

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.



www.gaucherconnection.ca

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Why register for a registry?

Diagnosis with Gaucher disease can be a difficult and emotionally draining process for patients and their families. The diagnostic process is often long and complicated, and it's only the beginning. When a diagnosis is made, patients and their families must work with their health care teams to learn about the disease, make lifestyle changes, learn about treatment options and make important treatment decisions.

In the midst of all this, patients will eventually be asked if they want to participate in the Gaucher Registry.

What's a registry?

A disease registry is a collection of information about different patients with a specific disease or condition. It can be as unsophisticated as papers in a filing cabinet collecting information about a single physician's patients, or it can be a large and comprehensive database including information from tens of thousands of patients from around the world. Disease registries are one of several important tools researchers use to look at the natural progression of a disease as well as the effects of different treatment options over time.

“ The hope is that the information collected in the Gaucher Registry will eventually lead to **earlier diagnosis**, earlier treatment **and better disease management** for people with Gaucher disease. ”

What's the Gaucher Registry?

The International Collaborative Gaucher Group (ICGG) Gaucher Registry is a cooperative observational Gaucher disease registry. It was created to gather information to improve the understanding of Gaucher disease and is designed to ultimately provide the Gaucher community with information about patients with Gaucher disease from around the world. The official objectives of the registry are:

- To enhance the understanding of the variability, progression, and natural history of Gaucher disease with the ultimate goal of better guiding and assessing therapeutic intervention
- To provide the Gaucher medical community with recommendations for monitoring patients and to provide reports on patient outcomes to help optimize patient care
- To evaluate the long-term effectiveness of enzyme replacement therapy

Why do we need a registry for Gaucher disease?

Researchers have a variety of tools they can use to study diseases. However, rare diseases present researchers with unique challenges. Simply put, when studying rare diseases, it can be difficult to collect information on how the disease and its treatment affect different people. The fewer patients there are available to study, the less reliable the results will be. Disease registries help researchers overcome some of the challenges of studying a rare disease by collecting as much information as possible about as many patients as possible into a single place.

Gaucher disease is a perfect example of a rare disease. Many medical professionals agree that since

Gaucher disease is rare, accurate and complete information on the disease is especially important. The hope is that the information collected in the Gaucher Registry will eventually lead to earlier diagnosis, earlier treatment and better disease management for people with Gaucher disease.

What can the Gaucher Registry do for me?

The information collected through the Gaucher Registry not only helps researchers to develop better disease management tools and strategies over the long term, but it can also assist you and your health care team in managing and monitoring your disease today and over time, regardless of whether you are currently being treated with medication. For example, a physician with a patient enrolled in the Gaucher Registry can produce a Patient Case Report before every appointment. This report uses charts, graphs and text to give a Gaucher-related health status overview and to show progress on important measures such as bones, blood, liver, spleen and quality of life. Health care professionals can review this report with their patients to illustrate how their health is progressing over time.

Because many patients feel more in control when they take an active role in their health care, the Patient Case Report can also be used to help keep the lines of communication open. For example, patients enrolled in the Gaucher Registry can ask their physicians or nurses to share their Patient Case Reports with them at future visits.

What about my privacy?

Patient privacy is a top concern for the Gaucher Registry. Information about patients can only be submitted with their written authorization. People enrolled in the registry will not be identified by name, as patient names are not collected. Instead, every patient in the registry is randomly assigned an identification number which is only given to the patient's physician. Any information that could link a patient to his or her physician is also kept confidential. All information in the registry is collected in a way that prevents the identification of specific patients.

Who can participate?

Anyone who has been diagnosed with Gaucher disease is eligible to participate in the Gaucher Registry. Participation is completely voluntary. After joining the registry, a person's medical information is pooled with other participants and used to assist researchers studying trends and addressing specific questions about treating Gaucher disease.

How can I join?

Patients wishing to participate in the Gaucher Registry are enrolled by their physicians, who explain the registry before asking their patients to complete an authorization form. If you are not enrolled in the Gaucher Registry, talking to your physician is an important first step. Ask your physician if he or she is enrolled in the registry and let your physician know you are interested in participating.

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.

Education and Awareness The Gaucher Scholarship Program

Genzyme Canada and the National Gaucher Foundation are providing **two post-secondary scholarships** for people with Gaucher disease.

