Gastroenterology Associates of S.W. Florida, P.A.

*Board Certified in Gastroenterology*

# HEMOCHROMATOSIS

***What is hemochromatosis?***

Hemochromatosis is an inherited disorder of excessive body accumulation of iron. It is common among the white population, affecting approximately 1 in 400 individuals of European ancestry. Hemochromatosis patients are believed to absorb excessive amounts of iron from the diet. Since the human body has limited ways of eliminating the absorbed iron, the iron accumulates over time in the liver, bone marrow, pancreas, skin, and testicles. This accumulation of iron in these organs causes them to function poorly.

***What are the symptoms of hemochromatosis?***

Patients with early hemochromatosis have no symptoms, and are unaware of their condition. The disease may then be discovered when elevated iron blood levels are noted by routine blood testing. In males, symptoms may not appear until 40-50 years of age. Iron deposits in the skin cause darkening of the skin. Since females lose iron through menstrual blood loss, they develop organ damage from iron accumulation 15- 20 years later than men on average.

Iron deposits in the pituitary gland and testicles cause shrinkage of the testicles and impotence. Iron deposits in the pancreas cause a decrease in insulin production resulting in diabetes mellitus (please read the Diabetes Mellitus article. Iron deposits in the heart muscle can cause heart failure as well as abnormal heart rhythms. Iron accumulation in the liver causes scarring of the liver (cirrhosis) and an increased risk of developing liver cancer. For further information on the consequences of cirrhosis, please read the Cirrhosis article.

***How is hemochromatosis diagnosed?***

Initial screening for hemochromatosis involves blood tests for blood iron and ferritin levels. Ferritin is a blood protein that serves as an indicator of the amount of iron stored in the body. Blood iron and ferritin levels are usually low in patients with iron deficiency anemia, and are high in patients with hemochromatosis and other conditions that cause an increase in body iron levels. Since ferritin can also be elevated in certain infections, such as viral hepatitis and other inflammations in the body, ferritin increase alone is not sufficient to accurately diagnose hemochromatosis.

The most accurate test for hemochromatosis is measuring the iron content of liver tissue obtained by a biopsy. A biopsy involves the removal of a sample of liver tissue for analysis and is usually performed with a needle under local anesthesia. After numbing the skin and the underlying tissues, the doctor inserts a needle into the liver through the right lower rib cage, sometimes under ultrasound guidance. The tissue obtained by the needle is studied under a microscope for liver damage or cirrhosis. The amount of iron in the liver is usually significantly elevated in hemochromatosis.

The gene for hereditary hemochromatosis was identified in 1996. The gene is referred to as HFE. The HFE gene can be identified in blood testing of 90 percent of patients with northern European ancestry.

***How is hemochromatosis treated?***

The most effective treatment of hemochromatosis is to reduce the blood iron by removing blood using phlebotomy (withdrawal of blood from the arm veins). One unit of blood (250 mg of iron) is usually withdrawn every one to two weeks until the patient becomes mildly anemic. The frequency of phlebotomies is then decreased to every 2-3 months to maintain mild anemia. When hemochromatosis is diagnosed early and treated effectively, damage to the liver, heart, testicles, pancreas and joints can be completely prevented, and patients can return to normal health. In patients with established cirrhosis, effective treatment can improve heart function, skin color, and diabetes. However, the cirrhosis is irreversible, and the risk of developing liver cancer remains.

***Screening for hemochromatosis***

Since hemochromatosis is common, and early diagnosis is important for successful treatment, screening for this disorder is important. Ferritin and iron levels are obtained in patients with unexplained liver disease, darkening skin, heart disease or heart rhythm abnormalities, impotence, diabetes mellitus, and relatives of patients with hemochromatosis. Some doctors even recommend blood iron and ferritin levels to be included in routine screening blood tests. Greater patient and physician awareness of this common inherited disorder can lead to earlier and more successful treatment and avoidance of preventable illness.

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