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HEMOCHROMATOSIS

P. Brindha
Lecturer, Sree Balaji College of Nursing, Bharath University, Tamilnadu

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Abstract:

Hereditary hemochromatosis is a disorder that causes the body to absorb too much iron from the diet. The three types of hemochromatosis are primary hemochromatosis, also called hereditary hemochromatosis; secondary hemochromatosis; and neonatal hemochromatosis. The symptoms includejoint pain fatigue or feeling tired unexplained weight loss abnormal bronze or gray skin color, abdominal pain. Iron may build up in the organs and cause complications, including cirrhosis, or scarring of liver tissue, diabetes, irregular heart rhythms or weakening of the heart muscle, arthritis, erectile dysfunction. The diagnosis is usually made by collecting medical and family history, a physical exam, routine blood testsand biopsy. The hemochromatosis is treated by phlebotomy. Phlebotomy rids the body of extra iron. This treatment is simple, inexpensive, and safe.

Introduction:

Hereditary hemochromatosis). Iron is important because it is part of hemoglobin, a molecule in the blood that transports oxygen from the lungs to all body tissues. However, too much iron in the body leads to iron overload a buildup of extra iron that, without treatment, can damage organs such as the liver, heart, and pancreas; endocrine glands; and joints

Causes and Risk Factors:

Factors that increase risk of hereditary hemochromatosis include:

- ✓ Having 2 copies of a mutated HFE gene. .
- ✓ Family history- having a first-degree relative
- ✓ Ethnicity.
- ✓ Sex- Men are more likely than women

Gene mutations that cause hemochromatosis. A gene called HFE is most often the cause of hereditary hemochromatosis. The child inherits one HFE gene from each of thier parents. The HFEgene has two common mutations, C282Y and H63D. Genetic testing can reveal whether child have these mutations in HFE gene

Primary Hemochromatosis:

Inherited genetic defects cause primary hemochromatosis, and mutations in the *HFE* gene are associated with up to 90 percent of cases

Secondary Hemochromatosis:

Hemochromatosis that is not inherited is called secondary hemochromatosis. The most common cause of secondary hemochromatosis is frequent blood transfusions in people with severe anemia

Neonatal Hemochromatosis:

Neonatal hemochromatosis is a rare disease characterized by liver failure and death in fetuses and newborns

Signs and Symptoms:

Some people with hereditary hemochromatosis never have symptoms. Early signs and symptoms often overlap with those of other common conditions. Common symptoms include:

- ✓ Joint pain
- ✓ Abdominal pain
- ✓ Fatigue
- ✓ Weakness

Later signs and symptoms of the disease may include:

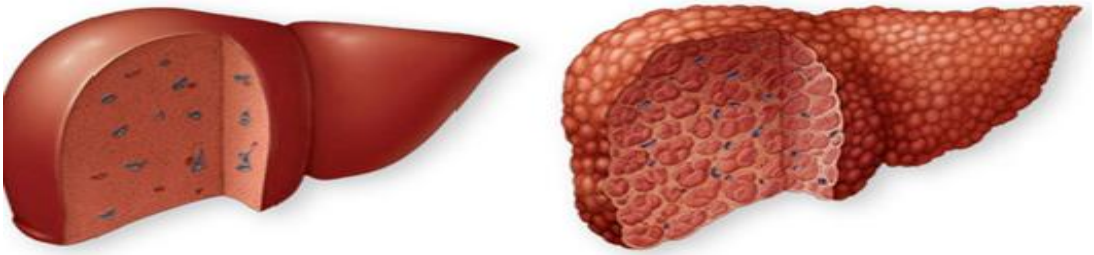
- ✓ Diabetes
- ✓ Loss of sex drive
- ✓ Impotence
- ✓ Heart failure
- ✓ Liver failure

Complications:

Without treatment, iron may build up in the organs and cause complications, including

- ✓ cirrhosis, or scarring of liver tissue
- ✓ diabetes
- ✓ irregular heart rhythms or weakening of the heart muscle
- ✓ arthritis
- ✓ erectile dysfunction

The complication most often associated with hemochromatosis is liver damage. Iron buildup in the liver causes cirrhosis, which increases the chance of developing liver cancer.



A normal liver (left) shows no signs of scarring. In cirrhosis (right), scar tissue replaces normal liver tissue.

Diagnosis:

- ✓ **Medical and Family History:** Taking a medical and family history help to diagnose hemochromatosis. The family history of arthritis or unexplained liver disease.
- ✓ **Physical Exam:**
  - examines a patient's body
  - uses a stethoscope to listen to bodily sounds
  - taps on specific areas of the patient's body
- ✓ **Blood Tests:** Blood tests can determine whether the amount of iron stored in the body is higher than normal
  - The transferrin saturation test shows how much iron is bound to the protein that carries iron in the blood. Transferrin saturation values above or equal to 45 percent are considered abnormal.
  - The serum ferritin test detects the amount of ferritin - a protein that stores iron - in the blood. Levels above 300 μg/L in men and 200 μg/L in women are considered abnormal. Levels above 1,000 μg/L in men or women indicate a high chance of iron overload and organ damage.
  - If either test shows higher-than-average levels of iron in the body, can order a special blood test that can detect two copies of the *C282Y* mutation to confirm the diagnosis.
- ✓ **Liver Function Tests:** These tests can help identify liver damage.
- ✓ **MRI:** An MRI is a fast and noninvasive way to measure the degree of iron overload in your liver.
- ✓ **Testing for Gene Mutations:** Testing your DNA for mutations in the HFE gene is recommended to find out high levels of iron in blood.
- ✓ **Liver Biopsy:** A procedure that involves taking a piece of liver tissue for examination with a microscope for signs of damage or disease.

Treatment:

Treatment for hemochromatosis is by drawing blood. This process is called phlebotomy. This treatment is simple, inexpensive, and safe. The serum ferritin levels periodically monitor iron levels. The goal is to bring serum ferritin levels to the low end of the average range and keep them there. Depending on the lab, the level is 25 to 50 μg/L.Serum ferritin tests every 6 months or once a year will help determine how often a patient should have blood drawn.

Diet and Nutrition:

Iron is an essential nutrient found in many foods. People with hemochromatosis absorb much more iron from the food they eat compared with healthy people. People with hemochromatosis can help prevent iron overload by

- ✓ eating only moderate amounts of iron-rich foods, such as red meat and organ meat
- ✓ avoiding supplements that contain iron
- ✓ avoiding supplements that contain vitamin C, which increases iron absorption

Prognosis:

Treating hemochromatosis before organs are damaged can prevent complications such as cirrhosis, heart problems, arthritis, and diabetes by treating hemochromatosis can stop the progression of liver damage in its early stages and lead to a normal life expectancy.

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