



**Pictured:** Horseracing legend Ruby Walsh launched a campaign to reduce the numbers of people suffering unnecessarily from haemochromatosis

## Bonded by blood



Early diagnosis and treatment of hereditary haemochromatosis prevents complications and results in a normal life expectancy, writes **Margaret Mullett**.

**H**ereditary haemochromatosis (HH) is a disorder where the body absorbs too much iron from the diet. The iron is then deposited in, and can ultimately damage, organs such as the liver, heart and pancreas. When a person has too much iron in their body they are said to have 'iron overload'.

The treatment of HH involves the removal of some blood at regular intervals and is called therapeutic phlebotomy. It uses the same procedure as a normal blood donation.

Initially the treatment can mean weekly or twice weekly phlebotomy to rapidly reduce the ferritin levels. After a normal level has been achieved, maintenance may only require three or four sessions per year for the remainder of life.

Ireland has the highest level of this condition in the world. Approximately one in 83 Irish people are predisposed to develop Haemochromatosis.

The underlying cause is the inheritance of a mutated gene. The faulty gene stops the normal body iron control working properly. No two people are alike and symptoms will vary from person to person. Symptoms tend to occur after the age of 40, but may be earlier or later.

### WHO SHOULD BE TESTED?

If you have the above mentioned symptoms or if you have family members with haemochromatosis you should be tested. If your brother, sister, child, or parent has haemochromatosis then you are at increased risk of having the condition.

Your GP can order blood tests to check your iron levels. It is ideally taken after an overnight fast. Both 'serum ferritin' and 'transferrin saturation' are measured.

Serum ferritin (SF) is an iron storage protein. A SF of more than 200ug/l in women and >300ug/l in men suggests iron overload. However, the raised SF could be caused by other reasons and should be taken in combination with transferrin saturation (TS).

TS is a protein that carries iron from the gut around the body. A TS result of more than 45% is strongly suggestive of haemochromatosis and should prompt a genetic test. This test for the known mutations will confirm the diagnosis.

Haemochromatosis is a recessive disorder, which means it only develops if you receive two mutated copies of the gene, one from each parent. When a person has one mutated copy, he or she

### WARNING SIGNS

#### The most common symptoms include:

- Fatigue, general weakness and lethargy.
- Joint pain, knuckles and first joint of the first two fingers are commonly affected.
- Abdominal pain.
- Sexual dysfunction /Loss of Libido
- Discoloration or bronzing of skin

#### Other signs that your GP can test for include:

- Irregular heart beat
- Diabetes
- Hormonal changes
- Enlarged liver

is called a carrier. Carriers do not usually load iron. One in five Irish people are carriers and if two carriers have a child there is a 25% chance that they will have haemochromatosis.

### DIET

Vitamin and mineral supplements containing iron should be avoided if you suffer from haemochromatosis. Consumption of red meat should be reduced and alcohol consumption should be limited.

A healthy, varied diet is recommended. Avoid vitamin C in tablet form, cereals fortified with iron and drink plenty of tea, as tannin prevents the absorption of iron.

### AIMS OF THE IHA

The Irish Haemochromatosis Association aims to encourage vital early diagnosis by raising awareness of the condition, both in the medical profession and within the general public.

The association provides support and information for those suffering with haemochromatosis and encourages the screening of members' extended families.

The organisation is also petitioning the Irish Blood Transfusion Service to extend the clinics for haemochromatosis patients to ensure that valuable blood can be used for transfusion. ■

#### For more information:

- w:** [haemochromatosis-ir.com](http://haemochromatosis-ir.com)  
**e:** [info@haemochromatosis-ir.com](mailto:info@haemochromatosis-ir.com)  
**t:** +353 1 873 5911