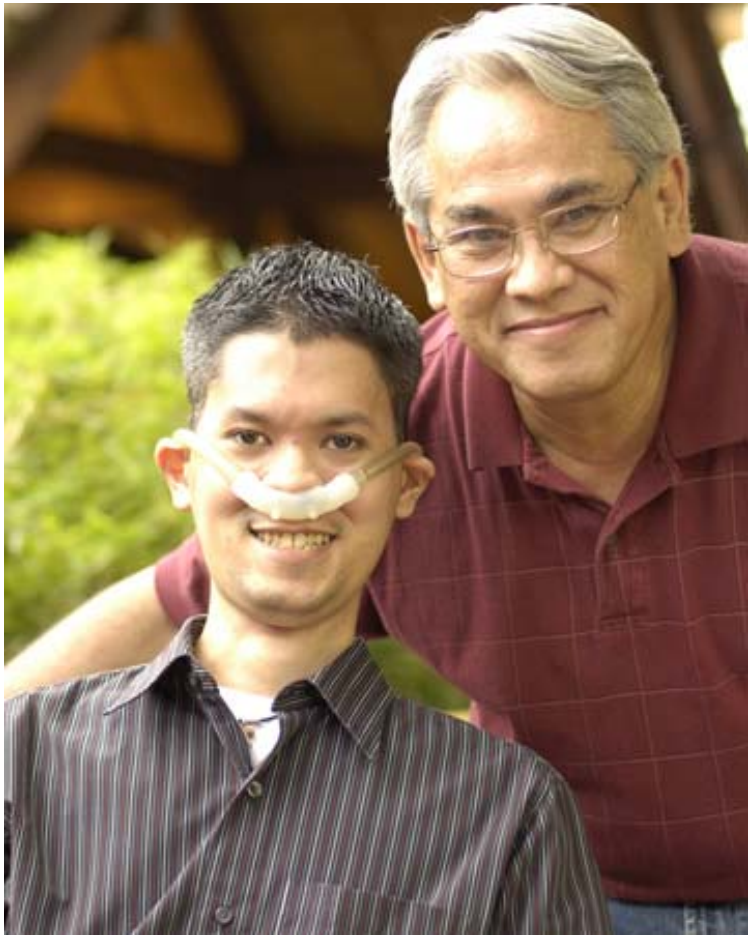
Rare Diseases and the Media

Bo Piela
Senior Director
Corporate Communications

Our Role

- Provide treatments for four ultra-rare disorders:
  - Gaucher disease
  - Fabry disease
  - MPS I
  - Pompe disease

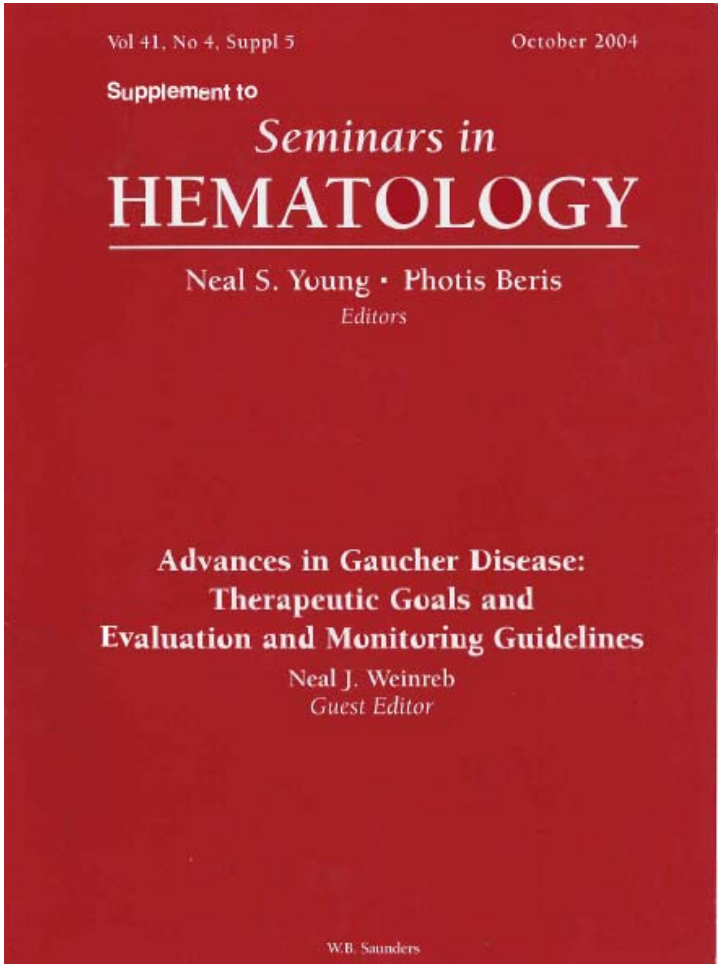


Our Responsibility

- Facilitate early diagnosis
- Advance understanding of disease and product
- Enable access to treatment
- Advance the field

