

Patient Information Sheet:

OPTIMAL COMMUNITY SCREENING FOR HAEMOCHROMATOSIS

WHAT IS HEREDITARY HAEMOCHROMATOSIS (HH) ?

This is the most frequently inherited (genetic) disorder in the Caucasian population. The disease is characterised by massive amounts of iron being accumulated in the body risking damage to the liver, pancreas, heart, joints and endocrine glands, and often causing diabetes, gonadal failure and pigmentation of the skin. The normal amount of total iron in an adult is approximately 3.5 grams mainly carried as haemoglobin in the blood or stored as ferritin and used by the body for its growth. If the excess iron is left to accumulate for a long time, organ damage will occur and when diagnosed at a later age (at 40-50 years of age) irreversible damage to the organs would have already caused serious consequences.

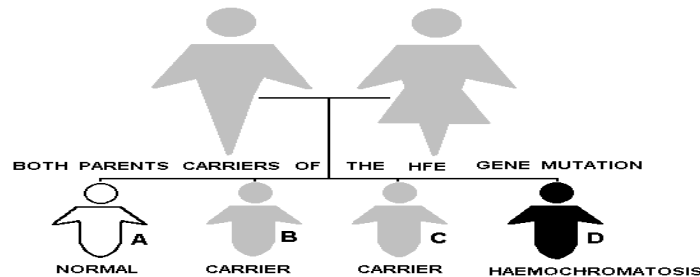
Haemochromatosis can also be acquired in association with related medical problems.

WHO IS LIKELY TO GET HEREDITARY HAEMOCHROMATOSIS ?

Genetic diseases are often more common in specific community groups. HH is most common amongst white North Europeans. About 1 in 8 Caucasians are carriers (heterogeneous) of the faulty gene (mutation) and are mostly unaffected by the disorder. Approximately 1 person in 250 will inherit a 'double dose' (homozygous) of the faulty gene and 85 % of these will develop a severe form of the disease, the detection of which may occur as late as middle age, by which stage organ damage has most probably already occurred.

WHAT CAUSES HEREDITARY HAEMOCHROMATOSIS ?

Hereditary Haemochromatosis is a result of an inborn error of metabolism and is caused by inheriting a 'double dose' of a faulty gene (HFE), which causes massive amounts of iron to accumulate in the body. Screening of asymptomatic patients is performed by measuring levels of iron and iron binding capacity in the blood. If possible iron overload is suggested by screen results or clinical symptoms, diagnosis is made by measuring iron concentration, transferrin saturation and serum ferritin in blood samples as an indication of the degree of iron storage levels. Confirmation of diagnosis is made by genetic (DNA) testing showing the presence of the mutation C282Y and/or H63D in the HFE gene. The HFE gene is located on the short arm of chromosome 6.



HOW IS THE DISEASE TRANSMITTED ?

This disease is transmitted by autosomal recessive mode i.e. When two unaffected carriers of the HFE gene mutation become parents, then for each pregnancy:

- There is a 1-in-4 (25%) chance that the child will inherit the normal gene from each parent and will be completely free of the gene mutation and will not be affected by Haemochromatosis (figure A).
- There is a 2-in-4 (50%) chance that the child will inherit both a regular copy and a faulty copy of the HFE gene and will be a carrier of the faulty gene, unaffected by the disease, just like his/her parents (figures B and C).
- There is a 1-in-4 (25%) chance that the child will inherit the faulty gene from each parent (figure D). Without early diagnosis and monitoring this child will develop symptoms of Haemochromatosis in 85% of cases.

WHAT ARE THE BENEFITS OF PARTICIPATION IN THIS PROJECT ?

Early diagnosis of HH in young, asymptomatic adults will prevent the disease from developing harmful consequences by appropriate preventive management. Testing is particularly relevant for persons with history of the disease in their family. Targetted testing amongst patients with symptoms related to haemochromatosis (eg. diabetes and heart muscle disease) is intended to identify patients sufficiently early to reverse or prevent further complications. Testing is available for all men and women over the age of 16.

WHAT DOES PARTICIPATION INVOLVE ?

Participation involves collection of 5 mL of blood to measure the level of iron and iron-binding capacity to screen for possible iron overload. If iron overload is suspected by the screen result or by clinical symptoms, 10 mL of blood is collected for serum ferritin, iron and transferrin saturation to assess iron storage levels and for confirmatory genetic tests for the presence of the gene mutation HFE C282Y and/or H63D.

WHAT ARE THE RISKS OF PARTICIPATION IN THIS PROJECT ?

Possible physical adverse effects include discomfort, swelling and bruising at the needle puncture site. Other risks include potential discrimination by any life-insurance company that may require disclosure of abnormal genetic test results. (There are currently no anti-discrimination laws for results of genetic test, but these issues of discrimination are being addressed by professional and peer support genetic groups to government and insurance groups)

DOES IT AFFECT ONE'S HEALTH TO BE A CARRIER OF THE HFE GENETIC MUTATION ?

Carriers of the HFE gene mutation are mostly unaffected by the disorder Haemochromatosis.

DOES HAEMOCHROMATOSIS DISEASE AFFECT ONLY MALES ?

No. Haemochromatosis disease can affect both males and females. Males however, are affected in larger numbers than the females and are usually diagnosed earlier in life. This occurs probably because females lose blood during menstruation and childbirth or may have a lower intake of iron and this protects them from iron overload.

IS IT POSSIBLE TO TEST FOR THE DISEASE OR TO DETERMINE IF YOU ARE A CARRIER OF THE FAULTY HFE GENE ?

Yes. In the first instance the degree of iron storage levels in the blood (Serum Ferritin) and the level of iron in the blood (Transferrin Saturation Concentration) are determined. If necessary a genetic test may be carried out for the presence of the HFE gene mutations.

WHY IS A PERSON'S ANCESTRY RELEVANT TO GENETICS CARRIER TESTING ?

There are different faults (mutations) found in genes. Some faults are specific to particular ethnic groups or geographical regions in the world and are more common among individuals who are of particular ancestries, origin and descent. Hereditary Haemochromatosis is more common in Caucasians. Our staff will need to ask about your origin and find out whether you are of white European ancestry.

IF I DO NOT HAVE FAMILY HISTORY OF HH DISEASE IS TESTING STILL RELEVANT ?

Yes. Over 95% of people affected by HH have no family history of the disease because the HFE faulty gene is silently passed down through the generations. However, if there is a family history, then the risk is much greater. People who have a known family history of HH should seek further information through genetic counselling. For counselling call (02) 9926 7324 and the NSW Haemochromatosis Support Society's number is (02) 4473 9099.