



Welcome to the Spring Newsletter...

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AGM

The AGM will be held on Saturday, 26 May 2007, at the Irish Blood Transfusion Service (IBTS), St James's Hospital, Dublin 8. The IHA would like to thank the IBTS and the Medical Director Dr Murphy for making the facilities available to us and for generously sponsoring refreshments. Family and friends are welcome. Coffee will be available from 10.30 am. The brief business meeting will be followed by guest speakers; Dr Barry Kelleher, Consultant Gastroenterologist at the

Mater Hospital, Dublin, Professor Moira O'Brien, President of the Irish Osteoporosis Society and Mr Paul Nestor who will talk about haemochromatosis from the patients' perspective. The talks will be followed by a Question and Answer 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.

Election Of Directors

The directors are responsible for the day to day running of the organisation for the members.

Nominations for appointment to the Board of Directors should be in writing and signed by at least two members of the company. Such nominations shall be submitted to the Secretary at least ten days before the AGM.

The address of the secretary is:

Ann Campbell
7, Ashleigh Crescent, Castleknock,
Dublin 15

CITY CHANNEL TELEVISION PROGRAMME

A programme on Haemochromatosis was produced by City Channel on the 7th February, 2007. This programme was sponsored by Pfizer and has been shown in Galway, Cork and Dublin.

TV3 presenter, Lisa Cannon interviewed a number of people including Dr Willy Murphy, Brendan Meehan and Margaret Mullett. Dr Murphy, Medical Director of the IBTS, discussed the change of policy by the IBTS on blood donation. Brendan Meehan gave a graphic account of the chronic fatigue and joint pain that he suffered for five years before he was eventually diagnosed, aged 40, with haemochromatosis. He had been so tired that he fell asleep watching TV. He wasn't able to concentrate on reading and he couldn't attend a business meeting in the afternoon, as he was liable to fall asleep. Since he was diagnosed Brendan has had weekly phlebotomy and is now able to enjoy his life, his work and his young family again.

Thanks to Brendan and Dr Murphy for agreeing to participate in this programme. City Channel and Pfizer have kindly granted the IHA permission to use the DVD of the programme to help raise awareness of Haemochromatosis.

If any existing or new Support Group, or if any newly diagnosed member would like a copy of the DVD, it is available by contacting Margaret at ph: 01-4922705 or email: margaretmullett@ireland.com



TV3 presenter, Lisa Cannon

"Tired? aching? Lost interest in sex? This could be the surprising reason..."

Daily Mail ARTICLE

An article on Haemochromatosis was published in *The Daily Mail* on 27th February, 2007. This article raised a lot of interest and was mentioned on TV/AM. The following are extracts from the article in which Clara Smith shares her story:

"I would have been spared so much misery, if the disease had been spotted earlier in my life," said businesswoman Clara Smith, now 62. Clara started feeling ill in her 30s. She had bad headaches, palpitations and sore knees. She went to the doctors quite a lot. Clara was diagnosed with an under-active thyroid and given a hormone to correct it but still didn't really feel well. Her health remained poor for several years and although she had various tests, nothing serious showed up. "I think my doctor probably regarded me as a 'heart sink' patient – someone who regularly turns up in surgery with vague symptoms that have no obvious cause. Part of the problem was that I always looked well, tanned and slim, but I felt terrible." Unknown to her then, a bronzing of the skin is another sign of the disease. She was tired all the time so at one point her doctor suggested she might

have CFS (Chronic Fatigue Syndrome), a condition many doctors believe is best treated with a form of psychotherapy. On another occasion she was given an iron supplement.

The key to her chronic ill health was eventually discovered on the internet by her daughter, Susan. In Susan's own words: "I'd been looking at sites that dealt with some of my mother's symptoms, when I got an e-mail about thyroid problems that also mentioned haemochromatosis as a possible cause. I'd never heard of it but as soon as I looked it up everything fell into place. There were all my mother's symptoms; the tiredness, the tightness in the chest, the pains, everything."

Even when Susan made the connection, a nasty shock awaited them in Clara's medical records. "It turned out that my iron blood levels had actually been tested 15 years earlier," said Clara. "The results showed that far from not having

enough iron, my levels were sky high. But it was considered so unimportant that I was not even told the result. Instead the doctor just said I had 'plenty of everything' and to 'go off and enjoy life'. Actually that was the last thing I did; that doctor's decision to ignore those results set me up for 20 years of misery."

Susan had herself been suffering the same sort of symptoms as her mother for years. She was tested and the results of the genetic test confirmed that she had haemochromatosis. "It came as a shock," she said, "and I'm still coming to terms with it, but hopefully being diagnosed early will allow me to avoid more serious problems." Susan's brother, now in his 40s, had no symptoms and thought he was perfectly healthy. But as it turned out, he too had a positive test result with hugely raised iron levels. Susan and her brother were very fortunate to be diagnosed in time.

