

UNDERSTANDING  
**Pompe Disease**  
From diagnosis to action



genzyme

genzyme

# UNDERSTANDING Pompe Disease

From diagnosis to action



A 2-month-old baby is admitted to the hospital with breathing problems brought on by a bad cold. An x-ray reveals a greatly enlarged heart.

A 2-year-old child who has just started walking has an odd walk that is a cross between a waddle and a limp. As an infant, he had trouble turning over and holding up his head.

A college student becomes winded and her muscles ache when she climbs a flight of stairs.

*All three scenarios above describe people who have Pompe disease.*

*This rare inherited muscle disease shows up in different ways in infants, children, and adults. If you or your child has been diagnosed with Pompe disease, it can be difficult to imagine how this disease will affect your health or the health of those you care for. And because the disease is rare, it is not easy to know where to turn for information, guidance, and support.*

*We hope that gaining a better understanding of Pompe disease will help you as you begin to move from diagnosis to action.*

## How this booklet can help you

**This booklet provides information about Pompe disease and how it is diagnosed and managed. It also offers practical tips on living with Pompe disease. As you read through the booklet, keep in mind:**

### ***You are not alone.***

There are active patient groups and caring health professionals throughout the world who can help you manage the challenges of living with Pompe disease.

### ***Being informed can help you take a more active role in your care.***

Learning about Pompe disease and disease management can help you work with your healthcare team to get the best possible care.

### ***Treatment is available.***

Over the last several years, a great deal has been learned about Pompe disease through the collective efforts of healthcare professionals, scientific experts, and patients worldwide. Myozyme® (alglucosidase alfa), the first and only enzyme replacement therapy (ERT) to treat the underlying cause of Pompe disease, has been approved for use in several countries around the world. Although ERT is not a cure, it can slow or halt the progression of Pompe disease.

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*The information in this booklet is not meant to take the place of professional medical advice. Always consult with your healthcare provider if you have questions and/or concerns about your medical condition.*

# What is Pompe disease?

Pompe disease is a rare neuromuscular disorder that occurs in infants, children, and adults who inherit a defective gene from their parents. It is one of more than 40 genetic diseases that are known as **lysosomal storage disorders**.

Pompe disease is a progressive disease, which means it gets worse over time. The disease causes progressive muscle weakness and often leads to breathing problems. Since the disease affects muscles, it is similar to other **neuromuscular disorders**, such as the muscular dystrophies. Pompe disease is also considered a **metabolic muscle disease** because muscle weakness occurs as a result of changes that take place inside the body's cells. Since glycogen (GLY-co-jen) builds up in muscle cells, the disease is often considered a **glycogen storage disease**.

Pompe disease is named after J.C. Pompe, the Dutch doctor who first described the disorder in 1932 after observing an infant with severe muscle weakness and a greatly enlarged heart.

There are several ways to pronounce the name of the disease. In different parts of the world, you may hear "pom-PAY," "POM-puh," or "pom-PEE."



*Pompe disease affects people from around the world*

Pompe disease is also referred to as:

- Acid alpha-glucosidase deficiency
- Acid maltase deficiency (AMD)
- Glycogen storage disease (GSD) type II
- Glycogenosis II
- Lysosomal alpha-glucosidase deficiency

**NOTE:** In scientific articles, the enzyme acid alpha-glucosidase may be written as acid  $\alpha$ -glucosidase or shortened to  $\alpha$ -glu or AGLU. It is also commonly shortened to GAA. GAA actually stands for glucosidase acid alpha, which is the name given to the gene (not the enzyme).

## How many people have Pompe disease?

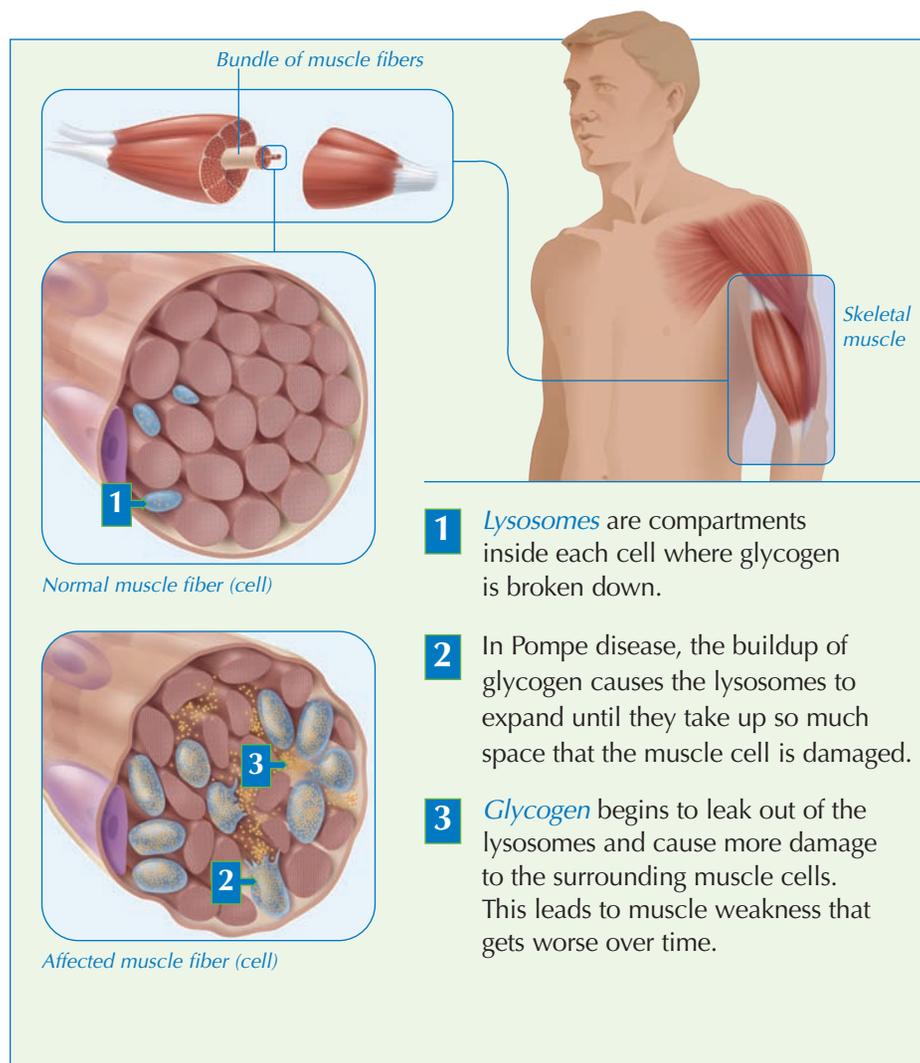
It is estimated that there are between 5,000 and 10,000 people around the world living with the symptoms of Pompe disease.

