Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

WELCOME TO THE SUMMER NEWSLETTER 2017

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DROGHEDA MEETING MAY 11TH

The Drogheda meeting at the Boyne Valley Hotel, which was organised by Nurses Anna Capplis and Majella Jobling was attended by 75 people. Dr John Keohane, Dr Sengupta and Nurse Anna Capplis gave an excellent overview of Haemochromatosis. Ms Tina Selby represented the IBTS. Anna mentioned that the Haemochromatosis team at the Louth County Hospital now look after a record 1,200 patients. The Q and A session that followed the presentations gave people a great opportunity to seek expert advice on their individual concerns. Anna gave an update on the recent meeting in Birmingham.

LAUNCH OF AWARENESS CAMPAIGN AT FARMLEIGH MAY 24TH

Minister Simon Harris launched the campaign and promised his support to the IHA. World Rowing Champion, Niall O' Toole and Dublin footballer Jack McCaffrey also took part in the photoshoot.

Jack is on the ball for the IHA

Rowing in to raise awareness are Brian Keegan, Jack McCaffrey, Margaret Mullett, Niall O' Toole and Jim Jackman.

BIRMINGHAM CONFERENCE - MARCH 31ST

Prof Pierre Brissot from Rennes University is a leading authority on Haemochromatosis. At the recent Haemochromatosis conference in Birmingham, Prof Brissot spoke on the significance of measuring Transferrin Saturation as well as Ferritin. He specified that both tests are very important but that the measure of transferrin saturation is, in his view, probably more important than the measure of serum ferritin. **He explained that Ferritin is the iron STORAGE protein in the body, and transferrin is the iron TRANSPORT protein – which he illustrated by using the analogy of a loaded ship moving materials from place to place. His premise was that once the transport system for iron was overloaded, excess "unbound" iron was the result, leading to toxicity and overload.** It became clear therefore that transferrin saturation is a crucial test in the diagnosis of HH – even more so than serum ferritin which can be elevated by other factors such as metabolic syndrome, alcohol consumption and infections/inflammations.
Only once iron stores are reduced (and serum ferritin levels very low) does the transferrin saturation level drop and again become a meaningful measure. In maintenance (i.e. after de-ironing and with venesection frequent enough to prevent iron stores building up again) transferrin saturation again becomes a crucial measure, with a level of 75% or more indicating that the system is beginning to struggle and that iron loading may be recommencing. For this reason, **Prof Brissot is of the opinion that serum ferritin should be maintained at under 50 microgrammes per litre and transferrin saturation at under 50 percentage points; he referred to this as the 50/50 rule.**
Prof John Porter from UCL London also spoke about the role of TS testing. He agreed with Prof Brissot that during de-ironing it was not essential to be measuring TS because it is known that the iron transport protein remains overloaded until storage iron reaches normal levels –indicated by the measurement of serum ferritin.
Dr Edward Fitzsimons, Consultant Haematologist at NHS Greater Glasgow and Clyde also spoke at the meeting. He too made it clear that monitoring transferrin saturation is crucial in maintenance. There was debate at the meeting about the levels that should be targeted. Dr Fitzsimons explained that the new guidelines from the British Society for Haematology are currently in preparation and would include firm recommendations as to the use of all the tests.

JARLATH'S STORY: WHY EARLY DIAGNOSIS IS CRUCIAL

Jarlath has been a member of the IHA for several years and has been an incredible support to the organisation. He has manned the IHA stand at the Ploughing Championship every year as well as manning the stand in Galway, on the Awareness Day. When you read his story you will realise why he is so anxious to raise awareness of Haemochromatosis and the importance of early diagnosis.