Our Tools

- Support medical education
  - Symposia
  - Grand rounds
- Develop evidence-based disease management
  - Guidelines
  - Registries
  - Publications
- Generate press coverage

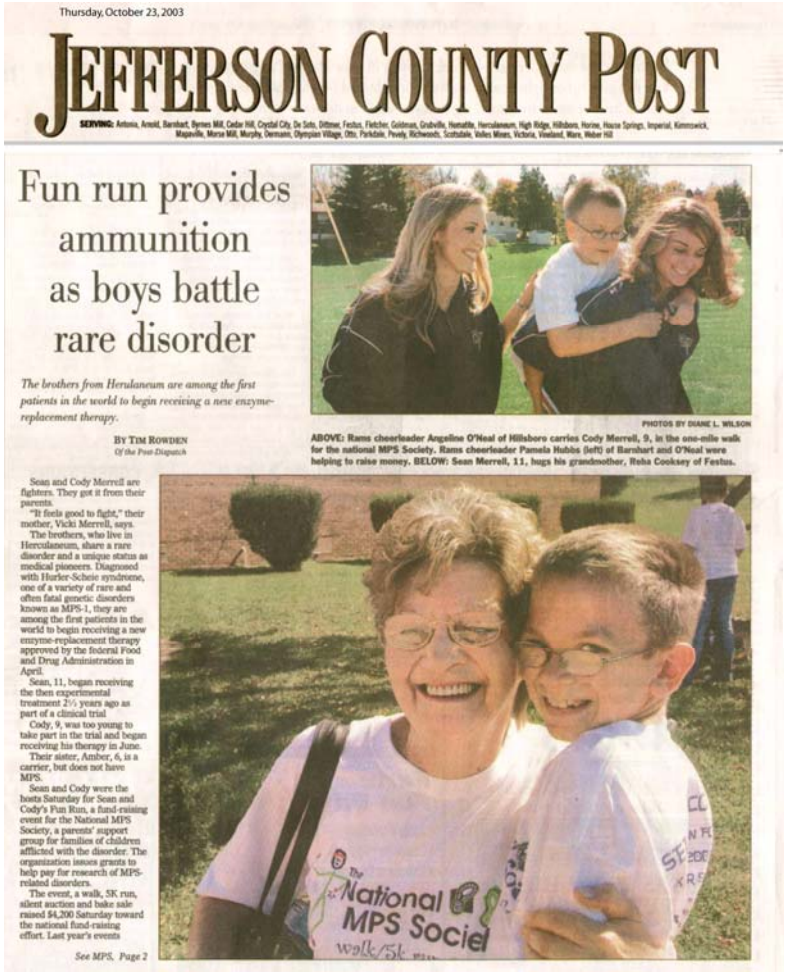


Challenges

- Rare diseases are rarely covered:
  - Small patient populations
  - Unfamiliar subject
  - Complex medical story

Opportunities

- Focus on individual patient stories



Thursday, October 23, 2003

JEFFERSON COUNTY POST

SERVING: Antonia, Arnold, Barnhart, Byrnes Mill, Cedar Hill, Crystal City, De Soto, Dittmer, Festus, Fletcher, Goldman, Grubville, Hematite, Herculaneum, High Ridge, Hillsboro, Horine, House Springs, Imperial, Kimmswick, Maxville, Morse Mill, Murphy, Dermann, Olympian Village, Otto, Parkdale, Pevely, Richwoods, Scotsdale, Valles Mines, Victoria, Vineland, Ware, Weber Hill

Fun run provides ammunition as boys battle rare disorder

The brothers from Herulaneum are among the first patients in the world to begin receiving a new enzyme-replacement therapy.

BY TIM ROWDEN
Of the Post-Dispatch

Sean and Cody Merrell are fighters. They got it from their parents.

"It feels good to fight," their mother, Vicki Merrell, says.

The brothers, who live in Herculaneum, share a rare disorder and a unique status as medical pioneers. Diagnosed with Hurler-Scheie syndrome, one of a variety of rare and often fatal genetic disorders known as MPS-1, they are among the first patients in the world to begin receiving a new enzyme-replacement therapy approved by the federal Food and Drug Administration in April.

Sean, 11, began receiving the then experimental treatment 2½ years ago as part of a clinical trial.

