

# Horizons

A Newsletter for the Gaucher Community From the Genzyme Corporation

## Understanding Insurance Appeal

*Resources That Can Help*

### **Genzyme Case Managers:**

*Providing Personalized Support for Over 20 Years*

### **The National Gaucher Foundation:**

*Bringing the Gaucher Community Together*



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

## Contents

Understanding Insurance Appeal:  
*Resources That Can Help* .....3

The National Gaucher Foundation:  
*Bringing the Gaucher Community  
Together* .....7

Genzyme Case Managers:  
*Providing Personalized Support  
for Over 20 Years* .....12

Patient Profile:  
*Melissa Landau Steinman* .....14

Also Inside - Special Insert:



## Foreword

Anyone facing the challenge of having Gaucher disease type 1, or having a family member with the disease, must also learn how to access adequate insurance coverage to pay for the treatment. This issue's article on *Understanding Insurance Appeal* will provide patients with resources to help them navigate this process with the assistance of programs and foundations, such as the Genzyme Care Coordination, the Foundation for Health Coverage Education (FHCE), and the Patient Advocate Foundation (PAF). Learn what to do when an appeal is denied and other ways to find a helping hand.

Providing support on another front, the National Gaucher Foundation (NGF) has been bringing the Gaucher community together with educational campaigns, financial assistance, mentoring programs, and fundraising. Read more about the foundation that CEO/Executive Director of NGF, Rhonda Buyers, calls "small but mighty," beginning on page 7.

This issue of *Horizons* features a special insert, *Horizons for Kids* with a glossary of *The Wild World of Words in Gaucher Disease Type 1*, information on the Gaucher Pen Pals program, and an exclusive interview with Joseph McCabe, an active 14-year old who has Gaucher disease type 1.

Learn about the work of Genzyme Case Managers throughout the United States and the support they are offering patients with Gaucher disease type 1. For over 20 years, the patient-support program at Genzyme has helped both patients and their providers with personalized support and services. An adult patient profile, on regulatory attorney Melissa Landau Steinman, can be found on page 14. Steinman learned she had Gaucher disease type 1, when she was 5 months into her first pregnancy, and motherhood and Gaucher disease have been indelibly linked ever since. Read about her experiences with Gaucher disease type 1 during and after childbirth.

As with every issue, we would love to hear your comments and feedback, so help shape your own *Horizons*, and send us a note.

—Your team at Genzyme

---

Cerezyme® (imiglucerase for injection) is indicated for long-term enzyme replacement therapy for pediatric and adult patients with a confirmed diagnosis of type 1 Gaucher disease that results in one or more of the following conditions: anemia (low red blood cell count), thrombocytopenia (low blood platelet count), bone disease, hepatomegaly or splenomegaly (enlarged liver or spleen).

### Important Safety Information

Approximately 15% of patients have developed immune responses (antibodies). These patients have a higher risk of an allergic reaction (hypersensitivity). Use Cerezyme® (imiglucerase for injection) carefully if you have had an allergic reaction to the product in the past. Symptoms suggestive of allergic reaction happen in 6.6% of patients, and 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. Side effects related to Cerezyme 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, abdominal pain, vomiting, 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. Cerezyme is available by prescription only. For more information, consult your physician.

**Please see accompanying full Prescribing Information on pages 10-11.**

Patients are encouraged to report negative side effects of prescription drugs to the FDA. Visit [FDA.gov/medwatch](http://FDA.gov/medwatch), or call 1-800-FDA-1088.



## Understanding Insurance Appeal: *Resources That Can Help*

By Matthew T. Corso

**P**atients with Gaucher disease type 1 and other lysosomal storage diseases now have a variety of resources to help them navigate the sometimes thorny issue of insurance coverage. Nonprofit organizations, advocacy groups, and Genzyme, the manufacturer of a treatment for Gaucher disease type 1, all provide much-needed guidance to patients and their families (**see Table 1 on page 4**). Some also provide financial assistance.

Anyone facing the challenge of having Gaucher disease type 1, or having a family member with the disease, must also learn how to access adequate insurance coverage to pay for the treatment, which can cost an average of \$200,000 per year. That challenge becomes distressing if a family learns that its insurance company either denies coverage for treatment of the disease or has strict limits on the costs it will cover.

When a patient's insurance company denies coverage, the first step is to appeal the denial, experts say. This can be a daunting process. Each state regulates insurance practices, so patients must make sure the appeal is properly worded and directed to the correct agency. That's where the patient assistance resources prove their value.

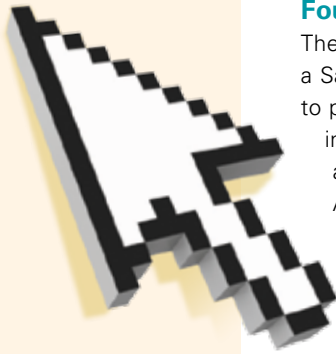
### Genzyme Care Coordination

One of the first resources that patients and their families can consult is a Genzyme Case Manager. Genzyme is a Massachusetts company that manufactures Cerezyme® (imiglucerase for injection), a treatment for patients with Gaucher disease type 1.

When a patient works with Genzyme Care Coordination, they are matched up with a dedicated Case Manager who will assist in handling treatment, access, and insurance issues, according to Nicole Bertoldi, a Genzyme Case Manager. Bertoldi is part of a team of Case Managers who include

## Table 1. Resources for Patients With Gaucher Disease

- Foundation for Health Coverage Education  
800-234-1317  
[www.CoverageForAll.org](http://www.CoverageForAll.org)
- National Gaucher Foundation  
800-504-3189  
[www.gaucherdisease.org](http://www.gaucherdisease.org)
- Genzyme Care Coordination  
800-745-4447



third-party payers about Gaucher disease type 1; (2) educating individuals about available health insurance and helping them review their policies; and (3) coordinating the exchange of information between physicians, third-party payers, and patients to obtain coverage approval for enzyme replacement therapy.

### Foundation for Health Coverage Education (FHCE)

The Foundation for Health Coverage Education (FHCE) is a San Jose, California, nonprofit group whose mission is to provide easy-to-understand “public and private health insurance eligibility information to help more people access coverage.” In partnership with the National Association for Health Underwriters, the FHCE has launched the *Coverage for All* campaign to assist uninsured Americans with identifying public and private healthcare coverage options. The FHCE says that it is dedicated to identifying the uninsured, distributing information and eligibility requirements, and encouraging more people to enroll, thereby lowering the ranks of the uninsured.

The foundation has a 24-hour-a-day, 7-day-a-week, toll-free hotline (800-234-1317), as well as an informative Website, [www.CoverageForAll.org](http://www.CoverageForAll.org). The FHCE says that every month it receives more than 100,000 queries through its Website and toll-free helpline. Consumers are first asked to take a basic 5-question eligibility quiz. Then they are presented with a personalized list of public and private healthcare coverage options. The FHCE says that most consumers qualify for public healthcare coverage.



Nicole Bertoldi

professionals from nursing, social work, and insurance industry backgrounds.

Many insurance plans have a *lifetime maximum* (LTM), which is a limit on medical expenses that an insurance company agrees to pay during a subscriber’s lifetime. However, an estimated 79% of patients with Gaucher disease type 1 who are being treated with Cerezyme (imiglucerase for injection) therapy have insurance with *no* LTM, according to Genzyme.

Regarding patients with Gaucher disease type 1 on Cerezyme therapy who claim insurance coverage for their disease, Bertoldi said, “We don’t often see denials,” since the drug is already approved on most payer or insurance plan formularies.

However, when insurance companies deny coverage to patients with Gaucher disease, Genzyme Case Managers provide support to patients and their requesting physicians, who often must assist in preparing an appeal. The Genzyme Case Managers are assigned to specific regions of the country so that they are familiar with the insurance policies in the states they cover. “Lots of people just don’t know the ins and outs of their insurance plans,” Bertoldi said.

