Health Library

Hereditary Hemochromatosis

What is hemochromatosis?

Hereditary hemochromatosis is one of the most common genetic disorders in the U.S. It is passed down from parents through their genes. This disorder causes your body to absorb too much iron from the food you eat. Iron is a mineral found in many foods. But too much iron is toxic to your body. The excess iron is stored in your body's tissues and organs. Over time, the iron builds up in your body (iron overload). It may damage your tissues and organs. Too much iron in your heart, liver, pancreas, and pituitary can cause severe problems.

There are other types of hemochromatosis. They include:

- **Juvenile hemochromatosis.** This affects teens and young adults ages 15 to 30. It leads to a severe iron overload. This can cause heart and liver disease.
- **Neonatal hemochromatosis.** In this type, iron builds up very quickly in a baby's liver before birth. This can cause severe organ damage.

- **Transfusion-related hemochromatosis.** This type occurs in people with certain blood disorders who need many blood transfusions. Over time, these transfusions can cause iron overload.

What causes hemochromatosis?

Hemochromatosis is a genetic disease. This means it is passed down from parents through their genes. It is most common in whites whose families are from Northern Europe. Men and women are equally affected by the disease.

You may be born with this condition if you inherit two hemochromatosis genes, one from each parent. If you have only one of these genes, you are called a carrier of the gene. You don't have symptoms. But you have a greater chance of having a child with this disease.

If parents without hemochromatosis have a child with the disorder, there is a 25% chance that any additional child may be born with the disease. First-degree relatives of people with hemochromatosis should be screened for the disorder.

It's more common for men with this condition to have too much iron. Men also tend to show symptoms at a younger age than women. This is likely because women lose iron each month when they have their period.

What are the symptoms of hemochromatosis?

Each person's symptoms may vary. Symptoms may include:

- Lack of energy (lethargy) and weakness
- Irritability
- Depression
- Joint pain
- Bronze or yellowish skin color
- Loss of body hair
- Impotence in men
- In women, not having a period
- Infection

Untreated or severe hemochromatosis may lead to the following:

- Liver function problems and an enlarged liver
- Abnormal heart rhythm
- Heart failure
- Enlarged spleen
- Diabetes

Symptoms may look like other health problems. Always see your healthcare provider for a diagnosis.

How is hemochromatosis diagnosed?

This disease is usually found through a routine blood test. Your provider will take your medical history and give you a physical exam. You may also have 1 or more of these tests:

- **Iron levels.** People with hemochromatosis have higher levels of iron in their blood.

- **Transferrin saturation (TS) test.** This blood test measures the percentage of transferrin and other proteins that have too much iron. It is helpful in finding the disease early.
- **Ferritin levels.** Ferritin is a protein in the blood. It increases when iron levels in the body increase. It rises most significantly when iron levels are very high.
- **Liver biopsy.** A small sample of liver tissue or cells is removed and checked under a microscope.
- **Genetic testing.** This blood test looks for the gene changes that cause hereditary hemochromatosis.

How is hemochromatosis treated?

Your healthcare provider will create a treatment plan based on:

- Your age, overall health, and medical history
- How sick you are
- How well you handle certain medicines, treatments, or therapies
- If your condition is expected to get worse
- Your opinion or preference

Treatment may include:

- **Phlebotomy.** This procedure removes blood from your body. This is done regularly at first, until iron levels return to normal. Then it can be done once or twice a year as needed.
- **Chelation therapy.** This treatment uses medicine to remove iron from your body.
- **Avoiding iron and vitamin C supplements**
- **Avoiding too much alcohol**
- **Treatment of the resulting diseases or conditions**

If your iron levels return to normal before any organs are damaged, you can live a normal lifespan with this disorder.

What are the complications of hemochromatosis?

If not treated, hemochromatosis can lead to:

- Liver function problems and an enlarged liver
- Abnormal heart rhythm
- Heart failure
- Enlarged spleen
- Diabetes

Living with hemochromatosis

Hemochromatosis is a life-long condition. It can cause problems if iron levels in the blood are not kept at normal levels. Because of this, regular treatment with phlebotomy or chelation therapy is needed to reduce iron levels. Work with your healthcare provider to check and manage your iron levels. You should avoid iron and vitamin C supplements. You should not drink too much alcohol.

Key points about hemochromatosis

- Hereditary hemochromatosis is one of the most common genetic disorders in the U.S
- It causes your body to absorb too much iron from the food you eat.

- The excess iron is stored in body tissues and organs. Over time it builds up and may damage tissues and organs.
- Early symptoms may include lethargy and weakness, irritability, depression, joint pain, yellowish skin, and loss of body hair.
- Regular treatment with phlebotomy or chelation therapy is needed to reduce iron levels.

Next steps

Tips to help you get the most from a visit to your healthcare provider:

- Know the reason for your visit and what you want to happen.
- Before your visit, write down questions you want answered.
- Bring someone with you to help you ask questions and remember what your provider tells you.
- At the visit, write down the name of a new diagnosis, and any new medicines, treatments, or tests. Also write down any new instructions your provider gives you.
- Know why a new medicine or treatment is prescribed, and how it will help you. Also know what the side effects are.
- Ask if your condition can be treated in other ways.
- Know why a test or procedure is recommended and what the results could mean.
- Know what to expect if you do not take the medicine or have the test or procedure.
- If you have a follow-up appointment, write down the date, time, and purpose for that visit.
- Know how you can contact your provider if you have questions.

Centers, Programs & Services

- Huntsman Cancer Institute

Related Documents

Drug Reference

- Deferasirox

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