

Client: Example Client ABC123  
123 Test Drive  
Salt Lake City, UT 84108  
UNITED STATES

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 6/5/1965  
**Gender:** Female  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**Familial Mediterranean Fever (MEFV) Sequencing**

ARUP test code 2002658

FMF FGS Specimen whole Blood

Familial Mediterranean Fever (MEFV) Int Negative

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at [www.aruplab.com](http://www.aruplab.com). Incidental findings are not reported unless clinically significant but are available upon request.

TEST PERFORMED - 2002658  
TEST DESCRIPTION - Familial Mediterranean Fever (MEFV) Sequencing  
INDICATION FOR TEST - Not Provided

**RESULT**  
No pathogenic variants were detected in the MEFV gene.

**INTERPRETATION**  
No pathogenic MEFV gene variants were detected by sequencing the coding region and intron-exon boundaries. This individual is likely not a carrier of, nor is affected with, familial Mediterranean fever (FMF). However, a diagnosis of FMF has not been excluded, since this individual may have a pathogenic MEFV variant that is not detected by this assay.

**RECOMMENDATIONS**  
Medical management should rely on clinical findings and family history. If the etiology for the patient's symptoms remains unclear, consideration should be given to ordering the Periodic Fever Syndromes Gene Panel (ARUP test code 2007370). Genetic consultation is suggested.

**COMMENTS**  
Reference Sequence: GenBank # NM\_000243.2 (MEFV)  
Nucleotide numbering begins at the "A" of the ATG initiation codon.  
Likely benign and benign variants are not included in this report, but are available upon request.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.

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

Background Information for Familial Mediterranean Fever (MEFV) Sequencing:

Characteristics: Recurrent episodes of inflammation, fever, abdominal pain, chest pain, joint pain, skin eruptions and the development of renal amyloidosis.  
Prevalence: 1 in 1,000 worldwide.  
Inheritance: Primarily autosomal recessive; some activating mutations appear to be autosomal dominant.  
Cause: Pathogenic MEFV gene mutations.  
Clinical Sensitivity: Approximately 80 percent.  
Methodology: Bidirectional sequencing of the entire MEFV coding region and intron-exon boundaries.  
Analytical Sensitivity and Specificity: 99 percent.  
Limitations: Diagnostic errors can occur due to rare sequence variations. Regulatory region, intronic mutations and large deletions/duplications will not be detected.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at [www.aruplab.com](http://www.aruplab.com).

See Compliance Statement C: [www.aruplab.com/CS](http://www.aruplab.com/CS)

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
FMF FGS Specimen	20-225-400589	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Familial Mediterranean Fever (MEFV) Int	20-225-400589	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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