Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

WELCOME TO THE SPRING NEWSLETTER 2016

In this issue:

LIMERICK MEETING M0NDAY APRIL 4th

The meeting took place at the South Court Hotel, Raheen, and was well attended. The speakers were Dr Hilary O'Leary MRCPI, Regional Hospital and Dr Willie Murphy, Medical Director, Irish Blood Transfusion Service (IBTS).
Dr Hilary O'Leary gave a very comprehensive and interesting overview of Haemochromatosis.
The talk was followed by a helpful question and answer session. Some patients raised the issue of inequity of access to venesection. Because of lack of capacity, the venesection clinic in the Regional Hospital is closed to new patients. Consequently, some patients are required to attend Ennis and Nenagh General Hospitals for venesection.
Dr Murphy confirmed that the IBTS hopes to open a clinic for HH sufferers in June and is in talks with Barringtons Hospital. As well as taking pressure off University Hospital Limerick and the hospitals in Ennis and Nenagh, the new service will give the IBTS a whole new source of blood for transfusions.

Sincerest thanks to Dr O' Leary and Dr Murphy for generously giving up their time to help the Irish Haemochromatosis Association.

Dr Willie Murphy and Dr Hilary O 'Leary

COFFEE MORNING TUESDAY MARCH 15TH 2016

A very enjoyable coffee morning was held in Bushy Park Road, Dublin on Tuesday 15th March 2016. The event was attended by friends and neighbours and €1,800 was raised for the IHA. It proved a great way to raise both awareness and money. Please contact us at 01 8759331 if you are interested in hosting a coffee morning or if you have any suggestions for a fund-raising event.

ANNUAL GENERAL MEETING SATURDAY MAY 28th 2016

The meeting will take place at the Irish Blood Transfusion Service (IBTS) St James's Hospital, James's Street, Dublin 8.
The IHA would like to thank the IBTS and the Medical Director Dr Murphy for once again making the centre available to us for the AGM and for generously sponsoring coffee and lunch. Family and friends are welcome. Coffee will be available from 10.30 am. A brief business meeting beginning at 11am will be followed by guest speakers. Details are on the enclosed sheet.
The talks will be followed by a Q&A session. The meeting will conclude with lunch. For catering purposes, please let us know as soon as possible if you will be attending by returning the reply slip posted out with the official AGM announcement or by phoning 01 8735911.

PROFILE OF BEN MARRIS
PRESIDENT OF HAEMOCHROMATOSIS AUSTRALIA

Ben Marris is President of Haemochromatosis Australia and is the newly elected President of the International Alliance of Haemochromatosis Associations (IAHA). In 2009 Ben was awarded the Medal of the Order of Australia for service to the community through health, charitable and palliative care organizations.

Ben and his wife Jane have lived in Tasmania for over 40 years. They were what is known as 'Ten Pound Poms'. This is a colloquial term to describe British subjects who migrated to Australia under the Assisted Passage Migration Scheme established by the Government of Australia.

The following interview with Ben was originally published in the Australian newsletter.

"Tasmania has proved to be a wonderful place to bring up our three children and we now enjoy having 10 grandchildren living here. My professional background was in Social Work and I spent much of my career working with young offenders and later became Director of Corrective Services for Tasmania. Apart from my family, my passion is for wooden boats and sailing. When I turned 40 Jane said that I could have another girlfriend provided she had at least 4 tons of lead in her bottom. Saona has rather more than that. She is a 40ft ketch, built of Huon Pine in 1936.

Saona, Bens 40ft ketch

I give thanks regularly for modern medical technology. We are so lucky.

I was diagnosed with Haemochromatosis in my early 40s after some years of arthritis and chronic stomach discomfort. I had been to GPs, chiropractors and an acupuncturist. When I was referred to a rheumatologist he immediately did my iron studies and then arranged a liver biopsy. The genetic test came later. It took two and a half years of weekly venesection to get my ferritin down to normal. My stomach settled immediately but the joint damage was permanent. I have had four hip replacements and an ankle arthrodesis, all of which enables me to get on with a normal life.

