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. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient’s phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**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: Whole Blood**

Specimen Requirements:
- In EDTA (purple top) tube:
  - Infants (2 years): 3-5 ml
  - Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:
- In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping:

### Special Instructions

Disclaimer: This information is confidential and subject to change without notice. It may not be reproduced in whole or part unless authorized in writing by an authorized EGL representative.
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.