SPRING 2013

Canadian HEMOCHROMATOSIS SOCIETY
Société canadienne de l'hémochromatose

Iron Filings

Providing information, awareness and support

"Tis healthy to be sick sometimes"
– Henry David Thoreau

By Mary Lennox, Ottawa, Ontario

My story is summed up by this quote. I lived a busy lifestyle as a professional who when asked about my health would often respond, "I am in excellent health!" As a 59 year old woman who had a demanding career that took a great deal of energy, I was always proactive in my wellbeing. I was, and still am, a lifetime member of Weight watchers for the past 16 years. I exercised at least 4 days a week. Walking was my favorite thing. I also began a regime of weights and resistance to fend off osteoporosis. And then I got my "inheritance".

The feeling of being unwell really began during the fall and winter of 2011/12. I felt like I was constantly going to get the flu. I went to my general practitioner and was encouraged to track my symptoms for at least a month. I took my temperature every day and wrote down these vague statements – fatigue; sore muscles; chest congestion; tender abdomen. Winter turned into spring and things improved. I felt like my vibrant self all summer.

Fall began, and the weekend of September 15 and 16, 2012, sticks out clearly in my mind. On Saturday morning of September 15th I began a new exercise class with my sister. One hour of a combination of step, cardio and weights. During the class, I felt like I was struggling a bit to keep up. The next day on Sunday September 16th I woke up a bit sore. But it was Terry Fox Day. Terry Fox - my hero! I have organized the Terry Fox Run for the past 7 years in my village outside of Ottawa. I was up early, setting up the registration and all the details that go with the event. I pushed through knowing I was not quite right. I would rest later.

It took me the entire week to recover. In fact I have not felt healthy since that weekend. As the fall went along I knew the flu-like symptoms I had suffered the previous winter were back. Something was wrong and I was going to get to the bottom of it. In 2006 I had undergone an executive health appointment. In October I decided to book another one to use the data as a second opinion. My doctor (who is just excellent) was tackling my issues one by one. We were considering thyroid or adrenal issues. I needed a look at the big picture for myself. As part of the program for the executive medical, I received a comprehensive report called Preventative Health Assessment. I studied the report from cover to cover and two things jumped out. One – I now had something called Metabolic Syndrome. Two – my ferritin levels had doubled in the past 6 years. I had no clue what either meant. I went to work researching for hours. I kept coming back to one thing: Hemochromatosis – the Celtic Curse.

"This is what I have," I stated to my poor husband one evening. "It all fits!" My heritage matched, as both parents were descendants of the British Isles. I was postmenopausal and the symptoms were similar to the ones I found in my research. I posted an article with respect to the "Celtic Curse" one day on my Facebook page. My first cousin responded to me, saying that she was experiencing iron overload and had been doing blood donations from time to time. That sealed the deal for me mentally. Now how do I get confirmation? I called my doctor's office the next day to report the genetic possibility.

Arrangements were made for the DNA to be completed. I was asked what side of the family this cousin came from and what gender. "Our mothers are sisters." Hmmm. Silence followed. The next day I picked up the requisition for my blood work and went right away to have it completed. While the blood was being drawn, I asked the technician to confirm the tests to me. "We are sending blood to CHEO (Children's Hopital of Eastern Ontario) for DNA testing for hemochromatosis."

"Good. I just learned my cousin has some iron issues," I said.

"Is your cousin a male?"

"No, she is female and our mothers are sisters."

"It is very rare that it's found in women," stated the technician. Today I understand it is a myth.

All during the month of November, I received guidance and valuable information from the

Continued on next page

"Tis healthy to be sick sometimes" - *Continued*

Canadian Hemochromatosis Society through their Facebook page and other correspondence, and finally got to the right tests and the right diagnosis. On December 18th I was given the news that I have a double C282Y mutation, the gene combination that puts me most at risk for iron overload. My ferritin level is 474 ng/mL. My transferrin saturation level is 80%. I am in the early stages of iron overload.

