



Haemochromatosis

What is haemochromatosis?

Haemochromatosis is a common inherited disorder that causes the body to absorb and store too much iron

How common is hemochromatosis?

The carrier rate affects 1 in 10 (heterozygote). The homozygote state seen in up to 85% of haemochromatosis affects approximately 1 in 250 people.

What are the symptoms?

Many people have no symptoms in advanced cases. Damage to the liver can slowly lead to cirrhosis if the illness is not treated. Damage to the pancreas can result in diabetes mellitus. Other symptoms include arthritis, heart problems, impotence and fatigue

Are there special tests for iron overload?

Transferrin saturation over 50% may indicate iron overload. An additional test is serum ferritin level, and a HFE gene test should also be arranged.

How can hemochromatosis be treated?

250ml of blood is removed each week until iron stores go down to a normal level. It may take from several months to several years to remove all excess iron. After the iron stores are reduced to normal, the therapy should then be continued every 2 to 4 months for life to prevent re-accumulation of iron. 250mg Iron is in 250ml of blood. The aim is to achieve a ferritin count that remains below 50.

What is the outlook for patients?

Those who are treated early can look forward to a completely normal active life. When the illness has advanced to the stage of cirrhosis, the situation is more serious. Liver cancers can occur in up to 25% of these patients.

Who is most likely to get hemochromatosis?

The gene is inherited from both parents. Hemochromatosis is most often diagnosed between the ages of 40 and 60, but it has been detected in younger and older people. Women frequently develop symptoms at a later age than men since women normally lose significant amounts of iron through menstruation, pregnancy and lactation.

Does having anemia rule out iron overload?

No. There are many forms of anemia, and it is possible for a person to have both anemia and haemochromatosis.

Is there any relationship between diet and iron overload?

Haemochromatosis develops in individuals eating normal diets. Rarely, people have developed iron storage problems after taking heavy amounts of iron tonics and medications over a long period. A normal, balanced diet is recommended. No one should take iron supplements without a doctor's advice. People who are receiving treatment for haemochromatosis do not have to follow a special diet. There is no evidence that the condition is worsened by including moderate amounts of iron-rich foods such as red meat and organ meats in the diet and they may also be advised to avoid vitamin C supplements, which promote iron absorption.

What effect does alcohol have on haemochromatosis?

Alcohol seems to intensify the problem. Anyone with a liver problem is advised to abstain from alcohol or have only an occasional drink. Moderate, alcohol does not pose a health problem for people with hemochromatosis who do not have liver disease

What are the implications for your family?

Doctors usually recommend that first-degree relatives (parents, siblings, and children) of people with known haemochromatosis undergo screening. For example, there is a 25% chance that a full brother or sister of a patient with hereditary hemochromatosis (with two copies of the C282Y mutation) will have hemochromatosis.

What is the prognosis?

Most people with haemochromatosis have a normal life expectancy, but survival is shortened in people who develop cirrhosis or diabetes mellitus. Studies suggest that liver cancer is the leading cause of death among people with hemochromatosis, followed by cirrhosis, heart disease, and diabetes.

Haemochromatosis HFE gene

Of all patients with haemochromatosis:

- 85% C282Y homozygote
(Two major abnormal genes)
- 3% C282Y H63D compound heterozygotes
(One major & one minor abnormal gene)
- 2% C282Y heterozygote
(One major abnormal gene)
- 10% that remain 50% heterozygote for H63D alone
(One minor abnormal gene)

What is the risk of developing haemochromatosis in my lifetime?

◆ Normal genotype	No increased risk	cys/cys	his/his
◆ Homozygote C282Y	>x2000 fold risk	tyr/tyr	his/his
◆ Compound C282Y/ H63D	x25 fold risk	cys/tyr	his/asp
◆ Heterozygote C282Y	x5 fold risk HH	cys/tyr	his/his
◆ Homozygote H63D	x8 fold	cys/cys	asp/asp
◆ Heterozygote H63D	x2 fold	cys/cys	his/asp

The major haemochromatosis gene C282Y may not be fully apparent:

- 50% of homozygotes fully express the disease and were symptomatic
- 20% of homozygotes have increased hepatic iron stores BUT are asymptomatic
- 30% of homozygotes NOT express disease