CONDITION

Hemochromatosis

Hemochromatosis is a condition in which excess iron builds up and may damage organs.

WHAT IT IS
Excess iron buildup in the body

BODY SYSTEM
Liver, joints, heart, pancreas, and hormone system

MAIN SYMPTOMS
Fatigue, joint pain, abdominal pain, skin darkening

WHAT CAUSES IT
Inherited genes or repeated blood transfusions

HOW IT IS DIAGNOSED
Blood tests, genetic testing, and sometimes liver imaging

MAIN TREATMENT
Regular blood removal or iron-chelating medicine

CAN IT BE CURED?
No, but iron overload can be controlled

IS IT AN EMERGENCY?
Sometimes, with chest pain or sudden confusion

LONG-TERM OUTLOOK
Often good when treated before organ damage

Overview

Hemochromatosis is a condition in which too much iron builds up in the body. Your body needs iron to make healthy red blood cells, but it has no simple way to remove large amounts of excess iron.

The most common form is hereditary hemochromatosis. This means gene changes passed through families cause the gut to absorb more iron than the body needs. Iron usually accumulates slowly over many years.

Extra iron is stored mainly in the liver. It can also collect in the joints, heart, pancreas, skin, and organs that make hormones. Without treatment, this may lead to liver scarring, diabetes, heart problems, or other lasting damage.

Many people have no symptoms when the condition is found. Early diagnosis and treatment

can remove excess iron before serious organ damage occurs. Treatment can control iron levels, although it cannot remove the inherited tendency to absorb too much iron.

Symptoms

Hemochromatosis often causes no symptoms at first. When symptoms develop, they can be vague and may resemble many other conditions.

Possible symptoms include:

Iron buildup can also cause symptoms related to an affected organ. Liver damage may cause yellow skin or eyes, swelling in the abdomen, or easy bruising. Heart involvement may cause breathlessness, ankle swelling, palpitations, or an irregular heartbeat. Damage to the pancreas can contribute to diabetes, causing increased thirst and frequent urination.

Having one or more of these symptoms does not mean you have hemochromatosis. Blood tests and, in some cases, genetic or imaging tests are needed to find the cause.

- Ongoing tiredness or weakness
- Pain or stiffness in the joints, especially the knuckles
- Pain or discomfort in the upper abdomen
- Bronze, grey, or darker-looking skin
- Reduced interest in sex
- Erectile problems
- Irregular or absent menstrual periods
- Unexplained weight loss

Causes and risk factors

Hereditary hemochromatosis is usually linked to changes in the HFE gene. The HFE gene helps the body control how much iron it absorbs from food. Certain changes can weaken that control.

A person generally needs to inherit a relevant HFE gene change from both biological parents to be at risk of iron overload. Even then, not everyone develops harmful iron buildup. A genetic result shows susceptibility; it does not by itself show how much iron is stored or whether organs have been damaged.

Hereditary hemochromatosis is more often associated with significant iron overload in men and in people who no longer menstruate. Regular blood loss through menstruation and pregnancy can delay iron accumulation, though it does not remove the inherited tendency.

Less common inherited gene changes can also cause hemochromatosis, sometimes at a younger age.

Secondary iron overload is not inherited in the same way. It can result from repeated blood transfusions, certain long-term blood conditions, or some liver conditions. Each transfusion adds iron, and the body cannot actively remove the excess.

Alcohol does not cause inherited hemochromatosis, but heavy alcohol use can increase the risk of liver damage when iron levels are high.

Diagnosis

Diagnosis usually begins with blood tests.

Transferrin saturation estimates how much of the blood protein that carries iron is being used. A raised result can suggest that the body is absorbing too much iron.

Ferritin is a protein that stores iron. A high ferritin level may reflect increased iron stores, but it can also rise because of inflammation, infection, liver injury, alcohol use, or metabolic conditions. A high ferritin result alone does not confirm hemochromatosis.

Blood tests may be repeated because results can change. Liver tests, a full blood count, and tests for diabetes or other causes of abnormal iron results may also be needed.

If blood results suggest hereditary hemochromatosis, an HFE genetic test can look for common gene changes. The meaning of the result is considered alongside transferrin saturation, ferritin, symptoms, family history, and ancestry.

Magnetic resonance imaging, or MRI, can estimate how much iron is stored in the liver without surgery. A liver biopsy, which removes a small tissue sample with a needle, is now needed less often. It may be used when the diagnosis remains unclear or when detailed information about liver scarring is required.

Close biological relatives may be offered testing after a person is diagnosed with hereditary hemochromatosis. Testing generally starts with adult siblings, parents, or children, guided by the family's genetic results.

Treatment and management

The main treatment for hereditary hemochromatosis is therapeutic phlebotomy , also called venesection. This is similar to donating blood. A set amount of blood is removed so the body must use stored iron to make new red blood cells.

At first, blood may be removed regularly until iron stores reach the agreed target. Blood counts and iron tests are checked during this phase. Once excess iron has been reduced, less frequent maintenance treatment is usually needed to prevent it building up again.

The schedule is adjusted to your iron results, general health, symptoms, and ability to tolerate treatment. You should not change or stop planned treatment based only on how you feel, because iron can accumulate without causing obvious symptoms.

Phlebotomy can prevent many complications and may improve fatigue, skin darkening, and some liver or heart problems. Established joint damage, diabetes, hormone problems, or advanced liver scarring may not fully reverse.

Iron-chelating medicines bind to iron so it can leave the body. They may be considered when regular blood removal is unsuitable, particularly for people with transfusion-related iron overload or certain blood conditions. These medicines require monitoring and are not the usual first treatment for uncomplicated hereditary hemochromatosis.

