Warde Medical Laboratory

LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 10/04/2023 15:02 Received: 10/04/2023 15:02

Test Name	Result	Flag	Ref-Ranges	Units	Site
Familial Medit Fever Mutation	**See Below**	**AB**			QCRL

RESULT: HETEROZYGOUS POSITIVE FOR THE M694V VARIANT IN THE MEFV GENE

Interpretation: DNA testing has demonstrated that this individual is positive for one copy of the M694V variant in the pyrin (MEFV) gene. Therefore, this individual is predicted to be at least a carrier of Familial Mediterranean Fever (FMF). This test cannot rule out the potential presence of a rare variant in the MEFV gene or a clinical diagnosis of FMF. Clinical correlation and genetic counseling are recommended.

Laboratory testing supervised and results monitored by

Familial Mediterranean Fever (FMF) is an autosomal recessive disorder characterized by recurrent episodes of fever and peritonitis with pain. FMF is most common in non-Ashkenazi Jewish, Armenian, Arab, and Turkish populations and has a carrier frequency as high as 1 in 3 to 1 in 7 in these populations. The carrier frequency of FMF in Ashkenazi-Jews is also high (up to 1 in 5) but FMF is not common in this population due to the predominance of a variant for a mild form of FMF (p.Glu148Gln, E148Q). FMF is caused by pathogenic variants in the pyrin (MEFV) gene on chromosome 16p13. This assay analyzes 10 MEFV variants: p.Phe479Leu (p.F479L, c.1437 C>G), p.Met680Ile (p.M680I, c.2040G>C or A), p.Ile692del (p.I692del, c.2074-2076del), p.Met694Val (p.M694V, c.2080A>G), p.Met694Ile (p.M694I, c.2082G>A), p.Lys695Arg (p.K695R, c.2084A>G), p.Val726Ala (p.V726A, c.2177T>C), p.Ala744Ser (p.A744S, c.2230G>T), and p.Arg761His (p.R761H, c.2282G>A) and two low penetrance variants (p.Glu148Gln (p.E148Q, c.442 G>C) and p.Pro369Ser (p.P369S, c.1105C>T)) that account for approximately 80%-90% of FMF pathogenic variants in Mediterranean populations (about 70% in the Arab population).

The 12 pathogenic variants listed above are detected by polymerase chain reaction (PCR) amplification of portions of the Mediterranean Fever (MEFV) gene (NM_000243), single nucleotide primer extension, and detection of fluorescent primer extension products on an automated capillary DNA analyzer. Although rare, false positive or false negative results may occur. All results should be interpreted in the context of clinical findings, relevant history, and other laboratory data. Health care providers may also contact your local Quest Diagnostics' genetic counselor or call for assistance with the interpretation of these
results.

This test is performed pursuant to a license agreement with Orchid Biosciences Inc.

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

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This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols Institute San Juan Capistrano. It has not been cleared or approved by the FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.
Reviewed and signed by Laboratory testing supervised and results monitored by Tina Hambuch-Hawks, Ph.D., DABMGG, CGMB, Signed on
Test Performed at:
Quest Diagnostics Nichols Institute
33608 Ortega Highway
San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

Performing Site:
QCRL: QUEST DIAGNOSTICS REFERENCE LAB CAPISTRANO 33608 Ortega Highway San Juan Capistrano CA 92675

Reported Date: 2023.10.04 15:02

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

F404000035
WX0000003827
Printed D&T: 10/04/23 15:02

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002365

Kajal V. Sitwala, MD, PhD - Medical Director
Form: MM RL1