To help patients and their doctors through the insurance process, Genzyme Case Managers assist by: (1) educating



### The National Gaucher Foundation

The National Gaucher Foundation (see story on page 7) is another valuable patient resource that concentrates on patient education and financial assistance for insurance premiums through its CARE and Care+Plus programs.

## What Happens When a Patient's Appeal Is Denied?

In some states, the law allows the patient to request that a case be sent for an external review. To date, 18 states (see Table 2) have external review boards, including some of the most populated states, such as California, Florida, Illinois, New Jersey, New York, and Texas. Insurance companies will send a patient's appeal to a company that they contract with to review the denial, the appeal, or any new information a patient may provide. This third-party company then makes a recommendation to the insurance company about the procedure in question. Typically, the external review board is made up of nurses, attorneys, and physicians who are specialists in the procedure for which a patient is requesting coverage.

In states without external review boards, a patient can contact an organization such as FHCE, Genzyme, or PAF to determine the next step.

## Changes in Healthcare Reform

An important question still unfolding is how the healthcare reform law signed by President Obama will affect patients with Gaucher disease type 1. Starting in 2014, health insurance companies will not be able to charge higher premiums or deny coverage for people with pre-existing conditions.

Genzyme's Bertoldi said that the most notable changes that would likely affect insurance coverage of the Gaucher community are the following:

- The elimination of LTMs (although the reform law exempts some types of health plans from this requirement) and restrictions on annual benefit limits.
- The creation of *pre-existing condition insurance plans* (PCIPs), which were designed to provide access to health insurance for people with pre-existing conditions who might otherwise be refused coverage or be subjected to waiting periods or exclusions. Some states opted into the federal funding for this, and others chose to maintain their existing high-risk pools. PCIPs vary from state to state and may have high out-of-pocket costs or premiums.
- A reduction in some of the out-of-pocket liabilities for Part D prescription drugs for Medicare-eligible patients with Gaucher disease type 1.
- Extension of health insurance coverage to dependents up to the age of 26.
- Regulations that give consumers in new health plans in every state the right to appeal decisions, including claim denials by insurance companies. Consumers will have the right to appeal decisions made by their health plan to an outside, independent decision-maker, no matter what state they live in or what type of healthcare coverage they have. States will work to establish or update their external appeals process to meet new standards, and consumers who are not protected by a state law will have access to a federal external review program.

## A Helping Hand From Genzyme

For patients who are faced with the constraints of an LTM, Genzyme can assist them in monitoring treatment costs because of the significant cost of enzyme replacement therapy.

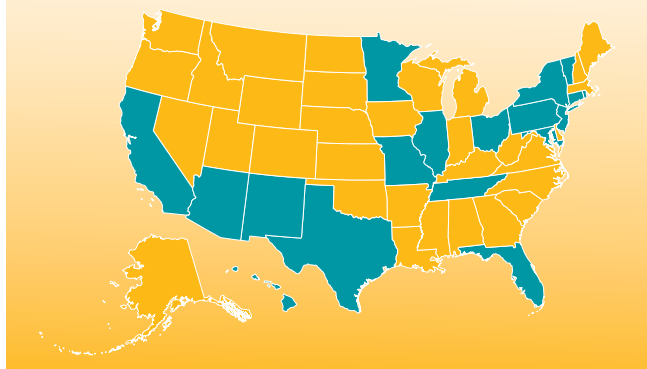
"Often we investigate coverage of enzyme replacement therapy under a pharmacy benefit, as there are cases where the lifetime maximum is only applicable to services covered under the medical benefit," said Bertoldi. "We might also see if there is an option to switch to a different plan during an open enrollment period if they have employer-based coverage. The patient may also have the opportunity to secure new coverage under a spouse's group policy, or there may be an individual plan offering or high-risk pool in his or her state that the patient could be eligible for. We would also evaluate their eligibility for publicly sponsored programs such as Medicaid or Medicare."

In instances where there may be no immediate insurance option available to patients and/or when a gap exists between periods of insurance coverage, Genzyme offers a charitable access program. "To be eligible to receive Cerezyme (imiglucerase for injection) under this program, a patient must be referred by his/her physician who recommends treatment based on medical need and meets other eligibility criteria," according to Bertoldi. ■

**Table 2. States That Provide External Review Boards for Denial of Insurance Coverage**

In some states, the law allows the patient to request that his or her case be heard by an external review board. So far, the following states have external review boards:

- Arizona
- California
- Connecticut
- Florida
- Hawaii
- Illinois
- Maryland
- Minnesota
- Missouri
- New Jersey
- New Mexico
- New York
- Ohio
- Pennsylvania
- Rhode Island
- Tennessee
- Texas
- Vermont

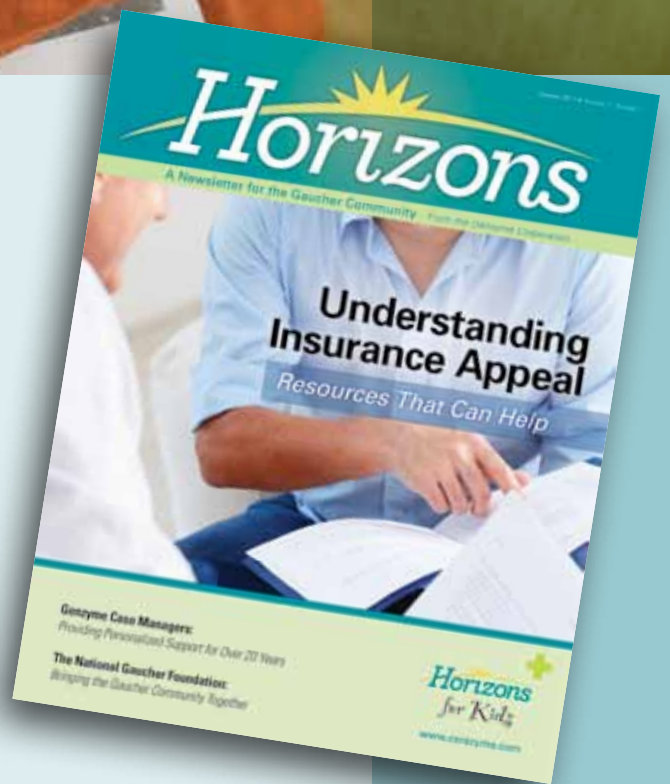




# We'd Love to Hear From You

Let us know how we can make *Horizons* even better.

Please send us your feedback by filling out the enclosed business reply card or emailing our publisher ([petercis1@yahoo.com](mailto:petercis1@yahoo.com)).



# The National Gaucher Foundation: *Bringing the Gaucher Community Together*

By Brenda Lange

**T**reatment to manage many of the debilitating effects of Gaucher disease type 1 has been FDA-approved for approximately 20 years, and for over 25 years, no one nonprofit organization has been more supportive of Gaucher research than the National Gaucher Foundation (NGF).

However, raising funds for research represents only a portion of the small nonprofit's efforts. "We may be small, but we're mighty," said CEO/Executive Director Rhonda Buyers with a chuckle.

## How It Began

The Foundation started when several families who had experienced Gaucher disease type 1 firsthand raised millions of dollars with the goal of finding a treatment and a cure. Over the years, that funding helped support the research that led to the approval of a targeted enzyme replacement therapy in 1991, the first treatment produced by Genzyme. The first board president of the National Gaucher Foundation traveled the country, speaking out and raising money; three of her six children were patients with Gaucher disease type 1.

As word spread, more people were diagnosed and put on the drug, and the needs of the Gaucher community changed, said Buyers. "We, the Foundation, needed to meet those needs. We needed to help find and educate the patients and communities, particularly those within the Ashkenazi Jewish community,\*"



Rhonda Buyers,  
CEO/Executive  
Director of the NGF.

she added. "Education became a big part of what we were doing, and we stepped it up."

## The Early Years

In 2000, the NGF began a massive educational push around the country to rival anything done by organizations many times its size. Especially in areas with dense Ashkenazi populations, such as New York City, Philadelphia, Miami, and Los Angeles, Buyers' staff and volunteers visited Jewish community centers, synagogues, daycare facilities, and schools. "We sent home flyers in schoolbags, did advertising, and even stuck posters on bulletin boards," remembered Buyers. At that time, her staff consisted of just three women, including her.