As with many rare diseases, it is difficult to know exactly how many people are actually affected. Based on studies conducted in the Dutch population and in New York City, it is estimated that Pompe disease occurs in 1 in 40,000 live births worldwide.

Pompe disease affects both men and women equally. Although the disease occurs in all ethnic groups, it does appear at a higher rate in the African American population, as well as in those of Southern Chinese and Taiwanese descent.

## What causes Pompe disease?

In people with Pompe disease, there is a defect in a gene named GAA. This gene is responsible for making an enzyme called acid alpha-glucosidase (AL-fa glue-CO-sih-days) within the lysosome, a compartment in the cell. Enzymes are proteins that do specific jobs to help keep the cells in the body working normally. The job of acid alpha-glucosidase is to break down lysosomal glycogen, a form of sugar stored in muscle cells throughout the body. In people with Pompe disease, this enzyme is either missing or in short supply.



## The spectrum of Pompe disease

Although the genetic defect that causes Pompe disease is present at birth, symptoms may show up at any time from infancy through adulthood.

In fact, one of the most striking features of the disease is how much it varies from one person to another primarily in:

- The age at which symptoms first appear
- How fast the disease progresses
- The degree to which other organs are affected

**In general, the earlier symptoms show up, the more severe the disease is likely to be.** The disease progresses rapidly in infants who have little or no enzyme activity.

By contrast, Pompe disease tends to progress more variably in children and adults who have at least some acid alpha-glucosidase enzyme activity.

*Pompe disease is a single disease that affects people differently*

## How do you get Pompe disease?

Pompe disease is genetically inherited or passed on when both parents have a defective gene. Genes are made from DNA, the chemical material that contains the instructions for every process in the human body and every other feature that makes us unique. Our genes are arranged on 23 pairs of chromosomes. One of those pairs (called sex chromosomes) determines whether a person will be male or female. The other 22 pairs (called autosomes) determine traits that are not related to a person's sex, such as eye color and height. Pompe disease affects males and females equally because it is inherited through a gene that is carried on one of the 22 pairs of autosomes.

### Some genes contain codes for making the enzymes that help the body function

If there is a mutation, or change, in a gene responsible for making a specific enzyme, then that enzyme may not work properly or may not be made. In Pompe disease, there are mutations in the gene for acid alpha-glucosidase. Pompe disease is an autosomal recessive disorder. This means you only inherit Pompe disease if you receive 2 copies of the defective GAA gene — 1 from each parent. Known Pompe mutations can be identified using genotype testing, which examines the DNA in a blood sample. This is sometimes done when Pompe disease is suspected or when a presumed diagnosis has been made but not confirmed and may also be done when there is a family history of Pompe disease.

### Inheriting the gene

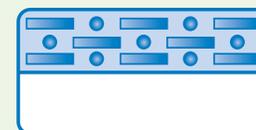
The chart on the next page shows what can happen if both parents are carriers of the defective gene. With each pregnancy, there is a 25% chance that the child will develop Pompe disease.

#### Other possible situations

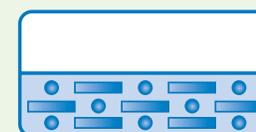
- If 1 parent has Pompe disease and the other is *not* a carrier, then all children will be carriers (and none will develop the disease).
- If 1 parent has Pompe disease and the other is a carrier (a very rare situation), then there is a 50% chance of having a child who will develop the disease and a 50% chance of having a child who is a carrier.

### Autosomal Recessive Inheritance

#### Parents



Father Carrier



Mother Carrier

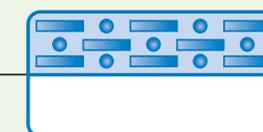
 = Normal gene

 = Abnormal (defective) gene

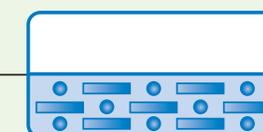
#### Children



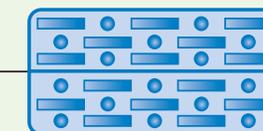
Unaffected Individual (25%)



Unaffected Carrier (25%)



Unaffected Carrier (25%)



Affected Individual (25%)

### If both parents are carriers (for each pregnancy):

- There is a 1 in 4 chance that the child will inherit 2 normal copies of the gene and **be unaffected**.
- There is a 2 in 4 chance that the child will inherit an abnormal copy from 1 parent and a normal copy from the other parent and **become a carrier**. Carriers will not develop Pompe disease but may pass on 1 defective gene to their children.
- There is a 1 in 4 chance that the child will inherit 2 abnormal copies of the gene and **develop Pompe disease**.

