

Volume 13, Number 1

# HORIZONS

A Newsletter for the Gaucher Community

**SPECIAL EDITION**

## Resources for the Gaucher Community

- Websites
- Patient Support
- Understanding the Family Tree

Introducing [www.gauchercare.com](http://www.gauchercare.com) on page 13

[www.gaucherdisease.org](http://www.gaucherdisease.org)  
[www.cerezyme.com](http://www.cerezyme.com)



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## PUBLISHING NOTE

In this special edition of HORIZONS, we present a wide array of information for the Gaucher community, including the introduction of Genzyme's newest online resource, [www.gauchercare.com](http://www.gauchercare.com). This website, like much of the other content of this issue, is designed to help you and your family learn more about Gaucher disease, treatments, and the resources that are available to you.



This special issue was developed in response to your feedback, requesting that one issue of HORIZONS be devoted to this type of information.

In the following pages, you'll find a collection of articles and resources on Gaucher disease. We hope that this information may help you discuss Gaucher disease more comfortably with your health care providers, friends, and family.

—Your team at Genzyme

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# expression of hope

## A Global Program of Goodwill and Awareness.

Throughout this issue of HORIZONS, we highlight works from the Expression of Hope exhibit, a worldwide program created as a means to generate awareness and understanding of the incredible strength and courage of the thousands of people living with lysosomal storage disorders (LSDs). Numerous patient organizations from around the world, such as the National Gaucher Foundation (NGF), became involved in Expression of Hope and encouraged their members to submit artwork which shares their feelings of hope and explores the realities, perceptions, and individual experiences of living with an LSD, such as Gaucher disease.

In collaboration with the Center for Art and Community Partnerships at Massachusetts College of Art, in Boston, Massachusetts, U.S.A., Expression of Hope also invited eleven artists to participate in the program and to create a total of 15 artworks representing their individual collaboration with patients around the world.

Artists and patients spent many hours in virtual collaboration with each other via phone, video conference and email in order to foster a mutual understanding of how to best capture and express the essence of the patients' experiences in living with an LSD. If you would like to learn more about this program or view the artwork, visit [www.expressionofhope.com](http://www.expressionofhope.com).



# Gaucher Disease Overview

## What is Gaucher Disease?

Gaucher (pronounced go-shay) disease is an inherited genetic condition that causes fatty deposits to build up in the cells of certain organs and bones. It can cause a wide variety of symptoms. Gaucher disease affects fewer than 10,000 people worldwide.

**Gaucher Cells.** Our bodies contain thousands of active substances called enzymes. The enzyme glucocerebrosidase (pronounced **gloo-ko-ser-e-bro-sy-daze**) breaks down a certain type of fat molecule (glucocerebroside). In Gaucher disease, there is not an adequate supply of the affected enzyme. As a result, the lysosomes of cells (specialized compartments in the cell that contain enzyme) fill up with undigested fat. These cells are referred to as Gaucher cells.

**Disease History.** Gaucher disease was first described in 1882 by the French physician Philippe Charles Ernest Gaucher upon autopsy of a 32-year-old woman with an enlarged spleen.

## The Three Types of Gaucher Disease

Experts have identified three different types of Gaucher disease:

**Type 1 (non-neuronopathic).** The most common form, Type 1 affects an estimated one in 40,000 to 60,000 individuals in the general population. Type 1 does not affect the brain or nervous system. Some patients with Type 1 Gaucher disease have no symptoms, while others develop serious consequences that can be life-threatening.



ARTIST: Alice Boynton

*"My career as a textile artist was cut short by Gaucher disease, and I switched from an art studio to a home studio. Each phase of my disease helped to shape my career into the most enjoyable outcome imaginable. I now paint watercolors and oils of still lifes, portraits and landscapes, a positive outcome from a disease."*

**Type 2 (acute neuronopathic).** Type 2 Gaucher disease is more rare, affecting fewer than one in 100,000. However, people with Type 2 generally have more severe effects than those with Type 1. Children develop signs and symptoms of Type 2 Gaucher disease within the first year of life and may suffer major neurological problems as well as other symptoms. Most of these patients do not live past age two.

**Type 3 (chronic neuronopathic).** Type 3 is also rare and affects fewer than one in 100,000 people. This form also causes neurological signs and symptoms, but they are generally less severe than those caused by Type 2 Gaucher disease. Signs and symptoms appear in early to late childhood, and patients with Type 3 Gaucher disease can live well into adulthood.

## Who Gets Gaucher Disease?

Gaucher disease is inherited, or caused by genes passed down from

an individual's parents. To learn more about how Gaucher disease is inherited, see "Understanding the Family Tree" on page 4. Gaucher disease is not gender-specific and its signs and symptoms may appear in affected individuals at any age, although Types 2 and 3 are most commonly diagnosed in childhood. Although individuals from any ethnic or racial background may develop Gaucher disease, Type 1 Gaucher disease is most prevalent among Jews of Ashkenazi (Eastern European) descent. Among this group, one in 450 people has Gaucher disease.

## Signs and Symptoms of Type 1 Gaucher Disease: A Self-Evaluation

The following self-evaluation highlights the signs and symptoms of Gaucher disease. If you experience some of these symptoms, it may be important to notify your physician or health care provider.