With the expert help of Marketing Director Rosina Papantonio, hired in 1999, the impact of every ad was measured and all data tracked to determine the success of each marketing effort. If one approach didn't work well, it was pulled and another one tried.

"Every penny mattered, and we had to be sure that every one we spent and everything we did was wise. We made a lot of positive movement within the [Gaucher] community, and I'm very proud of that," said Buyers.



Rosina Papantonio

\*Ashkenazi Jews are of Eastern, Central, and Northern European ancestry. People of Ashkenazi Jewish descent have a much higher risk of Gaucher disease type 1 as compared with non-Jewish individuals.

**"We, the Foundation, needed to meet those needs. We needed to help find and educate the patients and communities, particularly those within the Ashkenazi Jewish community."**

—Rhonda Buyers

## The NGF Today

A decade later, the Foundation has a new president of the board of directors. Brian Berman, who has Gaucher disease type 1, was the first person to receive the enzyme treatment at a young age. Buyers said that he had been severely affected with the complications of Gaucher disease type 1, but went on to become a successful businessman, marry, and raise four children.



**Brian Berman**

“To me, running the Foundation on a daily basis, I look to him because he has a good head for business and technology, and is very innovative and forward-thinking. He helps us all keep up with what’s going on in the world of Gaucher,” Buyers said.

The Foundation has decided to take its message to the general population. The new marketing campaign recently kicked off with ads in *Prevention* magazine, which reaches more than 10 million readers. “This wider exposure means that people are hopefully going to get diagnosed earlier and will be able to be treated more effectively sooner,” said Buyers, adding that she hopes more research dollars will enable a treatment to be found for types 2 and 3.

NGF’s work with the European Gaucher Alliance extends its reach around the world, as does its newly launched website [www.gaucherdisease.org](http://www.gaucherdisease.org). The site provides information about the disease, as well as ways in which the Foundation helps patients and their families.

Some of the monies raised by the Foundation support its Care and Care+Plus Programs. These financial assistance programs help patients with the costs of treatment that often run into six figures annually. They also help with high insurance premiums and ancillary needs, such as transportation to infusion sites or childcare.

One of the Foundation’s initiatives is its mentoring program, which matches previously and newly diagnosed patients and/or their parents with individuals who have more experience either living with the disease or coping with a family member’s illness.

## Being Frank

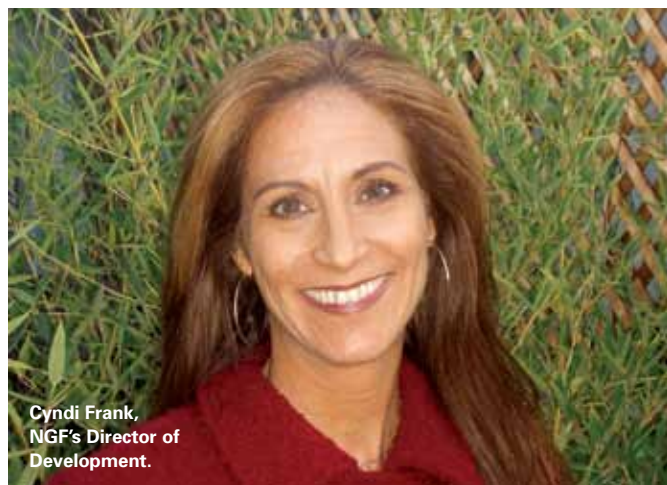
The National Gaucher Foundation’s Director of Development, Cyndi Frank, is both a mentor and a patient with Gaucher disease type 1. At age 12, she developed a large bruise on her leg that wouldn’t go away—and, in fact, grew larger. After a misdiagnosis of leukemia and months



of watchful waiting, her physician finally diagnosed her with Gaucher disease type 1.

“In 1976 there was no treatment and no Internet [to turn to for information],” Frank remembers. “I was very sick. They told me I’d be in a wheelchair by the time I was 20 and probably wouldn’t live past 30.” By 23, Frank’s spleen weighed 20 pounds, bruises covered her body, and she required blood transfusions with every menstrual period. Her overall vitality had suffered. At 25, she underwent a successful partial splenectomy, which returned some of her health and energy. In 1989, she entered the clinical trials of enzyme replacement therapy at the National Institutes of Health, and the treatment was so successful that her outlook on life changed radically.

“When I realized I might live a long life, I had to figure out what to do with it,” she said. “Now I had the freedom to feel my emotions, to meet a man, and think about a family. Now I had to start thinking long term.” Married for 21 years to one



**Cyndi Frank,**  
NGF’s Director of  
Development.

of her best college friends, Frank now runs regularly, eats a vegetarian diet, gets lots of sleep, and follows a healthy lifestyle in addition to her regular biweekly infusions of Cerezyme® (imiglucerase for injection) enzyme replacement therapy.

A recurrence of symptoms at age 40 led Frank to her current work with the Foundation. "I met Rhonda, we clicked, I had direct experience (with the disease *and* fundraising), and they didn't have anyone doing this job," she said. "This has been the most amazing experience. Knowing I can have this positive impact on others is like medicine for me. It's so fulfilling."

Frank currently mentors 15 patients with Gaucher disease type 1 and their families. She credits the nationwide network of mentors—who undergo training sessions—for their dedication to helping others cope with the disease.

"The first time I met someone else with Gaucher disease type 1, I was 28," she recalled. "It's huge to connect with someone who can validate you're not different." Frank calls and emails her mentees, following up to make sure they're doing well. She becomes friends with the parents and feels that she has become a role model for their children.

### Bringing the Gaucher Community Together

"The service our mentors perform is amazing," said Buyers. "They are there for others in their time of need. They help others realize that they can get through it too. We're fortunate that we have treatments available," she adds. "Many rare diseases don't."

The Foundation hosts a major conference every couple of years, with the most recent 2-day event held in Atlanta in November 2010. Speakers arrived from around the country

to educate and mingle with more than 200 attendees. "We talked everything Gaucher," Buyers said, "from symptoms, things people were dealing with, and the drug shortage in 2009 and how it affected people. The researchers and clinicians had a chance to present their studies, as well as talk directly to patients."

The Foundation also conducts dozens of patient meetings around the country every year—smaller versions of the national conference. A complete calendar can be found on the Website. "These meetings give patients and parents a chance to meet others they may have been communicating with by phone or e-mail or listserv for months or years," Buyers said. "They can get firsthand information directly from those doing the research."

### New Beginnings

When Buyers was looking for a nonprofit foundation with which to work in 1994, she had never heard of Gaucher disease. Today it is her passion. "When you see kids dealing with a rare disease every day of their lives... I saw some people go from wheelchairs to walking, and many of their symptoms have been treated... I thank God every day," she said.

As the tiny but mighty NGF staff and board move forward into a new decade, they will continue to work for the individuals who need them the most. "We're going to keep growing the initiatives," Buyers promised. "This is just the tip of the iceberg. For everyone who hears about Gaucher, there are many more who need to hear about it. I want the word 'Gaucher' to become much more common... we owe it to that population to give this awareness to them, and we intend to keep on going." ■

### Indications and Usage

Cerezyme® (imiglucerase for injection) is indicated for long-term enzyme replacement therapy for pediatric and adult patients with a confirmed diagnosis of type 1 Gaucher disease that results in one or more of the following conditions: anemia (low red blood cell count), thrombocytopenia (low blood platelet count), bone disease, hepatomegaly or splenomegaly (enlarged liver or spleen).

### Important Safety Information

Approximately 15% of patients have developed immune responses (antibodies). These patients have a higher risk of an allergic reaction (hypersensitivity). Use Cerezyme® (imiglucerase for injection) carefully if you have had an allergic reaction to the product in the past. Symptoms suggestive of allergic reaction happen in 6.6% of patients, and 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. Side effects related to Cerezyme 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, abdominal pain, vomiting, 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. Cerezyme is available by prescription only. For more information, consult your physician. To learn more, please see the enclosed full product information or contact Genzyme at 1-800-745-4447 (option 2).