## What are the symptoms of Pompe disease?

Pompe disease is a genetic disorder that is always present at birth for those who are affected. However, symptoms may show up at any time from infancy through adulthood.

### Pompe disease is a single disease, but it affects people differently

Historically, Pompe disease had been described by physicians as either early-onset or late-onset, depending on when the patient's signs and symptoms first appear. However, Pompe disease may be best understood as a spectrum.

When symptoms occur during the first few months of life, Pompe disease progresses very rapidly and is almost always fatal by the age of 1 year, usually due to heart failure.

When symptoms occur after infancy, Pompe disease progresses more variably but can cause great difficulties as muscles throughout the body become weaker and weaker. The muscles most often affected are those used for breathing and mobility (the ability to move around).



*Pompe disease progression varies with every patient*

### Pompe disease presents in different ways in children and adults of all ages

**A 3-month-old baby** who seemed “normal” at birth is diagnosed with an enlarged heart. At 6 months he cannot sit up without support. Tests confirm the baby has Pompe disease. He becomes very weak and needs a feeding tube to help him gain weight. Even with his physical problems, he is a happy baby who greets everyone he sees with bright eyes and a smile.

**A 6-year-old** is having trouble climbing the stairs and doesn't have the energy to make it through gym class. After many tests, she is diagnosed with Pompe disease. Though she is too weak to play sports, she is able to ride a bicycle all through her childhood. In her twenties, her muscles become so weak that she must trade the bicycle for an adapted car — and a wheelchair. Rather than dwell on the loss of mobility, she sees the wheelchair as a way to stay active and independent.



**A 37-year-old man** starts falling asleep during the day and has trouble breathing when he is lying down. After 2 years of being tested and examined by different healthcare providers, he is diagnosed with Pompe disease. Looking back, he can recall that even though he was active in sports as a child, he could never quite keep up with the other kids on the team. He is still walking, though with some difficulty, and enjoys working full-time.

## Pompe disease in infants

Pompe disease in infants is a life-threatening condition that affects all of the major body organs. Without disease-specific treatment, the disease progresses rapidly. The infant may quickly become gravely ill. Without treatment, infants with Pompe disease are not likely to survive past the age of 1 year. The chart below lists the major symptoms.

### Major symptoms of Pompe disease in infants

#### Muscles

- Severe muscle weakness
- “Floppiness” due to loss of muscle tone
- Head lag — when you try to pull the baby up to a sitting position by grasping the hands or arms, the head drops back
- Frog-like position of legs
- Failure to meet developmental milestones such as rolling over, sitting up, crawling, and walking — or loss of milestones

#### Lungs

- Difficulty breathing
- Frequent respiratory infections
- Respiratory failure

#### Digestive (GI) Tract

- Feeding problems that occur as it becomes harder to breathe, suck, or swallow
- Failure to thrive or gain weight as rapidly as other babies the same age
- Enlarged liver
- GI discomfort: vomiting, regurgitation
- Enlarged tongue

#### Heart

- Enlarged heart
- Heart failure
- Heart rhythm changes

## Pompe disease in children and adults

Pompe disease in children and adults tends to progress more variably than in infants. Symptoms and severity can vary widely from one person to another. Major breathing problems, such as respiratory failure, can shorten the life span of people with Pompe disease. However, many are able to adapt to the challenges that the disease presents and continue with their lives. The chart below describes the major symptoms.

### Major symptoms of Pompe disease in children and adults

#### Muscles

- Progressive muscle weakness in the legs and hips
- Difficulty climbing stairs, running, or getting up from a chair
- Walking with swaying hips or a waddle
- Frequent trips and falls (loss of balance)
- Gradual loss of motor milestones that have been achieved, such as walking, running, or jumping (children)
- Low back pain
- Scoliosis (curvature of the spine)

#### Lungs

- Difficulty breathing, especially after exertion or when lying on your back
- Morning headaches, daytime sleepiness, shortness of breath and other signs of respiratory insufficiency
- Frequent respiratory infections, such as bronchitis and pneumonia

#### Digestive (GI) Tract

- Difficulty gaining or maintaining weight
- Problems chewing and/or swallowing

## What are some of the common health problems of Pompe disease?

Muscle weakness can lead to many different health problems. Yet not all people with Pompe disease have the same problems. For some, the impact will be severe, while for others, it will be milder.

In either case, when medical problems occur, it can be difficult to realize that they could be related to Pompe disease. But if you know what to expect, you may be better able to seek medical care that is tailored to specific needs. **Keeping track of symptoms and getting regular checkups can also help prevent health problems from becoming more serious.**

### Breathing problems may develop slowly or come on suddenly

Pompe disease weakens the diaphragm, one of the main muscles used for breathing (located just below the lungs and above the abdomen). It can be hard to breathe deeply, especially when lying on the back. Patients may feel short of breath and have trouble sleeping soundly. Patients may get morning headaches. It may also be hard to cough strongly enough to clear mucus from the lungs. This increases the risk for lung infections such as pneumonia.