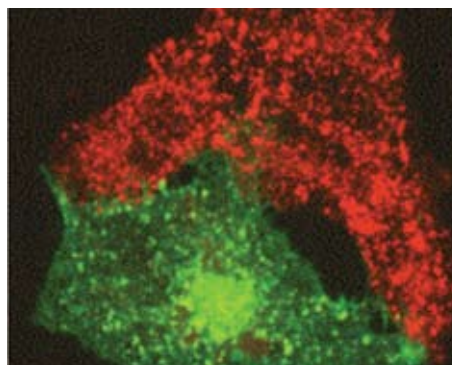
RESEARCH

Dr Mary McCaffrey, University College Cork (UCC)

Dr Mary McCaffrey is a Senior Lecturer in the Biochemistry Department, University College Cork. Her research group in UCC is studying the transport pathway by which iron and other molecules are taken into cells. For the past year they have undertaken a Health Research Board (HRB) funded study to investigate the functional properties of the HFE protein. By characterising the localisation and function of both the Wild type (normal) and mutated forms of HFE, they hope to come to a better understanding of human iron metabolism.

The most common mutation associated with Haemochromatosis patients is in the gene called HFE. This gene makes a protein which is displayed on the surface of particular cells in the body. Dr. McCaffrey's research group is examining the proteins made by the normal HFE gene and mutant proteins found in Haemochromatosis individuals. In order to do this they developed a model cell system of protein expression in liver cells in a laboratory setting.

Using a microscope and a technique called 'immunofluorescence' they can visualise the interactions of these protein with iron in the liver cells. Attached is the type of picture that can be seen. This picture shows the protein made by the normal HFE gene which is coloured in **green** and its interactions with iron which is in a complex with an important iron regulator molecule called transferrin receptor (Tfn-R) coloured in **red**. The HFE is binding the Tfn receptor which takes iron into cells, and prevents it from binding and internalising iron. On the other hand, when the C282Y



mutant of HFE is expressed no such effects on iron binding at the cell surface is seen.

Using this developed cell model system Dr. Mary McCaffrey's group will be able to visualise various mechanisms leading to iron uptake in liver cells.

Furthermore, this research team is also interested in understanding more about Heparin, a recently discovered molecule believed to be important in iron regulation. Heparin is a hormone produced by the liver which controls iron uptake from the diet. Information on this molecule will also help contribute to a better understanding of the complex questions of how the body controls its iron levels.

This important research will undoubtedly lead to a deeper knowledge and a better understanding of the issues surrounding Haemochromatosis.

The IHA would like to express thanks to Dr Mary McCaffrey, who kindly provided us with the information for this article.

PROGRESS REPORT FROM DR MURPHY, IBTS

April 2007

The plans to take blood from people with haemochromatosis at the IBTS are under way. At this stage, people who have been donors within the last two years and who now find that they have haemochromatosis can continue to donate provided they still meet all the other IBTS criteria for blood donors. In addition, people who have been told they have the genetic makeup for haemochromatosis, but don't yet have any iron overload, can also donate blood provided they meet the donation criteria.

However the main project is to set up blood donation clinics for people who are undergoing regular phlebotomy for haemochromatosis and who are eligible to donate blood for transfusion. The aim is to have the first of these clinics in place by the summer, to learn from the experiences there, and then to expand the programme to other parts of the country next year. Dr Brigid Gallagher is now employed fulltime in setting up this programme at the Stillorgan Centre. The detailed planning of the first clinic has unearthed several problems to be sorted out. Most of these are due to the levels of security that are built into the control of the blood testing system. Some are more mundane.



Dr Brigid Gallagher, IBTS

To address these issues we are asking the haemochromatosis clinics only to refer people to us who are likely to be able to donate for transfusion, and who want to do it – that is people who are not excluded for one of the many reasons that prevent anybody from donating. These include having lived in the UK or the North of Ireland for a year or more between 1980 and 1996, having had a blood transfusion in

Ireland after 1980, along with several other reasons. We will assess people referred to us, and if they are generally eligible to donate we will take a blood donation from them. Often people who can usually donate are deferred for a few weeks or months because of some temporary reason – recent travel to parts of the world where malaria and other tropical

“ people who have been told they have the genetic makeup for haemochromatosis, but don't yet have any iron overload, can also donate blood provided they meet the donation criteria. ”

diseases are endemic, childbirth, dental surgery, among others. When this happens to a donor who has haemochromatosis, we will generally take the unit of blood as required for the person's treatment on the day, but discard it. By the next time they are due to have a unit withdrawn, we will usually be able to use it for transfusion purposes.

The first clinic will be at the IBTS centre in Stillorgan in Dublin – we have a small facility there where donors attend, usually by appointment, for regular donations. The idea is that on one day of the week the Stillorgan clinic will be reserved for collections from people with haemochromatosis. If all goes well, we should be able to set up a robust system where people throughout the country who are willing and able to donate blood for transfusion will be able to do so as part of the management of their haemochromatosis – ideally at regular blood donation clinics as well as at designated clinics in the major cities.

The IBTS is looking forward to hosting the AGM at the National Blood Centre in May, when we will provide an update on progress to date and discuss the plans for the future.

