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 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, MK, 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.

**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 (EDTA)**

**Specimen Requirements:**
- EDTA (Purple Top)
- Infants and Young Children (2 years of age to 10 years old): 3-5 ml
- Older Children & Adults: 5-10 ml
- Autopsy: 2-3 ml unclotted cord or cardiac blood

**Specimen Collection and Shipping:**
Ship sample at room temperature for receipt at EGL within 72 hours of collection. Do not freeze.

**Type: DNA, Isolated**

**Specimen Requirements:**
- Microtainer
- 3µg
- Isolation using the Perkin Elmer™Chemagen™ Chemagen™ Automated Extraction method or Qiagen™ Puregene kit for DNA extraction is recommended.

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

### Related Tests
- Hereditary Periodic Fever Syndromes: Sequencing Panel