LRAT	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD,AR,X,XR,G	100	25 of 25
NMNAT1	Leber Congenital Amaurosis, Cone Rod Dystrophy	AR	98.94	72 of 75
PCYT1A	Spondylometaphyseal Dysplasia, Cone-Rod Dystrophy, Leber Congenital Amaurosis	AR	99.98	22 of 22
PRPH2	Choroidal Dystrophy, Macular Dystrophy, Patterned Dystrophy Of Retinal Pigment Epithelium, Retinitis Pigmentosa, Foveomacular Vitelliform Dystrophy, Cone Rod Dystrophy, Retinitis Pigmentosa, Retinitis Punctata Albescens, Stargardt Disease	AD,AR	100	188 of 188
RD3	Leber Congenital Amaurosis	AR	100	13 of 13
RDH12	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD,AR	100	122 of 122
ROM1	Retinitis Pigmentosa	AD,AR,X,XR,G	100	20 of 20
RPE65	Leber Congenital Amaurosis, Retinitis Pigmentosa	AD,AR	100	231 of 231
RPGRIP1	Leber Congenital Amaurosis, Cone Rod Dystrophy, Meckel Syndrome	AR	99.33	146 of 159
SPATA7	Leber Congenital Amaurosis, Retinitis Pigmentosa	AR	97.02	43 of 43
TUBB4B	Leber Congenital Amaurosis, Deafness	AD	100	3 of 3
TULP1	Leber Congenital Amaurosis, Retinitis Pigmentosa	AR	99.9	82 of 82
USP45	Leber Congenital Amaurosis	AR	99.08	4 of 5

*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.

References

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