Familial Mediterranean Fever

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of familial Mediterranean fever. Carriers may show mild, treatable symptoms of the disorder. You and your partner or donor would both have to be carriers of familial Mediterranean fever for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

We recommend that you share this information with all of your health care providers. Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning and their own personal clinical management.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for familial Mediterranean fever, each of your children has a 1 in 4 (25%) chance to have the condition.

Familial Mediterranean Fever Explained

What is Familial Mediterranean Fever?

Familial Mediterranean fever is an inherited disorder that causes recurrent episodes of painful inflammation in the abdomen, chest, joints, the membrane surrounding the brain and spinal cord, and other parts of the body. These episodes typically last 12 to 72 hours, depending on severity, and include fever, rashes, and headaches. Affected individuals are usually symptom-free between episodes. These episodes usually begin in childhood, adolescence, or adulthood. If familial Mediterranean fever is not treated, abnormal protein can accumulate in the body and cause organ damage. Kidney damage or failure can occur due to amyloidosis. Other symptoms of familial Mediterranean fever can include infertility due to inflammation of the reproductive organs and arthritis in the knees, ankles, and hips.

Many carriers for familial Mediterranean fever are asymptomatic their entire lives. However, studies have shown that some carriers can have symptoms ranging from classic to mild familial Mediterranean fever.

Prognosis

A normal, possibly symptom-free life span is expected with early and regular treatment. Familial Mediterranean fever can only be life-threatening if left untreated or if a person is not responsive to treatment.

Treatment

Treatment involves colchicine, a drug that helps reduce inflammation, or other drugs that may be used to help prevent inflammation.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/familial-mediterranean-fever
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/familial-mediterranean-fever/
National Society of Genetic Counselors
https://www.nsgc.org/

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Revised: 4/2022