Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7 *Summer/Autumn 2009*

Welcome to the Summer/Autumn Newsletter...

In this issue:

GEORGE HOOK INTERVIEW WITH DR MAURICE MANNING

On 28th May, George Hook interviewed Dr Maurice Manning on Newstalk 104. Maurice who suffers from Hereditary Haemochromatosis (HH) has been a great spokesperson for the IHA.
In 2006, Maurice chaired the working party report on HH that was set up by Minister Harney to examine the nature and extent of Haemochromatosis in Ireland and to advise her on the actions necessary to address the problems caused by Haemochromatosis. Maurice stressed the importance of asking your GP to test for HH, if you felt tired and cranky, had joint pain or any of the other symptoms. He explained that the blood tests for ferritin and transferrin were relatively cheap and could easily be organised by the GP. Maurice mentioned that in contrast to the HSE, which was very difficult to engage with, Mary Harney had been a great support to the IHA. He highlighted the difficulties of engaging with the HSE making the analogy that contacting the HSE was as difficult as getting on to the Complaints Department in Ryanair. The HSE does not take HH seriously enough and it is clear that a high profile medical person who would champion the cause and get it out from under the radar is very much needed.
On a personal note, the IHA would like to offer its heartiest congratulations to Maurice who has recently been appointed Chancellor of the National University of Ireland (NUI).

Sandra Gunne and Mary Campbell

Monica Vaughan, Phillipa Moran & Noelle Moran

FORTHCOMING EVENTS REGIONAL MEETINGS: TO BE ARRANGED

A number of regional meetings will take place in the coming year. We would appreciate help in organising these information meetings. If you have any suggestions for venues or speakers, please contact us at 01 8735911 or email: **margaretmullett@ireland.com**

NATIONAL PLOUGHING CHAMPIONSHIP

The National Ploughing Championship 2009 will take place in Cardenton, Athy, Co. Kildare from Tuesday 23rd to Thursday 25th September. We would appreciate if any volunteers could help man the stand. If you are available, please contact: **margaretmullett@ireland.com** or leave a message on the voice mail 01 8735911

EFAPH MEETING IN PORTO ON FRIDAY 12TH JUNE 2009

The AGM of the European Federation of Patients with Haemochromatosis was organised in conjuction with the International Bioiron Meeting in Porto, Portugal in June. Dr Matthew Lawless and Dr John D Ryan from the Mater Hospital, Dublin, presented excellent papers on Haemochromatosis at the International Bioiron meeting. Frances Mullaney was elected on to the EFAPH council and will continue to be the Irish representative. More information on this meeting will be published in the next edition of this newsletter.

ARE YOU INTERESTED IN FORMING A SUPPORT GROUP IN PORTLAOISE?

If so please contact Eamonn Keane
'I am a 54 year old male who has recently been diagnosed with Hereditary Haemochronatosis (HH). I had never heard of it until it was mentioned that I may have to be tested for Haemochromatosis. My diagnosis came about through a routine blood test which showed that my ferretin level was higher that normal. There was a certain reluctance to do the genetic test as I was not suffering from any of the symptoms and because there was no family history of HH. I now know that because I have HH there had to be a family history of it. At the very least my mother and father had to be carriers and God only knows if any of their brothers or sisters had HH. I am probably the first in my family to be tested. My iron levels were not high enough to have caused any organ damage, so it was detected in time. At the moment we are waiting for the results of my wife's genetic test. Depending on her results, it will be clear if my three adult children have to be tested.
When I first heard of HH I looked it up on the internet and what I read scared me and phrases like 'chronic life-long disease' kept cropping up. However, the good news is that if HH is diagnosed and treated before organ damage has occured, then the patient will have a normal life expectancy. As soon as my diagnosis was confirmed I joined the IHA. I felt that if there was a support group out there I should be part of it. Like any organisation the wider its network and the more members it has, the more effective it is. I am very interested in establishing a local support group of the IHA in Portlaoise.
So if there are any HH sufferers or family members of HH sufferers in the Portlaoise area who would be interested in helping to set up this support group please contact me on 086-8143575 or by email: **epkeogh@eircom.net.'**

Member Notice:
Did you leave your glasses behind at the AGM? If so contact Joan at the IBTS on: 01 4322800.

