Greenwich Hospital

What is Hemochromatosis?

PATIENT/FAMILY INFORMATION SHEET

What is Hemochromatosis?

Hemochromatosis is a condition that occurs when your body absorbs too much iron. Your body has no natural way to rid itself of the excess iron. It stores the iron in your tissues, joints, liver, pancreas, kidneys, and heart.

What causes the different types of hemochromatosis?

Hereditary (genetic) Hemochromatosis is caused by inheriting two copies of the HFE gene mutation that allows excess iron to be absorbed from your food.

Acquired (secondary) Hemochromatosis can develop from chronic liver diseases and blood conditions. The excessive intake of iron through vitamins, medicine or a diet high in iron can cause temporary hemochromatosis.

Are you at risk of inheriting hemochromatosis?

1. Your risk of inheriting hemochromatosis increases with the number of family members with hereditary hemochromatosis and how closely they are related to you.

Family Members with Hemochromatosis	Chances of Inheriting 2 Copies of HFE Gene Mutation
• None (General Population)	1 in 200 or 0.5%
• Aunt, uncle, cousin	Less than 1 in 60 or less than 2%
• One parent but no sibling	1 in 20 or 5%
• Sibling but no parent	1 in 4 or 25%
• Sibling and parent	1 in 2 or 50%
• Both parents	1 in 1 or 100%

Source: Retrieved May 18,2005 www.dnadiret.com

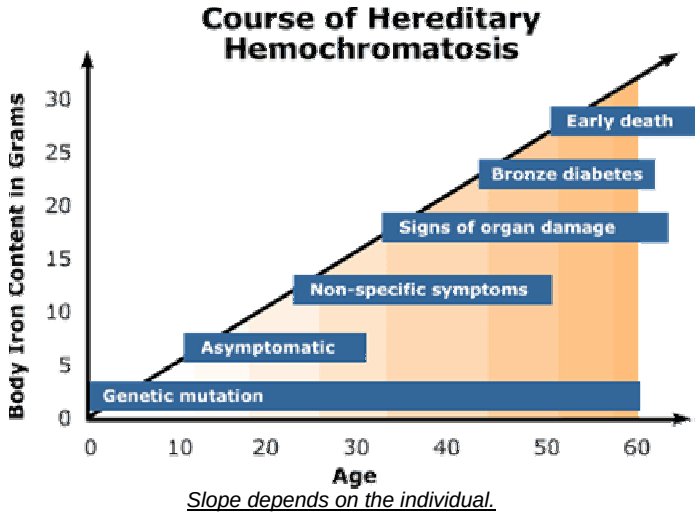
2. If you are of Northern European descent (British, Dutch, German, Irish or French) you are more likely to be a carrier of the HFE gene mutation that causes hereditary hemochromatosis.

Who will develop symptoms of hereditary hemochromatosis?

- A man is 5 times more likely to develop symptoms of hereditary hemochromatosis.
- Menopause or a hysterectomy may increase your risk of excess iron storage and developing symptoms of hereditary hemochromatosis.

When do symptoms of hereditary hemochromatosis occur?

Symptoms of hereditary hemochromatosis occur later in life. Excess iron builds up slowly over years as indicated on the chart below:



Slope depends on the individual.

Source: Retrieved May 18, 2005, from www.cdc.gov

What are the symptoms of hereditary hemochromatosis?

Early symptoms of hereditary hemochromatosis are like those of many other conditions:

- Joint Pain
- Fatigue
- Lack of energy
- Weight loss
- Abdominal pain
- Loss of sex drive

If hereditary hemochromatosis is untreated, later symptoms develop as iron is stored in your body.

Complication	Description
Liver damage	• Liver inflammation • Liver failure
Heart problems	• Irregular heart beat • Heart disease • Heart failure
Decreased function of testicles in men and ovaries in women	• Loss of body hair in men • Breast enlargement in men • Shrinkage of testicles • Early menopause in women • Decreased sex drive in men & women
Damage to the pancreas	• Diabetes
Other problems	• Arthritis • Darkened skin color (bronze to metallic gray)

How is hereditary hemochromatosis diagnosed?

1.Your doctor will take a thorough medical history. He may ask you about:

- Your health conditions and the symptoms you are having.
- Family health history.
- Medications, vitamins or supplements that you are taking.
- Your diet.
- History of blood loss.
- Sexual dysfunction.

2. A physical examination.
3. Routine blood tests.
4. If your doctor suspects hereditary hemochromatosis, additional blood tests can be ordered to determine whether you have excessive iron stored in your body.
   - Serum transferrin saturation
   - Total iron-binding capacity (TIBC)
   - Serum iron
   - Serum ferritin
5. A genetic test for HFE gene mutation can help confirm the diagnosis of hereditary hemochromatosis.
6. A liver biopsy may be needed to measure how much iron has been stored in your liver and if there is any damage.
7. Your doctor may order other additional test.

How is hereditary hemochromatosis treated?

Hemochromatosis is treated by removing the excessive iron stored in your body through a therapeutic phlebotomy. This is the intentional removal of a specified amount of blood on a regular basis. Your blood is removed the same way it is drawn from donors at blood banks. Depending on how elevated your iron tests are your doctor will order a pint of blood to be taken. You will need to do this once or twice a week for several months to a year and sometimes longer. When your iron has returned to a normal level, you will need to see your doctor regularly for physical examinations and blood tests. He will then determine how often you will need your therapeutic phlebotomies to maintain the normal iron level.

The earlier you know your risks of inheriting hemochromatosis, the earlier you can have your iron levels monitored and treated, and the better your chances are of living a longer, healthier life.

Sources:
- **CDC @ www.cdc.gov Retrieved May 2005.**
- **NDDIC @ http://digestive.niddk.nihgov Retrieved May 2005.**
- **DNA Direct @ www.dnadirect.com Retrieved May 2005.**
- **Yale New Haven Hospital @ www.ynhhs.org Retrieved May 2005.**