I feel terrible most days. I cannot imagine if this diagnosis had dragged on for years as is often the case. I experienced my first phlebotomy in early February. My first one did not make me feel any better, but it was a simple procedure. I am slated for one every two weeks for 3 months as a start. So it truly is healthy to be sick sometimes. My entire family has been alerted. I can be an advocate for this condition. And most of all – when I get back to myself – I will never take my "excellent health" for granted!

May
National Awareness Month Event Calendar

Several information sessions and awareness events have been planned to raise the profile of hereditary hemochromatosis in Canada. The following is a list of events occurring before, during, and after the month of May. Be sure to come out and attend (or tune into) these and other happenings in your area throughout the year. Your support will provide the momentum to keep events like these running and hemochromatosis awareness spreading.

Friday to Sunday, April 26 to 28
Canadian Home Builders' Association of Northern BC Home Show
Prince George Kin Centre,
2187 Ospika Boulevard
Prince George, British Columbia

The Canadian Hemochromatosis Society is once again hosting a booth at the CHBA Northern BC Home Show. Stop by our booth to visit with others who have hemochromatosis and to learn more about the disorder, including screening, diagnosis, treatment and management.

Monday, April 29 @ 7 pm
Hemochromatosis Information Seminar
Prince George Civic Centre
808 Civic Plaza
Prince George, British Columbia
Room: 201

Sunday, May 5
Radio Interview: Dr. Joe Schwarcz Show – The Right Chemistry
CJAD 800 Montreal
Approximate air time: 3:30 pm EDT
Listen live at www.cjad.com

Wednesday, May 8 @ 7 pm
Hemochromatosis Information Seminar
Future Inns Moncton Hotel and Conference Centre
40 Lady Ada Blvd
Moncton, New Brunswick
Room: Fundy

Saturday, May 11 @ 11 am
Hemochromatosis Information Seminar
Best Western Hotel Charlottetown
238 Grafton Street
Charlottetown, Prince Edward Island
Room: Stanhope A
Continental breakfast served

Tuesday, May 14 @ 7 pm
Hemochromatosis Information Seminar
Grand Lake Fire Hall
793 Grand Lake Road
(beside the Mayflower Mall)
Sydney, Nova Scotia

Wednesday, May 15 @ 7 pm
Hemochromatosis Information Seminar
Sydney Mines Old Post Office Building
2 Fraser Avenue
(corner of Main Street & Fraser Avenue)
Sydney Mines, Nova Scotia

Wednesday May 22 @ 7 pm
Hemochromatosis Information Seminar
Hilton Garden Inn Halifax Airport
200 Pratt & Whitney Drive,
Room: Cape Breton
Enfield, Nova Scotia

More radio show interviews are being planned for the month of May, while July, August and September will see CHS bring information sessions to more communities in British Columbia and Alberta. Watch our website www.toomuchiron.ca, Facebook page www.facebook.com/TooMuchIron and Linked in Group (http://linkd.in/X12vnz or search for Canadian Hemochromatosis Society while in Linked in) for all event updates.

Volunteers will be sending out Letters to the Editor in their local papers to promote hemochromatosis awareness during May Awareness Month. If you would like a template of a letter to send to your local newspaper, please email office@toomuchiron.ca with your request.

Let Me Carry Your Voice Into Talks with Governments & Colleges

Bob Rogers,
Canadian Hemochromatosis Society - Executive Director and CEO

In our Fall 2012 newsletter, I shared a few examples of how a lack of sound medical practice from family physicians is contributing to the frequent misdiagnosis and underdiagnoses of hereditary hemochromatosis. This may be due to a lack of initial and continuing medical education about this disorder. I am informed that medical students receive a mere 30 minutes of education on hemochromatosis during their studies at university. Specialists may assume family doctors are knowledgeable, but they seldom are. Only two provinces in Canada, Alberta and British Columbia, have provincial health approved best practice medical guidelines (protocols) published to assist family doctors in this area of medicine. The Foundation for Medical Practice Education at McMaster University in Ontario has produced an excellent continuing medical education module accredited for family physicians, yet the Foundation only serves a small subset of family doctors in Canada.

