



Welcome to the Summer/Autumn Newsletter 2012

In this issue:

- Interview with Philip Maree in the Sligo Champion
- A Patient's Perspective - Michelle O'Donnell
- Annual General Meeting

FEATURES

- Saturday May 26th
- Flora Women's Mini Marathon Monday June 4th
- Presentation - Prof Suzanne Norris

- Haemochromatosis Awareness Day Thursday May 31st
- Letter to Minister Reilly
- Life as a Way of Golf: 50 Years of Golf Nut Wisdom by Ivan Morris

PHILIP MAREE INTERVIEW in the SLIGO CHAMPION by HARRY KEANEY



Being an iron man is not always a good thing

It was only when Sligo-based Garda Sergeant Philip Maree was diagnosed with Haemochromatosis and began to talk to others about it that he realised a number of other people also had the condition. And with no prior symptoms, his own diagnosis came as a surprise. Indeed, initially it was something of a shock. Sgt. Maree found out that he had Haemochromatosis through a regular check-up with his GP and he stresses the importance of having not only regular check-ups but also ensuring the appropriate blood tests are done. "I am not sure all doctors test for levels of ferritin, or iron, in the blood" he said. Once Sgt. Maree's

level was found to be high, he was referred to a consultant. "Then I had to get blood drawn until my ferritin level dropped to 50 or below". After that he was called back every six months to check his ferritin. Sgt Maree felt lucky to be diagnosed early when his ferritin level wasn't very high. "The level of your ferritin depends on how early you are diagnosed", he said going on to point out that for most people, the only symptom of haemochromatosis may be tiredness. "They could be mistaken and start taking iron supplements which only increases the iron overload". Another important factor to take into consideration ensuring early diagnosis is the cost of treatment which he describes as "huge". "It was costing my insurance €730 every time just to have a unit of blood taken in a procedure that would take at maximum 30 minutes."

Sgt Maree's message is "Get yourself checked out. It's a very simple blood test. The sooner you are diagnosed, the lower your ferritin levels will be and the shorter the length of treatment".

Sincerest thanks to Sgt Maree for all his help and for organising and manning the information stand in Sligo on the Awareness Day.

Important : Email Alert
If you have not received an email from us in the last month we probably do not have your correct email address. To help us rectify this please send Catherine Geoghegan kategeog@gmail.com an email with your full name and Haemochromatosis as the subject. Unfortunately, several emails have recently bounced back and this means that we can't contact you by email.

A PATIENT'S PERSPECTIVE MICHELLE O'DONNELL



Michelle O' Donnell from Belfast is an Occupational Therapist in St James's Hospital. She was tested for Haemochromatosis (HH), when her sister who suffered from chronic fatigue, tested positive. Michelle had never heard of HH and initially did not take it very seriously. She just thought it was excess iron. However, when her genetic test results came back she also tested positive as did her two other siblings. Both parents were then tested and were positive which meant that all six family members had two copies of the HFE gene. Michelle's mother had been suffering from chronic fatigue for 30 years, palpitations in more recent years and arthritis in her joints. Michelle's father was diagnosed with diabetes. Apart from an aching finger Michelle has had no symptoms. After nine venesections the pain is now gone. Because of what she says is a lack of awareness in Northern Ireland amongst GPs, Michelle sent them leaflets from the IHA. Michelle stressed the importance of awareness amongst the public and the medical profession. The IHA would like to thank Michelle for sharing her story with us and for her interview with Catriona Perry on RTE news on the Awareness Day. Michelle was also interviewed by Matt Cooper and by Shane Hickey for the Health Supplement of the Independent.

ANNUAL GENERAL MEETING, SATURDAY MAY 26TH

The AGM took place at the Irish Blood Transfusion Service in St. James Hospital, James's Street, Dublin 8, on Saturday May 26th 2012 and was attended by over 80 people. The attendance included visitors from Denmark, Spain and the UK. One member from the Aran islands travelled by boat, bus, train and taxi to get to the meeting! Margaret Mullett gave the Chairperson's report and the Treasurer's report was read by Brendan Keenan. The IHA would like to thank the IBTS 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, Ms Michelle O' Donnell and Dr Willie Murphy for their excellent presentations.

One of our Directors, Frank O'Meara resigned this year. Leonora Mullett was elected on to the board in his place. The other Directors have agreed to stay on as board members for another year.

Frank O'Meara
Sincerest thanks to Frank O'Meara for his great help to the Association, both before and since joining the board. We greatly appreciate the manner in which Frank negotiated with the VHI regarding the money that was being paid out for venesection to the various hospitals. Thanks also to Frank for organising and manning a stand for the Awareness Day in Cork. Frank and fellow board member Kathy O'Dwyer organised a very successful meeting in Cork last October. Frank intends to continue his negotiation with the VHI on behalf of the IHA. No doubt he will make further progress.