Cody, 9, was too young to take part in the trial and began receiving his therapy in June.

Their sister, Amber, 6, is a carrier, but does not have MPS.

Sean and Cody were the hosts Saturday for Sean and Cody's Fun Run, a fund-raising event for the National MPS Society, a parents' support group for families of children afflicted with the disorder. The organization issues grants to help pay for research of MPS-related disorders.

The event, a walk, 5K run, silent auction and bake sale raised $4,200 Saturday toward the national fund-raising effort. Last year's events

See MPS, Page 2

PHOTOS BY DIANE L. WILSON

ABOVE: Rams cheerleader Angeline O'Neal of Hillsboro carries Cody Merrell, 9, in the one-mile walk for the national MPS Society. Rams cheerleader Pamela Hubbs (left) of Barnhart and O'Neal were helping to raise money. BELOW: Sean Merrell, 11, hugs his grandmother, Reba Cooksey of Festus.

Opportunities

- Focus on Firsts

Newsday
Tuesday, June 17, 2003

Enzyme Therapy Scores A New Win

Boost for kids with rare genetic disease

By Jamie Talan
STAFF WRITER

Ten-year-old Sean Merrell and his family have logged thousands of weekly miles flying from their home in Herculaneum, Mo., to a hospital in Manhattan to be part of a study testing an experimental treatment for a rare genetic disease.

Thanks to the Merrells' commitment and that of others like them in proving the treatment's disease-slowing benefits, the Food and Drug Administration last month approved the new therapy, involving a genetically derived enzyme replacement.

It's the first treatment for Sean's illness, known as Hurler-Scheie's disease. Sean, his younger brother, Cody, and about a thousand other children in the United States inherited the genetic mutation, which causes a buildup of sticky sugar molecules whose normal job is to build connective tissue throughout the body. Children with the disease lack an enzyme that breaks down the sugar molecules so they can be easily eliminated from the body when they've completed their work.

The accumulation of this sticky substance can cause a long list of problems, including reduced growth; rubbery, thick skin; cloudy corneas; heart problems; joint stiffness and shortness of breath. Sean had a number of these medical problems, yet doctors failed to diagnose the genetic illness until he was 8.

"We are very lucky to have this treatment," said Vicki Merrell, Sean's mother. The boy has been part of a two-year study at New York University School of Medicine. Now that the enzyme treatment, which must be continued over a lifetime, has been approved, the family is trying to figure out how to pay for it. It could cost around $1,500 a week, a cost few could afford without the best insur-

See ENZYMES on A36

With the enzyme therapy that Sean Merrell, 10, has been receiving from his Manhattan doctor, Gustavo Charria, to combat Hurler-Scheie's disease, the boy's skin is no longer rubbery, his body is more flexible and he's more active.

New Enzyme Therapy Fights Genetic Disorder

ENZYMES from A31

ance coverage. The Merrells are still waiting to hear whether their insurance company will pay for the medicine.

Before the study, Sean had undergone heart surgery to unclog his valves, and carpal tunnel surgery for his wrists. "He is definitely doing better," his mother added. "He's got a lot more energy." Tests also have shown his liver and spleen — which had doubled in size — have returned to normal.

Enzyme replacement therapy for genetic diseases has quietly made its entrance into medicine during the last decade. While not holding the promise of gene therapy — a technique that permanently replaces the missing gene itself — a genetically engineered enzyme delivered on a routine basis carries out the task of a missing enzyme. In Hurler-Scheie's disease, it delivers the missing enzyme, called idurenidase, which goes into the organs and chews up the sticky substance.

There are about 40 fat-storage diseases like Hurler, called lysosomal-storage disorders. The most common is Gaucher's disease, which causes spleen enlargement, skin pigmentation and bone lesions. Federal scientists found during the 1960s that human placentas contained the missing enzyme involved in Gaucher's disease. They spent decades devising a way to replace the enzyme in patients, using the placentas.

By 1991, when they were successfully testing the enzyme in humans, federal scientists calculated that it would take about 20,000 human placentas to get enough of the substance to treat just one Gaucher's patient for a year.

Because there are 30,000 such patients in the world, that method was not realistic.

Scientists at Genzyme, a biotech company in Cambridge, Mass., began tinkering with the enzyme from human placenta and eventually created a genetically engineered form now used to treat patients.

The company went on to develop a similar enzyme-replacement therapy for another genetic storage disease, Fabry, and now Hurler. Costs of developing these medicines run into hundreds of millions of dollars, and the market is too small to recoup the investment, said Dr. Gerald Cox, medical director of Genzyme. Knowing that most families can't possibly find $50,000 to $200,000 a year to pay for treatment, Cox said the company has made "a vow that no patient will go without therapy."