The Gaucher scholarship program, designed to increase awareness of Gaucher disease and to promote the well-being of members of the Gaucher community, will provide winners with \$2,500 each to pursue their studies at a CEGEP, college or university in the 2009-2010 academic year. Winners will be selected by Genzyme Canada and the National Gaucher Foundation based on a combination of a creative and persuasive essay, excellent recommendations and academic standing.

To apply, students must be enrolled in a program spanning at least eight months in the 2009-2010 academic year, and lasting at least two years overall, in an accredited college or university. They must also complete and send in an application form and a release form, write and submit a 1000-word essay, and ask someone who knows them well (but is not a family member!) to send in a recommendation.

If you or someone you know is right for this scholarship, visit the Gaucher Connection Program's interactive www.gaucherconnection.ca platform and click on **Scholarship** in the left-hand menu bar for more details.

Q & A Dialogue: Questions and Answers about Insurance

Adequate insurance is an important part of anyone's financial plan. It protects dependents from the consequences they may face when death or disability results in a loss of needed income. A chronic illness, however, changes the insurance equation, limiting some insurance options while making others more attractive.



Group insurance plans

"If you work for a company and they have a group plan, then you are entitled to benefits," says Martin Firestone, a benefits consultant in Toronto, Ontario. Depending on the type of group insurance offered, people with Gaucher disease can have access to the same life, critical illness, long-term care, disability and travel coverage as any other employee within the company, with no medical questions asked.

However, according to Mr. Firestone, these types of programs have a major drawback.

"You leave the company, you leave all those benefits behind," he says. "The best idea, if you can, is to apply and get approved for an individual policy."

Individual insurance plans

Individual insurance plans stay with a person regardless of employment status, as long as they continue to pay the premium. However, not all insurance products are available to people with Gaucher disease. For example, living benefits insurance, which includes long-term care, disability and critical illness coverage in case someone is unable to continue working, is not available to people with Gaucher disease.

Life insurance, on the other hand, is available to most people with Gaucher disease. While those with type 2 or type 3 Gaucher disease will be unable to get individual life insurance, those with type 1 Gaucher disease are able to get individual life insurance coverage. This coverage, however, comes with a rating, which means the insurer will want more premium for the same coverage.

“That’s a good thing,” says Mr. Firestone. “It’s better to get a policy rated than to get no policy at all.”

According to Mr. Firestone, people with Gaucher disease typically have ratings resulting in premiums between one and a half and two times those paid by individuals in perfect health. The final decision will be made by the insurance company based on information provided in your application, a report from a nurse who will perform various tests, and a statement from your physician. People with Gaucher disease may be declined completely depending on other medical conditions.

The effect of the rating on the premiums paid will depend on whether you choose temporary (term) insurance or permanent (universal life) insurance.

The premiums paid with term insurance, which can be purchased for 10- or 20-year terms, will remain the same for the length of the term and will increase to a new level for the next 10 or 20 years. Term insurance expires when you reach age 85.

Universal life insurance premiums, on the other hand, will never increase and the insurance will never expire. While the premiums are more expensive, this type of insurance can be advantageous for people with Gaucher disease.

“The key here is that people with a rating, in this case a 150% rating, can have that rating removed, virtually issuing a policy that is standard,” says Mr. Firestone.

Having the rating removed can make a big difference over the long term. For example, a 50-year-old with Gaucher disease purchasing \$100,000 worth of 10-year term insurance will pay \$195,000 in premiums by the time the term expires at age 85. If the same person chose 20-year term insurance they would pay \$132,000.

“With permanent insurance,” says Mr. Firestone “They would have put out \$45,000 because of the reduced rating, and also still have the insurance for life.”

Travel insurance

People with Gaucher disease are also able to get travel insurance. As with anyone applying for travel insurance, their insurance company will ask questions about their health that can result in approval or denial of coverage. While there are no questions directly related to Gaucher disease, insurance companies are interested in stability, meaning no changes in medications and no investigations pending.

“When it comes to travel insurance, you can get it,” says Mr. Firestone. “Just be careful the products you buy and the coverage you get.”

For more information, visit the Gaucher Connection Program’s interactive www.gaucherconnection.ca platform, click on **My Community** in the left-hand menu bar and then click on **Media Library**. Look for the videos on insurance coverage. You can also contact Mr. Firestone at **1-877-636-4441** or at **martyf@dot-if.com**.

