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facts about...

HAEMOCHROMATOSIS

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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 the iron is 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, recently described and called the HFE 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 carriers. Their iron absorption may be slightly higher than normal but most do not absorb enough iron to cause any significant health problems. If two carriers marry, each of their children has a 25% chance of inheriting two haemochromatosis genes and a 50% chance of inheriting one haemochromatosis gene. Carriers can now be identified by a new gene test for the HFE 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 300 has the disease haemochromatosis while about 12% of our Australian population are carriers of one haemochromatosis gene. This means that in a city of the size of Sydney there are approximately 12,000 people affected by the disease and 400,000 people carrying one haemochromatosis gene. This makes haemochromatosis one of the commonest genetic diseases in our society, although many people are only mildly affected.

What are the symptoms of Haemochromatosis?

Symptoms vary considerably among patients. The symptoms resemble those of many other medical conditions, making diagnosis difficult. The symptoms may include fatigue, weakness, weight loss, abdominal discomfort and joint pain. A tanned appearance, not due to sun exposure, 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 (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 Haemochromatosis?

- Blood relatives of patients (particularly close relatives such as brothers, sisters and children).
- Individuals with symptoms of the disease.
- Individuals with diabetes, arthritis and certain heart problems.

If you have some of the symptoms mentioned above, do not overreact 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?

A blood relative of someone with haemochromatosis should have a simple blood test for the HFE gene to see if they are at high risk of haemochromatosis. This blood test is available (after discussion with your general practitioner) through usual pathology laboratories. In people without any family history of haemochromatosis, the two most useful initial blood tests are:

- Serum transferrin saturation (best test).
- Serum ferritin (may be normal early in the disease).

These tests can be done on the one sample of

blood. If the tests are abnormal on at least two occasions, a further blood test looking for the HFE gene may be all that is required to confirm the diagnosis of haemochromatosis. Some patients will require a liver biopsy to confirm the diagnosis and/or assess the degree of damage to the liver. Liver biopsy involves removal of a small piece of liver with a needle under local anaesthetic. It is a safe procedure when done by a medical practitioner experienced in the technique.

Screening of relatives

After a diagnosis of haemochromatosis is made, all close relatives over the age of 10 years should be screened for haemochromatosis. Close relatives include brothers, sisters, parents and children. Cousins, aunts and uncles should also be tested, although the risk is much lower. Family screening will include an examination by a doctor, with blood testing as described above, to determine which family members are likely to have the disease. In some cases, blood tests will need to be repeated in 2 years as sometimes the excess iron does not become apparent until later life. Early diagnosis and treatment of family members with the disease is essential to prevent organ damage.

Is there a treatment?

Yes, By removing about 500ml of blood, as in 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. Treatment should continue for the rest of the person's life.

Venesection treatment must only be done by medical or nursing staff experienced in this technique. Venesections can be done at some hospitals, some pathology laboratories or some general practices. On occasions, venesections can be organised through blood banks, after referral from a specialist. Immediately prior to a venesection, rest for 15 minutes and drink 500ml of fluid. After the venesection, stand up slowly and sit in a chair for 15 minutes, keeping pressure on your arm for 5 minutes where the needle was inserted.

How effective is the treatment?
What is the outcome?

There is good evidence that with removal of excess iron, patients feel better, stronger, their tan 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. 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 advised that patients with haemochromatosis do not take iron tablets of any type, nor vitamin tablets containing iron or vitamin C (ascorbic acid) as vitamin C can increase iron absorption. People with haemochromatosis may also consider reducing red meat intake (eg. to approximately 90-120 g/day). Alcoholic drinks in small quantities (eg. two glasses or less per day) are not usually harmful, but if there is liver damage your doctor may advise you to have no alcohol.

Are there support groups?

Yes. Some large hospitals have support groups for patients and relatives with liver diseases and haemochromatosis. If not, you could contact the: Haemochromatosis Society of Australia, 412 Musgrave Road, Coopers Plains, QLD, 4108 (Tel: (07) 3345 7583)or the Digestive Health Foundation (see below).

This information booklet has been designed by the Digestive Health Foundation as an aid to people who have Haemochromatosis or for those who wish to know more about it. This is not meant to replace personal advice from your medical practitioner.

The Digestive Health Foundation (DHF) is an educational body committed to promoting better health for all Australians by promoting education and community health programs related to the digestive system.

The DHF is the educational arm of the Gastroenterological Society of Australia, the professional body representing the Specialty of gastrointestinal and liver disease in Australia. Members of the Society are drawn from physicians, surgeons, scientists and other medical specialties with an interest in GI disorders.

Since its establishment in 1990 the DHF has been involved in the development of programs to improve community awareness and the understanding of digestive diseases.

Research and education into gastrointestinal disease are essential to contain the effects of these disorders on all Australians.

Further information on a wide variety of gastrointestinal conditions is available on our website.

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