HEREDITARY
SENSORY
NEUROPATHY

London Health Sciences Centre
St JOSEPH'S HEALTH CARE LONDON
Pathology and Laboratory Medicine

Orderable – E-order/Requisition

Turnaround Time: 3 months

Laboratory:
Molecular Diagnostics Lab

Requisition:
MOLECULAR DIAGNOSTIC REQUISITION

Method of Analysis:
DNA Sequence Analysis

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

Alternate Name(s):

HSN1

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:

Hereditary sensory neuropathy type I (HSN1) is the most common hereditary disorder of peripheral sensory neurons, and is an autosomal dominant condition resulting in progressive degeneration of dorsal root ganglia and motor neurons with onset in the second or third decades. Mutations in SPTLC1, encoding serine palmitoyltransferase, long chain base subunit-1 have been shown to be responsible for this condition1,2. Early reports in the literature suggest that mutations both in exon 5 (C133Y and C133W)1 and exon 6 of SPTLC1 (V144D)1 can be responsible for this condition.

References

1. Dawkins JL et al (2001) Mutations in SPTLC1, encoding serine palmitoyltransferase, long chain base subunit-1, cause hereditary sensory neuropathy type I. Nat Genet 2001 Mar;27(3):309-12
2. Bejaoui K et al (2001) SPTLC1 is mutated in hereditary sensory neuropathy, type 1 Nat Genet 2001 Mar;27(3):261-2

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Critical Information Required:

Pedigree required

Storage and Shipment:

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