POMPE
SUPPORT NETWORK

My Pompe Treatment

A guide to empower and inform people with Pompe disease on their treatment journey

Late Onset Pompe Disease

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Contents

How can I use this guide?

This guide is designed for people with Late Onset Pompe Disease, to help them understand the care they will receive at their specialist centre and the treatments available.

If you have recently been diagnosed with Late Onset Pompe Disease, this guide will support you in making informed choices about treatment, by helping you get the information you need.

It also provides tools to prepare for check-ups, including space to record symptoms between visits to your specialists to help you make the most of your appointments.

What is Pompe disease?

Pompe disease is a rare genetic condition that is currently diagnosed in less than 200 people in the United Kingdom.[1]

The symptoms of Pompe disease are caused by a build up of **glycogen** in the body which causes damage, especially to the heart and muscles. The most common symptom is progressive muscle weakness, but symptoms and their severity can vary from person to person.[2]

In the body, **glycogen** is broken down into glucose (a form of sugar your body uses for energy) with the help of an enzyme called acid alpha glucosidase (GAA). In Pompe disease, the body either does not make enough of this enzyme or it makes enzyme that does not work properly. Generally, the less working enzyme a person has, the more serious the symptoms will be.[2]

The most severe form of Pompe disease is known as Infantile Onset Pompe Disease ((IOPD). Symptoms start early in life and progress rapidly.

A separate guide for those with IOPD is available from **www.pompe.uk**

The other form of Pompe disease is known as Late Onset Pompe Disease (LOPD).

Late Onset Pompe Disease

People with LOPD have low or very low GAA enzyme levels, but some enzyme is working. Symptoms can develop in children (usually after the age of 1 year), teenagers, and adults. These symptoms include gradual muscle weakness and breathing difficulties, but LOPD doesn't usually affect the heart.[2]

The symptoms and their severity can vary widely between individuals. The first symptoms may be things like finding it difficult to climb stairs, falling over or stumbling. Over time, those with LOPD are likely to experience muscle weakness, pain and soreness. Breathing problems, morning headaches and fatigue are also common.[2, 3]

More information on LOPD can be found on the following websites:
www.pompe.uk
www.agsd.org.uk

Care and treatment for LOPD

As the symptoms and progression of LOPD can vary widely from person to person, your care and treatment will be tailored to your specific needs. Some people may only experience mild muscle weakness, while others may develop more severe challenges, including difficulty breathing and significant weakness in the muscles around the hips, shoulders, and spine. This variation means that each person's experience with LOPD is unique.[2]

A range of healthcare professionals may be involved in your care and further details of these are given on page 5.[2]
Their aims are to:

- Support the health of your muscles, bones and lungs.
- Support you to ensure that your diet helps your body to work its best.
- Improve your quality of life.

It is important to know that currently available treatments do not cure Pompe disease but can help slow down its progression and make symptoms milder and more manageable, although the results can vary from person to person.[2]

Guiding principles

Medical experts in Pompe disease develop guidelines for the care, treatment and monitoring of people with LOPD based on up to date research and evidence. The guidelines are updated when new evidence emerges.

European guidelines were published in 2024.[2] When UK guidelines are updated they are available at:
www.bimdg.org.uk/guidelines/lsd-guidelines

Enzyme replacement therapy

Treatment with enzyme replacement therapy (ERT) is available for patients with LOPD, but ERT may not be suitable for everyone.

ERT is offered to those who are most likely to benefit, while avoiding the additional burden of ERT in patients less likely to benefit from the treatment.

Am I eligible for treatment with ERT?

If you are having trouble with muscle weakness or breathing problems, you may be able to start ERT treatment.[4] In some cases, treatment may not be suitable, for example, if you have a severe or terminal illness unrelated to Pompe disease that greatly shortens your life expectancy. Your Pompe specialist will discuss your treatment options with you and the commitment to the treatment schedule and having regular check-ups necessary when receiving ERT.[4]

How does ERT work?