**Please see accompanying full Prescribing Information on pages 10-11.**



200 UNITS

imiglucerase for injection

400 UNITS

DESCRIPTION

Cerezyme® (imiglucerase for injection) is an analogue of the human enzyme β-glucocerebrosidase, produced by recombinant DNA technology. β-Glucocerebrosidase (β-D-glucosyl-N-acylsphingosine glucosylase, E.C. 3.2.1.45) is a lysosomal glycoprotein enzyme which catalyzes the hydrolysis of the glycolipid glucocerebroside to glucose and ceramide.

Cerezyme® is produced by recombinant DNA technology using mammalian cell culture (Chinese hamster ovary). Purified imiglucerase is a monomeric glycoprotein of 497 amino acids, containing 4 N-linked glycosylation sites (Mr = 60,430). Imiglucerase differs from placental glucocerebrosidase by one amino acid at position 495, where histidine is substituted for arginine. The oligosaccharide chains at the glycosylation sites have been modified to terminate in mannose sugars. The modified carbohydrate structures on imiglucerase are somewhat different from those on placental glucocerebrosidase. These mannose-terminated oligosaccharide chains of imiglucerase are specifically recognized by endocytic carbohydrate receptors on macrophages, the cells that accumulate lipid in Gaucher disease.

Cerezyme® is supplied as a sterile, non-pyrogenic, white to off-white lyophilized product. The quantitative composition of the lyophilized drug is provided in the following table:

Table with 3 columns: Ingredient, 200 Unit Vial, 400 Unit Vial. Rows include Imiglucerase (total amount)\*, Mannitol, Sodium Citrates (Trisodium Citrate, Disodium Hydrogen Citrate), Polysorbate 80, NF, and Citric Acid and/or Sodium Hydroxide.

\*This provides a respective withdrawal dose of 200 and 400 units of imiglucerase.

An enzyme unit (U) is defined as the amount of enzyme that catalyzes the hydrolysis of 1 micromole of the synthetic substrate para-nitrophenyl-β-D-glucopyranoside (pNP-Glc) per minute at 37°C. The product is stored at 2-8°C (36-46°F). After reconstitution with Sterile Water for Injection, USP, the imiglucerase concentration is 40 U/mL (see DOSAGE AND ADMINISTRATION for final concentrations and volumes). Reconstituted solutions have a pH of approximately 6.1.

CLINICAL PHARMACOLOGY

Mechanism of Action/Pharmacodynamics

Gaucher disease is characterized by a deficiency of β-glucocerebrosidase activity, resulting in accumulation of glucocerebroside in tissue macrophages which become engorged and are typically found in the liver, spleen, and bone marrow and occasionally in lung, kidney, and intestine. Secondary hematologic sequelae include severe anemia and thrombocytopenia in addition to the characteristic progressive hepatosplenomegaly, skeletal complications, including osteonecrosis and osteopenia with secondary pathological fractures. Cerezyme® (imiglucerase for injection) catalyzes the hydrolysis of glucocerebroside to glucose and ceramide. In clinical trials, Cerezyme improved anemia and thrombocytopenia, reduced spleen and liver size, and decreased cachexia to a degree similar to that observed with Ceredase® (alglucerase injection).

Pharmacokinetics

During one-hour intravenous infusions of four doses (7.5, 15, 30, 60 U/kg) of Cerezyme® (imiglucerase for injection), steady-state enzymatic activity was achieved by 30 minutes. Following infusion, plasma enzymatic activity declined rapidly with a half-life ranging from 3.6 to 10.4 minutes. Plasma clearance ranged from 9.8 to 20.3 mL/min/kg (mean ± S.D., 14.5 ± 4.0 mL/min/kg). The volume of distribution corrected for weight ranged from 0.09 to 0.15 L/kg

(0.12 ± 0.02 L/kg). These variables do not appear to be influenced by dose or duration of infusion. However, only one or two patients were studied at each dose level and infusion rate. The pharmacokinetics of Cerezyme do not appear to be different from placental-derived alglucerase (Ceredase®).

In patients who developed IgG antibody to Cerezyme, an apparent effect on serum enzyme levels resulted in diminished volume of distribution and clearance and increased elimination half-life compared to patients without antibody (see WARNINGS).

INDICATIONS AND USAGE

Cerezyme® (imiglucerase for injection) is indicated for long-term enzyme replacement therapy for pediatric and adult patients with a confirmed diagnosis of Type 1 Gaucher disease that results in one or more of the following conditions:

- a. anemia
b. thrombocytopenia
c. bone disease
d. hepatomegaly or splenomegaly

CONTRAINDICATIONS

There are no known contraindications to the use of Cerezyme® (imiglucerase for injection). Treatment with Cerezyme should be carefully re-evaluated if there is significant clinical evidence of hypersensitivity to the product.

WARNINGS

Approximately 15% of patients treated and tested to date have developed IgG antibody to Cerezyme® (imiglucerase for injection) during the first year of therapy. Patients who developed IgG antibody did so largely within 6 months of treatment and rarely developed antibodies to Cerezyme after 12 months of therapy. Approximately 46% of patients with detectable IgG antibodies experienced symptoms of hypersensitivity.

Patients with antibody to Cerezyme have a higher risk of hypersensitivity reaction. Conversely, not all patients with symptoms of hypersensitivity have detectable IgG antibody. It is suggested that patients be monitored periodically for IgG antibody formation during the first year of treatment.

Treatment with Cerezyme should be approached with caution in patients who have exhibited symptoms of hypersensitivity to the product.

Anaphylactoid reaction has been reported in less than 1% of the patient population. Further treatment with imiglucerase should be conducted with caution. Most patients have successfully continued therapy after a reduction in rate of infusion and pretreatment with antihistamines and/or corticosteroids.

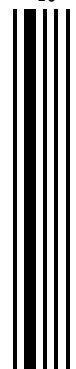
PRECAUTIONS

General

In less than 1% of the patient population, pulmonary hypertension and pneumonia have also been observed during treatment with Cerezyme® (imiglucerase for injection). Pulmonary hypertension and pneumonia are known complications of Gaucher disease and have been observed both in patients receiving and not receiving Cerezyme. No causal relationship with Cerezyme has been established. Patients with respiratory symptoms in the absence of fever should be evaluated for the presence of pulmonary hypertension.

Therapy with Cerezyme should be directed by physicians knowledgeable in the management of patients with Gaucher disease.

Caution may be advisable in administration of Cerezyme to patients previously treated with Ceredase® (alglucerase injection) and who have developed antibody to Ceredase or who have exhibited symptoms of hypersensitivity to Ceredase.



## Carcinogenesis, Mutagenesis, Impairment of Fertility

Studies have not been conducted in either animals or humans to assess the potential effects of **Cerezyme**<sup>®</sup> (imiglucerase for injection) on carcinogenesis, mutagenesis, or impairment of fertility.

## Teratogenic Effects: Pregnancy Category C

Animal reproduction studies have not been conducted with **Cerezyme**<sup>®</sup> (imiglucerase for injection). It is also not known whether **Cerezyme** can cause fetal harm when administered to a pregnant woman or can affect reproductive capacity. **Cerezyme** should not be administered during pregnancy except when the indication and need are clear and the potential benefit is judged by the physician to substantially justify the risk.

## Nursing Mothers

It is not known whether this drug is excreted in human milk. Because many drugs are excreted in human milk, caution should be exercised when **Cerezyme**<sup>®</sup> (imiglucerase for injection) is administered to a nursing woman.

## Pediatric Use

The safety and effectiveness of **Cerezyme**<sup>®</sup> (imiglucerase for injection) have been established in patients between 2 and 16 years of age. Use of **Cerezyme** in this age group is supported by evidence from adequate and well-controlled studies of **Cerezyme** and Ceredase<sup>®</sup> (alglucerase injection) in adults and pediatric patients, with additional data obtained from the medical literature and from long-term post-marketing experience. **Cerezyme** has been administered to patients younger than 2 years of age, however the safety and effectiveness in patients younger than 2 have not been established.

