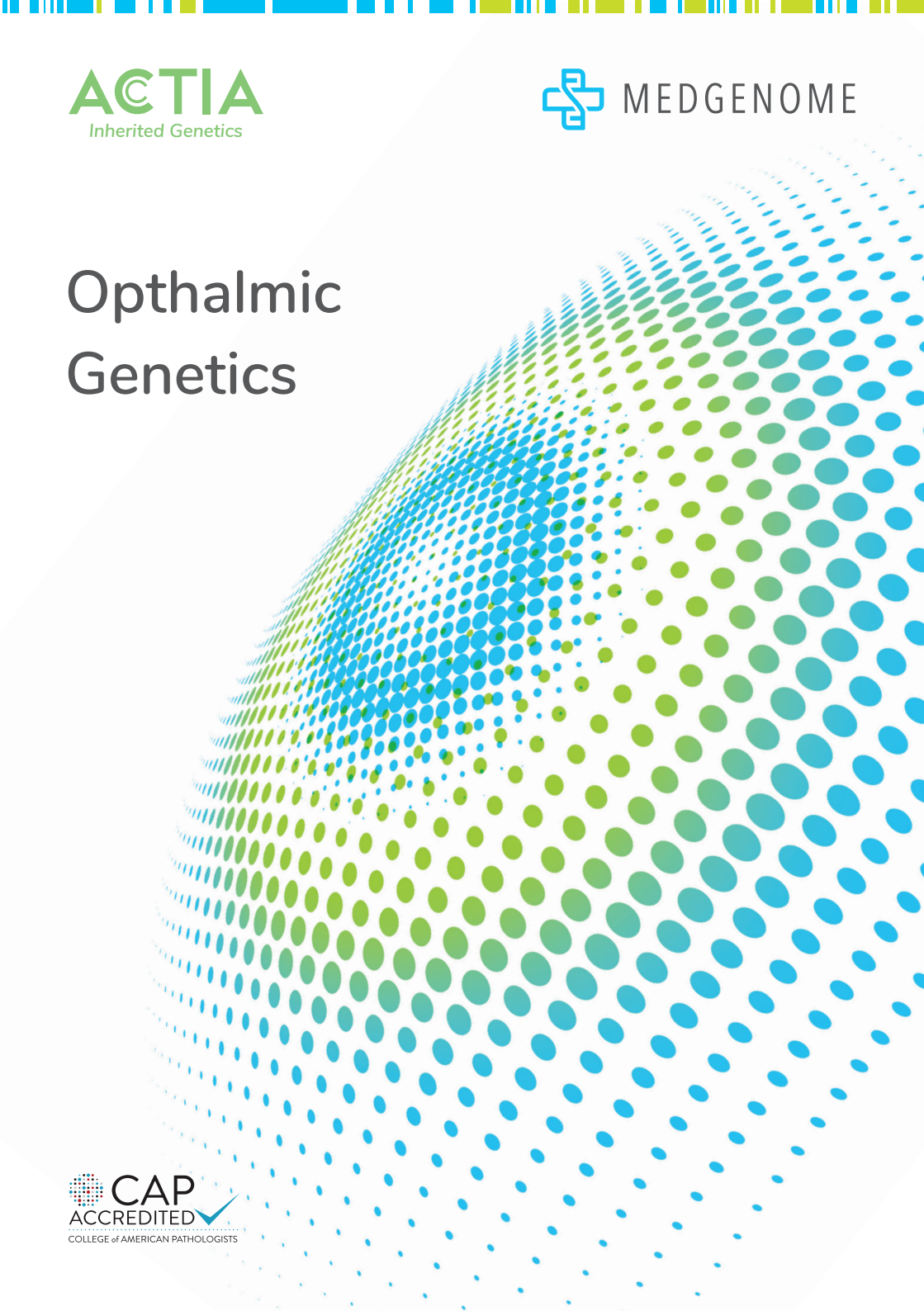
ACTIA
Inherited Genetics

MEDGENOME

Opthalmic Genetics



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What is Opthalmic Genetics?

