ANNUAL GENERAL MEETING SATURDAY MAY 28TH

The meeting which took place at the Irish Blood Transfusion Service (IBTS) in St James's Hospital was attended by over 80 people. The IHA would like to thank the IBTS and Dr Murphy for once again making the centre available to us and for generously sponsoring morning coffee and lunch. We would also like to thank Professor Suzanne Norris, Dr Willie Murphy and Dr Anthony Ryan for their excellent presentations. Mr Frank O'Meara spoke on his concerns regarding the costs claimed and paid by VHI for phlebotomy procedures. A very informative question and answer session followed the talks.

DIRECTORS

Frances Mullaney (Fran), who joined the Board in 2003, resigned this year. Frank O' Meara was elected on to the Board in her place.

Fran's contribution to the Association has been invaluable and her youth, enthusiasm and ability have been a great help in progressing the Association to where it is today. Fran was elected on to the EFAPH Council in 2009. She has represented the IHA at meetings in Rennes, London, Barcelona, St Gallen and Porto. Her fluent knowledge of French and German has been a great help in establishing links and friendships with our European Colleagues. Fran will be missed by the board of both the IHA and EFAPH. She promises to keep in touch and returned to Dublin on June 6th to run the marathon for the IHA.

Fran is now a Director with Fleishman–Hillard UK and is based in London. We wish her every success and hope that she will be back in Ireland and on the Board before too long.

DR ANTHONY RYAN - A CHANCE DIAGNOSIS

Dr Anthony Ryan spoke from the patient perspective at the AGM. To help raise awareness of HH, Anthony and Theresa O' Connor, were at a later stage, interviewed by City Channel.

'I found out I had the Haemochromatosis (HH) genotype through my job as a scientist. Some time ago I was asked by a colleague to give a blood sample for a study of Haemochromatosis. My blood was supposed to be for comparison purposes only, to be compared with the blood of people who had the condition. But, as it turned out, I had the genotype.

Because of my job, I had heard of HH before, and I wasn't overly surprised: it's very common in Ireland. I had no symptoms at all, apart from tiredness now and then, and I still put that down to normal living: having kids, commuting to work and the occasional late night. But I knew that even in the absence of symptoms, there was still a chance that I could have iron overload, without knowing it. And of course, there is always the risk that symptoms could appear later in life.

So when I found out I immediately had my ferritin measured to determine blood iron levels, and I had this repeated at regular intervals thereafter. My first few measurements were within the normal range but then it went up slightly – so I was advised to start phlebotomy – regular bleeding to keep iron levels down. As it happened my ferritin had gone down again of its own accord before I even started treatment – but in the meantime I was lucky to be able to talk with some colleagues who explained to me that, with my genotype, the normal practise is to keep ferritin/iron levels low for the sake of safety. It's certainly well worth the effort, if it stops symptoms from appearing.

The treatment cannot at all be described as difficult. I've never had to wait more than a few minutes and the atmosphere is pleasant and relaxed. In addition, I'm lucky that I work close to the clinic and the treatment fits in well with a relaxing coffee break, with free biscuits! And the staff at the clinic do a great job to keep the atmosphere cheerful.

The way I found out is, I suppose, a bit like finding myself on the other side of the microscope. But I'm glad I did. The more we learn about the human genome, the more we're beginning to understand that all of us carry genetic variations that might make us get a disease some day. With haemochromatosis, we have the good fortune to know exactly how to identify the right genetic variation, there is an effective treatment and we can alert our relatives and maybe enable them to avoid iron overload completely.

It's little more than a minor inconvenience, just a few trips to the clinic per year, if it's caught in time.

As far as testing is concerned, there's nothing to lose and everything to be gained! If anything, I feel very fortunate to have been identified so early.

EMAIL ADDRESS FOR MEMBERS

Sending you an email is the quickest and least expensive way of getting in touch.

If you have not received an email from us recently we probably do not have your correct email address. To help us rectify this please send Catherine Geoghegan an email with your full name and Haemochromatosis as the subject to kategeog@gmail.com

NATIONAL PLOUGHING CHAMPIONSHIP (NPC)

The NPC will take place at the same venue as last year, Cardenton, Athy, Co Kildare from Tuesday 20th to Thursday 22nd September 2011. The IHA will have a stand again this year and we would greatly appreciate your help in manning the stand. If available to help, please contact Margaret at 01 4922705 or email margaretmullett@ireland.com

BRIAN LENIHAN

Sincerest sympathy to the Lenihan family and to former Minister, Mary O' Rourke, on the untimely and very sad death on June 10th of former Minister for Finance, Brian Lenihan.

In 2004, when Brian was Junior Minister for Health, he met with IHA representatives and gave the Association a once-off government grant of €10,000.

Brian had a special interest in the IHA, his late father suffered from the condition and Mary O'Rourke is Honorary President of the IHA. May he rest in peace.

Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

Summer/Autumn 2011

Welcome to the Summer/Autumn Newsletter

In this issue:

FEATURES

HAEMOCHROMATOSIS AWARENESS DAY, THURSDAY JUNE 2ND

Model Jenny Lee Masterson, Sophie Mullett (aged 5) and Paul Harrington at the launch of the Haemochromatosis Awareness Day.

The second Haemochromatosis Awareness Day took place on June 2nd 2011 and it was a great success. There were 34 information stands throughout the country. Without the incredible help and support from members and friends this would not have been possible. Sincerest thanks to everyone who gave so willingly of their time to help man the stands. The volunteers were surprised to meet so many people who had either heard of Haemochromatosis or knew someone with the condition.

The media coverage included articles in the Health Supplements of the *Irish Examiner* May 27th, *Irish Times* May 31st and the *Daily Mail* June 7th. Thanks to Ray Weldon, Therese O'Connor and Tom Doorley for their interesting and informative interviews.

Dr Barry Kelleher and Gavin O'Donnell were featured on TV3 Ireland AM on the morning of June 2nd. Mark Cagney who conducted the interview is a great supporter of the IHA and never fails to mention the fact that he also has HH.

Drivetime interview with Paul Harrington

Professor Suzanne Norris and Paul Harrington were interviewed by Mary Wilson on Drivetime . Paul who is a well known pianist and singer, won the Eurovision in 1994.
Paul was diagnosed in 2003. Before that he was constantly tired and said that he wanted to sleep all day like a 'teenager in Summer'. He felt physically heavy and unwell as if he was carrying a lump of lead around with him.
After thorough investigation by his GP and consultant it emerged that Paul had Haemochromatosis. He had never heard the word and thought it sounded like a 'made up' name.
It reminded him of a disease Bobby Ewing was supposed to have in Dallas!

Paul started treatment immediately and for the first three months he had his blood taken every week. For the past two years he has not needed to have any venesections. He is feeling great, is eating healthily, and has started on a serious exercise regime for the first time in his life.

Prof Norris mentioned that she would like to see the ferritin test included as part of a routine blood test.
Presentation on page three.

At the Gorey Shopping Centre: Jim Browne, Chris Mc Carthy and Dave McGrath

At the HAD Mullingar stand were Grainne O Rourke and Ann Mitchell

At the Marshes Shopping Centre in Dundalk were Nurse Anna Capplis, Kathleen Quigley, Gerry Brennan and Nurse Majella Jobling

GAVIN O' DONNELL'S STORY

The IHA would like to thank Gavin for sharing his story with us and for his interview with Dr Barry Kelleher on TV3 Ireland AM on the morning of the Awareness Day.

Gavin (39) works as a Civil Engineer in a Design Consultancy. Originally from Cork, he has been living in Dublin for 16 years where he is married with two sons. Although his work is mainly office-based, Gavin likes to play tennis and golf and is a keen supporter of Munster and Irish Rugby. He shares his story with us below:

'As I approached mid-30s I was feeling generally fit and healthy, though a little lethargic, especially in the evenings where I found that I did not have much energy. I put this down to the fact that I was working hard and not exercising as much as I should. While I have been asthmatic all my life, it has never been a problem for me and I had no other known medical issues.

In May 2008 a blood sample taken as part of a routine health screening organised by my employer raised alarm bells. The liver function parameters were elevated in my sample, particularly the Bilirubin level, which was more than double the upper range limit. In addition, my iron biochemistry was recorded at 50% higher than the upper range limit. I was advised to get a full screen of bloods done through my GP. I went to Dr Emer O'Reilly in the Grafton Medical Centre and got the blood test done. It came back with a very high Ferritin level (around 1,350 where the normal level is less than 300), and Dr O'Reilly immediately organised an appointment to see Dr Barry Kelleher at the Mater Hospital.

Dr Kelleher organised a genetic test and when I met with him he confirmed that I had Haemochromatosis(HH) and that I would need treatment for many months in the form of weekly venesection (having blood taken) to reduce my iron levels.

Like most people, I had not even heard of the condition before I was diagnosed. So, it was all a little surprising and I was not looking forward to the idea of blood-letting every week. But Dr Kelleher was very helpful, explaining things clearly and preparing me for the treatment. I

I cannot stress the importance of a routine health screening for anyone in their mid-thirties or older.

started the treatment in early April 2009, and continued weekly from there. I underwent an ultra sound to have my liver assessed, which was shown to be larger than it should be. So Dr Kelleher then organised a biopsy of my liver to check its condition definitively. The result of my biopsy showed that my liver had suffered damage as a result of the iron loading over the years. Thankfully, Dr Kelleher was satisfied that the damage was not of major concern and would also be treated by the weekly sessions. So, after 35 sessions over the course of the next year (to March 2010) my Ferritin levels were down to less than 100. However, by November 2010 my Ferritin levels were back up over 500, so I went back to the Mater for further treatment. As I understand it, my rate of re-accumulation of iron is unusually rapid, so I will need to attend the clinic more regularly than I initially thought (probably about 4 or 5 times every 6 months or so).