## ADVERSE REACTIONS

Since the approval of **Cerezyme**<sup>®</sup> (imiglucerase for injection) in May 1994, Genzyme has maintained a worldwide post-marketing database of spontaneously reported adverse events and adverse events discussed in the medical literature. The percentage of events for each reported adverse reaction term has been calculated using the number of patients from these sources as the denominator for total patient exposure to **Cerezyme** since 1994. Actual patient exposure is difficult to obtain due to the voluntary nature of the database and the continuous accrual and loss of patients over that span of time. The actual number of patients exposed to **Cerezyme** since 1994 is likely to be greater than estimated from these voluntary sources and, therefore, the percentages calculated for the frequencies of adverse reactions are most likely greater than the actual incidences.

Experience in patients treated with **Cerezyme** has revealed that approximately 13.8% of patients experienced adverse events which were judged to be related to **Cerezyme** administration and which occurred with an increase in frequency. Some of the adverse events were related to the route of administration. These include discomfort, pruritus, burning, swelling or sterile abscess at the site of venipuncture. Each of these events was found to occur in < 1% of the total patient population.

Symptoms suggestive of hypersensitivity have been noted in approximately 6.6% of patients. Onset of such symptoms has occurred during or shortly after infusions; these symptoms include pruritus, flushing, urticaria, angioedema, chest discomfort, dyspnea, coughing, cyanosis, and hypotension. Anaphylactoid reaction has also been reported (see WARNINGS). Each of these events was found to occur in < 1.5% of the total patient population. Pre-treatment with antihistamines and/or corticosteroids and reduced rate of infusion have allowed continued use of **Cerezyme** in most patients.

Additional adverse reactions that have been reported in approximately 6.5% of patients treated with **Cerezyme** include: nausea, abdominal pain, vomiting, diarrhea, rash, fatigue, headache, fever, dizziness, chills, backache, and tachycardia. Each of these events was found to occur in < 1.5% of the total patient population.

Incidence rates cannot be calculated from the spontaneously reported adverse events in the post-marketing database. From this database, the most commonly reported adverse events in children (defined as ages 2 – 12 years) included dyspnea, fever, nausea, flushing, vomiting, and coughing, whereas in adolescents (>12 – 16 years) and in adults (>16 years) the most commonly reported events included headache, pruritus, and rash.

In addition to the adverse reactions that have been observed in patients treated with **Cerezyme**, transient peripheral edema has been reported for this therapeutic class of drug.

## OVERDOSE

Experience with doses up to 240 U/kg every 2 weeks have been reported. At that dose there have been no reports of obvious toxicity.

## DOSAGE AND ADMINISTRATION

**Cerezyme**<sup>®</sup> (imiglucerase for injection) is administered by intravenous infusion over 1-2 hours. Dosage should be individualized to each patient. Initial dosages range from 2.5 U/kg of body weight 3 times a week to 60 U/kg once every 2 weeks. 60 U/kg every 2 weeks is the dosage for which the most data are available. Disease severity may dictate that treatment be initiated at a relatively high dose or relatively frequent administration.

Dosage adjustments should be made on an individual basis and may increase or decrease, based on achievement of therapeutic goals as assessed by routine comprehensive evaluations of the patient's clinical manifestations.

**Cerezyme**<sup>®</sup> should be stored at 2-8°C (36-46°F). After reconstitution, **Cerezyme** should be inspected visually before use. Because this is a protein solution, slight flocculation (described as thin translucent fibers) occurs occasionally after dilution. The diluted solution may be filtered through an in-line low protein-binding 0.2 µm filter during administration. Any vials exhibiting opaque particles or discoloration should not be used. DO NOT USE **Cerezyme** after the expiration date on the vial.

On the day of use, after the correct amount of **Cerezyme** to be administered to the patient has been determined, the appropriate number of vials are each reconstituted with Sterile Water for Injection, USP. The final concentrations and administration volumes are provided in the following table:

	200 Unit Vial	400 Unit Vial
Sterile water for reconstitution	5.1 mL	10.2 mL
Final volume of reconstituted product	5.3 mL	10.6 mL
Concentration after reconstitution	40 U/mL	40 U/mL
Withdrawal volume	5.0 mL	10.0 mL
Units of enzyme within final volume	200 units	400 units

A nominal 5.0 mL for the 200 unit vial (10.0 mL for the 400 unit vial) is withdrawn from each vial. The appropriate amount of **Cerezyme** for each patient is diluted with 0.9% Sodium Chloride Injection, USP, to a final volume of 100 – 200 mL. **Cerezyme** is administered by intravenous infusion over 1-2 hours. Aseptic techniques should be used when diluting the dose. Since **Cerezyme** does not contain any preservative, after reconstitution, vials should be promptly diluted and not stored for subsequent use. **Cerezyme**, after reconstitution, has been shown to be stable for up to 12 hours when stored at room temperature (25°C) and at 2-8°C. **Cerezyme**, when diluted, has been shown to be stable for up to 24 hours when stored at 2-8°C.

Relatively low toxicity, combined with the extended time course of response, allows small dosage adjustments to be made occasionally to avoid discarding partially used bottles. Thus, the dosage administered in individual infusions may be slightly increased or decreased to utilize fully each vial as long as the monthly administered dosage remains substantially unaltered.

## HOW SUPPLIED

**Cerezyme**<sup>®</sup> (imiglucerase for injection) is supplied as a sterile, non-pyrogenic, lyophilized product. It is available as follows:

200 Units per Vial NDC 58468-1983-1  
400 Units per Vial NDC 58468-4663-1

Store at 2-8°C (36-46°F).

Rx only

**Cerezyme**<sup>®</sup> (imiglucerase for injection) is manufactured by:  
**Genzyme Corporation**  
500 Kendall Street  
Cambridge, MA 02142 USA

Certain manufacturing operations may have been performed by other firms.

Cerezyme and Genzyme are registered trademarks of Genzyme Corporation.

6LE0005D



## Genzyme Case Managers: *Providing Personalized Support for Over 20 Years*

*By Cheryl Alkon*

Learning that you've been diagnosed with Gaucher disease type 1 is life-changing, so it may feel reassuring to know that someone at Genzyme—the company that manufactures Cerezyme® (imiglucerase for injection)—is looking out for you: your Case Manager.

"The first person to speak to a patient, other than a doctor, is often someone from Genzyme," said Michael Clavadetscher, a Genzyme Case Manager who works closely with patients who live in South Florida. "It's important to put their mind at ease."

Genzyme Case Managers are there to offer help in whatever way it is needed, both immediately after diagnosis, as well as through the years. Established more than 20 years ago, the company's patient support department has a history of partnering with both patients and their providers to provide personalized support from diagnosis through treatment plan.

"It can help reassure a patient that Gaucher type 1 is rare, but treatable," said Clavadetscher. "There are doctors throughout the country who are extremely knowledgeable."

### **Helping Patients Navigate Treatment**

Cerezyme (imiglucerase for injection), which is used to treat Gaucher disease type 1, is typically infused into the bloodstream for 1 to 2 hours, every 2 weeks. Medication dosing is based on a patient's weight, but costs an average of \$200,000 for 1 year of treatment. The price is based on the research and development costs, manufacturing and distribution costs, and costs associated with the rarity of the condition. It is estimated that less than 10,000 people in the world have been diagnosed with Gaucher disease type 1.

Because of the expense, many patients are understandably concerned about how insurance will cover the cost of Cerezyme (imiglucerase for injection) therapy. Genzyme Case Managers can help shine a light on the often confusing path of health insurance coverage to determine what the patient's insurance policy will cover, what it may not, and, if necessary, how to make up the difference.

"It's overwhelming," said Clavadetscher. "There are restrictions with insurance, to know what is covered and what needs authorization, or precertification. We're here to act in a patient's best interest."

