

# Single Gene Disorders - Autosomal Dominant Inheritance

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Familial Mediterranean fever

## Familial Mediterranean fever

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### Description

Familial Mediterranean fever is an inherited condition characterized by recurrent episodes of painful inflammation in the abdomen, chest, or joints. These episodes are often accompanied by fever and sometimes a rash or headache. Occasionally inflammation may occur in other parts of the body, such as the heart; the membrane surrounding the brain and spinal cord; and in males, the testicles. In about half of affected individuals, attacks are preceded by mild signs and symptoms known as a prodrome. Prodromal symptoms include mildly uncomfortable sensations in the area that will later become inflamed, or more general feelings of discomfort. The first episode of illness in familial Mediterranean fever usually occurs in childhood or the teenage years, but in some cases, the initial attack occurs much later in life. Typically, episodes last 12 to 72 hours and can vary in severity. Episodes generally occur once a month, and in affected women of reproductive age, attacks often correspond with menstruation or ovulation. However, the length of time between episodes can range from days to years. During these periods, affected individuals usually have no signs or symptoms related to the condition. However, without treatment to help prevent attacks and

complications, a buildup of protein deposits (amyloidosis) in the body's organs and tissues may occur, especially in the kidneys, which can lead to kidney failure.

## Frequency

Familial Mediterranean fever primarily affects populations originating in the Mediterranean region, particularly people of Armenian, Arab, Turkish, or Jewish ancestry. The disorder affects 1 in 200 to 1,000 people in these populations. It is less common in other populations.

## Causes

Familial Mediterranean fever is caused by variants (also known as mutations) in the MEFV gene. The MEFV gene provides instructions for making a protein called pyrin (also known as marennosin), which is found in white blood cells. This protein is involved in the immune system, helping to regulate the process of inflammation. Inflammation occurs when the immune system sends signaling molecules and white blood cells to a site of injury or disease to fight microbial invaders and facilitate tissue repair. When this process is complete, the body stops the inflammatory response to prevent damage to its own cells and tissues. Variants in the MEFV gene reduce the activity of the pyrin protein, which disrupts control of the inflammation process. An inappropriate or prolonged inflammatory response can result, leading to fever and pain in the abdomen, chest, or joints. Normal variations in the SAA1 gene may modify the course of familial Mediterranean fever. Some evidence suggests that a particular version of the SAA1 gene (called the alpha variant) increases the risk of amyloidosis among people with familial Mediterranean fever.

Learn more about the genes associated with Familial Mediterranean fever

MEFV

SAA1

### Inheritance

Familial Mediterranean fever is usually inherited in an autosomal recessive pattern, which means both copies of the MEFV gene in each cell have variants. The parents of an individual with an autosomal recessive condition each carry one copy of the altered gene, but they typically do not show signs and symptoms of the condition. Research shows that about one-third of people with familial Mediterranean fever have a single MEFV gene variant, though most experts believe there is a second variant somewhere else in the gene that has not been detected by genetic testing. In rare cases, this condition appears to be inherited in an autosomal dominant pattern. An autosomal dominant inheritance pattern describes cases in which one copy of the altered gene in each cell is sufficient to cause the disorder. In autosomal dominant inheritance, affected individuals often inherit the variant from one affected parent. However, another mechanism is believed to account for some cases of familial Mediterranean fever that were originally thought to be inherited in an autosomal dominant pattern. A gene variant that occurs frequently in a population may result in a disorder with autosomal recessive inheritance appearing in multiple generations in a family, a pattern that mimics autosomal dominant inheritance. If one parent has familial Mediterranean fever (with variants in both copies of the MEFV gene in each cell) and the other parent is an unaffected carrier (with a variant in one copy of the MEFV gene in each cell), it may appear as if the affected

child inherited the disorder only from the affected parent. This appearance of autosomal dominant inheritance when the pattern is actually autosomal recessive is called pseudodominance.

#### Other Names for This Condition

Benign paroxysmal peritonitis Familial paroxysmal polyserositis FMF MEF Recurrent polyserositis Reimann periodic disease Siegal-Cattan-Mamou disease Wolff periodic disease

#### Additional Information & Resources

#### Genetic Testing Information

Genetic Testing Registry: Familial Mediterranean fever

Genetic Testing Registry: Familial mediterranean fever, autosomal dominant

Genetic and Rare Diseases Information Center

Familial Mediterranean fever

Patient Support and Advocacy Resources

National Organization for Rare Disorders (NORD)

Clinical Trials

ClinicalTrials.gov

Catalog of Genes and Diseases from OMIM

FAMILIAL MEDITERRANEAN FEVER, AUTOSOMAL DOMINANT

## FAMILIAL MEDITERRANEAN FEVER; FMF

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