

The Gaucher Connection



For people with Gaucher disease, their families and their health care providers.

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The Gaucher Connection newsletter is part of the Gaucher Connection Program brought to you by Genzyme Canada. The Gaucher Connection Program is endorsed by the National Gaucher Foundation of Canada.



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The Gaucher Registry: What can it do for you?

Margaret Mackrell knows the value of the Gaucher Registry.

For more than a decade, Ms. Mackrell has worked alongside Dr. Julian Raiman treating patients with Gaucher disease at Toronto’s SickKids hospital. She also works very closely with Gaucher nurse co-ordinator Mary Anne Patterson and Dr. Dominick Amato at nearby Mount Sinai Hospital’s Gaucher Centre of Excellence. As one of North America’s largest Gaucher clinics, the Gaucher Centre of Excellence provides registry information on patients from ages 18 to 88, meaning Ms. Mackrell has significant registry experience and has seen first hand both the direct and indirect benefits participation in the registry holds for patients. With a new registry platform on the horizon, Ms. Mackrell anticipates the registry will soon be even more useful to patients and their health care team.

Value of registry in giving information to patients

Of the many benefits to participation in the Gaucher Registry, the one often most apparent to patients is the availability of information to help them track important symptoms of their disease. This information is often provided

to patients in the form of patient case reports – personalized information sheets that use charts, graphs and text to illustrate a patient’s progress over time. According to Ms. Mackrell, authorized health care professionals can print patient case reports from the registry for their patients. These can then be used during visits to show patients how they are progressing over time in several important areas, and patients can take their results home with them after the visit.

Not all health care professionals use the patient case report forms. Some health care teams prefer to use their own internal systems to communicate with their patients, which means you won’t necessarily be given a patient case report at your next visit. However, this doesn’t mean the registry is not being used to provide information directly to patients. According to Ms. Mackrell, data from the registry are routinely used to help educate patients about reasons they should or should not be on enzyme replacement therapy.

“If we get to the point to where we think they should start treatment, we can talk about patients who have started treatment,” says Ms. Mackrell. “You can show the graphs.”

If, on the other hand, the health care team feels a patient does not need enzyme replacement therapy, they can also provide the data needed to support their decision.

“If we don’t believe the patients need to be on treatment, then we have a good background to say there’s maybe 6,000 patients in this registry and of that number so many are on treatment, but there is a subset of patients who are not on treatment and may never need treatment.”



Margaret Mackrell

“ In Canada, we are giving the registry a tremendous amount of information on the natural history of the disease. ”

Value of registry in research and teaching

Participation in the Gaucher Registry is especially important for patients not just because of what they gain from the registry on an individual basis, but also because of what they gain collectively by providing information for researchers. More than anything else, Ms. Mackrell says the registry benefits patients by providing the data needed to study a rare disease.

This is especially true in Canada where patients do not necessarily receive enzyme replacement therapy. According to Ms. Mackrell, when research developed using registry data is published, there may be concerns about its applicability to patients who are not on enzyme replacement therapy. This is because most of the patients in the registry are on enzyme replacement therapy. In the United States, where the majority of patients with Gaucher disease are started on enzyme replacement therapy, this is less of a concern. However, in the rest of the world, where many patients never start enzyme replacement therapy or receive varying dosing regimes, it is not always clear if the information provided through registry-based research is applicable.

Because many of the Canadian patients providing information to the registry are not being treated with enzyme replacement therapy, the “natural history” information they provide to the registry is especially important, as it helps researchers to paint a true picture of the disease.

“In Canada, we are giving the registry a tremendous amount of information on the natural history of the disease,” says Ms. Mackrell. “I think that’s important for Canadians reading *The Gaucher Connection* to realize.... It’s as important for us to learn about why patients manage who are not on treatment as much as the response to treatment.”

While information for researchers is important, the direct benefits to patients can take months or years to become apparent. According to Ms. Mackrell, however, patients can see a direct short-term benefit through the quality of care delivered by their health care team as the data generated are used to train the health care professionals who interact with patients on a daily basis.

“When we’re in clinic and teaching fellows and residents who are rotating through the metabolic genetics program, one of the benefits of having the registry for such a rare disease is an accumulation of, number one, natural history data and also the effects of enzyme replacement therapy.”

Improved access for the future

While the Gaucher Registry provides important information and education to patients, researchers and health care professionals alike, some feel the interface used to enter and access information is somewhat cumbersome. According to Ms. Mackrell, this leads to some health care professionals not participating in the registry. In addition, as researchers learn more about the disease, some are advocating that more information about different aspects of the condition be collected.

To address these concerns, registry administrators have undertaken an initiative – known as RegistryNXT! – designed to help them understand and meet the needs of those participating in the registry. Based on this initiative, during which they consulted with health care professionals like Ms. Mackrell, Gaucher Registry officials will soon unveil a new web-based platform that better serves patients and health care professionals through improved data capture and access.