If a person is facing out-of-pocket costs for treatment, for example, Clavadetscher will help connect him or her with programs sponsored by the National Gaucher Foundation (<http://www.gaucherdisease.org/psp.php>) or the Patient Access Network (<https://www.panfoundation.org/>) to help find financial assistance.

### **Making Therapy Convenient**

Since Gaucher disease type 1 treatment is lifelong and requires infusion for 1 to 2 hours every 2 weeks, Genzyme Case Managers often work closely with their patients to help coordinate care. Infusion therapy may occur, in a specialist physician's (such as a hematologist) office, in an outpatient hospital setting, or in a patient's home, explained Clavadetscher.

"Particularly in a remote place in the middle of the country, it takes the coordination of insurance and making sure the hospital can get the drug however possible," he said.

## Handling Life's Logistics

Many patients plan to travel, attend college outside of their home state, or simply want to be available for life's experiences without feeling tethered to their enzyme replacement therapy (ERT) schedules. Daniel Marsh, 74, has lived with Gaucher disease type 1 for more than 30 years, and began taking Cerezyme (imiglucerase for injection) in 1991 when it first became available. Today, he lives half of the year in Swartz Creek, Michigan, and half of the year in Naples, Florida, and relies on his Genzyme Case Manager to make sure he receives his treatment in both locations.

When his insurance coverage changed from Blue Cross to Medicare, receiving treatment got tougher, he said. Medicare requires facilities to order the medication directly from Genzyme, rather than ordering it from a mail-order pharmacy.

"When Medicare took over, it became very difficult," said Marsh. "[The hospital] where I used to get it, they would not put up the amount of money." Marsh's Case Manager worked to find him another location where he could get Cerezyme. "We found another hospital only 10 minutes from my home that would do it."

Nicole Wilson, a Genzyme Case Manager for Massachusetts, New Hampshire, and Vermont, recalled another situation where her patient wanted to fit ERT into her life, rather than change her life to fit it into the treatment schedule.

"I have a patient who is going to college out of state, and her family has an HMO plan, which can be very restrictive," Wilson

said. "Her family is changing its plan to a PPO plan so she can see people in that state for treatment.

Due to the reality of being a college freshman in a new setting, the patient also decided to return home once a month for her treatment. Once she is a sophomore, Wilson said, the patient will work with her college's health services department to remain on campus for ERT.

One of Clavadetscher's newly diagnosed patients didn't want to let her medication schedule get in the way of an important celebration. Based in Florida, the woman wanted to travel to Chicago to visit her first grandchild, and so worked closely with her doctor to figure out how to make it happen. "She had the infusion in South Florida, and then flew out to Chicago the next day to see her grandchild. She came back a couple of weeks later and was infused the day she came back," Clavadetscher said. That way the patient avoided the fatigue that sometimes occurs in patients who first start treatment or who miss an infusion. "She wanted to be in the best possible health when it was her time to be in the hospital [with the new baby]," he said. Ongoing, close contact between Genzyme Case Managers and patients enables such scenarios to happen as smoothly as possible.

"We know our patients," said Wilson. "We work with them. We know when they have kids. We tend to know a lot of details about them, and we check in regularly. I think our patients appreciate that." ■

## New Program Announcement



  
**Cerezyme**  
imiglucerase for injection

Genzyme is pleased to announce  
the launch of our new program

## Cerezyme Co-pay Assistance Program

This new program will help eligible individuals with commercial insurance who are prescribed Cerezyme with their drug related co-pay expenses, including co-insurance and deductibles, regardless of financial status.

**With the introduction of this program in July 2011 Genzyme intends to cover eligible individuals' co-pay expenses, including co-insurance and deductibles, at 100% for the remainder of 2011.**

### How to apply:

Simply visit [www.cerezyme.com/copay.aspx](http://www.cerezyme.com/copay.aspx) to complete the online application or call your Genzyme Case Manager at 1-800-745-4447, option 3 to learn more about the program, eligibility, and begin the application process.

  
A SANOFI COMPANY

Cerezyme® is a registered trademark of Genzyme. ©2011 Genzyme Corporation. All Rights Reserved. CZ-US-P388-06-11

# Patient Profile: Melissa Landau Steinman

By Cheryl Alkon

**M**otherhood and Gaucher disease type 1 have always been intimately linked for attorney Melissa Landau Steinman.

Steinman was completely unaware of Gaucher disease before becoming pregnant with her first child 13 years ago. Five months into what had been an uneventful pregnancy, she talked to her cousin, also pregnant, who had been tested for genetic conditions. The cousin learned that she had an abnormal gene that caused Gaucher disease type 1; she was a type 1 carrier and could potentially pass on the disease to a child or grandchild.



"I got tested for these genetic Jewish diseases, and you should really get tested too," Steinman, now 42, recalled her cousin telling her. "I asked my OB-GYN, and she said, 'We don't do that here.' It was something I had completely never heard of, and by that point, I was pretty darn pregnant."

Steinman, who lives in Chevy Chase, Maryland, took the blood test at a nearby hospital. Later that day, she learned, over the phone, that she had the disease. "They told me, 'Normally we wouldn't counsel you about this over the phone, but because of the timing, you need to know you actually have Gaucher disease.'"

## A New Reality

Steinman was *homozygous*, which means that she carried a pair of genes that tested positive for Gaucher disease type 1, the most common form of this uncommon disease. She had never shown any symptoms, which can include an enlarged liver or spleen, a low blood platelet count, a low white blood cell count, anemia, fractures related to bone tissues thinning or dying, or misshapen bones (called *Erlenmeyer flask deformity*).

"It was obviously a surprise," Steinman said. Her doctors told her to be fully tested postpartum to see if there were signs of Gaucher disease type 1 that had not been causing outward pain or problems.

Steinman and her then-husband needed to decide quickly what to do next with the information. They could have the baby tested in utero, tested at birth, or not tested at all for Gaucher disease type 1. Doctors tested the umbilical cord blood of her son Charles, now 13, as soon as he was born. He is a Gaucher disease type 1 carrier, meaning that only one of his two genes for the disease is affected.

## Fighting for Herself and Her Sons

Steinman became immersed in new motherhood and returned on a part-time basis to her job as a regulatory lawyer at the Venable law firm in Washington, DC. She only got tested for the effects of Gaucher disease type 1 two years later, when she wanted to get pregnant again. She still felt fine, though she recalled that she'd had postpartum hemorrhaging after Charles was born.

"It could have been something that lots of people have, but I said 'Gee, was this the Gaucher?'" Her doctor couldn't tell her definitively. "The tough thing is that the symptoms are a little hard to track because so few people have Gaucher disease, and so few have it while pregnant," she said.

Steinman learned that her liver and spleen were enlarged but that she didn't have the bone problems common in many people with the disease. She began enzyme replacement therapy with Cerezyme® (imiglucerase for injection) before getting pregnant again.

"I had always felt fine, and I had to make the decision about whether to start the treatment at all," she said. "I knew I'd gotten lucky the first time I was pregnant, and I thought maybe I wouldn't be the second time. The fact was that I had a mild iteration of the disease."

Once she became pregnant a second time, Steinman stopped her infusions. "I was much less nervous and much more relaxed during this pregnancy," she said. "I had a lot more energy than you would think for someone who had a small child and was pregnant at the same time."

Steinman's second child, a boy named James, was also checked at birth via cord blood testing for Gaucher disease

type 1. Like his mother, he is homozygous for the disease but shows no symptoms of it. Steinman said that her now 9-year-old, known as Jamie, has had annual bloodwork to monitor for symptoms.

"We've taught him from a young age that this is part of life, that he has this part of his genes that makes him special," she said. "He hasn't really had to deal with any negative consequences at all, other than getting a blood test once a year. It's been really good in that respect."

Being in a place where her symptoms are mild and being so knowledgeable about Gaucher disease type 1 puts both Steinman and Jamie in a unique situation. "We know so much more about these kids," she said. "It used to be that kids were diagnosed really late, when they were really sick and had irreversible bone issues. Many doctors didn't recognize Gaucher, and people were sometimes misdiagnosed." Now, both she and Jamie know the signs and symptoms they need to be aware of.



