HOW DO I TALK TO MY FAMILY ABOUT Gaucher?
This booklet will help you talk to your family about Gaucher. Each section includes different approaches to speaking with your family. Each family is different, so choose the approach that works best for you and your family.

**What is family?**

There is no one-size-fits-all family. Your family is the group of people who are important to your well-being. They might be related to you biologically, legally (through adoption or marriage), or emotionally (through affection, dependence, obligation, or cooperation).
Why should I talk to my family about Gaucher?

Gaucher is a rare condition (relative to other chronic health conditions). Most people do not know what it is and might never have heard of it. You may find that many doctors and other healthcare professionals do not know much about it.

It is important to help your family and healthcare providers understand what Gaucher is, what it means for you, and how your diagnosis might impact your family. Some topics to share are major signs and symptoms and how the condition is passed down through families. Turn the page for more information on Gaucher.

Sharing your story will help your relatives understand their family health history. In turn, it is helpful for you to know your whole family health history, including Gaucher and other conditions that might run in your family. Putting the whole picture together will help you and your relatives:

• Identify risks due to shared genes.
• Talk to each other about health (including quality of life).
• Summarize health information to give to healthcare providers.
What is Gaucher?

Gaucher (pronounced go-shay) is an inherited condition that causes deposits to build up in certain organs and bones. Our bodies contain thousands of active substances called enzymes. For example, your stomach has enzymes that help break down the food you eat. In healthy people, the enzyme glucocerebrosidase (pronounced gloo-ko-ser-e-bro-sy-daze) helps the cells of the body break down a certain type of molecule (glucocerebroside). People with Gaucher do not have enough of this enzyme. As a result, cells fill up with this substance and no longer work normally. These cells are referred to as Gaucher cells.

Linda’s story

In 1990 at my annual check-up, my doctor noticed my platelets were low and my spleen and liver were enlarged to five times their normal sizes. I was anemic, I bruised easily, and I kept getting sicker. After a year of testing for various diseases including lupus and leukemia, my Primary Care Physician sent me to a Hematologist/Oncologist who fortunately specialized in Gaucher. After a bone marrow test I was finally diagnosed in 1991. My bones were just showing signs of the disease, but enzyme replacement therapy (ERT) improved my symptoms. My platelets
There are three major types of Gaucher. Type 1 is the most common form. The symptoms are extremely variable. In some people symptoms can be seen in early childhood, while others are diagnosed as senior citizens. Type 1 Gaucher does not affect the brain. Gaucher Types 2 and 3 affect the body and the brain. Babies with Type 2 can be very sick and usually do not survive the second year of life. Children with Type 3 tend to have more severe Gaucher and some effects on the brain.

Type 1 Gaucher occurs more often in the Ashkenazi Jewish population (Eastern European descent) compared to the general population. However, it is important to remember that Type 1 Gaucher can occur in individuals of any ethnicity.

Type 2 and Type 3 Gaucher are very rare.

started to go up, I was no longer anemic, and after a couple years my liver and spleen returned to their normal sizes.

It put things in perspective for me as I sat there receiving treatment with cancer patients who did not know if they would live or die. I was able to become pregnant, deliver a healthy baby boy, and now keep up with my 14-year-old son. I exercise regularly. I live a healthy, productive life with Gaucher. My future is very hopeful!
Who is at risk?

- Gaucher occurs in both males and females.
- Gaucher affects people of all races and ethnicities, all over the world.
- Gaucher is more common in people of Ashkenazi (Eastern European) Jewish descent.
- The signs and symptoms of Gaucher may appear in affected individuals at any age.
- The signs and symptoms of Gaucher may be very different within families, even between siblings.
- When both parents are carriers, there is a 1 in 4 (25%) chance that any child born will have Gaucher.
- If one parent has the disease and the other parent is a carrier, there is a 1 in 2 (50%) chance that any child born will have Gaucher.
- When both parents have Gaucher, all of their children will inherit the disease. [Turn to page 9 for a chart showing how Gaucher is passed down through generations.]
The signs and symptoms of Gaucher may appear in affected individuals at any age.

**Major signs and symptoms**

- Anemia (low red blood cell count)
- Fatigue (tiredness)
- Low platelet count
- Easy bruising
- Large spleen and liver
- Easy bleeding that is difficult to stop
- Bone pain, bone crisis (severe bone pain often accompanied by fever), and the chance of easily broken bones
- Reduced bone mineral density
How are individuals affected?

Receiving a diagnosis of a genetic condition can be a life-changing event for individuals and families. Fortunately, much has been learned about Gaucher—what causes it, how its symptoms can be managed, and treatments that may prevent, stop or reverse major symptoms.