In Hurler patients, the enzyme-replacement therapy breaks down the sticky substance. The recent study, and an earlier one, proved that this reduces symptoms, said Dr. Gustavo Charria, a scientist at New York University School of Medicine who is also Sean Merrell's doctor and a lead scientist in the study.

For Sean, that means his skin is no longer rubbery, he's more active and his body is more flexible. For the past six months, he's received his weekly infusion, an intravenous drip over a four-hour period, closer to home, in Missouri, and he returns to Manhattan every three months for follow-up.

"These patients will need medication for life," said Charria, who has been following six other patients with the same genetic disease.

Enzyme-replacement therapy is making a huge difference in the lives of children with the first few genetic diseases studied. At NYU, Charria sees the benefits through one little boy who just celebrated his third birthday. "He runs circles around everybody," the doctor says with a laugh. The boy has a similar storage disorder called Pompe's disease, which affects muscles and the heart.

"Without medication, he simply would have died," Charria said.

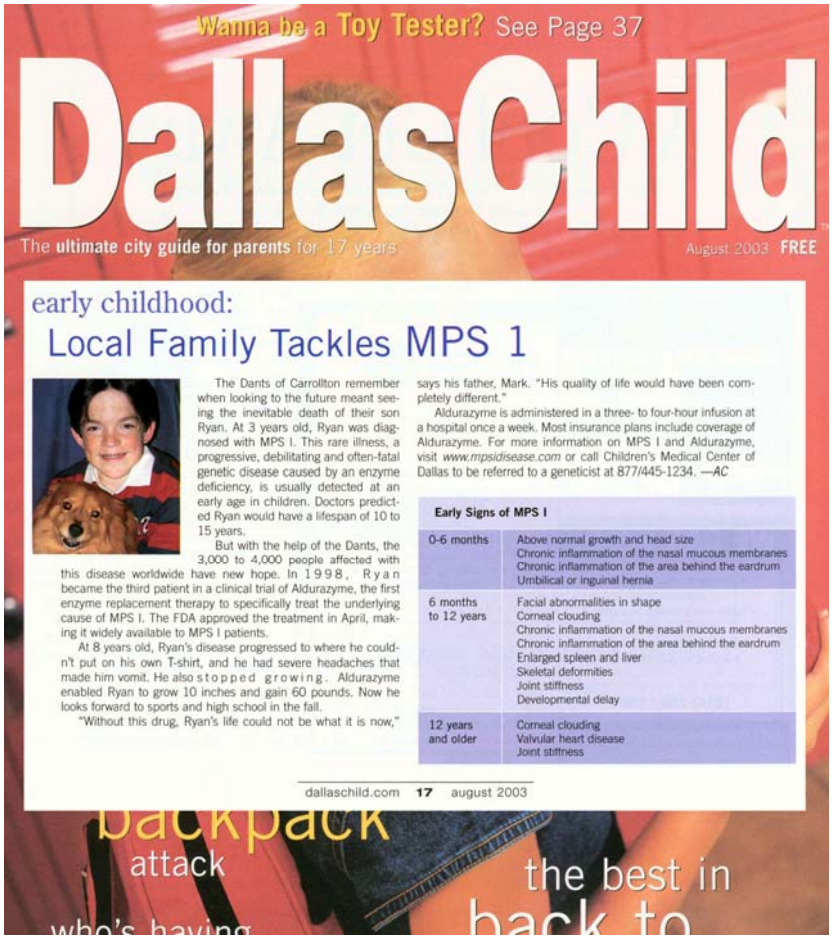
The problem with these therapies is that the enzymes can't pass through into the brain. Eventually, the harmful substances accumulate there, resulting in brain damage, Charria said.

In time, the hope is that gene therapy will progress, so a permanent repair of such genetic diseases can be made. But many gene therapy trials have been put on hold after the death of a young male patient in 1999 at the University of Pennsylvania. It's not clear how scientists will overcome the obstacles.

"But for now, we are watching these children grow and thrive, their symptoms not gone, but in control," Charria said of enzyme therapy recipients. "What more could you ask for?"

Opportunities

- Make it local



Wanna be a Toy Tester? See Page 37

DallasChild

The ultimate city guide for parents for 17 years

August 2003 FREE

early childhood:

Local Family Tackles MPS 1

The Dants of Carrollton remember when looking to the future meant seeing the inevitable death of their son Ryan. At 3 years old, Ryan was diagnosed with MPS I. This rare illness, a progressive, debilitating and often-fatal genetic disease caused by an enzyme deficiency, is usually detected at an early age in children. Doctors predicted Ryan would have a lifespan of 10 to 15 years.