Recently, at a community health fair, I met Jeff who was diagnosed at age 50 with a serum ferritin of over 4000 µg/L. Due to his late diagnosis, he has a compromised liver, type 2 diabetes, severe depression and mood swings, bronze discolouration of his skin and considerable weight loss. He doesn't consume alcohol, preferring tea, yet during the long arduous evaluation period for his liver condition he was placed on an eight month watch to assess if he was an alcoholic. At 54, he is living in a care home because he is unable to care for himself. Unbelievable and unnecessary!

I have had conversations with health authorities recently in Prince Edward Island, Nova Scotia and Newfoundland to discuss heading up committees to ensure protocols for hemochromatosis are produced and sanctioned by the health authorities. Yet several months later, and in one case several years, no protocols exist in these provinces. Why? How many more "Jeffs" do there need to be to have this disorder placed on the minds of practicing family physicians?

I will be in Ontario and the Maritimes this May and I will be asking many tough questions with the aim to getting an agreement that more action needs to be done to raise more awareness and to further educate and assist family practice medical practitioners about hereditary hemochromatosis. I am interested in hearing our readers' diagnosis experiences. Drop me a line so that I can carry your voice into future discussions with government, colleges and specialists.

DUCA Supports Hemochromatosis Community

The Canadian Hemochromatosis Society is pleased to announce its new corporate sponsor: DUCA Financial Services Credit Union.

DUCA has a strong history of providing help for those in need. In fact, this is how the credit union took shape. In 1954, a number of Dutch immigrants, who were unable to access funds from large financial institutions, came together. Today DUCA serves over 41,000 members throughout the Greater Toronto Area and holds $1.3 billion in assets, making it one of the largest credit unions in the province. DUCA is open to all residents of Ontario.

DUCA is committed to the communities it serves. In 2012 alone, DUCA donated over $437,000 to a wide range of causes that focus on numerous issues, including health and long term care, educational programs, international development, economic development and housing, environmental causes and disaster relief.

"At DUCA, our goal is not only to help our members achieve financial security but also to support the communities in which they live. Supporting organizations like the Canadian Hemochromatosis Society is a fundamental part of who we are. It all comes back to the cooperative values that are at the core of our business," says Richard Senechal, President and CEO of DUCA.

In addition to DUCA's charitable giving, they also return a percentage of profits to members through a unique profit sharing program. Since the inception of this program back in 1999, DUCA has shared over $68 million in profits with members. It's one of the ways DUCA proves they are "not your average bank".

"We are happy to have the strong support of DUCA in Ontario," says Bob Rogers, Executive Director of the Canadian Hemochromatosis Society. "Both of our organizations believe in creating and maintaining strong, healthy communities, and it's a pleasure to work toward this goal together."

More information on DUCA can be found by visiting www.duca.com.

Richard Senechal, President and CEO of DUCA.

DUCA
Come bank with us.

Focus on the Patient

Q and A with Dr. Paul Adams

We speak with Dr. Paul Adams, a Professor of Medicine and Chief of Gastroenterology at the University of Western Ontario and practicing gastroenterologist at University Hospital in London, Ontario, for his view on treating hemochromatosis and where he thinks future research is headed.

Q. What screening test do you use for screening a patient?

A. If I am screening a clinic patient or family member, I use transferrin saturation, serum ferritin, *HFE* genotyping and clinical judgement.

Q. Not all homozygotes or heterozygotes of the *HFE* gene mutations express signs of iron overload. Have there been any findings that link modifier genes (genes that modify the effects produced by other genes) or environmental factors to the manifestation of iron overload?

A. This is an area of ongoing research. The HEIRS (Hemochromatosis and Iron Overload Screening) team has had several genetic research studies in this area and is currently using exome sequencing [selectively sequencing the short, functionally important regions of the genome] to look for new modifying genes.

Q. You were a principal investigator in the HEIRS Study. Can you tell me more about it?

A. The HEIRS Study was a large population based study that screened over 100,000 participants for iron overload. The participants were racially diverse and participants began with a survey, a genetic test, serum ferritin and transferrin saturation blood test. Participants with elevated iron tests or a positive genetic test (C282Y homozygote) were called back for a more detailed clinical examination. The study reported on the prevalence of the genetic mutations across different racial groups, the biochemical expression of the disease in patients with the typical genetic test and also on many aspects of the ethical, legal and social implications of genetic testing.

