



## Welcome to the Winter Newsletter...



### In this issue:

#### FEATURES

- Regional Meetings in Letterkenny, Waterford, Cork and Dublin
- Upcoming Meetings in Monaghan and Galway
- EFAPH
- Haemochromatosis Research In Portugal
- Mary's Story
- Ploughing Championship
- Insurance Information

## REGIONAL MEETINGS

### IN LETTERKENNY, WATERFORD, CORK AND DUBLIN

Regional meetings took place in Limerick, Letterkenny, Waterford, Cork and Dublin. The high attendance at the meetings indicated the level of interest in Haemochromatosis.

The IHA would like to thank the excellent speakers who gave so generously of their time and expertise. The question and answer sessions which followed the lectures were very helpful and greatly appreciated. The IHA would like to thank Noel Hynes for his help in organising the Cork meeting, also John Hughes of Coughlan Coleman Hughes and the other insurance companies who helped sponsor this event.

## Regional Meetings 2007 Photo Gallery

### Letterkenny

Date: 15th October  
 Venue: Mount Errigal Hotel, Letterkenny  
 Speaker: Dr. Brian Hennessy, Consultant Haematologist, Letterkenny General Hospital (below left)



Dr. Brian Hennessy



From Left: Dr Mireille Sweeney, Ms Sally Shortt and Ms Ann McGrath

### Waterford

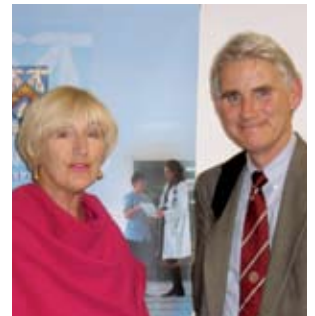
Date: 19th October  
 Venue: Waterford Regional Hospital, Waterford  
 Speaker: Dr Suzanne Norris, Consultant Hepatologist, St James Hospital Dublin



Dr Suzanne Norris

### Dublin

Date: 14th November  
 Venue: Robert Adams Lecture Theatre, Beaumont Hospital  
 Speaker: Prof Frank Murray and Dr Stephen Patchett, Consultants in Gastroenterology, Beaumont Hospital



Ms Ann McGrath and Prof Frank Murray

### Cork

Date: 25th October  
 Venue: Boole Lecture Theatre, University College Cork  
 Speaker: Dr Orla Crosbie, Consultant Gastroenterologist, Dr Mary Cahill, Consultant Haematologist, CUH



Dr Dairmuid Quinlan and Dr Mary Cahill



From left: Helen McHugh, Prof Frank Murray, Mary Judge and Dr Stephen Patchett

# EFAPH

European Federation of Patients with Haemochromatosis



The General Assembly Meeting of EFAPH was held in London in September. At present there are 14 member countries. Portugal has an association with 50 members. Dr Gracca Porto is their representative and has written an account of their association for this newsletter. The Spanish Association has 120 members.

The Netherlands Haemochromatosis Association was started in 2000 by Philip De Sterke, whose mother had HH and died in 1995. Holland has a population of 16 million. The association has 1,250 members and collaborates with the Clinic and Research Centre in Nijmegen. Philip has helped draw up evidence based guidelines for the diagnosis and treatment of HH.

The British Society was established in 1991 and has a 1,000 members.

The Norwegian Haemochromatosis Association (NHA) was founded in 1999 and now has 300 members. The NHA receives annual government economic support. The NHA hopes that in time every Norwegian citizen will be tested for HH before the age of 20 and those affected with HH given treatment. They would recommend that tablets with a

higher concentration of iron should be available only on prescription, as is the case in Austria, Holland, Belgium, France and Portugal.

## AIMS OF EFAPH

- **Raise awareness of HH in the medical community and public.**  
*Due to misinterpretation of the non-specific symptoms of Haemochromatosis there is a delay in diagnosis and an underestimation of the prevalence of the disorder.*
- **Raise funding to create EU-guidelines for diagnosis and therapy of Haemochromatosis and for Haemochromatosis Research Protocol for phlebotomy and use/disposal of veneselected blood needs to be developed. There is a great variation in the cost of venesection and of genetic testing throughout Europe.**
- **The creation and development of associations in each of the 27 member states of the European Union and the initiation of Competence Centres.**

The next General Assembly Meeting of EFAPH is in Saint Gallen (Switzerland) 20th September, 2008 .



From Left: Fran Mullaney, Philip DeSterke, Margaret Mullett and Dr. Gracca Porto



From left: Stuart McDonald, Kate Roddy, Valerie Hadfield, Maria Sanchez, Françoise Courtois

## MARY'S STORY – A CHANCE ENCOUNTER

'I got married at 21 and seven years later my son was born. During my son's childhood I suffered from extreme fatigue. While other mothers were out for a walk with their baby, I struggled with housework which never seemed to get done. I was prescribed iron supplements and also iron injections but I never felt any better. At age 40 I stopped menstruating and gained weight. Thinking that I was pregnant, I went to my GP who diagnosed an underactive thyroid. At that stage, I had a lot of pain in my right hand, knees and elbows.