But with the help of the Dants, the 3,000 to 4,000 people affected with this disease worldwide have new hope. In 1998, Ryan became the third patient in a clinical trial of Aldurazyme, the first enzyme replacement therapy to specifically treat the underlying cause of MPS I. The FDA approved the treatment in April, making it widely available to MPS I patients.

At 8 years old, Ryan's disease progressed to where he couldn't put on his own T-shirt, and he had severe headaches that made him vomit. He also stopped growing. Aldurazyme enabled Ryan to grow 10 inches and gain 60 pounds. Now he looks forward to sports and high school in the fall.

"Without this drug, Ryan's life could not be what it is now," says his father, Mark. "His quality of life would have been completely different."

Aldurazyme is administered in a three- to four-hour infusion at a hospital once a week. Most insurance plans include coverage of Aldurazyme. For more information on MPS I and Aldurazyme, visit *www.mpsidisease.com* or call Children's Medical Center of Dallas to be referred to a geneticist at 877/445-1234. —*AC*

Early Signs of MPS I

0-6 months	Above normal growth and head size Chronic inflammation of the nasal mucous membranes Chronic inflammation of the area behind the eardrum Umbilical or inguinal hernia
6 months to 12 years	Facial abnormalities in shape Corneal clouding Chronic inflammation of the nasal mucous membranes Chronic inflammation of the area behind the eardrum Enlarged spleen and liver Skeletal deformities Joint stiffness Developmental delay
12 years and older	Corneal clouding Valvular heart disease Joint stiffness

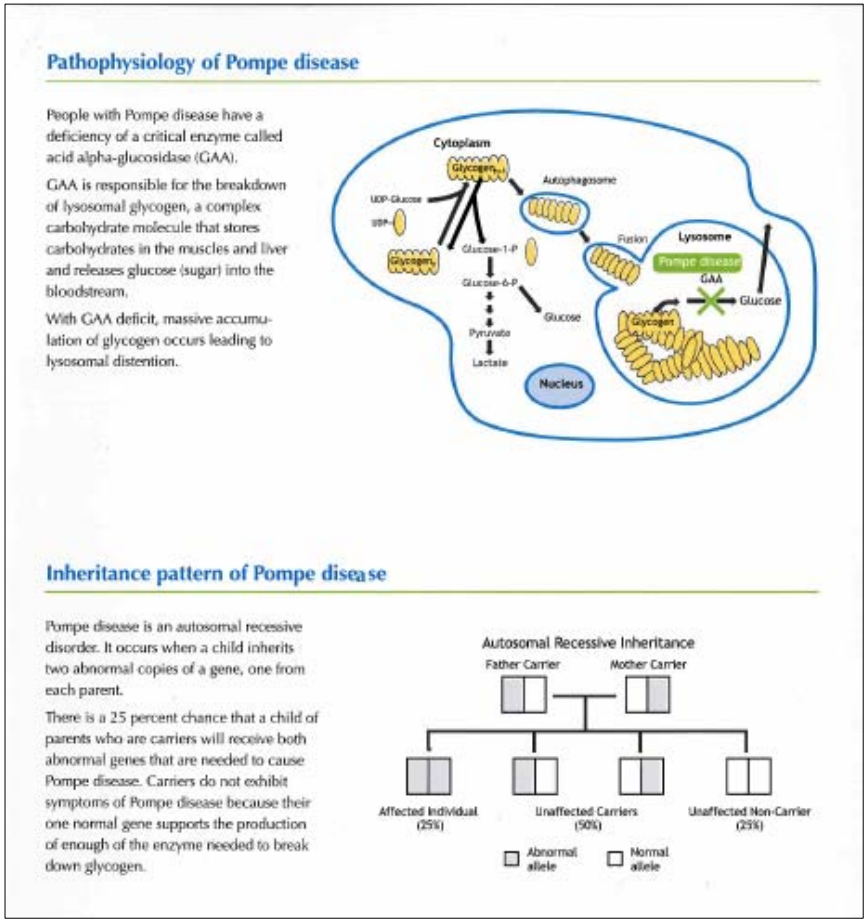
dallaschild.com 17 august 2003

backpack attack

the best in back to

Opportunities

- Make it easier for journalists



Pathophysiology of Pompe disease

People with Pompe disease have a deficiency of a critical enzyme called acid alpha-glucosidase (GAA).

GAA is responsible for the breakdown of lysosomal glycogen, a complex carbohydrate molecule that stores carbohydrates in the muscles and liver and releases glucose (sugar) into the bloodstream.

With GAA deficit, massive accumulation of glycogen occurs leading to lysosomal distention.