LAUNCH OF THE IMPACT DOCUMENT ON HAEMOCHROMATOSIS

This event took place at the AGM of the Irish College of General Practitioners (ICGP) in Galway on May 9th and represented a great step forward in raising awareness of Haemochromatosis (HH) in Ireland. The document is the result of a collaborative effort between the ICGP and the Irish Haemochromatosis Association (IHA). Clear guidelines for diagnosis and management are given and this publication should be welcomed and read by every GP in the country. This was also one of the recommendations published in an earlier report presented to Minister Mary Harney in June 2006.

Pictured at the recent ICGP AGM at the launch of the Quality in Practice Committee Impact Document on the "Diagnosis and Management of Haemochromatosis in General Practice" were Dr Mark Walsh, Dr Margaret O'Riordan, Dr Ann Nicholson GP and Margaret Mullett, IHA

FLORA MINI MARATHON-MONDAY JUNE 1ST 2009

Madeline Costello and Kay De Loughry

Once again the sun shone for the women's mini-marathon. Several members of the IHA and their friends participated and others who were not able to take part contributed generously.
Sincerest thanks to all those who helped in any way.

UPDATE ON VENESECTION CHARGES
FROM MINISTER MARY HARNEY

In relation to the issue of charges for venesection services, which was highlighted in the previous issue, the Minister has written to the IHA as follows: 'the Department has asked The HSE to review current charging practices and variances in treatment provision in all hospital locations in order to ensure that there is a consistent policy in relation to the provision of venesection services throughout the country and that HH patients are seen as outpatients.'
The IHA hopes that the outcome of the introduction of a consistent policy will mean that charging policies will be discontinued at hospitals that currently charge for this service rather than the reverse which will see charges being introduced where previously there had been no charge for this life saving procedure.

In reply to Dail Question 176, regarding the Long Term Illness Scheme (LTI), the Minister said that there are no plans to extend the list of eligible conditions for the LTI. This is a great disappointment to members of the IHA as the IBTS is not able to offer phlebotomy to people who would otherwise have to pay for it, on the basis that it could be seen as offering a degree of financial incentive to donate blood. This would be contrary to good practice and to the requirements of European law on promoting unrenumerated donations.

REPORT FROM AGM

Dr Niall Breslin

The AGM took place on Saturday May 23rd and was attended by more than 90 members. As there were no new nominations for the Board the current directors were re-elected. Sincerest thanks to the Board members for their very hard work and commitment, which is given on a voluntary basis. The IHA would like to thank Dr Niall Breslin, Dr Thecla Ryan and Mr Arthur Lappin for their excellent presentations. Special thanks also to Dr Willie Murphy and the IBTS for generously sponsoring morning coffee and lunch. A very informative question and answer session followed the talks.

UPDATE ON STILLORGAN HAEMOCHROMATOSIS BLOOD DONATION CLINIC

Almost 400 patients with Hereditary Haemochromatosis donate their blood at the Stillorgan Friday Clinic as part of the management of their condition. The hospitals and the patients are very pleased with the service provided at the clinic.
The clinic has the capacity to collect more donations from HH patients and would welcome new referrals.
If you wish to be referred to the Stillorgan Clinic, it is best to ask at your local hospital or GP service. All the hospitals in Dublin have the official IBTS referral forms and these forms can be posted out to your GP if requested.
The D'Olier St Clinic has not worked out, so far, mainly because of Industrial Relations issues.
It is hoped to send the BloodMobile to park near the Mater Hospital on 17th July 2009. Patients attending the Liver Centre on Eccles Street will be given the option to call in to the BloodMobile Bus (a 3-bedded unit) to donate their blood on that day. The management of their condition would remain entirely with the Clinical Nurse Specialist in the Liver Centre.
If you would like more information about this please ask at your clinic or phone the IBTS at 01 4322800.

