





Pathology and Laboratory Medicine

# Orderable - E-order/Requisition

Turnaround Time: 6 weeks

# **Alternate Name(s):**

LHON



#### Laboratory:

Molecular Diagnostics Lab

## **Specimen:**

Whole blood-2 x 4 mL Lavender EDTA top Vacutainer tube



#### **Requisition:**

MOLECULAR DIAGNOSTIC REQUISITION

### **Collection Information:**

Blood samples <u>must</u> be maintained at room temperature.



### Method of Analysis:

PCR and restriction digest

# **Reference Ranges:**

See report



#### **Test Schedule:**

As required, Monday to Friday 0800-1600 hours

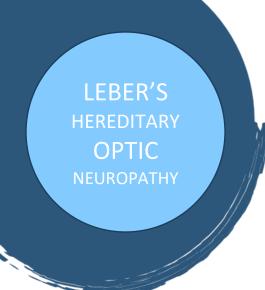
## **Interpretive Comments:**

Please note that these samples will be subjected to direct mutational analysis which may not account for all cases of the disease diagnosed.

Further information will be supplied with the test result.

#### Analysis:

Lebers hereditary optic neuropathy (LHON), is a maternally inherited disease resulting in optic nerve degeneration and cardiac dysrhythmia1. Three mitochondrial DNA point mutations (nt.#3460G>A, nt.#11778G>A and nt.#14484T>C) in the gene coding for subunits ND#1, ND#4 and ND#6 respectively of NADH-CoQ oxidoreductase, are thought to account for the greatest proportion (60-80%) of Leber's cases 1,2,3,4. Further, less common mutations associated with LHON can be found in segments of the mitochondrial genome coding for both cyt b and the subunits ND#1,#2,#5, and #63,4. The decision as to whether other mutations should be screened if the 3460, 11778 and 14484 mutations are absent is dependent on the clinical situation.







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### References

- 1. Wallace et al. Science (1988) 242:1427-1430.
- 2. Newman et al. Am. J. Opthalmol. (1991) III:750-762
- 3. Wallace et al. Science (1992) 256:628-632
- 4. Shoffner et al. Am. J. Hum. Genet. (1992) 51:1179-1186

# **Critical Information Required:**

Pedigree required.

# **Storage and Shipment:**

Must be received within 5 days of collection, shipped at room temperature by courier/overnight delivery.