HAEMOCHROMATOSIS

(Iron Overload Disease)

Haematology Department
Auckland City Hospital
Patient Information Leaflet
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HAEMOCHROMATOSIS

INTRODUCTION;

Iron in small quantities is essential to life, particularly for the function of haemoglobin, the blood protein which carries oxygen to the tissues. Normally iron is taken into the body from food via the intestine (known as "absorption"). Once absorbed the body has no way of getting rid of excess iron. Some people have a disease called ***haemochromatosis*** in which too much iron is absorbed. If this disease is not diagnosed and treated iron can damage vital organs and shorten a person's life

WHAT IS HAEMOCHROMATOSIS?

Haemochromatosis is a genetic (family inherited) disorder in which too much iron is taken into the body over and above the needs of the body. It is caused by an abnormal gene.

A gene is a code for a family likeness or characteristic. There are millions of genes located on our 23 pairs of genetic material (called Chromosomes) that we inherit from our parents, half from each parent.

Individuals inheriting one haemochromatosis gene and one normal gene are called ***carrier****s*. Their iron absorption may be slightly higher than normal but they do not absorb enough iron to cause any significant health problems. However, if two carries marry, each of their children has a 25% chance of inheriting two haemochromatosis genes and a 50% chance of inheriting one haemochromatosis gene.

Individuals inheriting two haemochromatosis genes will absorb far too much iron. This iron slowly builds up in the liver, heart, pancreas and other hormonal glands, and joints. It takes many years to build up iron to a level which causes damage to these organs, but by the time the damage occurs, it is often too late for the organ to repair itself and some permanent damage may remain.

HOW COMMON IS HAEMOCHROMATOSIS?

About one person in every 400 has the disease, while about 10% of our population are carriers of one haemochromatosis gene (but other wise unaffected). This makes haemochromatosis one of the most common genetic diseases in our society, although many people are only mildly affected.

WHAT ARE THE SYMPTOMS OF HAEMOCHROMATOSIS?

Symptoms vary considerably among patients, and may resemble those of many other medical conditions, making diagnosis difficult. The symptoms may include fatigue, weakness, weight loss, abdominal discomfort and joint pain. Grey skin pigmentation may also occur. Other symptoms may develop later as a result of organ damage to the liver, heart (palpitations, shortness of breath, chest pain), pancreas (thirst or frequent urination as a result of diabetes), or other hormonal deficiencies due to hypopituitarism (loss of sex drive or body hair). However, most young people with the disease have no symptoms or only minor symptoms in the early stages of the disease.

WHO MAY BE AT RISK OF HAVING HAEMOCHROMATOSIS?

1. Blood relatives of patients (particularly close relatives such as brother, sisters and children)

2. Individuals with symptoms of the disease.

3. Individuals with diabetes, arthritis and certain heart problems

If you have some of the symptoms mentioned above, do not over-react and conclude that you have haemochromatosis because there are many other causes for such symptoms. See your family doctor and discuss your concerns.

HOW IS THE DIAGNOSIS MADE?

The two most useful screening blood tests for haemochromatosis are:

1. Serum transferrin or TIBC and saturation

2. Serum ferritin (may be normal early in the disease)

If the above tests are abnormal on at least two occasions, the following tests may be required to confirm the diagnosis

1. Haemochromatosis gene testing. Studies to date suggest that at least 90% of patients with haemochromatosis have a mutation of this gene, which can be detected on a blood test.

2. A Liver biopsy. This may be done if the liver function tests are abnormal and it involves removal of a small piece of liver with a needle under local anaesthetic.
   The biopsy is examined for the amount of iron in the liver and potential damage to the normal liver cells from the build-up of iron.

Once the diagnosis of haemochromatosis is made, additional investigations may be undertaken to assess if there is any organ damage from the iron overload. These tests might include a chest x-ray and ECG and blood tests for liver function, thyroid function, glucose level and testosterone level.

SCREENING OF RELATIVES:

After a diagnosis of haemochromatosis is made, all close relatives over the age of 10 - 15 years, should be screened for haemochromatosis. Close relatives includes brothers, sisters, parents and children. Cousins, aunts and uncles may also be tested, although their risk is much lower.

Family screening will include the following blood tests:

1. Serum transferrin ot TIBC and saturation

2. Serum ferritin

3. Haemochromatosis gene testing.
   This is indicated where the affected family member has been shown to have the haemochromatosis gene mutation.

Early diagnosis and treatment of family members with the disease is essential to prevent organ damage.

HOW RECESSIVE INHERITANCE WORKS WHEN BOTH PARENTS ARE CARRIERS

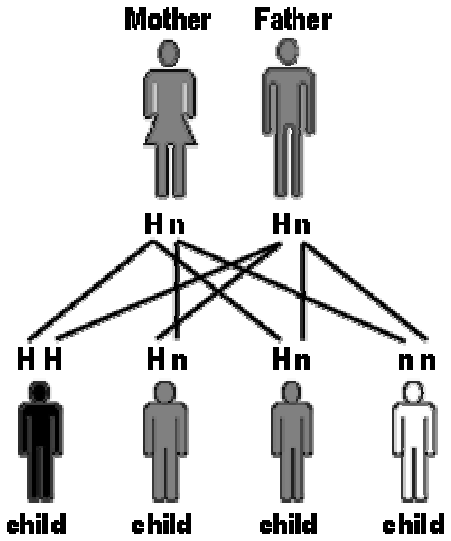
HH = person with haemochromatosis

Hn = carrier of Haemochromatosis

Nn = normal

H = gene for Haemochromatosis

N = normal gene



IS THERE TREATMENT?

Yes. By removing about 500mls of blood, as in routine blood donation, usually once a week, the body is stimulated to make more blood and this uses up the excess iron. This is called ***"venesection treatment".*** Depending on the amount of iron in the body the initial treatment may take one or two years. Blood tests are done to monitor the iron removal. Once the excess iron has been removed, venesections are done about four times a year to prevent iron building up again. This treatment needs to be life long.

HOW EFFECTIVE IS TREATMENT? WHAT IS THE OUTCOME?

There is good evidence that with removal of excess iron, patients feel better, stronger, their grey colour lessens, liver size decreases, diabetes may improve, and heart function improves. If treatment has been commenced early, damage to the liver and other organs may be completely prevented, with life expectancy equivalent to those who do not have the condition. If cirrhosis (liver scarring) is present it is usually not reversed by treatment but should not get worse. Without venesection treatment, iron continues to build up and organ damage continues.

It is not possible to treat haemochromatosis with a low iron diet, since iron is present in most foods, and it is the iron already in the body which will cause damage. However, it is strongly advised that patients with haemochromatosis do not take iron tablets of any type, nor vitamin tablets containing vitamin C (ascorbic acid). Alcoholic drinks in small quantities (e.g. 2 glasses or less per day) are not usually harmful, but if there is liver damage your doctor may advise you to have no alcohol

QUESTIONS OR CONCERNS

If you have any questions about Haemochromatosis or any of the material in this pamphlet you should talk to your family doctor.