

Using Genetic Testing for Diagnosis and Risk-Assessment for Familial Mediterranean Fever (FMF)

Indications:

- Suspicion of FMF based on clinical observation
- Favorable response to colchicine
- History of FMF in first-degree relatives
- Known familial mutation in *MEFV*

Benefits:

Genetic testing for FMF

- Can confirm a suspected clinical diagnosis of FMF and identify the familial mutation(s).
- Can identify presymptomatic family members of patients, enabling preventative treatment.
- Can distinguish presymptomatic family members from heterozygous carriers, who do not need and should not receive preventative treatment.

Background:

- FMF is an autosomal recessive disorder affecting Mediterranean populations (Arabs, Armenians, Jews, Turks).¹ Prevalence ranges from 1 in 400 to 1 in 1,000 persons.²
- Type 1 FMF is characterized by short, recurrent episodes of fever, inflammation, or serositis, which can lead to amyloidosis and renal failure in some patients. In type 2 FMF, amyloidosis is the first clinical manifestation.³
- Treatment with colchicine decreases inflammatory attacks and can prevent renal amyloidosis, especially if started early.⁴
- FMF has been linked to mutations in the gene *MEFV*.¹
- Genetic testing can confirm a diagnosis of FMF during the early disease stage or in asymptomatic individuals and facilitate prophylactic treatment.

References: 1. O'Neill MJ (2010) OMIM 249100. 2. Ben-Chetrit E, Touitou I (2009) *Arthritis Rheum* 61(10): 1447. 3. Shohat M, Halpern GJ (2009) *GeneReviews* 4. Haghigat M et al. (2006) *SEMJ* 7(2).

Ordering Information: Please see other side.

Ordering Information for Familial Mediterranean Fever (FMF) Testing

Indications for Testing

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Ordering Information for Single Gene Tests

Gene(s)	Test Code
<i>MEFV</i>	252797

Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

**For test information, sample requirements, or to request a sample shipping kit,
please contact Client Services at 1-866-647-0735 or visit us on the web at
www.correlagen.com.**