ERT provides the body with working enzyme, which helps to reduce the buildup of glycogen in the cells. Studies have shown that ERT can help with breathing, walking, muscle strength and quality of life, while reducing fatigue and the risk of becoming wheelchair dependent.[5-11]

Since the enzyme in ERT is gradually broken down by the body, it must be given at regular intervals and is a lifelong treatment.

It can take several months for the effects of ERT to be seen. You will be carefully monitored to see if your health is improving as expected, and if the treatment is working.

Where can I find out more about the different treatments available?

Your Pompe specialist will be able to advise you on the treatment options available.

You can also find information about treatments in our **Pompe Disease Treatments booklet**, available at **www.pompe.uk**

How is ERT given?

ERT is given through an infusion, which is a slow drip through a small tube placed into a vein, typically in the arm, allowing the medication to enter the bloodstream gradually. **Each infusion takes 3-6 hours.**[2]

Some people have a portacath (port) fitted.[3] This small device is placed under the skin through a minor surgical procedure and allows infusions to be given directly through the port, avoiding the need for repeated needle insertions into a vein. While ports can offer convenience and comfort, they also carry potential risks such as infection and blockage. Because of these risks, not everyone undergoing ERT will have a port fitted.

Find more information here:

Needle fear:

Ports:

If you are unable to scan the QR codes, links are provided on page 9.

Where do the infusions take place?

Treatment is usually started in a hospital or clinic, where a clinical nurse specialist will set up your infusion, monitor you during the infusion and manage any side effects.

Once everything looks stable, it may be possible to continue the infusions at home:[2, 3]

- You may learn to self-administer.
- A family member or friend can be trained to help you.
- A home care provider or nurse can come and help you at home.
- Having your infusions at school, college or the workplace may also be possible.

If you need more guidance on home infusions or home care providers, please contact your specialist centre.

What happens when I go on holiday?

As you need a constant supply of the missing enzyme to stop glycogen building up and causing damage, missing infusions should be avoided.

If you are going on holiday, you may be able to make arrangements with your specialist centre so that you do not miss your infusions. It's always best to consult your specialist centre well in advance to explore your options and plan accordingly.

What happens if ERT is not working for me?

Switching to a different ERT may be considered if muscle weakness and/or breathing has not stabilized or improved with ERT treatment. It may also be considered if you have severe infusion-related reactions that cannot be managed.[4]

When ERT may need to stop

As every person's experience of Pompe disease and response to treatment can be different, sometimes ERT may not help. There may also be other reasons why a decision to stop ERT may be considered. Any decision to stop

treatment would only be taken after extensive discussions with you and your family.[2] The situations where stopping ERT may be considered should be discussed with you when treatment starts. If ERT is stopped, you would continue to receive care and support from your healthcare team.

Stopping treatment might be considered if you have severe reactions during infusions, high levels of antibodies to the ERT, or no stabilisation or improvement in muscle strength or breathing. Having another life-threatening disease at an advanced stage would be another reason for considering stopping ERT treatment. Those receiving ERT may also decide they no longer wish to continue with treatment.[4]

Can I restart ERT?

If ERT was stopped because it did not appear to be working but the condition worsens faster without it, restarting therapy may be considered.[4]

Making informed choices about treatment

As there is more than one ERT available, the most appropriate treatment for you will be something you will discuss with your specialist. It's important to feel confident that you have as much information as you need. Conversations with your Pompe specialist can help you understand what's involved and what to expect from treatment.

Questions you may want to ask

Treatment decision-making

- [ ] What are the treatment options?
- [ ] How can I get a second opinion if I feel unsure about the treatment options?
- [ ] Why am I being offered this particular treatment?

Effectiveness & expectations of ERT

- [ ] How does ERT work for Pompe disease, and what can I expect in terms of slowing disease progression?
- [ ] Will treatment help with specific symptoms?
- [ ] Are there symptoms that treatment does not help with?
- [ ] How will I know if the treatment is working?
- [ ] Does the effectiveness of treatment reduce over time?
- [ ] What are the options if symptoms don't improve or they worsen?

