FAMILIAL AMYLOIDOTIC POLYNEUROPATHY – TTR

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

Orderable – E-order/Requisition

Turnaround Time: 4-6 weeks **STAT: 4 weeks**

Laboratory:
Molecular Diagnostics Lab

Requisition:
MOLECULAR DIAGNOSTIC REQUISITION

Method of Analysis:
All coding exons and 20 bp of flanking intronic sequence are enriched using an LHSC custom targeted hybridization protocol (Roche Nimblegen), followed by high throughput sequencing (Illumina). Sequence variants and copy number changes are assessed and interpreted using clinically validated algorithms and commercial software (SoftGenetics: Nextgene, Geneticist Assistant, Mutation Surveyor; and Alamut Visual). All exons have >300x mean read depth coverage, with a minimum 100x coverage at a single nucleotide resolution. This assay meets the sensitivity and specificity of combined Sanger sequencing and

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.

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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