PROFESSIONAL Margaret Mullett, Chairperson, Irish Haemochromatosis Association

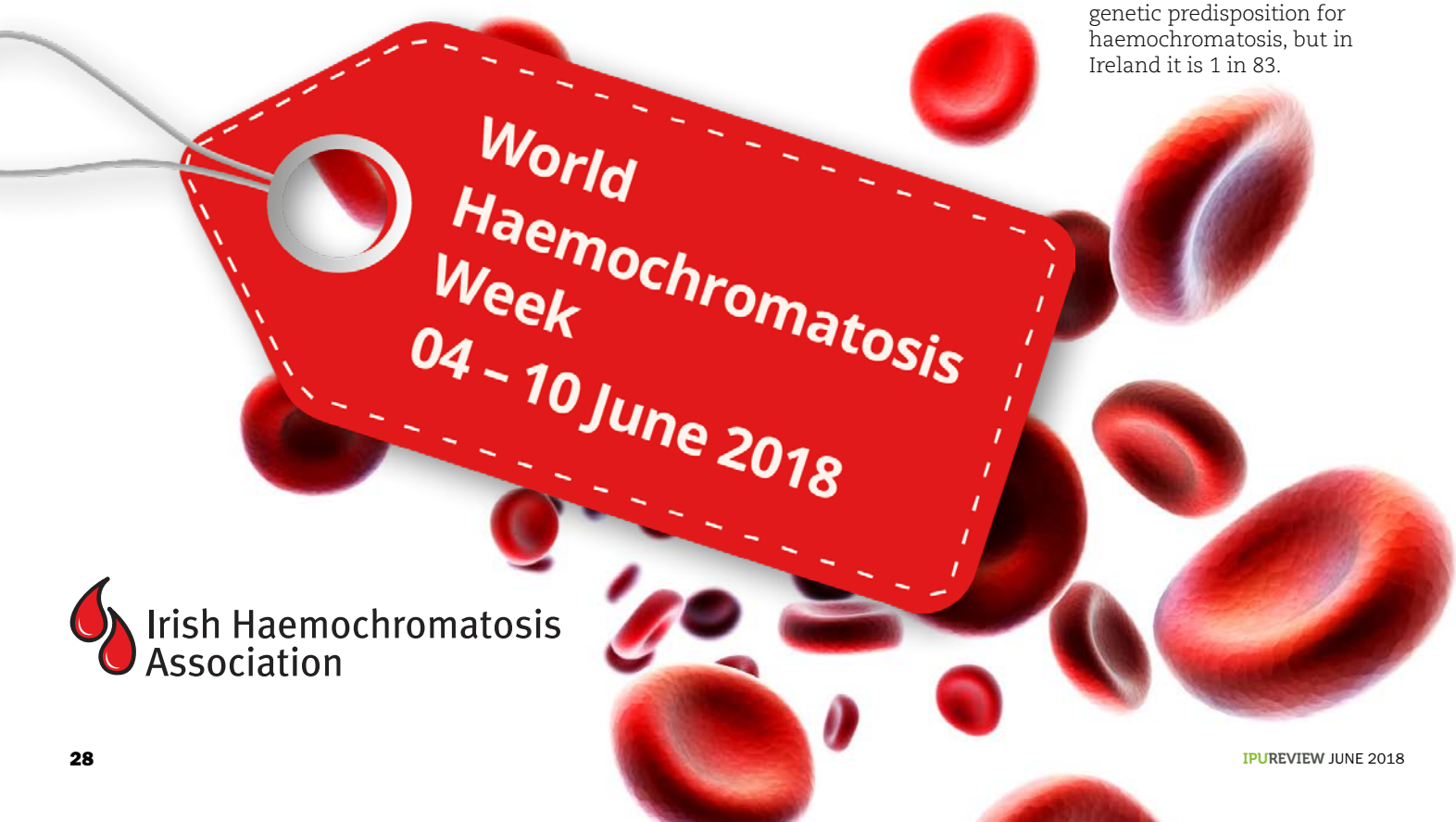
Haemochromatosis – are we pumping too much iron?