**Melissa's sons, James (left) and Charles (right).**

### An Attorney's Advocacy

Steinman's unique position—as a person with mild Gaucher disease type 1 and the mother of a child with the same genetic presentation—has made her passionate about issues related to Gaucher disease type 1.

"I feel a calling to participate in clinical trials" for new ways to treat Gaucher, she said, both because of the disease's rarity (only 1 person in 40-60,000 is diagnosed with Gaucher) and for her children. She is currently participating

in a research study with an investigational oral medication to see how effective it is as compared to enzyme replacement therapy. She has also served on patient advisory boards for Genzyme and two other biopharmaceutical companies, and has taken part in several clinical studies of new medications and drug protocols. In addition, Steinman does *pro bono* legal work for charities and nonprofits such as the Parent's Guide to Cord Blood Foundation, which provides guidance on cord blood medical research, banking, and storage, and the Robin Hood Foundation.

"I've tried to get involved in the community because I hope I can guide things along for the future. With my son, it's sort of a dual perspective as a patient and a parent."

As a regulatory attorney focusing on the marketing, advertising, and consumer protection law, Steinman has more than a patient's perspective on how drugs are approved for use in the United States, as well as how the health insurance industry operates.

Wayne Rosenfield, PhD, a Connecticut psychologist who has been friends and partners-in-advocacy with Steinman for several years (they have worked together in several Gaucher organizations and patient advisory groups), says of Steinman, "She possesses good human values. I know her to be extremely bright, goal-directed, and have directly observed her to be a strong advocate. Her commitment to advocacy comes from not just being the person directly affected, but also being the mother of a child with Gaucher." ■

### Indications and Usage

Cerezyme® (imiglucerase for injection) is indicated for long-term enzyme replacement therapy for pediatric and adult patients with a confirmed diagnosis of type 1 Gaucher disease that results in one or more of the following conditions: anemia (low red blood cell count), thrombocytopenia (low blood platelet count), bone disease, hepatomegaly or splenomegaly (enlarged liver or spleen).

### Important Safety Information

Approximately 15% of patients have developed immune responses (antibodies). These patients have a higher risk of an allergic reaction (hypersensitivity). Use Cerezyme® (imiglucerase for injection) carefully if you have had an allergic reaction to the product in the past. Symptoms suggestive of allergic reaction happen in 6.6% of patients, and 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. Side effects related to Cerezyme 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, abdominal pain, vomiting, 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. Cerezyme is available by prescription only. For more information, consult your physician. To learn more, please see the enclosed full product information or contact Genzyme at 1-800-745-4447 (option 2).

**Please see accompanying full Prescribing Information on pages 10-11.**

# Now Recruiting GAUCHER disease ORAL compound clinical research trials

Eliglustat tartrate  
(Genz-112638)  
is an investigational  
oral compound which  
aims to partially inhibit  
the production of  
glucosylceramide  
(GL-1) in Gaucher  
cells

**The ENGAGE and ENCORE studies are designed to determine the safety and efficacy of eliglustat tartrate (Genz-112638) in patients with Gaucher disease type 1.**

The **ENGAGE** study is recruiting patients at least 16 years of age with splenomegaly and anemia and/or thrombocytopenia and who have never been or currently are not being treated for the disease. The **ENCORE** study is recruiting clinically stable patients at least 18 years of age who have been treated with enzyme replacement therapy for at least 3 years. The **EDGE** study is recruiting patients to evaluate maintenance of treatment goals comparing twice daily dosing vs. once daily dosing.

These studies require:

- Patients to have a confirmed diagnosis of Gaucher disease type 1
- Patients to be excluded if they have a clinically significant disease, other than Gaucher disease type 1

## To participate or learn more about these studies, contact:

■ **[www.clinicaltrials.gov](http://www.clinicaltrials.gov)** search

**ENGAGE:** NCT00891202 **ENCORE:** NCT00943111 **EDGE:** NCT01074944

■ **Genzyme Medical Information** at **1-800-745-4447**  
(option 2) or **[medinfo@genzyme.com](mailto:medinfo@genzyme.com)**

One of the world's leading biotechnology companies, Genzyme is dedicated to making a major positive impact on the lives of people with serious diseases.

# Horizons for Kids

Special  
Section!

## The Right Treatment for You

After you are diagnosed with Gaucher disease type 1, you and your parent or caregiver will choose a doctor for you that you feel comfortable with. Then you'll find out about a treatment that will get you on the road to feeling better.

Gaucher disease does not yet have a cure or treatment to correct the genetic condition. But fortunately there are treatments to help keep the disease under control.

You may be getting Cerezyme® (imiglucerase for injection) infusion for your treatment. Cerezyme has been used in thousands of patients since 1994. It acts like a naturally occurring enzyme, which is a protein in your body that targets the Gaucher cells. It works by breaking up the fat within the cells into smaller parts, called *glucose* and *ceramide*. Then the glucose and ceramide can be removed from the cell naturally. Infusion treatments usually take 1-2 hours and can be done at a treatment center or in your doctor's office. These infusions are usually scheduled by your nurses and/or doctors to create the best treatment plan for you.

Remember, the doctor is there to help YOU. So feel free to ask your doctor ANYTHING!



# “What does that mean?”

## The Wild World of Words in Gaucher Disease Type 1

When you first find out you have Gaucher disease type 1, you’ll meet with doctors, nurses, and other healthcare workers who will explain everything to you. That’s when you may start hearing odd-sounding words that you’ve never heard before.

If the doctor uses a word you do not know or says anything you don’t understand, ask him/her to explain it. Don’t be afraid to jump in and ask, “What does that mean?”

Below are some of the words you might hear used when talking about Gaucher disease type 1:

**Bone mineral density**—Amount of calcium and other minerals that are in your bones. People with Gaucher disease type 1 sometimes feel bone pain or other problems because of low bone density.

**Cerezyme**<sup>®</sup>—(also known as imiglucerase for injection) A medicine for Gaucher disease type 1 that patients get through injection or “infusion.”

**Enzyme**—A *protein* in your body that changes a substance into something the body can use. For example, *enzymes* help you turn food into *glucose*, which is a good sugar that your body uses for energy.

**Enzyme replacement therapy**—Treatment that replaces the missing enzyme in patients with Gaucher disease type 1.

**Gaucher disease**—Caused by a change to the gene that makes an enzyme called *glucocerebrosidase*. There are three main types of Gaucher disease, with type 1 the most common. It usually affects organs (like the spleen and liver), blood, and bones.

**Genes**—Your body contains thousands of *genes*, which carry information that determines your “characteristics,” or things that decide how your body is made up, or how you look or feel. Genes are passed down to you through your parents. For example, if both of your parents have green eyes, you might get green eyes from them. You also get the gene for Gaucher disease type 1 from your parents.

**Genetic disorder**—When a gene is different than it should be, it is called a *mutation*. A mutation in a single gene can cause a problem with how our body works, known as a *genetic disorder*, such as Gaucher disease type 1.

**Glucocerebrosidase**—An enzyme that breaks down a fat (called a *lipid*) that our bodies make. The fat is called *glucocerebroside*. If your body does not make enough glucocerebrosidase, it cannot turn glucocerebroside into a useful substance. This happens in Gaucher disease type 1.

**Glucocerebroside**—A type of fat the cells in our body produce. Our body uses glucocerebroside to build cell membranes. Cell membranes are kind of like a jacket that surrounds the cell. When cells get old, our body makes new replacement cells by recycling material from the old cells, including glucocerebroside. If your body cannot break down the fatty glucocerebroside, it builds up in your cells and leads to Gaucher disease type 1.

**Hepatomegaly**—When your liver is larger than it should be. This can happen when you have too much glucocerebroside (defined above).

**Hepatosplenomegaly**—When both your liver and spleen are larger than they should be.

**Imiglucerase for injection**—Another name for Cerezyme<sup>®</sup>, a drug used in enzyme replacement therapy to treat people with Gaucher disease type 1.