“The new platform is going to be web based. In a perfect world if you’re sitting in clinic...with a patient, it would be ideal to just go into the patient case report form,” says Ms. Mackrell. “You could actually share it with your patient without printing it out ahead of time. And at the same time you could also dynamically enter data into the registry about their current status.”

Ms. Mackrell anticipates that increased access to data will help to empower patients and that, at some point in the future, patients will have access to their own improved patient case reports via the web.

“We are facing a whole new generation of patients who have advanced computer skills and may feel comfortable accessing and retrieving their information online. I do think that the new registry platform will be revolutionary. My hope is that the new registry platform will help to facilitate data sharing and collection and revolutionize the care of our patients in clinic.”

For more information, visit the Gaucher Connection Program’s interactive www.gaucherconnection.ca platform and click on **The Gaucher Registry** in the left-hand menu bar.

Gaucher Guidebook

Tools for Tracking

Since its launch in 2008, the goal of the Gaucher Connection Program has been to help the Canadian Gaucher disease community manage the disease through the provision of information, support and interactive tools. Ultimately, those behind the program hope to assist patients in maintaining their independence as they make important treatment decisions and navigate the disease management process. To help patients track appointments, test results and changes in their disease, the Gaucher Connection Program's interactive www.gaucherconnection.ca platform provides patients with a variety of useful disease management tools.

My Test Tracker

My Test Tracker is an online tool used to help patients keep track of their progress by monitoring test results. Your test results allow your medical team to track your disease status over time to ensure your treatment plan is effective and to make changes to your treatment if they are needed. By getting your test results from your doctor and entering them in the My Test Tracker tool, you will be able to keep track of changes in your disease status and monitor your own progression.

The My Test Tracker tool allows you to record information on your biomarkers, blood, spleen, liver and bone tests. After entering your information you can review individual entries, or use the graphing tool to chart your progress.

My Calendar

Of course, a tool to track test results is of little use if there are no results to track. That's where the My Calendar tool comes in. This easy-to-use appointment tracker allows you to record the date, time and location of each of your appointments, and even features a straight-to-your-inbox e-mail reminder service.

My Health Diary

Your health care team needs to know how your disease affects your life between appointments. My Health Diary helps you to keep track of day-to-day health status changes between appointments and can be useful in helping you to see long-term trends that might otherwise slip by unnoticed. You can use the My Health Diary tool to record details of how you feel, changes in your quality of life and symptoms such as pain, fatigue, bleeding or bruising.

For more information on these tools, visit the Gaucher Connection Program's website at www.gaucherconnection.ca, log in using your username and password and click on the **My Health Tools** link on the left-hand side of the screen.

Q & A Dialogue: Answering Questions

**How do you tell your boss you have Gaucher disease?
What about your friends and family? Your teacher?
Or that cute guy or girl who's becoming more than just a friend?**

In some cases you may not want to say anything at all. That's a personal choice only you can make. However, you may eventually need to explain to your boss why you need to miss work for tests. You may need to tell classmates why there's a home infusion pump in your book bag. It's even possible that cute guy or girl is wondering what Gaucher disease could mean for your future together.

Chances are few of these people will ever have heard of Gaucher disease. Some of them may even be surprised to hear you have this condition at all. This means if you do decide to tell people about it, they will probably have a lot of questions. The best thing you can do is to be prepared.

What do I say?

You probably already know a lot about Gaucher disease. Discussions about platelet counts and liver volume might be second nature to you at this point. But think back to your initial diagnosis and some of the questions that ran through your mind. (If you were too young to remember, ask your parents.) The important thing is to start with the basics and move on from there. Use simple, straightforward explanations of the science behind Gaucher disease, how it can affect you and what steps you're taking to manage the disease.

To help you prepare for some of these conversations talk to your nurse or genetic counsellor, or visit the Gaucher Connection Program's website at www.gaucherconnection.ca and click on the Gaucher Disease link on the left-hand side of the screen.

We'd like to hear from you

How did you tell the important people in your life about Gaucher disease? What helped? What went well? What could have gone better? Let us know by sending a short note to gaucherconnection@genzyme.com.

Profile

Karen MacDonald Carey Running with it!

Karen MacDonald Carey describes carrying the Olympic torch as “the most amazing 300 metres I have ever run.” That says a lot, considering the many thousands of times she has run 300 metres over the last 12 years.

Running with the torch

Ms. MacDonald Carey’s time in the Olympic torchlight began with a suggestion from her grandson who, upon learning that Olympic organizers were looking for torchbearers to participate in the relay, suggested that his marathon-running Nana should apply.

So she did.

After entering her name in an online draw, Ms. MacDonald Carey won the opportunity to write a 500-word essay explaining how she lived actively. The essay was submitted to a panel of judges, who selected her for the relay. That was last August.