Stopping ERT

- [ ] Under what circumstances might stopping ERT be considered?

Administration and logistics

- [ ] Where is ERT administered and by who?
- [ ] Can I learn to do it myself?
- [ ] How can I schedule infusions around work, childcare, school or other commitments?
- [ ] What happens if an infusion is missed or I go on holiday?
- [ ] How is the response to ERT monitored, and how often will follow-up appointments be needed at the hospital?

Interaction with other treatments and lifestyle

- [ ] Are there any medicines, supplements, or dietary items that might interfere with ERT?

Side effects

- [ ] What are the common side effects of ERT, and how can they be managed?
- [ ] Are there any long-term risks associated with ERT?

Who will be involved in your care?

There will be a team of healthcare specialists who work together to provide the best care for you.[2, 12] Each specialist has a specific role in managing your condition. They may include:

Pompe Specialists

Metabolic disease specialist
Your main point of contact and responsible for your overall care.

Specialist metabolic nurse
Co-ordinates assessments, treatment and supportive care.

Therapists

Speech and language therapist
Helps with any speech, swallowing and eating difficulties that may arise due to muscle weakness in the face and throat.

Respiratory therapist
Assists with breathing exercises and treatments including the use of equipment recommended by pulmonologists, such as ventilators.

Physiotherapist
Helps improve movement, strength and coordination through exercise and massage.

Occupational therapist
Helps with the skills needed for everyday tasks, such as washing and dressing. They can recommend adaptive tools or techniques to make life easier.

Other Specialists

Audiologist
Monitors and treats hearing difficulties.

Cardiologist
Will check for heart enlargement and any other heart issues.

ENT specialist
Monitors and treats issues with the ears, nose or throat.

Gastroenterologist
Assesses and manages any digestive issues.

Genetic counsellor
Can explain how the disease is inherited and the chance of existing or future family members being affected.

Neurologist/Neuromuscular specialist
Monitors how well the muscles, brain and nerves are working.

Dietitian
Will provide guidance on proper nutrition to support health and muscle function.

Respiratory physician
Monitors breathing and lung function and may recommend treatments or help with ventilation if necessary.

Psychologist
May provide counselling for individuals and families to cope with the emotional challenges of living with Pompe disease.

Orthopaedic surgeon
To monitor bones, muscles, and joints to address any problems.

Tests and assessments

You will have regular visits to your specialist centre where tests and assessments will be conducted to monitor your health and response to treatment.

Before starting treatment

Baseline assessments provide a starting point to measure how symptoms of your Pompe disease are improving or worsening over time and with treatment. Your assessments are part of your care plan which is tailored to meet your individual needs and age (assessments for children or adolescents with LOPD may be adjusted to suit their age and development).[2]

Follow-up and assessment of response to treatment

The aim of treatment is to stabilise your condition. Your doctor will decide which tests and assessments are suitable for you, to see if your treatment is working for you. As the follow-up measures are chosen for each individual's needs, you may have different tests to someone else who has Pompe.

Your follow-up assessments will be more frequent when you first start ERT; after that you will usually visit your specialist centre at least once a year.[2]

Tests and assessments you may have include:[2]

- Tests of muscle strength
- 6-minute walk test
- Lung function
- Polysomnography, also known as a sleep study, to monitor things like breathing and heart rate while you sleep
- Speech assessment
- Other assessments such as chest x-rays, ECG (to check heart rate and rhythm), and video fluoroscopy (a type of swallowing assessment) may be performed if needed.

Making the most of visits to your specialist centre

It is important to attend all check-ups to help your doctors keep track of your Pompe disease and response to treatment and to give you the opportunity to discuss any concerns you may have.

To make the most of your clinic appointments, it can be helpful to write down things you wish to talk about.

We have prepared note sheets that you can complete on paper or your electronic device, these can be found at: **www.pompe.uk**

Navigating the journey: Support for you and your family

Learning that you have LOPD can bring a mix of emotions, and it's common to feel lost in the vastness of information, treatment plans and day-to-day management. Remember, you are not alone on this journey, and there are many ways to find help and comfort, including both practical and emotional support.

