Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

Winter 2014

Welcome to the Winter Newsletter 2014

In this issue:

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

IRISH LIFE CENTRE MONDAY OCT 6TH

IHA board members, Margaret Mullett (on the left) and Ann Mc Grath were delighted to be given the opportunity to meet and raise awareness of HH with the Staff of Irish Life at a Health and Wellbeing Fair in October.

Elite Athletes and Haemochromatosis

Professor Jean–Francois Toussant

Genes are associated with high level sport performance. Iron is essential for the manufacture of red blood cells, muscular metabolism and heart function.
Jean–Francois Toussant is a Professor at the National Institute of Sport, Expertise and Performance (INSEP) in Paris. He presented a paper at the European Iron Club meeting in Verona in September. His team assessed HFE mutation frequency (H63D, C282Y, S65C) in elite members of French teams requiring energetic power- Nordic ski (n=77), rowing (n=18) and judo (n=34) and elite athletes of a non-energetic sport petanque, a form of boules (n=41).
These frequencies were compared with 219 control subjects matched by age, gender and geographical origin. Blood samples were tested for red blood cells and iron parameters. **It was shown that the frequency of HFE mutations in elite athletes of aerobic sport is twice as high as those athletes of the non energetic sports and of control subjects.**
For athletes with Olympic, World or European medals, HFE mutation frequency is 12 fold higher. Biological parameters including haemoglobin, hematocrit, red blood cells and ferretin of all athletes are higher than those of the control group.
Phenotypes linked to high level sport performance are associated with a higher frequency of HFE gene mutation in elite athletes who reached Olympic, World and European podiums in Nordic ski, rowing and judo.

Editors note:
At least two members of the IHA are GAA All Stars! The All Star awards are considered to be the most coveted sporting awards in the country.

David Beggy

David Beggy is an All-Ireland winning Gaelic footballer from County Meath. David won two All-Ireland Senior Football Championships in 1987 and 1988. He received two all stars while playing with Meath as well as playing Rugby with Leinster.

Seamus Leydon

Seamus Leydon is another All Star. He was a vital part of Galway's great three-in-a row side of the Mid 60s. Apparently, he was the scourge of all right half backs up and down the country for a decade.

EFAPH Meeting September

The tenth AGM of EFAPH, the European Federation of Associations of Patients with Haemochromatosis, took place in Verona in September. Delegates from France, Germany, Belgium, Norway, UK, Switzerland, Denmark, Portugal, Spain and Hungary attended the meeting and the IHA was represented by board members, Ann McGrath and Margaret Mullett.
Professor Pierre Brissot, Rennes, reported on the recent developments in Haemochromatsois research which had been discussed at the European

EFAPH
European Federation of Associations of Patients with Haemochromatosis

Iron Meeting (EIC). The meeting ran concurrently with the EFAPH meeting in Verona. Three prizes were awarded by EFAPH to young scientists for their work on Haemochromatosis.
Much of the current research involves Hepcidin. Prof Brissot mentioned that only 30% of males and 1% of females with homozygous C282Y mutation got severe Iron overload.

Fr Richie Higgins tells his story

Fr Higgins has been a member of the IHA since it started in 1995.He believes that he was one of the very first people diagnosed with Haemochromatosis (HH) in Ireland.

"I was a priest working in Glasgow. I led a very busy life and played golf and tennis to keep fit. In later years I used to tire easily and I assumed it was due to work and exercise.

In 1970, I was on a golfing holiday in Wexford and I became weak and collapsed. I returned to my home in Athenry and visited the doctor who confined me to bed for a week and treated me for pleurisy.

On returning to Glasgow my G.P. immediately sent me for an X-Ray. Following the X-Ray I was hospitalised. I presented with an enlarged heart, a significant murmur and a diseased mitral valve. My skin was pigmented. For weeks I was treated for a heart condition and x-rayed regularly to see if the heart had diminished in size as a result of the bed rest.

Due to my colour the consultants thought it was a form of Leukaemia. They did not refer to the high iron content in my body nor did they consider carrying out a biopsy. They did not realise that the iron deposits in my heart had damaged the mitral valve and enlarged the heart. It would appear that they had never encountered HH.

It was suggested that I had venesection two or three times a week. After several treatments the x-rays showed that the size of the heart had diminished – obviously some of the iron deposits had been removed.