- I experience bone and/or joint pain.
- My stomach feels full quickly with small portions.
- My abdomen is distended.
- I have an enlarged liver and spleen.
- I am very tired most of the time and require frequent naps.
- I have anemia.
- I have a family member who has been diagnosed with Gaucher disease.
- I avoid physical activities due to skeletal pain and/or fatigue.
- I bleed easily and it is difficult to stop.
- I bruise easily.

# Awareness of Gaucher Disease: Understanding the Family Tree

**G**aucher disease affects people from all ethnic groups. However, it is more common in Jewish people with ancestors from Eastern Europe.

Because Gaucher disease is rare and causes a wide variety of symptoms, its diagnosis can be challenging. Individuals may experience symptoms of the disease a long time before an accurate diagnosis is made. This is a concern because the longer a person has Gaucher disease without treatment, the more damage is likely to occur. The earlier the disease is diagnosed, the earlier it can be managed, possibly preventing further health problems.

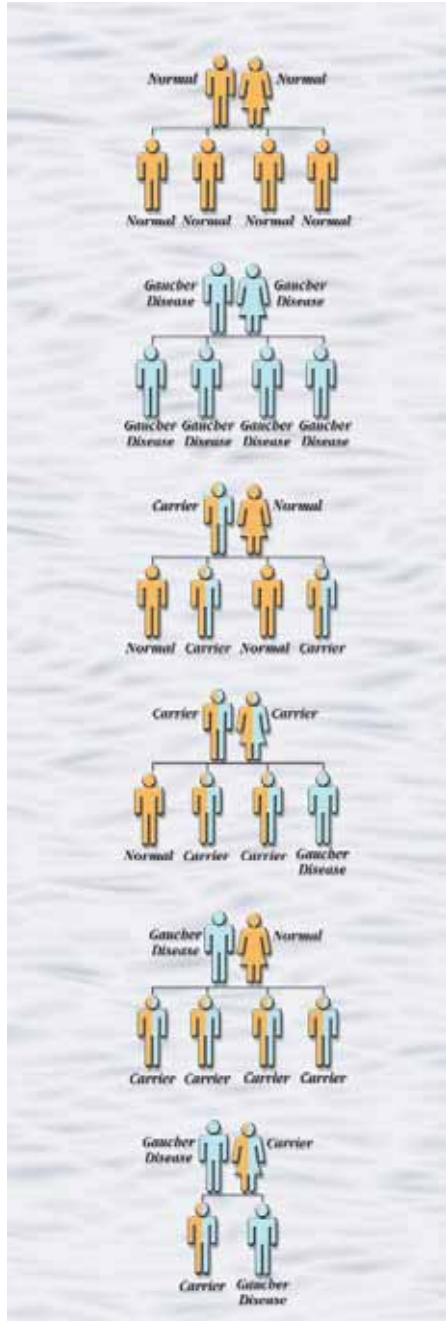
Gaucher disease is inherited, and family members of people with the disease are also at risk for having it or are likely to be carriers of the Gaucher gene. To have Gaucher disease, you must inherit two copies of the Gaucher gene—one from your mother and one from your father. People who have only one copy of the Gaucher gene are carriers. Carriers do not display signs and symptoms of Gaucher disease and usually do not know that they are carriers, but can pass the Gaucher gene on to their children. If a mother and father are both carriers, both can pass on the abnormal gene, and the child would then have Gaucher disease.

One way to increase the likelihood of early diagnosis is to understand who is at risk for developing Gaucher disease. When one person in a family has the disease, others may also be at risk. A family medical tree is a record

of health and illness within a family. It resembles the family tree you might draw to record your genealogy, except that it also includes medical information. It can

help you understand how Gaucher disease has affected relatives, both living and deceased, as well as how the gene may affect future generations.

The accompanying chart on the next page is designed to help you gather and organize relevant information about your family members and their status as potential carriers of Gaucher disease. Together with a knowledge of inheritance patterns, it will help you to better understand how Gaucher disease occurs and how it is passed along. Share this family tree chart with your genetic counselor, health care provider, or a Genzyme Patient Care Liaison to better understand you and your family's risk factors for Gaucher disease.



ARTIST: Grant Bowen

"I am 11 years old. I am a swimmer and play basketball and take art once a week. My painting is a bunch of bright flowers. I really like to use bright colors. I am trying to be an artist when I grow up."

# The Medical Family Tree

Creating a medical family tree can help you understand the inheritance pattern in your family and your risk of passing on Gaucher disease. You can fill out this chart to get started. A medical genetic counselor or a Genzyme Patient Care Liaison (see the article on page 12 regarding Patient Care Liaisons) can assist you from there. Complete the information about yourself first, and then be sure to fill in both sides of your family. We encourage you to cut this chart out and share it with your family and health care providers.

