POMPE
SUPPORT NETWORK

My Child's Pompe Treatment

A guide for parents and carers

Infantile Onset Pompe Disease

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Contents

How can I use this guide?

This guide is designed for parents and carers of children with Infantile Onset Pompe Disease, to help them understand the care they will receive at their specialist centre and the treatments available.

If your child has recently been diagnosed with IOPD, 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.

The other form of Pompe disease is known as Late Onset Pompe Disease (LOPD). Symptoms may appear in childhood or adulthood and progress more slowly.

A separate guide for those with LOPD is available from:
www.pompe.uk

Infantile Onset Pompe Disease

This severe form of Pompe disease is a result of a complete, or near complete deficiency of the enzyme GAA, with symptoms appearing in the first few weeks or months of life.[2]

For babies with IOPD, early signs may include low muscle tone (floppiness), difficulty feeding and breathing problems. The muscles of the heart may be thickened (the medical term for this is hypertrophic cardiomyopathy). If left untreated, the condition can quickly become life-threatening.[2]

Some children may have a less severe form called Non-Classic Infantile Pompe Disease. Symptoms are usually first noticed after 6 months of age and may include low muscle tone, frequent respiratory infections, and an enlarged tongue and liver.[2]

This form of IOPD is rare, progresses more slowly, and the heart problems may be less severe.[2]

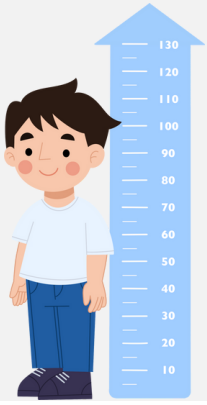
Care and treatment for IOPD

In recent years, advances in care and the introduction of enzyme replacement therapies have greatly improved survival rates and outcomes for children with IOPD.[2]

However, 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.[2]

Your child will be supported by a range of healthcare professionals and further details of this are given on page 5. Their aims are to:[2]

- Support the health of your child's heart, lungs and muscles.
- Support their development, e.g. growth and learning to walk.



Guiding principles

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

You can check the latest guidelines on this website:
www.bimdg.org.uk/guidelines/lsd-guidelines

Early care

At diagnosis, infants with severe forms of Pompe disease are often very sick and need specialised care. Many of these infants need to stay in the hospital for an extended time, usually at least three months, until their heart shows signs of improvement.

Enzyme replacement therapy

Following diagnosis of IOPD, your child will be assessed to see if enzyme replacement therapy (ERT) would be suitable for them. Your doctors will consider if your child is well enough to benefit from treatment and discuss this thoroughly with you.[2]

Reasons why treatment may not be suitable are included in the UK guidelines.

How does ERT work?

ERT provides the body with the working GAA enzyme, which helps to reduce the build up of glycogen in the cells.

Studies have shown that treatments can help reduce the thickening of the heart muscle, improve movement, delay the need for breathing support, and help patients live longer compared to patients who received no treatment.[3-5] The latest research has also shown that starting treatment as soon as possible after diagnosis is key for the best outcomes.[2]

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. Your child will be carefully monitored to see if their health is improving as expected, and if the treatment is working.

Is there anything that could reduce the effectiveness of ERT?

In some cases, children may respond to the treatment by producing antibodies, which can make ERT less effective.

Antibodies are part of the immune system's response to foreign substances and can sometimes reduce the effectiveness of treatments.

Doctors use CRIM (Cross-Reactive Immunologic Material) classification which can predict your child's response to treatment:[2]

- **CRIM-positive** children produce a little of their own enzyme, which means they are less likely to develop high levels of antibodies against ERT.
- **CRIM-negative** children do not produce any enzyme of their own, making them more likely to develop antibodies that reduce the treatments' effectiveness. In this case, additional treatment may be given to help manage this immune response and improve the effectiveness of ERT. This is known as **immunomodulation.**

Immunomodulation is additional medicine that helps to stop the immune system from attacking the treatment and reducing its effectiveness.

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.

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.

Some children will have a port (portacath) fitted. This is a device that remains under the skin and allows for the infusion to happen via the port, without the need to put a needle in the vein each time ERT is given).

Each infusion takes about 3-6 hours.[2]

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 specialist centre, where a doctor who knows about Pompe disease can monitor how well it's working and check for any side effects. Your child will receive their first ERT infusions in hospital. Once everything looks stable, it may be possible to continue the infusions at home. Families can learn how to give the infusion to their child or have a home care nurse visit their home to give the infusion. This can make treatment more comfortable and convenient, but your doctor will advise you on what's best for your child.[2]

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 your child needs 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 your child does not miss their 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 my child?

Increasing the dose or switching to a different ERT may be considered if muscle weakness and/or breathing has not stabilised or improved with ERT treatment.[2]

When ERT may need to stop

As every child'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 the family.[2]

The situations where stopping may be considered are part of the guidelines that your Pompe specialist works to. These should be discussed with you when treatment starts. You can also find the latest guidelines at: **www.bimdg.org.uk/guidelines/lsd-guidelines**

If ERT is stopped, your child would continue to receive care and support from your healthcare team.

