Cake Sale September 2013

Thanks to Katie Thurston and her colleague Monica Harding at IDA who held a cake sale and raised €355 for the IHA.

THANKS TO THE VOLUNTEERS FROM PHARMACEUTICAL COMPANY, GLAXO SMYTH KLINE

Tom Lane, Aidan Egan, Marianne Durkan, Jacqueline Jennings and Richard Hayes used their volunteer day to man a stand for the Irish Haemochromatosis Society at the National Ploughing Championships in Co.Laois on Thursday September 26th.

A SINCERE THANKS

The IHA would like to sincerely thank Eddie Arthurs who did a sponsored cycle in aid of Haemochromatosis. The cycle was done in memory of his late father, Thomas Arthurs, and €500 was raised as a result.

CLARIFICATION RE VHI PAYMENT FOR VENESECTION

Benefit is payable for therapeutic phlebotomy by the VHI when serum ferritin is above 50ug and the patient has an established diagnosis of Haemochromatosis consistent with the diagnostic criteria specified by the VHI.

The VHI guidelines concerning eligibility for payment of GP charges provide that the patient at some point must meet the following criteria:

1. HH (genetically tested) with evidence of iron overload (ferritin >450), or
2. HH with evidence of iron overload (ferritin 350-400) on two occasions six months apart, or
3. Secondary iron overload with serum ferritin >350, and providing the ferritin at the time of venesection is 50 or above.

The claim must be supported by a lab report attached to the claim form.

It is advisable for patients to discuss their diagnosis with their GP or the Hospital Consultant carrying out their venesection in order to determine eligibility for payment by VHI.

Regarding venesection in Hospital, the VHI will only cover the cost of venesection if the ferritin at the time of venesection is 50 or above.

THE PLOUGHING CHAMPIONSHIP 24TH 25TH 26TH SEPTEMBER 2013

Kay Colm and Phillipa

This year a record crowd of 228,000 attended the Ploughing Championships at Ratheniska, Stradbally, Co Laois. Thanks to the volunteers who gave so generously of their time to man the stand. The perfect weather helped to make the event enjoyable and the volunteers were delighted to find that there was a great increase in the level of awareness of Haemochromatosis.

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.

If your email address has changed or if you would prefer to receive the Newsletter by email, contact Kate by emailing kategeog@gmail.com with Haemochromatosis in the subject line.

Your ongoing support through the annual subscription is greatly appreciated and 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.

Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

Winter 2013

Welcome to the Winter Newsletter 2013

In this issue:

WORLD AUTHORITY ON HAEMOCHROMATOSIS AT COLLEGE OF PHYSICIANS FRIDAY OCTOBER 18TH 2013

Dr. Paul Adams, Professor of Medicine and Chief of Gastroenterology at the University of Western Ontario, in London, Ontario, Canada recently lectured on Haemochromatosis to a prestigious gathering of Irish Consultants at the College of Physicians in Dublin.

Prof Adams was awarded an Honorary Fellowship from the Royal College. The degree was conferred by Prof John Crowe who is the current President of the RCPI and is another leading authority on Haemochromatosis.

Prof Adams has been working in the field of haemochromatosis and iron overload since 1977. He has published over 300 papers on the topic in various medical journals.

Haemochromatosis and Iron Overload Screening Study (HEIRS)
Nearly a decade ago, Prof Adams received a $34 Million National Institutes of Health (NIH) grant to lead the Haemochromatosis and Iron Overload Screening Study (HEIRS).To ensure that he got this sizeable grant, a 7,000 page proposal was submitted to the NIH.

The HEIRS study screened 101,168 people for iron overload.
The study investigated the prevalence of iron overload in a multi-ethnic population to determine the optimal use of diagnostic blood testing including genetic testing. His innovative research has had far reaching implications for attitudes on genetic testing for all medical diseases. The information learned through HEIRS study has been used by major policy decision makers will be utilized for policy development to standardize patient care for the diagnosis and treatment of haemochromatosis.

Dr. Paul Adams

Prof Adams has enjoyed a long career in research and has worked all over the world including Australia and France. His fascinating talk at the R.C.P.I. was both amusing and informative! Here are some random snippets from the lecture.

