# **Genetic Insights:** quick reference guide for healthcare providers

This guide is intended to facilitate a discussion between a provider and their patient.

## Genetic Insights test results: **Hereditary hemochromatosis - carrier**

### Key results

A DNA variant associated with being a carrier of *HFE*-associated hemochromatosis was found in the *HFE* gene.

People with *HFE*-associated hemochromatosis, also known as hereditary hemochromatosis, have a increased risk of iron overload. Carriers do not have the condition and are likely not at increased risk for iron overload.

### Next steps

| Clinical recommendations | Resources |
|---|---|
| *Genetic Insights is a screening test and is not intended for diagnosis.*<br><br>*It is important to review your patient's personal and family health history for increased incidence or early onset of health conditions, including hereditary hemochromatosis, for accurate risk assessment.* | **Have questions?**<br><br>Call **1.866.GENE.INFO** (1.866.436.3463) to speak to a specialized Quest genetic counselor or geneticist available to healthcare providers to discuss test selection and results. |
| *Consider referring your patient to a genetic counselor if they have questions.*<br><br>*Genetic counselors can provide counseling on the implications of this test result and next steps for your patient.* | Your patient can contact a genetic counselor through their online Genetic Insights dashboard at no additional cost.<br><br>To find a genetic counselor practicing in your patient's area for an in-person session, please visit FindAGeneticCounselor.NSGC.org |

**Patient conversation starters:**

Patient conversation starters summarize the preceding information in plain language to support meaningful conversations between you and your patient.

FindAGeneticCounselor.NSGC.org

Your Genetic Insights test is a screening test. Your result indicates you are a carrier for hereditary hemochromatosis. As a next step, let's review your personal and family health history and address any questions or concerns you might have.

You may want to talk with a genetic counselor. Genetic counselors are experts in genetics and can help you understand this result.

You can access a genetic counselor through your Genetic Insights dashboard at no additional cost to you, and we can discuss a referral to a local genetic counselor.

**Visit QuestDiagnostics.com/Genetic-Health-Screening for more information about this test.**

## What is hereditary hemochromatosis?

*HFE*-associated hemochromatosis (also known as hereditary hemochromatosis) is an inherited condition characterized by increased absorption of dietary iron which may lead to organ damage, including liver cirrhosis, if left untreated.[1]

Not all people with hereditary hemochromatosis will develop iron overload or associated symptoms and complications.[1] Treatment is available for those who do.[2,3]

The most common type of hereditary hemochromatosis is caused by a DNA variant in each copy of the *HFE* gene.[1]

People who are carriers have a variant in just 1 copy of the *HFE* gene and are most likely not at increased risk for iron overload due to *HFE*-associated hemochromatosis.[1,2]

Regardless of this result, personal and family history are important risk factors for iron overload.[1,2]

**Patient conversation starters:**

Hereditary hemochromatosis is an inherited condition that can cause too much iron in the blood and body. If not treated, the iron can build up over time and can cause damage to certain organs.

Hereditary hemochromatosis is caused by having a DNA variant in each copy of the *HFE* gene.

People who have a DNA variant in only 1 copy of the *HFE* gene are carriers of the condition. Carriers do not have the condition and are unlikely to be at increased risk for iron buildup.

## What this result means for family members

*HFE*-associated hemochromatosis is an autosomal recessive condition. Family members may have the same DNA variant. The variant was most likely inherited from a parent who is at least a carrier of *HFE*-associated hemochromatosis. Siblings and other relatives may also be carriers.

People who are carriers can pass the variant on to their children. If the carrier's partner (or sperm/egg donor) is also a carrier of *HFE*-associated hemochromatosis, then each child they have has a 1 in 4 chance of having the condition.

People who are carriers may wish to share their test results with family members so they may consider discussing hereditary hemochromatosis with their healthcare provider.

**Patient conversation starters:**

Hereditary hemochromatosis runs in families.

That means your family members may have the same DNA variant in their *HFE* gene. You most likely inherited this variant from 1 of your parents. Your siblings may also be carriers.

You may want to share this result with your family members so they can consider talking to their doctor about hereditary hemochromatosis.

## Additional resources

The following advocacy groups have additional information and resources about hereditary hemochromatosis:

**American Liver Foundation:** LiverFoundation.org
**Iron Disorders Institute, Hemochromatosis.org:** Hemochromatosis.org

### References


1. Barton JC, Edwards CQ. HFE Hemochromatosis. April 3, 2000. Updated December 6, 2018. In: Adam MP, Ardinger HH, Pagon RA, et al, editors. GeneReviews® [Internet]. University of Washington, Seattle; 1993-2022. https://www.ncbi.nlm.nih.gov/books/NBK1440/
2. Bacon BR, Adams PC, Kowdley KV, Powell LW, Tavill AS. Diagnosis and Management of Hemochromatosis: 2011 Practice Guideline by the American Association for the Study of Liver Diseases. *Hepatology*. 2011;54(1):328-343. doi:10.1002/hep.24330
3. Kowdley KV, Brown KE, Ahn J, Sundaram V. ACG Clinical Guideline: Hereditary Hemochromatosis. *Am J Gastroenterol*. 2019;114(8):1202-1218. Published correction: *Am J Gastroenterol*. 2019;114(12):1927. doi:10.14309/ajg.0000000000000315