Familial Mediterranean Fever

Familial Mediterranean fever (FMF) is a genetic disorder. It is characterized by recurring short episodes of fever, as well as inflammation in the abdomen, chest, and joints. In some cases, there may be abnormal protein buildup in the kidneys. People with FMF most often have two variants in the MEFV gene.

Overview Scientific Details

Jamie, you **do not have the variants** we tested.

You could still have a variant not covered by this test.

How To Use This Test

This test does not diagnose any health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Carrier Status tutorial
See Scientific Details

Intended Uses

- Tests for **multiple variants** in the MEFV gene.
- To identify carrier status for FMF.
- Informs individuals with one variant or certain combinations of variants in the MEFV gene that they may be at risk for developing symptoms of FMF.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of the M680I, M694I, M694V, or R761H variant.

Important Ethnicities

- This test is most relevant for people of **Arab**, **Armenian**, **Sephardic Jewish**, and **Turkish** descent.

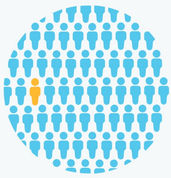
You are likely not a carrier.

This result is most relevant for people of Arab, Armenian, Sephardic Jewish, and Turkish descent.

We ruled out the most common variants for FMF in people of Arab, Armenian, Sephardic Jewish, and Turkish descent.

You still have a chance of being a carrier for FMF.

We cannot estimate your chances because sufficient data is not available.



About Familial Mediterranean Fever

Also known as: FMF, Benign Paroxysmal Peritonitis, Recurrent Polyserositis

When symptoms develop

FMF can develop anytime from early childhood to adulthood. For most people with the condition, the first episode occurs before the age of 20.

Typical signs and symptoms

- Periodic episodes of fever
- Inflammation in the abdomen, chest, and joints
- Skin rash
- Abnormal protein buildup in the kidneys

Ethnicities most affected

This condition is most common in people of Arab, Armenian, Sephardic Jewish, and Turkish descent.

How it's treated

During a fever episode, anti-inflammatory drugs may be used to manage fever and inflammation. In addition, doctors often prescribe certain medications to prevent fever attacks and kidney damage, especially for people who have the M694V variant.

Read more at: MedlinePlus GeneReviews Mayo Clinic National Organization for Rare Disorders (NORD)

Consider talking to a healthcare professional if you are concerned about your results.

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Learn more

Share your results with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

Learn more

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Overview Scientific Details

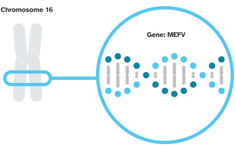
FMF is caused by variants in the MEFV gene.

MEFV

The MEFV gene contains instructions for making a protein called pyrin. The pyrin protein plays a role in the body's immune response. Certain variants in the MEFV gene prevent the protein from functioning properly, which can trigger an inappropriate immune response, leading to symptoms like fever or inflammation.

Read more at MedlinePlus

Chromosome 16



You have no variants detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*		Additional Information
M680I **Gene:** MEFV **Marker:** rs28940580	C Typical copy from one of your parents	C Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 5, 6, 7, 20] | ClinVar*
M694I **Gene:** MEFV **Marker:** rs28940578	C Typical copy from one of your parents	C Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 6, 7, 9, 20] | ClinVar*
M694V **Gene:** MEFV **Marker:** rs61752717	T Typical copy from one of your parents	T Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 6, 7, 9, 20] | ClinVar*
K695R **Gene:** MEFV **Marker:** rs104895094	T Typical copy from one of your parents	T Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [6, 7, 9, 16] | ClinVar*
V726A **Gene:** MEFV **Marker:** rs28940579	A Typical copy from one of your parents	A Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 6, 7, 9, 20] | ClinVar*
A744S **Gene:** MEFV **Marker:** rs61732674	C Typical copy from one of your parents	C Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 6, 9, 19] | ClinVar*
R761H **Gene:** MEFV **Marker:** rs104895097	C Typical copy from one of your parents	C Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 6, 9] | ClinVar*

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

Post-test carrier risk for FMF is the chance of still being a carrier for the condition if you do not have the variants tested. This chance depends on how common it is to be a carrier for FMF and whether the variants we tested tend to be found in people of your ethnicity.

Because you do not have the variants we tested, your chances of still being a carrier are lower than for someone who has not been tested. However, we cannot provide an exact estimate because the information needed to calculate post-test carrier risk is not available for your ethnicity.

Test Details

Indications for Use

The 23andMe PGS Carrier Status Report for Familial Mediterranean Fever is indicated for the detection of seven variants in the MEFV gene. This test is intended to be used to determine carrier status for FMF in adults. This report also describes if a result is associated with personal risk for developing symptoms of FMF, but it does not describe a person's overall risk of developing symptoms. This test is most relevant for people of Arab, Armenian, Sephardic Jewish, and Turkish descent.

Special Considerations

- There are currently no professional guidelines in the U.S. for carrier testing for this condition.
- The E148Q variant is one of five founder variants commonly observed in ethnic groups originating from the Mediterranean basin, such as Arabs, Armenians, Sephardic Jews, and Turks. This variant is not included in this test because it is currently considered a variant of uncertain significance.
- Symptoms of FMF may vary between people with the condition even if they have the same genetic variants.
- In some cases, people with only a single MEFV variant can experience symptoms of FMF. In addition, some studies have identified individuals who meet clinical criteria for FMF but do not have any MEFV variants.

Test Performance Summary

Carrier Detection Rate & Relevant Ethnicities

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified by this test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

Arab	71-96%	[2, 7, 8, 15, 16]
Armenian	92%	[9]
Sephardic Jewish	75-89%	[3, 7, 12, 19]
Turkish	72-92%	[1, 4, 6, 11, 13, 21, 22]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

1. Akin H et al. (2010). "MEFV mutations in patients with Familial Mediterranean Fever from the Aegean region of Turkey." Mol Biol Rep. 37(1):93–8.
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8. Jarjour RA. (2010). "Familial Mediterranean fever in Syrian patients: MEFV gene mutations and genotype-phenotype correlation." Mol Biol Rep. 37(1):1-5.
9. Kriegshäuser G et al. (2018). "Clinical and genetic heterogeneity in a large cohort of Armenian patients with late-onset familial Mediterranean fever." Genet Med.
10. Manna R et al. (2019). "Familial Mediterranean Fever: Assessing the Overall Clinical Impact and Formulating Treatment Plans." Mediterr J Hematol Infect Dis. 11(1):e2019027.

See all references

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
Nov. 3, 2021	The carrier detection rate range was updated for Turkish.
Dec. 17, 2018	Familial Mediterranean Fever report created.