The Information:

- One pint of Guinness contains less than 3% of the iron that we need daily, so one would need to drink 33 pints per day to get the required level!
- One in four Irish people are carriers of the gene for HH which meant that a quarter of the people at the lecture were carriers.
- Haemochromatosis is under diagnosed in those that have it and over diagnosed in those who don't. High ferritin could be due to fatty liver, alcohol or inflammation.
- Many people who have the genes do not have the illness and are known as non-expressing HH patients.
- There is a very high prevalence of HH in Iceland which may be accounted for by the fact that the Vikings took back Irish women as wives to Scandinavia. There is also a very high incidence of HH in Brittany and many of Prof Adams' patients come from the North of Portugal.
- Lim, is a young researcher in Prof Adam's department. He has a maths degree and by analysing 100,000 results from a database he has come up with an equation that will predict the percentage possibility of a person having a positive gene test for HH. The Ferritin and TS are punched into the equation.
- A man of 40 with a Ferritin of 2,523 and a T.S. of 61 has an 88% chance of having HH. On the other hand a person with a Ferritin of 1,000 and a T.S. of 25 has a 2.3% chance of having a positive gene test for HH. An app will soon be available for this calculation.

A special thanks to Prof Adams and Prof Crowe for inviting a representative from the IHA to attend this stimulating lecture.

CORK INFORMATION MEETING

Tuesday December 3rd at 7.30 pm
Venue: Marymount Hotel, Douglas, Cork
Speakers: Dr Matthew Murphy, Consultant Gastroenterologist, South infirmary Victoria Hospital, Cork.
Dr Orla Crosbie, Consultant Hepatologist /Gastroenterologist, Cork University Hospital.
Information on this meeting will be included in the next newsletter

KILKENNY MEETING THURSDAY NOVEMBER 7TH AT PEMBROKE HOTEL

Sincerest thanks to the staff of the Hepatology Unit at St Luke's Hospital, Kilkenny for their excellent presentations at the recent meeting in Kilkenny. The speakers were Dr Zaid Heetun, Specialist Registrar, and Clinical Nurse Specialists Angela Buggy, Pauline Carroll and Noreen Maher. The presentation was followed by a very helpful question and answer session which was particularly useful to people who were newly diagnosed. Approximately 50 people attended.

Nurse Angela Buggy, Dr Zaid Heetun , Nurse Noreen Maher and Nurse Pauline Carroll. All four speakers work in the Hepatology Unit in St Lukes and they gave up their time to alert people to the importance of considering haemochromatosis as a diagnosis when people are suffering from chronic fatigue and joint pain.

Dr. Zaid Heetun explained that Hereditary Haemochromatosis is the most common cause of iron overload. The highest frequency in the world is found in Ireland and 1:83 Irish people are at risk compared to 1:300 in US.

Haemochromatosis is caused by an abnormal gene, known as the HFE gene. This results in the loss of the 'stop' signal for iron absorption. The gene is commonly called the Celtic gene and it is thought that the Vikings brought it to Ireland. It was probably a useful mutation at the time of the famine when iron was in short supply.

Clinical manifestations

- Usually the iron overload for the first 20 years of life is clinically insignificant (0-5 g iron storage). Iron overload without disease.
- For the next twenty years from 20 -40 (10-20 g iron storage). Iron overload with organ damage.
- Over 40 years the iron storage may be >20g iron storage.

Symptoms

- Chronic fatigue is the most common symptom.
- Other symptoms include Arthritis, Diabetes, Impotence, Cardiomyopathy, Skin Pigmentation, and Liver disorders.

Diagnosis

- A Ferritin test and a Fasting Transferrin Saturation (TS) test are organized by GP or the hospital.
- If both the Ferritin (>300) and TS (>45%) are raised then proceed with a genetic test.

Genetic Test

- Each person has two copies of the HFE gene, one copy inherited from their father and one copy from their mother. Each copy may be either normal or have either the C282Y or the H63D mutation.
- The most common mutation of the HFE gene is known as C282Y and the next most common mutation is the H63D.

Gene mutation does not always lead to iron overload

This is because of variations in gene penetrance and also the involvement of other genes such as Hepcidin and Ferroportin.

The Hepatology Nurses, Pauline, Angela and Noreen explained the treatment protocol at St Luke's Hospital and mentioned that part of their role was to advise screening of family, spouse/ partner where applicable and to act as patient advocate. They ensure continuity in care particularly after the initial treatment is finished and ensure compliance in follow up care.

