Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

Winter 2012

Welcome to the Winter Newsletter 2012

In this issue:

The IHA would like to wish all members a Happy Christmas and a peaceful New Year

CYCLING FROM SHANGHAI TO DUBLIN

Two young men, Brendan Kaye (on the left), 29, from Chicago and Ben Shuker, 25, from Brisbane are cycling from Shanghai to Dublin to raise awareness of Haemochromatosis. It is a 10,000km bicycle ride. They arrive in Dublin on the 7th December. They are taking six months to cross two continents and 11 countries.

Brendan's uncle has been diagnosed with HH and is awaiting a liver transplant. Brendan chose to end the trip in Dublin for three reasons: Firstly, Ireland is one of the furthest points in Europe from China, secondly, Brendan is of Irish descent and lastly, HH is particularly common in Ireland.

KERRY INFORMATION MEETING

Dr Grace Creedon and Dr Naveed Sultan at the Kerry meeting

The meeting took place on Thursday, November 15th 2012 at 7.30pm in the Carlton Hotel Tralee.
The speakers were Dr Naveed Sultan, Consultant Physician, Kerry General Hospital
Dr Grace Creedon, BSc., Ph.D., Specialist Medical Scientist, Kerry General Hospital
Dr Donal Daly, MICGP, General Practitioner, Listowel, Co Kerry

This meeting was one of the most successful ever. Over 130 people attended and the three speakers covered every aspect of the diagnosis and treatment of Haemochromatosis (HH).
Dr Naveed Sultan gave a very comprehensive overview of the disorder. His presentation included an interesting summary of the history of Haemochromatosis. Trousseau first described a case of iron overload in 1865. This disorder was originally known as Bronze Diabetes and it only became known as Haemochromatosis in 1889.
The HFE gene for Haemochromatosis (HH) was only identified in 1996. People who have HH have a mutation in that gene, the most common one's being the C282Y and the H63D mutation.
Genetic testing is now carried out in the laboratory in Kerry General by Dr Grace Creedon and her colleagues.
Grace mentioned that since this testing commenced in 2001, they have tested almost 10,000 samples. She explained the procedure used to extract and test the DNA, as well as giving a breakdown of the percentages who had the different mutations. Minors under 16 years of age are not tested.
Dr Donal Daly has diagnosed many patients with HH and they are being treated at his surgery. He gave a very interesting account of four different case histories.
The excellent presentations were followed by a lively question and answer session in which all three speakers participated.

The IHA would like to thank the speakers and also Nurse Mary Devane for her help in organising the meeting.Thanks also to Deirdre Walsh of Kerry Radio for a great interview on Wednesday 14th with Jim McCarthy, Raphael Crowley and Dr E Shannahan.

HAEMOCHROMATOSIS ARTHROPATHY

by Dr Patrick Kiely PhD, FRCP

Joint pain is very common in patients with Hereditary Haemochromatosis (HH). Of 244 respondents to the British Haemochromatosis Society members' survey (2004-08), joint pains or arthritis were the most frequent symptoms reported in as many as 72%.

A similar survey of 199 patients with HH in Germany, published in 2010, found an identical very high percentage with joint pains. Interestingly almost one in five of these patients had first developed painful joints despite venesection. The distribution of joint involvement and the clinical features of arthritis in HH patients have similarities with findings in traditional osteoarthritis (OA), but also some unique features. The frequency of joint involvement in the survey from Germany shows that in keeping with OA the knees and hips are commonly involved. However, the high frequency of OA-like involvement of the 2nd and 3rd metacarpophalangeal joints (MCP, knuckles of the index and middle fingers) and the ankles is a characteristic feature of HH, rarely seen in traditional OA.

Whilst we don't understand why these joints should be preferentially involved, the finding of arthritis that has the clinical features of 'osteoarthritis' in this unusual pattern provides an opportunity for new cases of HH to be diagnosed. So much so that any patient presenting under the age of 60 with what appears to be OA in these joints, especially if the 2nd and 3rd MCP joints or the ankles are involved, should have iron studies performed.

There is much that is not understood about the arthritis that occurs in HH patients.

Fundamental questions remain unanswered, such as what exactly causes the arthritis, why particular joints are involved, and why venesection usually makes no difference to existing joint pain, nor prevents new joints becoming affected. An obvious assumption is that it is the iron in the joints that causes joint damage, either directly or indirectly, and there are strands of evidence to support this. However we don't know why venesection should be a successful means of improving some aspects of the disease (e.g. in the liver) but make little or no difference to the joints.

Research is required to answer important questions, including whether joint pains are dependent or independent of the development of iron overload, whether current venesection regimes are effective in removing iron from joints and whether venesection might be more effective for the joints if started earlier.

Perhaps there would be more success in preventing the development of arthritis if iron were not allowed to build up in the joints, a strategy that could be tested in patients with the genetic abnormality before developing iron overload.