The colour of my skin had always been dark, but it became increasingly darker and then turned grey and metallic. My doctor referred me to a rheumatologist who couldn't decide what type of arthritis I had, but then decided that it was osteo-arthritis. Numerous drugs were prescribed and still there was no improvement.

In 1993, I had a chance encounter with a doctor who was on a visit to Ireland. He remarked on my colour and suggested that I should be tested for Haemochromatosis. I tested positive. At this point in time, haemochromatosis was practically unheard of. I was referred to Professor John Crowe at the Mater Hospital. My ferritin was 1,100 and my liver was greatly enlarged. Under the excellent care of Prof. Crowe and his team, my HH was brought under control. I had weekly phlebotomies in Donegal, carried out by my caring GP. After 110 phlebotomies, I began to feel like a new person. Never in my life had I felt so energetic.

That euphoria lasted about five years. My iron levels hadn't risen but suddenly my energy level dropped drastically and I lost weight. I developed bowel

problems and became depressed. Back in the Mater for investigations, Prof. Crowe found that I had Coeliac Disease.

It was a great shock and another condition to contend with. So I wasn't absorbing iron or nutrients and therefore not storing any. Osteoporosis was later diagnosed and then B12 deficiency.

I am now on a strict gluten free diet and feeling better. Everything is balanced and being managed. My husband and I are now enjoying the company of our first grandchild.

Sincerest thanks to Prof Crowe, Ann-Marie and the excellent team at the Liver Clinic and GI Unit, for their great help and attention over the years.'

The IHA would like to thank Mary for sharing her interesting story with us.

*While other mothers were out for a walk with their baby, I struggled with housework which never seemed to get done.*

# HAEMOCHROMATOSIS RESEARCH IN PORTUGAL

*Background Information from Margaret Mullett*

Dr Porto is particularly interested in the penetrance of Haemochromatosis or why HH manifests itself differently among individual patients. Not all people who are homozygous for C282Y are predisposed to the same degree of iron overload. Even within families, where all members have the same HFE gene mutations, each person can display a different combination of symptoms. Some people do not load any iron whereas others will absorb high levels.

The penetrance is the proportion of individuals with a mutation, causing a particular disorder, who exhibit clinical symptoms of that disorder. Complete penetrance means the gene or genes for a trait are expressed in all the population who have the genes and clinical symptoms are present in all individuals who have the disease-causing mutation. Reduced or incomplete penetrance is when clinical symptoms are not always present in individuals who have the disease-causing mutation.

Phenotype incorporates the observable characteristics of an organism for example; hair colour, weight or the presence or absence of a disease. Genotype is the genetic composition whereas phenotype is the product of the genotype and the environment.

Much of the research carried out by Dr Porto and her team is in the hope of identifying the markers and factors that influence the penetrance of HH.

## Haemochromatosis Mutation in Portugal

*Dr Graça Porto, MD, PhD, Clinical and Research Centre, Porto, Portugal*

'Like in many European regions, Hereditary Haemochromatosis (HH) is recognised as one of the most common genetic disorders in northern Portugal. It is known that the allele\* frequencies of the C282Y mutation varies within Europe with a clear gradient pattern of decreasing frequencies from north to south. The highest frequencies are found in populations from Ireland, United Kingdom and Brittany, and also in Scandinavian populations.

This geographical distribution serves as an argument both to support the original idea proposed by Marcel Simon that the haemochromatosis mutation originated in a Celtic population which subsequently spread by population migration, and the later suggestion that the Viking conquests and migrations could also have played a significant role in the spread of the mutation.

Celts occupied the Portuguese territory during the 2nd and 1st millennium B.C. and it is recognized that their influence

allele\* alternative forms of a gene, eg; 'blue' eyes: 'brown' eyes

was more consistently maintained in the northern regions of the country, in contrast with the south where North African populations had a stronger influence. Curiously, the present distribution of the C282Y in the normal Portuguese population shows also a gradient of distribution from north to south, with allele frequencies in the north similar to those observed in more northern European populations, in contrast with the lower frequencies found in the south. Whether this heterogeneous distribution is due to the demographic history of the country or is due to other selective forces acting differently in the two parts of the country is still a matter of debate.

## The Haemochromatosis Clinical and Research Centre in Porto

Created in 1985, the Haemochromatosis Clinic at Santo António General Hospital (HGSA) in Porto has grown for the last 20 years as a reference Clinical and Research Haemochromatosis Centre (HC) in Portugal. The Haemochromatosis Clinic includes a medical school and a research institute.