I continued to give two units of blood per week. After 22 weeks in Glasgow Hospital I asked to be transferred to Galway University Hospital under Professor Mc Carthy. He carried out several tests including a liver biopsy which was necessary to analyse the iron content in the liver.

> I would like to advise people never to let a HH diagnosis get you down, you won't die from it; you can control it and live a normal life as I do.

After I left hospital I was still very weak and weighed only nine stone. After a period of convalescence at home, I spent two years in a Nursing home where I acted as Chaplain to the residents. During that time I had to visit the hospital twice a week for venesection and injections. It was a long process of recovery. The heart came back its normal size, the mitral valve repaired itself and the murmur was non existent. By this time I had had over 150 units of blood removed. Two years later I was ready to take up an appointment as a hospital chaplain. I remained there for 14 years which suited me because I could be treated in the hospital.

My case is a success story. My recovery was remarkable and I was able to return to busy and very fulfilling parish work. Without my good health, this would not have been possible.

Now I have retired as Parish Priest but I help out in Maree and the work is not too strenuous. I still play golf regularly. I celebrated my 80th birthday this year and as I was only 36 when I was first diagnosed, I think I am the longest in the country to still have venesection every three months. There were nine siblings in my family and five out of the nine were diagnosed with Haemochromatosis.

I want to say thanks to Professor McCarthy and all the nurses and doctors in University College Galway and the Health Centre in Gort for their excellent care and attention.

To conclude I would like to advise people never to let a HH diagnosis get you down, you won't die from it; you can control it and live a normal life as I do".

We thank Fr Richie for sharing his story and wish him many more happy years in his home in Oranmore, Co Galway.

Clonmel Meeting Thursday, Nov 6th 2014

(Education Centre, South Tipperary General Hospital)

Sareen Walsh, Eleanor Maher, Dr Clare O' Leary, Breda Foley and Prof Paud O' Regan

The speakers were Prof Paud O'Regan, Dr Clare O 'Leary FRCPI, Nurse Breda Foley and dietician Sareen Walsh.
The fact that the blood venesected in Clonmel Hospital is not being used for transfusion was mentioned. Prof O 'Regan raised the question as to why it is not possible for the IBTS mobile unit to come to Clonmel periodically to collect blood from Haemochromatosis patients? Dr Clare O 'Leary gave a comprehensive overview of the diagnosis and treatment of Haemochromatosis and explained the genetics of the disorder.
Nurse Breda Foley discussed relevant issues of concern to HH patients. Breda and her nursing colleagues have been an ongoing support to the IHA.

Dietary advice from Sareen Walsh

- Aim to eat a healthy well balanced diet
- Avoid all supplements that include iron & vitamin C. These include multivitamins which may contain vitamin C, multivitamins & minerals which may contain both iron and vitamin C and tonics which again may also contain both
- Note the difference between haem iron and non haem iron. Haem iron is well absorbed so intake should be reduced. Examples are red meat, offal, oysters and mussels
- Examples of non haem iron are green leafy vegetables, pulses (peas, beans and lentils), dried fruit, nuts and seeds. No necessity to reduce intake
- Do not drink vitamin C rich drinks with mealsá
- Tannins (tea), Calcium and phylates reduce iron absorption
- Avoid fortified bread and cereals

Hepcidin – Dr Dan Johnstone PhD

While most of us can manage our iron overload through routine venesection, this is not feasible for everybody. Some people have a strong aversion to needles, others have blood vessels that are difficult to cannulate, and others may have non-iron-related anaemia that would be worsened by blood removal. For these reasons, there has been considerable interest in ways of controlling the small molecule hepcidin.

Hepcidin is a hormone that is produced by the liver and circulates in the bloodstream. Since its discovery in 2000, hepcidin has been a focal point of the iron research field, as it appears to be the master regulator of body iron levels, controlling how much iron is absorbed from the diet and how much is released from the body's cells. People with haemochromatosis naturally have low hepcidin levels; this is believed to be what causes them to accumulate more iron than is healthy. Scientists believe that by finding a way to increase hepcidin levels or activity, they will be able to restore normal iron control to haemochromatosis patients. In a recent study, a research group in Boston screened over 10,000 different chemicals to see how they affected the production of hepcidin. From this screen they identified 16 chemicals that were capable of increasing hepcidin production. In particular, two of these chemicals, named ipriflavone and vorinostat, were particularly potent.

