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Education

Hemochromatosis

What is hemochromatosis?

Hemochromatosis is a disease that causes iron deposits to build up throughout the body. The buildup of iron can severely damage or destroy organs. If the disease is detected early, it can be treated and the damage prevented.

Hemochromatosis is also called iron overload disease.

How does it occur?

Hemochromatosis is caused by an inherited tendency to store too much iron.

The liver, heart, pancreas, and hormone levels are most affected by the slow buildup of iron in the body.

- Liver cells become scarred. The liver helps you digest food and use medicines. It also helps rid the body of toxins and waste products. Damage to the liver makes it hard for your liver to function normally. Scarring of the liver is called cirrhosis.
- Your heart muscle can become damaged, leading to heart problems, including heart failure.
- The pancreas, which produces insulin, becomes damaged. The pancreas may make less insulin than your body needs. When you have too little insulin, the level of sugar in your blood rises and you may become diabetic.
- Abnormal levels of thyroid hormone and sex hormone can cause fatigue, infertility, and erectile dysfunction (trouble having and keeping an erection).

About 3 of every 1,000 people in the US have hemochromatosis, especially those of English, French, Swedish, or Portuguese descent. Men are much more likely to have symptoms than women. Before menopause women are protected somewhat from the disease because they lose quite a bit of iron during menstruation and childbirth.

Some people do not have the disease but are carriers. Being a carrier means that you can pass the gene for the disease on to your children.

What are the symptoms?

You may not have any symptoms for years. Symptoms usually appear in middle age. They include:

- fatigue (the most common symptom)
- joint pains (especially in the fingers, hips, and knees)
- a change in your skin color to gray or brown
- episodes of rapid heart rate
- erectile dysfunction
- irregular or no menstrual periods, and trouble getting pregnant
- symptoms of diabetes, such as frequent urination and excessive thirst
- symptoms of cirrhosis of the liver, including nausea, loss of appetite, swelling of the abdomen, abdominal pain, and vomiting of blood.

How is it diagnosed?

Hemochromatosis can be diagnosed from blood tests. These tests can detect the problem before symptoms appear.

For people who have no symptoms and no known family history of hemochromatosis, the disease is usually discovered through blood tests done for some other reason. When these blood tests show signs of excess iron or liver damage, specific blood tests for iron overload disease can be done.

If blood tests for hemochromatosis are positive, you may have a liver biopsy. When you have a liver biopsy your health care provider numbs the skin over the area of the liver (by the lower right rib cage). He or she then inserts a hollow needle and removes some liver tissue. The tissue is examined for abnormal stores of iron and signs of liver damage.

How is it treated?

The treatment is very simple: excess iron is removed from your body by removing blood. When your level of iron is high, you may need to have a pint of blood removed each week until your iron level is normal. Your iron levels can be checked with blood tests. These tests will determine if, when, and how much blood needs to be taken. When blood removal has lowered your iron levels to normal, you will probably need to repeat the treatment every 3 to 4 months to maintain normal levels.

If your liver or other organs are damaged, problems resulting from the damage will also be treated. You will likely need to continue taking medicine for problems such as:

- diabetes
- thyroid problems
- erectile dysfunction
- heart disease.

Joint pain can be treated with anti-inflammatory medicines.

Once liver scarring has begun, it may progress to serious liver disease and liver failure. A liver transplant may be an option in this case.

An important part of treatment is to avoid drinking alcohol and taking medicines that can worsen liver damage.

How long do the effects last?

Once you start having symptoms, they usually continue even though you are having treatments to remove excess iron. This means you will need to continue treatment for heart, thyroid, liver, erectile dysfunction, and joint problems.

If you do not have any symptoms of hemochromatosis, you will have regular checks of your iron levels so blood can be removed when your levels get too high. This will prevent symptoms and organ damage.

How can I help prevent hemochromatosis?

If you have a family history of hemochromatosis, you should have genetic testing or blood tests to see if you have the disease or may be a carrier. Early and continued treatment, including regular blood tests, can prevent your iron levels from becoming too high. This will prevent organ damage and allow you to have a normal life.

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