Optic Atrophy: Deletion/Duplication Panel

Test Code: MD234  
Turnaround time: 2 weeks  
CPT Codes: 81228 x1, 81406 x1

**Condition Description**

Optic atrophy is characterized by visual impairment, temporal optic disc pallor, color vision deficits, and centrocecal scotoma of variable density. Optic atrophy may be observed in both a syndromic and nonsyndromic context. Please note, if Leber hereditary optic neuropathy (LHON) is suspected, please order LHON mitochondrial pathogenic variant testing.

References:

- OMIM
- GeneReviews

**Genes**

MFN2, OPA1, OPA3, TIMM8A, TMEM126A, WFS1

**Indications**

This test is indicated for:

- Confirmation of a clinical diagnosis of optic atrophy.
- Carrier testing in adults with a family history of optic atrophy.

**Methodology**

**Deletion/Duplication:** DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which 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 24 hours of collection. Do not refrigerate or 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 Disorder: Comprehensive Sequencing and Deletion/Duplication Panels.
- Optic Atrophy: Sequencing Panel.