Interesting and Informative Comments from the AGM

A Wexford man who was diagnosed 12 mths ago commented that he had an initial ferritin level of 7000! He felt

lucky to have been referred to Prof Norris and is now very well and in the maintenance phase.

In response to a question on raised Transferrin Saturation ("T.S") Prof Norris explained that high T.S. was of no relevance when someone has already been diagnosed. However, Ferretin is important. Prof Norris does ultra sound on all her HH patients. Regarding liver scan, she is guided by symptoms.

There is now a new calibration machine for ferritin. Cut off value is 133 (100 is now 133)

Prof Norris was asked if HH has anything to do with depression. She said that there was no scientific evidence but that there is a natural depression around any chronic illness. As the iron levels come down people begin to feel better.

Ivan Morris, a Limerick member, mentioned that his son of 32, who was diagnosed at 22, now follows a macrobiotic vegetarian diet. His Ferretin has come down and he has not needed a venesection in two years. Ivan suggested a possible study to look into the benefits of a macrobiotic lifestyle. Another member thought that if you cut out all red meat, you might die of boredom!

Dr Willie Murphy IBTS explained to members that when blood is used for transfusion, it is used as a medicine to treat patients. The procedure is highly regulated and it is vital that very strict protocol is followed.

Dr Brigid Gallagher is in charge of the very successful IBTS clinic in Stillorgan. She looks after 560 HH patients who attend the clinic and have their blood taken and used for transfusion. We would hope that this model would be replicated elsewhere.

The chairperson thanked the nursing staff for the exceptional care and attention they give to the patients in the nurse-led venesection clinics, throughout the country.

FLORA WOMEN'S MINI MARATHON MONDAY JUNE 4TH

It was a great bonus to have such perfect weather for this year's marathon. Sincere thanks to all who supported the event either by participating or fundraising.

Anna Capplis, Majella Jobling and Charlotte Boland relaxing after the mini marathon.



PRESENTATION ON HEMOCHROMATOSIS

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

Prof. Norris explained that Haemochromatosis (HH) 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 those of 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 5) Prevalence: 1 in 200 in NW Europe and 1 in 80 in Ireland have the two mutations.

Stages of development of HH

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

- Genetic predisposition but no other abnormality: C282Y positive, normal ferritin. 0 – 20 years of age; 0 – 5 gm Fe storage, clinically insignificant iron overload
- Iron overload without disease: C282Y positive; ferritin raised ears of age; 10 – 20 gm Fe storage
- 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: OA, acute synovitis/ joint destruction

Organ damage and HH

Liver blood abnormalities	75%
Weakness, lethargy	74%
Skin pigmentation	70%
Diabetes	48%
Arthralgia	44%
Impotence	45%

Symptoms often present for 8 – 11 yrs before diagnosis

Arthritis and HH

- 50% of HH patients develop arthritis if untreated
- Cause unknown – maybe iron deposition in cartilage?
- Removal of blood has minimal effect on arthritis symptoms
- May be worse symptom of HH affecting quality of life
- Starts in the small joints (fingers, hands)
- Progresses to larger joints (shoulders, knees, hips)
- Can mimic Rheumatoid Arthritis (but RHF negative)
- Chondrocalcinosis seen on x-rays

General work up

- Chest X-ray, (ECHO if indicated)
- X-ray of affected joints
- Ultra Sound,
- Liver Function Tests
- Blood Sugar, Urinalysis, Oral Glucose Tolerance Test
- ECG

Is the gene defect always expressed?

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

Review of 100 HH patients attending OPD at St James's Hospital

- 73% C282Y+/+; 10% H63D+/+; 17% C282Y/H63D
- Age at presentation: 18 – 80 years
- 21% female
- Ferritin at presentation: 48 – 4675 (20% > 1000)
- 15% diabetic, 19% with comorbid liver disease
- 33% had liver biopsies: 24% cirrhotic

Screening for HH target populations

- Symptomatic patients
- Abnormal LFTs with raised iron markers
- NIDDM, particularly with hepatomegaly
- Cardiac disease, atypical arthropathy,
- Male sexual dysfunction
- Asymptomatic patients
- Close relatives of a HH patient
- Individuals with abnormal iron markers found on routine testing, or unexplained abnormal LFTs

Screening Tests

- **Transferrin:** is a protein that binds and transports iron in serum
- Transferrin saturation (%): indicates the amount of iron that is available to be moved from storage to utilisation. Normal range 10-45%
- **Serum ferritin:** is a measure of the amount of iron stored as ferritin in all the body tissues. Normal range

for men 20-250, women 10-150 mg/L

Should Diabetics be screened for Haemochromatosis

- Italy, 894 diabetics (type 2) studied
- Screened for HH: ferritin and TS
- Confirmed by liver biopsy
- HH prevalence was 6 times higher than controls
- Denmark, 716 diabetics (type 1) studied
- HFE genetic testing
- HH prevalence was 5 times higher than controls
***high ferritin in diabetes (type 2) often due to liver fat*

Haemochromatosis and Mortality

The degree of iron overload has a direct impact on life expectancy.

