

## Leber Congenital Amaurosis: Deletion/Duplication Panel

**Test Code:** MD137

**Turnaround time:** 2 weeks

**CPT Codes:** 81228 x1, 81479 x1

### Condition Description

Leber congenital amaurosis (LCA) is characterized by poor vision beginning between birth or early childhood, nystagmus, an initially normal fundus exam and a nonrecordable electroretinogram (ERG). In addition, other typical findings include defective pupillary responses, photophobia, and the characteristic Franceschetti's oculo-digital sign. Over time, macular coloboma and pigmentary retinopathy may develop. Due to the early manifestation of LCA, other syndromic or nonsyndromic conditions may be incorrectly diagnosed as LCA. LCA is most commonly inherited in an autosomal recessive manner.

**Please note, the *NMNAT1* gene is not included in the NGS panel at this time due to presence of at least four pseudogenes. For clinicians that would like *NMNAT1* analysis in the event that all other genes test negative, we request that you contact EGL directly.**

References:

- Daiger *et al.* (1998) *Invest Ophthalmol Vis Sci* 39:S295.
- OMIM
- GeneReviews

### Genes

[AIPL1](#), [CABP4](#), [CEP290](#), [CNGA3](#), [CNGB3](#), [CRB1](#), [CRX](#), [GNAT2](#), [GUCY2D](#), [IMPDH1](#), [IQCB1](#), [KCNJ13](#), [LCA5](#), [LRAT](#), [OTX2](#), [PDE6C](#), [PDE6H](#), [RD3](#), [RDH12](#), [RPE65](#), [RPGRI1](#), [SPATA7](#), [TULP1](#)

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of Leber congenital amaurosis.
- Carrier testing in adults with a family history of Leber congenital amaurosis.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

### Detection

**Deletion/Duplication:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic pathogenic variants. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

**Submit only 1 of the following specimen types**

#### Type: DNA, Isolated

**Specimen Requirements:**

Microtainer

3µg

Isolation using the Perkin Elmer™ Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

**Specimen Collection and Shipping:**

Refrigerate until time of shipment in 100 ng/µL in TE buffer. Ship sample at room temperature with overnight delivery.

#### Type: Whole Blood (EDTA)

**Specimen Requirements:**

EDTA (Purple Top)

Infants and Young Children ( 2 years of age to 10 years old: 3-5 ml

Older Children & Adults: 5-10 ml

Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**

Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

## Special Instructions

Please include fundus photographs, electroretinogram (ERG) findings, visual field findings, and visual acuity, if available, for expert review and clinical correlation with test results.

## Related Tests

- Eye Disorders: Comprehensive Sequencing and Deletion/Duplication Panels.
- Leber Congenital Amaurosis: Sequencing Panel.