SPRING | 2010

Canadian HEMOCHROMATOSIS SOCIETY

Iron Filings

Providing information, awareness and support

OUT OF THE BLUE

BY PATRICK HANEY

My experience with hereditary hemochromatosis began the month before my 33rd birthday. In August 2009, despite feeling generally healthy I was declined extended disability insurance as a result of some routine blood analysis the insurance company completed. Essentially, my liver enzymes were elevated.

"Out of all the symptoms that could be attributed to HH, I only ever experienced joint pain in the thumb and forefinger of both my hands."

My liver enzymes were elevated? It was a surprise for sure, so a trip to my general physician ensued where he spent about six weeks sending me for numerous blood tests trying to rule out any major infectious diseases. In the end, his biggest concern was hepatitis A or B, which didn't make sense to me, but was the most logical explanation about my liver enzymes. He also lined me up for an abdominal ultrasound which I was able to get for mid-October and produced more questions as it showed an enlarged liver and spleen and enlarged lymph nodes in my abdominal cavity.

I soon had a referral to a gastroenterologist, which took another six weeks so there was more waiting and wondering. Life went on as usual, but I did stop drinking because we knew my liver was compromised in some way. The gastroenterologist I saw at the end of November was straightforward. He asked me about joint pain, ordered a few more lab tests, and then tested my ferritin. This was the first time I heard about ferritin.

My wife was away at a work trip the day my lab results arrived in the mail. My ferritin result came back off the charts with the first number high in the 9000s. Before she was home that night, I spent some time with "Dr. Google" and realized the gravity of my situation. My wife came in the door and I said "I think I know what I have". It was called hereditary hemochromatosis (HH).

Because of my enlarged nodes and the extreme level of ferritin, the specialist wasn't convinced it was just HH, but could also be leukemia or lymphoma. Before I knew it, I was ordered for an urgent CT scan. I was left with a message of impending doom right before the Christmas holidays along with waiting

on the genetic test results for HH, which I would not receive until early in 2010. Furthermore, no one in our family has ever been diagnosed with HH, at least that I was aware of, which made the doctors even more skeptical it was solely HH.

I received a positive genetic test for HH (homozygous C282Y) in mid-January. I was referred to a hematologist, who ruled out the additional scare of lymphoma and got me started on a weekly phlebotomy regime in February which will continue as long as my hemoglobin stays reasonable and my ferritin levels are lowered to a target of 50.

Even with my out of the ordinary lab results, it still took five months to get my diagnosis with HH. I am a young, healthy man with no other medical problems. I did not have unusual abdominal pain or fatigue. Out of all the symptoms that could be attributed to HH, I only ever experienced joint pain in the thumb and forefinger of both my hands. However, I had gone to the doctor about this four years ago, which he and I both chalked up to repetitive stress from daily computer work. The doctor suggested I wear wrist guards, which I have been religiously wearing since and have helped reduce my wrist and hand pain, but not eliminated it. My ferritin level continues to be high (in the high 5000s) and I have a number of appointments lined up with specialists (including an MRI scan and echocardiogram) to help assess the extent of damage my iron overload has had on my system.

The purpose of my story is that had I not had the test from my insurance company, I am sure I would have suffered more severely from HH within the next decade. I now encourage my friends and acquaintances to have their ferritin tested by their doctors whether they have HH in their family or not particularly if they have any of the following symptoms:

- Arthritis, especially in their hands
- Chronic fatigue
- Loss of sex drive (libido) or impotence
- Lack of normal menstruation (amenorrhea)
- Abdominal pain
- High blood sugar levels
- Low thyroid function (hypothyroidism)
- Abnormal liver function tests, even if no other symptoms are present

Transferrin saturation percentage (TSAT) is also an important diagnostic that if elevated, is a good indicator that HH is present.

I've been told that I must be the quarterback of my own health and stay on top of my treatment and conditions related to HH. I've pushed to see not only a hematologist, but also other specialists like cardiologists, gastroenterologists and so on because HH is a multi-system disorder especially depending on the severity of its impact when diagnosed.

I remain hopeful that I will successfully work through my treatment, lower my ferritin, manage any associated conditions and live my life to the fullest, despite my HH. I wish the best to those of you similarly affected with HH as well.

Medical research indicates that end organ damage begins at a when serum ferritin is in excess of 1000ug/mL

UPCOMING EVENTS

May Awareness 2010

While the mission of the Canadian Hemochromatosis Society is to promote awareness of hemochromatosis all year round, May is a special month when the society ramps up its publicity campaign in an effort to inform the entire nation about Canada's most common genetic disorder.