Prolonged shallow breathing can result in respiratory insufficiency. This is a condition that occurs when reduced airflow into the lungs causes carbon dioxide to build up in the blood. This may cause shortness of breath, morning headaches, or sleep problems. It may make it hard to function during the day.

Respiratory insufficiency increases the risk for developing chest infections and respiratory failure, which means patients cannot breathe on their own. **Seeking treatment at the first sign of breathing problems can help prevent this medical emergency.**

### Weak muscles can cause eating problems

Pompe disease can weaken the muscles used for biting, chewing, and swallowing food. Infants who are very weak may not be able to suck from a breast or bottle. In infants, vomiting or regurgitation (when swallowed food mixes with stomach acids and flows back toward the throat) may also occur.

Eating problems may also prevent children and adults with Pompe disease from getting the nutrition their bodies require. They may lose weight or have trouble keeping it on. If breathing problems disrupt sleep, patients may lose their appetites or be too tired to eat.

### Weakness of the skeletal muscles can lead to mobility problems

Pompe disease weakens muscles throughout the body that enable patients to walk, keep their balance, stand up straight, and move freely. Increasing weakness and lack of muscle use can make muscles stiff and tight, which can be painful. Extra stress on joints and ligaments may also cause muscle aches and lower back pain. If muscles or tendons get too tight, they can “freeze” or become fixed in one position, forming a contracture. This can prevent normal movement. Muscle weakness may also lead to the development of scoliosis, a sideways curvature of the spine, which, in severe cases, can make it harder to breathe.

The pages that follow describe how Pompe disease is diagnosed, as well as ways to manage these medical problems and the discomfort they may cause.



## How is Pompe disease diagnosed?

Diagnosing Pompe disease can be challenging because many of the symptoms are similar to those of other diseases. In addition, symptoms often develop slowly and may not present themselves at the same time. It may be easier to diagnose infants with Pompe disease because the rapid progression and more pronounced symptoms may prompt healthcare providers to perform more extensive testing. Many healthcare providers, though, have never seen a patient with Pompe disease. In most cases, healthcare providers have to rule out other more common possible causes before considering a diagnosis of Pompe disease. If a patient complains only of breathing problems — or only of muscle weakness — Pompe disease may be overlooked. The patient may then receive another diagnosis, such as carnitine deficiency, hypothyroidism, or limb-girdle muscular dystrophy.

To learn more about the signs and symptoms shared by these diseases, visit [www.pompe.com](http://www.pompe.com).

### Medical specialists who assist in diagnosis

- Neuromuscular specialists and neurologists
- Metabolic specialists
- Geneticists
- Cardiologists
- Pediatricians and developmental pediatricians
- Pulmonologists and respiratory specialists
- Primary care physicians
- Emergency room specialists

If there are no specialists in your local area who are familiar with Pompe disease, ask your family doctor to refer you to a major medical center that is known for diagnosing rare diseases. If the nearest center is too far away, urge your doctor to consult with an expert there about how to handle your case. As many people with Pompe and other rare diseases have learned, it is important to work closely with your healthcare provider to make sure you get the best care possible.

## Tests that confirm the diagnosis

A number of tests may be done to help diagnose Pompe disease and determine the extent of muscle weakness or how far the disease has progressed. However, an enzyme assay is commonly used to confirm a diagnosis of Pompe disease. This biochemical test measures the activity of acid alpha-glucosidase enzyme in a small sample of skin, muscle, or blood. The enzyme assay may be performed using different samples, which include:

- Dried blood spot
- Cultured skin fibroblasts
- Lymphocyte or leukocyte (blood)
- Muscle

A diagnosis of Pompe disease is confirmed if the test shows there is less than normal or no enzyme activity. You may find regional differences in the availability of these tests and how healthcare professionals use them to diagnose Pompe disease.

## Other clinical tests that may be performed

Healthcare providers may also choose to order other tests. These tests may point toward a muscle disorder or detect which muscles and organs may be affected and to what extent. These additional tests may include:

- **Blood tests** to check the level of creatine kinase (CK), an enzyme that is released at higher levels when muscle injury occurs.
- **Electromyography** (ee-LEK-tro-my-AH-graph-ee) (EMG), a test that records electrical activity in affected muscles (but may show a normal pattern in children and adults with Pompe disease).
- A **chest x-ray**, often performed in infants to see if the heart is enlarged.
- An **echocardiogram** (eh-co-CAR-dee-oh-gram) or echo test, an ultrasound that can show if the heart muscle has become too thick or is not functioning properly.
- An **electrocardiogram** (ee-LEK-tro-CAR-dee-oh-gram) (EKG) that can detect abnormal patterns of heartbeats and electrical activity.

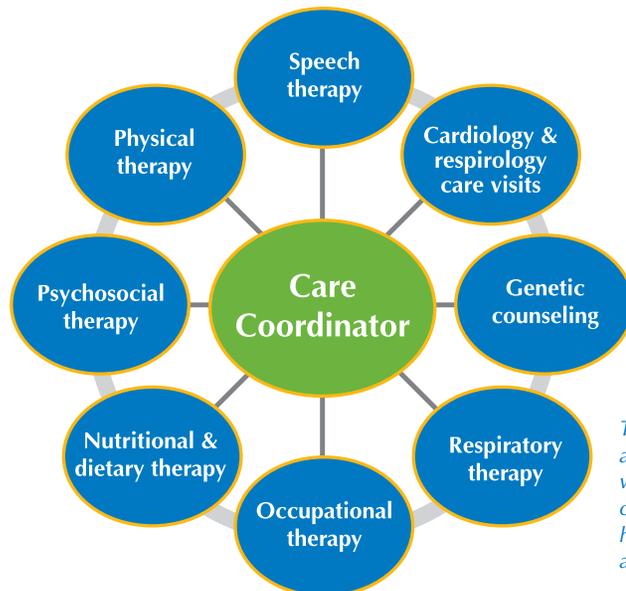
To find out more about these tests for diagnosing Pompe disease, visit Genzyme's Pompe Community website at [www.pompe.com](http://www.pompe.com).

