ANNUAL GENERAL MEETING SATURDAY MAY 29TH

The meeting which took place at the Irish Blood Transfusion Service (IBTS) in St. James's Hospital and was attended by over 100 people. The IHA would like to thank the IBTS and the Medical Director, Dr. Murphy for once again making the centre available to us and for generously sponsoring morning coffee and lunch. We would also like to thank Dr. Barry Kelleher, Dr Willie Murphy and Mr Michael Hallissy for their excellent presentations.
Mr Frank O Meara spoke on his concerns regarding the exorbitant costs claimed and paid by VHI for phlebotomy procedures .This is a major issue with many members and we greatly appreciate the fact that Frank has been very actively involved on our behalf in negotiations with VHI.
A very informative and helpful question and answer session followed the talks.

Two of our Directors resigned this year and two new Directors were elected. The other Directors have agreed to stay on as board members for another year. We welcome the new Directors - Kathy O Dwyer from Cork and Brenda Flannery from Dublin.
At the AGM it was proposed that the first sentence of Article 28 of the Articles of Association of the IHA be amended to read "The board of directors of the Company shall comprise at least seven directors" (instead of "The board of directors shall comprise seven directors")
This proposal was approved and accepted by all present.

Former Directors

Rosaleen Mc Court, is stepping down because of family and work commitments. Over the years, her contribution to the Association has been invaluable and without her it is probable that the Association would not have survived. As a board member, her sound advice, good humour and good judgement have been a great help and she has become a great friend to all on the board. Rosaleen travelled to meetings from Roscommon and has been on the board since the beginning .She was treasurer for several years. We wish Rosaleen every success and will miss her. However, we are delighted that Rosaleen is now an Honorary Director of the Association and will be there for us.

Brendan Gallagher is resigning for Health reasons. Brendan joined the Board in 2005 and has been treasurer for the last five years. The IHA were very fortunate to have Brendan who is an Accountant, as our treasurer. Thanks to Brendan we applied for Lottery money in 2008 and were successful in getting a sizable donation. Brendan was a great addition to the board and despite chronic pain he was always upbeat and in the best of humour.We wish this very admirable, capable and courageous man every success in the future and the best of health.

A PATIENT'S PERSPECTIVE
Michael Hallissey

Michael spoke at the AGM from the patients' perspective. Michael is aged 45 and comes from Killarney. He now lives in Kildare with his wife and two children. Originally he trained and worked as a teacher but has been an IT consultant for the last 20 years. He was diagnosed with Haemochromatosis in August 2008. He says it was a red letter day for him as he finally knew what was wrong with him!
Over 20 years ago he played a lot of both football and hurling at his college in Cork. He would come home from school and have a nap before heading out to play another game. He thinks that maybe the iron helped him at this stage but he does remember chronic tiredness in his early twenties. As time moved on he lived abroad. The tiredness was always there. As he moved into his thirties he felt very lethargic and chronically fatigued. He visited his GP who put him on iron tablets. After three years he started to develop soreness in his knuckles. His dad had just been diagnosed with Haemochromatosis. His sister had also been tested and had HH. Michael who is from a family of ten then decided it was time for him to be tested and was very relieved to know that there was a reason for his symptoms. His iron levels were over a thousand. He now goes to Nurse Liz Ellis in St James's and has regular venesection. He says "the treatment is fantastic and Liz is brilliant".
We thank Michael and appreciate the fact that he also went on the Morning Show on TV 3. He was a natural performer and almost immediately over 20 calls came in to the IHA.

EMAIL ADDRESSES FOR MEMBERS

If you have not received an email from us recently we probably do not have your correct email address. To help us rectify this please send Catherine Geoghegan, an email with your full name and Haemochromatosis as the subject. kategeog@gmail.com

NATIONAL PLOUGHING CHAMPIONSHIP(NPC) TUES 21ST TO THURS 23RD SEPTEMBER

The NPC will take place at the same venue as last year, Cardenton, Athy, Co Kildare from Tuesday 21st to Thursday 23rd September.
The IHA will have a stand again this year and would greatly appreciate your help in manning the stand. Please contact Margaret at 01 4922705 or email margaretmullett@ireland.com

Irish Haemochromatosis Association

Carmichael Centre, North Brunswick Street, Dublin 7

Summer/Autumn 2010

Welcome to the Summer/Autunm Newsletter

In this issue:

FEATURES

HAEMOCHROMATOSIS AWARENESS DAY, THURSDAY JUNE 3RD

Mark, Lisa, Nadine, Sinead and Paul highlighting the Awareness Day.

Our first Awareness Day was a great success and sincerest thanks to everyone who helped out on the day. We had approximately 150 people helping in various stands throughout the country. The feed back was great and all the volunteers reported that they enjoyed the interaction with the public, met some lovely people and had good fun as well as getting the message out there! Please consider helping out next year as it is a very worthwhile experience.

