What Your Test Results Mean

Testing results indicate that you are a carrier of familial Mediterranean fever. Carriers typically show no symptoms of familial Mediterranean fever; however, carriers are at an increased risk of having a child with familial Mediterranean fever. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor.

Familial Mediterranean Fever Explained

Familial Mediterranean fever is an inherited disorder characterized by recurrent episodes of painful inflammation in the abdomen, chest, joints, and sometimes the heart, the membrane surrounding the brain and spinal cord, and in males, the testicles. An attack of inflammation usually lasts 12 to 72 hours, depending on severity. Affected individuals also experience fever, rashes, and headaches. Those with familial Mediterranean fever do not typically experience symptoms until childhood or the teenage years. In some cases, symptoms do not appear until adulthood.

If familial Mediterranean fever is not treated, abnormal protein may accumulate in the body and cause organ damage. Kidney damage or failure may occur due to amyloidosis; infertility may be caused due to inflammation of the reproductive organs; and arthritis may result in the knees, ankles and hips. Treatment generally involves colchicine, a drug that helps to reduce inflammation, or other drugs that may be used to help prevent inflammation.

How the Genetics Work

Familial Mediterranean fever is an autosomal recessive disorder caused by mutations in the MEFV gene. All individuals have two copies of the MEFV gene. Carriers of familial Mediterranean fever have a mutation in one copy of the MEFV gene while individuals with familial Mediterranean fever have mutations in both copies of the MEFV gene, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?

Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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