Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

WELCOME TO THE WINTER NEWSLETTER 2015

In this issue:

TRALEE PRESENTATION BY DR JAMES RYAN MRCPI

Dr James Ryan, Consultant Physician and Endocrinologist, Bon Secours Hospital, Tralee, gave an excellent presentation on Haemochromatosis followed by a very helpful question and answer session.
The meeting was attended by 65 people including nurses from both Tralee General and the Bons Secours Hospital.

History of Haemochromatosis

1865: A case of Iron overload was first described by Trousseau
1871: Iron overload was associated with diabetes
1888: Known as bronze diabetes because of the skin pigmentation
1889: Name Haemochromatosis was suggested
1935: Postulated that Haemochromatosis was hereditary
1996: HFE gene was identified

Symptoms:

- Fatigue
- Joint Pain
- Weight loss
- Abdominal pain
- Loss of libido

Dr Ryan stressed that not all iron overload is Haemochromatosis and not all patients with C282Y have clinical disease or shortened life expectancy.

Attending the Tralee Presentation were: Gerard McCarthy, Nurse Helena Kilbridge, Nurse Noirin Taylor , Frank O' Sullivan and Nurse Mary Devane.

Diagnosis of Haemochromatosis:
- Clinical suspicion
- Onset ages generally 40-50
- Biochemical testing (Ferritin and Transferrin saturation)
- Genetic confirmation

Venesection
- Venesection treatment
- Ferritin should be monitored every 2-4 donations until < 200 ng/mL and then every 1-2 donations until ferritin is <50-100 ng/mL. Haemoglobin should be < 12.5g/dL

Case History
Dr Ryan also gave guidelines on testing testosterone levels. He mentioned a case study where a 50 yr old man presented with low libido over a two year period. He was consequently found to have a low testosterone level of 7.6 nmol/L (10 – 30) and a ferritin of 1075ug/L (15-300). A genetic test confirmed that he had Haemochromatosis.

Testosterone replacement and venesection treatment commenced and was very successful.

Sincerest thanks to Dr Ryan for a great meeting.

EFAPH MEETING IN COLOGNE OCTOBER 16th – 17th 2015

The picture includes Haemochromatosis Nurses: Hazel Ruddock, Yvonne Gammell Majella Jobling, Fiona Colclough and Anna Capplis at the EFAPH meeting in Cologne

The 11th Annual General Meeting of the European Federation of Patients with Haemochromatosis (EFAPH) took place in Cologne and was followed by a meeting of the International Alliance of Haemochromatosis Associations (IAHA).

The German Haemochromatosis Association organised and hosted the excellent conference. The meeting was attended by patient representatives from 12 European countries and also delegates from Brazil and Australia. The IHA was represented by Ann McGrath and Margaret Mullett. Five Irish Nurses attended: Fiona Colclough from Beaumont, Yvonne Gammell and Hazel Ruddock from Tallaght as well as Anna Capplis and Majella Jobling from Louth County Hospital. Representatives from the various Haemochromatosis (HH) Associations gave an account of the work being carried out in their countries. Excellent presentations on various aspects of Haemochromatosis were given by leading authorities.

More rare forms of Haemochromatosis are coming into focus and the need for a network of Reference Centres or Centres of Expertise was confirmed.
Professor Pierre Brissot, France, updated the delegates with feedback from the recent international bio-iron conference in Hangzhou, China. This important scientific conference, which takes place every two years, covers much more than Haemochromatosis, so it was useful for us to have a lay-level summary of relevant developments.

The IAHA will soon be launching its own website which will include links to all of the national organisations

Arthritis
Dr. Patrick Kiely, rheumatologist from St. George's Hospital, London, said that people often experience significant joint pain for five or more years before diagnosis. He said that while it is well known that fingers and wrists can be involved, ankles are also frequently affected. It is uncommon for people to have arthritis in the ankles unless they have either had traumatic injury or have iron overload.

Dr. Barbara Butzeck said that between 50 and 70% of people with Haemochromatosis are affected, many aged 30 or 40 at onset, and they often feel that they are getting too old too soon. From severe pain and mobility problems to the inability to have nice shoes, the impact of this condition on the quality of life is pervasive and can be much under-rated.

Dr. Kiely reported that, while de-ironing does not repair arthritis, there are good options for pain management that can be helpful. He suggested that while non-steroidal anti-inflammatory medications are often sufficient, in more serious cases medications such as Amitriptyline may be appropriate. He strongly supports aids such as foot orthotics and considers that joint replacement technology is improving and can be a good solution.

Prof. Pascal Guggenbuhl discussed some promising current experimentation with injections of Anakinra into affected joints.
Dr Sephanie Finzel of University of Erlangen, Germany, gave a very technical address on the developing use of low radiation computer tomography in the treatment and understanding of arthritis in the hands and feet. **The mechanism by which Haemochromatosis leads to arthritis is not well understood. It is not simply a matter of iron loading in the joints.** Various theories have been proposed but it remains unclear whether preventing iron overload from the beginning will prevent or mitigate the development.

The last session of the three day meeting was focussed on the organisation of an international association (IAHA). Officers were elected, finance discussed, and priorities set for the next few months. The IAHA will soon be launching its own website which will include links to all of the national organisations and start to demonstrate a collaborative approach to tackling the issues surrounding the diagnosis and treatment of genetic Haemochromaosis.