Mental health

A Pompe disease diagnosis can be life-altering, bringing stress and emotional challenges for those affected and their families.

Support is available through patient organisations, disease-specific support groups, and professional counselling. Individuals and families can also connect with each other online and through Pompe meetings and events.

For information on where to get mental health support go to the Mental Wellbeing pages on the Pompe Support Network website:
www.pompe.uk

Mental wellbeing:

For help from a trained counsellor:

Genetic counselling

Understanding the genetic aspects of Pompe disease is important for you and your family. Genetic counselling explains how the disease is passed on, the chances of other family members being affected, and options for family planning and testing. It helps you make informed decisions about ongoing care and future pregnancies.

If you have not been referred to a genetic counsellor, please speak to your specialist healthcare team.

Physical activity

Regular exercise helps maintain muscle function and improve quality of life. A study showed that exercise improved endurance, strength, and stability in people with Pompe disease.[13]

It is important that any exercise programme is tailored to the individuals' abilities to avoid overexertion, which can damage muscles.[12, 13]

Please speak to your specialist healthcare team or specialist physiotherapist for the best advice on exercise programmes.

Find more information here:

If you are unable to scan the QR codes, links are provided on page 9.

Diet

While there is no special diet for Pompe disease, it is important to have a balanced diet filled with protein, vitamins, minerals, and antioxidants to support muscle health. A supervised high-protein diet may be recommended.

Adequate vitamin D and calcium intake are important for bone health, as people with Pompe disease are at risk of developing osteoporosis.[2, 12]

Please speak to your specialist healthcare team or dietitian who can provide a personalised diet plan.

Find more information here:

Lifestyle modifications

Proper rest, stress management, and regular check-ups are essential for managing the disease. Establishing a routine with regular rest may help with your fatigue. Home adaptations, like handrails or assistive devices, can also help improve your mobility and help maintain independence.

Find more information here:

If you are unable to scan the QR codes, links are provided on page 9.

Practical support

Patient organisations can provide advice about practical support including benefits and social care.

Find more information here:

Useful resources for you

Patient organisations in the UK

Pompe Support Network
t: 07590 270261 (Support); 01730 231554 (Office)
e: hello@pompe.uk
w: www.pompe.uk

Association for Glycogen Storage Disease (AGSD UK)
t: 0300 123 2790
e: info@agsd.org.uk
w: www.agsd.org.uk

International patient organisation

International Pompe Association
www.worldpompe.org

Video links

Florida University: Pompe Disease Exercises
www.youtube.com/playlist?list=PLr_fqSI-B7YAcWgw1HzJxyoK69swJRXP1

Social media

Pompe Support Network
www.facebook.com/PompeSupportNetwork

AGSD UK
www.facebook.com/AGSDUK

Cure Pompe Disease
www.facebook.com/groups/128829060576

Where to find information about clinical trials

Speak to your specialist healthcare team if you are interested in finding out about clinical trials. You can also look on patient organisation websites, which usually have research news including updates on clinical trials. There are a number of trial registries you can check too:

- www.clinicaltrials.gov
- www.clinicaltrialsregister.eu/ctr-search/search
- www.trialsearch.who.int

