

UCLA DIAGNOSTIC MOLECULAR PATHOLOGY LABORATORY
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Familial Mediterranean Fever Mutation

↑CPT

83890; 83900; 83901 (x2); 88384; 83912

↑Synonyms

FMF; MEFV Mutation

↑Test Includes

Identification of common mutations in the MEFV gene

↑Laboratory

Molecular Pathology

↑Availability

Monday-Friday, 0700-1700

↑Turnaround Time

3-28 days

↑Specimen

Whole blood

↑Volume

4 mL

↑Container

Lavender top (EDTA) tube

↑ Storage Instructions

All specimens should be sent to the Laboratory immediately after collection, preferably by overnight delivery. Specimens should be kept at room temperature or refrigerated but not frozen.

↑ Causes for Rejection

Blood samples frozen and thawed will yield low quality DNA; specimens inadequately identified

↑ Reference Range

No MEFV mutation detected

↑ Use

Familial Mediterranean fever (FMF) is an inherited disorder. It is transmitted in an autosomal recessive pattern and affects mostly ethnic groups living around the Mediterranean basin: Jews, Armenians, Turks, Arabs, etc. The frequency of the disease gene in these populations is very high, with a carrier rate of about 1/7 in North African Jews and Armenians. It is characterized by recurring bouts of fever, most commonly with severe abdominal pain due to peritonitis. Attacks can also include arthritis and pleurisy. Colchicine was successfully introduced in 1972 as a prophylactic therapy for eliminating FMF attacks and preventing amyloidosis, which could otherwise lead to renal failure in FMF patients.

↑ Limitations

FMF primarily affects North African Jews, Armenians, Turks, and Arabs, in which a founder effect has been demonstrated. Eight founder-effect mutations were identified in exon 10, each segregating with one ancestral haplotype. Besides these eight mutations, additional mutations have been observed in exon 2, exon 5, and exon 10 of the FMF gene. The panel of 12 mutations offered in this test will pick up 80% to 90% of Mediterranean carriers, but less in other ethnic groups. Thus, a negative result in this test does not rule out a diagnosis of FMF, and a finding of only one mutation may be supportive of the diagnosis in the face of relevant clinical symptoms.

↑ Methodology

Reverse Hybridization Blot. DNA is isolated and amplified with biotinylated primers. Amplicons are hybridized to a strip that contains specific probes for E148Q, P369S, F479L, M680I (G>C and G>A), I692del, M694V, M694I, K645R, V726A, A744S, and R761H mutations and corresponding wild-type. After hybridization, the arrays are washed and then incubated with conjugate and substrate. A purple color indicates hybridization of patient DNA to each specific probe target.