Making informed choices about treatment

When starting treatment, 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?

What medical care will your child have?

When your child is being treated for Pompe disease, there will be a team of healthcare specialists who work together to provide the best care, based on your child's needs.[2,6] Each specialist has a specific role in managing your child's condition. They may include:

Pompe Specialists

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

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

Therapists

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

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

Physiotherapist
Helps improve your child's movement, strength and coordination through exercise, appropriate positioning and supportive aids.

Occupational therapist
Helps with the skills needed for everyday tasks, such as feeding 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 feeding or digestive issues.

Genetic counsellor
Can explain how the disease is inherited, the risk of future babies being affected, and what other family members may be at risk.

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

Nutritionist/Dietitian
Will provide guidance on proper nutrition to support growth, health and muscle function.

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

Psychologist
May provide counselling for your child and the family to cope with the emotional challenges of living with Pompe disease.

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

Tests and assessments

Before starting treatment

Initial tests will help doctors understand your child's health before starting treatment and provide a baseline so that the effects of ERT can be measured.

Your doctors will ask you for information about your child's past and current health conditions and medications, and use age-appropriate measures to understand how Pompe disease is affecting them. They will also do some laboratory tests – these are tests on samples that your child will provide, such as a urine or blood sample.[2]

During the first year of ERT treatment

Your child will be closely monitored to check their health and response to treatment. How often they are seen at your specialist centre will depend on your child's needs.

After 1 year on treatment

You will continue to visit your specialist centre at least once a year.

What are the tests and assessments?

The tests and assessments your child has will follow the recommendations in the UK guidelines and depend on their age and health, **(www.bimdg.org.uk/guidelines/lsd-guidelines)**. They may include:

Heart tests

Tests that check heart health including chest X-rays, heart rhythm tests (ECG), heart scans (ECHO), blood pressure.

Lung function

Polysomnography (sleep studies), test breathing patterns during sleep. Pulse oximetry, uses a small device that clips onto a finger or earlobe to measure oxygen levels in the blood. Older children will have standard lung function tests.

Swallowing assessment

Video fluoroscopy, a type of X-ray to identify any issues with swallowing.

Mobility and endurance

6-minute walk test and a suitable mobility assessment.

Brain health

A brain MRI and neuropsychometric assessment which looks at things like thinking, learning, memory and problem-solving skills. Performed at age 5, 10 and 15 years.

Bone health

Including vitamin D levels.

Hearing tests

Hearing tests.

Health and wellbeing

Suitable quality of life questionnaire.

Other tests if needed

Speech and Language Therapy (SALT) review, EEG that measures electrical activity in the brain, bladder ultrasound scan, DEXA bone density scan, spine x-ray.

Laboratory tests

Tests on urine or blood samples that assess Pompe disease activity, indicators of damage to the heart and muscles and overall health. The development of antibodies to ERT is also monitored.

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 child's 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.


Getting ready for appointments at your specialist centre

Infantile-Onset Pompe Disease


Enlarged tongue

Developmental delays

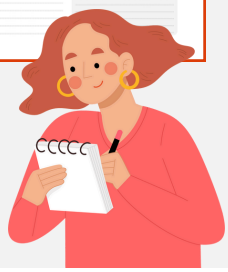
Behaviour changes

Sleep problems

Frequent colds or illnesses

Other symptoms or concerns

Other things I want 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 families and children

Learning that your child has IOPD often brings a mix of emotions, and it's common for parents to feel lost in the vast amount of information, treatment plans, and day-to-day management. Remember, you are not alone on this journey, and there are many ways to find support and comfort, both practically and emotionally.

Mental health

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

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

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

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

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 and appropriate exercise can have physical and mental health benefits for children with Pompe disease.

It is important that any exercise programme is specifically designed for the abilities and age of your child to avoid overexertion or muscle damage.

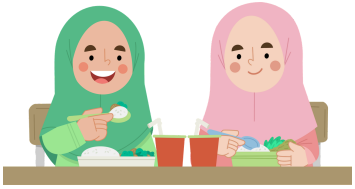
Please speak to your specialist healthcare team or specialist physiotherapist for the best advice on exercise for your child.

Find more information here:

Diet

While there is no special diet for Pompe disease, it is important for your child to have a balanced diet rich in protein, vitamins, minerals, and antioxidants to support muscle health.[6]

Please speak to your specialist healthcare team or nutritionist/dietitian who can provide a personalised diet plan to boost energy and muscle function.



Find more information here:

Lifestyle modifications

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

Find more information here:

Practical support

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

Find more information here:

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

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

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

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6. 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.

QR code links

Needle fear:
www.worldpompe.org/resources
Ports:
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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