MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: March 10, 2021
Effective Date: March 23, 2021

Hereditary Sensory and Autonomic Neuropathy, Type I, Serum

Test ID: HSAN1

Useful for:

Diagnosis of patients with clinical features suggestive of hereditary sensory and autonomic neuropathy, type I caused by variants in *SPTLC1* and *SPTLC2*

Monitoring of patients with hereditary sensory and autonomic neuropathy, type I caused by variants in *SPTLC1* and *SPLTC2*

Genetics Information:

Deoxysphingolipids (dSLs) are elevated in patients with hereditary sensory and autonomic neuropathy type I (HSAN1) due to variants in *SPTLC1* and *SPTLC2*, and are useful to support a diagnosis of HSAN1.

Elevations in dSLs may also be seen in patients with order disorders including type 2 diabetes, metabolic syndrome, mitochondrial disease, glycogen storage disease type I, and possibly disorders of serine biosynthesis.

Additional testing is required to determine the specific cause of elevated dSLs.

Methods:

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Reference Values:

Sphinganine: < or =18.0 ng/mL
1-deoxysphinganine: < or =0.25 ng/mL
1-deoxymethylsphinganine: < or =0.04 ng/mL
Sphingosine: < or =80.0 ng/mL
1-deoxysphingosine: < or =0.05 ng/mL
1-deoxymethylsphingosine: < or =0.09 ng/mL

Specimen Requirements:

Patient Preparation:	Fasting 8 hours
Collection Container/Tube:	
Preferred:	Serum gel
Acceptable:	Red top
Submission Container/Tube:	Plastic Vial
Specimen Volume:	1 mL
Minimum Volume:	0.5 mL

Specimen Stability Information:

Specimen Type	Temperature	Time
Serum	Frozen (preferred)	90 days
	Refrigerate	24 hours

Cautions:

This assay is not intended to but may detect neuropathies other than hereditary sensory and autonomic neuropathy, type I.

CPT Code:

82542

Day(s) Setup: Varies **Analytic Time:** Varies

Questions

Contact Melissa Tricker-Klar, Laboratory Technologist Resource Coordinator at 800-533-1710.