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Hemochromatosis

Hemochromatosis is a condition in which there is too much iron in the body. It is also called iron overload.

Causes

Hemochromatosis may be a genetic disorder passed down through families.

- People with this type absorb too much iron through their digestive tract. Iron builds up in the body. The liver, heart, and pancreas are common organs where iron builds up.
- It is present at birth, but may not be diagnosed for years.

Hemochromatosis may also occur as a result of:

- Too many blood transfusions in a person with blood disorders such as thalassemia or certain anemias. Over time this may lead to iron overload.
- Long-term alcohol use and other health conditions.

This disorder affects more men than women. It is most common in white people of northern European descent.

Symptoms

Symptoms may include any of the following:

- Abdominal pain
- Fatigue, lack of energy, weakness
- Generalized darkening of skin color (often referred to as bronzing)
- Joint pain
- Loss of body hair
- Loss of sexual desire
- Weight loss

Exams and Tests

Your health care provider will perform a physical exam. This may show liver and spleen swelling, and skin color changes.

Blood tests may help make the diagnosis. Tests may include:

- Ferritin level
- Iron level
- Percentage of transferrin saturation (high)
- Genetic testing

Other tests may include:

- Blood sugar (glucose) level
- Alpha fetoprotein
- Echocardiogram to examine the heart's function
- Electrocardiogram (ECG) to look at the electrical activity of the heart
- Imaging tests such as CT scans, MRI, and ultrasound
- Liver function tests

The condition may be confirmed with a liver biopsy or genetic testing. If a genetic defect is confirmed, other blood tests can be used to find out if other family members are at risk for iron overload.

Treatment

The goal of treatment is to remove excess iron from the body and treat any organ damage.

A procedure called phlebotomy is the best method for removing excess iron from the body:

- One half liter of blood is removed from the body each week until the body's iron stores are depleted. This may take many months to do.
- After that, the procedure may be done less often to maintain normal iron stores.

Why the procedure is needed depends on your symptoms and levels of hemoglobin and serum ferritin and how much iron you take in your diet.

For people unable to undergo phlebotomy, medicines may be prescribed to reduce the body's iron.

Other health problems such as diabetes, decreased testosterone levels in men, arthritis, liver failure, and heart failure will be treated.

If you are diagnosed with hemochromatosis, your provider may recommend a diet to reduce how much iron is absorbed through your digestive tract. Your provider may recommend the following:

- Do not drink alcohol, especially if you have liver damage.
- Do not take iron pills or vitamins containing iron.
- Do not use iron cookware.
- Limit foods fortified with iron, such as 100% iron-fortified breakfast cereals.
- Avoid uncooked shellfish.

Outlook (Prognosis)

Untreated, iron overload can lead to liver damage.

Extra iron may also build up in other areas of the body, including the thyroid gland, testicles, pancreas, pituitary gland, heart, or joints. Early treatment can help prevent complications such as liver disease, heart disease, arthritis or diabetes.

How well you do depends on the amount of organ damage. Some organ damage can be reversed when hemochromatosis is detected early and treated aggressively with phlebotomy.

Possible Complications

Complications include:

- Cirrhosis
- Liver failure
- Liver cancer

The disease may lead to the development of:

- Arthritis
- Diabetes
- Heart problems
- Increased risk for certain bacterial infections
- Testicular atrophy
- Skin color changes

When to Contact a Medical Professional

Contact your provider if symptoms of hemochromatosis develop.

Contact your provider for an appointment (for screening) if a family member has been diagnosed with hemochromatosis.

Prevention

Screening family members of a person diagnosed with hemochromatosis may detect the disease early so that treatment can be started before organ damage has occurred in other affected relatives.

Alternative Names

Iron overload; Blood transfusion - hemochromatosis

References

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