William Murphy, National Medical Director

REGIONAL SUPPORT GROUPS

Midwestern Haemochromatosis Support Group (MWHSG)

This group includes Limerick City and County, North Kerry, Clare and North Tipperary. A meeting was held by the IHA in the Kilmurray Lodge Hotel, Limerick, in November 2006. As a result of this meeting the MWHSG support group was set up. The main focus of the group is to assist and support any newly diagnosed patients. The MWHSG aims to highlight the lack of facilities available to sufferers and to see what can be done to reduce the waiting list for venesection. To date the MWHSG have mailed 223 GPs within the area, to inform them of the establishment of the support group. The most recent meeting took place in Bobby Byrne's Pub in O'Connell Avenue, Limerick on Monday, 16 April 2007. For information on further meetings or if you are interested in joining the MWHSG, contact Denise McAuliffe ph: 087-8298461 or by email: denisemcauliffe@ireland.com



Cork

Ursula Kiely is helping to organise a Cork Support Group. If you are interested in getting involved, Ursula can be contacted at ph: 021-4632648 or mobile: 086 8154440 or by email: klyrsl@yahoo.com
Former Chairperson of the IHA, Noel Hynes, is happy to give advice to recently diagnosed people. Noel can be contacted at ph: 021-4361371
Former Director of the IHA, Eoghan O'Brien, can be contacted at ph: 021-4677133 or mobile: 086-0854 070

Monaghan

Brendan Gallagher, a Director and also the Treasurer of the IHA of the Monaghan Group, can be contacted at ph: 047 72802 and email: bfgallagher@eircom.net

Do you think you have the dedication and enthusiasm to start a support group in your area?

If so we'd love to hear from you! Please get in touch with us and we will include your Support Group details in the next Newsletter and on the HH Website.

EFAPH

The European Federation of Patients with Haemochromatosis has recently welcomed Iceland as the 14th member of EFAPH. Iceland was settled by Norwegian and Celtic (Scottish and Irish) immigrants during the late 9th and 10th centuries A.D. It is estimated that 50% of the inhabitants are of Celtic origin. The present population of Iceland is 307,261. It is estimated that 1 in 200 inhabitants are Homozygous for the C282Y mutation and 12-15% of the people are carriers of one copy of the mutated gene.

A delegation from (EFAPH) recently met MEP General Morillion (member of the European Parliament and Vice President of EFAPH). He expressed his willingness to help the Association and suggested that EFAPH should meet with each

of the officials responsible for health matters in the three main European political groups. Following these meetings, a meeting would be set up with the President of the Health Committee within the Parliament to achieve:

- The creation and development of associations in each of the 27 Member States of the European Union
- Funding for the production of various information documents regarding diagnosis and treatment
- Recognition of EFAPH as a federal public entity

Web site of European Federation of Associations Of Patients with Haemochromatosis:

www.european-haemochromatosis.eu



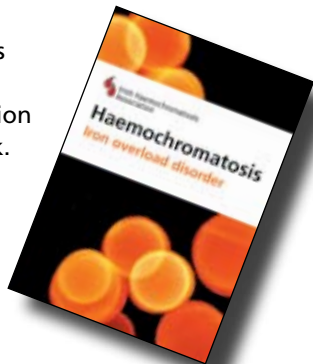
INCIDENCE OF COMPLICATIONS BRITISH SURVEY

A survey of the members of the British Haemochromatosis Society showed the following incidence of complications among its members:

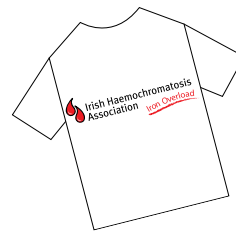
| | |
|---|-----|
| Arthritis | 60% |
| Chronic fatigue | 57% |
| Impaired sexual function | 50% |
| Chest pains, cardiomyopathy & shortness of breath | 40% |
| Skin tan | 34% |
| Abdominal pain | 27% |
| Loss of body hair | 25% |
| Liver disease | 20% |
| Menstrual problems | 15% |
| Diabetes | 13% |

IRISH HANDBOOK ON HAEMOCHROMATOSIS

The Irish Haemochromatosis Association has successfully completed an updated version of the Australian Handbook. This booklet is available to members.



WOMEN'S MINI MARATHON 2007



Spring is in the air. We hope you all have your applications in and your running shoes at the ready for this year's Women's Mini Marathon on June 4th! T-Shirts are available from Margaret at ph: 01-4922705.

We look forward to seeing you there.

RENEWAL OF MEMBERSHIP

If you have not already renewed your membership, please send your subscription to:

Brendan Gallagher (Treasurer),
Irish Haemochromatosis Association
2, Annagh Close, Latlurcan, Monaghan

YOUR CONTRIBUTIONS

We hope you have enjoyed this issue. If there are any issues you would like us to cover, or any stories you would like to share, please contact Margaret at margaretmullett@ireland.com.

Please email us if you are willing to receive the Newsletter by email.