Of the approximately 5000 genetic diseases and syndromes known to affect humans, at least one-third involve the eye. Due to advances in molecular genetics and sequencing methods, there has been an exponential increase in the knowledge of genetic eye diseases and syndromes.

Prevalence

- *More than 60% of cases of blindness among infants are caused by inherited eye diseases such as congenital cataracts, congenital glaucoma, retinal degeneration, optic atrophy, eye malformations and corneal dystrophies.*

What are the common Genetic Opthalmic Disorders ?

Ophthalmic Disorders can be classified according to the type of genetic abnormality

Monogenic

- *Corneal dystrophies*
- *Oculocutaneous Albinism*
- *Norrie disease*
- *Retinoschisis*
- *Choroideremia*
- *Inherited retinal degenerative diseases (non-syndromic and syndromic)*
- *Inherited optic neuropathies*
- *Colour vision deficiencies*
- *Retinoblastoma*

Developmental

- *Anterior segment dysgenesis*
- *Aniridia*
- *Anophthlamos/ microphthalmos/ nanophthalmos*
- *Coloboma*

Complex

- *Cataract*
- *Keratoconus*
- *Fuchs endothelial corneal dystrophy*
- *Age-related macular degeneration*
- *Glaucoma*
- *Pseudoexfoliation syndrome*
- *Diabetic retinopathy*

Why do you need to test for Genetic Opthalmic Disorders?

- *There is also evidence now that the most common vision problems among children and adults are genetically determined (Eg: strabismus, amblyopia, refractive errors such as myopia, hyperopia and astigmatism)*
- *Genetic ophthalmic disorders include a large number of ocular pathologies which have autosomal dominant, autosomal recessive or X-linked inheritance patterns, or are complex traits with polygenic and environmental components*
- *The presence of a particular ocular sign known to be associated with a specific systemic disease and is often the deciding factor in confirming the diagnosis of that disease, like dislocated eye lens confirms Marfan's syndrome; characteristic red spot in eye is associated with Tay-Sach's diseases; blue sclera suggests Osteogenesis Imperfecta; Aniridia indicates WAGR syndrome*
- *Additionally, most of the ocular genetic disorders like Coloboma, Retinitis Pigmentosa, and Glaucoma show systemic, as well as, non-systemic manifestation*

When do you need to get tested for Genetic Ophthalmic Disorders

Genetic testing as early prescribed can detect the exact underlying defect at molecular level. In-depth clinical information in combination with the identified genetic cause would ideally give an exact diagnosis of a disease condition.

Genetic testing can guide in the treatment of certain genetic eye diseases, where therapies already exist or are in development, such as for Leber congenital amaurosis.

The rarer the disease, the more likely it is caused by variations in a single gene. Therefore, it is appropriate to offer genetic testing to patients whose clinical features indicate a Mendelian disorder for which the causative gene is known.

E.g: 'glaucoma' can be a congenital disease and in some cases may have a later onset.

- Babies with congenital glaucoma – it is recommended to test CYP1B1 and LTBP2
- Children and teens with glaucoma – it is recommended to go with a panel of PITX2, FOXC1, PAX6, LMX1B, and mutations in the MYOC gene if a family history of glaucoma exists
- Adults younger than age 50 with glaucoma and a strong family history may harbour mutations in the MYOC gene
- Patients with optic nerve disease and a family history of normal tension glaucoma but no personal history of elevated intraocular pressure - OPTN gene

Confirmation of a clinical diagnosis through genetic testing will likely change medical management and allows for genetic counseling.

Who needs to get tested?

- Individuals presenting with the symptoms of an eye disorder
- Individuals with a standard preliminary test showing the possibility of an eye disorder
- Individuals with a positive family history
- Individuals without a positive family history but if any individuals in the family with symptoms resembling a specific disease condition
- Prenatal testing is recommended only in families with affected individuals

Why Recommend Actia for Patients with Genetic Opthalmic Disorders?

Actia offers a broad range of pre-designed gene mutation panels which have been developed with in-depth disease understanding of the genetic disorder incorporating the latest research in that particular domain.