Q. What are some studies that have sprouted from data and results of the HEIRS study?

A. The HEIRS study group continues to use the data to study hemochromatosis but also other diseases. An HEIRS study showed that there was no correlation between dietary intake of iron and serum ferritin. A research paper on celiac disease has been completed and we have done another genetic study in iron deficiency.

Q. That's interesting. In the case of iron overload, serum ferritin is an indirect measurement of the amount of iron in the body. Please explain how iron introduced into the body through diet does not increase ferritin levels.

A. You do absorb the excess iron from food but we were unable to show a difference in those that have a high iron diet and those with a low iron diet. In Sweden, they have removed iron fortification of food with a very slight decrease in the serum ferritin of the population.

Q. How have the results from the HEIRS Study affected how you treat *HFE*-linked hemochromatosis sufferers?

A. I am opting not to recommend phlebotomy for patients with mild elevations in ferritin or in those who do not tolerate the treatment.

Q. What do you consider as "mild elevations"?

A. A mild elevation in ferritin would be less than 500 µg/L . If we had used the HEIRS study to determine normal ranges it would have been up to 550 µg/L in men.

Q. There is anecdotal evidence that sufferers with elevated ferritin levels below 1000 µg/L experience physical and emotional symptoms consistent with iron overload at even higher levels, and end up being unable to work or live vibrantly. After diagnosis and phlebotomy treatment, they report being well again and go back to contributing to their family and community. In terms of the positive impact on a patient's overall well-being, why would you not recommend treatment to individuals with mild elevations?

A. We do recommend phlebotomies to any C282Y homozygote with an elevated ferritin. When we have studied quality of life after phlebotomy we have not been able to show a consistent benefit. There are patients who report a worse quality of life or worsening of arthritis. If this occurs, we stop treatment in mild cases.

Q. How many hemochromatosis patients do you see in a week?

A. I see about 25 patients a week that think they may have hemochromatosis. Usually 1 or 2 in the group will have the typical genetic test. A common misconception is that an elevation in serum ferritin always means iron overload. In the general population, obesity, daily alcohol consumption and inflammation are more common causes of a ferritin elevation than iron overload.

Q. At what stage of iron overload do most sufferers get referred to you?

A. The average patient is in their 5th decade. In the context of a family study, we see younger patients.

Q. What is the worst case of iron overload that you have seen in an individual?

A. The worst complication of hemochromatosis is cirrhosis with liver cancer. This is uncommon. Some of the most severe cases of iron overload I have seen are juvenile hemochromatosis with a new genetic mutation in hemojuvelin. This gene was discovered by a research team based in Vancouver.

Q. What current research into the treatment of hemochromatosis do you think shows the most promise?

A. Advances in genetic research tools will soon allow us to analyze all of the genes in one patient for under $1000. This whole genomic sequencing could lead to the discovery of new genes that influence the natural history of hemochromatosis.

Q. Any words of advice for those recently diagnosed with hereditary hemochromatosis?

A. It is a mistake to attribute every symptom you have from head to toe to a variation in your genes. Don't always assume that it is a bad diagnosis. There is some data suggesting that hemochromatosis patients have lower cholesterol and may have a better long term survival than the general population.

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Among Ourselves

This column appears regularly in every issue of Iron Filings and features stories about our dedicated volunteers.

SALUTE TO ONTARIO'S VOLUNTEERS

As a national non-profit organization, volunteers help the Canadian Hemochromatosis Society reach far and wide into areas of Canada with its message of awareness, information and support. This issue's column is dedicated to all our Ontario volunteers, past, present and future.

