

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 5/1/1981
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 3004434

FMF Specimen whole Blood

FMF Interp

Negative

RESULT

No pathogenic variants were detected in the MEFV gene.

INTERPRETATION

No pathogenic variants were detected in the MEFV gene. 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 methodology and limitations of this test.

RECOMMENDATIONS

Medical management should rely on clinical findings and family history. If this individual has a family history, determination of a causative familial variant in an affected family member is necessary for optimal interpretation of this negative result. Further testing may be warranted if there is a familial variant that is not detectable by this assay. 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 recommended.

COMMENTS

Likely benign and benign variants are not reported. Variants in the following region(s) may not be detected by NGS with sufficient confidence in this sample due to technical limitations: None

This result has been reviewed and approved by [REDACTED]

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

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 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.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
FMF Specimen	23-357-103639	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
FMF Interp	23-357-103639	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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 23-357-103639
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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