Congratulations to Ben Marris from Australia who was elected President of the IAHA and to Paulo Santos from Brazil who is the newly elected Vice President. The next meeting of EFAPH is planned in Innsbruck, Austria in 2016 and information is available at **www.efaph.eu.**

TRANSFERRIN SATURATION AS THE KEY MEASURE FOR DIAGNOSIS

Iron moves out of the stomach and into the duodenum (first part portion of the small intestine) where the majority of the absorption of iron takes place. Absorbed iron is grabbed by finger like projections called villi which line the surface of the intestinal wall. The iron is then passed into the bloodstream where it is met by transferrin.

Transferrin is the main protein which transports iron around the body. When working normally, transferrin binds to iron and transports it to all tissues, vital organs and bone marrow so that normal metabolism, DNA synthesis and red blood cell production takes place. Absorbed iron that is not needed for metabolism etc. is placed within cells as ferritin. Measurement of Serum ferritin gives a measure of the stored iron in the body.

Iron is also transported from the spleen where red cells are broken down. Normally transferrin is about 25-35% saturated with iron. Transferrin molecules that are heavily saturated lose the ability to tightly bind iron. When too much iron is present for transferrin to carry, trouble can develop. The iron may go to the liver, heart and other organs restricting their ability to function properly. If the transferrin is over 40-45% saturated it is an indication that the person may be suffering from Haemochromatosis.
At the recent meeting in Cologne, Professor Pierre Brissot compared transferrin to a barge carrying goods. At a certain point there is no more room for additional cargo!.
He stressed the fact that transferrin saturation (TS) should be used as the key measure for diagnosis. Where TS is elevated Haemochromatosis is highly probable. However, once the diagnosis has been made it is important to use Serum Ferritin as the measure of overload. He mentioned that TS can remain very high until storage iron levels in the body are normalised by venesection. A multi-centered study has observed that when TS levels are above 80%, plasma NTBI (non-transferrin bound iron) and labile plasma iron (LPI) which are potentially toxic forms of iron are frequently noted. This indicates that the evaluation of TS may be of interest for treatment as well as diagnosis.

NATIONAL PLOUGHING CHAMPIONSHIP (NPC)

The National Ploughing Championship is the largest open air show in Europe and is a great way to target a huge cross section of the Irish public.

We would like to thank all our volunteers who manned the Haemochromatosis stand on Tuesday 22nd September.

Sincerest thanks also to GlaxoSmith Kline (GSK) who manned the stand on Wednesday 23rd and Thursday 24th September.

Brendan Keenan and John McGillicudy at NPC

Team West from GlaxoSmithKline who manned the stand

Nicky Mulhall and members of the GSK team

WEXFORD INFORMATION MEETING TUESDAY NOVEMBER 17th

A report of this meeting in Whites Hotel will be included in the next newsletter.

WATERFORD BRANCH OF THE IHA

Two Waterford based members, Paddy Early and Milo Walsh are very interested in setting up a Waterford Branch of the Irish Haemochromatosis Association. If anyone in the Tramore/ Waterford area is interested in getting involved please contact:
Milo Walsh at 0876239370
milowalsh@hotmail.com
Volunteers would help with the Awareness Day and distribute information and brochures in the area.

SILVER SURFER 2015

Margaret Mullett was this year's winner of the Silver Surfer Awards 2015. She was presented with her award in the Helix Theatre in DCU by Age Action CEO Eamon Timmins and broadcaster George Hook.

PHIL TELLS HER STORY

"In 2005, I felt constantly tired and wondered what was wrong with me. On one occasion, I brought the children to see a film and slept right through.They had to wake me up when everyone was leaving! I went to the doctor and explained to him how I was feeling. He sent me for blood tests and when the results came back he told me I had Haemochromatosis. I thought I was dying! The doctor explained that I was suffering from too much iron in my body. He explained to me that both of my parents had the gene and this is why I have Haemochromatosis.

The doctor told me to advise my siblings and my husband to get blood tests to see if they also had Haemochromatosis. Luckily when my husband finally had the blood test it turned out that he doesn't have the gene which meant none of my children have Haemochromatosis.

None of my siblings have Haemochromatosis either.
Arrangements were made for me to go into a day ward in the Mater hospital to have a liver biopsy to see if the iron had damaged my liver. Thankfully, it hadn't but my iron level was 2,050 when it should have been under 200. The nurse explained that to reduce my iron levels, I had to give pints of blood. I'm terrified of getting a normal blood sample taken so when I went to get my first pint taken, I was so nervous and my hands were sweating. But when the nurse came in to take my blood she couldn't have made me more relaxed, she was brilliant! I couldn't have got though the first year without her - thanks Ann Marie!

I have been attending the Mater Hospital for 10 years. At the beginning I was going every week. Now I only have to go every 6 months to get a blood test to see if my iron levels have gone up, if they have I give a pint of blood.
I feel very lucky to have been diagnosed in time and now have the energy to look after my nine grandchildren."
Thanks to Phil for sharing her interesting story.

RENEWAL OF SUBSCRIPTION

Membership renewal forms are enclosed with this newsletter. Please complete and return to our treasurer Brendan Keenan. If you have recently sent in your subscription please ignore this reminder.
Should you no longer wish to be a member, please let us know as the cost of posting and printing is very high. Let us know if you would prefer to receive the newsletter by email.
If your email address or phone number has changed, please contact Kate by emailing kategeog@gmail.com with Haemochromatosis in the subject line.
Your ongoing support through the annual subscription is greatly appreciated and it is one of the main sources of income for the Association.
Sincerest thanks to each of you who have made donations to the IHA, in addition to the annual fee. Unfortunately we cannot thank each of you individually as the organisation has a large administrative burden.