During the month of May CHS encourages the mayors of Canadian cities and the provincial Health Ministers to proclaim the need to become aware of hemochromatosis, the dangers of iron overloading, and the need for more Canadians to be tested.

The tradition of Hemochromatosis Awareness Month began in 1987, to honour CHS's feat in helping to identify 523 new diagnoses. In some families, as many as three or more hemochromatosis sufferers were found. In one extreme case, nine members from one family were identified with the disorder. However, despite the ongoing effort to increase the awareness of hemochromatosis, CHS calculates there are over 125,000 Canadians suffering from hemochromatosis, most of them completely unaware that they even have the disorder. Too many hemochromatosis patients will go untested, undiagnosed, and untreated unless the awareness is raised.

During the awareness campaign in May, CHS coordinates interviews with radio, television, and newspapers across the nation to raise awareness about hemochromatosis in Canada. CHS volunteers are also fully mobilized to distribute print packages and electronic information, as well as setting up information booths in malls, hospitals, pharmacies, and blood banks in every province. Last year the society organized seventeen significant activities and events in five provinces, and this year there will again be many opportunities for you and your family to join the campaign. For this year's May Awareness Month schedule, please refer to the CHS website at www.toomuchiron.ca/May2010

Canadians Need a Guideline for Diagnosing Hemochromatosis in Every Province

A medical guideline or protocol is a document that instructs the criteria, management, and treatment of a specific medical condition; it provides recommendations to doctors and plays a critical role in assisting their diagnostic decision. A protocol is especially important for disorders that are vague in their symptoms, such as hemochromatosis. It is astonishing that only two provinces, Alberta and British Columbia, have established guidelines and protocols for hereditary iron overload, the most common genetic disorder in Canada. In the rest of the country, doctors have only their personal knowledge to rely on. At the CHS we hear time and time again stories of HH patients being diagnosed with anemia and receiving treatments that will only make their conditions worse.

The protocols in Alberta and British Columbia are more or less the same. They outline the differences between hereditary and acquired iron overload. The former is the typical form of hemochromatosis which is caused by mutations in the HFE gene, while the latter relates to either iron loading anemia or transfusion iron overload.

The protocols then give a total of five recommendations, the first of which deals with the symptoms and advises that patients should be tested for iron overload if they are experiencing arthritis, congestive heart failure or cardiomyopathy, adult-onset diabetes, persistent elevation of liver enzymes or cirrhosis, secondary hypogonadism, increase skin pigmentation. Patients with persistently elevated serum ferritin not explained by an underlying inflammatory disease are recommended to also be tested.

Recommendations two through four all deal with testing and follow-up. They first advise doctors to use either transferrin saturation or saturation of total iron binding capacity to test for iron overload on a fasting blood sample. If the "fasting transferrin saturation" (fTS) is below 0.45, no further testing is required. If fTS is between 0.45 and 0.60, repeat test within a month, and if fTS is still above 0.45, DNA testing for HFE mutations should be considered. Also, if fTS is ever above 0.60, DNA testing is strongly advised.

Recommendation three further specifies that persons of European descent who have one or more first-degree relatives with a confirmed or presumptive diagnosis of HH should also receive DNA testing.

Recommendation four is a follow-up on the DNA testing. There are three recognized mutation patterns in the HFE gene. The first is the C282Y/C282Y combination (one C282Y mutation from each parent), which causes the most common form of hereditary hemochromatosis. The second possibility is a C282Y/H63D combination, where H63D is a less severe mutation in the HFE gene and has a less than three percent chance of developing iron overload. Some people may also have only one C282Y mutation or a C282Y/normal combination. In this case the risk of developing iron overload is even lower than three percent and further ferritin testing is not required.

Finally, recommendation five specifies treatments for hemochromatosis patients. This section emphasizes that the volume and frequency of phlebotomy need to be individualized according to the patient's age and clinical circumstances, and that ferritin level should be monitored regularly to assess response to therapy. Once patients have been successfully depleted of excess iron stores, a program of maintenance therapy should be established and any end organ damage should be reassessed periodically.

The Canadian Hemochromatosis Society is willing to work with doctors in every province to formulate and reach approval for a guideline to diagnose hemochromatosis.

Ross Gilley, Outgoing CHS President

Ross Gilley, President

After two years of dedicated service, Ross Gilley, the current President of the Canadian Hemochromatosis Society (CHS), is about to pass on his presidency to CHS's Vice President, Frank Erschen. Ross was first introduced to the CHS in 2002 after his father was diagnosed with liver cancer. Ross was subsequently diagnosed with hemochromatosis and has been intimately involved with the society's work ever since.

