GENE Seeker

You know the risk of your child being a carrier of a genetic disease.

Familial Mediterranean fever

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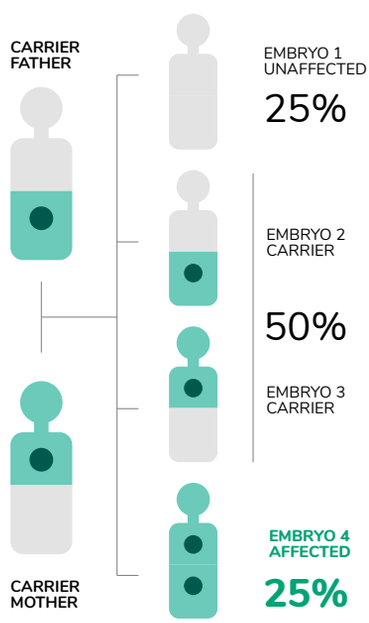
What is Familial Mediterranean fever?

Familial Mediterranean fever (FMF) is an autoinflammatory disorder characterized by recurrent short episodes of fever and serositis resulting in pain in the abdomen, chest, joints and muscles.

Disease onset usually occurs before the age of 30 with an earlier onset corresponding to a more severe phenotype. FMF can be divided into 2 types: FMF type 1 and 2. Type 1 is characterized by attacks (as often as once a week or every few years) of fever and serositis lasting 1-4 days and resolving spontaneously. Stress, exposure to cold, fat-rich meals, infections, certain drugs and menstrual cycles are possible attack triggers. Mild symptoms (myalgia, headache, nausea, dyspnea, arthralgia, low back pain, asthenia and anxiety) precede attacks and last about 17 hours. Attacks manifest as fever (38°C-40°C lasting 12-72 hours and not responding to antibiotics), diffuse or localized abdominal pain (often mimicking acute abdomen), constipation (diarrhea in children), arthralgias (in large joints), arthritis (in upper/lower limb/knee joints) and chest pain caused by pleuritis and/or pericarditis (see this term). In 7-40% of patients cutaneous involvement is also present. Amyloidosis type AA (see this term) can be a serious long term complication. FMF type 2 describes a phenotype where amyloidosis occurs as the first and only manifestation of the disease.



What is the next step if I'm a carrier of Familial Mediterranean fever?

If you are found to be a carrier of Familial Mediterranean fever, it is important that your partner be tested for the same genetic disorder.

What if my partner is not a carrier?

If your partner's test for Familial Mediterranean fever negative, the chance to have an affected child is low. However there is currently no test able to detect all existing mutations, so there is always a residual risk that the person who has done the test is a carrier of other less frequent mutations.

What if both me and my partner are carriers of Familial Mediterranean fever?

When both parents are carriers of Familial Mediterranean fever, the probability of having a child with Familial Mediterranean fever is 25%.

We recommend that you discuss your results with your doctor or genetic counselor in order to know more about reproductive options.

If both you and your partner are carriers, speak with your doctor or genetic counselor about reproductive options.

1 frequency — **65** people

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