## Iron Overload and Hemochromatosis

In the United States, most cases of "iron overload" are caused by a genetic condition known as "hereditary hemochromatosis." Serious and sometimes fatal health problems may result from the excess iron that accumulates in the body. These complications may include severe weakness or fatigue, unexplained joint or abdominal pain, impotence, infertility, and loss of menstrual periods. The diseases associated with hemochromatosis include liver cancer, cirrhosis, arthritis, diabetes, and heart failure.

#### **Overview on Iron Overload and Hemochromatosis**

This section provides basic information on hemochromatosis, the signs and symptoms, diagnosis, treatment, questions regarding blood donation, prognosis and public health issues related to hemochromatosis.

#### Iron Overload Disease due to Hereditary Hemochromatosis

Additional background information, a diagram depicting the course of hereditary hemocromatosis, CDC program activities, and priorities for 2001 can be found here.

Screening for Iron Overload due to Hereditary Hemochromatosis

Information on identifying people early with evidence of iron overload.

#### **Information for Patients and Their Families**

You can live a healthy life, *if* you get treatment early.

#### **Frequently Asked Questions**

Definitions, signs and symptoms, risk factors, detection, prevalence, treatment, blood donation, diet, causes, history, and important information regarding the CDC and Hemochromatosis.

# Iron overload and Hemochromatosis Overview on Iron Overload and Hemochromatosis

Hemochromatosis is a condition in which the body accumulates excess amounts of iron. Hereditary hemochromatosis is one of the most common genetic diseases in humans. In the United States, as many as one million people have evidence of hemochromatosis, and up to one in every ten people may carry the gene for the disorder. Serious and sometimes fatal health problems may result from the excess iron that accumulates in the body as a result of this genetic disorder. Some diseases associated with hemochromatosis are arthritis, cirrhosis of the liver, diabetes, heart failure, and liver cancer.

### Signs and Symptoms

Symptoms associated with hemochromatosis most often appear in middle age, although some people may develop problems earlier. These symptoms reflect the tissue and organ damage that result from the disease.

- Early disease symptoms of hemochromatosis are nonspecific and include fatigue, heart palpitations, joint pain, non-specific stomach pain, impotence, and loss of menstrual periods. Abnormalities of liver function tests can also occur in the absence of other symptoms.
- Later disease symptoms include gray or bronze skin pigmentation, cirrhosis of the liver, liver
  cancer, diabetes mellitus, hypopituitarism, decreased pituitary or gonadal function, heart disease or
  heart failure, joint disease, chronic abdominal pain, severe fatigue, and certain infections.

Excess iron in the diet, alcohol use, infections, iron lost through blood donations and menstruation, and other environmental factors may affect the clinical course of hemochromatosis. For example, alcohol use may worsen the disease, whereas iron lost through blood donations or menstruation may lessen the severity.

## **Diagnosis**

Early detection of hemochromatosis is essential because the disease's potentially serious complications can be prevented by early therapy. A sensitive and relatively inexpensive screening test for iron overload is the transferrin saturation test (serum iron divided by the total iron binding capacity). Quantitative phlebotomy is now considered the diagnostic test of choice by most health care providers.

Currently, routine medical care or checkups usually do not include testing for hemochromatosis and many cases may remain undetected.

People who have a close blood relative with hemochromatosis, as well as persons who have the signs and symptoms compatible with hemochromatosis (as described above), should talk with their health care provider about the possibility of being evaluated for hemochromatosis.