



Hemochromatosis

Hemochromatosis is a metabolic disorder that causes your body to absorb too much iron from the food you eat. This excess iron is then stored in your organs, especially your liver, heart, and pancreas. Over time, iron accumulation can lead to life-threatening conditions such as liver disease, heart problems, and diabetes.

Primary vs Secondary Hemochromatosis

There are two main types of hemochromatosis: primary (or hereditary) hemochromatosis and secondary hemochromatosis. Primary hemochromatosis is an inherited condition caused by a genetic mutation. This form of the disease is most common in Caucasians of Northern European descent and affects one in 300 people in the United States. Secondary hemochromatosis, on the other hand, is usually the result of another disease or condition, such as anemia or chronic liver disease.

Lack of Early Warning Symptoms

One of the challenges with hemochromatosis is that it often goes undiagnosed, partially due to its nonspecific symptoms. In the early stages of the disease, symptoms may be vague or even nonexistent. As the disease progresses, symptoms may include fatigue, joint pain, abdominal pain, loss of libido, and skin darkening.

Diagnostic Tests

Diagnosing hemochromatosis involves a combination of medical history, physical examination, and tests. Blood tests are the primary method for diagnosing hemochromatosis. These tests measure the amount of iron in your blood and your liver's function. The two main blood tests used to screen for hemochromatosis are serum transferrin saturation and serum ferritin. The serum transferrin saturation test measures the amount of iron bound to a protein (transferrin) that carries iron in the blood. Transferrin saturation values greater than 45% are considered too high. The serum ferritin test measures the amount of stored iron in your body. Normal levels are generally between 20 and 500 nanograms per milliliter (ng/mL) for men and between 20 and 200 ng/mL for women. Levels significantly above these ranges may indicate hemochromatosis. If these blood tests show high levels of iron in your body, your doctor may recommend a liver biopsy to confirm the diagnosis and check for liver damage. A small sample of liver tissue is taken and examined under a microscope during a liver biopsy.

Treatment

The treatment for hemochromatosis aims to reduce the amount of iron in your body to normal levels and manage any organ damage that has occurred. The primary treatment is a simple and effective process called phlebotomy. During phlebotomy, a unit of blood is removed from your body on a regular schedule. This procedure helps lower the amount of iron in your body. In addition to phlebotomy, you may need to make specific lifestyle changes. These can include avoiding iron supplements and multivitamins containing iron, limiting vitamin C supplements (which can increase iron absorption), limiting alcohol consumption (to reduce the risk of liver damage), and avoiding raw shellfish (people with hemochromatosis are at risk of serious infection from certain bacteria in raw shellfish).

In conclusion, hemochromatosis is a serious but manageable condition. Understanding this disease's causes, diagnosis, and treatments is the first step toward managing it effectively. If you or a loved one has been diagnosed with hemochromatosis, remember that you are not alone and that resources and treatments are available to help you on your journey.

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