"When I was about 40, I began to feel tired, lethargic and had low libido. I went to the doctor but as the doctor didn't know the cause of my problems I stopped attending the GP.
As the years went by, things got worse. I was getting headaches, loss of concentration and poor memory. This made it very difficult to do my job and it was very stressful. **Every night I went to bed exhausted and woke up just as tired after 8 hours sleep.**
I decided I had to do something about this. I went to the GP and insisted that he did a 'Wellman" full medical examination. One of the blood tests was Serum Ferritin. My level was 2580ng where it should have been under 300ng. The Transferrin Saturation was 98% instead of 45-50%. A genetic test showed that I was homozygous for the C282Y mutation. This confirmed a diagnosis of Haemochromatosis. I was referred to a Gastroenterologist who commenced weekly phlebotomies for nine months until my level was normal. This was a slow process as it was often difficult to get blood from my veins, the smallest needle had to be used and the phlebotomy would have to be stopped as my blood pressure frequently dropped to around 90/65 mmHg. A liver biopsy showed that I had cirrhosis. This was very worrying as I rarely drank alcohol. I felt very irritable. In the meantime I saw a psychiatrist for 2 years who put me on medication for anxiety and depression. During this time my heart was going out of rhythm very often. It would get so bad it would bring me to my knees. I saw a cardiologist, and after numerous outpatients appointments and nine admissions, two of which were emergency admissions to ICU, I had 2 heart ablations for Atrial Fibrillation and a pacemaker fitted.
The iron had caused a lot of damage to my heart. I was dizzy all the time but as I was on a lot of medication, I thought it was the cause. My joints were very painful. A few sessions with a physiotherapist gave some relief. I could no longer work so I was put on disability benefit.

I was 57 when I was diagnosed in 2007. I am now 68 and my life has greatly improved. I no longer have headaches or dizziness. My heart is doing well and my blood pressure has come back up to normal. I have regular scans to monitor my liver. My last phlebotomy was in 2011. I am still tired and my joints are still painful but the pains in my stomach are greatly reduced.

My wife is a carrier for HH. We have 3 children, one son doesn't have HH, our daughter is a carrier, our other son was diagnosed when he was 23. He has twice yearly phlebotomy but because he was diagnosed early he has none of the symptoms I had. This is why I strongly believe that early diagnosis is so important".

DR BARRY KELLEHER'S PRESENTATION

Dr Barry Kelleher is a Consultant Gastroenterologist and Hepatologist at the Mater Hospital, Dublin. At the AGM, Dr Kelleher gave a very clear overview of the diagnosis and treatment of Haemochromatosis and answered many of the questions that are commonly asked. Dr Kelleher outlined the historical background of Haemochromatosis from 1865 when it was known as bronze diabetes, to the discovery of the HFE gene in 1996, up to the present day and the latest developments.

Iron deficiency affects 500 million people worldwide but it is very rare in the Western world. Increasing affluence, longevity and dietary fortification lead to a greater risk of iron overload in those who are genetically predisposed.
Not everyone with the genes for Haemochromatosis will develop symptoms and the penetrance is said to be variable. Penetrance refers to the proportion of people with a particular genetic change who exhibit signs and symptoms of a genetic disorder.

The answer may lie with HEPCIDIN. This is a hormone secreted by the liver and first found in the urine of mice.
Hepcidin has now been shown to play a critical role in the regulation of iron absorption from the diet. It has also been shown that patients with all forms of HH have low or undetectable levels of this molecule, leading to inappropriately increased absorption of iron from the diet relative to the level of iron stores already in the body.

In the future, HH patients may be treated with Hepcidin injections in order to control the amount of iron taken up from the diet, similar to the manner in which insulin injections control the amount of sugar in the body.

DIET

- A Normal diet
- Avoid excess alcohol
- Avoid iron supplements
- Avoid Vitamin C

TESTING

- All siblings should be genetically tested
- All offspring should be genetically tested at age 18
- Parent, genetically tested but if older just test for ferritin
- All other relatives should be informed

LIVER DAMAGE

Not everyone is likely to have liver damage if ferritin <1000 and the Liver Function Tests are normal.
If ferritin is >1000 a liver biopsy is carried out. This involves removing a small slender core of tissue with a biopsy needle. The results are processed in a week.
A Fibroscan is a non-invasive alternative to a Biopsy. In this case the velocity of shear waves through the liver is measured.

Most people will not have liver damage but if they do, it is good to know about it. There is a change in management going forward. Patients should be watched for liver failure. Scope tests should be carried out to check for varices. To screen for Hepatocellular carcinoma (HCC) there should be an ultrasound every six months.

The protocol in relation to the removal of iron is a little controversial, however Dr Kelleher recommends the following;

- When to start - Ferritin >200-500
- When to finish - Ferritin of 100
- Maintenance - 2-3 a year or as needed

Each unit of blood taken removes 250 mg iron.

