Hereditary Periodic Fever Syndromes: Sequencing Panel

Test Code: MM180
Turnaround time: 6 weeks
CPT Codes: 81402 x1, 81479 x1

Condition Description

Hereditary periodic fever syndromes (HPFS) is a group of familial autoinflammatory disorders with heterogeneous genetic causes. There are seven types of HPFS that have been characterized. Clinical features continue to be used reliably to assign patients to this general disease category. Identification of the precise genetic defect is important to permit carrier testing and early prenatal diagnosis. Molecular analysis is likely to expand the clinical spectrum of HPFS and may also provide data relevant to prognosis and future therapeutic intervention. HPFS is a very rare disorder. Although each HPFS presents with particular symptoms, globally they share intermittent, apparently unprovoked episodes of fever and inflammation. HPFS can be inherited in an autosomal dominant or autosomal recessive manner.

References:
- OMIM

Genes
ELANE, LPIN2, MEFV, MKK, NLRP3, PSTPIP1, TNFRSF1A

Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of hereditary periodic fever syndromes.
- Carrier testing in adults with a family history of hereditary periodic fever syndromes.

Methodology

Next Generation Sequencing: In-solution hybridization of all coding exons is performed on the patient's genomic DNA. Although some deep intronic regions may also be analyzed, this assay is not meant to interrogate most promoter regions, deep intronic regions, or other regulatory elements, and does not detect single or multi-exon deletions or duplications. Direct sequencing of the captured regions is performed using next generation sequencing. The patient's gene sequences are then compared to a standard reference sequence. Potentially causative variants and areas of low coverage are Sanger-sequenced. Sequence variations are classified as pathogenic, likely pathogenic, benign, likely benign, or variants of unknown significance. Variants of unknown significance may require further studies of the patient and/or family members.

Detection

Clinical Sensitivity: Unknown. Pathogenic variants in the promoter region, some pathogenic variants in the introns and other regulatory element pathogenic variants cannot be detected by this analysis. Large deletions will not be detected by this analysis. Results of molecular analysis should be interpreted in the context of the patient's clinical and/or biochemical phenotype.

Analytical Sensitivity: ~99%

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood
Specimen Requirements:
In EDTA (purple top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

Type: Isolated DNA
Specimen Requirements:
In microtainer: 60 ug
Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight
Related Tests

- Hereditary Periodic Fever Syndromes: Deletion/Duplication Panel