Questions & Answers

Profile

Gloria DiFolco Acting Out

Gloria DiFolco is used to putting herself in other people's shoes. As an actor, she has spent her career trying to imagine and understand the feelings of the characters she plays. So she can understand the negative reactions of friends and associates when she explains she has Gaucher disease and has to undergo infusions every two weeks. She doesn't get upset, but she loves to change their attitude.



Ms. DiFolco was diagnosed with Gaucher disease more than 20 years ago at a time when there were few treatment options available. While keeping in mind the precautions needed to protect herself from serious injury, she all but forgot about her condition for the next 15 years. During this time, she focused on her career in film, television and theatre. It was only when she moved to Kingston, Ontario, that she found a family doctor who happened to have another patient with Gaucher disease. It was this doctor who told her about the many treatment advances that had been made over the years and who referred her to local specialists. These specialists, after monitoring her condition for a few years, recommended she start enzyme replacement therapy.

At first, Ms. DiFolco's infusions were easily integrated into her life as she remained in Kingston and in close proximity to the hospital. But eventually, the work of someone who makes her living on stage, on screen and behind the scenes was bound to interfere with her infusion schedule.

Acting career

Ms. DiFolco doesn't define herself as a Gaucher patient. Rather, she seems to identify herself as an actor and a lover of the dramatic arts who happens to have Gaucher disease.

Ms. DiFolco's love of acting started at a very early age. "At the elementary school I went to we used to do plays, but they were very professional in the way they were produced. They had a professional theatre director come in and direct us, and then we performed them outside of the school a few times," says Ms. DiFolco. "So basically my passion for acting started there, and I just kept it going all my life."

Since then, Ms. DiFolco has been involved in "a lot of little independent films that haven't really gone anywhere," multiple theatre productions and a few television shows. She once played a small role opposite Sophia Loren in *Between Strangers*, she works in film production and every summer she goes to Middlebury College in Vermont to direct an Italian-language play for the school's summer language program.

"I'm not anybody famous," she says, "but somebody who has had the opportunity to do what they like to do."

All of this means Ms. DiFolco travels regularly for work, sometimes for weeks or months at a time. In addition, she goes to Italy every year, where she occasionally works and where she always spends time visiting family.

A balancing act

Ms. DiFolco's ability to work and travel without interrupting her treatment has been a story of teamwork, compromise and co-operation, all of which has happened with minimal involvement from Ms. DiFolco herself.

"I think the first challenge was Vermont," she says, referring to a summer working at Middlebury College.

After trying without success to find someone locally who could administer ^{Pr}Cerezyme[®], Ms. DiFolco's health care team, in cooperation with Genzyme, found a way to ensure she was able to continue receiving her treatment. "With the help of Genzyme – who I have to say is incredibly organized and really good at what they do – they arranged for me to have my enzyme replacement therapy done in this very small town in Quebec," she says. "There was a pharmacy there and the pharmacist's office is where I would have the enzyme replacement therapy. I would meet this nurse there and she would do it every two weeks."

Infusions in Italy were made possible, as well, with co-operation between Ms. DiFolco's health care team, the Ontario government and Genzyme. "They arranged for me to go to a hospital in Rome. I go to the hematology ward in the pediatric unit and I have this enzyme replacement therapy," she says. "I just have to show up and everything is there and ready to go."

That's not to say Ms. DiFolco is not sometimes frustrated by the limitations imposed by her disease. She admits she might like to ski or play soccer or participate in other activities that put her at risk of bone injury or hemorrhaging. And she acknowledges that some days she has little appetite to fight traffic and weather, go to the hospital, deal with the bureaucracy of the Italian health care system, and sit for hours for an infusion. But overall, Ms. DiFolco says her experiences, especially in the pediatric hematology unit, as well as the recognition that her parents immigrated to a country where her infusions are covered by the health care system, have made her thankful for the advantages she has.

"When you've been taking your enzyme replacement therapy in a pediatric ward where kids are getting chemo and they're smiling, it totally puts everything in perspective," she says. "Once you sit there and you see what's going on around you, you sit down and say, 'Wow. You know what? My situation isn't great, but it's really not bad.' "

It's this attitude shift Ms. DiFolco loves to bring about in others who have a negative reaction when they learn about her disease.

"What I like to see is the reaction they have when I say to them, 'Really, you know what? There are worse things in life. There are worse things and if this is really the worse thing that's going to happen to me in terms of health, I'm good with it.' And all of a sudden they change and they say, 'Oh. You know what? You're right! You're absolutely right!' "

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