In 2001, Marjorie Lounder was one of CHS' first regional organizers, then called support group coordinators, located in Ottawa. Her husband Jim had suffered for twenty years from symptoms and diseases unknowingly related to hemochromatosis: diabetes, arthritis, abdominal pain, confusion, mood swings and chronic fatigue, and underwent two hip replacement surgeries. He died in 1997, with autopsy reports showing hemochromatosis as the cause. Marjorie's response to Jim's premature death and her loss, preventable with "a simple blood test done years ago" was to start a local support group, to spread awareness about the disorder and prevent the "scourge of hemochromatosis" from devastating families. Marjorie also became a member of CHS' volunteer Board of Directors. For well over a decade, Marjorie and her group, including Elaine Robinson, actively served their community, hosting information booths, holding support group meetings and recruiting speakers. We are grateful for all the work Marjorie and her group have done to represent CHS and spread awareness of hemochromatosis.

When CHS' online forum began in November 2005, Kingston's Bob McLeod was active on the forum, sharing his experiences and information. Not long after, Bob was appointed as the forum moderator where he did tremendous work, providing supportive answers and information to the online questions, and ensuring spam was kept off the forum. We were sad to see Bob resign from the position in 2011 due to family and work pressures.

From Brampton, Linda Perkins donated much of her time to getting CHS' public service announcements aired on several radio stations across Ontario and Canada during May Awareness Month in 2008 and 2009. She also filled the role of area contact in 2004 and 2005.

In Toronto, strong CHS supporters Lynn and Allan Day take time out from their busy lives to coordinate and even sponsor special events such as the Toronto Network Event in February 2012, plan fundraisers, and host volunteer meetings. Lynn explains what her involvement with CHS means to her by saying, "Being a volunteer with CHS has increased my personal awareness and better my understanding of the disorder. This has been a big help for me to increase the awareness of friends and family who may also be afflicted."

Also hailing from Toronto is Past President and Past Chair of CHS' Board of Directors, Frank Erschen. Frank continues to provide his time and expertise to help direct the Society's activities towards its mission and goals. Kay Easun is another Torontonian who ran a local support group and still continues to recruit volunteers, help at special events and plan awareness-raising activities. CHS has also recruited a new volunteer who is dedicated to helping CHS open and operate a branch office in Toronto this year.

Anna Kyle, Gloria Haché and Diane Jodouin are some more caring Ontarians who have given their time to the cause. Of course we can't possibly mention every volunteer that CHS has ever had in Ontario, there just isn't the room! What we can say is that every single committed volunteer gives CHS the strength required to reach all Canadians at risk for hemochromatosis. CHS salutes you.

As you can see, there are many ways that you can volunteer. We are currently looking for a Regional Organizer for our Toronto Chapter to assist Kay Easun. If you are interested in filling this role, or would like to organize a Chapter in your community, join an existing Chapter, or find out more about CHS volunteer opportunities, check out our website at www.toomuchiron.ca, send an email to program@toomuchiron.ca, or call us at 1-877-BAD-IRON (1-877-223-4766).

CHS Snapshot

The newly formed Montreal Chapter organized an information session held last November 20th. Pictured here are volunteers Mike Sung, Mark Johns, Regional Organizer Juliana Pavelka-Johnston, Tom Tian and CHS President Patrick Haney. Patrick presented the information to a receptive audience that included patients, students, a researcher looking at a possible predictor for hemochromatosis, and two rheumatologists. Kudos to all involved!

Special Acknowledgements

The Canadian Hemochromatosis Society gratefully acknowledges our current Partners and Corporate Sponsors:

Genetrack Biolabs Inc.

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Canadian Blood Services

Province of British Columbia

Government of Alberta

DNA 101: Research into past, present and future

By June Wong, PhD, Vice President, Laboratory Operations, Genetrack Biolabs Inc.

Hereditary hemochromatosis is a deadly but treatable disease which is found predominantly in individuals of European descent. The gene mutation most commonly associated with hemochromatosis is C282Y. Almost 85 to 90% of all individuals who are clinically affected with hemochromatosis test positive for the C282Y gene mutation, and studies have shown that in non-clinically diagnosed individuals, 10 to 33% of individuals who test positive for 2 copies of the C282Y mutation eventually go on to experience the morbidity associated with hereditary hemochromatosis.