National Haemochromatosis Awareness Day is on Thursday 7 June. The Irish Haemochromatosis Association (IHA) is a support group for haemochromatosis patients and their families, and it will be running events across 27 venues around Ireland from 4 – 10 June to raise awareness of haemochromatosis, both in the medical and the general population.

What is haemochromatosis?

Iron overload, or haemochromatosis, is a serious condition in which too much iron is absorbed and stored in the body. It can cause liver cirrhosis, liver cancer, arthritis, diabetes and general fatigue.

Haemochromatosis is hereditary and is more common in Ireland than anywhere in the world. About 1 in 200 people of European origin have the genetic predisposition for haemochromatosis, but in Ireland it is 1 in 83.



Irish Haemochromatosis Association

What are the symptoms?

The most common symptoms noticed by people with haemochromatosis include:

- Fatigue, general weakness and lethargy;
- Joint, knuckle and the first joint of the first two fingers are commonly affected and experience pain;
- Abdominal pain;
- Sexual dysfunction;
- Discolouration of or bronzing of skin; and
- Mood swings and irritability.

The early biochemical signs of haemochromatosis tend to be:

- Increased serum ferritin and transferrin saturation; and
- Abnormal liver function tests (LFT's).

Symptoms are often attributed to other causes, leading to a delay in diagnosis. When undetected and untreated, iron overload can result in severe organ damage and premature death.

Diagnosis

A simple blood test to check your iron status can confirm or rule out iron overload. If both the serum ferritin (SF) and the transferrin saturation (TS) levels are raised, then a genetic test is carried out to confirm the diagnosis. SF reflects body iron stores; an SF of >200ug/L in pre-menopausal women, and >300ug/L in men and post-menopausal women, suggests that the patient may be iron overloaded. This should prompt a test for fasting TS.

Transferrin saturation

Fasting transferrin saturation of >45% is strongly suggestive of haemochromatosis and should prompt a test for the HFE gene.

Genetic testing

Genetic testing should be performed for C282Y and H63D mutations in the HFE gene. Everyone has two HFE genes, one inherited from each parent. It is a fault that occurs in the gene which causes the mutation. Haemochromatosis is inherited in an autosomal manner and to have haemochromatosis you must inherit a faulty gene from each of your parents. If you have only one faulty gene, you are said to be a carrier.

Treatment

When diagnosed early, haemochromatosis responds well to the recommended treatment. This is known as venesection or phlebotomy, and involves the removal of a unit of blood. Each 500ml of blood withdrawn contains 250mg iron. The aim is to bring the SF level down to 50 – 100 ug/L.

The frequency of venesection depends on initial SF. It may take many months to unload iron. Initially, the treatment can mean weekly or biweekly 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 the patient's life.

Diabetes and haemochromatosis

Dr Mensud Hatunic, Consultant Endocrinologist at the Mater Hospital, said diabetes develops in 30% of patients with haemochromatosis. A significant proportion of patients with newly diagnosed haemochromatosis have abnormal glucose levels. It is recommended that all patients with haemochromatosis are screened for diabetes. Appropriate haemochromatosis treatment with venesection will improve glucose levels, prevent progression to diabetes and improve diabetes control with already established diabetes in haemochromatosis.

Nutrition and lifestyle

- Patients should eat a well-balanced diet and drink plenty of water;
- Iron supplements and foods with iron fortification should be avoided;
- Iron-containing products are contraindicated in individuals with haemochromatosis and iron-free vitamins should be prescribed;
- Vitamin C should not be taken with meals; and
- Alcohol consumption should be kept to a minimum, as moderate to heavy alcohol consumption increases the risk of cirrhosis by 10%.

Pharmacist vigilance

Patients should not be prescribed iron supplements unless they are shown to be deficient in iron. In the past, people suffering from chronic fatigue, lack of vitality and generalised aches and pains, especially in the joints, were sometimes prescribed iron supplements, whereas in reality, they were suffering from extreme tiredness because of an excess of iron.

Brochures on haemochromatosis are available for pharmacies on request. Please phone 01 873 5911 or email info@haemochromatosis-ir.com for more information. The Irish Haemochromatosis Association website is at www.haemochromatosis-ir.com.

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