Hereditary Periodic Fever Syndromes: Deletion/Duplication Panel

Test Code: MD180
Turnaround time: 2 weeks
CPT Codes: 81228 x1

Condition Description

Hereditary periodic fever syndromes (HPFS) is a group of familiar 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, MVK, 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

Deletion/Duplication Analysis: DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region. Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

Detection

Deletion/Duplication Analysis: Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

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: 10 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 delivery.

Related Tests

- Hereditary Periodic Fever Syndromes: Sequencing Panel