Your Family Tree

### GRANDMOTHER

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

### GRANDFATHER

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

### AUNTS/UNCLES

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

### YOU

### AUNTS/UNCLES

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

### MOTHER

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

### CHILDREN

### FATHER

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

### BROTHERS

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

### SISTERS

### SISTERS

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

### COUSINS

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

### COUSINS

Initials \_\_\_\_\_ State of Residence \_\_\_\_\_

Year of Birth \_\_\_\_\_ Living  Yes  No

Gaucher Disease  Yes  No  Don't know

Carrier  Yes  No  Don't know

Genotype (if known) \_\_\_\_\_

# Celebrating 16 Years of Serving the Gaucher Community

In 2007, Genzyme celebrates 16 years of serving the Gaucher community. Founded in 1981, Genzyme is one of the world's largest biotechnology companies. Since the beginning, Genzyme has been committed to developing innovative solutions for major unmet medical needs. Genzyme currently has many established products and services helping patients in nearly 90 countries. For the Gaucher community, some of those services include: The Gaucher Registry, the Registry Patient Case Report, Personal Treatment Trackers, Case Managers, Patient Care Liaisons, and some excellent online resources (see pages 13-14).

In 1991, Genzyme introduced Ceredase® (alglucerase injection) worldwide as the first product to treat a lysosomal storage disorder. During that year, the Food and Drug Administration granted marketing approval for Ceredase in the United States to treat Type 1 Gaucher disease. By the end of 1991,

several hundred patients had been receiving Ceredase treatment. In 1994, Cerezyme® (imiglucerase for injection), a genetically engineered form of Ceredase, became available for Type 1 Gaucher patients.

Over the past 16 years, Ceredase and Cerezyme therapy have helped over 5,000 Gaucher patients. Cerezyme has the longest track record for helping Gaucher patients and is indicated to treat Type 1 Gaucher patients who have one or more of the following conditions:

- Anemia (low red blood cell count)
- Low platelet count
- Spleen enlargement
- Liver enlargement
- Bone disease

Cerezyme therapy has been shown to be safe and effective and helps relieve or reverse many of the signs and symptoms of Type 1 Gaucher disease. However,

treatment experiences may vary. Cerezyme therapy is not a cure for Gaucher disease: it does not correct the underlying genetic defect. In order to continue to benefit from the treatment, symptomatic patients must receive intravenous infusions for the rest of their lives, even though they may feel better. That's because, once therapy stops, Gaucher cells may build up again and symptoms may come back.

Cerezyme is made in a sophisticated biopharmaceutical manufacturing facility, using a biotechnology scientific process that utilizes Chinese Hamster Ovary (CHO) cells that have been genetically modified to produce human glucocerebrosidase (GCR). CHO cells are used in over 70 biopharmaceutical products such as insulin for diabetic patients, human growth hormone for growth disorders, erythropoietin (EPO) for anemia, and factor VIII for patients with blood clotting disorders.

## IMPORTANT SAFETY INFORMATION

Side effects related to Cerezyme® (imiglucerase for injection) administration have been reported in less than 15% of patients. Each of the following events occurred in less than 2% of the total patient population. Reported side effects include nausea, vomiting, abdominal pain, diarrhea, rash, fatigue, headache, fever, dizziness, chills, backache, and rapid heart rate. Because Cerezyme® therapy is administered by intravenous infusion, reactions at the site of injection may occur: discomfort, itching, burning, swelling or uninfected abscess. Symptoms suggestive of allergic reaction include anaphylactoid reaction (a serious allergic

reaction), itching, flushing, hives, an accumulation of fluid under the skin, chest discomfort, shortness of breath, coughing, cyanosis (a bluish discoloration of the skin due to diminished oxygen), and low blood pressure. Approximately 15% of patients have developed immune responses (antibodies); periodic monitoring by your physician is suggested. Patients should notify their physician immediately if they experience any side effects with treatment. For more information, consult your physician. To learn more, please see full product information; contact Genzyme at 800-745-4447, or visit [www.cerezyme.com](http://www.cerezyme.com). Cerezyme is available by prescription only.

## The Gaucher Registry

Disease registries are large, often multi-national observational databases that collect clinical data on patients with a particular disease. By collecting, analyzing, and disseminating data on disease progression and patient responses to long-term disease management strategies, registries may help improve disease understanding and keep medical professionals up-to-date on the latest advances. Registries can also help patients better understand their treatment goals.

Sponsored by Genzyme, the Gaucher Registry is the first and world's largest cooperative observational database on Gaucher disease. The International Collaborative Gaucher Group (ICGG) established the Registry in 1991 to track Gaucher patient outcomes of routine clinical practice. All Gaucher patients are eligible for inclusion. By enrolling in the Registry, your physician will receive reports on your care to monitor your disease status and be able to access information on current treatment guidelines and practice patterns.

The Gaucher Registry maintains its integrity by a commitment to patient privacy and confidentiality, as well as a commitment to the quality of the data. To ensure the most comprehensive data, the Gaucher Registry is open to all patients with Gaucher disease, regardless of treatment modality. To learn more about being enrolled in the Gaucher Registry, speak to your doctor and visit [www.gaucherregistry.com](http://www.gaucherregistry.com).



# The Importance of an Annual Evaluation: A Patient's Story

Many of you know Warren McCollom, and many more have heard or read his story. In fact, Warren wrote about his experiences with Gaucher disease in a previous edition of *HORIZONS*. Warren's story is interesting in that his treatment has evolved over the years. Warren was diagnosed in 1974, well before there was an available treatment for type 1 Gaucher disease. He was very proactive in finding doctors who were familiar with Gaucher disease. He also educated himself about the disease and discovered (by reading a letter to the editor written by Dr. Roscoe Brady in *Good Housekeeping* magazine) that research was being done to develop a treatment for type 1 Gaucher disease. Warren has continued to be an advocate for his own health care over the years.

