Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

WELCOME TO THE SUMMER NEWSLETTER 2015

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LAUNCH OF AWARENESS CAMPAIGN AT FARMLEIGH MAY 29th

Minister Leo Varadkar launched the campaign and promised his support to the IHA. Professor Suzanne Norris and members of the IHA attended the launch. Prof Norris said that "ill -health from Haemochromatosis and the development of serious complications such as cirrhosis can be prevented by simple treatment. Life expectancy in treated non-cirrhotic patients is normal."

Minister of Health Leo Varadkar with Molly Geoghegan and Clare Bolger.

Photographed with Minister: Brendan, Phil, Niall, Michelle, baby George , Margaret, Noel, Clare, Molly and Ann.

ANNUAL GENERAL MEETING SATURDAY MAY 23rd 2015

The AGM took place at the Irish Blood Transfusion Service in St James's Hospital, Dublin 8 on Saturday May 23rd and was attended by 80 people. Margaret Mullett gave the Chairperson's report and the Treasurer's report was given by Brendan Keenan. The board of directors agreed to stay on for another year. Kate Geoghegan took over as secretary from Ann Campbell who will stay on as a board member.

The IHA would like to thank the IBTS for once again making the centre available to us and for generously sponsoring morning coffee and lunch.

We would also like to thank Dr Barbara Ryan, Dr Patrick Kiely and Miriam Mulkerrin Mason for their excellent presentations.

PRESENTATION BY DR BARBARA RYAN

M.D, M.Sc., F.R.C.P.I.

Consultant Gastroenterologist, Adelaide and Meath Hospital, Tallaght

What is Haemochromatosis?
- A genetic condition
- Mutations of certain genes which produce proteins involved in controlling iron metabolism
- Cause increased iron absorption from the gut
- Excess iron is deposited in tissues, such as the liver, pancreas, and heart, joints- this damages the functioning of the tissues involved

Causes
- Failure of normal control of iron absorption
- Increased iron absorption over a long period of time
- Initially excess iron stored as ferritin
- Accumulation of ferritin results in production of haemosiderin and release of iron
- Excess iron -> free radicals - damages the tissues

In Ireland
- 1 in 80 people homozygotes
- 1 in 5 people are carriers
- Highest frequency in the world

Why is HH So Common in Ireland?
- The gene originated in a Celtic / Viking ancestor 2000-4000 years ago
- Improves iron absorption
- Therefore is advantageous when nutrition is poor
- Reliance on potatoes / the Famine
- Excessive consumption of tea

Clinical Stages
- O-20 yrs - clinically insignificant iron accumulation (0-5g iron excess)
- 20-40 yrs – iron overload without disease (10-20g iron excess)
- >40 years/post menopausal females – iron overload with clinical disease (> 20g iron excess)

Clinical Manifestations

Liver disease - More likely if consuming >28 units alcohol per week
- Manifestations of Liver Disease include
- Abnormal liver tests
- Fibrosis
- Cirrhosis
- Increased risk of Hepatocellular cancer (20x)

Cardiac – 15%
- Dilated cardiomyopathy
- Conduction defects (irregular heart rhythm)
- Venesection

Diabetes – 48%
- Venesection may reduce insulin requirements
- 1.3% of type II

Other Endocrine Manifestations
- Hypothyroidism
- Osteoporosis
- Early menopause
- Sexual dysfunction
- May respond to venesection
- Skin pigmentation -70%
- Fatigue is the most common symptom
- Abdominal pain

Arthropathy
- Small joints of hand
- Wrists, knees, hips, shoulders
- Elevated circulating iron increases susceptibility to infections
- Listeria, Yersinia, Vibrio

Diagnosis
- Transferrin saturation
- Ferritin
- Genetic testing
- Liver biopsy

Genetic Testing (HFE)
- C282Y homozygous
- C282Y/ H63D compound heterozygote
- Carrier
- H63D homozygote

Who should be screened for HH?
- First degree relatives
- Abnormal liver tests
- Unexplained early arthropathy
- Unexplained sexual dysfunction
- Cardiomyopathy
- Type II diabetes?
- Patients with unexplained fatigue

Treatment
- Venesection removes 250mg
- 10g overload will take 40 venesections to clear
- When?
- Ferritin >200 females; 300 males
- Weekly
- Maintain hydration
- Avoid vigorous exercise x 24hours
- Keep Hb ~11g/dl
- aim to bring Ferritin <50 and TS <50%

Dietary Recommendations
- Avoid iron supplements
- Avoid Vitamin C supplements
- Avoid uncooked shellfish
- Mild alcohol consumption
- Limit iron rich foods – e.g. red meat

Prognosis
- Normal in non-cirrhotic and non-diabetic patients
- Causes of death
- Cirrhosis
- Hepatocellular cancer
- Cardiomyopathy
- Diabetes
- Early diagnosis ensures excellent prognosis- normal life expectancy and normal quality of life

Summary
- HH is the most common genetic condition in Ireland
- 1/5 is a carrier
- Early diagnosis is key
- Early diagnosis and treatment leads to normal life expectancy
- Avoid excess iron in the diet

Dr Ryan's excellent overview of Haemochromatosis was followed by a lively Q and A session.

INFORMATION MEETING IN TRALEE, THURSDAY OCTOBER 8TH

A meeting will take place in Fels Point Hotel, Tralee on Thursday October 8th at 7.30 pm. The speaker is Dr James Ryan, Consultant Endocrinologist , Bon Secours Hospital, Tralee. All are welcome.