Ben Marris is President of Haemochromatosis Australia

I give thanks regularly for modern medical technology. We are so lucky. These services were not available to our parents and are still not available to many people on this planet. I can get angry that I was not diagnosed earlier, but I am so grateful to that rheumatologist. Without his diagnosis where would I be now?

Since 2001 I have been involved with Haemochromatosis Australia. When I was President of Arthritis Tasmania we obtained some funding to do some haemochromatosis research and family tracing. We started the "Iron Overload Disorder Support Group of Tasmania" but later decided to wind that up and become a branch of Haemochromatosis Society Australia

I would like to see the Association having a much stronger presence all across Australia. I would like every member of the medical profession to know about haemochromatosis and to be really alert to it. Our goal is that no Australian should experience the symptoms of haemochromatosis. Nor should anyone else. It's a big task, but not impossible. Having a large and supportive membership will be essential."

The IHA is a member of the International Alliance of Haemochromatosis Associations (IAHA) and we greatly appreciate Ben's work on behalf of HH sufferers everywhere.

BEAUMONT HOSPITAL PRESENTATION FEBRUARY 15TH

On Monday February 15th Professor Frank Murray and Dr Aoibhlinn O'Toole gave an excellent overview of Haemochromatosis. The meeting was attended by over 100 people and concluded with a very informative Q&A session. The following extract covers some of the main points that were discussed.

Haemochromatosis (HH) is a clinical syndrome caused by the toxic effect of excess iron which accumulates in and damages organs. Nearly all absorption of dietary iron occurs in the duodenum.

Hepcidin is now considered to be the principal hormone involved in iron regulation. People with haemochromatosis have low hepcidin levels; this is believed to be what causes them to accumulate more iron than is healthy.

Scientists believe that by finding a way to increase hepcidin levels or activity, they will be able to restore normal iron control to haemochromatosis patients.

Inheritance

The gene that controls Haemochromatosis has been identified and is known as HFE. While several different mutations to this gene have been discovered, there are two main mutations or faults that cause HH. These are referred to as C282Y and H63D. The C282Y mutation is associated with most cases of HH. The H63D mutation seems to have less impact as do the other much rarer types. It is thought that the Haemochromatosis mutation arose in a single Celtic or Viking ancestor who inhabited north western Europe centuries ago.

Autosomal Recessive Disorder

HH is a recessive gene disorder. That means for the condition to be passed on, both mother and father must have one copy of the abnormal HFE gene. About one in seven people have one abnormal HFE gene. They are referred to as a 'carrier' because they carry a gene which may cause their children to inherit the disorder. Carriers won't develop the condition themselves.

If two carriers have children, their child has:

- a 50 per cent chance of inheriting one mutated HFE gene and becoming a carrier
- a 25 per cent chance of inheriting both mutated HFE genes and therefore being at risk of excess iron absorption and developing symptoms of haemochromatosis
- a 25 per cent chance of inheriting two normal genes

Dr Aoibhlinn O'Toole

Professor Frank Murry

Heterozygous: Having one copy of the abnormal HFE gene, for example C282Y or H63D – also known as a 'carrier'.

Homozygous: Two copies of the same gene abnormality, for example C282Y and C282Y.

Modifiers of Haemochromatosis

Sex Male: Penetrance higher among males

Alcohol: Hemochromatosis subjects who drink >60 g alcohol per day 9 times more likely to develop cirrhosis.

One particular survey showed that 28% of male C282Y homozygotes had evidence of iron-overload–related disease.

Sequence of abnormalities

Transferrin saturation is almost always increased in patients with hemochromatosis.

Later, serum ferritin levels increases, indicating the accumulation of iron in tissues.

Four stages of disease

Genetic predisposition with no other abnormality

Iron overload without symptoms

Iron overload with early symptoms

Iron overload with organ damage, particularly cirrhosis

Symptomatic organ damage

Generally begins in midlife, often with non-specific symptoms such as unexplained fatigue or joint pain.

Liver disease usually predominates.

Endocrine disorders (diabetes, hypogonadotropic hypogonadism, impotence, and hypothyroidism),

Cardiac problems (arrhythmias and heart failure), and Joint disease (destructive arthritis)

Symptoms

Better screening techniques mean that >75% of patients are asymptomatic.

