



LEG WEAKNESS

TROUBLE BREATHING

TROUBLE CLIMBING STAIRS

EXCESSIVE FATIGUE

DAYTIME SLEEPINESS

DIFFICULTY SWALLOWING

ABNORMAL WALK

DIFFICULTY REACHING OVERHEAD

MORNING HEADACHES

DIFFICULTY GETTING UP OUT OF A CHAIR

FALL EASILY

IT'S NOT IN YOUR HEAD

IT'S IN YOUR MUSCLES

If these symptoms seem familiar, you may want to learn about Pompe disease¹⁻³

It's real.

POMPE disease



IT'S TIME
TO

GET
ANSWERS

You may be experiencing some unexplained and concerning symptoms

Perhaps you've had recent difficulty climbing stairs, getting up out of a chair, or reaching above your head. Maybe you've felt related muscle weakness in your shoulders, legs, or hips, or have even felt short of breath. You may have noticed that your symptoms have also been getting worse over time.

It's important to trust yourself and discuss these symptoms with your doctor. If any of these symptoms seem familiar, they could be signs of a neuromuscular disorder.¹⁻³

One possibility is a rare condition called Pompe disease.¹⁻³ Although Pompe disease is not common, it is a good idea to discuss any health concerns you may have with your doctor.¹⁻⁴ This brochure was created to help you talk to your doctor about symptoms you may be experiencing.



How do you feel?

Let's start with a quick question: Have you experienced or had difficulty with any of the following activities, and have they been getting worse over time?^{1-3,5}

- > Trouble walking
- > Trouble climbing stairs
- > Trouble getting up out of a chair
- > Combing your hair
- > Reaching for items overhead
- > Holding arms up to wash your hair
- > Picking things up off the floor
- > Difficulty breathing while lying down
- > Difficulty breathing while talking
- > Morning headaches
- > Daytime sleepiness

These issues may seem unrelated, but when it comes to Pompe disease, they're not. These are some of the most common signs and symptoms described by actual patients.^{1-3,5} And in some cases, their challenges have steadily worsened over the course of months or years.¹

If any of this sounds familiar, do not hesitate to speak with your doctor. While your condition is not likely to be Pompe disease, the information in this brochure is intended to help you talk about your symptoms and discuss testing options with your doctor. Something to remember about Pompe disease is that early diagnosis is important.^{1,4,6}

KNOW YOUR SYMPTOMS

No symptom you're experiencing is unimportant. Although the above symptoms and changes in physical ability may seem unrelated, Pompe disease can present itself in many ways.^{1,2} Speak up and get the answers you need.



ABOUT POMPE DISEASE

Pompe disease is a condition where the body does not make enough of an important enzyme. The enzyme is called acid α -glucosidase or “GAA” for short.¹ It helps metabolise (process) a sugar in your muscles called glycogen. Muscles use glycogen for energy, but when it builds up and is not processed, muscles don’t work properly. As the glycogen continues to build up, muscles may also become damaged.²

As a result, Pompe disease may cause muscle weakness—especially in the hips and legs—and breathing difficulty.^{2,5,6} These symptoms are progressive, which means they get worse over time.¹

How Pompe disease may feel^{1,2,7}

- > Basic daily activities, such as getting out of a chair or climbing stairs, may become more difficult over time
- > Excessive fatigue, feeling really tired and not improving after getting more sleep
- > Morning headaches and daytime sleepiness may occur due to trouble breathing while sleeping (sleep apnoea)
- > Shortness of breath may occur due to weakness in the diaphragm (a muscle that helps the lungs)

Whether or not a person has Pompe disease comes down to the genes that they have received from their parents.¹ People do not develop this condition because they've done anything "wrong" to their bodies or have been exposed to anything "bad" in the environment.^{1,2}

Pompe disease may develop and progress in different ways from one patient to the next.² Some patients progress more rapidly than others. An important thing to remember is that the earlier a diagnosis is made, the earlier that doctors are able to help patients.^{1,4}

Pompe disease symptoms can be present at birth, or develop at any time during infancy, childhood or adulthood.¹



TRUST YOUR
INSTINCTS

If you relate to any of the information presented in this brochure, ask your doctor about testing options for Pompe disease. Always remember to trust yourself and to speak with your doctor about your health concerns.





IMPORTANCE OF RECOGNITION

Since Pompe disease is rare and sometimes can seem like other, more common muscle conditions, diagnosis can be challenging and take time.^{8,9} All of the following conditions listed below share symptoms similar to Pompe disease.^{1,2} Your doctor may have already talked to you about one, or even more of these.

If you have learned about any of these conditions while trying to better understand your symptoms, you may want to consider asking your doctor about Pompe disease.

Common-condition checklist^{1,2}

- > Muscular dystrophy (MD)
- > Limb-girdle muscular dystrophy (LGMD)
- > General myopathy/ unspecified myopathy
- > Polymyositis
- > Myasthenia gravis
- > Sleep apnoea (trouble breathing while sleeping)

LEARN MORE

What you understand and share with your doctor may make a big difference. No detail about your symptoms or overall health is unimportant or insignificant.

Testing and diagnosis

Different tests may be used to narrow down the possible causes of your symptoms.⁵ These may include muscle, movement, and breathing tests.^{1,10} A specific blood-based assay or DNA test is also available for Pompe disease.^{5,11} Your doctor will determine which test is best for you.

If you think you may have Pompe disease, speak up. Since the disease may get worse over time, it is important to have it diagnosed as soon as possible.^{1,6} The earlier Pompe disease is diagnosed, the more doctors are able to help with disease management.¹



REACH
OUT

Talk to your doctor about testing options for Pompe disease. Testing results may help answer questions about your symptoms.⁴ The results may also quickly determine the best next steps for your care.⁴

POMPE DISEASE

DISCUSSION CHECKLIST

It is important to discuss all of your symptoms with your doctor. Below is a checklist of common symptoms related to Pompe disease.^{1-3,5,9,12}

Check the symptoms you are having and bring this list to your doctor's appointment. It is important to get a definitive diagnosis as soon as possible.^{4,6}

Ask your doctor about testing options for Pompe disease.

Respiratory Issues^{1,9}

- Feeling breathless during and/or after exercise
- Trouble breathing while sleeping (sleep apnoea)

Additional Signs^{1,2,12}

- Morning headaches
- Daytime sleepiness
- Difficulty chewing/swallowing
- Unexplained weight loss
- Tongue weakness
- Acid reflux

Muscle Issues^{2,3,5}

- Difficulty climbing stairs
- Difficulty getting up out of a chair
- Walking with swaying hips or a waddle
- Frequent trips and falls (loss of balance)
- Trouble reaching over your head
- Difficulty washing/brushing your hair

SPEAK UP

Let your doctor know if your symptoms have become worse over time. Symptom progression is another sign of Pompe disease.¹



IT'S TIME
TO



GET
ANSWERS

See the checklist on the back of this panel for potential Pompe disease symptoms you may want to discuss with your doctor.

Additional Pompe Disease Resources

Please contact your physician if you would like to discuss your symptoms and concerns.

Additional patient information regarding Pompe disease and other muscle disorders awareness, education, and support services is available on itsinyourmuscles.com.au



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It's real.

POMPE
disease

A rare commitment to the Pompe community

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