A WORD OF WARNING

High iron can also be caused by alcohol and by fat. Obesity trends and the high alcohol consumption per capita in Ireland are serious causes of concern. According to a recent report, Irish men have the highest body mass index in Europe and Ireland is set to become the most obese country in Europe, with the UK, within a decade.

TAKE HOME MESSAGE

- Haemochromatosis is relatively common in Ireland
- Penetrance is variable
- Treatment is relatively straight forward
- New molecular developments will help
- Important to enjoy life

MINI MARATHON MONDAY JUNE 5th

Heartiest Congratulations to the amazing Murray family. Fourteen of the extended family did the marathon in memory of their beloved brother Des who died last year as a result of complications caused by Haemochromatosis.

HAEMOCHROMATOSIS AWARENESS DAY THURSDAY JUNE 1ST

Sincerest thanks to all the people who helped man the stands in 32 venues throughout the country. It would not have been possible to organise the event without the support of members of the association and friends. The general consensus was that there is now a much greater awareness of Haemochromatosis.

Beth O'Connor and Josephine Hicks in Dundrum

Paddy Early & Milo Walsh in Waterford

Marie and Angela Broderick with Margaret Murphy in Frascati Shopping Centre

ANNUAL GENERAL MEETING SATURDAY MAY 27TH 2017

The AGM of the Irish Haemochromatosis Association which took place in the IBTS was attended by 62 people. Margaret Mullett gave the chairperson's report and the treasurer's report was given by Brendan Keenan. Due to work commitments, Kathy O' Dwyer stepped down as a board member. The other board members agreed to stay on. Maurice Manning was proposed for re-election by Niall Rabbitt and seconded by Brendan Keenan. Ann Mc Grath was proposed for re-election by Fran Mullaney and seconded by Paddy Early.
The board expressed their appreciation of the work Kathy had done over the years, especially with regard to organising the three information stands in Cork on the Awareness Day.

World Rowing Champion, Niall O'Rourke, spoke of his long journey to a diagnosis.
For several years while training and rowing competitively, Niall felt totally exhausted and had severe joint pain. He hoped that when he eased off on all the physical activity and training he would be less tired. The reverse happened and he was even more fatigued. He pumped himself with iron tablets and vitamin C, all to no avail, he even felt worse.
Eventually he realised that he needed to consult his GP and in due course he was diagnosed with Haemochromatosis . He attended Nurse Liz Ellis in St James's Hospital for his venesections and over a period of months, his levels came down. Niall got back his energy and the joint pains eased. He is very appreciative of the fact that he now knows the cause of his exhaustion and has been a great help to the IHA in our campaign to raise awareness.
Mr Andy Kelly, CEO of the IBTS spoke on behalf of the IBTS. He hopes that in due course the IBTS will extend their programme for Haemochromatosis patients nationwide.
Dr Barry Kelleher's excellent presentation is covered elsewhere in the newsletter. The meeting concluded with lunch which was generously sponsored by the IBTS.

Eamon Murphy, Maureen Flannery and Wyn Murphy at the AGM 2017

Niall Rabbitt at the AGM

NATIONAL PLOUGHING CHAMPIONSHIP

The National Ploughing Championship is set to return to Screggan, Tullamore, Co Offaly on September 19th, 20th & 21st . Please let us know if you are free to help man the IHA stand.

FIBROSCAN AND VHI

The IHA contacted the VHI to enquire about cover for Fibroscans. We were told that the VHI provides benefit for these types of ultrasound scans depending on your healthcare plan. Any members of the IHA who have queries in relation to any benefits or aspects of their health cover, can contact the VHI Customer Service Team on 1890 444444 or 056-444 4444.

MODELS OF CARE DISAPPOINTING NEWS

In reply to a recent query from the IHA, regarding the Implementation of the Models of Care, the following reply was sent by the HSE.
'While the Models of Care were approved by the HSE Leadership unfortunately funding was not secured in the 2017 Estimates process to support their implementation, in particular the expansion of phlebotomy services outside the acute hospital setting. We have recently met with the new Medical Director of the IBTS and presented him with both models of care for his consideration, we also discussed the need for fixed or mobile blood clinics around the country for individuals with HH to access for phlebotomy'.