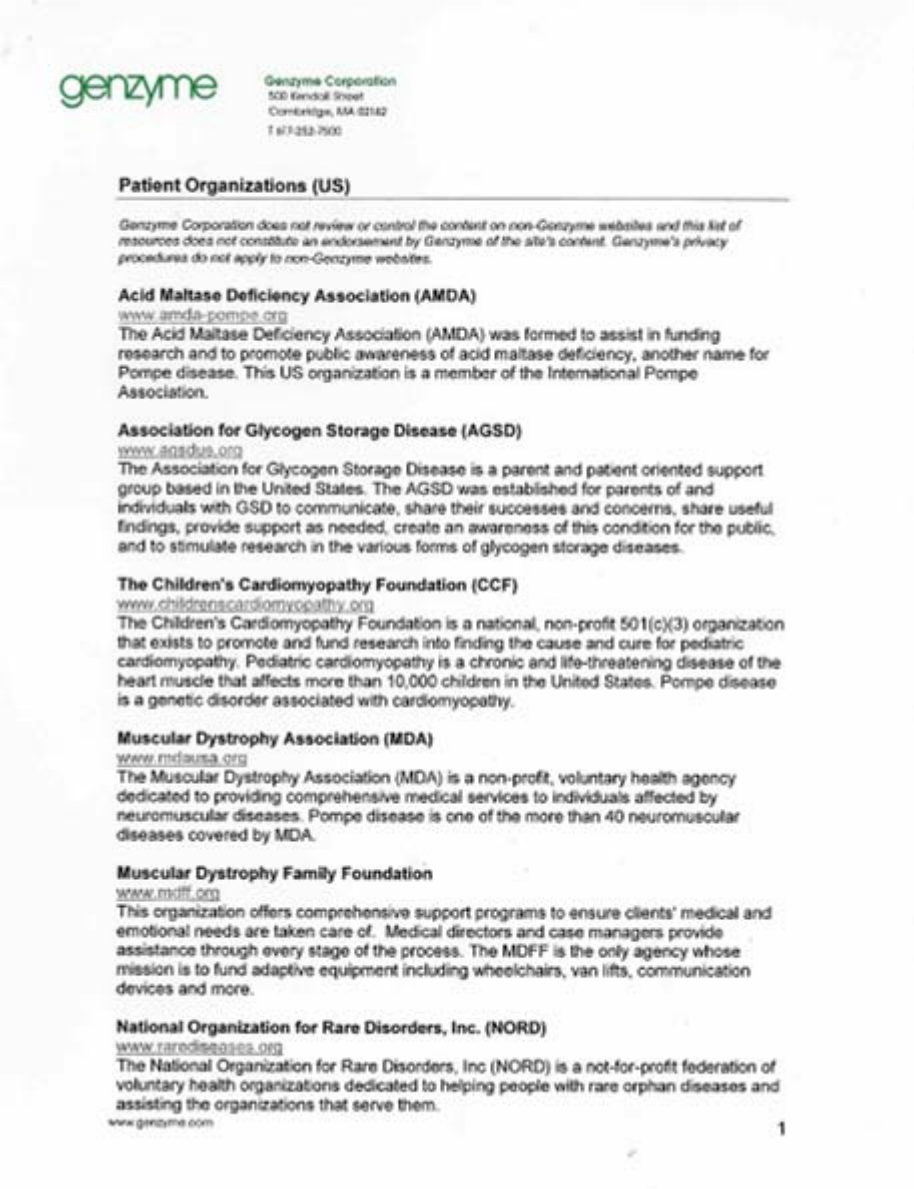
Inheritance pattern of Pompe disease

Pompe disease is an autosomal recessive disorder. It occurs when a child inherits two abnormal copies of a gene, one from each parent.

There is a 25 percent chance that a child of parents who are carriers will receive both abnormal genes that are needed to cause Pompe disease. Carriers do not exhibit symptoms of Pompe disease because their one normal gene supports the production of enough of the enzyme needed to break down glycogen.

Opportunities

- Collaborate



genzyme

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Patient Organizations (US)

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Acid Maltase Deficiency Association (AMDA)
www.amda-pompe.org
The Acid Maltase Deficiency Association (AMDA) was formed to assist in funding research and to promote public awareness of acid maltase deficiency, another name for Pompe disease. This US organization is a member of the International Pompe Association.

Association for Glycogen Storage Disease (AGSD)
www.agsdus.org
The Association for Glycogen Storage Disease is a parent and patient oriented support group based in the United States. The AGSD was established for parents of and individuals with GSD to communicate, share their successes and concerns, share useful findings, provide support as needed, create an awareness of this condition for the public, and to stimulate research in the various forms of glycogen storage diseases.