Then, on December 20, 2009, Ms. MacDonald Carey – a 53-year-old medical centre supervisor with four children, five grandchildren, Gaucher disease and a passion for running – carried the Olympic flame for a small fraction of its journey from Olympia, Greece, to Vancouver, British Columbia, for the 2010 Winter Olympics.

Because Ms. MacDonald Carey was the first torchbearer of the day, she had about half an hour to mingle and pose for photographs with the family, friends and strangers who had gathered in the pre-dawn Hamilton, Ontario darkness.

“In the beginning when I first got dropped off, it was my family and friends and they were like ‘Karen, Karen, Karen!’ ” says Ms. MacDonald Carey of the 30 minutes prior to her run. “And then all of a sudden I kept on hearing ‘Karen!’ but I’m looking and I’m thinking ‘I don’t know those people.’ But they were calling my name because they knew they would get my attention because they heard my family call my name. They all wanted to hold the torch and take pictures. It was amazing!”

Then, came the moment when the torch was lit.

“I did not expect to be as moved as I was when they lit the torch,” she says. “I was looking at the torch, and when it lit on fire and I looked up at it I just burst into tears. I don’t know why, it just really got me. It was very moving.”

Her tears didn’t stop her from running. After the torch was lit, Ms. MacDonald Carey jogged down the street, screaming for joy the whole way.

“I felt like part of me was running for all those people who can’t run,” she says. “My sister Gale has Gaucher disease much more severely than I do and she would never be able to run that. So I felt like I was carrying that for her too and for all other people who have disabilities that you don’t necessarily see.”

After 300 meters she hugged the next runner in the relay, they “kissed torches” to pass the flame, and the next runner continued on his way. Immediately afterwards, her flame was extinguished, the fuel was removed from the torch and she was in a shuttle bus with a memory to last a lifetime.

“I loved every second of it.”

Living actively

Although carrying the torch represents a high point in Ms. MacDonald Carey’s career as a runner, it is certainly not her only achievement. Over the past 12 years, she has run 18 full marathons and a multitude of other long-distance races.

Ms. MacDonald Carey started running as a way to increase her stamina for martial arts training – Ms. MacDonald Carey has a second-degree black belt in karate. However, she soon discovered she only had



time for one sport. As she fell in love with running and started to worry that increasing levels of physical contact in karate might lead to broken bones, she soon found herself training for marathons.

“ I know that sometimes the Gaucher may slow me down, but I also know that for me it can be as simple as remembering to put one foot in front of the other and eventually I will cross the finish line. ”

That’s not to say that running didn’t initially cause her some concern. People she knew questioned the wisdom of running when her bones were at risk. However, when given the opportunity to speak with a Connecticut-based Gaucher disease expert who was in Toronto for a meeting, Ms. MacDonald Carey not only found reassurance, but a new level of motivation. She asked him if she was doing herself harm by running.

He said, “Absolutely not. If you can run, run. Because one day you might not be able to. So just keep running.”

This advice continues to drive Ms. MacDonald Carey today. On days when motivation is lacking, Ms. MacDonald Carey tells herself she doesn’t have to run; she gets to run.

“Some days when I’m running and I’m really struggling and other people are way ahead of me I think, ‘Wow, is this from Gaucher?’” she says. “But then I think, ‘At least I’m out here. There’s lots of people who can’t be out here and would love to be out here...I’m fortunate. Very fortunate.’”

Ms. MacDonald Carey doesn’t claim to be fast. A typical marathon will take her between four-and-a-half and five hours, and trail runs take longer. She does, however, claim to listen carefully to what her body is telling her.

“When I found running five days a week was too much for me I added some cycling and swimming and dropped to three times a week running,” she says. “If I am really tired and my hemoglobin is low I take it easy and maybe run slower or skip a run.... I try to push myself but not to the point of exhaustion.”

Inspiration – a family affair

Ms. MacDonald Carey says she finds her motivation to run in those around her. In addition to carrying her sister in her heart on every run, she cites her mother – an avid hiker – as her source of inspiration. Ms. MacDonald Carey has also run marathons with her two daughters and a son-in-law, she trains with a supportive group of running friends, and she always runs races with her husband.

“He is a wonderful man who was my high-school sweetheart and we have been married for 35 years,” she says. “When we are running and he can see I am struggling he will actually run backwards in a race to make sure I am OK. He always slows down and waits so we can cross the finish line hand in hand.”

As for what she would do should she ever have to stop running?

“I actually don’t think about it too often. I would find something else,” she says. “Whatever I could do to keep myself up and active and keep myself going. I guess it would depend on what my limitations were. It would be pretty hard to keep me down, I think.”

In the meantime, Ms. MacDonald Carey continues to run. She recently participated in Hamilton’s Around the Bay 30k both as a volunteer and a runner, and hopes to run at least two more marathons this year.

“I know that sometimes the Gaucher may slow me down, but I also know that for me it can be as simple as remembering to put one foot in front of the other and eventually I will cross the finish line.” ■

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