## How is Pompe disease managed?

Until recently, the only therapeutic option for Pompe disease was supportive care. Enzyme replacement therapy (ERT) with Myozyme is now available and has the ability to treat the underlying cause of Pompe disease. Although ERT is not a cure, providing the missing enzyme may slow or halt the progression of muscle weakness and improve muscle function.

In addition to treatment, supportive care will continue to be an important part of Pompe disease management. The goal of supportive care is to help individuals with Pompe disease keep up their strength and prevent health problems caused by muscle weakness. Supportive therapies are tailored to the specific needs of patients. These therapies can ease the burden of living with Pompe disease by helping individuals feel better emotionally and physically.

### A team approach to supportive care



*This chart depicts some, but not all, of the health professionals who may be involved in your care. Always talk with your healthcare team before starting any type of supportive therapy.*

### Development of standards of care

Standards of care are guidelines for healthcare providers to follow when diagnosing and treating diseases. These guidelines can help ensure earlier diagnosis and aid in managing a disease like Pompe. They can also help patients receive medical care tailored to the disease and get better follow-up care. One professional group, the American College of Medical Genetics (ACMG), has developed guidelines for the diagnosis and management of Pompe disease.

Genzyme's Pompe Community website ([www.pompe.com](http://www.pompe.com)) provides information on new developments in Pompe disease management as it becomes available.

#### Play an active role to help get the care you need

You may find that many healthcare providers have never heard of Pompe disease. And those who offer supportive care may not know how to meet your specific needs. If that's the case, it may take additional steps to get the kind of care you need. Here are some steps you can take:

- **Reach out to Pompe patient organizations or a muscular dystrophy association** for information, resources, and support. They can suggest therapists in your area who have been trained to treat neuromuscular disorders.
- **Try to learn as much as you can about Pompe** so that you can ask your healthcare providers about therapies that might be helpful for you.
- **Contact Genzyme** (see page 30) for more information on a wide range of support services offered to patients with Pompe disease and their families.

This active role may not be familiar to you, but it is the kind of role that may be helpful when you are affected by a rare disease like Pompe. (For more suggestions on how to obtain care for Pompe disease, see page 21.)

## Types of supportive care

### Respiratory therapy

Respiratory therapy can provide the critical support needed to manage breathing problems. A respiratory therapist can teach exercises to strengthen breathing muscles. As the muscles used for breathing get weaker, mechanical ventilation may be needed. Mechanical ventilation uses machines that support breathing by sending air to the lungs. It may be provided in one of the following two ways:

**Noninvasive ventilation** may be used to manage early-stage breathing problems. Portable machines supply air through a mask that fits over the nose or both the nose and mouth. At first, a ventilator may be needed only at night while you sleep. As breathing problems become more severe, the ventilator may be needed during the day as well.

Two types of noninvasive ventilators are often recommended for people with Pompe disease. They are bilevel airway pressure (BiPAP) ventilators and continuous positive airway pressure (CPAP) ventilators. Both allow you to vary the amount of air delivered to the lungs while you inhale. This way, your muscles are still doing some of the work of breathing. CPAP ventilators provide positive pressure that is constant when a patient inhales and exhales. This can require more work for the muscles to exhale. For people with Pompe disease, the CPAP machine is generally used to treat sleep apnea (when breathing stops for short periods during sleep).

**Invasive ventilation** provides more intensive respiratory support to infants, children, and adults with Pompe disease. It is used when a serious lung infection or worsening breathing problems result in respiratory failure. In these emergency situations, there is often an urgent need to get air into the lungs as quickly as possible. This may be done manually by inserting a tube directly into the windpipe through the mouth or nose, called intubation (in-too-BAY-shun). It may also be done surgically by inserting the tube into the windpipe. This procedure is called a tracheostomy (tray-key-OSS-toe-me). The breathing tube is then connected to a ventilator that provides breathing support.

*The information in this booklet is not meant to take the place of professional medical advice. Always discuss your questions and concerns with your healthcare provider.*

### Dietary therapy

If you have eating problems that make it difficult to maintain a healthy weight, a registered dietitian can help you plan well-balanced meals to get the calories and nutrients you need.

To help prevent muscle wasting in older children and adults with Pompe disease, doctors may prescribe a high-protein, low-carbohydrate diet. This diet is high in meats, poultry, and fish and low in breads and starchy foods. This type of dietary therapy has not been shown to provide consistent results.

Some children and adults have had improvements in lung function and muscle strength after following a carefully supervised high-protein diet. For others, the benefits have been modest at best. Combining a high-protein diet with daily exercise (also carefully supervised) may produce better results.

Always consult your healthcare provider before beginning any diet and/or exercise program. Some studies have suggested that adding alanine, an amino acid (one of the building blocks of protein), to the diet may also be helpful.

