“I steadily became severely underweight... it was obvious something was wrong.”

-Hillary
What you need to know about Pompe disease

We want to help you get the answers you need about Pompe disease. This brochure includes details about the disease, as well as additional online resources for you.

Pompe (pronounced pom-PAY) disease is rare: only about 1 in 40,000 people have the disease. While it occurs quickly in newborns, it can slowly develop in adults as well. Pompe disease makes it hard for some muscles in the body to work as they should.

Signs and symptoms in adults

When Pompe disease happens in adults, it can start as muscle weakness. It happens because the body is missing a key enzyme that helps certain muscle cells break down sugar (glycogen). Pompe patients may have:

- Weakness in their hips, legs, and shoulders
- Trouble climbing stairs
- Shortness of breath

Visit www.pompe.com to learn more about Pompe disease. And always remember to talk to your doctor.

What if I’ve already talked to my doctor?

If you have symptoms like these, please talk to your doctor. Even if you are already being treated for another illness, discuss your symptoms again. Pompe disease can sometimes seem like other more common illnesses, such as muscular dystrophy, general myopathy, and even sleep disorders.
As early as 6 years old, Brian remembers struggling to keep up on the playground and then suffering from weakness and back pain before being diagnosed with Pompe disease.

"I live a very full life, and Pompe disease is part of it. But Pompe disease doesn’t define my life.”
What causes Pompe disease?

Pompe disease is a genetic disease passed down from your parents. It occurs because the body has trouble breaking down glycogen.

When glycogen is not processed normally, it can build up in muscle cells. When this happens over time, muscles stop working correctly. As a result, it can become difficult to walk due to weakness in the hips and legs. It can also cause shortness of breath due to trouble with muscles that help us breathe.

Pompe disease is manageable

The good news is that doctors have answers for Pompe disease. The disease can be managed. The earlier patients are diagnosed, the sooner disease management can begin.

Visit www.pompe.com to learn more about Pompe disease. And always remember to talk to your doctor.

Where can I learn more?

You can learn more about Pompe disease at www.pompe.com. This online resource includes more details about the disease, as well as additional signs and symptoms.
“I had to advocate for myself... When I got the diagnosis, I felt relieved that it wasn’t all in my head.”

Allison
Age 26
In her early 20s, Allison started having difficulty breathing, especially at night when lying down to sleep, and would frequently wake up gasping for air.
You have options

There are reliable and accurate testing options for Pompe disease. The sooner patients can be diagnosed, the more help doctors are able to provide.

You are not alone

If you have more questions and concerns about Pompe disease, help is available. The patients featured in this brochure once had questions about their health. After they were diagnosed with Pompe disease, they were able to develop a disease management plan with their healthcare providers.

Neurologists and neuromuscular specialists are particularly focused on Pompe disease. They work with teams of professionals who help diagnose the disease. If you are experiencing symptoms, the important thing to remember is you are not alone. Discuss your symptoms with your doctor so that you may be referred to specialists who regularly work with Pompe disease.

Visit www.pompe.com to learn more about Pompe disease. And always remember to talk to your doctor.

What else can I do?

When you visit www.pompe.com, you’ll also see information about Genzyme Support Services. Through this program, Genzyme is committed to help those who are diagnosed with Pompe disease. The program includes medical information, advocacy, treatment support, and more. There are doctors who specialize in Pompe disease and we encourage you to discuss next steps with your doctor if you are experiencing any symptoms.
“Even though Pompe has taken away muscle strength from me, it can’t take away my internal strength.”

Dianna
Age 53

Diagnosed in her 20s, Dianna suffered for years from a limp and underdeveloped hips before a blood test confirmed she has Pompe disease.
The good news is Pompe disease is manageable.

If you would like more information about Pompe disease, please visit www.pompe.com.

This website also includes personal perspectives and video testimonials from actual patients with Pompe disease. It is dedicated to raising Pompe awareness and helping patients find medical information, additional resources, and even advocacy support.

And, as with any health questions you may have, we encourage you to speak with your doctor.

References:

A rare commitment to the Pompe community

For over 30 years, Genzyme has pioneered the development and delivery of patient-centric programs and services for individuals affected by rare and debilitating diseases. With a focus on rare diseases and multiple sclerosis, we are dedicated to making a positive impact on the lives of the patients and families we serve.

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