The Children's Cardiomyopathy Foundation (CCF)
www.childrenscardiomyopathy.org
The Children's Cardiomyopathy Foundation is a national, non-profit 501(c)(3) organization that exists to promote and fund research into finding the cause and cure for pediatric cardiomyopathy. Pediatric cardiomyopathy is a chronic and life-threatening disease of the heart muscle that affects more than 10,000 children in the United States. Pompe disease is a genetic disorder associated with cardiomyopathy.

Muscular Dystrophy Association (MDA)
www.mdausa.org
The Muscular Dystrophy Association (MDA) is a non-profit, voluntary health agency dedicated to providing comprehensive medical services to individuals affected by neuromuscular diseases. Pompe disease is one of the more than 40 neuromuscular diseases covered by MDA.

Muscular Dystrophy Family Foundation
www.mdff.org
This organization offers comprehensive support programs to ensure clients' medical and emotional needs are taken care of. Medical directors and case managers provide assistance through every stage of the process. The MDFF is the only agency whose mission is to fund adaptive equipment including wheelchairs, van lifts, communication devices and more.

National Organization for Rare Disorders, Inc. (NORD)
www.rarediseases.org
The National Organization for Rare Disorders, Inc (NORD) is a not-for-profit federation of voluntary health organizations dedicated to helping people with rare orphan diseases and assisting the organizations that serve them.

www.genzyme.com 1

A reality

- Cost story supersedes clinical story

The Boston Globe

SATURDAY, APRIL 29, 2006

High-priced Genzyme drug is OK'd

1st remedy for rare Pompe disease

By Stephen Heuser
GLOBE STAFF

Genzyme Corp. won federal approval yesterday for the first drug to treat a rare genetic disorder called Pompe disease, an intravenous treatment that will be one of the most expensive in the world at more than $200,000 per patient annually.

The drug, Myozyme, treats an incurable disease that causes muscle wasting in adults and fatal heart and lung failure in infants. It is currently known to affect only about 1,000 people.

"This is a special day for people across the Pompe community and at Genzyme," said Henri Termeer, the Cambridge company's chief executive.

By developing life-saving treatments for extremely rare illnesses, Genzyme has become the largest drug company in Massachusetts and one of the biggest biotechnology companies in the world. It has also emerged as a target for critics who say the healthcare system can't afford a growing stream of such high-priced drugs.

Myozyme joins the company's first two drugs for similarly rare genetic disorders, Cerezyme and Fabrazyme, which together treat just over 6,000 patients and last year made Genzyme over $1 billion. Myozyme could generate more than $100 million annually by 2010, according to analysts.

"We've been waiting for this for a long time," said Marylyn House of San Antonio, secretary of the Acid Maltase Deficiency Association, a Pompe patients

GENZYME, Page A4

SMALL MARKET, LARGE PRICETAG

CEREZYME (Approved in 1994)
Treats Gaucher disease
Patients on drug: 4,500
Sales last year: $932m

FABRAZYME (Approved in 2003)
Treats Fabry disease
Patients on drug: 1,700
Sales last year: $305 million

MYOZYME (Approved yesterday)
Treats Pompe disease
Patients on drug: 280
Sales: N/A

SOURCE: Genzyme Corp. GLOBE STAFF

Reprinted from THE WALL STREET JOURNAL.

WEDNESDAY, NOVEMBER 16, 2005

Why Genzyme Can Charge So Much for Cerezyme

BY GEETA ANAND

Why is the price of Genzyme Corp.'s drug for Gaucher disease $200,000 a year for the average patient?

The reason is simple -- Genzyme charges that much today, 14 years after the first version came to market, because it can. There is no competition, patients are desperate and most insurers pay.

Genzyme says it keeps the price high to help it pay for the hunt for other drugs and also to fund programs that allow it to give away a small part of its production.

The company makes a profit of more than 90% on the drug, excluding market-patients. So Ms. Lees and others say they must struggle to keep their insurance coverage, because they can single-handedly drive up the costs of the plans they join to unaffordable levels. Because the drug is a treatment, not a cure, the high costs to insurers continue indefinitely. Genzyme says there are only about 4,500 patients on Cerezyme, the Gaucher medicine.

When Genzyme first brought the drug to market in 1991, it was difficult and expensive to produce, notes Henri Termeer, the company's chief executive. It took 22,000 human placentas to make enough treatment for one patient annually. The drug cost $1.90 for each unit their drugs.

Genzyme's profit on Cerezyme has also allowed it to bring new treatments to market for two other rare diseases. It has purchased many small companies to expand into a diversified drug company with cancer, kidney disease and diagnostic products, among others.