Tube feeding may be required for infants who are not gaining weight or for children and adults who are severely underweight or have serious swallowing or breathing problems. Liquid food is carried directly to the stomach or intestines. This can be done through a nasal tube that goes through the nose, throat, and esophagus or by a gastrostomy (gas-TROSS-toe-me) tube, or G-tube, that is surgically attached to the stomach wall. Another option is a gastrojejunostomy (GAS-troh-jee-ju-NOSS-toe-me) tube, or G-J tube, that is attached to the small intestine.

### Patient Experience

*A 27-year-old woman diagnosed with Pompe disease always had trouble gaining weight. But after starting a special diet and exercise program supervised by a metabolic specialist, her health improved dramatically. The extra nutrition she gets from overnight tube feeding helps keep the weight on and gives her more energy to get through the day.*

### Physical therapy

Physical therapy can improve balance, posture, and muscle tone; support mobility; maintain flexibility and range of motion; and ease muscle pain and stiffness. The goal is to help people stay healthy, strong, and mobile.

The therapy may include the use of exercises, massage, machines, and adaptive devices such as foot splints or wheelchairs. Stretching exercises can improve a young child's posture and help prevent contractures (muscle tightness).

A physical therapist can help people learn new ways to sit, stand, or move around as muscle weakness progresses. The therapist can also teach individuals how to use different muscles to do the work of muscles they can no longer use.

### Occupational therapy

Occupational therapy helps individuals with muscle weakness learn new ways to complete daily tasks at home, in school, and at work. The therapy may consist of specific exercises to help maintain strength and dexterity (the skill and ease with which you use your hands). It can teach how to use adaptive devices that make it easier to do activities of daily living (such as bathing, dressing, and cooking), participate in school activities, or perform job duties.

Occupational therapists may also recommend special equipment or changes that can be made in the classroom or workplace to help people with progressive muscle weakness function well in these environments.

*The information in this booklet is not meant to take the place of professional medical advice. Always discuss your questions and concerns with your healthcare provider.*

### Patient Experience

*A 13-year-old with Pompe disease is showing signs of scoliosis. He exercises with a physical therapist to help strengthen his muscles and hopefully delay the need for a wheelchair. He also keeps up his strength by playing tennis and swimming.*

## How do I find medical care for Pompe disease?

Your general practitioner or your child's pediatrician may recommend that you see a Pompe disease medical expert. There are only a few medical centers around the world that specialize in treating and managing Pompe disease.

However, many clinics and rehabilitation centers treat similar neuromuscular disorders, such as Muscular Dystrophy. There are also a number of medical centers at major universities that specialize in caring for patients with rare genetic disorders.

### Tip

*For help locating comprehensive care centers for neuromuscular disorders or genetic diseases, see pages 29-30.*

*If you do not have access to a center, you might contact specialists at one of these centers and ask them to consult with your local healthcare providers by phone or e-mail. Or ask your local healthcare providers to contact the specialists on your behalf.*

### Comprehensive care centers can offer a range of services in one place

- Genetic screening and counseling
- Diagnostic procedures, such as muscle biopsy or enzyme level testing
- Lab tests
- Sleep studies
- Early intervention for developmental delays
- Visits with specialists, such as cardiologists, pulmonologists, and neurologists
- Supportive therapy to manage symptoms
- Regular checkups
- Flu vaccines
- Support groups
- Help with insurance claims

Many of these facilities also participate in clinical trials to evaluate treatment for Pompe disease.

### *The medical specialists who may be involved in your care have a wide range of skills*

In addition to the healthcare professionals who provide supportive care, your care team may include the specialists listed below.

#### Care coordinator

This healthcare provider takes the lead in planning and arranging for tests and disease management. This person may be a general practitioner, a pediatrician, or a specialist.

#### Experts who diagnose and treat medical problems caused by Pompe disease

- **Neuromuscular specialist or neurologist:** diagnoses and treats neuromuscular problems
- **Geneticist/metabolic specialist:** diagnoses and counsels family members about genetic diseases
- **Cardiologist:** monitors and treats heart problems
- **Pulmonologist or respiratory specialist:** monitors and treats breathing problems
- **Gastroenterologist:** treats digestion and feeding problems
- **Orthopedist:** treats joint and bone problems, such as scoliosis and contractures

#### Providers who address psychosocial and practical needs

- **Social worker or psychotherapist:** offers emotional support and assists with practical matters or financial concerns
- **Genetic counselor:** provides guidance on genetic issues, such as family planning, carrier testing, and prenatal screening

### Is there a treatment for Pompe disease?

Yes. Myozyme® (alglucosidase alfa) is the first and only approved enzyme replacement therapy (ERT) for Pompe disease. Myozyme is a recombinant, or genetically engineered, form of the human enzyme acid alpha-glucosidase (GAA). Prior to the approval of Myozyme, there were no approved treatments for Pompe disease, only palliative and supportive care. Currently, there are more than 900 people with Pompe disease receiving treatment with Myozyme worldwide.

#### Enzyme replacement therapy (ERT)

Enzyme replacement therapy (ERT) with Myozyme treats the underlying cause of Pompe disease by replacing the missing or deficient enzyme, acid alpha-glucosidase. Enzyme replacement therapy is a lifelong treatment that is given at regular intervals through an intravenous infusion (an injection given over time directly into a vein). It is important to remember that every individual reacts differently to the enzyme and results of therapy may vary depending on how advanced the disease is at the time of treatment initiation. Therefore, it may take months to see any effect of Myozyme treatment, and some patients may not experience any visible results. Early diagnosis and treatment are critical to optimizing patient outcomes and may alter the course of disease progression.