Family Screening recommendations:

- First degree relatives of confirmed case of H.H.
- Children: When of age to give informed consent
- Spouse: If negative, no need to screen children
- Elevated Liver Blood tests
- Symptomatic individuals

Treatment

- Venesection is the cornerstone of treatment. It involves the removal of a unit of blood weekly, or every 2nd week until ferritin is less than 50. The length of treatment will depend on how high the Ferritin and Transferrin Saturation were originally.
- Before treatment, the Haemoglobin (Hb) is measured and must be over 11.5.g/dl
- Blood bag weighed ¼ grams (250mg) iron per 500mls of blood to be removed
- Once ferritin is down to below 50, give a break for around 3 months
- Restart venesection when ferritin is above 300, lower if patient is symptomatic
- Note: Increased venesection leads to increased iron absorption

Follow up care

- Once treatment is initiated, patients are not at risk of further complications
- Ferritin should be checked every 6 to 12 months
- Liver blood tests once a year or every 6 to 12 months if abnormal
- Abdominal Ultra Sound and Alpha Feta Proteins AFP in Cirrhotics

Diet

- The common view is that a low iron diet is of little benefit and is not advised
- Modest alcohol consumption
- Preferably no alcohol
- No iron tablets or vitamins containing iron
- No fortified processed foods e.g. Iron supplemented bread/cereals

NEW IBTS HEREDITARY HAEMOCHROMATOSIS CLINIC SERVICE

The Blood Transfusion Service has introduced a new free Phlebotomy Venesection Clinic for people diagnosed with Hereditary Haemochromatosis. The new Cork clinic, opening in January 2014 but taking appointments now, will complement the existing service available at the Dublin D'Olier Street Clinic.

Attendance is by appointment only and patients must have a definite diagnosis of hereditary haemochromatosis and a signed prescription from their doctor or clinic nurse. Patients do not need to become blood donors but can if they wish.

When will the service start?
The IBTS has advised us that phlebotomy service for Cork will begin on 20th January 2014. It is currently taking bookings for phlebotomy in Cork from January onwards. Patients can call 021-4807400 to make an appointment. The phlebotomy service in Dublin is already up and running, and appointments can be made at 01 474 5000.

Will the service extend to other regions?
The IBTS hopes to do its best to get it running in Limerick, Galway and the North East within a year.

The IBTS advise us that the real costs in areas that do not have a current fixed clinic have to be worked out. This will be done based on the experience of the clinics in Dublin and Cork, and ensure a robust funding model.
To avail of the Dublin and Cork clinics, patients do not need to be living there provided they are prepared to travel to the clinic.
Prescription form and more details of the service are available at **www.giveblood.ie**

Free Phlebotomy Venesection Service
Dublin - D'Olier Street Clinic
Every Mon/Wed/Thurs 10-11.20 a.m.
(except Bank Holidays)
Cork - St Finbarrs IBTS Clinic
Every Monday 12.30 - 14.30 p.m.
(except Bank Holidays)

MARGARET RANKIN

FOUNDER OF HAEMOCHROMATOSIS AUSTRALIA

Margaret Rankin is the founder of Haemochromatosis Australia. For over 20 years she was the President and the main driving force of the Association. More recently she has stepped down and has been appointed to the newly created position of Patron. Margaret has been a great support to the Irish Haemochromatosis Association.

Marg's Story
'As a specialist nurse in a hospital coronary care unit, I loved my job but I suffered from total exhaustion and at the end of a nursing shift it felt as if I'd run a marathon. I suffered from joint pains and had trouble studying and concentrating. I would throw myself on the bed and cry for no reason and felt totally frustrated. Then I would sleep for the rest of the day. I not only slept the day away, but backed up in the night and slept soundly until morning when I had to go to work. I thought if I got enough sleep I would wake up refreshed and everything would be OK. It got to the stage where I couldn't cope with shift work anymore and had to transfer to a regular daytime job.

I was afraid my continual tiredness and loss of sex-drive would harm my marriage. One doctor after another dismissed my concerns as a 'bit of arthritis' or 'another neurotic nurse' saying there was nothing they could do.

I was 46 years old and still going downhill, when a radiologist at the hospital where I worked, took an interest in my case. He took me seriously and did a series of blood tests. The results showed my iron levels were up around 1,400 ug/l instead of 150-200. The Transferrin saturation was 98 per cent. I was ordered a liver biopsy to check if the liver was still functioning alright. Fortunately the results came back negative. The doctor explained that I had haemochromatosis which is an inherited disorder.

My first reaction was relief-at last I knew what was wrong with me, it had a name. At this time (1989) the gene for haemochromatosis had not yet been discovered.

I began treatment right away. This involved having a pint of blood taken every week for a year. After I'd lose a pint I'd feel very week for 24 hours. Although my condition is now under control, the disorder has left its mark and I have had two hip replacements. In the end I had to retire early and I feel very angry that I wasn't diagnosed earlier.

I started the support group in 1991 because little known about this very common genetic disorder. Twenty years later I received the Order of Australia for my work in setting up the patient support group.'

Thanks to Margaret for allowing us to include her story and for her ongoing help to the IHA.