New updated technologies, helpful customer service, and clear result interpretation along with counselling sessions with our expert genetic counsellors, make us equipped to provide you the best available support for your patients and families with Genetic Opthalmic Disorders

Actia offers the following Opthalmic gene panel tests

1. Corneal dystrophy gene panel
2. Bardet-Biedl syndrome gene panel
3. Cataract gene panel (congenital/ developmental)
4. Choroideremia - CHM gene - Deletion duplication
5. Congenital stationary night blindness gene panel
6. Leber congenital amaurosis gene panel
7. Microphthalmia and anophthalmia gene panel
8. Optic atrophy gene panel
9. RB1 gene analysis
10. RB1 gene deletion/duplication analysis
11. Retinal Degeneration gene panel
12. Usher syndrome (USH2A) deletion/ duplication analysis
13. Usher syndrome gene panel

Test methodology

Next Generation Sequencing (NGS)

Using genomic DNA extracted from blood, the coding regions of all the genes are captured and sequenced simultaneously by NGS technology on an Illumina platform. The sequence data that is generated is aligned and analyzed for sequence variants.

Multiplex Ligation-dependent Probe Amplification (MLPA)

Deletion and duplication analysis of genomic DNA is carried out by MLPA. This method allows for the amplification of multiple targets with only a single primer pair.

Test sample requirements

Blood (3-5ml in EDTA tubes)

or

Extracted DNA samples
(1µg high quality DNA)

Required forms

- Relevant clinical information including all the clinical presentations and symptoms
- Test request form

Turnaround time

The time taken for generating a clinical report will be maximum of

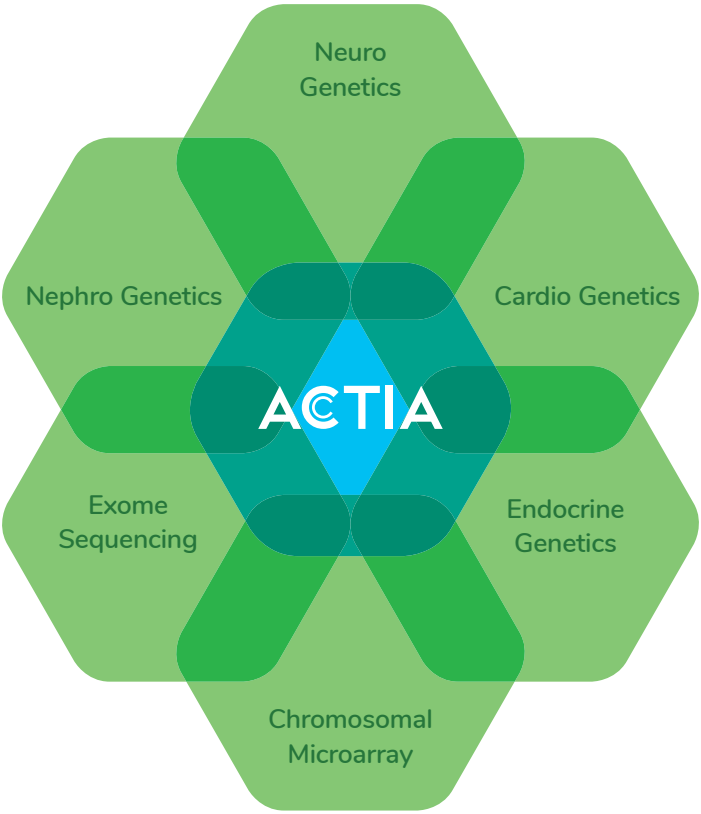
- 6 weeks for NGS
- 3 weeks for MLPA
- 3 weeks for Sanger Sequencing

Free genetic counselling

Actia offers all your patients FREE pre & post test genetic counselling with our expert and certified genetic counsellors.

Best available support for your patients and families via

- Latest technologies
- Helpful customer service
- Clear result interpretation
- Counselling sessions with our Genetic Counsellors



MedGenome Labs Ltd.
3rd Floor, Narayana Netralaya Building,
Narayana Health City, #258/A,
Bommasandra, Hosur Road,
Bangalore – 560099

Toll free no: 1800 103 3691

www.medgenome.com | *diagnostics@medgenome.com*

Bangalore | Chennai | Kochi | Mumbai | Delhi