When asked about the most important accomplishment of the society during the past two years, Ross replied that it is the improvement in spreading the awareness for iron overload. "I have seen a real progress in the society's work as we lengthened and improved the lives of more and more hemochromatosis patients. This could only have been accomplished through improved distribution of information and an increase in awareness."

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Ross Gilley notes that more Canadians are getting tested for hemochromatosis every year, not because they have developed symptoms themselves, but because one of their close relatives has been diagnosed with the disorder. "This increase in testing is a direct result of the society's effort to educate the public about hemochromatosis. It is, after all, the most common genetic disorder of the country, and we take real pride in knowing we at the CHS are improving the lives of thousands of Canadians. "Gilley attributes much of this success to the work of Bob Rogers, the Executive Director of CHS, and the growing team of volunteers across the country. All have been doing wonderful work in recent years keeping the society on track with the members' needs".

When asked to suggest future improvements for CHS, Ross commented that while provincial governments are starting to get the message about hemochromatosis, much more work remains ahead. "Governments understand dollars and cents. We need to help them realize that early detection of hemochromatosis reduces acute demands on our medical system later in life. When a patient's iron level is pro-actively kept under control with regular phlebotomies, we as a society don't need to spend thousands of dollars for liver transplants and joint replacements in the future. In partnership with governments, we have an exciting opportunity to positively impact both individual lives and our fiscal reality. That's a powerful package of benefits."

Gold Medal Given to Hemochromatosis Expert

As the Canadian athletes broke the Winter Olympics record in gold medal count, Dr. Paul Adams was also earning a gold medal for his research in hemochromatosis.

Dr. Paul Adams of the Lawson Health Research Institute in London, Ontario has dedicated his professional life to the diagnosis and treatment of iron overload. This February during the 2010 Canadian Digestive Diseases Week in Toronto, Adams was awarded the 2010 Gold Medal for excellence from a leading foundation in Canada for his continual research in hemochromatosis, which encompasses the identification of the fundamental genetic determinants of the disorder and several effective new treatments.

Nearly a decade ago, Adams received a $34 million dollar grant from the National Institutes of Health in the United States to conduct research investigating the prevalence of hemochromatosis in a multi-ethnic population, which helped determine the optimal use of diagnostic testing such as genetic testing. Nowadays Adams has established himself as an international authority on iron overload; his innovative research has not only contributed to the standardization of patient care for the diagnosis and treatment of hemochromatosis, but has also made an impact on genetic testing for all medical diseases.

Introducing CHS's New President: Frank Erschen

Frank Erschen

This year Frank Erschen, the Vice President of the Canadian Hemochromatosis Society, will become the new President for 2010-2012. Similar to past Presidents of the CHS, Erschen is an insightful leader who believes passionately in the cause. Erschen has a tremendous respect for his predecessors and aims to build on their success while implementing progressive plans to lead CHS to new heights in the future.

Ten years ago, like the majority of his fellow Torontonians, Frank was oblivious to hemochromatosis. This changed, however, in the early 2000s, when he was diagnosed with the genetic disorder. Frank became involved with CHS when his iron level peaked in 2007 and very soon after was invited to join the Board due to his impressive corporate, governance and community experience. For most of the past twenty-nine years, Erschen has worked in various roles at prominent corporations including IBM and the BMO Financial Group, where he retired as an executive in 2006. He has chaired or been an active participant in many fundraisers for charities in Canada and the U.S. These experiences allowed him to contribute greatly to the operation and governance of CHS. In 2008 Frank Erschen became the Vice President of CHS.

"I am truly excited and honoured with the opportunity to lead CHS through its next wave of change" said Erschen, who has witnessed a remarkable growth in the society since 2007. "CHS has a rich history of very devoted and insightful leaders, starting with our amazing founder Marie Warder and most recently with the two Presidents I've had the pleasure of working with and supporting: Elizabeth Minish in 2007 and Ross Gilley from 2008 to 2010. Their courage, foresight, and intensity of purpose have distinctively transformed the society on all fronts."

Erschen commented that his most important task as the new President is to balance the 30 year old tradition of the society with the need to strategically evolve. "Our unrelenting tradition of caring is what sustained the organization for the last three decades. However, we also need to focus on new ways to extend our reach." He thinks that maintaining a steady and substantial growth is perhaps the biggest challenge facing CHS at the moment. More specifically, the society's fundraising capabilities must be further strengthened to meet the challenges of growth.

