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

**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 telangiectactic 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 (11778G>A / 3460G>A / 14459G>A / 14484T>C) are detected by Sanger pyrosequencing analysis.

**Detection**

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

**Reference Range**

Qualitative assay.

**Specimen Requirements**

**Type: Whole Blood**

Specimen Requirements:

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

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Related Tests**

Autosomal Dominant Optic Atrophy (Kjer): OPA1 Gene Sequencing (DL)