Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

Autumn/Winter 2011

Welcome to the Autumn/Winter Newsletter

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EUROPEAN PARLIAMENT DEBATE TUESDAY SEPT 20, 2011

The lunch-time debate on Haemochromatosis which took place on Tuesday, September 20, 2011 in Brussels was organised by Dr Francoise Courtois, General Secretary of EFAPH. The aim of the discussion was to help raise awareness of Haemochromatosis in the European Community and to highlight ways of promoting early diagnosis and treatment of Haemochromatosis.

The meeting was attended by specially invited guests including German MEP, Mr Joe Leinen, Bulgarian MEP, Mrs. Antonyia Paranova, French MEP, Mrs Corinne Lepage as well as representatives of EFAPH and several EU umbrella organisations.

Mr. Jo Leinen (Chair) welcomed the representatives of EFAPH and their guests to the European Parliament. The introduction was followed by presentations from three guest speakers: Professor Pierre Brissot, Dr. Barbara Butzeck and Margaret Mullett.

Professor Pierre Brissot, Professor of Medicine, Rennes, France, gave an excellent and simplified account of Haemochromatosis. He mentioned that there could be more than two million people in Europe predisposed to iron overload.

Dr. Barbara Butzeck MD, President of (EFAPH) is a Haemochromatosis patient who is also a medical doctor. Barbara was not aware of the disease before she was diagnosed. She mentioned the importance of the involvement of international experts and researchers in iron metabolism and the growing network that EFAPH has established with EU umbrella organizations.

Margaret Mullett

Margaret Mullett, Chairperson of the Irish Haemochromatosis Association (IHA) spoke from the patient's perspective. Margaret explained how Haemochromatosis had affected the life of her family in two different ways: Firstly, there was the death of her husband, Dr. George Mullett, who was diagnosed with Haemochromatosis (HH) only 6 weeks before his death. He died awaiting a heart transplant necessitated by iron overload, due to HH.

'Check your iron before taking supplements'.

Secondly, her five adult children, when genetically tested were all found to have HH. This was due to the fact that Margaret also carried the two mutated genes for HH. Fortunately, because of this early diagnosis and subsequent treatment, the family should have a normal life expectancy. This presentation helped convince the audience of the importance of early diagnosis. A lively debate followed the presentations.

Mrs Parvanova, Bulgarian MEP who is a medical doctor supported the suggestion to check Transferrin Saturation and Serum Ferritin at the age of 30 and again at 40. This would be much cheaper than chronic disease management. Mrs. Parvanova also announced an awareness campaign planned for 2012 on food supplements. Most supplements contain iron and Vitamin C which increases the absorption of iron in the body. She suggested that EFAPH might participate in this campaign e.g. with the Slogan: 'Check your iron before taking supplements'.

Mrs Corinne Lepage MEP, Vice President of the ENVI Committee suggested that a written parliamentary question on the importance of early diagnosis be sent to the European Commission.

AGM OF THE EUROPEAN ASSOCIATION THURSDAY SEPT 8, 2011

The AGM of EFAPH (European Federation of Associations of Patients with Haemochromatosis) took place in Brussels on Sept 8, 2011. Ann McGrath, who is a member of the IHA Board, was one of eleven members elected on to the EFAPH Board. EFAPH was founded in Rennes in 2004 with three member countries.

There are now eleven members including Ireland and the UK. New members from Belgium, Italy and Switzerland were welcomed.
Delegates from the various member countries reported on the work of their Associations.

Suggestions for future initiatives such as a European Awareness Day were discussed.The relative costs of Venesection and Genetic testing in the various member countries were compared. By comparison, the cost in Ireland was high and there was great variation throughout the country.

Congratulations from the IHA to Janet Fernau who was honored with an MBE in the Queen's Birthday list for her work in establishing and running the British Haemochromatosis Society.

MEETING AT THE QUEENSLAND INSTITUTE FOR MEDICAL RESEARCH OCT 5, 2011

While on holiday in Brisbane in October, Margaret Mullett had the great honour of being invited to a lunch-time meeting with Professor Lawrie Powell and his research team at the Queensland Institute for Medical Research. Sincerest thanks to Margaret Rankin who organised the meeting. Margaret founded the Australian Association and was until recently its President.

Professor Lawrie Powell is probably the foremost authority on Hereditary Hemochromatosis in the world. It was he who convinced the world in 1989 that Hereditary Hemochromatosis was a genetic disease and his research provided the impetus for the work that was to lead to the cloning of the HFE gene in 1996.

