

WHAT YOU SHOULD KNOW ABOUT HEMOCHROMATOSIS

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What Is Hemochromatosis?

Hemochromatosis is an inheritable condition where too much iron builds up in your body due to a genetic defect. Although most patients have no symptoms, in some patients, because the body cannot get rid of iron, the excess iron is deposited in different organs, leading to organ damage, lower quality of life, and shorter life expectancy.



What Are the Risk Factors?

Hemochromatosis is caused by mutations in different genes that more commonly occur in White people of European descent, with some of the mutations more likely to lead to symptoms and poorer health effects than others. Risk factors that lead to worse outcomes include:

- Male sex
- Older age at presentation
- Alcohol consumption
- Type 2 diabetes mellitus
- Homozygous p.C282Y genetic variant

What Are the Symptoms?

Most people with hemochromatosis do not have any symptoms. However, those with more severe forms of the condition may have symptoms due to iron deposits in organs such as the liver, heart, pancreas, endocrine glands, and joints. This can lead to symptoms, including arthritis and joint pain; symptoms from diabetes or high blood glucose, such as increased urination, increased thirst, numbness and tingling, and vision changes; loss of libido and symptoms of low thyroid levels, such as weight gain and weakness; palpitations; shortness of breath; leg swelling; tiredness; light-headedness; and, in advanced cases of liver damage, easy bruising, bleeding, jaundice, and swelling of the legs. Deposits in the skin can cause browning.

How Is It Diagnosed?

Your doctor may obtain blood tests of your iron levels based on your family history, symptoms, or other abnormal blood test results. Your doctor may also order a genetic test or other imaging tests, such as an MRI.

How Is It Treated?

For patients with iron levels above a certain threshold, phlebotomy (drawing about a pint of blood at a time) on a regular basis is the most common way to decrease body stores of iron to maintain levels within a target range. Patients with this condition should avoid drinking alcohol and should limit their intake of red meat, iron-enriched grains, and vitamin C supplements (which can increase iron absorption).

Questions for My Doctor

- How serious is my hemochromatosis?
- Should I change my diet?
- Should I undergo treatment?
- Am I at risk for cirrhosis or liver failure?
- Should I see a specialist?

For More Information



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National Institute of Diabetes and Digestive and Kidney Diseases

www.niddk.nih.gov/health-information/liver-disease/hemochromatosis