**HEREDITARY SENSORY NEUROPATHY**

**Orderable – E-order/Requisition**

**Turnaround Time:** 3 months

**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**

**Critical Information Required:**

Pedigree required

**Storage and Shipment:**

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