His interest in Haemochromatosis was stimulated when he treated four women who had Haemochromatosis. The fact that all four were non-drinkers fascinated him. The literature at that time stated that HH was due to alcoholism and affected almost exclusively men. This led to original research and a lifelong professional interest in the subject.
Professor Powell established an internationally recognized liver research group, which made significant contributions to the understanding of HH.
Professor Powell feels that it is very important to make sure all relatives of Haemochromatosis sufferers are alerted to the necessity of being tested for HH.
When asked about the implication of high Transferrin Saturation (TS), Professor Powel felt that there was no need to be unduly worried as high TS reflects the genetic defect and not iron overload

Margaret Rankin and Professor Lawrie Powell

Research on rare forms of Haemochromatosis is on-going at the Queensland Institute of Medical Research

RARE FORMS OF HEREDITARY HAEMOCHROMATOSIS

Research on rare forms of Haemochromatosis is on-going at the Queensland Institute of Medical Research.
The majority of cases of Hereditary Haemochromatosis (HH) are caused by mutations in the HFE gene located on chromosome 6. This disorder is termed type 1 or HFE-associated HH (HFE-HH).
In Ireland, around 93% of HH patients are homozygous for carry 2 copies of the C282Y mutation in HFE. It is estimated that as many as 1 in 83 people in Ireland have this genotype. This is the highest frequency in any country and fits with the concept of a Celtic origin

for the mutation.
The common H63D and rarer S65C mutations in HFE are often screened for by genetic testing. However these mutations are mild and are very rarely associated with iron overload. Other rare disease-causing mutations in HFE have been identified, but these are infrequent and only occur in isolated families.

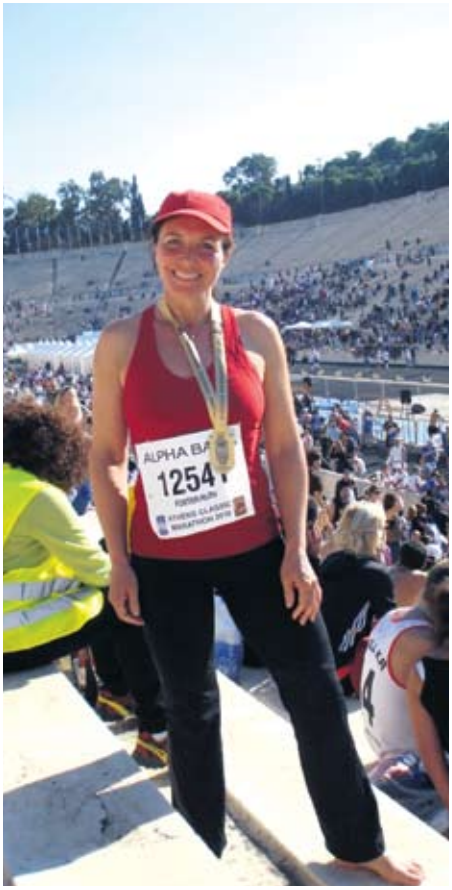
Rare forms of Haemochromatosis, also termed non-HFE Haemochromatosis, have all been identified in the last few years and are classified according to the gene responsible. There are four main types of non-HFE haemochromatosis (type 2A, type 2B, type 3 and type 4) caused by mutations in four different genes.

The study of these rare disorders and the molecules involved has greatly increased the understanding of how the body regulates iron balance.

Thanks to Daniel F Wallace PhD and V. Nathan Subramaniam PhD for giving the IHA permission to include this interesting article in the Newsletter.

Professor Powell and his research team at the Queesnland Institute

ATHENS 2500th MARATHON



Ruth Clayton

In 2010, Ruth Clayton from Brussels, Belgium, set a goal to run her first marathon and raise money for the Irish Haemochromatosis Association. The marathon was a very special one as it took place in Greece and took the same route as the fabled Greek soldier, Pheidippides. He ran from the town of Marathon (the namesake of the race) to Athens 2500 years ago to announce that the Battle of Marathon was over. Ruth successfully completed the marathon in 4 hours and 29 minutes and was supported by her sister and two friends who accompanied her to Greece. Ruth was very thankful to all her friends and family who gave moral and financial support and together raised €1500 for the IHA.