Quality of life

For some people, discomfort, pain, and disability due to skeletal disease can seriously impact quality of life. Today, with early diagnosis and appropriate treatment, some people with Gaucher may not experience such severe and debilitating bone involvement.
Diagnosis

Early diagnosis and treatment are very important for people with Gaucher. However, Gaucher is not very well known, and many people endure months and years of misdiagnosis before receiving a definitive diagnosis of Gaucher. Definitive diagnosis can be made with a simple blood test that measures glucocerebrosidase enzyme activity. The blood sample can be taken in the healthcare provider’s office, but may need to be sent to a specialized medical center for analysis. Diagnosis can take a long time because signs and symptoms of Gaucher are often shared with other disorders, such as:

- Leukemia or lymphoma
- Multiple myeloma
- Idiopathic thrombocytopenic purpura (ITP)
- Neimann-Pick disease
How are families affected?

A diagnosis of Gaucher in any person may reveal an increased risk that others in the family may carry the gene, or in fact have Gaucher. This includes siblings, parents, and children as well as more distant relatives such as cousins. Genetic counseling for families with Gaucher is important to help address who should be tested for Gaucher in order to determine if they have Gaucher or if they are carriers.

Hints for health

• Early diagnosis may reduce risk of serious medical complications.

• Gaucher is a lifelong condition. Visit your doctor regularly for check-ups. Specific monitoring over time will help your physician keep track of your liver and spleen size, platelet levels, and bone health.

The following are some tests your physician may perform:

• Physical examination

• MRI (magnetic resonance imaging)

• Blood work to review anemia and platelet levels
Ben’s story

My name is Ben, and I have Gaucher. So does my paternal grandmother.

My grandmother, Fran, passed the gene to my dad and Aunt Barbara. They are both carriers. My aunt married Bill, who does not have the gene for Gaucher. I have two cousins—Stephen is a carrier, and Kim is not.

My dad married another carrier—my mom. This means that each of their children—me, my brother and sister—had a 1 in 4 (25%) chance of having Gaucher, a 1 in 2 (50%) chance of being a carrier, and a 1 in 4 (25%) chance of not having the Gaucher gene at all. Molly was born first—she doesn’t have the Gaucher gene! I’m the middle child—I have Gaucher. Mike is a carrier.
You share a lot with your family—including what can make you sick.

Family members can have genes, habits, diet, and environment in common, so they share risk for the same health problems.

Most common diseases (heart disease, diabetes, etc.) are known as “multifactorial” conditions. They are caused by a combination of genes, lifestyle, and environment. People with similar genes may not develop the same illness if they make different choices or live in a different environment.

Melissa’s story

One Thanksgiving, my cousin told the family she was tested and is a carrier for Gaucher. She told us all we should get tested. I was pregnant at the time and my OB-GYN referred me to a specialist, who explained the Ashkenazi Jewish testing panel to me. It turns out both my parents are carriers, and I have Gaucher. I now have a son with Gaucher, too.
On the other hand, many conditions are caused by a specific change in the DNA of a single gene, called a mutation. Many of these conditions, like Gaucher, are rare. In these cases, genes play a much bigger role than lifestyle and environmental influences. These conditions usually develop when an individual is born with a specific gene mutation(s).

Some conditions are more common in particular racial or ethnic groups. For example, Gaucher is more common in people with an Ashkenazi (Eastern European) Jewish background, though it is not limited to that group. Other conditions occur equally across populations.

It is important to discuss all types of health conditions—common and rare, mild and fatal, acute and chronic—with your family and doctors. Knowing about the conditions that run in your family can help you make informed decisions about your health.

It is important to be honest when talking to family and to watch out for misinformation. My sister married someone who is not Jewish, so she did not think it was necessary for him to be tested. I had to emphasize to her that this does not only affect Jewish people.
What is family health history?

Family health history is a collection of information about health conditions that run in your family, as well as the eating habits, activities, and environments that your family shares. Your family's health is one part of the entire history of your family. Knowing about health conditions in your family can help you make healthy choices.

How can family health history affect my health?

You inherit many things from your parents and grandparents. They pass on culture and values through photos, recipes, stories, spiritual practices, and music. You also inherit how you look—for example, your height and the color of your eyes. Small structures in cells called genes carry information for these characteristics and guide your body through growth and development. Changes in genes may also lead to a risk in the family for developing certain health conditions, such as Gaucher.
Genes are the instructions inside each of your cells. Since everyone has slightly different genes, everyone has a different set of instructions. Genes are one reason why you are unique!

