FAMILIAL AMYLOIDOTIC POLYNEUROPATHY – TTR

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

Alternate Name(s):
FAP
TTR

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:
Familial Amyloidotic Polyneuropathy (FAP) is a neurodegenerative disorder characterized by extracellular deposition of transthyretin (TTR) amyloid fibrils, particularly in the peripheral nervous system (PMID:11569892, PMID:8095302). A number of mis-sense mutations in the human prealbumin gene have been directly linked to FAP.

Storage and Shipment:
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