Symptoms generally begin in midlife.

Non-specific: weakness, lethargy, weight loss, Hepatomegaly, Abnormal liver blood tests, Arthralgias, Amenorrhoea (absence of Menstruation), Reduced libido, Heart failure, irregular heartbeat, Diabetes.

Gender differences: Iron loading in girls and young women is more gradual than it is in older women due to menstruation and pregnancy.

Rate of symptomatic organ disease in boys and men is three times the rate in women of reproductive age.

Treatment

Weekly phlebotomy

1 unit venesected = 200-250mg of iron

Target Ferritin less than 50ng/ml

Maintenance phlebotomy every few months

In Summary

Be Reassured: Organ damage due to haemochromatosis is unusual today, due to earlier and more accurate diagnosis. Disease progresses in only a minority of untreated patients.

NB Family members should be screened.

Sincerest thanks to Prof Frank Murray, Dr Aoibhlinn O'Toole and Nurse Fiona Colclough for a great meeting.

WEXFORD MEETING TUESDAY NOVEMBER 17TH 2015

Nurses Mary Geoghegan, Imelda Mernagh and Ann Murphy (at the Wexford meeting)

The meeting which was held in Whites Hotel was a great success. Despite treacherous weather conditions, there was a record attendance of over 50 people.

Dr Colm Quigley, Consultant Physician, Wexford General Hospital gave an excellent presentation which was followed by a very helpful Q & A session. Wexford Hospital has 220 patients. The following is a summary of Dr Quigley's presentation: Haemochromatosis is the most common genetic disorder in white people of European origin If you have a Celtic heritage, ancestors from Ireland, Wales, Scotland, or Great Britain, then you are at high risk for carrying the HFE mutations for hereditary hemochromatosis (HH), also known as iron overload disease or iron storage disease.

The gene is located on a particular chromosome known as Chromosome 6. In Ireland 1 in 5 are 'carriers' which means they have one copy of the mutated HFE gene. To have Haemochromatosis you need to have inherited a gene from both of your parents.

One in 83 Irish people have two copies of the gene and are predisposed to load iron. Early identification prevents complications. If undetected and untreated, iron accumulates in the tissues and may cause:

- Joint pains
- Tiredness and Weakness
- Sexual dysfunction
- Slate grey skin discolouration
- Liver damage (cirrhosis)
- Diabetes mellitus (pancreas)
- Heart muscle failure.

The treatment is known as phlebotomy or venesection and is the removal of blood.

Investigations carried out in Wexford General Hospital include:

- Genotyping, iron studies
- Liver function
- Liver texture ultra sound
- Alpha feto protein
- Blood sugar
- Tracking blood tests

Avoid

- Iron supplements
- Foods fortified with iron
- Raw oysters (Yersinia infection)
- Moderate alcohol intake < 14 units
- Tea is good

Sincerest thanks to Dr Quigley and Nurses, Imelda, Mary and Ann for their help in organising this very successful meeting.

A MODEL OF CARE FOR HAEMOCHROMATOSIS

Aisling Phelan has been appointed as project manager by the HSE, to look into developing a Model of Care for patients with Haemochromatosis and a Model of Care for Venesection. A set of national guidelines is currently being developed.
A rough draft of the guidelines will be discussed in the Working Group meeting on the 27th April. Professor Suzanne Norris, Dr Maurice Manning and Margaret Mullett will represent the IHA at this meeting.

AWARENESS DAY THURSDAY JUNE 2ND 2016

This year the Awareness Day is in Thursday June 2nd. There will be information stands in shopping centres throughout the country. Volunteers are required to man the stands. If you can assist please phone 01 8735911 or email: margaretmmullett@gmail.com

If anyone has media contacts, please let us know as we need your help to raise awareness of the event.

Proposed venues for 2016 :The finalised list of venues will be posted on the web-site and on facebook.

At the Beaumont Meeting: Angela and Marie Broderick

RENEWAL OF SUBSCRIPTION

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.

Should you no longer wish to be a member, please let us know as the cost of posting and printing is very high. Also let us know if you would prefer to receive the newsletter by email.