DR NIALL BRESLIN'S PRESENTATION

Dr Breslin is a Consultant Gastroenterologist at Adelaide and Meath Hospital, Tallaght. At the AGM, Dr Breslin gave a very clear overview of the Diagnosis and Treatment of Haemochromatosis.
'Hereditary Haemochromatosis (HH) is a genetically determined disorder caused by mutations of certain genes which produce proteins involved in iron metabolism. These mutations cause increased intestinal iron absorption. The physical ailments relate to iron deposition in tissues, such as the liver, pancreas, and heart. The haemochromatosis gene is called HFE. The letters HFE don't stand for anything in particular. Everyone has HFE genes, one inherited from each parent. This gene is found on the short arm of chromosome 6. Where there is a fault (mutation) in a gene, this can change the protein produced by the gene and can lead to iron overload. The most common mutations are C282Y and H63D. In C282Y the amino acid Cysteine has been replaced by Tyrosine at position 282. In H63 D Histidine has been replaced by aspartate at position 63.

Frequency in Ireland
- 1 in 80 people are C282Y homozygous i.e. have two copies of the C282Y mutation
- 1 in 25 are C282Y/H63D are compound heterozygous i.e. have one of each mutation
- 1 in 5 carry one copy of the C282Y mutation (C282Y heterozygous)

Who should be screened for haemochromatosis?
First degree relatives and patients with the following symptoms:
- Abnormal liver tests
- Unexplained early arthropathy
- Unexplained sexual dysfunction
- Cardiomyopathy
- Type II diabetes
- Patients with unexplained fatigue

Treatment
Venesection is the standard treatment for HH
- When Ferritin greater than 200 microgrammes per litre in females; greater than 300 ug/l males
- Each Venection removes 250mg of iron per 500ml
- 10g overload will take 40 venesections to clear

During venesection treatment, patients are advised to:
- Maintain hydration
- Avoid vigorous exercise for 24 hours
- Keep Hb ~11g/dl
- Aim to keep Ferritin less than 50 and TS less than 50%

Dietary recommendations
- Avoid iron supplements
- Avoid Vitamin C supplements
- Avoid uncooked shellfish
- Mild alcohol consumption
- Limit iron rich foods e.g. red meat

Early diagnosis ensures excellent prognosis!

POSSIBLE CONNECTIONS BETWEEN COELIAC DESEASE AND HAEMOCHROMATOSIS

Dr Thelca Ryan, School of Biochemistry, Dublin City University gave an informative presentation on iron uptake and metabolism in the body system and spoke of the possible connections between Coeliac Disease and Haemochromatosis.
Coeliac Disease and C282Y Hereditary Haemochromatosis have opposite effects on iron absorption. Coeliac Disease (CD) is a malabsorption disorder of the small intestine and it also has a hereditary component.
The common symptoms are anaemia, malabsorption of minerals and infertility. People with CD have an intolerance to gluten which is found in wheat, barley and rye. Coeliac Disease and C282Y Homozygous Hemochromatosis have a similar increasing incidence across Europe. Both have gradients of frequency which increase from Turkey to North West Europe and culminate in a high frequency in Ireland. These two gradients follow the path of the Neolithic settlers who reached the edge of Europe at Ireland. The C282Y mutation is estimated to be some 2000 years old. Coeliac Disease is likely to be a much older disorder. Haemochromatosis may protect sufferers against iron deficiency whereas Coeliac Disease seems to prevent the penetrance of Haemochromatosis (by penetrance is meant the extent to which iron loading and consequent clinical problems develop in people with the genetic predisposition to the disease). Case histories show that iron overload and diagnosis of hereditary hemochromatosis often follows successful coeliac treatment. Also, British patients with Coeliac Disease showed a greater occurrence of mutation in the gene (HFE) controlling hemochromatosis, which might indicate that enhanced iron production is an adaptation to the reduced nutrient absorption associated with Coeliac Disease.

IRON UPTAKE AND METABOLISM IN THE BODY

Normally 1 to 2 mg iron per day is taken up by the cells in the intestine. The iron is absorbed via the layer of cells known as enterocytes, which line the intestinal wall. Hepcidin regulates iron uptake. When the body iron stores are full, more hepcidin is produced by the liver. Persons affected with HH absorb 3-4 mg/day instead of the normal 1-2 mg. There is an inability to down regulate intestinal iron absorption even when iron stores are high.
It has been shown that patients with all forms of HH have low or undetectable levels of hepcidin.
In the future it is hoped that HH patients may be able to be treated with hepcidin injections in order to control the amount of iron taken up by the diet.