- Genes carry instructions that tell your cells how to work and grow.
- A person has two copies of each gene, one from the mother and one from the father.
- A change in a gene is called a mutation.
- Genes are inside cells. Every part of your body is made up of billions of cells working together.
- Genes are arranged in structures called chromosomes. Most people have 23 pairs of chromosomes. Copies of the chromosomes are found in each cell.
- Chromosomes are made up of DNA. DNA is the special code that spells out the instructions in your genes.
- Gaucher is caused by a mutation in a single gene.
How can my choices affect my health?

Many things affect your health. Some things—such as your genes—are outside of your control. Every person has many gene changes (mutations). Sometimes these changes have no effect or are even slightly helpful. But sometimes, as with Gaucher, they can affect your health.

Other things—such as what you eat, whether you smoke, whether you exercise, and what you do for a living—can be influenced by the choices you make. Though you can’t change your genes, you can change your behavior.

Karen’s story

Several years ago, my dad had a triple bypass. My siblings and I all ran to get blood tests. We knew that if our father, who was very diligent about his health, had high cholesterol, then we could all have it. When my results came back, my doctor thought I had cancer. Eventually a hematologist realized it was Gaucher. My son had symptoms of Gaucher but had never been diagnosed. With my diagnosis, we realized he has it, too.
I learned that I inherited Gaucher from both parents, so I told my immediate family right away. We do not jump up and down and shout that we have a genetic disease, but within our family everyone knows. It is to their benefit to share this information.

Though you can’t change your genes, you can change your behavior.
Why is it important to let my family know I have Gaucher?

Gaucher is an inherited condition. This means it is passed on through generations. If you or someone in your family has Gaucher, other family members, adults or children, may also have Gaucher and not know it.

Knowing this family health history can help you make smart choices for yourself and your family. For example, you can:

• Get yourself and your partner tested for Gaucher and carrier status.
• Make informed reproductive choices.
• Get treatment at the first signs of symptoms.

Enrique and Disleny’s story

There is always communication within our family, because someone always has something. My father has Gaucher; his illness inspired me to study to become a nurse. In a class we were asked about an illness that runs in the family. I used Gaucher, and my teacher had never heard of it. I hope to help my family and eventually find a cure.
How do I talk to my family about Gaucher?

Gaucher may cause symptoms that are different from common conditions like heart disease, diabetes, and cancer, but the steps you take to talk to your family about it can be similar.

**Talk to your family**

Your relatives are the best source of information about your family. This means you are a source of information for them in return. Family history is often shared during conversations at events like birthday parties, weddings, reunions, religious gatherings, holiday dinners, and funerals. These events provide an opportunity to talk to family members about their lives. You can ask them questions about their health and tell them what you know about yours.
How do I talk to my family about Gaucher?

Use what you have

Test results, letters from your doctor, or other information you received about your diagnosis can be shared with family to help them understand your circumstances. It may help explain their chances of having Gaucher or the chance of children inheriting Gaucher from their parents. Websites and brochures that have been useful for you might also be useful for your family members. This booklet can be passed along to help explain family health history and Gaucher.

Plan an individual conversation

After you have brought up Gaucher, you may want to talk with certain family members in more detail. Some people respond better to one-on-one talks rather than conversations in a large group setting. This will give you the chance to address any questions they have and ask questions of your own.

These individual talks should take place when both you and your family member feel up to it: you should not feel rushed or tired. Give the conversation the energy and attention it deserves. Also make sure your family member does not feel threatened. Let him or her know you are open to questions and that it is not a one-time conversation; the discussion can continue in the future.
Bring support
If you have a friend or family member who is a medical professional or who understands Gaucher and has been a help to you throughout your diagnosis and treatment, it could be useful to have him or her there for support during the conversation.

Send a letter
You may wish to send a letter to your family with an update on your health and information about Gaucher. Your physician or genetic counselor may be able to help you write this letter and identify who in the family would benefit from being informed.
How do I prepare for the talk?

Know the facts

Know the facts about Gaucher so you can answer any questions your family members have or refer them to other resources. Use your own experiences and information you learned from your doctor and others. Remember, Gaucher varies from person to person, so talk to your healthcare provider for medical advice. [Turn to page 2 for more details on Gaucher that you can share with family.]

Know how it might impact your family

For each family member you talk to, understand what your diagnosis might mean for him or her. For example, other family members may have an increased chance of being a carrier for

Suzanne’s story

When I was first diagnosed with Gaucher, my husband and I were devastated, and actually a bit confused. After the geneticist explained the inheritance of Gaucher, it all made sense. But the challenge was to explain it to my parents and the rest of our family. My father and mother did not want to accept the fact that they had passed a mutated gene to their child. There was a tremendous amount of guilt.

