The Oregon Clinic - Gastroenterology
Hemochromatosis

What is hemochromatosis?
Hemochromatosis is a disease of iron overload. It is caused by a malfunction in the intestine that causes the body to absorb too much iron. One of the places this excess iron is stored is in the liver. When too much iron is stored in the liver it can cause liver damage. In hemochromatosis excess iron is stored not only in the liver but in other organs such as the pancreas, heart, brain, and bones.

Hemochromatosis is a genetic disease. It is caused by genetic mutations that are passed on from parents to their children. It is the most common genetic disease in the United States and affects one in every 200-300 people.

Another type of hemochromatosis is caused by taking too much iron supplements or receiving too many blood transfusions. This is called secondary hemochromatosis or hemosiderosis. This is not a genetic disease but can lead to some of the same problems.

Cause
The exact mechanism that causes the body to absorb too much iron is not fully understood. Having a single hemochromatosis gene mutation occurs in about one of every 10 people. However, to get the disease, a person must have two copies of a hemochromatosis gene; they have to get one mutated gene from each parent. Thus, people who have a first degree relative (parent, brother, or sister) that has hemochromatosis should be screened for the disease by their doctor.

Symptoms
The disease can occur at any age but typically is found in middle age. Women often are diagnosed later because they lose iron from menstrual bleeding and thus do not develop iron overload until later in life. Often people do not have any symptoms and the disease is found on routine blood testing.
Symptoms of hemochromatosis are due to iron overload and damage to certain organs. In the liver, this damage can cause cirrhosis and sometimes people become jaundiced (turn yellow). Iron in the pancreas can cause diabetes or trouble digesting food. Iron in the heart can cause heart failure. Iron in the bones and joints can cause arthritis.

**Diagnosis**
The earlier this disease is diagnosed the better. Untreated hemochromatosis can lead to severe organ damage. One way to find this disease is to measure the level of ferritin in the blood. Ferritin carries iron around the body and is present in very high levels in patients with hemochromatosis. To confirm the diagnosis the doctor will often obtain a liver biopsy to measure how much iron is in the liver tissue. There is also a blood test that can detect the most common gene mutations that cause hemochromatosis.

**Treatment**
The only way to treat hemochromatosis is to help the body get rid of the excess iron. A large portion of the body’s iron is in the red blood cells. Removing one pint of blood every week helps the body get rid of this excess iron although it can take several months to get the job done. Once the iron levels are back to normal, removal of one pint of blood every 1-2 months keeps the iron level where it should be.

Treatment for hemochromatosis can prevent organ damage. However, existing organ damage cannot be totally reversed and, in those cases, treatment is mainly aimed at preventing further harm to the organs.

**Summary**
Hemochromatosis the most common inherited disease in the United States and causes iron overload in the body. A family history of this disease in a first degree relative is a reason to be screened for hemochromatosis. Early diagnosis allows treatment to begin that can prevent organ damage.