Warren started Ceredase® (alglucerase injection) treatment in 1991. After a few months, it was clear that Warren was responding to treatment. His liver and spleen reduced in size, and his anemia and platelet count improved. As a result, Warren went on a *drug holiday*.

However, during Warren's drug holiday, his symptoms returned and some worsened. Warren soon reinitiated therapy and was enrolled in the Gaucher Registry. He began receiving annual comprehensive evaluations, his



WARREN MCCOLLOM

dose and the frequency of treatments were adjusted based on whether or not he was achieving his therapeutic goals. Warren currently receives weekly Cerezyme® (imiglucerase for injection) infusions. He does not let this stop him from living his life and even fits in an annual trip to his hometown in Illinois for two to three weeks every summer. Warren works with his doctor to schedule treatments around this trip.

Warren encourages all patients with Gaucher disease to have annual comprehensive evaluations, whether on treatment or not. He also suggests you review the results of tests with your doctor and request copies of all your medical records. He recommends you follow your doctor's treatment recommendations and talk with him/her well in advance of taking any trips that exceed the interval of your specific treatment schedule.

*Warren is an active member of the National Gaucher Foundation and is always happy to talk with other patients. You can contact Warren through his email: [wlmccollom@earthlink.net](mailto:wlmccollom@earthlink.net)*

Treatment responses may vary. Please see important safety information about Cerezyme on page 15.

## Tips for Adhering to Treatment

Patients being treated with Cerezyme® (imiglucerase for injection) are more likely to benefit when they understand the disease and the treatment process. Before treatment begins, patients and family members may want to be counseled about infusion therapy, treatment-associated risks, possible therapeutic outcomes, and the need to keep infusion appointments. For some patients, it may be difficult to accept that treatment will be needed throughout their life. However, adhering to a treatment schedule is an essential component of taking control of one's health. Some tips that may assist patients in adhering to treatment include:

- Maintaining an open dialogue with your doctor and other health care providers.
- Talking with individuals with Gaucher disease about their experiences and contacting Gaucher patient associations, support groups, and education programs to learn more.
- Talking with a counselor.
- Rewarding yourself for sticking with treatment.
- Making treatment more enjoyable for children, by reading books or playing games with individuals being treated.
- Keeping a diary to monitor changes in your health.
- Following progress and check for improvements over treatment and maintenance of therapy.
- Planning for trips in advance, including working with your Genzyme Case Managers to ensure uninterrupted treatment.
- Using the Personal Treatment Tracker on page 10.

# Patient Case Reports and Therapeutic Goals

The Gaucher Registry Patient Case Report (PCR) is an important component of the Gaucher Registry and provides a useful summary of all of the key evaluations that have been part of a patient's comprehensive monitoring for Gaucher disease. It is a useful tool for tracking the progression of disease. It may also help to ascertain when the patient's next evaluations need to be scheduled according to the Recommended Schedule of Assessments. The PCRs are provided in both a table and graph format, allowing easy review of outcomes. The data contained in the report include the following information:

- Cerezyme® (imiglucerase for injection) status (if applicable)
- Blood and other lab values
- Liver and spleen volume
- Skeletal involvement
- Quality of life

The graphs (on the next page) provide a simple way to evaluate trends over time in individual parameters. For instance, a downward slope on the hemoglobin or platelet count graph would indicate that these parameters were deteriorating and perhaps re-evaluation by the physician may be necessary.

For patients enrolled in the Registry, the Registry Patient Case Report enables the physician,

nurse, genetic counselor, and patient to monitor response to therapies and to assess whether the patient is meeting individual therapeutic goals. This report provides an opportunity to review treatment history and outcomes.

The PCR provides a summary of the patient's disease course and may be a useful tool for reviewing the patient's disease status with the physician and other health care providers.

Treatment goals are included on the PCR, and this information allows the patient and health care providers to compare the patient's current disease status with these goals.

## Bone Disease

### Treatment Goals

Patients	Goals	Timeframe
All patients	<ul style="list-style-type: none"> <li>■ Lessen or eliminate bone pain</li> <li>■ Prevent bone crises</li> </ul>	Years 1 to 2

Treatment response may vary.

Patients with advanced bone disease may require orthopedic intervention, physical therapy and/or other medications.

Development and maintenance of therapeutic goals for patients with Type 1 Gaucher disease requires an integrated approach. All patients should receive an initial comprehensive assessment of potential disease compartments: anemia, thrombocytopenia, hepatomegaly, splenomegaly, bone disease, growth retardation in pediatric patients, pulmonary involvement, functional health and well-being, and a thorough physical examination and assessment of biomarkers. The information obtained should then be used to establish therapeutic goals for all disease compartments.

The specific therapeutic goals for bone disease are to lessen or eliminate bone pain and bone crises within 1 to 2 years of treatment. The onset or development of bone pathology during treatment should prompt investigation for loss of mechanical bone integrity requiring changes in therapy with orthopedic intervention.

