Educational Handouts for Clients

Haemachromatosis – a patient's guide

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What is it?

Haemochromatosis is a genetic disorder of Iron metabolism very common in those of Celtic, Anglo and Northern European descent. Absorption of iron through the intestine is uncontrolled even when body saturation levels have been reached—the excess iron is very toxic to body organs. The consequences do not occur until this has been happening for several years.

It is a silent killer eventually causing liver cirrhosis (and cancer), heart damage, arthritis, diabetes and sexual dysfunction. Some undiagnosed iron overload sufferers, whose main symptom is fatigue, take iron tablets for years without medical advice — these supplements are a serious danger to people with haemochromatosis.

Early diagnosis and treatment prevents these complications. Those individuals with no organ damage have a normal life span.

Those who already have significantly damaged organs can have more serious problems and need ongoing monitoring and treatment, such as insulin for diabetes. Liver cirrhosis is associated with liver cancer, and ongoing surveillance is recommended for those so affected as some cancers can be detected early when surgery or transplant is still possible.

Blood tests that are done to check for haemochromatosis include an iron saturation test and a serum ferritin. Most affected (but not all) have an increased iron saturation of >55% (often expressed as >0/55), and/or a raised serum ferritin. The former test is more specific — the serum ferritin can be "falsely" elevated in situations of illness including infection, inflammation, cancer or liver disease.

Gene Test

There is a mutation in a gene, designated the HFE gene which can identify most patients with haemochromatosis. Once this abnormality is found in an individual, the diagnosis and screening of members of their family is relatively easy.

A heritable, but non-HFE, form of iron overload occurs in some black populations of African origin.

Inheritance

Haemochromatosis is common in people of northern European origin, affecting at least one in 200. More than one in ten are carriers of the condition, the latter usually without any clinical consequences. In most cases a genetic abnormality can be identified.

Years ago haemochromatosis probably protected those who lost a lot of blood in battles or from childbirth.

To inherit two abnormal genes, both parents of an affected person must have either: 1) one copy of the abnormal gene (they are both carriers or heterozygote), or 2) one is a carrier and the other has haemochromatosis, or 3) both parents can have haemochromatosis.

Children of an affected person are all at least carriers. If the second parent is a carrier, 50% will have haemochromatosis.

Suspected Haemochromatosis

Of those with abnormal iron tests (iron saturation > 0.55 and/or a raised serum ferritin), many will be homozygous with two copies for the HFE gene mutation.

The gene test is useful in making the initial diagnosis, as well as detecting other affected family members. The 10% of cases without the common gene mutation have other genes or factors — some are unidentified as yet.

In those diagnosed on blood iron studies and/or clinical findings with negative gene test, a liver biopsy should be done to help confirm the diagnosis and to exclude liver cirrhosis. A liver biopsy is not needed in those with the mutation to make the diagnosis, but is done in those with a very high ferritin level and/or abnormal liver tests to check for liver damage.

Family Screening

First-degree relatives (parents, brothers, sisters, children) of an affected person with the HFE mutation should be screened with iron studies (saturation, serum ferritin) and the gene test. The latter is particularly useful in identifying young people who may be affected but as yet have normal iron tests.

Population Screening

Patient support groups have welcomed the gene test and have pushed for screening of populations at risk. However, the presence of the gene mutation does not always correlate with disease expression. There are other genetic or environmental factors, which contribute to the amount of iron loaded and any clinical consequences. Before the gene test was available, it was recommended that first degree relatives with normal iron studies had iron tests repeated every 3 to 5 years.

Obviously in a family known to have the HFE gene, the testing of other members is important and easy.

Mass screening of Europeans using iron saturation and/or ferritin tests has been proposed and is controversial. Some are dubious that such an approach would be cost effective. Others believe that the testing would prevent expensive end-stage treatments and save lives.

Studies aimed at providing information about screening are currently underway. Certainly those found to have increased iron saturation and ferritin should have the gene test performed.

Those with liver disease and/or arthritis, unexplained heart disease, diabetes or cirrhosis should also be tested.

Treatment

Treatment is life long, but usually relatively simple. The aim is to reduce the total amount of body iron and then to maintain a low iron level to prevent further damage (ferritin less than 100Ug/L).

This is done by regular blood removal (also known as Phlebotomy or Venesection), with monitoring of blood ferritin levels. Each ml of blood removed contains approximately 1 mg of iron — the body replaces the blood by drawing iron from the tissues. The amount of blood removed at each session should be no more than 10% of the blood volume (8mls/kg), and may need to be less in those with bad heart disease such as angina.

How many treatments needed will depend on the rate of blood replacement and the severity of the iron overload.

It is important to remove the excess iron as quickly as possible to switch off ongoing organ damage, so initially venesections should be done once or twice weekly if tolerated.

Once the iron levels are low, venesections are then needed three or four times a year to maintain the body iron at a safe level.

A normal diet can then be eaten, although excessive red meat and liver should be avoided.

For those with iron overload, iron tablets and vitamin C supplements which increase iron absorption should be avoided, as well as raw oysters and other shellfish which can transmit a dangerous organism.

Drinking alcohol in unsafe amounts often causes symptoms to develop sooner because alcohol increases the absorption of iron from the diet.

Blood Donation

It is very likely that as many as one in 200-blood donors have undiagnosed early haemochromatosis and that their blood is currently being transfused safely. However in some countries, healthy people with haemochromatosis are retired as blood donors when the diagnosis is made. The concern when there is a financial charge for therapeutic venesections relates to the fact that the desire to donate blood is then not entirely altruistic. Other blood services allow those with early-uncomplicated haemochromatosis to donate as long as all other donor criteria are fulfilled.

Haemochromatosis Support Group

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