

## **Hemochromatosis**

### **What is hemochromatosis?**

Hemochromatosis is a common disorder of iron metabolism resulting in iron overload and affecting about 1 in 250 individuals of Northern European descent. It is an inherited disorder, but to actually develop problems from hemochromatosis you must inherit two abnormal genes, receiving one from each parent. If you have inherited both abnormal genes, you will absorb increased amounts of iron from your diet and will gradually accumulate excess iron, primarily in the liver. Over many years, these increased iron deposits in the liver can result in liver disease such as cirrhosis and cancer of the liver.

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

Symptoms from hemochromatosis are often vague and nonspecific and may include weakness, lack of energy, upper abdominal pain, and weight loss. Parts of the body other than the liver can also be affected by hemochromatosis and cause more specific symptoms. For example, patients may develop arthritis and have joint pain. Patients may have involvement of the heart and develop abnormal heart rhythms or symptoms of heart failure. Patients can develop abnormalities in the pancreas including diabetes. Early in the disease, patients may not have any symptoms at all.

### **How is hemochromatosis diagnosed?**

Currently, the most common way that patients with hemochromatosis come to medical attention is by having abnormal levels of iron in the blood identified during routine blood tests. Thus, if you have any of the symptoms mentioned above, or if screening blood tests are abnormal, you should be evaluated for hemochromatosis. Typically, if hemochromatosis is suspected, the patient will be asked to have a liver biopsy. A liver biopsy is a procedure, performed using local anesthesia, where a needle is inserted to remove a small specimen of liver tissue so that it can be examined microscopically. When the liver biopsy is performed, the liver tissue that is removed is tested for iron.

### **How is hemochromatosis treated?**

Once the diagnosis of hemochromatosis is confirmed, treatment is simple and involves a procedure called "therapeutic phlebotomy" or "blood-letting." This is done by removing blood each week until the excess iron stores are reduced to a normal level. This procedure is the same one used for blood donation and can take as long as 6 to 12 months of weekly phlebotomy to fully deplete the excess iron stores. Once the excess iron stores are depleted, then patients should have "maintenance phlebotomy" every 2 to 4 months for the rest of their lives.

### **Can hemochromatosis be confused with other liver diseases?**

Patients with various types of liver disease or certain other conditions can have abnormal blood iron studies. The only way to definitively diagnose hemochromatosis is by way of liver biopsy.

### **Should family members be screened?**

Since hemochromatosis is an inherited disorder, once the disease has been treated, it is recommended that all first-degree relatives (e.g., brothers, sisters, parents, children) be screened for hemochromatosis with routine blood iron tests.

### **Summary**

Hemochromatosis is a common disorder. It can be easily identified before there are any complications and can be treated in a safe and inexpensive manner.

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