



Hemochromatosis

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Hemochromatosis, the most common form of iron overload disease, is an inherited disorder that causes the body to absorb and store too much iron. The extra iron builds up in organs and damages them. Without treatment, the disease can cause these organs to fail.

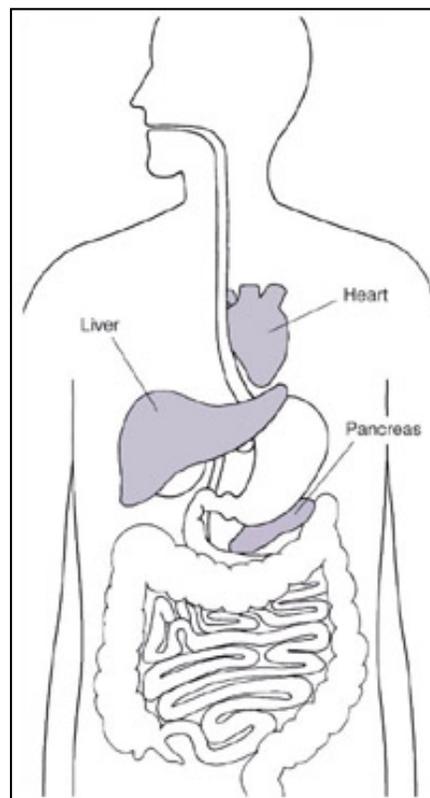
Iron is an essential nutrient found in many foods. The greatest amount is found in red meat and iron-fortified bread and cereal. In the body, iron becomes part of hemoglobin, a molecule in the blood that transports oxygen from the lungs to all body tissues.

Healthy people usually absorb about 10 percent of the iron contained in the food they eat to meet the body needs. People with hemochromatosis absorb more than the body needs. The body has no natural way to rid itself of the excess iron, so it is stored in body tissues, especially the liver, heart, and pancreas.

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Causes

Genetic or hereditary hemochromatosis is mainly associated with a defect in a gene called *HFE*, which helps regulate the amount of iron absorbed from food. There are two known important mutations in *HFE*, named C282Y and H63D. C282Y is the most important. When C282Y is inherited from both parents, iron is overabsorbed from the diet and hemochromatosis can result. H63D usually causes little increase in iron absorption, but a person with H63D from one parent and C282Y from the other may



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rarely develop hemochromatosis.

The genetic defect of hemochromatosis is present at birth, but symptoms rarely appear before adulthood. A person who inherits the defective gene from both parents may develop hemochromatosis. A person who inherits the defective gene from only one parent is a carrier for the disease but usually does not develop it. However, carriers might have a slight increase in iron absorption.

Scientists hope that further study of *HFE* will reveal how the body normally metabolizes iron. They also want to learn how iron injures cells and whether it contributes to organ damage in other diseases, such as alcoholic liver disease, hepatitis C, porphyria cutanea tarda, heart disease, reproductive disorders, cancer, autoimmune hepatitis, diabetes, and joint disease. Scientists are also working to find out why only some patients with *HFE* mutations get the disease.

Juvenile hemochromatosis and neonatal hemochromatosis are two forms of the disease that are not caused by an *HFE* defect. Their cause is unknown. The juvenile form leads to severe iron overload and liver and heart disease in adolescents and young adults between the ages of 15 and 30, and the neonatal form causes the same problems in newborn infants.

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Risk Factors

Hereditary hemochromatosis is one of the most common genetic disorders in the United States. It most often affects Caucasians of Northern European descent, although other ethnic groups are also affected. About 5 people in 1,000 (0.5 percent) of the U.S. Caucasian population carry two copies of the hemochromatosis gene and are susceptible to developing the disease. One person in 8 to 12 is a carrier of the abnormal gene. Hemochromatosis is less common in African Americans, Asian Americans, Hispanic Americans, and American Indians.

Although both men and women can inherit the gene defect, men are about five times more likely to be diagnosed with the effects of hereditary hemochromatosis than women. Men also tend to develop problems from the excess iron at a younger age.

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Symptoms

Joint pain is the most common complaint of people with hemochromatosis. Other common symptoms include fatigue, lack of energy, abdominal pain, loss of sex drive, and heart problems. Symptoms tend to occur in men between the ages of 30 and 50 and in women over age 50. However, many people have no symptoms when they are diagnosed.

If the disease is not detected early and treated, iron may accumulate in body tissues and may eventually lead to serious problems such as

- arthritis
- liver disease, including an enlarged liver, cirrhosis, cancer, and liver failure
- damage to the pancreas, possibly causing diabetes

- heart abnormalities, such as irregular heart rhythms or congestive heart failure
- impotence
- early menopause
- abnormal pigmentation of the skin, making it look gray or bronze
- thyroid deficiency
- damage to the adrenal gland

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Diagnosis

A thorough medical history, a physical examination, and routine blood tests help rule out other conditions that could be causing the symptoms. This information often provides helpful clues, such as a family history of arthritis or unexplained liver disease.