Dr Kelleher runs an early morning clinic in the Mater, at 7.00am on a Thursday, which suits me very well. I am in and out, and into work without any disturbance to my schedule. Like most people I was a little apprehensive about the treatment, but this disappeared after the first session. The staff in the Mater are excellent and make everything very easy. I have found I have more energy since I started the treatment, which was an immediate positive benefit. However, the essential benefit is the reduction of my iron levels, which had grown to potentially dangerous levels. My liver was scarred and was getting worse, slowly. If this had not been picked up in the way it was, it wouldn't have taken too long for my liver and other organs to start deteriorating, causing serious health problems for me.

I cannot stress the importance of a routine health screening for anyone in their mid-thirties or older. I didn't realise the risks to health at what was a relatively young age. However, I am lucky that my employer organised the screening for me, many need to do this for themselves – I don't know how long it would have taken me to do it on my own!

Thankfully, I feel I am lucky to have had the disorder picked up and I have a clear treatment plan to tackle the issue into the future. Oh, and I have the necessary health insurance to cover the cost of the treatment, without which I would have had to reconsider how I got my treatment.'

Nurse Geraldine Lennon at the Monaghan HAD

PRESENTATION ON HEMOCHROMATOSIS - PROF SUZANNE NORRIS

Professor Suzanne Norris who is a Consultant Hepatologist at St James's Hospital in Dublin was the principal speaker at the AGM.

Professor Norris explained that Haemochromatosis is a condition of iron overload due to abnormal regulation of iron absorption in the duodenum (gut). It is due to inheritance of a mutated gene (HFE gene) which stops the body from correctly regulating iron intake. Patients with HH continue to absorb iron from the diet despite excess stores of iron. Excess iron is toxic and may cause irreversible damage to body tissues and organs in which it is stored.

Inheritance of HH

HH is the most common genetic disorder in Caucasian (white) populations, concentrated in Celts and Nordic ancestry.
The two mutations in the HFE gene found in HH patients are C282Y (90%) and H63D (5%). Carrier status (one copy of the mutation) is more common (1 in 10).
Prevalence: 1 in 200 in NW Europe and 1in 80 in Ireland have the two mutations.

Stages of development of HH

The clinical condition evolves in a series of stages beginning with:

(a) Genetic predisposition but no other abnormality:
C282Y positive, normal ferritin.
0 – 20 years of age; 0 – 5 gm Fe storage, clinically insignificant iron overload

(b) Iron overload without disease:
C282Y positive; ferritin raised years of age; 10 – 20 gm Fe storage

(c) Iron overload with organ damage: ferritin raised; cirrhosis > 40 years of age; >20 gms Fe storage
Symptoms may only develop at late stages and that is why professor Norris believes that ferritin levels should be screened as part of a routine blood test

Iron Overload and organ damage

Liver:	cirrhosis, liver cancer (HCC)
Pancreas:	diabetes (often detected before HH)
Heart:	cardiomyopathy (heart failure) arrhythmias
Joints:	Arthritis

Arthritis and HH

If HH is untreated an average of 50% of HH patients develop arthritis. The cause is largely unknown. It may be worst symptom of HH affecting quality of life. It tends to start in the small joints (fingers, hands) and progresses to larger joints (shoulders, knees, hips). Symptoms can mimic Rheumatoid Arthritis (but RHF negative). Chondrocalcinosos seen on x-rays

> Excess iron is toxic and may cause irreversible damage to body tissues and organs in which it is stored.

Is the gene defect always expressed?

- Penetrance is highly variable
- Clinically significant disease may vary from 1 – 50% of C282Y+/+ homozygotes

Should children be screened? Yes

- When they can take responsibility for the result.
- Can give informed consent.
- Can understand the implications of the test

Dietary Advice

- All things in moderation
- Be wary of shellfish
- Drink tea as tannin prevents the absorption of iron
- Avoid vitamin C in tablet form
- Alcohol consumption should be restricted

Take Home Message - Early Diagnosis is Crucial

Survival is normal in HH patients where treatment is initiated before development of organ damage. Screening is a very important strategy to detect early disease and prevent complications: significant disease can be detected, treated, and progression prevented

Recommendations

The Department of Health and Children should set out a policy endorsing a framework for management of HH.

The Health Service Executive should aim to ensure early diagnosis of HH in order to prevent the onset of a range of serious medical conditions. Funding must be prioritised to develop a HH screening programme. Information obtained from this programme will provide essential guidance to policy makers in structuring a countrywide programme for HH service development.

FLORA WOMEN'S MINI MARATHON MONDAY JUNE 6TH 2011

Leonora Mullett and Fran Mullaney. Fran returned from London for the marathon.

Mary O Rourke and Kay De loughrey at this years marathon

It was a great bonus to have had such perfect weather for this years marathon.
Thanks to all who supported the event either by participating or fund raising.