Hepcidin is a hormone that is produced by the liver and circulates in the bloodstream. Since its discovery in 2000, hepcidin has been a focal point of the iron research field, as it appears to be the master regulator of body iron levels, controlling how much iron is absorbed from the diet and how much is released from the body's cells

As this study only looked at the effects on cells grown in culture, a lot more work needs to be done to show that these chemicals are both safe and effective in animal models. But it provides a few good candidates for follow-up studies and may, in a few years' time, lead to a suitable drug therapy for haemochromatosis patients who cannot tolerate regular venesection.

Sincerest thanks to Dr Johnstone and the Australian Association for permission to include this information. Dr Johnstone is a research scientist in Sydney and spent a very happy year in 2005 as a young doctor in Galway.

National Ploughing Championship (NPC) September 23rd -25th

This year there was a record crowd at the NPC. Sincerest thanks to our members and also volunteers who manned the stand over the three days.

Aidan, Michelle, Deirdre and Jackie from the Pharmaceutical company Glaxosmithkline.

Volunteers Olive and Tony McDevitt, Ann Dooley and Brendan Keenan

Factors influencing iron overload

Graça Porto, MD, Ph D

Institute for Molecular and Cell Biology, University of Porto, Portugal

The question of great variability in the degree of iron overload has intrigued scientists for a long time. The fact that almost all patients with classical Haemochromatosis (HH) have the same ancestral mutation (C282Y) in the HFE gene, would predict a great similarity in the clinical expression amongst patients. But that is not the case.

In 2002 it was shown that a large number of people with two copies of the C282Y gene do not develop iron overload and therefore other factors should be found to explain why some people have the disease and others do not.

So what are the modifiers of iron overload?

The modifiers of iron overload can be divided into two major categories: the environmental modifiers and the genetic modifiers.

The environmental modifiers are relatively well known and they include, besides the effects of sex and age, the life-style habits, body weight, alcohol consumption, blood loss, etc.

Regarding genetic modifiers, a group of scientists in Portugal, have for many years focused on the role of CD8 T lymphocytes as modifiers of iron overload. Their research has shown that HH patients with low numbers of circulating CD8 T lymphocytes have more severe iron overload. These lymphocytes are important cells of the immune system and have been shown to be capable of taking up and retaining a toxic form of iron in circulation.

Studies involving HH patients also demonstrated that low CD8 T cell numbers in HH patients is "genetically determined" and that this additional genetic characteristic is transmitted together with the inheritance of the HFE mutation.

The story of CD8 T lymphocytes and iron overload is not complete and many aspects are still to be discovered.

For example, it is now very important to understand the mechanism of how CD8 T lymphocytes handle the iron in circulation and if that function is altered in people with HH. Knowing all this may in future help to control the severity of iron overload in HH patients.

Thanks to Dr Graca Porto for sharing this information with us. Dr Porto is a member of the Scientific board of EFAPH and is invaluable to the association.

A more detailed account of this research is available on the web-site www.haemochromatosis-ir.com.

Athlone Information Meeting November 5th 2014 (at the Sheraton Hotel)

Dr Gerard Clarke

Oliver Brennan, Vincent Mc Cabe , Rosaleen Mc Court and Imelda Mc Cabe

Dr Gerard Clarke, FRCPI, Consultant Gastroenterologist, Portiuncula Hospital, Ballinasloe gave an excellent presentation followed by a very helpful and interesting question and answer session.

Dr Clarke feels that people should be more proactive with regard to their treatment. They should take responsibility for their condition and be aware of their blood levels, what their ferritin level is and what the normal level should be. He stressed the importance of the transferrin saturation level in the diagnosis of Haemochromatosis.

> People should take responsibility for their condition and be aware of their blood levels, what their ferritin level is and what the normal level should be.

Dr Clarke hopes that in due course, the blood venesected in Portiuncula Hospital would be used for transfusion there and in Roscommon and Galway Hospitals.

Sincerest thanks to Dr Clarke for his ongoing help and support to the IHA.

TG4 Programme Wednesday 17th September

An excellent programme on Haemochromatosis was recorded and produced by TG4 on September 17th. Dr. Sinéad Ní Bheirn, GP, interviewed two native Irish speakers, Felim McDonnacha and Aodh O'Coileain. Both men gave a very interesting and informative account of how they were diagnosed and how HH affects their life.

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 no longer wish to be included in our mailing list, we would appreciate it if you would let us know as the cost of posting and printing is 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

Facebook

We are now active on Facebook. Find us and like us at www.facebook.com/irishhaemochromatosisassociation