Blood tests can determine whether the amount of iron stored in the body is too high. The transferrin saturation test determines how much iron is bound to the protein that carries iron in the blood. The total iron binding capacity (TIBC) test measures how well your blood can transport iron. The serum ferritin test shows the level of iron in the liver. If either of these tests shows higher than normal levels of iron in the body, doctors can order a special blood test to detect the *HFE* mutation, which will help confirm the diagnosis. (If the mutation is not present, hereditary hemochromatosis is not the reason for the iron buildup, and the doctor will look for other causes.) A liver biopsy, in which a tiny piece of liver tissue is removed and examined under a microscope, may be needed. It will show how much iron has accumulated in the liver and whether the liver is damaged.

Hemochromatosis is often undiagnosed and untreated. It is considered rare and doctors may not think to test for it. The initial symptoms can be diverse and vague and can mimic the symptoms of many other diseases. Also, doctors may focus on the conditions caused by hemochromatosis—arthritis, liver disease, heart disease, or diabetes—rather than on the underlying iron overload. However, if the iron overload caused by hemochromatosis is diagnosed and treated before organ damage has occurred, a person can live a normal, healthy life.

Hemochromatosis is usually treated by a specialist in liver disorders (hepatologist), digestive disorders (gastroenterologist), or blood disorders (hematologist). Because of the other problems associated with hemochromatosis, several other specialists may be on the treatment team, such as an endocrinologist, cardiologist, or rheumatologist. Internists or family practitioners can also treat the disease.

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Treatment

Treatment is simple, inexpensive, and safe. The first step is to rid the body of excess iron. The process is called phlebotomy, which means removing blood the same way it is drawn from donors at blood banks. Depending on how severe the iron overload is, a pint of blood will be taken once or twice a week for several months to a year, and occasionally longer. Blood ferritin levels will be tested periodically to monitor iron levels. The goal is to bring blood ferritin levels to the low end of normal and keep them there. Depending on the lab, that means 25 to 50 micrograms of ferritin per liter

of serum. Depending on the amount of iron overload at diagnosis, reaching normal levels can take many phlebotomies.

Once iron levels return to normal, maintenance therapy, which involves giving a pint of blood every 2 to 4 months for life, begins. Some people may need it more often. An annual blood ferritin test will help determine how often blood should be removed.

The earlier hemochromatosis is diagnosed and treated in appropriate cases, the better. If treatment begins before any organs are damaged, associated conditions—such as liver disease, heart disease, arthritis, and diabetes—can be prevented. The outlook for people who already have these conditions at diagnosis depends on the degree of organ damage. For example, treating hemochromatosis can stop the progression of liver disease in its early stages, which means a normal life expectancy. However, if cirrhosis has developed, the person's risk of developing liver cancer increases, even if iron stores are reduced to normal levels. Appropriate regular follow-up with a specialist is necessary.

People who have complications of hemochromatosis may want to consider getting treatment from a specialized hemochromatosis center. These centers are located throughout the country. Information is available from the organizations listed under [For More Information](#).

People with hemochromatosis should not take iron supplements. Those who have liver damage should not drink alcoholic beverages because they may further damage the liver.

Although treatment cannot cure the conditions associated with established hemochromatosis, it will help most of them. The main exception is arthritis, which does not improve even after excess iron is removed.

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Tests for Hemochromatosis

Screening for hemochromatosis (testing people who have no symptoms) is not a routine part of medical care or checkups. However, researchers and public health officials do have some suggestions:

- Brothers and sisters of people who have hemochromatosis should have their blood tested to see if they have the disease or are carriers.
- Parents, children, and other close relatives of people who have the disease should consider testing.
- Doctors should consider testing people who have joint disease, severe and continuing fatigue, heart disease, elevated liver enzymes, impotence, and diabetes, because these conditions may result from hemochromatosis.

Since the genetic defect is common and early detection and treatment are so effective, some researchers and education and advocacy groups have suggested that widespread screening for hemochromatosis would be cost-effective and should be conducted. However, a simple, inexpensive, and accurate test for routine screening does not yet exist, and the available options have limitations. For example, the genetic test provides a definitive diagnosis, but it is expensive. The blood test for transferrin saturation is widely available and relatively inexpensive, but it may have to be done twice with careful handling to confirm a diagnosis and to show that it is the

consequence of iron overload.

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Hope Through Research

Current research in hemochromatosis is concentrated in four areas:

Genetics. Scientists are working to understand more about how the *HFE* gene normally regulates iron levels and why not everyone with an abnormal pair of genes develops the disease.

Pathogenesis. Scientists are studying how iron injures body cells. Iron is an essential nutrient, but above a certain level it can damage or even kill the cell.

Epidemiology. Research is under way to explain why the amounts of iron people normally store in their bodies differ. Research is also being conducted to determine how many people with the defective *HFE* gene go on to develop symptoms, as well as why some people develop symptoms and others do not.

Screening and testing. Scientists are working to determine at what age testing is most effective, which groups should be tested, and what the best tests for widespread screening are.

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For More Information

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