The Congenital Cataracts Panel is a comprehensive next-generation sequencing (NGS) panel that can be used to confirm a clinical diagnosis of congenital cataracts or identify at-risk individuals.

Congenital cataracts are lens opacities that are present from birth. Hereditary congenital cataracts are caused by pathogenic changes in genes that encode proteins related to cell transparency. Congenital cataracts may appear in isolation, or as part of a syndrome, and the degree of vision loss is variable between individuals.

**INCLUDED DISORDERS**
This panel includes genes associated with:
- Congenital Cataracts
- Sengers syndrome
- Axenfeld-Rieger syndrome
- Lowe syndrome
- Nance-Horan syndrome
- Branchio-oto-renal syndrome
- Marinesco-Sjogren syndrome
- EDICT syndrome

**PREVALENCE**
The prevalence of congenital cataract is estimated to be 1-15 in 10,000, with 1/4 to 1/3 of cases considered to be hereditary (Yi et al, 2011).

**INHERITANCE AND PENETRANCE**
Congenital cataracts are inherited in autosomal dominant, autosomal recessive, and X-linked fashions. Penetration is typically high.

**CLINICAL SENSITIVITY**
Pathogenic variants can be identified in approximately 40% of congenital cataract cases (Reis et al, 2013; Sun et al 2011). The Congenital Cataracts Panel includes all of the common genetic causes related to this disease.

**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
- Risk assessment for asymptomatic family members of proband with molecular diagnosis of congenital cataracts
INCLUDED GENES (38):

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ADDITIONS TO THE CONGENITAL CATARACTS PANEL:

Emerging Evidence Genes (6):

Emerging evidence genes can also be added to the Congenital Cataracts Panel. These genes do not have a clear association with congenital cataracts, but emerging evidence suggests that they may play a role in disease pathogenesis:

| CHMP4B | CRYBA4 | CRYGB | LIM2 | TDRD7 | VIM |

REFERENCES