In the Fall 2012 issue of Iron Filings, an article discussed the historic hemochromatosis connection to the French. The strong genetic association of this disease with the C282Y mutation, and the striking prevalence of this mutation in individuals of European ancestry has led researchers to investigate the history of C282Y, including the location in Europe where it first originated and how this mutation eventually spread throughout Europe. To tackle this study, scientists have conducted large scale screenings for the C282Y mutation in various indigenous populations throughout Europe in an effort to pinpoint the population distribution pattern of C282Y and retrace its mode of spread.

To follow is a table summarizing the distribution frequencies of C282Y in indigenous populations in various regions of Europe:

Country	# individuals screened	C282Y allele frequency (%)
Austria	758	4.6
Bulgaria	100	0
Czech Republic	239	4.8
Denmark	11,902	5.7
Estonia	442	3.5
Faeroe Islands	387	6.6
Finland (North)	173	5.2
Finland (East)	1150	3.4
France (Brittany)	8726	7.7
France (South)	353	2.6
Germany	919	4.2
Greece	297	1.0
Greenland	200	2.3
Hungary	1721	3.4
Iceland	321	5.1
Ireland	663	9.7
Italy	3158	1.7
Norway	2138	6.6
Poland	871	3.1
Portugal (North/Central)	259	5.2
Portugal (South)	381	2.2
Spain	1342	3.1
Sweden (Sami People)	151	2.0
Sweden (Umea)	206	7.5
Switzerland	189	3.7
UK (England)	368	6.0
UK (N.E. England)	117	7.7
UK (Jersey Islands)	411	8.3
UK (Orkney Islands)	103	4.9
UK (Scotland)	184	8.4
UK (Wales)	11,180	8.2

The population studies conducted to date show a very interesting and distinct distribution pattern for the C282Y marker in Europe. The highest frequencies of this mutation are found along the coastline of Europe. Furthermore, the frequency of C282Y is highest in Western Europe and declines significantly moving east. Also, the frequency is highest in Northern Europe and declines significantly moving south.

DNA studies have also shown that the C282Y mutation likely first arose in Europe approximately 60 to 70 generations ago. Assuming that each generation is 20 to 25 years, that would place the origin of this mutation to approximately 1200 to 1750 years ago, with subsequent rapid spread along the coastline of Europe and disseminating from Northern and Western Europe. The timing and pattern of spread of this mutation has a very close correlation to the migration of Vikings and locations of Viking settlements in Europe beginning around the same time, resulting in the "Viking hypothesis" for the C282Y marker which suggests that the C282Y mutation is associated with Vikings.

Further studies to investigate the "Viking Hypothesis" found that C282Y is found in greatest frequencies in regions that are known to be Viking settlements. For example C282Y is found in high frequencies in the Scandinavian countries including Iceland and the Faeroe Island, which are known to be colonized by Vikings. Further support of the Viking Hypothesis was indicated by studies which showed that the lowest frequencies of C282Y were found in regions of Europe that were not affected by Vikings such as Central Europe, the Balkans, the Mediterranean countries and Russia.

The genetic studies to date suggest that the C282Y mutation first arose in Europe and was spread by Vikings and later by the Normans throughout Europe, indicating a genetic link between hereditary hemochromatosis and Viking ancestry. Moving forward, ongoing studies in the research community will focus on forensic DNA testing of Viking skeletons to obtain definitive proof of the origins of the C282Y mutations and solidifying a Viking link to this prominent European genetic disease.

To order a hemochromatosis DNA test or for more information regarding *HFE* gene testing, go to www.hemochromatosisdna.org or contact the Canadian Hemochromatosis Society.

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CHEO Genetics Clinic: A Look at Genetic Services Related to Hemochromatosis

By Shawna Morrison, MS, CGC, Certified Genetic Counsellor, CHEO | Program manager of the Genetics Education Centre – Knowledge for Ontario (GEC-KO)

Located in Ottawa, the Regional Genetics Program at the Children's Hospital of Eastern Ontario (CHEO) is one of several centres in Ontario that provides genetic assessment, diagnosis, counselling and testing. The CHEO Genetics Program mainly serves Eastern Ontario and Western Quebec. More information can be found on our website www.cheo.on.ca/en/genetics.

