The Early-Onset Glaucoma Panel is a comprehensive next-generation sequencing (NGS) panel that can be used to confirm a clinical diagnosis of early-onset glaucoma or identify at-risk individuals.

Glucoma is a condition that is characterized by loss of vision as a result of damage to the optic nerves. Damage to the optic nerves is typically caused by increased intraocular pressure, and usually occurs in older adults. The risk for the disease is increased by comorbidities such as a high blood pressure and diabetes. However, some individuals experience early-onset glaucoma, with onset in early childhood or early adulthood, often as a result of a genetic condition. In these individuals, glaucoma may be present in isolation or as part of a syndrome.

METHODOLOGY AND ANALYTICAL SENSITIVITY

Next-generation sequencing technology is used to test clinically relevant portions of each gene, including coding exons, adjacent flanking bases, and selected introns/noncoding variants. Pathogenic and likely pathogenic variants are confirmed by orthogonal methods. Copy number variants, including intragenic deletions and duplications are detected to a resolution of a single exon. To request analysis of a specific single exon copy number variant, please contact our Client Services team prior to ordering. Analytical sensitivity and specificity of the assay is >99%.

INDICATIONS FOR TESTING

- Confirmation of a clinical diagnosis
- Glaucoma with onset in infancy or childhood
- Risk assessment for asymptomatic family members of proband with molecular diagnosis of early onset glaucoma

INCLUDED DISORDERS

This panel includes genes associated with:
- Primary open angle glaucoma
- Primary congenital glaucoma
- Axenfeld-Rieger syndrome
- Nail-Patella syndrome
- Frank-ter Haar syndrome

PREVALENCE

The prevalence of primary congenital glaucoma is 1 in 10,000 to 1 in 20,000 individuals (Papadopoulos et al, 2007), although it may be higher in some populations.

INHERITANCE AND PENETRANCE

Early-onset glaucoma is inherited either in an autosomal recessive or autosomal dominant fashions, and is typically highly penetrant.

CLINICAL SENSITIVITY

In individuals with familial primary congenital glaucoma, causative variants in CYP1B1 can be identified in 20-100% of cases, although only in 10-27% of simplex cases (Abu-Amero and Edward, 2004). Pathogenic changes in FOXC1 and PITX2 are identified in approximately 15-40% of Axenfeld-Rieger syndrome, a condition that often involves glaucoma as a symptom (D’haene et al, 2011).
INCLUDED GENES (13):

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<tr>
<th>COL4A1</th>
<th>FOXC1</th>
<th>LTB2P</th>
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<td>MFRP</td>
<td>OPTN</td>
<td>PITX2</td>
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REFERENCES