Major causes of death are cirrhosis (30%), liver cancer (30%), and cardiomyopathy (sudden death)

Death due to liver cancer is 10 – 119 times higher in HH patients than general population

BUT survival is normal in HH patients where treatment is initiated before development of cirrhosis.

EARLY DIAGNOSIS IS CRUCIAL

Should children be screened ?

- 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
- No vitamin C supplements
- Tea/coffee after each meal
- 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

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.

Important to beware of 'googling' as information could be based on old studies and may be out of date and alarmist.

HAEMOCHROMATOSIS AWARENESS DAY, THURSDAY MAY 31ST

The third Haemochromatosis Awareness Day took place on Thursday May 31st 2012. It was a great success. This year there were 38 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 generously of their time to help man the stands.

The media coverage for the Awareness Day included articles in the *Irish Independent* (Health & Living section), *Farmers Journal*, *Ireland's Own*, *Sligo Champion*, *Evening Echo*, *The Star*, *Irish Medical Times*, *Irish Times*, TV3, Matt Cooper Interview with Michelle O'Donnell and Margaret Mullett, RTE News, Live 95 FM, Clare FM, Dundalk FM and the Gerry Kelly show.

Special thanks to Mags Corbett who was our excellent communications officer for the campaign.



Anna Capplis and Shane Kelly in Drogheda on the Awareness Day



Saoirse and Zoe Corbett Fitzpatrick with Adrian Mullett enjoying the Awareness Day



Ann Hannan and Norma Kissane at Manor West, Shopping Centre in Tralee



Margaret Mullett and Brendan Keenan at the Healthcare Innovation Awards

LIFE AS A WAY OF GOLF: 50 YEARS OF GOLF BY IVAN MORRIS

Haemochromatosis sufferer and Limerick man Ivan Morris's, latest literary effort, *Life as a Way of Golf*, is a very entertaining commentary on the current state of the game and the golden era of Irish golf.

Ivan has operated off a handicap of 4 or less for over 50-years, making his mark as a multi-capped inter pro with Munster, a winner of the European Club's Cup, Irish Senior Cup and Barton Shield as well as nine scratch cups. However, at age 56, Ivan was 'struggling' to finish 18 holes without feeling very tired. He

was tempted to give up golf altogether but fortunately for Ivan a family member was diagnosed with Haemochromatosis and Ivan consequently found that he also HH. Since being diagnosed and treated Ivan has fully recovered his health, energy and consuming passion for golf.

Life as a Way of Golf (Book Hub Publishing, 2012) by Ivan Morris is available as an e-Book from the industry standards and in hard copy format from the publishers, at www.bookhub4u.ie (€15 + p & p.)

LETTER TO MINISTER REILLY

Margaret Mullett recently wrote to Minister Reilly on behalf of the IHA and suggested that Haemochromatosis which is a life long chronic illness should be covered by the General Medical Service and treatment should be free for all patients. She expressed her deep concern that there had been no improvement in equity of access to venesection, or resolution of the cost problems for Haemochromatosis patients. At present there is a many layered and unnecessarily expensive system for access to and payment for treatment.

Margaret also mentioned the huge disappointment for people with Haemochromatosis. Despite the stated commitment by the Minister and the HSE in the 2012 Service Plan to open three free- at-point of care walk-in clinics by 2012, the plug has been pulled and this is not going to happen.

The IHA were sure, and consequently has assured its members, that this programme was definite. The IBTS were determined to make this programme work and the 'about turn' must be a great disappointment to them.

In Limerick, in particular, it is almost impossible for patients to receive the necessary venesection treatment. The Limerick Regional is closed to referrals and patients are being advised to attend Ennis and Nenagh hospitals. Venesection is essential to prevent organ damage and even death. Surely it should be possible to work out a joint programme involving both the GP's and the IBTS. The IBTS would be assured of donors, the blood would be used for transfusion and it should be possible to work out an equitable price with both the GP's and the insurers.

As yet the Minister has not replied to this letter.

Meeting in Glencarn Hotel, Castleblaney, Co Monaghan May 30th

As part of the Haemochromatosis Awareness Day, an Information Evening on Haemochromatosis was organised by the staff of Monaghan General Hospital. The meeting took place in the Glencarn Hotel, Castleblaney, Co Monaghan on May 30th. Dr Muthalagu was the guest speaker. The evening was a great success and was attended by over 50 people