

Hemochromatosis - What is It?

A Genetic, Metabolic Disorder

Hereditary hemochromatosis is a **genetic, metabolic disorder that results in iron overload**; the body absorbs and retains too much dietary iron. It is a primary disorder of iron metabolism that can affect many organ systems including the liver, pancreas, heart, endocrine glands and joints. It is **potentially fatal, but easily treated if diagnosed early**, before the excess iron causes irreversible damage.

'Normal' Iron Levels

Iron is an essential nutrient for the human body. Too little can compromise many important functions and lead to various diseases. Similarly, too much can cause severe damage to organs and tissues, leading to disease and early death.

A normal diet provides between 10-20 mg of iron daily, of which the body absorbs only 1.0 to 1.5 mg through the intestinal tract. The rest of the iron not absorbed during digestion is excreted in the stool. Iron metabolism is a complex process. The body responds to increased or decreased demand by adjusting the amount it absorbs. Once iron is absorbed into the body, it is difficult to eliminate, and can only be lost in small amounts through blood loss, sweat, urine and the sloughing of skin and gut cells. Therefore, our body maintains a strict regulation of iron absorption.

Normally, the body has about 4,000 mg of iron, of which about 3,000 mg is contained in hemoglobin in the red blood cells. About 500 mg is bound to the storage protein ferritin, and 300 mg is stored in the liver. Transferrin, the protein that carries the iron from organ to organ around the body, helps regulate how and when iron is stored and transferred to bone marrow and other cells when needed for body processes.

Broken Iron Feedback System

In hereditary hemochromatosis (HHC), the feedback signal within this complex system is not working properly. The gut continues to absorb iron at 2-4 times the normal rate, despite the body already being overloaded with iron. In response, the level of ferritin, the protein that stores unused iron in body cells, increases in an attempt to contain excess iron. As the transferrin protein gets saturated with iron, other proteins not usually involved in iron metabolism bind the excess iron. Unfortunately, these other proteins readily enter cells to deposit the iron in organs and tissues where it causes damage from free radical activity leading to specific organ dysfunction, disease and death. The liver and heart are particularly vulnerable, but the pancreas, endocrine glands, testes and ovaries, joints and skin can also be affected.

Excess iron accumulation in HHC is chronic and ongoing, and while a normal body contains about 4 grams (4,000 mg) of iron, a hemochromatosis patient typically has at least 15-60 grams of iron upon diagnosis.

Development of Symptoms

It takes time for iron overload to reach a level that will cause organ damage and failure. **Men typically develop disease between 40 and 60 years of age, and women after menopause.** Diet, vitamin pills with iron, and alcohol consumption all can have an effect.