

# MEFV Gene

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## 1. Introduction

The *MEFV* gene provides instructions for making a protein called pyrin (also known as marenostrin). Although pyrin's function is not fully understood, it likely assists in keeping the inflammation process under control. 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 has been accomplished, the body stops the inflammatory response to prevent damage to its own cells and tissues.

Pyrin is produced in certain white blood cells (neutrophils, eosinophils, and monocytes) that play a role in inflammation and in fighting infection. Pyrin may direct the migration of white blood cells to sites of inflammation and stop or slow the inflammatory response when it is no longer needed. Pyrin also interacts with other molecules involved in fighting infection and in the inflammatory response. Research indicates that pyrin helps regulate inflammation by interacting with the cytoskeleton, the structural framework that helps to define the shape, size, and movement of a cell.

## 2. Health Conditions Related to Genetic Changes

### 2.1. Familial Mediterranean fever

More than 80 *MEFV* gene mutations that cause familial Mediterranean fever have been identified. A few mutations delete small amounts of DNA from the *MEFV* gene, which can lead to an abnormally small, nonfunctional protein. Most *MEFV* gene mutations, however, change one of the protein building blocks (amino acids) used to make pyrin. The most common mutation replaces the amino acid methionine with the amino acid valine at protein position 694 (written as Met694Val or M694V). Among people with familial Mediterranean fever, this particular mutation is also associated with an increased risk of developing amyloidosis, a complication in which abnormal protein deposits can lead to kidney failure. Some evidence suggests that variations in another gene, called *SAA1*, can further modify the risk of developing amyloidosis among people with the M694V mutation.

*MEFV* gene mutations lead to reduced amounts of pyrin or a malformed pyrin protein that cannot function properly. As a result, pyrin cannot perform its presumed role in controlling inflammation, leading to an inappropriate or prolonged inflammatory response. Fever and inflammation in the abdomen, chest, joints, or skin are signs of familial Mediterranean fever.

## 3. Other Names for This Gene

- FMF
  - marenostrin
  - Mediterranean fever
  - MEF
  - MEFV\_HUMAN
  - MRST
  - pyrin
  - TRIM20
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