HOW DO INDIVIDUALS GET REFERRED TO THE GENETICS CLINIC AT CHEO?

Individuals are generally referred to the Genetics Clinic by their primary care provider but can also be referred by a specialist such as a haematologist, hepatologist or gastroenterologist. Referrals come mainly from Ottawa and Gatineau, and as far out as Cornwall, Hawkesbury and Renfrew. Although we are situated at a Children's Hospital, we see patients of all ages.

A referral to the Genetics Clinic is usually initiated when an individual:

- has received a genetic test result which shows the individual has an inherited predisposition to hereditary hemochromatosis (their genetic test result shows that they carry two gene changes, one in each copy of the *HFE* gene) or
- has received a genetic test result which shows the individual is a carrier of hemochromatosis (their genetic test result shows they carry a gene change in one *HFE* gene) or
- is considering whether or not they would like to have genetic testing based on a family history of hemochromatosis or on biochemical evidence* of iron overload. In general, genetic testing would be offered in the following scenarios:
  - Any individual with biochemical evidence* of iron overload and/or in whom clinical suspicion is high

    *This means blood work iron indices have been persistently high, demonstrating a high serum ferritin and a high transferrin saturation
  - An asymptomatic (no symptoms), adult, first-degree relative (sibling, parent or child) of an individual identified to have one of the following genetic test results:

    a. C282Y/C282Y
    b. C282Y/H63D
    c. C282Y/S65C
    d. C282Y carrier

WHAT HAPPENS AT A GENETIC COUNSELLING APPOINTMENT?

Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease (National Society of Genetic Counselors, 2005).

If an individual is referred to the Genetics Clinic at CHEO specifically for genetic counselling (ie. the referring provider simply wants the patient to be provided with information about the condition) he/she can expect to attend a group information session delivered by a professional with a Masters degree in Genetic Counselling. The group information session was developed in response to the very high numbers of referrals for a personal and/or family history of hereditary hemochromatosis received in our clinic. (This in turn is due to the high frequency of hemochromatosis mutations in Caucasians of Northern European origins). To best serve the patient population in as timely a manner as possible, we feel this is an efficient and effective model. Immediately following the group information session, patients are provided with the opportunity to meet with the genetic counsellor individually, so that they

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While women are less likely to develop iron overload prior to menopause (presumably due to iron loss associated with menstruation and increased iron utilization associated with pregnancy), they are EQUALLY as likely to inherit the genetic predisposition to hemochromatosis and EQUALLY as likely to pass on a gene mutation as men.

Hemochromatosis is a condition that will not present until adulthood. Testing in childhood does not change medical management. In accordance with the recommendations of medical specialty societies in Canada and the U.S., testing of children should be deferred until the child is competent to decide whether they want the information.

IRONS for IRON

Canadian HEMOCHROMATOSIS SOCIETY
Société canadienne de l'hémochromatose

Take part in one of three inaugural golf tournaments being held this May in support of CHS' life-saving programs. Registration is $200 per person, and includes the green fee and dinner. Tax receipts will be issued for the portion supporting CHS. Space is limited, so sign up early for a great round of golf and help raise awareness of hemochromatosis! For more details and registration information, visit www.toomuchiron.ca, email office@toomuchiron.ca or call toll free 1 877 BAD IRON (1 877 223 4766).

Irons for Iron Vancouver
Thursday, May 16, 2013
Quilchena Golf & Country Club

Irons for Iron Calgary
Thursday, May 30, 2013
Speargrass Golf Course

Irons for Iron Cape Breton Island
Monday, May 20, 2013
The Lakes Golf Club

CHEO Genetics Clinic - *Continued*

may ask questions pertinent to their own family and make a choice about testing (if it has not already been performed) in a confidential setting. They are also invited to call their counsellor at any time after the session, as necessary. They receive a detailed letter summarizing the information presented in the group session as well as information and interpretation specific to their situation and reason for referral. We have solicited patient feedback in the form of a questionnaire to confirm that patients are satisfied with the group format.