My father had the most difficulty with the diagnosis. He blamed my disease on all the exercise I did, or said I caught the disease from someone else. It took my parents a few months to realize the importance of understanding and learning about Gaucher and exactly how it was transferred from them to me.
Once they accepted it, my parents became proactive about Gaucher, holding fundraisers and speaking to Jewish organizations. My family also got tested: My father had asymptomatic Gaucher, my mother is a carrier, and my two sisters are carriers.

My family helped me get through many hurdles dealing with multiple surgeries and the early years of coping with horrific symptoms. I am fortunate to be blessed with the most amazing, supportive team: my family.

**Get ready for different reactions**

Be sensitive to a person's wishes not to talk about certain topics. Some relatives might not be comfortable hearing about Gaucher or other health conditions right away. Make it clear that you are available to talk whenever they are ready.
Conversation starters

There are many ways to talk to your family about health and Gaucher. After you tell your family your story, you might want to let them know what symptoms people can have that might be signs of Gaucher.

Health problems associated with Gaucher

• Easy bruising
• Prolonged bleeding time after getting a cut
• Easy fatigue (tiredness)
• Bone pain
• Enlarged abdomen caused by enlarged liver or enlarged spleen

Questions about family to help build a family health history

• Do you know if your parents or grandparents took medicine on a regular basis? If so, what kinds and for what?
• Has anyone had problems in pregnancy or childbirth? What kinds of problems?
• Are there any diseases that run in our family?
• Is there anything else you would like to tell me about your life or health concerns in our family?
What now?

Keep talking to your family and doctors.

Be prepared to talk with family members whenever they are ready to have the conversation.

Stay up to date on Gaucher, for yourself and to inform your family.

Remember – you cannot change the fact that you have Gaucher, but you can take steps toward a healthier life.

Resources

Learn More and Get Involved
National Gaucher Foundation: www.gaucherdisease.org
Gaucher Care: www.gauchercare.com
Lysosomal Learning: www.lysosomallearning.com
Gaucher Associations Around the World: www.gaucher.org.uk

Diagnostic Testing and Registry
www.genetests.org
www.gaucherregistry.com

More Resources
Disease InfoSearch: www.geneticalliance.org
National Organization for Rare Disorders: www.rarediseases.org
Genzyme Medical Information: 800.745.4447 (option 2)
What should I do if my family does not want to talk about Gaucher?

Some family members might not understand why it is important to know this information. Others might be nervous about receiving a diagnosis of Gaucher themselves. It is normal to be anxious or uncomfortable if something is unfamiliar. We are all at different emotional places at different times and will process the information differently.

You may wish to start the conversation with other conditions that they are more familiar with that run in your family, like high blood pressure or asthma. Then move on to Gaucher. Make it clear that although there is no way to lower your risk for Gaucher, you can still lead your life just like they do, with hobbies, a career, and a family.

Try to get them involved even if they express denial. If they are not willing to take steps to learn more on their own behalf, they might be willing to do it for the rest of their family.

Ramon’s story

I have three children with Gaucher. When they were diagnosed, we let the whole family know about the condition. What really amazed me is the completely varied level of interest from our relatives.

My mom was a physician, and we discussed everything from my children’s diagnoses to their treatment. But I have a couple sisters who have not asked me anything about it, so we have not told them anything. I do not think they are aware they might be carriers like I am. I do not understand why they are not curious. I expected them all to be worried and to want to understand, but it is their choice.
If family members remain unwilling to talk about it, respect their wishes. Try to understand that it is not unusual for family members to react to the news differently.

Let them know you are available to talk when they are ready and if they have questions, and give them some resources for when they are ready to learn more [refer to page 23 for resources].

When family members do not want to talk about Gaucher, you might feel upset or alone. Seek support from doctors, friends, other family members, or other people you know with Gaucher.

The National Gaucher Foundation Mentor Program is another good place to turn to connect with others in the Gaucher community for information, advice and support. There are also online groups with Gaucher patients from around the world ready to listen, help and welcome newly diagnosed Gaucher patients and family.

We do not really share information with people unless they ask. We never hid anything. We found that some people just do not want to know. For those who want to know, we explain absolutely everything.

My wife’s family was very curious; her sister-in-law asked lots of questions about the condition to find out how her children might be at risk. My wife’s sister even helped us start an organization similar to the National Gaucher Foundation in our country, Venezuela.