Other care focuses on existing complications. This may include treatment for diabetes, heart rhythm problems, heart weakness, arthritis, hormone changes, or liver damage.

You should avoid iron supplements unless they have been specifically recommended. High-dose vitamin C supplements can increase iron absorption and may be discouraged in people with iron overload. A highly restrictive low-iron diet is usually not a substitute for treatment.

Complications

Complications are more likely when iron overload is severe or remains untreated for many years. They may include:

Removing excess iron lowers the risk of further damage. However, cirrhosis may not reverse completely. People who already have cirrhosis may need ongoing liver monitoring, including surveillance for liver cancer, even after iron levels have been controlled.

Joint symptoms can continue despite successful treatment because blood removal reduces stored iron but does not always repair established joint damage.

- Liver scarring, called fibrosis or cirrhosis
- Liver failure or liver cancer in people with advanced liver damage

- Diabetes caused partly by damage to the pancreas
- Arthritis, often affecting the knuckles, hips, or knees
- Abnormal heart rhythms or weakening of the heart muscle
- Reduced sex hormones, infertility, or sexual difficulties
- Changes in skin colour

Prevention

Inherited hemochromatosis cannot be prevented. A person is born with the gene changes that increase iron absorption.

Its complications can often be prevented or limited through early testing, monitoring, and treatment. If hereditary hemochromatosis is confirmed in your family, adult close relatives can discuss genetic and iron testing with a healthcare professional.

Avoiding unneeded iron supplements can reduce additional iron intake. Limiting alcohol is especially important if there is liver damage, because alcohol and iron overload can both injure the liver.

People with iron overload are advised to avoid raw or undercooked shellfish. Certain bacteria found in seawater and shellfish can cause unusually severe infections when iron levels are high. Normal food hygiene and thorough cooking reduce this risk.

Secondary iron overload cannot always be prevented because transfusions may be essential. When repeated transfusions are needed, regular iron monitoring can identify buildup and allow treatment.

Living with it and what to expect

Hemochromatosis is usually a lifelong condition, but iron overload can often be controlled. Treatment commonly has two stages: removing the iron that has already accumulated, then preventing it from building up again.

The first stage may involve frequent appointments over months. Maintenance treatment is usually less frequent, but blood tests and follow-up continue over the years. The exact pattern is based on how quickly your iron levels rise.

If treatment begins before organ damage, many people have a usual life expectancy and remain active. Existing cirrhosis, heart damage, diabetes, or other complications can affect

long-term health and require separate care.

Fatigue may improve as iron levels fall, but it does not always disappear. Joint pain is particularly likely to persist. New or continuing symptoms should not automatically be blamed on hemochromatosis, because another condition may need assessment.

Family testing can bring practical and emotional questions. A genetic change can be passed to children, but inheritance does not mean that every relative will develop iron overload. Genetic counselling may help explain results and family implications.

Regular follow-up is important even when you feel well. It tracks iron stores, checks whether maintenance treatment is adequate, and monitors any organs already affected.

When to get medical help

Arrange a routine medical appointment if you have persistent fatigue, joint pain, abdominal discomfort, skin darkening, or abnormal liver or iron blood tests. These symptoms have many possible causes and need appropriate assessment rather than self-diagnosis.

You should also seek routine advice if a parent, sibling, or child has confirmed hereditary hemochromatosis. Testing may be appropriate even if you have no symptoms.

Contact your healthcare team promptly if you have known hemochromatosis and develop worsening ankle or abdominal swelling, increasing breathlessness, yellow skin or eyes, new palpitations, or symptoms of diabetes.

Seek emergency help for:

These signs may reflect a serious heart problem, bleeding, advanced liver trouble, or severe infection. They require urgent assessment rather than waiting for a routine follow-up visit.

- Chest pain, severe breathlessness, fainting, or a sustained rapid heartbeat
- Vomiting blood or passing black, tar-like stools
- Sudden confusion, extreme drowsiness, or difficulty waking
- Severe weakness with yellow skin or rapidly increasing abdominal swelling
- Fever or severe illness after eating raw or undercooked shellfish

Common questions

Is hemochromatosis cancer?

No. Hemochromatosis is an iron-overload condition, not cancer. However, people who develop

cirrhosis from iron overload have a higher risk of liver cancer and may need regular surveillance.

Can I have hemochromatosis with a normal ferritin level?

Yes. Genetic susceptibility can be present before iron stores rise. Ferritin can also change over time. Diagnosis and monitoring use ferritin alongside transferrin saturation, genetic results, and other clinical information.

Do I need to stop eating red meat?

Most people do not need to remove all iron-containing foods. Treatment removes much more iron than diet changes alone. Avoiding iron supplements and maintaining a balanced diet are usually more practical than following a severely restricted diet.

Can I drink alcohol if I have hemochromatosis?

Alcohol can add to liver injury. The safest amount depends on whether you have liver damage and other health factors. People with cirrhosis are generally advised to avoid alcohol. Discuss your situation during routine care.

Can I donate the blood removed during treatment?

In some places, blood collected through therapeutic phlebotomy can be used for donation if you meet blood-service requirements. Policies differ, so the treatment centre or blood service can explain local arrangements.

Will my children inherit hemochromatosis?

They may inherit an HFE gene change, but this does not mean they will develop iron overload. Their risk also depends on the genes inherited from their other biological parent. Testing is generally considered when relatives are adults unless a rare childhood-onset form is suspected.

Sources

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