

Client: ARUP Example Report Only  
500 Chipeta Way  
Salt Lake City, UT 84108  
UNITED STATES

Physician: arup, arup

**Patient: FMF NGS, EXAMPLE**

**DOB**

**Sex:** Female

**Patient Identifiers:** 36417

**Visit Number (FIN):** 36736

**Collection Date:** 2/23/2022 07:23

**Familial Mediterranean Fever (MEFV) Sequencing**

ARUP test code 3004434

FMF Specimen	whole Blood
FMF Interp	<p>Negative</p> <p>INDICATION FOR TESTING Not provided</p> <p>RESULT No pathogenic variants were detected in the MEFV gene.</p> <p>INTERPRETATION No pathogenic variants were identified in the MEFV gene by massively parallel sequencing of the coding regions and exon-intron boundaries. This result decreases the likelihood of, but does not exclude, a diagnosis of familial Mediterranean fever (FMF). Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.</p> <p>RECOMMENDATIONS Medical management should rely on clinical findings and family history. If concern for a periodic fever syndrome remains, consideration should be given to ordering the Periodic Fever Syndromes Gene Panel (ARUP test code 2007370). Genetic consultation is suggested.</p> <p>COMMENTS Likely benign and benign variants are not included in this report.</p> <p>This result has been reviewed and approved by [REDACTED]</p> <p>BACKGROUND INFORMATION: Familial Mediterranean Fever (MEFV) Sequencing</p> <p>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.</p> <p>CAUSE: Pathogenic germline variants in the MEFV gene</p> <p>INHERITANCE: Autosomal recessive, although some heterozygous individuals may have symptoms</p> <p>CLINICAL SENSITIVITY: 75-90%</p>

**H=High, L=Low, \*=Abnormal, C=Critical**

Unless otherwise indicated, testing performed at:

**ARUP LABORATORIES | 800-522-2787 | aruplab.com**  
500 Chipeta Way, Salt Lake City, UT 84108-1221  
Tracy I. George, MD, Laboratory Director

Patient: FMF NGS, EXAMPLE  
ARUP Accession: 22-054-100580  
Patient Identifiers: 36417  
Visit Number (FIN): 36736  
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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 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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
FMF Specimen	22-054-100580	2/23/2022 7:23:00 AM	2/23/2022 7 24:05 AM	2/23/2022 7:26 00 AM
FMF Interp	22-054-100580	2/23/2022 7:23:00 AM	2/23/2022 7 24:05 AM	2/23/2022 7:26 00 AM

END OF CHART

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