Although current iron depleting strategies do not prevent the progression of arthritis in existing patients, much can be done to help decrease the pain from affected joints. Simple lifestyle changes can make a difference to the hips, knees and ankles.

It is important to avoid high resistance load bearing exercise through these joints such as racket sports, and contact sports like football and hockey.

For everyday activities shoes should have thick cushioned heels and soles (e.g. trainers) and a biomechanical assessment of gait by a podiatrist is recommended to see if an orthotic insole might improve weight bearing through the feet and minimise adverse loading through the legs. Building up compensatory muscle strength and stability around affected joints is also very beneficial, and the best exercises are swimming,cycling and the cross-trainer, supplemented by Pilates classes.

Recent advances in the fields of pain management and traditional OA have shown us the best ways to use existing pain killing (analgesic) drugs and given us new therapies to minimise pain. Pain killers can be divided by type, including simple analgesics, non steroidal anti-inflammatory drugs (NSAIDs) and centrally acting neuromodulators, and also by mechanism of action including peripheral (at the source of pain, i.e. the joint) and central (i.e. the brain). The doses of these drugs can be carefully titrated (adjusted) to maximise benefit, and agents with differing mechanisms of action can be used in combination.

It should be remembered that in HH patients these will suppress pain but not alter the effects of iron in the joints or the pathological processes leading to joint damage

At St Georges Hospital in London a Haemochromatosis arthropathy clinic is being set up,specifically for patients with HH. It will be led by Dr Kiely, supported by colleagues in chemical pathology, radiology and histopathology. It is planned to provide a rheumatology service for patients with HH, both with and without arthritis. To make progress in our understanding and treatment of Haemochromatosis arthropathy, we need to understand more about the processes that lead to joint damage.

The IHA would like to thank Dr Kiely for allowing us to include this very informative article. Dr Patrick Kiely is a Consultant Physician and Rheumatologist at St Georges Healthcare NHS Trust, London. At the AGM of the British Haemochromatosis Society on June 9th 2012, Dr Kiely spoke on arthropathy .

IRISH LIFE HEALTH AND WELLBEING WEEK NOVEMBER 2012

The IHA were invited to man a stand on Friday Nov 16th 2012 as part of the Health & Wellbeing Week, organised by Ann-Marie Walsh at Irish Life plc, Lower Abbey Street, Dublin 1. The IHA treasurer, Brendan Keenan, represented the Association and was very pleased to find that so many people were interested in learning more about the disorder. At least 50% of people had already heard of the disorder.

THE ANNUAL GENERAL MEETING OF EFAPH

The Annual General Meeting of the European Federation of Associations of Patients with Haemochromatosis (EFAPH) took place in Rennes from August 29th to September 1st 2012. Ann McGrath and Margaret Mullett represented the Irish Haemochromatosis Association. The member countries of EFAPH are Belgium, France, Germany, Hungary, Ireland, Italy, Norway, Spain and the United Kingdom.
The aim of EFAPH is to raise awareness of Hereditary Haemochromatosis in all European countries both in the health professionals and the general public.

EFAPH Expansion and Future Projects:

- It is planned to set up national associations in Austria and Norway.
- A European Network of Reference Centres (ERN) for iron metabolism diseases will be headed by Professor Graça Porto (Chair of EFAPH Scientific Committee). The situation is currently very diverse in Europe. In France the national reference centre for rare iron overloads of genetic origin is located in Rennes; it is headed by Pr. Pierre Brissot (CHU Rennes) and coordinates 9 French competence centres as well as genetic diagnostic labs. Within EFAPH only 3 countries have a National Reference Centre (France, Portugal and Norway); the most structured is the Rennes Centre. In Spain there is a coordination of expert centres and in 6 other member countries there are several uncoordinated expert centres: Italy, UK, Ireland, Germany, Hungary and Belgium.This project will be very time consuming for the EFAPH Scientific Committee; it implies a close coordination with each member country, so that in the long term there will be a national reference centre in each country.
- An epidemiological and clinical research programme on Haemochromatosis related arthropathies is contemplated.

It would be managed by the Scientific Committee of EFAPH in cooperation with Pr. Pascal Guggenbuhl (Rennes CHU).

THE FIRST EUROPEAN SURVEY ON GENETIC INFORMATION

In general the results of the survey showed that patients prefer to get information on their genetic tests from their General Practitioners and their Consultants. When questioned 70% of people felt that they were sufficiently informed about genetic testing and its implications on their health. The survey was sent to 9 European countries and the results will be analysed and available in due course.

Professor Pierre Brissot, Head of Liver Diseases, Rennes, gave a very interesting presentation on the different forms of Haemochromatosis.

Type 1 is the usual form and is related to the HFE gene (C282Y mutation). It is clinically expressed only at the adult stage. It is estimated that 2.2 million people in Europe have a genetic predisposition, but all will not have the disease. The prevalence is higher in Brittany (1 out of 200) and in Ireland (1 out of 80).
Rare forms can develop not only in adults but also in children or teenagers; the corresponding genetic mutations are different (4 types). They are most often very severe and can be observed worldwide. Haemochromatosis in children is a very rare, severe but curable genetic disease. Recent genetic advances have identified different forms of HH affecting children or adolescents.