-- Geeta Anand

(See related article: "The Most Expensive Drugs --- Uncertain Miracle: A Biotech Drug Extends a Life, But at What Price? --- For Ms. Lees, Treatment Bill Now Totals $7 Million; Her Bones Keep Crumbling --- Guilt of Another $1,400 Day" -- WSJ Nov. 16, 2005)

Be transparent

OUR COMMITMENT

- Patients
  - Unmet Medical Needs
  - Free Drug Programs
  - Humanitarian Programs
  - Cost of Treatment
- Community
- Environment
- Corporate Governance

The Cost of Enzyme Replacement Therapy

One of Genzyme's responsibilities as a pioneer in the development of treatments for rare disorders has been to participate in the debate surrounding the cost of these treatments. In the early 1990s, we developed the first therapy for people with Type 1 Gaucher disease, an extremely rare inherited disorder with potentially disabling and life-threatening complications. Since then, we have introduced treatments for three similar rare disorders, transforming the lives of patients throughout the world who previously had no other treatment options.

From the beginning, we have been transparent in our approach to setting a price for these products, and we have openly explained the factors affecting their cost with physicians, patients, insurers, government authorities, journalists and others. Given the interest in this subject, we feel it is important to share our perspective even more broadly by providing answers to some frequently asked questions:

What drugs has Genzyme developed for rare diseases?

Treatment	Disease	First Approved	Patients on therapy as of January 1, 2008
Cerezyme (imiglucerase for injection)	Type 1 Gaucher disease	1991 (first-generation product Ceredase)	5,200
Fabrazyme (agalsidase beta)	Fabry disease	2001	2,200
Aldurazyme (laronidase) with BioMarin Pharmaceutical	MPS I	2003	600
Myozyme (alglucosidase alfa)	Pompe disease	2006	900

Highlight

Gaucher Initiative Helps Patients in the Developing World

The Gaucher Initiative is a humanitarian program through which Genzyme provides Cerezyme to Gaucher patients in developing countries. It is one of several charitable access programs sponsored by the company. Face to Face documents the journey of Tonye Tierney to meet patients, their families and their physicians throughout the world and to see how the Gaucher Initiative continues to affect their lives

Redefining “media”

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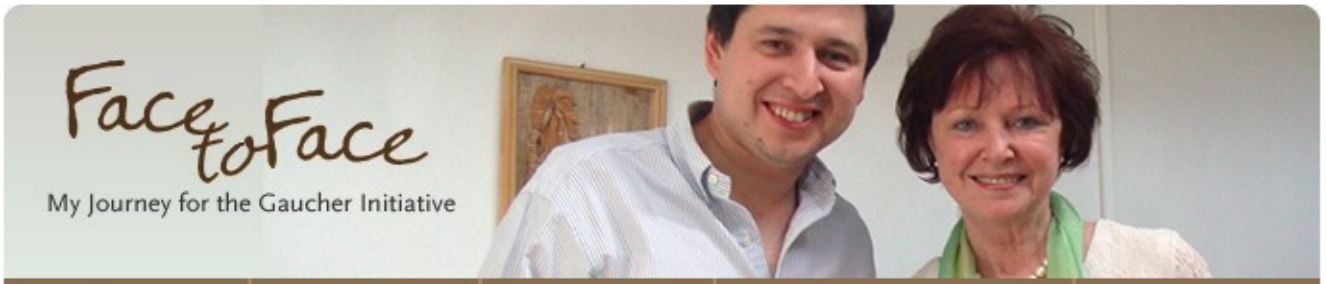

Journal Home | Destinations | About Tomye | The Gaucher Initiative

Through this journal I will be documenting my journey to meet the faces behind the Gaucher Initiative, a humanitarian program that provides Gaucher patients in developing countries with the enzyme replacement therapy, Cerezyme, free of charge. I have the rare opportunity to meet with patients and their families and physicians to see how the Gaucher Initiative has and continues to affect their lives.

More from Cairo

Friday, April 25, 2008

A second day of escape from the heat! Dr. Reda Abdellah of Project HOPE's Egypt staff organized a comprehensive training workshop day. This is a small part of the group of about 60 physicians from around the country who came and participated. We timed the photo op for lunch break, so most of the group not only stayed inside the cool building, but got a head start on the feast!

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More from Cairo

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Here I am with Dr. Abeer from Mansoura and Dr. Iman Mazouk from Alexandria, during a break in the training workshop at National Training Institute.

DESTINATIONS

Hanoi, Vietnam

Delhi, India

São Vicente, Cape Verde

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Written by Tomye Tierney
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