## Your Doctor's Role in Helping to Maximize Your Health





# Patient Resources

The following pages contain information to help you and your family learn more about Gaucher disease and the resources that are available to you.

Personal Treatment Tracker									
TEST	How often to have tests Acheived goals?		Track your treatment Year _____			Next DUE DATE	Track your treatment Year _____		
	If NO have the test EVERY:	If YES have the test EVERY:	Your test DATES	Your test RESULTS	Achieved your goals?		Your test DATES	Your test RESULTS	Achieved your goals?
Initial exam					<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
Complete checkup	12 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
<b>BLOOD TESTS</b>									
Hemoglobin	3 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
Platelet count	3 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
<b>SPLEEN/LIVER TESTS</b>									
Spleen volume	12 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
Liver volume	12 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
<b>BONE TESTS</b>									
MRI	12 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
X-ray	12 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
DEXA	12 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
<b>BIOCHEMICAL MARKERS</b>									
Chitotriosidase	3 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
ACE	3 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
TRAP	3 months	12-24 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N
<b>OTHER TESTS</b>									
SF-36® Health Survey	12 months	12 months			<input type="checkbox"/> Y <input type="checkbox"/> N				<input type="checkbox"/> Y <input type="checkbox"/> N

Note: This tracker highlights some of the minimum recommendations for routine monitoring. Depending on your health status, you may need additional or more frequent tests and exams. Be sure to talk with your doctor about these tests and any symptoms you may experience. To receive an enlarged copy of the Treatment Tracker, please order the Gaucher Patient Resource – 7 Tests brochure by calling 800-745-4447.

## HOW TO USE THIS TRACKER:

- Bring a copy of this treatment tracker to your next health care provider visit.
- Discuss these tests with your health care provider.
- Write down the dates of your tests and the results provided to you by your health care provider.
- Talk with your health care provider about whether your goals have been reached for each test.
- Mark a YES or NO in the *Achieved Your Goals?* Column.
- Compare results from visit to visit.
- Write down when you need to have these tests again in the *Next Due Date* column.
- Keep this tracker in a safe place and bring it with you to each health care provider visit.

# Genzyme Patient Resources

These support materials can be ordered by calling Genzyme Medical Information at 800-745-4447 (option 2) or by visiting [www.cerezyme.com](http://www.cerezyme.com).

## **Living with Gaucher Disease: A Guide for Patients, Parents, Relatives, and Friends**

This brochure describes (in patient-friendly language) Gaucher disease and the genetics, testing, diagnosing, and coping with this disorder. It is designed for families and patients who have been diagnosed with Type 1 Gaucher disease. Also available in Spanish.

## **Question and Answer Guide: A Guide for Individuals with Type 1 Gaucher Disease**

This brochure answers most frequently asked questions about Cerezyme® (imiglucerase for injection), including important safety information, dosing, and administration. The brochure is written in patient-friendly language and has a small resource section and glossary.

## **Inheritance of Gaucher Disease: Straight Talk for Patients and Families**

This brochure describes Gaucher disease and how it is inherited. It is written in patient-friendly language for families and patients who suspect or have been diagnosed with Gaucher disease. The brochure is also available in Spanish.

## **Making a Difference — Talking about Gaucher Disease**

This kit is designed to help patients better understand and discuss Gaucher disease. This information

will assist patients in communicating about Gaucher disease and will help them to teach others about the disease.

## **A Young Couple's Guide to Jewish Genetic Disorders and Screening**

This resource guide gives newly engaged couples information about disorders (such as Gaucher disease) that have high frequency in the Jewish population.

## **Type 1 Gaucher Disease: Setting Goals to Help Maximize Your Health. 7 Tests to Help You Get the Most from Your Treatment**

This brochure provides information to patients to help set specific goals and targets for treatment that will help patients and doctors monitor progress. The tests and treatment goals discussed in this booklet come from a set of guidelines developed by Gaucher Registry disease experts from around the world. Also available in Spanish.

## **Genzyme Treatment Support**

This brochure is designed for those living with Gaucher disease or any other lysosomal storage disorder. This brochure also describes Genzyme Treatment Support and case management services and answers frequently asked questions about health insurance.

## **GIGI and Type 1 Gaucher Disease**

This brochure presents the story of Gigi and can be used as a tool for parents or guardians to use when explaining Type 1 Gaucher disease and treatment to their child.

## **A Message to Elijah: An Educational Video on Type 1 Gaucher Disease and its Treatment**

This DVD shares the personal stories of Gaucher patients and is narrated by actor Elliott Gould. The video was created and produced by patient Michael Margolis and was a joint effort of the National Gaucher Foundation and Genzyme.

### **ANNOUNCEMENT:**

## **The National Gaucher Foundation Opens a National Office**

**National Office of the National Gaucher Foundation  
2227 Idlewood Road Suite 12  
Tucker, GA 30084**

**Toll Free: 800-504-3189 • Local: 770-934- 2910  
Fax: 770-934-2911  
[www.gaucherdisease.org](http://www.gaucherdisease.org)**



Please see the accompanying full Product Information for Cerezyme® (imiglucerase for injection)

## Genzyme Case Managers

There are approximately 40 case managers at Genzyme who act as patient advocates for individuals with lysosomal storage disorders, including Gaucher disease. Lee Ann Chavez, Senior Case Manager at Genzyme Patient and Product Services explains, "As case managers, our goal is to try to minimize the obstacles to care. We assist people with many challenges including insurance and other issues, whether or not Cerezyme® (imiglucerase for injection) is recommended for the treatment of the disease, we provide information and support to them. We try to help them understand their insurance coverage or try to help them understand what insurance options are available in their area."

