

WHAT TO DO NEXT

If you have Haemochromatosis it is vital that family members are screened for the condition so that treatment can be given before serious organ damage occurs. If caught in time, Iron Overload is treatable and the effects are preventable.

For the same reason, health care professionals need to be alerted to the early clinical features of HH. As people can suffer from the ravages of Haemochromatosis, yet still be completely unaware of the condition until it's too late...

So, if you have two or more of the mentioned symptoms or have a relevant family history, approach your doctor about ALL your symptoms or contact us now.

YOU can help by:

- Asking your doctor about iron overload
- Talking to others about it if you have HH
- Getting involved with the Society
- Refer patients for HH testing if you are a medical professional
- Making a donation to the Society

RELATED WEBSITES & SOCIETIES

- **South African Haemochromatosis Society website**
www.haemochromatosisza.org
- Canadian Haemochromatosis Society
<http://www.cdnhaemochromatosis.ca>
- Iron Disorders Institute
www.irondisorders.org
- Haemochromatosis Society UK
www.haemochromatosis.org.uk
- Other Links
www.haemochromatosis.co.uk

WHO ARE WE?

The Haemochromatosis Society of South Africa is a non-profit organization originally established by Marie Warder in 1987. The HSSA has recently been re-established with the primary aim of raising awareness of Haemochromatosis throughout South Africa. In order to sustain this we need your support!

The objectives of the HSSA are:

- To promote awareness of HH in the medical profession and amongst the SA public in order to ensure the early diagnosis and screening of potential HH sufferers.
- To provide informative literature to the public and publicise current debate and research regarding iron related disorders, as well as establish an online reference for questions and discussion.
- To establish support groups for, and a central registry of, those affected by HH, as well as a registry of doctors and specialists aware of HH and its treatment.

FOR MORE INFORMATION CONTACT HSSA

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Acknowledgements and thanks to the Prof AP MacPhail and Dr MJ Kotze for their kind assistance in the production and editing of this brochure.

IRON OVERLOAD

Do you suffer from...

- Abdominal pain/diarrhoea
- Cirrhosis of the liver
- Arthritis/joint pain
- Late-onset diabetes
- Irregular heartbeat
- Chronic fatigue
- Loss of libido/impotence
- Porphyria

"...then ask your doctor for an iron studies test!"



WHAT IS "IRON OVERLOAD"?

Hereditary Haemochromatosis (HH) is a genetic condition in which there is excessive absorption of iron from a normal diet leading to iron overload. As the body has no natural way of excreting iron due to the genetic defect, the excess iron accumulates in the liver, pancreas, heart and other organs, eventually causing organ failure. Symptoms could typically appear in middle-age after years of damage, although HH can affect young persons in their early 20's, as well as children.

Haemochromatosis is an inherited disorder due to a genetic defect. Several genetic risk factors have been identified, including mutations in the HFE, transferrin receptor-2 and ferroportin genes. Three main mutations account for the disease in more than 80% of cases in most populations.

Those who inherit a defective copy of the gene from both parents are homozygous and are at risk of developing the disease, whereas those who inherit from one parent are carriers who are unaffected or may show a smaller increase in iron absorption.

POSSIBLE FEATURES

Not all of these features are necessarily present, but the following are typical of HH:

- Chronic fatigue
- Arthritis - all joints but particularly between the knuckles of the 1st & 2nd finger (iron fist)
- Cirrhosis and enlarged liver
- Diabetes - due to damage to the pancreas
- Impotence, loss of libido & testicular atrophy
- Menstrual irregularities (amenorrhea)
- Abdominal pain (upper right quadrant)
- Mood swings/ depression
- Irregular heartbeat (arrhythmia)

ISN'T IRON OVERLOAD A RARE CONDITION?

Genetic studies have shown that approximately 17% of the Caucasian population of South Africa (i.e. up to 1 out of 6 people) are carriers of the most common HH gene mutation (C282Y). Furthermore, up to 1 out of 115 could have inherited it from both parents and, as homozygotes, are at serious risk of loading iron. According to the Center for Disease Control in Atlanta, US, Haemochromatosis is *THE MOST COMMON* Genetic Disease to affect the Caucasian population. Yet it continues to be largely unrecognised, misdiagnosed and considered to be rare by many doctors.

IDENTIFY AN AFFECTED PERSON AND SAVE A FAMILY

WHO IS AT RISK?

Most of the suffering and clinical symptoms associated with HH is *PREVENTABLE* if potential victims are detected at a young age or before organ damage occurs. Those at risk are persons of European descent, particularly the Irish, Scots, French and English; as well as those who have a family history of arthritis, diabetes, liver disease or heart failure. In particular, *ALL* relatives of an HH sufferer should be tested. As carriers do not necessarily develop symptoms HH can be passed on in a family unnoticed. However, the offspring of 2 carriers will have a 25% (1 in 4) chance of being homozygous. Many rural South Africans may also be affected by Acquired Iron Overload through eating food cooked in and drinking beer brewed in iron (potjie) pots. Mutations in the ferroportin gene have been identified as important contributing factors to iron loading.

TESTS AND TREATMENT

The diagnosis can be made by requesting iron studies which include determination of transferrin saturation (normally less than 45% in women and 50% in men) and serum ferritin (normally less than 200g/L). If high levels are confirmed other tests to assess organ damage, such as liver biopsy and x-rays, may need to be done. A DNA test (strip-assay) testing for multiple mutations in different genes is now available, which enables accurate diagnosis of HH without the need for an invasive liver biopsy.

Healthy individuals with the genetic defect can prevent iron overload through regular blood donation. There is ongoing treatment for HH through 'blood-letting' or 'phlebotomy'. This procedure reverses the buildup of excess iron and will help reduce or prevent complications. The frequency of the blood-letting will depend on the level of iron in the body and, initially, may need to be done every week. To prevent iron stores re-accumulating it is necessary to continue life long phlebotomy therapy every two to three months to keep transferrin saturation below 30%. There is no strict diet, as iron is present in all foods. Foods rich in iron include meat and liver. Iron and vitamin C supplements must be avoided since vitamin C greatly increases iron absorption and may cause cardiac palpitations in those with iron overload. Drinking tea (not Rooibos) with meals is beneficial because it inhibits iron absorption. *ALCOHOL* should be avoided particularly where there is a suspicion of liver damage.