When asked what he would like to accomplish during his presidency, Erschen said that he has three main objectives: achieving the purpose of CHS, fundraising, and continuous improvement. "Priority number one must always be fulfilling why we exist. The President, as with each of the Directors, has a role to play in creating awareness about hemochromatosis - the diagnosis, treatment, and realities of living with the disorder." Secondly, Erschen would like to fully utilize his fundraising expertise and attract more donations to support the society's mission. Last but not least, he also knows that the growth of the society is a continuous process, and that various improvements will have to be made along the way to fine-tune governance and operations. "One of my focus areas will be to continue the evolution of governance processes and the Board. In addition, we'll also focus on our operational framework, our geographical strategies, and our programs."

"I am truly excited and honoured with the opportunity to lead CHS through its next wave of change"

Iron Overload Community Information Session

THE IWK'S MARITIME MEDICAL GENETICS SERVICE IS HOSTING A FREE COMMUNITY INFORMATION SESSION ON HEREDITARY HEMOCHROMATOSIS (IRON OVERLOAD).

Hereditary hemochromatosis is a genetic condition common among people in the Maritime provinces that affects the body's metabolism of iron. Hereditary hemochromatosis is a genetic condition common among people in the Maritime provinces that affects the body's metabolism of iron.

When: **Tuesday, May 18th, 2010, 7PM**

Where: **O.E. Smith Auditorium**
Main Floor, Children's Site,
IWK Health Centre
5850 University Avenue
Halifax, Nova Scotia

THIS MULTI-DISCIPLINARY SESSION WILL INCLUDE PRESENTATIONS BY:

Patricia Steele, Genetic Counselor
(inheritance and genetic testing)

Dr. Sue Robinson, Hematologist
(management and treatment of the condition)

Dr. Kevork Peltekian, Hepatologist
(symptoms and complications caused by increased iron levels)

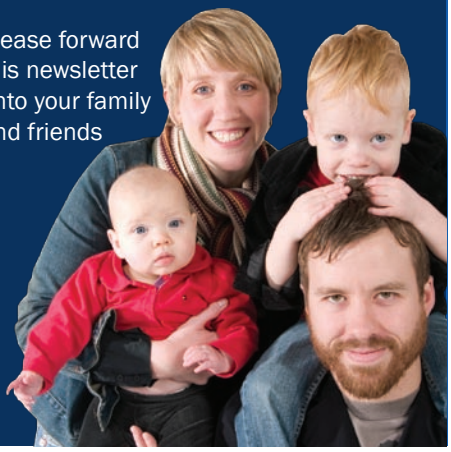
Treatment
Phlebotomy treatments (bloodletting) which are ongoing for life.

Reference reading
The Bronze Killer; Ironic Health; Iron Disorders Institute Guide to Hemochromatosis

Hereditary Hemochromatosis (HHC) is a genetic disorder that affects over 3,000,000 people in Canada.

There is a cure.
Awareness.

Please forward this newsletter onto your family and friends



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Support CHS and help prevent needless suffering and early death

Donation $_____

Annual Membership

- ☐ Regular ____ @ **$30**
- ☐ Senior ____ @ **$20**
- ☐ Family *(same address)* ____ @ **$45**

☐ Yes, please renew my annual membership automatically by using my credit card info

- ☐ Senior's Lifetime (55 +) ____ @ **$295**
- ☐ Lifetime ____ @ **$500**

Books & Accessories

- ☐ The Bronze Killer ____ @ **$20**
- ☐ Ironic Health ____ @ **$22**
- ☐ Wristband ____ @ **$8**
- ☐ Hemochromatosis DVD ____ @ **$21**
- ☐ Too Much Iron DVD ____ @ **$10**

Total $_____

☐ Credit card ☐ Cheque/Money order
(use credit card info area to the right)

- ☐ I have Hemochromatosis (HHC)
- ☐ I have a blood relative with HHC

- ☐ I would like my support acknowledged in the newsletter
- ☐ I would like to receive the E-Newsletter, The Magnet
- ☐ Send me information about planned giving or leaving a bequest in my will.

Contact Information

Name: _____

Address: _____

City: _____ | Province/State _____ | PC/Zip: _____

Tel: _____ | Email: _____

Official Revenue Canada receipts are issued for all memberships and donations.

Donation Options

I would like to make a ☐ Monthly ☐ Quarterly ☐ Annually ☐ Other _____
donation in the amount of $ _____

☐ **Charge my** ☐ VISA ☐ MC ☐ American Express

Card Number: _____ | Expiry date: _____

Signature: _____

☐ **I am enclosing ___ postdated cheques in the amount of $**_____
to the Canadian Hemochromatosis Society.

Please mail this form to:
Canadian Hemochromatosis Society
272 - 7000 Minoru Blvd. Richmond, BC V6Y 3Z5
Tel: 604 279 7135 | Toll Free: 1 877 BAD IRON

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