What Your Test Results Mean

Testing results indicate that you are a carrier of HFE-associated hereditary hemochromatosis (HFE-HH). Carriers typically show no symptoms of HFE-HH; however, carriers are at an increased risk of having a child with HFE-HH. 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.

HFE-Associated Hereditary Hemochromatosis Explained

HFE-HH is an inherited condition that causes the body to absorb too much iron from the diet. The excess iron is stored in the body’s tissues and organs, particularly the skin, heart, liver, pancreas, and joints. Excess iron cannot be excreted properly, thus causing damage to tissues and organs. Early symptoms may include fatigue, joint pain, abdominal pain, and loss of sex drive. Later signs and symptoms may include arthritis, liver disease, diabetes, heart abnormalities, and skin discoloration. In rare cases, symptoms may began before birth, resulting in liver damage that is apparent at birth or within the first few days of life. Treatment includes therapeutic phlebotomy, iron chelation therapy, and dietary changes.

How the Genetics Work

HFE-HH is an autosomal recessive disorder caused by mutations of the HFE gene. In general, individuals have two copies of the HFE gene. Carriers have a single mutation in one copy of the HFE gene while individuals with HFE-HH have mutations in both copies of HFE, one inherited from each parent. Risk for two carriers to have a child with HFE-HH is 25%; however, HFE-HH is very common in the European population and risk is 50% if one parent is affected and the other is a carrier.

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.