In the group information session, patients learn about:

- Hemochromatosis – what it is, the signs and symptoms of untreated iron overload
- the inheritance – basic genetics, how hemochromatosis runs in families and how it relates to them
- what tests are available – iron indices versus genetic testing
- the different genetic test results and what they mean
- how testing is arranged
- to whom testing should be offered
- the advantages and disadvantages of genetic testing

Instead, if an individual is referred to the Genetics Clinic at CHEO for a medical opinion about whether or not their symptoms and lab tests results are caused by hereditary hemochromatosis, they are seen by a Medical Geneticist (physician specialist in genetic conditions) for a private appointment, much like appointments in other medical clinics, though it typically lasts 45 minutes to 1 hour. That appointment includes a medical evaluation as well as genetic counselling. The physician is likely to order hemochromatosis testing and may also order other tests depending on the patient's medical history, family history and previous lab test results.

In summary, a referral to the Genetics Clinic can make, confirm or clarify a diagnosis of hemochromatosis. It may also help their health care provider arrange appropriate surveillance and management of their iron levels. Genetic counselling is a part of each type of clinical encounter. Counselling helps patients understand the implications of their genetic test result and what this information means for them and their families. Additionally, a genetic counselling session can help an individual decide if genetic testing is right for them. Genetic counselling enables patients to weigh the benefits of testing – which can be substantial – with the relatively rare unintended consequences (such as inadvertent disclosure of non-paternity).

OTHER GENETICS CLINICS IN ONTARIO ARE LOCATED AT:

- McMaster Children's Hospital (Hamilton)
- Kingston General Hospital (Kingston)
- London Health Sciences Centre (London)
- Trillium Health Partners formerly Credit Valley Hospital(Mississauga)
- Lakeridge Health Network (Oshawa)
- Mount Sinai Hospital (Toronto)
- North York General Hospital (Toronto)
- University Health Network (Toronto)
- Northern Regional Genetics Program/Sudbury Regional Hospital (Sudbury)

"We did it again!"

And we couldn't have done it without you. Together, we surpassed our goal.

Canadian Blood Services
it's in you to give

In 2012, you, as a member of our Canadian Hemochromatosis Society Partners for Life group, helped to donate 176 units of blood to Canadian Blood Services, exceeding our goal of 150 units. This special CHS Partners for Life group now has 92 registered members, compared with last year's 74.

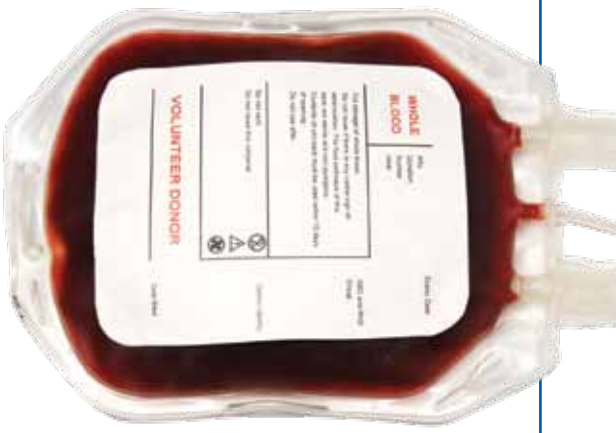
We have set our 2013 goal to 200 units, and we are well on our way to achieving that goal – we already have 27 units donated so far.

These uplifting numbers tell a story: Canadians with hemochromatosis have something to give, something that they are willing to give and need to give: their precious and life-saving blood.

With an estimated 110,000 Canadians affected by hemochromatosis, we have the unique and staggering potential to meet the needs of tens of thousands of hospital patients, if not more! We have the numbers, so now let's prove it.

Let's grow our Partners for Life group and continue to rally together to save or improve the lives of hospital patients across Canada.

If you are in the maintenance phase of your treatment and you are not already a donor, don't let your blood go to waste. Register with the CHS Partners for Life group from www.toomuchiron.ca or in person at the blood donor clinic, and go give blood! The Partner ID # to use is CANA002257 (four letters and six numbers).



For Canadian Blood Services donor clinic locations and schedules, visit www.blood.ca/clinics. Check your eligibility at www.blood.ca/eligibilty.