**Intravenous infusion**—A method for getting medicine directly into your blood. A needle is attached to a thin tube, which is attached to the bag of medicine. The needle is inserted into your vein—usually with a quick prick—and the medicine goes into your bloodstream.

**Lysosomal storage disorders**—Rare diseases caused by a problem with one or more enzymes. These diseases include Gaucher disease, Tay-Sachs disease, Fabry disease, and Pompe disease.

**Lysosome**—A special compartment (imagine a little room) inside a cell that contains enzymes for breaking down certain substances, like glucocerebroside.

**PORT-A-CATH**<sup>®</sup>—A device made of metal and plastic that is placed under your skin to help deliver the enzyme therapy throughout your body during treatments.

**Splenomegaly**—When your spleen is larger than it should be.

**Symptom**—Sign or feeling you have that can be caused by a disease, such as pain, tiredness, or bruising.

**Urticaria**—Itchy red bumps that suddenly appear on the skin, usually because of an allergic reaction.

Now that you know what these words mean, you’ll feel like you’ve learned a foreign language!

# Word Search

How many words below can you find that relate to Gaucher disease type 1?

(Note: Words may appear upwards, downwards, across, backwards, or diagonally!)

BONE  
CEREZYME  
ENZYME  
GAUCHER

GENES  
GENZYME  
GLUCOCEREBROSIDASE  
GLUCOCEREBROSIDE

HEPATOMEGALY  
HEPATOSPLENOMEGALY  
IMIGLUCERASE  
INFUSION

LYSOSOME  
SPLENOMEGALY  
SYMPTOM  
URTICARIA

T G S V J S O R Q P E K C A T V E F W Y  
D M D M A R G C H Y N G O O M D U X X L  
N W I P R E X W L C Z V Q U I I K N V A  
E N O B N N K Z Y W Y P D S R I G A R G  
S O G E V P V W B B M G O G V F W R G E  
I E S A D I S O R B E R E C O C U L G M  
E M K S J L A C S A B S Y X M G V Y Q O  
K P I D Y H B W T E X B H E G G Y J S N  
F E Y G A H B F R U M F M T N U Y R P E  
K F Q B L G X E B U N Y N E Z S D J L L  
K R T A F U C I R Y Z O K M H M H Z E P  
F D L Q A O C T R N F E I O D I N U N S  
V Y Y Y C M I E E T E K R S W E P V O O  
K A G U T C L G R S M A B O U V R V M T  
Y O L T A A D W J A Y Z H S L F R Z E A  
K G W R K H P H H J S M F Y J R N O G P  
W P I E M Y Z E R E C E P L M Z H I A E  
G A U C H E R O E L U V K T V K T K L H  
H E P A T O M E G A L Y T F O U C I Y T  
X Z E U G X X K D E P G H A O M Z E F Q

(Answer key on next page)

# “Wow, I know just how you feel!”

## Connecting with other kids living with Gaucher disease type 1

Living with Gaucher disease type 1 can cause changes in your life that can be difficult. And many of your friends may not be able to understand. During times like these, it’s good to know there are people and places you can go to that can help you cope with these changes.

The Cerezyme Website, [http://www.cerezyme.com/patient/living/cz\\_pt\\_living-gigi.asp](http://www.cerezyme.com/patient/living/cz_pt_living-gigi.asp), has interesting stories, like Gigi’s, a girl with Gaucher disease type 1. You can read about other kids your age who have similar experiences to yours.

### Gaucher Pen Pals Program

A great way to communicate with people your age who are also living with Gaucher disease type 1 is the *Pen Pals* program.

The company that makes the medication Cerezyme® (imiglucerase for injection), the Genzyme Corporation, has started the *Pen Pals* program to bring kids together who are going through the same experience. They may live with similar symptoms such as tiredness and bone pain, and may also have had enzyme replacement therapy. The program is open to kids and teenagers between the ages of 5 and 17 who are living with Gaucher disease.

Jennifer Pettengill, a senior case manager at Genzyme, who helped create the *Pen Pals* program, explained, “We match patients based on their interests and age. Our goal is to give each patient a connection to someone who is going through the same things they are.”

Pettengill added, “A lot of these patients can benefit from talking to someone their age with Gaucher. This program allows each patient to make more personal social connections. When it comes to the Gaucher community, there are meetings, which take place over the course of a few hours. Many kids and young adults, however, do not want to sit through these meetings. This program allows them to get involved in the Gaucher community in a unique way.”

To sign up for the *Pen Pals* program, just go to [www.cerezyme.com/patient/cz\\_pt\\_overview.asp](http://www.cerezyme.com/patient/cz_pt_overview.asp), click on the **Living with Gaucher** tab on the left, then click on **Pen Pals**. This will take you to a page where there is a link to download a questionnaire and permission form.

Join today!

## Patient Spotlight: Joseph McCabe

“For the infusions, a nurse puts a needle into my PORT-A-CATH®. The PORT-A-CATH is a small metal and plastic tool that is under my skin and helps with the infusions. It’s not fun having a PORT-A-CATH, but it helps with the infusions. After the infusion, I feel tired for about a day, but after that, I feel fine.”

Although Joseph’s mom feels he should not play sports, he finds other ways to stay active. Before he was diagnosed with Gaucher disease type 1, Joseph was a young boy who had little energy to play. His mother thought that maybe that was just the way he was. Then he started getting nosebleeds.

“He always got nosebleeds. They were awful. They would be so bad that I couldn’t even take him to preschool,” his mother recalled. “During his annual



Joseph McCabe

exam, we talked to the doctor about how Joseph got terrible nosebleeds, how easily he became fatigued, and how he bruised so easily when he had never been involved in contact sports. The doctor gave us a diagnosis of Lyme disease.”

Knowing that it wasn’t Lyme disease, Joseph’s mom turned to a specialist. “Dr Claussen is my angel. He asked so many questions about Joseph’s nosebleeds, bruising, and fatigue. The day after Christmas, when

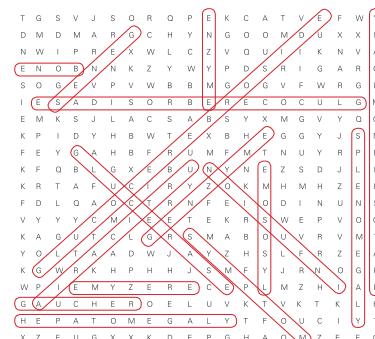
Joseph was 5 years old, Dr Claussen told me, ‘Joseph has Gaucher disease.’ I was so scared.”

Since then, Joseph has had his enzyme infusions at home, every 2 weeks through his PORT-A-CATH. “Joseph can relax, play video games, or do his homework while he is getting his infusions.”

Joseph gets exams every year from Dr Claussen, complete with blood work and tests called *magnetic resonance imaging*, or *MRI*, and *magnetic resonance spectroscopy*, or *MRS*.

“Even if his numbers don’t improve each year, we feel he won’t get any worse because he was diagnosed so early,” said Joseph’s mom. “It’s not scary anymore.”

### Word Search Answer Key





## We'd love to hear from you.

What information would you like to see covered in future issues of *Horizons*?

---

---

How can Genzyme better communicate information on Gaucher disease?

---

---

What channels are you using to keep informed?

- |  |  |
|--|--|
| <input type="checkbox"/> Your physician              | <input type="checkbox"/> Cerezyme supply website |
| <input type="checkbox"/> Your Genzyme case manager   | <input type="checkbox"/> Other _____             |
| <input type="checkbox"/> National Gaucher Foundation |  |

How would you like to receive your *Horizons* publications in the future?

- Print       Email \_\_\_\_\_       Digital download

Would you be interested in sharing your story of living with Gaucher disease?

If so, please fill in the following:

Name \_\_\_\_\_

Address \_\_\_\_\_

City \_\_\_\_\_ State \_\_\_\_\_ Zip \_\_\_\_\_

Email \_\_\_\_\_

Phone \_\_\_\_\_

Place  
Postage  
Here

*Horizons*

c/o Peter Ciszewski  
9 Lefurgy Ave.  
Hastings-on-Hudson, NY 10706