Orderable – E-order/Requisition
Turnaround Time: 4-6 weeks  STAT: 4 weeks

Alternate Name(s):
CMT
Charcot Marie Tooth Neuropathy

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:
Charcot-Marie-Tooth (CMT) disease is a genetically and clinically heterogeneous group of inherited disorders of the peripheral nervous system characterized by progressive loss of muscle tissue and touch sensation across various parts of the body. CMT type 1 (CMT1) is a demyelinating peripheral neuropathy characterized by distal muscle weakness and atrophy, sensory loss, and slow nerve conduction velocity. CMT type 2 (CMT2) is an axonal (non-demyelinating) peripheral neuropathy characterized by distal muscle weakness and atrophy, mild sensory loss, and normal or near-normal nerve conduction velocities. Most common mutation is associated with CMT1A (70%-80% of all CMT1) and involves duplication of PMP22, while PMP22 gene deletion is the most common cause (80%) of hereditary neuropathy with liability to pressure palsies (HNPP). Mutations in remaining genes are associated with less frequent subtypes of CMT, including autosomal dominant, recessive and X-linked forms of the disease.
CHARCOT MARIE TOOTH DISEASE

**Specimen Stability:**

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

MLPA copy number analysis. All variants interpreted as either ACMG category 1, 2, or 3 (pathogenic, likely pathogenic, VUS; PMID: 25741868) are confirmed using Sanger sequencing, MLPA, or other assays. ACMG category 4 and 5 variants (likely benign, benign) are not reported, but are available upon request. This assay has been validated at a level of sensitivity equivalent to the Sanger sequencing and standard copy number analysis (>99%; PMID: 27376475, 28818680).

**Test Schedule:**

As required, Monday to Friday 0800-1600 hours.