

LEBER'S
HEREDITARY
OPTIC
NEUROPATHY

Orderable – E-order/Requisition

Turnaround Time: 6 weeks

Alternate Name(s):

LHON

Specimen:

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

Collection Information:

Blood samples must be maintained at room temperature.

Reference Ranges:

See report

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 #6,4. 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.



Laboratory:
Molecular Diagnostics Lab



Requisition:
[MOLECULAR DIAGNOSTIC REQUISITION](#)



Method of Analysis:
PCR and restriction digest



Test Schedule:
As required,
Monday to Friday 0800-
1600 hours



LEBER'S
HEREDITARY
OPTIC
NEUROPATHY

References

1. Wallace et al. *Science* (1988) 242:1427-1430.
2. Newman et al. *Am. J. Ophthalmol.* (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.