A special thanks to Mark Cagney of TV3 for his help in highlighting the Awareness Day and for being part of the photoshoot. Also thanks to Paul (Pillar) Caffrey, Nadine Rooney, Lisa Cannon and Sinead Kissane. A special thanks to Brian McEvoy (brian@publicity.ie) who did the shoot *pro bono* and made everyone feel like movie stars. Many of the Regional Newspapers and Radio Stations covered the Day and again this was a great help in raising awareness.

Ann Mc Garry in St Stephen's Green SC

Dr Muthalagu who spoke at the awareness evening in Monaghan

As part of the Awareness Day, an Information Meeting on Haemochromatosis was organised by the staff of Monaghan General Hospital. It took place in the Four Seasons Hotel Monaghan and Dr Muthalagu was the guest speaker. The evening was a great success and was attended by over 200 people.
There were information stands in several shopping centres throughout the country. In Dublin there were stands in: St Stephen's Green Shopping Centre, The Ilac Centre, Superquinn Shopping Centre, Sutton, Nutgrove Shopping Centre, Rathfarnham, Swan Centre, Rathmines, Ashleaf Shopping Centre, Crumlin, Frascati Shopping Centre, Blackrock,Civic Offices Wood Quay.

Other venues around the country included:
Cork: Mahon and Douglas Shopping Centres, Tipperary: South Tipperary General Hospital, Clonmel, Galway: Corrib Shopping Centre and University College Hospital Galway, Donegal: Letterkenny General Hospital, Waterford: City Square Shopping Centre, Wexford: Gorey Shopping Centre, Kilkenny : Market Cross Shopping Centre, Dundalk: Louth County Hospital and The Marshes Shopping Centre, Limerick: Crescent Shopping Centre, Sligo: Sligo General Hospital, The Tesco Arcade Sligo, Roscommon: Tesco Shopping Centre and Manor West SC, Tralee.

ANN'S STORY

Ann and her husband enjoying an evening out in Brussels

In 1973, I left Mallow and headed for Brussels to work in the European Commission. I enjoyed working in very interesting jobs, and worked in Cabinets often for long hours. I played lots of sport, tennis, squash, golf, and travelled frequently. However, I was constantly very tired and my main symptom was chronic fatigue which I had for a long time.
I had a wonderful English doctor, Dr Vivian Piercy, who pursued the matter. She decided to test for Haemochromatosis (HH) and eventually that was diagnosed in 1990.
Needless to say I had never heard of Haemochromatosis, let alone pronounced it - and there was no Internet or Google in those days! A friend got some information from the *British Medical Journal* which explained that the condition was rare in women and rather serious. However, my sister who is a nurse in Cork got more information, and I was reassured that with regular on-going treatment I would lead a normal life. Fortunately this has proved to be correct.
I was referred to Professor De Groote, a specialist in a Louvain hospital who carried out a liver biopsy. The result was "idiopathic haemochromatosis without cirrhosis". My ferritin level was 1200 ng/ml which of course was high. I had an enlarged liver, and they also commented on the bronze pigmentation.
It was explained to me that I would need regular phlebotomy to control the ferritin and while all this was rather frightening at the time, since then, I have had regular blood lettings (phlebotomy) every two months without any problems.
Initially for a couple of years my treatment was carried out in Louvain at the Red Cross centre. At first the Professor thought it best to take my blood, separate the cells, and just give back my plasma. But, I didn't have a good reaction to this, the procedure was slow and it took a long time. Fortunately later it was decided to just simply take the blood.
I now go to Erasme Hospital in Brussels. I attend the day clinic there in the chemotherapy unit. I also have a yearly check up with Prof Decaux at the hospital. Depending on my results (ferritin, transferrin saturation) blood letting is either every 6 weeks or every 8 weeks – and always 300ml. The blood is not re-used. Sometimes my blood pressure becomes lower, but after a longer rest it returns to normal.
I seem to be the only one in my family to have Haemochromatosis. However, I know that two of my father's sisters died around the ages of 50 and perhaps they also had Haemochromatosis.
I am very lucky in that I was able to take time off work to attend hospital all these years. Because my treatment is ongoing, I have 100% reimbursement of medical costs. But in fact the treatment is not expensive. It is roughly €15 each visit, and the specialist and lab costs are around €150 per yearly visit.
I plan to take early retirement this year, and hope now to play more golf. Having Haemochromatosis has not prevented me in any way from working, sports or travel!!
The first article on HH I read in the Irish papers was about Brian Lenihan who had a liver transplant in May 1989 as a result of Haemochromatosis. I kept an article by Kathryn Holmquist where she wrote "rarer causes of cirrhosis include Haemochromatosis, a hereditary condition etc.' dated 27th July1992. Then I read the article Braced for the blood letting in the *IrishTimes* dated April 1995 regarding two Galway sisters which I found most interesting. It was in this article that I got the address of the Irish Haemochromatosis Association.

The IHA would like to thank Ann for sharing her very interesting story and wish her many great golfing years when she retires.

MUCKROSS TRANSITION YEAR STUDENTS

We were invited to talk to Transition Year students in Muckross College. It proved to be interesting and rewarding as hopefully the receptive students passed on the information to their parents.

