

Leber Hereditary Optic Neuropathy (LHON) Screening: Targeted Mutation Analysis

Test Code: QC

Turnaround time: 2 weeks

CPT Codes: 81401 x1

Condition Description

Leber Hereditary Optic Neuropathy (LHON) is a mitochondrial disorder characterized by central vision loss, usually permanent, due to atrophy of the optic nerve. LHON presents with acute or subacute painless loss of central vision acuity usually between 12 and 30 years of age. Typical ophthalmoscopic features of acute LHON include circumpapillary telangiectatic microangiopathy and swelling of the nerve fiber layer around the optic disc. Both eyes are usually affected, either concurrently or sequentially. Males are more likely to be affected than females and may be affected at earlier ages. Some females with LHON may develop a multiple sclerosis-like illness. LHON affects 1 in 25,000-33,000 of the Northern European population.

Mutations that cause LHON are found in mitochondrial genes that encode complex I subunits of the respiratory chain which may impair the ability of optic nerve cells to produce ATP. The mitochondrial mutations 11778G>A, 14459G>A, 14484T>C, and 3460G>A account for approximately 95% of LHON mutations. The 11778G>A mutations accounts for approximately 70% of LHON mutations in the Northern European and Australian populations. The 3460G>A, 14459G>A, and 14484T>C mutations each account for approximately 13-14% of LHON mutations.

Indications

This test is indicated for:

- Patients with clinical symptoms of LHON.
- Family members of individuals with LHON.

Methodology

Presence or absence of the mutations (11,778G>A / 3,460G>A / 14,459G>A / 14,484T>C) are detected by Sanger sequencing.

Detection

Over 90% of cases of LHON will be detected by this assay.

Reference Range

Qualitative assay.

Specimen Requirements

Submit only 1 of the following specimen types

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.

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.

Related Tests

[Autosomal Dominant Optic Atrophy \(Kjer\): OPA1 Gene Sequencing \(DL\)](#)