Case management specialists are employees of Genzyme and may assist individuals in many ways including:

- Provide information about Type 1 Gaucher disease and the risks and benefits of Cerezyme therapy
- Educating individuals about available health insurance and helping them review their policies
- Coordinating the exchange of information between physicians, insurance companies, and patients to obtain coverage approval for Cerezyme
- Serving as a resource to individuals and their families who are seeking alternative funding options

Case managers become experts in the regions in which they work and are able

to solve problems unique to that state or area. For example, Ms. Chavez says, "In Florida, I work with an aging population, many of whom need to travel long distances to treatment centers. I may assist them by identifying infusion sites closer to home or addressing issues related to transportation."

For people with Type 1 Gaucher disease receiving Cerezyme, understanding how an insurance plan covers the Cerezyme infusions is important. Some insurance plans may cover Cerezyme as an outpatient service under the major medical policy or under a prescription plan. A case management specialist can help determine insurance coverage. Before beginning treatment, it may be necessary for a doctor to get written confirmation of coverage before treatment begins. A case management specialist can work with the physician's office to obtain approval.

Genzyme is committed to ensuring that people who are prescribed Cerezyme therapy will have access to it. To help you through the insurance process, call Genzyme Treatment Support at 800-745-4447. A case management specialist will assist you in reviewing your current insurance coverage and, if necessary, try to obtain and maintain the insurance coverage you may need for long-term Cerezyme treatment. This service is voluntary and the information required to perform this service will be maintained as confidential by Genzyme.



LEE ANN CHAVEZ



ARTIST: Naomi Arai

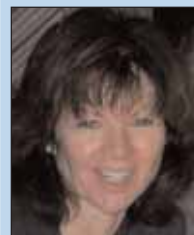
"I had severe bone problems in my hip joint since childhood, so I could not play outside or do any physical activity. For that reason, I love drawing. I would love to express my appreciation to all the people who support me with my drawing. In this work, I expressed about myself being free but physically challenged and also tried to express the strength, mercy, beauty and frailty of life in my own image."

## Genzyme Patient Care Liaisons

A priority of Patient Care Liaisons (PCLs) is to empower individuals living with lysosomal storage disorders. Erin O'Rourke, MS, CGC, a PCL from Genzyme Therapeutics' Mid Atlantic Region, says she and other PCLs achieve this goal in a number of ways, but most importantly by "educating patients about their disease. We listen to what patient's challenges are and provide assistance where we can."

Since lysosomal storage diseases are rare, PCLs spend time educating health care professionals and the community regarding these diseases. PCLs also work with physicians, nurses, genetic counselors and other providers to optimize patient care.

Gaucher disease is a progressive, life-long condition that affects each person differently in terms of severity and rate of progression. PCLs stress the importance of comprehensive evaluations to patients and encourage patients



ERIN O'ROURKE

to work with physicians to understand, set, achieve and maintain treatment goals. In the best case scenario, patients play an active role in their care by doing the following:

- Ask your physician about the results of your tests and learn how to compare them to previous tests
- Talk to your physician about achieving treatment goals
- Stick with your treatment schedule
- Maintain copies of all your medical records

# Gaucher Disease-Related Web Links

These links contain information and support from professional medical organizations, hospitals, national and international Gaucher disease groups, patient support organizations, Jewish organizations, medical centers specializing in Gaucher disease care and research, government sites, as well as Genzyme-sponsored websites.

These web pages and their content are maintained by the organizations listed below. With the exception of Genzyme websites (\*), Genzyme does not endorse any particular organization or the content contained on its website.

## Gaucher Disease Resources

### \*Cerezyme.com

Genzyme created this website for people living with or caring for someone with Gaucher disease. This site provides supportive information about the condition and treatment of Type 1 Gaucher disease. The site also provides links and other important information on Gaucher disease.  
[www.cerezyme.com](http://www.cerezyme.com)

### Children's Gaucher Research Fund

The Children's Gaucher Research Fund is a non-profit organization that raises funds to support research aimed at finding a cure for Gaucher disease as well as providing support to families who battle this disease.  
[www.childrensgaucher.org](http://www.childrensgaucher.org)



### \*GaucherCare.com

Gaucher Care is a new online resource for patients and healthcare professionals. This comprehensive website is designed to help patients and their families learn more about Gaucher disease and resources that are available. This information may also help patients discuss Gaucher disease more comfortably with their physicians and other health care professionals.  
[www.gauchercare.com](http://www.gauchercare.com)

### National Gaucher Foundation (NGF)

The National Gaucher Foundation (NGF) is a patient organization that offers a variety of services and programs for the Gaucher community. This includes free public educational materials such as posters and brochures with display holders.  
[www.gaucherdisease.org](http://www.gaucherdisease.org)