FLORA WOMEN'S MINI MARATHON MON JUNE 7TH 2010

The marathon was a greater challenge this year as unfortunately the rain came bucketing down. Sincerest thanks to all those who helped in any way either by participating or fund-raising.

BAG PACKING

The students in Holy Child Convent, Killiney bag packed for the IHA and raised over €350. As well as collecting this sizable amount of money the pupils also distributed information brochures.

DR. BARRY KELLEHER'S PRESENTATION ON HAEMOCHROMATOSIS

Dr. Barry Kelleher is a Consultant Gastroenterologist at the Mater Hospital, Dublin.
Dr. Kelleher explained that Hereditary Haemochromatosis (HH) is the most common cause of iron overload.
The highest frequency in the world has been identified in Ireland. In this country 1:83 are predisposed to iron overload whereas in the USA the figure given is 1:300. Increasing affluence, longevity and dietary fortification lead to a greater risk of iron overload in those genetically predisposed.
The first description of the disease was by Trousseau in 1865 and in 1889 the term Haemochromatosis was coined. Two mutations of the Haemochromatosis gene (HFE) were discovered in 1996 by Feder et al. C282Y and H63 D. Iron overload is associated with C282Y homozygosity (C282Y/ C282Y) and to a lesser extent with C282Y/ H63 D compound heterozygosity.

Presentation and Symptoms

Fatigue is the most common symptom affecting up to 60% of patients.
Other symptoms in the developed syndrome are Arthralgia/Arthritis, Hepatomegaly/cirrhosis, diabetes, cardiac failure and sexual dysfunction.
In general, the severity of the clinical illness is related to the iron burden.
The clinical condition evolves in a series of stages beginning with:

(1) Clinically insignificant iron overload (0-20 years/0-5g iron storage)
(2) Iron overload without disease (approximately 20-40 years/10-20 g iron storage)
(3) Iron overload with organ damage (greater that 40 years/greater than 20 g iron storage)

The outline above suggesting simple step-wise progression of iron overload overlooks the phenotypic variability.
Not all people with the genetic predisposition will develop significant iron overload.
The factors that influence the variability are not completely understood but research is ongoing.

Hepcidin

Hepcidin is a peptide hormone of 25 amino acids produced by the liver. It was first identified in human urine.
It is thought to be very important in iron metabolism. Hepcidin deficient mice develop iron overload whereas Hepcidin over-expressors have severe iron deficiency. Mutations in levels of hepcidin may explain why some people develop more serious disease. (More recent discoveries have shown that hepcidin directly reacts with ferroportin, a protein that transports iron out of cells that store it.)

Diagnosis

The simple blood tests for ferritin and transferrin can be carried out in the doctor's surgery. Ferritin measures the iron in your blood and Transferrin Saturation measures the iron carried in your body. If these tests are above the normal level, a genetic test should be carried out.

Family Screening is recommended for:

First degree relative of confirmed case of haemochromatosis.
Children of confirmed case of haemochromatosis.
Genetic testing of the spouse: If negative no need to screen the children; if carrier (C282Y or H63D) further genetic testing of children is indicated in those over the age of 20.

Treatment

Phlebotomy is the cornerstone of treatment and has been around since the middle ages. It is the removal of blood, similar to blood donation. Each unit of blood (500cc) contains 0.25 grams of iron. Phlebotomy should be carried out weekly until the serum ferritin falls below 50mg/l.
Once this goal has been reached, maintenance phlebotomy must be tailored to suit the individual patient by six monthly monitoring of fasting serum transferrin and serum ferritin. The haemoglobin should be checked before each phlebotomy and phlebotomy deferred if level is <11g/dl.
Prior to the commencement of therapeutic phlebotomy, some patients may require liver biopsy to estimate the degree of fibrosis or cirrhosis.

Who needs a biopsy?

People aged over 40 with abnormal liver enzymes or Ferritin over 1000 mg/l.
Rules are meant for interpretation and discretion is appropriate in recommending liver biopsy on the basis of age alone.

Other Types of HH

The majority of cases of HH are caused by mutations in the HFE gene located on chromosome 6. This disorder is termed type 1 or HFE associated HH.
There are other rare forms of Haemochromatosis not related to the HFE gene. The four main forms are caused by mutations in the genes hemojuvelin, hepcidin, transferrin receptor 2(TfR2ferroportin) and ferroportin.
Juvenille (JH) or type 2 Haemochromatosis is a more severe form of Haemochromatosis with an earlier age of onset than HFE-HH. Generally patients with JH present with clinical symptoms before thirty years of age and often in teen-age years. It is very important to diagnose and treat at an early age.

Take Home message

In Ireland HH is relatively common, the prognosis is excellent and treatment is relatively straightforward. New 'molecular' discoveries and ongoing research will help. It is important to enjoy life. Eat a balanced diet and avoid exclusionary iron-free diets. Take alcohol in moderation and no iron or vitamin C supplements.

Colm Kavanagh and Mike McDonagh at the Corrib Shopping Centre, Galway.

Anne Hannan in Manor West SC, Tralee