The reason Ruth chose to raise funds for the IHA was because a very dear Irish friend of hers, Sharon, was diagnosed with Haemochromatosis in 2009. Sharon who at the time lived in Belgium went through a lot of unnecessary anxiety before finally being diagnosed with Haemochromatosis. Happily, Sharon now receives regular treatment and her condition is well under control. Sincerest thanks on behalf of the IHA to Ruth and her friends for their very generous contribution.

THE PLOUGHING CHAMPIONSHIP IN ATHY SEPT 20-22nd 2011

Rose Tubridy and Frank O'Meara

Once again, the IHA would like to thank the voluntary helpers who gave so generously of their time to man the stand over the three days of the Ploughing championship. This is a very useful event for the Association in building awareness of HH and drawing public attention to the possibility of having iron overload.

CORK MEETING THURSDAY OCTOBER 20

Frank O'Meara, Dr Orla Crosbie, Maragret Mullett, Dr. William Stack and Cathy O'Dwyer

The Cork information meeting at Rochestown Park Hotel on October 20 was a great success and was attended by 70 people. Sincerest thanks to the excellent speakers, Consultant Haematologist, Dr Orla Crosbie and Consultant Gastroenterologist, Dr William Stack who gave so generously of their time and expertise.

In her presentation, Dr Crosbie explained that Ferritin is a soluble protein and is found in all human cells and in small amounts in plasma. It is able to accumulate and release iron rapidly. Plasma ferritin is closely correlated with body iron stores and hence its importance in diagnosing HH.

Dr Orla mentioned that causes of raised ferritin other than HH can be: alcohol, obesity, iron supplements, inflammation, infection, blood disorders and transfusion.

Dr Stack said that as a result of venesection there is an improvement in certain symptoms such as malaise, fatigue, skin pigmentation and liver function tests.

Other symptoms such as arthropathy are less responsive. To estimate the number of venesections required there is a formula that may be used: Initial ferritin x10-540/250. If the initial ferritin was 2000 then 78 phlebotomies would be required.

Dr Stack explained that Hepcidin is an iron- sensing protein which is made by the liver. It regulates iron absorption by the gut. People with HH are short of Hepcidin and consequently they do not receive the signal to stop absorbing iron.

At present there are many on-going studies on the function and synthesis of Hepcidin which hopefully will be of great benefit to HH patients. The meeting concluded with a very informative question and answer session.

Thanks to the two Cork Board members, Kathy O' Dwyer and Frank O'Meara for their great work in organising the meeting.

WEXFORD MEETING

Whites Hotel Wexford Tuesday November 15. The speaker is Dr. Colm Quigley, Medical Director, Wexford General Hospital. A report on this meeting will be included in the next Newsletter.

IMPORTANCE OF EARLY DIAGNOSIS

A young Irish pharmacist of 42 who was recently diagnosed with Haemochromatosis had no symptoms whatsoever. He is married with four children and decided to buy a bigger house. When he applied for a mortgage he was asked to give the name of his GP. He had never visited a doctor but to satisfy the Insurance Company he located a GP in his neighbourhood and decided that he might as well have a health check. It turned out that he had a ferritin of over 5,000 and a TS of 100%!

He is now being venesected on a frequent basis and is due to have a liver biopsy. He feels very lucky to have been diagnosed before organ damage had occurred.

His diagnosis emphasises the importance of the suggestion in Brussels to routinely test men of 30 for Ferritin and Transferrin Saturation. The test should be repeated at 40 and women should also be tested but at a slightly later stage.

RENEWAL OF SUBSCRIPTION

Membership renewal forms are enclosed with this newsletter. Please complete and return to our treasurer Brendan Keenan. Your ongoing support through the annual subscription is greatly appreciated and is one of the main sources of income for the Association. If you would prefer to set up a yearly Standing Order with your Bank, to pay your membership, please fill out the enclosed Standing Order Form and submit to you Bank.

If you no longer wish to be included on our mailing list, we would appreciate it if you would 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, please let us know by emailing kategeog@gmail.com with Haemochromatosis in the subject line. If you have recently sent in your subscription please ignore this reminder.

CORK MARATHON SEPTEMBER 25, 2011

Padraig McCarthy (Patrica), Olivia McCarthy, Eileen and Lucy Hunt, Catherine and Michelle McCarthy and Steven Clarke (Stephanie).

Catherine, Olivia, Michael and Michelle McCarthy and Eileen Hunt

Sincerest thanks to the Mc Carthy family from Goleen, Co. Cork who ran the Cork Marathon on behalf of the IHA and raised in excess of €1000!