NATIONAL PLOUGHING CHAMPIONSHIP, SEPTEMBER 22nd – 24th

The NPC will take place at Ratheniska, Co Laois. If you are available to help man a stand, please contact us margaretmmullett@gmail.com

LOURDA TELLS HER STORY

"I was diagnosed with Haemochromatosis (HH) in early 2006. For the previous year or two I had been feeling extremely tired and lacking in energy. In fact I had been on an iron supplement and vitamin C to try and give my energy a boost (quite the wrong thing as it transpired!). Working as a secondary school teacher, I found the tiredness really difficult.

I had severe lower abdominal pain, particularly in the morning. I changed the mattress in the bed because I thought that might be the problem! I had joint pain in my arms and legs.

My menstrual cycle had been erratic in my thirties and stopped altogether before I was forty. There was no medical reason for this. I didn't have any children.

Many years earlier I remembered getting blood tests done and the doctor commenting that my iron level was very high but nothing more was done about it. At around the same time my nephew did a medical for a job and was told his iron level was very high and that he should be tested for Haemochromatosis. I read up on Haemochromatosis and decided that putting all these things together, I

should get my GP to do a ferritin test.

When the result of the test came back the ferritin level was sky high. It was almost 2,400 ng/dl when it should have been under 200 ng/dl. My transferrin saturation was 67% when it should have been under 45%. The G P immediately ordered more blood tests and requested that the genetic test be done. The genetic test showed that indeed I did have Haemochromatosis, I was then referred to a haematologist to begin treatment. Interestingly, I was the first patient in this practice to be diagnosed with HH.

The venesections began in April 2006 and I had one every fortnight until October 2007 and then every month until August 2008. I found these two years very hard going but my ferritin levels came down to 60 ng/dl and I now have venesection four times a year.

However if I let it go too long without venesection my ferritin goes up very quickly and I might have to have two or three in two months to bring the level back down. I always know when I need to have a venesection as I get very tired and lethargic

I am under the care of a gastroenterologist and have had scopes done to check my organs. Thankfully there is no major damage. I still have joint pain in my fingers and leg. So far I am clear of diabetes but my last blood test was border line so sugar levels have now to be monitored.

There are six children in my family but I am the only one with HH, one sister is a carrier but the others, one brother and three sisters are normal.

I am very sallow skinned as are some of my family and my mother's family which is reflected in the old name for HH - Bronze diabetes.

I feel lucky that I discovered my HH early. I lead a full and happy life and hope to continue doing so for a long time yet."

Sincerest thanks to Lourda for sharing her very interesting story with us.

HAEMOCHROMATOSIS ARTHROPATHY

Dr Patrick Kiely

Dr Patrick Kiely, Consultant Physician and Rheumatologist, St Georges Healthcare NHS Trust, London, spoke at the AGM. He presented the results of the British Haemochromatosis Society survey on arthritis. Below is a synopsis of his presentation:

Joint pain is very common in patients with genetic Haemochromatosis (HH).

Of 244 respondents to the British Haemochromatosis Society members survey, joint pains or arthritis were reported, in as many as 72% of cases.

Dupuytren's contracture which is a progressive bend of the fingers occurred in 10% of those surveyed (13% in men, 8% in women).

Knees and hips are commonly affected but the high frequency of involvement of knuckles of the middle fingers and the ankles is a characteristic feature of HH. Any patient presenting under the age of 60 with what appears to be Osteoarthritis (OA) in these joints, should have iron studies performed.

Help for Arthritis Sufferers

- Simple lifestyle changes can make a difference to the hips, knees and ankles. It is important to avoid high resistance load bearing exercise 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.
- Anti-inflammatory drugs such as Paracetamol and Codeine based analgesics were most frequently used for joint symptoms with the majority reporting them to be effective.
- Amitriptylline and joint injections were less used but also reported to be effective. 23% of respondents had joint surgery, most frequently to the hip and knee.
- Results of the survey indicated that only 13.6% of those interviewed felt that venesection improved their joint pain.

UPDATE ON MEETING WITH MINISTER VARADKAR

Since the meeting with Minister Varadkar in March, the IHA represented by Maurice Manning and Margaret Mullett have met with Dr Colm Henry and Ms Dee Conroy of the HSE on Wednesday May 20th. Dr Henry is the designated person in the HSE in regard to HH.

At the meeting It was agreed that the views of persons with Haemochromatosis be sought on a number of issues with a view to helping formulate change in policy and practice. A questionnaire has been emailed out to members and many people have already responded.

Other items on the agenda included:

- Greater involvement by the HSE in raising awareness of Haemochromatosis, both in the medical profession and in the community
- Equity of access to venesection and treatment. As it stands there is a huge discrepancy in cost and availability of treatment throughout the country.
- Increased involvement of GPs in treatment.
- Role of IBTS - problems that are currently experienced regarding access to venesection and how this might be improved.

A follow up meeting with Dr Stephanie O'Keeffe and Dr Colm Henry is scheduled for Wednesday July 15th.

Dr Henry has already met with a consultant group to discuss a pathway of care for diagnosis and treatment of Haemochromatosis.

WOMEN'S MINI MARATHON JUNE 1st 2015

The IHA would like to thank all the ladies, including Carmen Alvarado pictured above, who ran the Women's Mini Marathon for the IHA.

HAEMOCHROMATOSIS AWARENESS DAY, THURSDAY JUNE 4th

Sincerest thanks to all the people who helped man stands in over 34 venues throughout the country. It would not have been possible to organise the event without the support of members of the Association and friends.

Pamela O'Donovan in Galway

Kay Kenny Bedford, Gay Murphy, Liz O'Callaghan and Denise Mc Auliffe in Limerick

Nessan O' Shea, Kathy O' Dwyer and Finbar O' Brien in Cork

Geraldine Mc Swiney and Ann Hannan in Tralee

Michael and Catherine Mc Carthy in Cork