#### Gene therapy

With gene therapy, scientists hope to be able to insert a normal copy of the GAA gene into the body so that it would be able to start making acid alpha-glucosidase on its own. So far, preclinical research (using animals) has focused on finding the best vehicle to transfer the gene into the body and reach the cells that will make enough enzyme to start clearing away glycogen and restoring muscle function. If this effort succeeds, it would be a major advance in the treatment of Pompe disease. Gene therapy is still in its early stages and testing has not yet begun in humans.

### *Clinical trials*

Although treatment for Pompe disease has been studied in several clinical trials, there is still much to be learned. Clinical trials offer physicians the opportunity to learn as much as possible about Pompe disease and treatment. To join a clinical trial, the volunteer patients must meet certain enrollment criteria based on their age, gender, the stage/severity of their disease, and other factors. The clinical investigator or physician leading the study must explain both the risks and possible benefits of the trial before volunteers can agree to participate. Volunteers must then sign a document called an Informed Consent Form, in which they acknowledge that they have been advised of the risks and benefits and voluntarily agree to participate in the trial. Patients can withdraw from such a trial at any time. This process of informed consent and other rules are in place to help protect the health and safety of patients.

**For up-to-date information about Pompe disease research studies throughout the world that are actively recruiting volunteer patients, visit [www.clinicaltrials.gov](http://www.clinicaltrials.gov).**



*When you feel ready, explore ways to help yourself*

## What can I do to make it easier to live with Pompe disease?

Pompe disease is a neuromuscular disease that causes changes over time. Because Pompe affects everyone differently, it is difficult to predict how these changes may affect your day-to-day life. Although you will likely find your own ways to cope with issues that arise, there are a number of strategies and sources of support that have helped people with other neuromuscular diseases adapt to the kind of challenges you may encounter.

The advice presented on the next few pages comes from Pompe patient advocates, family members, and professionals who understand the needs of people living with chronic neuromuscular diseases. Though not every tip suggested here may apply to your situation, some may be helpful to you as time goes by. Also turn to pages 29-30 for additional resources.

### Suggested strategies for helping children who are living with neuromuscular diseases

- **Helping your child be as independent and active as possible.** Giving children the chance to do tasks, chores, and activities that are tailored to their age and abilities can help them feel better about themselves. At every age, it may help to encourage involvement in sports, art, and music programs that meet your child's need for social interaction and physical activity.
- **Learning how to advocate for your child's special needs.** For example, work with your child's school to set up an Individual Educational Program (IEP). This type of plan spells out the services the school must provide for your child, such as a personal aide, special equipment, or adaptive physical education activities.
- **Taking some time for yourself.** If you are caring for someone with Pompe disease, it is very important for you to take breaks from caregiving. Finding someone reliable and trustworthy to provide care for a short time may reduce stress and allow you to renew your energy. If your child needs round-the-clock care, it is also important for you to be realistic about how much of the caregiving you can handle by yourself.

## Suggested strategies for adults living with neuromuscular diseases

Trying to balance your emotional, social, physical, and financial needs can be stressful. These strategies can help you reduce stress and put your energy into living as well as you can.

### Emotional health

**Caring for yourself.** Focus on what you can do to make life better for yourself. Learn about supportive therapies for Pompe disease, and talk with your healthcare team about which ones are right for you. Try to get enough rest and exercise. Educate yourself about Pompe disease so that you can be an active member of your healthcare team.

**Adjusting your expectations.** Accept that your abilities may change from day to day. Set some priorities, and put your energy into doing the things that are most important to you. Seek professional counseling if you are having trouble coping with the impact of Pompe disease on your family relationships, daily life, or personal goals.

### Work and social activities

**Finding ways to keep working if it is important to you.** As your physical abilities change, your employer may make accommodations that will help you in doing your job. For helpful ideas about what kind of accommodations would best meet your needs, visit the U.S.-based website of the Job Accommodation Network ([www.jan.wvu.edu](http://www.jan.wvu.edu)) and click on “Individuals with Disabilities”.

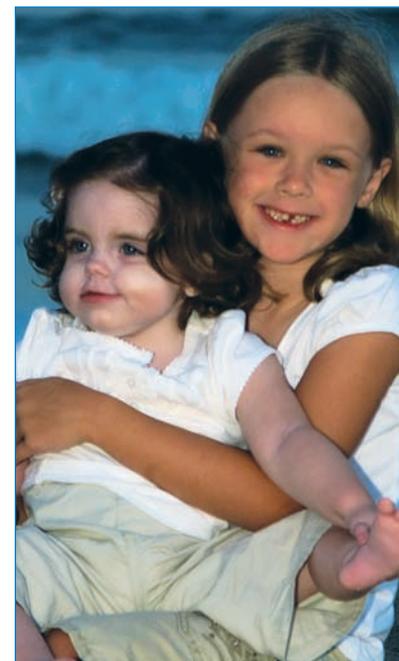
**Keeping up your social life.** As much as possible, try to spend some of your day enjoying hobbies or interests and being with people you like. If you cannot do a hobby as well as you used to, see if you might be able to do it differently. Or try new activities that use the skills and strengths you still have.

### Support from others

**Learning to ask for help.** Ask family, friends, and neighbors for help with specific tasks like taking you to your healthcare provider, going shopping, or keeping your child company for an hour or 2 each week. Let others know you need their companionship, as well.

**Building a network of support.** In addition to your circle of friends and family, reach out to religious groups, government and social service agencies, and healthcare providers in your local community who may be able to help you get home healthcare, equipment, financial assistance, transportation, or other services.