The relative importance of Ferritin and Transferrin blood tests to HH sufferers.
Ferritin is a complex protein formed in the intestine and the amount found in serum is directly related to iron storage in the body. It is a huge molecule and each molecule can hold up to 4,500 atoms of iron. Increased ferritin levels may indicate iron loading. When there is too much ferritin, it changes into insoluble Haemosiderin. This can accumulate in various organs. Transferrin is a protein found in the blood. It transports iron from the intestine into the blood. Transferrin molecules that are heavily saturated with the metal lose the ability to tightly bind iron. If the serum ferritin is greater than 200 microgrammes per litre in females and greater than 300 ug/litre in men and if it is also the case that the transferrin saturation is more than 45% in women and more than 50% in men, then a diagnosis of HH is considered and the genetic test is suggested.

A PATIENT'S PERSPECTIVE- ARTHUR LAPPIN

Arthur Lappin is a film producer who has lived in Durrow, Co Laois for the last 10 years. His own journey towards a positive HH diagnosis has led him to take a strong stance in encouraging others to play a more significant role in the management of their own health. Nine years ago, as he approached 50, Arthur took the decision to be proactive about maintaining his own health and underwent an expensive and thorough health check. The results showed that he had an elevated level of ferritin and he took this report to his GP. The GP suggested that this was likely to be only the result of an infection and not a major cause for concern. Arthur took the advice and went away thinking that all was great!
Two years later, feeling very tired and with little energy or zest for life and a reduced libido, Arthur again considered the state of his health. He was working very hard but aches and pains that appeared to have no particular explanation were bothering him and he wondered if this was normal. He

was referred to a Rheumatologist who carried out a full scan but did not find arthritis of any significance.
At this point, Arthur mentioned to the Consultant that during his twenties, his brother - a young intern in a Dublin hospital - had conducted some blood tests on him and asked for a liver function report, the results of which were somewhat abnormal. They showed that he had mild jaundice and from that time on, Arthur, who had been a regular blood donor, was no longer eligible to donate blood. In hindsight, Arthur feels that he had unknowingly been treating his condition. The Rheumatologist then referred Arthur to Professor Suzanne Norris who confirmed that his ferritin was high and a genetic test showed that he was a compound heterozygote. He went to Nurse Liz Ellis in St James's and had 12 venesections over 12 weeks. After six or seven venesections great zest for life came back and thankfully this has remained. Quite dramatically the fatigue disappeared. Arthur informed his siblings and suggested that they should be tested. One brother was found to be a carrier and the other brother has HH.
Arthur now encourages people around his own age or younger to have themselves checked out for HH, at least for ferritin and transferrin saturation. His mother died in her early 50's and Arthur wonders whether HH could have been the cause of her heart complaint and subsequent untimely death. **The important message is that we must not always take what the doctors say as gospel and should not hesitate to seek further help and investigation if we feel that is the correct option.**

AWARENESS CAMPAIGN FOR PHARMACISTS

The IHA is encouraging pharmacists to work with the Association in raising awareness of HH and has sent brochures and an information letter to all pharmacies around the country. The most common complaints of people suffering with HH are chronic fatigue, joint pain, diabetes, irregular heart beat, enlarged liver and loss of sex drive. All of these symptoms are encountered daily in community pharmacies and many are remedied by taking a suitable tonic.

In the past people suffering from chronic fatigue may have been prescribed iron supplements, whereas in reality they were suffering from extreme tiredness because of an excess of iron. Iron containing products are contra-indicated in individuals with haemochromatosis and only iron free vitamins should be prescribed. During the coming months, the IHA intends to distribute brochures to libraries throughout the country.

THE MEDIA

The IHA is continually seeking new approaches to move the Association forward and welcomes any suggestions from members that will support it in achieving greater awareness of HH. Thanks to the help and suggestions of some members Articles on HH have recently been published in the *Sunday Mirror, the Health* and *Living Supplement of the Independent*, the *Galway Advertiser* the *Southern Star* and the *Medical News*.