Medical references

1. Pompe Support Network. Pompe Disease. Available online: www.pompe.uk/ (Accessed: 29 May 2025)
2. Parenti G, Fecarotta S, Alagia M, Attaianese F, Verde A, Tarallo A, et al. The European reference network for metabolic diseases (MetabERN) clinical pathway recommendations for Pompe disease (acid maltase deficiency, glycogen storage disease type II). *Orphanet J Rare Dis*. 2024; 19(1):408.
3. Association for Glycogen Storage Disorders UK. Pompe Disease (GSD2) Diagnosis & Follow up - Late Onset. Available online: www.agsd.org.uk/all-about-gsd/gsd-variants/pompe-disease-gsd2/gsd2-section1b-tab1/ (Accessed: 1st May 2025).
4. Schoser B, van der Beek N, Broomfield A, Brusse E, Diaz-Manera J, Hahn A, et al. Start, switch and stop (triple-S) criteria for enzyme replacement therapy of late-onset Pompe disease: European Pompe Consortium recommendation update 2024. *Eur J Neurol*. 2024; 31(9):e16383.
5. van der Ploeg AT, Clemens PR, Corzo D, Escolar DM, Florence J, Groeneveld GJ, et al. A randomized study of alglucosidase alfa in late-onset Pompe's disease. *N Engl J Med*. 2010; 362(15):1396-406.
6. van der Ploeg AT, Barohn R, Carlson L, Charrow J, Clemens PR, Hopkin RJ, et al. Open-label extension study following the Late-Onset Treatment Study (LOTS) of alglucosidase alfa. *Mol Genet Metab*. 2012; 107(3):456-61.
7. Güngör D, de Vries JM, Brusse E, Kruijshaar ME, Hop WC, Murawska M, et al. Enzyme replacement therapy and fatigue in adults with Pompe disease. *Mol Genet Metab*. 2013; 109(2):174-8.
8. Güngör D, Kruijshaar ME, Plug I, D'Agostino RB, Hagemans ML, van Doorn PA, et al. Impact of enzyme replacement therapy on survival in adults with Pompe disease: results from a prospective international observational study. *Orphanet J Rare Dis*. 2013; 8:49.
9. Kuperus E, Kruijshaar ME, Wens SCA, de Vries JM, Favejee MM, van der Meijden JC, et al. Long-term benefit of enzyme replacement therapy in Pompe disease: A 5-year prospective study. *Neurology*. 2017; 89(23):2365-73.
10. van der Meijden JC, Kruijshaar ME, Rizopoulos D, van Doorn PA, van der Beek N, van der Ploeg AT. Enzyme replacement therapy reduces the risk for wheelchair dependency in adult Pompe patients. *Orphanet J Rare Dis*. 2018; 13(1):82.
11. Güngör D, Kruijshaar ME, Plug I, Rizopoulos D, Kanters TA, Wens SC, et al. Quality of life and participation in daily life of adults with Pompe disease receiving enzyme replacement therapy: 10 years of international follow-up. *J Inherit Metab Dis*. 2016; 39(2):253-60.
12. Kishnani PS, Steiner RD, Bali D, Berger K, Byrne BJ, Case LE, et al. Pompe disease diagnosis and management guideline. *Genet Med*. 2006; 8(5):267-88.
13. van den Berg LE, Favejee MM, Wens SC, Kruijshaar ME, Praet SF, Reuser AJ, et al. Safety and efficacy of exercise training in adults with Pompe disease: evalution of endurance, muscle strength and core stability before and after a 12 week training program. *Orphanet J Rare Dis*. 2015; 10:87.

QR code links

Needle fear:
www.worldpompe.org/resources
Ports:
https://www.royalfree.nhs.uk/patients-and-visitors/patient-information-leaflets/portacath-guide
Mental wellbeing:
www.pompe.uk/mental-wellbeing
For help from a trained counsellor:
www.pompe.uk/mental-wellbeing/counselling-support
Information of physical activity:
www.worldpompe.org/resources/patient-focused-publications
Exercise and physical therapy:
www.worldpompe.org/resources/patient-focused-publications
Information on diet:
www.worldpompe.org/resources/patient-focused-publications
Nutrition and dietary therapy:
www.worldpompe.org/resources/patient-focused-publications
Information on lifestyle modifications:
www.worldpompe.org/resources/patient-focused-publications
Adapting to living with Pompe disease:
www.pompe.uk/support-and-advice/equipment-support
Information on practical support:
www.pompe.uk/support-and-advice/benefits-advice-and-social-care

POMPE
SUPPORT NETWORK

Based in the United Kingdom, we are a network of individuals, families, scientists and healthcare professionals who aim to improve the lives of all people living and working with Pompe disease. The network is run by members of the Pompe community, for the benefit of the Pompe community.

Pompe Support Network
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www.pompe.uk/publications-library/guidelines

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