Hemochromatosis is a disorder in which the body absorbs too much iron from food. The most common cause is a genetic disorder called hereditary hemochromatosis, or HHC. Normally, any iron that the body doesn’t need is excreted. But patients with HHC store the excess iron throughout their body, including in the pancreas, liver, and skin. The iron deposits damage these organs and tissues and can be fatal. The hereditary form is the most common genetic disorder in the United States.

Some patients develop hemochromatosis after years of taking large doses of iron pills or receiving many blood transfusions. This form, which is rare, is called acquired hemochromatosis.

What are the symptoms?
Joint pain is the most common symptom of HHC. Patients may also experience fatigue, abdominal pain, jaundice (yellowing of skin and eyes), and a change in skin color to bronze or gray. Over time, hemochromatosis can cause arthritis, liver disease, liver failure, liver cancer, heart disease, and diabetes. Because iron overload takes years to develop, symptoms may not occur until a person is well into their 30s, 40s, or 50s.

How is it diagnosed and treated?
Hemochromatosis is diagnosed through blood tests that measure iron levels. Sometimes a liver biopsy is done to check for the amount of iron in the liver and for liver damage.

Hereditary hemochromatosis is treated by drawing blood to remove excess iron from the body (phlebotomy) and prevent it from building up in organs. For about the first 18 months, blood is drawn once every 2 to 6 months for the rest of the person’s life. Phlebotomy cannot cure HHC but it can help control symptoms.

Patients with HHC should also limit their intake of iron-rich foods such as red meats and iron-fortified cereals. Iron supplements should be avoided completely. Patients should limit their intake of alcohol, and those with liver damage should avoid drinking alcohol altogether. Because vitamin C increases the amount of iron absorbed in the intestines, patients should limit vitamin C supplements to 500 mg/day.

Because early detection and treatment will prevent the serious long-term complications such as heart and liver disease, first-degree relatives (mother, father, brothers, sisters) of patients with HHC should undergo genetic screening to determine whether they have HHC as well.

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