### \*Gaucher Registry

In addition to helping understand the natural progression of Gaucher disease, the Gaucher Registry is designed to help physicians understand the long-term effects of treatment.  
[www.gaucherregistry.com](http://www.gaucherregistry.com)

### International Gaucher Disease Association

This international site provides over 200 links for patients with Gaucher disease and their doctors.  
[www.gaucher.org.uk/overseas.htm](http://www.gaucher.org.uk/overseas.htm)

### \*Lysosomal Learning

Online resource for information about lysosomal storage disorders and the science behind the diseases.  
[www.lysosomallearning.com](http://www.lysosomallearning.com)

## General Genetic and Disease

### American Society of Hematology

The mission of the American Society of Hematology is to further the understanding, diagnosis, treatment, and prevention of disorders affecting the blood, bone marrow, and the immunologic, hemostatic and vascular systems, by promoting research, clinical care, education, and training.  
[www.hematology.org](http://www.hematology.org)

### ClinicalTrials.gov

This site provides regularly updated information about federally and privately supported clinical research in human volunteers.  
[www.clinicaltrials.gov](http://www.clinicaltrials.gov)



### GeneTests

The GeneTests website is a publicly funded medical genetics information resource developed for physicians, other healthcare providers, and researchers, available at no cost to all interested persons.  
[www.geneclinics.org](http://www.geneclinics.org)

### Genetic Alliance

Genetic Alliance increases the ability of genetic advocacy organizations to achieve their missions and leverages the voices of millions of individuals and families living with genetic conditions.  
[www.geneticalliance.org](http://www.geneticalliance.org)

### Global Organization for Lysosomal Diseases

The Global Organization for Lysosomal Diseases is an international collaboration of scientific and medical associations, patient groups, and commercial organizations dedicated to improving the lives of all patients with a lysosomal disease.  
[www.goldinfo.org](http://www.goldinfo.org)

### Health Care Information Resources

This site provides information about health and disease to better inform consumers. This service does not offer advice about health or health care and cannot substitute for a health care practitioner.  
[hsl.mcmaster.ca/tomflem/top.html](http://hsl.mcmaster.ca/tomflem/top.html)

### National Organization for Rare Disorders

The National Organization for Rare Disorders is dedicated to helping people with rare diseases and assisting the organizations that serve them.  
[www.rarediseases.org](http://www.rarediseases.org)

## National Society of Genetic Counselors

The National Society of Genetic Counselors promotes the professional interests of genetic counselors and provides a network for professional communications.

[www.nsgc.org](http://www.nsgc.org)

## National Tay-Sachs & Allied Diseases Association

The National Tay-Sachs & Allied Diseases Association is dedicated to the treatment and prevention of Tay-Sachs, Canavan and related genetic diseases, and to providing information and support services to individuals and families affected by these diseases, as well as the public.

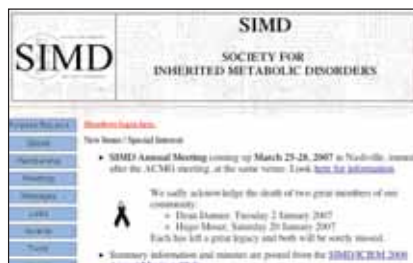
[www.ntsad.org](http://www.ntsad.org)



## Society for Inherited Metabolic Disorders

The Society for Inherited Metabolic Disorders was founded to develop a group of metabolic experts who could advise the government on medical treatment of inborn errors of metabolism and to create centers for the diagnosis and treatment of patients with metabolic diseases.

[www.simd.org](http://www.simd.org)



## Office of Rare Diseases

The goals of the Office of Rare Diseases are to stimulate and coordinate research on rare diseases and to support research to respond to the needs of patients who have any one of the more than 6,000 rare diseases known today.

[rarediseases.info.nih.gov](http://rarediseases.info.nih.gov)

## Jewish Organizations, Support Groups, and Genetic Disease Sites

### Bikur Cholim

Bikur Cholim is a multi-faceted medical and health care related agency dedicated to helping patients and their families receive the full complement of health care services.

[www.bikurcholim.org](http://www.bikurcholim.org)

### Center for Jewish Genetic Diseases

The Center for Jewish Genetic Diseases at The Mount Sinai Medical Center in New York City is the first center in the world devoted to the study of diseases that affect Ashkenazi Jews.

[www.mssm.edu/jewishgenetics](http://www.mssm.edu/jewishgenetics)

### Chai Lifeline

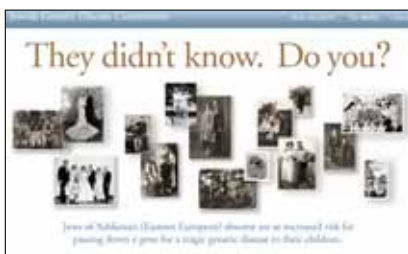
Chai Lifeline is dedicated to helping children suffering from serious illness as well as their family members.

[www.chailifeline.org](http://www.chailifeline.org)

### Victor Center for Jewish Genetic Diseases at Albert Einstein Medical Center

In this section of Albert Einstein Medical Center website, you will find information on Keyv K. and Hortense M. Kaiserman Ashkenazi Jewish Genetic Disease Screening Program.