**Seeking out other patients and families of people with Pompe disease for encouragement and advice.** There are patient organizations around the world that can help you connect with people in your area (see resources on pages 29-30).



### Planning for financial needs

When you are facing the prospect of lifelong medical care, it is important to understand what resources may be available to you so that you can plan for your financial needs. It is also important to review what type of health insurance coverage you have. Financial assistance is available to help patients with Pompe disease and their families cover certain expenses, but it may take some persistence to find it. To learn more, contact the groups listed on pages 29-30.

## Choosing the right equipment for your needs

Adaptive equipment helps people with Pompe disease stay mobile and enables them to be more independent. To choose the right equipment for your needs, be sure to get advice from your physician, occupational and/or physical therapist, and other parents or individuals with Pompe disease. Think about how your needs may change over time and try out equipment before any purchase. The chart below lists examples of the types of equipment used by people with Pompe disease.

### Communicating and being independent

**Adaptive devices** *Mobile telephone, computer, intercom, universal remote, medical alert systems*

**How they help**

- Stay connected to the outside world
- Make you feel safer and more secure in your home
- Control electronic equipment in your home, such as lamps, TV, DVD player

### Getting around

**Adaptive devices** *Foot splint, folding cane, scooter, wheelchair, adapted car*

**How they help**

- Provide support if you are still able to walk
- Stay active and independent if you are no longer able to walk

### Sitting, standing, resting

**Adaptive devices** *Cushion, seating shell, stand-up chair, hospital bed, foam mattress*

**How they help**

- Increase comfort by taking pressure off weak muscles
- Make it easier to get up from a chair or bed

### Toileting and self-care

**Adaptive devices** *Grab bars, pedestal sink, handheld shower or adjustable-height shower heads, roll-in shower, bath bench, shower chair, lifter, raised toilet seat, adapted toilet*

**How they help**

- Make it easier to get to all areas of the bathroom
- Improve safety
- Afford privacy and independence

## Where can I find more information and support?

Though Pompe disease is rare, there are patient groups and organizations throughout the world that offer information, advice, and support to people living with Pompe disease.\*

### International Pompe Association (IPA)

A federation of Pompe disease patient groups worldwide, the IPA provides referrals to patient group contacts in different countries, organizes international conferences, and coordinates global activities related to Pompe disease. Through its website, the IPA reports the latest news on clinical trials, treatment studies, and standards of care for Pompe disease. Individual patient experiences are highlighted in the Patient Testimonials section.

**Telephone: +31-35-548-0461**

**[www.worldpompe.org](http://www.worldpompe.org)**

### World Alliance Neuromuscular Disorder Associations (WANDA)

WANDA assists patients and families with finding neuromuscular disorder associations in different countries and providing links to websites and other sources for more information on neuromuscular disorders. Patients and families can also learn how to form a new neuromuscular disorder association.

**[www.worldmuscleforum.org](http://www.worldmuscleforum.org)**

**[www.wandaweb.org](http://www.wandaweb.org)**

### Additional Patient Groups

#### Acid Maltase Deficiency Association (AMDA)

**[www.amda-pompe.org](http://www.amda-pompe.org)**

#### The Pompe's Group of the Association for Glycogen Storage Disease

**[www.agsd.org.uk](http://www.agsd.org.uk)**

#### United Pompe Foundation (UPF)

**[www.unitedpompe.com](http://www.unitedpompe.com)**

\* With the exception of Genzyme's own website, the websites referenced in this booklet are maintained by third parties over whom Genzyme Corporation has no control. As such, Genzyme Corporation makes no representation as to the accuracy or any other aspect of the information contained on such websites.

## Resources available from Genzyme

As a partner in the effort to manage the challenges of living with Pompe disease, Genzyme offers a wide range of support services to people with Pompe disease and their families.

### **Medical Information**

The Medical Information group can answer questions about Pompe disease and provide you with information.

### **Patient Advocacy**

The mission of Patient Advocacy is to demonstrate Genzyme's commitment to patients and cultivate relationships with the patient community for lasting impact. The Patient Advocacy team works to:

- Champion the patient perspective and integrate it within Genzyme's internal decision-making process
- Harmonize, coordinate and develop collaborations with patient groups to optimize patient care

### **Pompe Community**

Visit [www.pompe.com](http://www.pompe.com) to download copies of this booklet, learn more about Genzyme's services, and find links to other resources.

### **Pompe Registry**

The Pompe Registry, initiated in September 2004 and sponsored by Genzyme, is an ongoing, global observational program that tracks natural history and treatment outcomes of patients with Pompe disease. The Registry is dedicated to improving the understanding of Pompe disease and helping to optimize care of patients living with this rare disease. The Pompe Registry is open to all patients with Pompe disease, regardless of treatment status. Visit [www.pomperegistry.com](http://www.pomperegistry.com) for more information or ask your physician about participation in the Pompe Registry.

*For information on the services described above,  
use the following contact information:*

Call +1-617-768-9000

Monday–Friday 8:00 am to 6:00 pm, US Eastern Time

## Where do I go from here?

When you're dealing with a disease that spans the spectrum from mild muscle weakness to severe disability, there will always be uncertainty about what the future may hold. And everyone affected with Pompe disease must find a way to cope with daily struggles and lifelong challenges.

Learning about Pompe disease, taking time for yourself, tapping into support resources, and reaching out to others for advice, comfort, wisdom, and understanding can help make your journey easier as you move from diagnosis to action.