## Looking for Novel Genetic Modifiers of HH

Clinical research on Haemochromatosis at our centre has focused, for the last 10 years, on the search for phenotypic and genotypic markers of disease expression and progression, in an attempt to explain the clinical heterogeneity and variable penetrance of the disease. The researchers have previously shown that a large proportion of HH patients have abnormally low numbers of CD8+ T lymphocytes (a sub-population of cells of the immune system) and that those abnormalities were correlated with a more severe expression of iron overload.

Those results prompted them to routinely use the phenotypic marker of CD8+ T lymphocyte numbers as a predictor of the clinical severity in HH. Patients with the lowest numbers of those cells are the ones predicted to have the highest iron accumulation as measured by quantitative phlebotomies (total body iron stores).

Based on that evidence they next looked for genetic markers associated with the transmission of lymphocyte numbers to see if they could also be used as markers of disease expression. They recently found that a genetic region localized around the microsatellite marker D6S105 in chromosome 6 is associated with the transmission of CD8+ T lymphocyte numbers and its inheritance may constitute a risk factor for a more severe expression of HH.

The research team are presently trying to narrow the region of interest to find a more sensitive marker so that in future we may predict with more certainty which homozygous subjects are at a higher risk of developing the more severe symptoms of iron overload.'

Dr Graça Porto is the EFAPH representative for Portugal and is responsible for the Haemochromatosis Clinical and Research Centre, Porto, Portugal).



# INFORMATION ON INSURANCE

Any members who have issues with their Health or Life Insurance should contact Frank Falvey or Paul Holohan of the Irish Insurance Federation at 01 6761914 for advice and information. They will investigate any complaints and will advise you of your rights. Brendan Gallagher at 047 72802 is a director of the IHA and he will also be very happy to help you with insurance issues. Carl Widger is Sales Director with GK Wealth Management Ltd. Carl has provided us with the following information from New Ireland Assurance and from Hibernian Insurance. For further information and help, Carl can be contacted at 061 312144 and email: [carl.widger@gki.ie](mailto:carl.widger@gki.ie)

## New Ireland Assurance

The Underwriting Manager of NIA has kindly supplied the IHA with the following information which should be helpful to members.

'Each applicant for life cover is considered on his/her own merits. In relation to Haemochromatosis a report from the applicant's GP will always be required. We will not usually require a medical examination. We may require up to date blood tests if these are not available from an applicants doctor.

In assessing such a medical history we need to establish:

1. When the condition was diagnosed
2. Age at diagnosis
3. Initial and on going treatment
4. Blood test results at diagnosis and since
5. Results of any other tests
6. Presence or absence of any complications
7. The nature and severity of these complications same

In most cases the condition will be well controlled and there will be no complications and such applicants will be acceptable at normal rates. If an applicant's condition is not controlled, they are not compliant with treatment or there are associated complications then the underwriting decision will depend on the facts of the case and could vary. Applicants may be accepted at an additional premium or their application may be postponed for a period of time.

In accordance with the Disability Act 2005 we do not ask for or take into account the results of any genetic test that may have been carried out in relation to Haemochromatosis or any other condition.

## Hibernian Insurance

The Underwriting Manager has given a general guide line but says it is impossible to give a single answer as each case will be evaluated on its own merit.

1. Diagnosis within 6 months — postpone
2. Confirmed diagnosis, no complications, normal serum ferritin levels — standard rates.
3. Evidence of non-compliance with treatment, complications or significantly elevated serum ferritin levels — postpone
4. Where there are other conditions in conjunction with Haemochromatosis, terms will be considered on an individual basis.
5. From an underwriting perspective, we usually ask for a General Medical Questionnaire and request a report from the GP.
6. Where there is no recent serum ferritin results available or where the last result was considered abnormal we may request our own ferritin test.
7. A similar approach applies for both Life and Critical Illness Contracts.

# PLOUGHING CHAMPIONSHIP

Sept 25th - 27th Tullamore

The Irish Haemochromatosis Association would like to thank Stephen Geary and Michael Campbell of Europlan Health and Safety for generously sponsoring a stand for the IHA on the three days of the Ploughing Championship in Tullamore.

The stand was a fantastic success and we were delighted to meet many members of the IHA as well as people who had friends with HH.



Clockwise from above:  
Nora Coyne and  
Philip Granger,  
Denise McAuliffe,  
Stephen Geary and  
Ann Campbell



## Upcoming Meetings

### Monaghan

Date: Thursday 7th February 7:45pm  
Venue: Glencairn Hotel, Castleblayney  
Speaker: Dr Muthalagu,  
Endocrinologist / Gastroenterologist  
Monaghan General Hospital

### Galway

Date: Wednesday 23rd January 8:00pm  
Venue: GMIT, Dublin Road, Galway  
Speaker: Dr John Lee, Gastroenterologist,  
University Hospital, Galway

## Renewal of Membership

Application form is included. IHA would like to wish all its members a happy and peaceful Christmas.

## Help Us Keep You Updated!

If you haven't provided us with your mobile number and email address already, please do so.