[www.einstein.edu/jewishgenetic](http://www.einstein.edu/jewishgenetic)



### Jewish Genetic Disease Consortium

The Jewish Genetic Disease Consortium was created as a means by which a number of smaller, individual organizations could join together to heighten awareness of Jewish genetic diseases with a strong and unified voice.

[www.jewishgeneticdiseases.org](http://www.jewishgeneticdiseases.org)

## Chicago Center for Jewish Genetic Disorders

The Chicago Center for Jewish Genetic Disorders is a critical effort to provide public and professional education and to empower community members to seek out information and prevention strategies.

[www.jewishgeneticscenter.org](http://www.jewishgeneticscenter.org)



## Hadassah

Hadassah, the Women's Zionist Organization of America, is a volunteer women's organization that focuses on health issues.

[www.hadassah.org](http://www.hadassah.org)

## Jewish Genealogical Society

The Jewish Genealogical Society is a nonprofit organization dedicated to collecting, preserving, and disseminating knowledge about Jewish genealogy.

[www.jgsny.org](http://www.jgsny.org)

## Jewish Genetic Diseases: A Mazornet Guide

This website compiles helpful information and resources concerning Jewish genetic diseases.

[www.mazornet.com/genetics](http://www.mazornet.com/genetics)

## Rofeh International

Rofeh International provides medical referral and support services to the needy.

[www.rofeh.org](http://www.rofeh.org)

## Russian American Medical Association

Russian American Medical Association was created to facilitate and enable Russian American physicians and other health care professionals to excel in patient care, teaching and research, and to pursue their aspirations in professional, humanitarian, and community affairs.

[www.russiandoctors.org](http://www.russiandoctors.org)

# Raising Disease Awareness: Tips for Patients

**A**wareness of storage disorders such as Gaucher disease remains very low. According to the National Gaucher Foundation (NGF), more than nine out of 10 Jewish Americans are unaware of Gaucher disease. As referenced in earlier articles (pages 2 and 3), Jewish people of Eastern European (Ashkenazi) heritage have an increased risk of passing down a gene for Gaucher disease to their children. Approximately one in 450 people in this population may have the disorder, and the carrier rate is approximately one in 14. As a comparison, one in 40,000 to 60,000 people in the general population have Gaucher disease.

Knowing your heritage remains one of the most significant steps in raising one's awareness of Gaucher disease and its risk factors. Testing is also important and may help to prevent serious health issues, unnecessary pain, and distress. Gaucher disease can be detected through a simple blood test. However, Gaucher disease may not be included in all Jewish genetic disease testing panels, including carrier screening. People must ask for it, but they can't ask if they do not know about the disease.



ARTISTS: Troy Hagenbart / Jen-Hao Hsiao

Troy: "I was struck when Jen-Hao told me he had to travel for his treatment and would do anything for his health. This shows not only strength and courage but a knowledge that we should always be grateful for the things that truly make up our life."

Jen: "I like this image very much. It is quite different from what I imagined. I am very pleased with Troy's interpretation and am excited about the program. I hope to see the exhibition and what's next."

Gaucher patients need to know the importance of timely diagnosis and treatment. The genetic component of the disease means that family members are at much greater risk of the disease than the general population.

Patients and family members can help in spreading the word about Gaucher and other genetic diseases. The following list was adapted from the NGF and highlights several simple activities that patients and family can do to help raise awareness:

- Send a letter to the American College of Obstetricians and Gynecologists urging them to include carrier testing for Gaucher disease in standard panels.
- Schedule talks by Genetic Counselors or Patient Care Liaisons (see page 12) at local organizations.
- Encourage at-risk young couples involved in family planning to get tested.
- Advise family members of patients to go to area testing/treatment centers.
- Pass out Gaucher disease materials at health-related events.
- Show educational videos at health-related events.
- Inform immediate and extended families of patients, and consider filling out the family tree on page 5.
- Present Gaucher information in a way that is not alarming, stressing the value of early diagnosis.
- If you are Jewish, ask your synagogue, Jewish Community Center, or related organization to disseminate information about the disease.
- If you are a patient, your first-hand story will be taken seriously when shared with others.

## IMPORTANT SAFETY INFORMATION

Side effects related to Cerezyme® (imiglucerase for injection) administration have been reported in less than 15% of patients. Each of the following

events occurred in less than 2% of the total patient population. Reported side effects include nausea, vomiting, abdominal pain, diarrhea, rash, fatigue, headache, fever, dizziness, chills, backache,



and rapid heart rate. Because Cerezyme® therapy is administered by intravenous infusion, reactions at the site of injection may occur: discomfort, itching, burning, swelling or uninfected abscess. Symptoms suggestive of allergic reaction include anaphylactoid reaction (a serious allergic reaction), itching, flushing, hives, an accumulation of fluid under the skin, chest discomfort, shortness of breath, coughing, cyanosis (a bluish discoloration of the skin due to diminished oxygen), and low blood pressure. Approximately 15% of patients have developed immune responses (antibodies); periodic monitoring by your

physician is suggested. Patients should notify their physician immediately if they experience any side effects with treatment. For more information, consult your physician. To learn more, please see full product information; contact Genzyme at 800-745-4447, or visit [www.cerezyme.com](http://www.cerezyme.com). Cerezyme is available by prescription only.

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