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Patient Report

Patient Age/Gender: 1 day Male

Specimen Collected: 26-Oct-21 11:30

Familial Mediterranean Fever by NGS | **Received: 01-Nov-21 15:38** **Report/Verified: 01-Nov-21 15:52**

Procedure	Result	Units	Reference Interval
FMF Specimen	Whole Blood		
FMF Interp	See Note [i1]		

Test Information

i1: FMF Interp

BACKGROUND INFORMATION: Familial Mediterranean Fever (MEFV) Sequencing

CHARACTERISTICS: Familial Mediterranean fever (FMF) is a genetic condition characterized by recurrent but short-lived attacks of fever, abdominal pain, joint pain, and/or skin rashes. Symptoms and frequency of these attacks are highly variable. Renal amyloidosis is another common complication in untreated individuals and may be the only manifestation in some patients.

CAUSE: Pathogenic germline variants in the MEFV gene

INHERITANCE: Autosomal recessive, although some heterozygous individuals may have symptoms

CLINICAL SENSITIVITY: 75-90%

GENE TESTED: MEFV (NM_000243)

METHODOLOGY: Probe hybridization-based capture of all coding exons and exon-intron junctions of the targeted MEFV gene, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and to confirm reported variants that do not meet acceptable quality metrics. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions (indels) from 1-10 base pairs in size. Indels greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced.

LIMITATIONS: A negative result does not exclude a diagnosis of familial Mediterranean fever. This test only detects variants within the coding regions and intron-exon boundaries of the MEFV gene. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Regulatory region variants and deep intronic variants will not be identified. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations caused by the presence of pseudogenes, repetitive, or

Test Information

i1: FMF Interp

homologous regions. This test is not intended to detect low-level mosaic or somatic variants, gene conversion events, complex inversions, translocations, mitochondrial DNA (mtDNA) mutations, or repeat expansions. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.