You may find it helpful to contact your patient support group.

**Association of glycogen storage disorders (AGSD)**  
Tel: 0300 123 2790  
https://agsd.org.uk/

**The Pompe Support Network**  
www.pompe.uk

**Contact us**  
LDU Box 135, Addenbrooke’s Hospital, Hills Road, Cambridge. CB2 0QQ  
Tel: 01223 274634  
Email: lucambridge@nhs.net  
www.cuh.nhs.uk/addenbrookes-hospital/services/lysosomal-disorders

Please note the department is open Monday to Friday 08:30-17:00.

If you are unwell outside of these hours you should seek medical advice from your local healthcare team.

If your emergency doctor needs specific advice they should call the hospital switchboard on 01223 245151 and ask for the metabolic consultant on call.

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**Other formats:**

If you would like this information in another language or audio, please contact Interpreting services on telephone: 01223 256998, or email: interpreting@addenbrookes.nhs.uk

For Large Print information please contact the patient information team: patient.information@addenbrookes.nhs.uk

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We are now a smoke-free site: smoking will not be allowed anywhere on the hospital site. For advice and support in quitting, contact your GP or the free NHS stop smoking helpline on 0800 169 0 169.

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What is Pompe disease?
Pompe disease is a lysosomal storage disorder. Your body is made up of trillions of individual cells and each cell contains a small part called the lysosome. This is primarily responsible for breaking down and recycling worn out cell components. Inside the lysosome are enzymes which perform this very important housekeeping role. In Pompe disease your Pompe enzyme is not working effectively, allowing these products to build up and become stored in very specific areas of your body. There are many different enzymes in your lysosomes, and deficiencies of each enzyme will cause a different disease. All of these different diseases together are called lysosomal storage disorders.

How will this affect me?
In Pompe disease the cells most commonly affected can be found in the muscles. This means you may experience or be at risk of the following:

- Progressive muscle weakness
- Walking difficulties
- Breathing difficulties
- Fatigue

The progress of the disease in adults is extremely variable with some people not really noticing any symptoms whilst others are more severely affected.

If you have any questions about the effects and progression of the disease please do ask your doctor or specialist nurse.

Is there any treatment?
Pompe-specific treatment is currently available in the form of enzyme replacement therapy (ERT). All medication is supplied to you directly at home using a specialist medical homecare delivery company.

Enzyme replacement therapy
This is an intravenous infusion usually administered every one to two weeks. It provides you with extra enzyme that will supplement your own and help to reduce the amount of stored material in your cells. This is initially administered in hospital but after that you receive this treatment at home.

You can only receive this treatment for your Pompe disease if you attend one of the UK national referral centres at least once per year for regular assessment and review.

How did I get Pompe disease?
Pompe disease is a genetic condition which means that you inherited it from your parents.

There are instructions in your genes for making lysosomal enzymes and a small change in your DNA can be enough to stop the enzyme from working properly. You have two copies of every gene, having received one from each parent. To be affected by Pompe disease you need to have two copies of the gene with the DNA change on it – one from each parent. Only receiving one affected copy means you are a carrier and will not be affected.

Please do talk to your doctor or specialist nurse who can explain this in more detail.

What do I need to do next?
- Do ask your doctor or nurse for more information.
- Do attend your regular appointments with the lysosomal disorders team so that we can ensure you receive the most appropriate care and can keep you informed of any new developments.