Type 2 Juvenille Haemochromatosis is related to mutations of 2 different genes (Hemojuvelin and Hepcidin). These forms are extremely rare and result in hypogonadism and heart failure.

Type 3 Haemochromatosis (mutations of transferrin receptor 2) has been reported in young patients and is extremely rare.

Type 4 Haemochromatosis is caused by a mutation in Chromosome 2 (Ferroportin). It is rare and occurs in adults. It is caused by a dominant gene unlike the other forms of HH which are recessive.

REPLY FROM DR JAMES REILLY'S SECRETARY TO THE IHA

'The Minister for Health, Dr. James Reilly, T.D., has asked me to thank you for your recent email concerning free-at-point-of-care walk in clinics for haemochromatosis patients mentioned in the HSE's 2012 Service Plan.
The Minister is anxious that a model of care for haemochromatosis patients that would provide equal access to patients in the most cost-effective manner possible be established .
As you may be aware, Dr William Murphy, has been seconded from the IBTS to the HSE to undertake a range of activities relating to transfusion medicine. Part of Dr Murphy's brief is to develop a model of care for haemochromatosis. The Minister understands that Dr Murphy is currently in discussion with all relevant parties regarding a comprehensive plan for haemochromatosis care.
The plan entails every patient with haemochromatosis having a venesection at either a GP surgery or at an IBTS clinic. Under the plan the HSE would pay the GP for every venesection but the IBTS would not be paid for any venesection that would become part of the national blood supply. Most importantly, the patient would not have to pay for the service.

The Minister is aware that discussions in regard to this plan are at an early stage and he looks forward to hearing the outcome'

THE UNEXPECTED CONSEQUENCES OF IRON OVERLOAD: SPOOF THRILLER

James Minter was diagnosed with Hemochromatosis in 2004. He is a member of the British Association. James is a fiction writer based in the UK. He has recently completed a spoof thriller book called "The Unexpected Consequences of Iron Overload." The aims of the book are to raise awareness of Iron Overload, raise funds for Haemochromatosis education and research and hopefully to entertain fellow suffers. The book is on www.amazon.co.uk and www.amazon.com in both paperback and eBook (Kindle) formats. The paperback retails for £10 and the Kindle version is £1.28.

INFORMATION MEETING AT RESOURCE CENTRE BALBRIGGAN WEDNESDAY 21st N0VEMBER

The Irish Haemochromatosis Association were invited to talk on Haemochromatosis to members and staff of the Fingal Traveller's organisation. Two of the IHA members, Jim Browne and Margaret Mullett spoke from a personal perspective and explained the hereditary nature of Haemochromatosis. Many of those present were familiar with HH and family members had already been diagnosed.
Jim stressed the importance of alerting brothers, sisters, parents and children of sufferers to the possibility that they may also have the mutated genes and should be genetically tested. Many traveller families are very large and one young man at the meeting had 16 siblings. Because a lot of marriages are with other members of the Travelling Community, it is very likely that HH is even more common than in the general population. Suzie Mc Carthy, Primary Healthcare Co-ordinator at the centre is determined to help raise awareness of HH by including information in the travellers spring newsletter and distributing brochures and posters.
The IHA would like to express their thanks for the invitation and appreciated the opportunity to meet representatives of the organization.

RENEWAL OF SUBSCRIPTION

Membership renewal forms and standing order forms are enclosed with this newsletter. Please complete and return to the 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 no longer wish to be included in our mailing list and would like to cease membership of the Association, 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 Kate:kategeog@gmail.com
The new email address for Margaret is margaretmmullett@gmail.com and for Denise McAuliffe it is denisekellymcauliffe@gmail.com

DUNDALK INFORMATION MEETING TUESDAY MARCH 5th 2013

An Information meeting on Haemochromatosis has been organised by the HH specialists nurses from the Louth County Hospital. The venue is the Fairway Hotel, Dublin Road, Dundalk. The speakers are Dr Sengupta and Dr Keohane and the time is 7.30 pm. All are welcome.

THE PLOUGHING CHAMPIONSHIP IN NEW ROSS SEPT 25-27 th 2011

Therese and Jarlath Huges with Denise McAuliffe and Brendan Keenan

This year the National Ploughing Championships,were held in New Ross in Co Wexford.
Over 187,000 people attended the three-day event, which was opened by President Michael D Higgins on Tuesday 25th Sept. Poor weather affected the first two days of the event, while high volumes of traffic were also an issue at times.
The organisers said they regretted that some motorists had long delays getting to and from the parking sites over the course of the event.
The IHA would like to thank the voluntary helpers who gave so generously of their time to man the stand over the three days. This year because of weather and traffic conditions their work though rewarding was more difficult than previous years.