



A Guide to Infantile Pompe Disease

Understanding Infantile Pompe Disease
and treatment management

Association for Glycogen Storage Disease UK.
In collaboration with specialist centres in
Birmingham, London and Manchester



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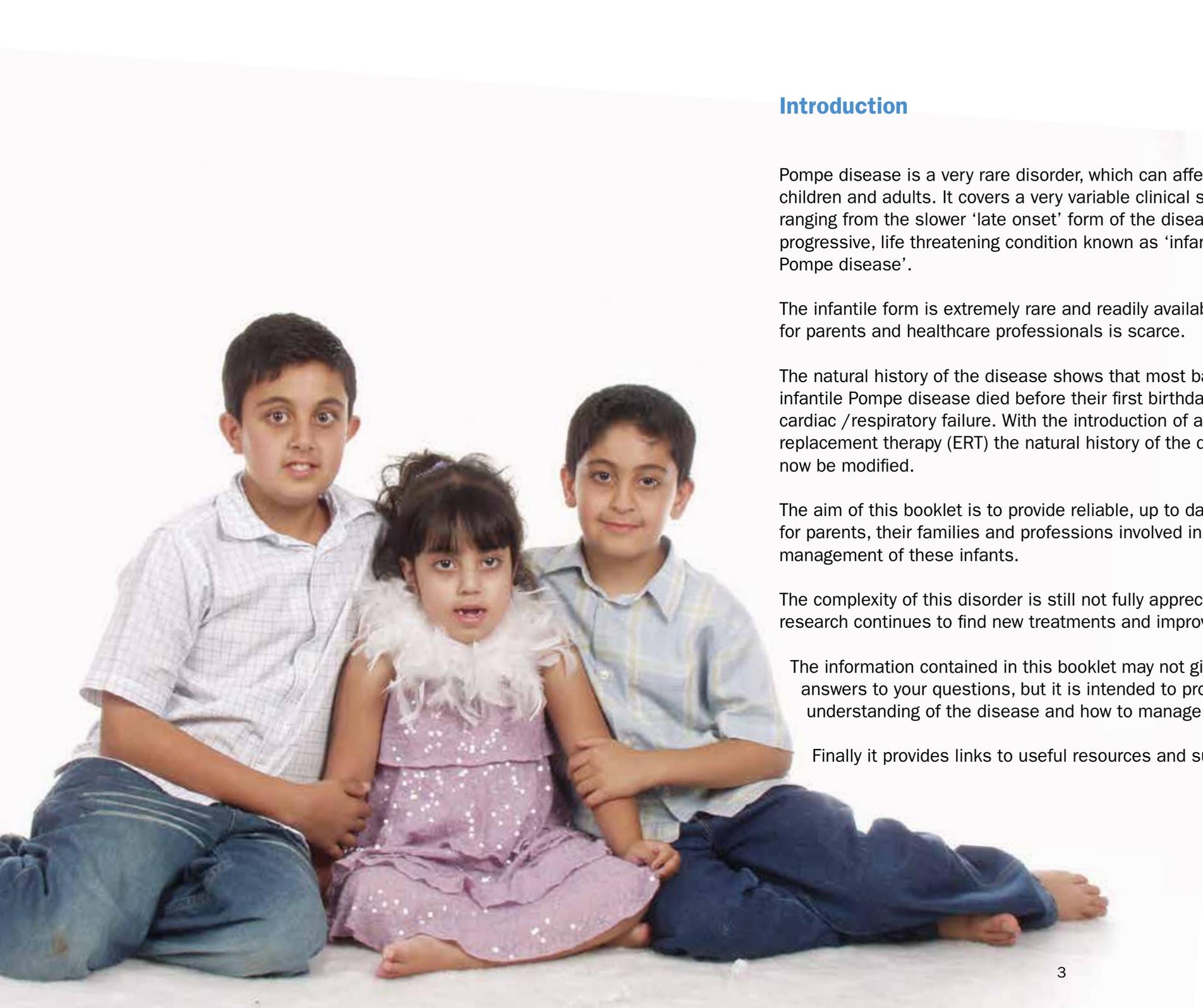
This booklet has been produced in collaboration with the specialist centres for the treatment of infantile Pompe disease in the UK.

We would like to express our gratitude to the co-authors for their contributions and their excellent co-operation.

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Introduction

Pompe disease is a very rare disorder, which can affect infants, children and adults. It covers a very variable clinical spectrum, ranging from the slower 'late onset' form of the disease to a rapidly progressive, life threatening condition known as 'infantile onset Pompe disease'.

The infantile form is extremely rare and readily available information for parents and healthcare professionals is scarce.

The natural history of the disease shows that most babies with infantile Pompe disease died before their first birthday from cardiac /respiratory failure. With the introduction of a new enzyme replacement therapy (ERT) the natural history of the disease can now be modified.

The aim of this booklet is to provide reliable, up to date information for parents, their families and professions involved in the care and management of these infants.

The complexity of this disorder is still not fully appreciated and research continues to find new treatments and improve outcomes.

The information contained in this booklet may not give all the answers to your questions, but it is intended to provide a better understanding of the disease and how to manage it.

Finally it provides links to useful resources and support groups.

Pompe Disease

Pompe disease is a rare, progressive muscle-wasting disease that can affect infants, children and adults.

This disorder is caused by an inherited metabolic deficiency or dysfunction of the lysosomal enzyme, acid alpha-glucosidase (GAA).

GAA is needed for the breakdown of glycogen (a form of sugar) within the lysosomes.

Lysosomes (little enzyme packages) are found in virtually all cells within the body and their purpose is to digest or recycle waste products of the cell's metabolism.

Without GAA the lysosomes can not break down any glycogen and this is primarily found in muscle cells and the liver, but the major clinical effects are on the structure of the muscle and its function.

The build up of glycogen causes the lysosomes to expand until they take up so much space that the muscle cell is damaged. Glycogen begins to leak out of the lysosomes causing damage to the surrounding muscle cells leading to muscle weakness.



Normal muscle fibre

Affected muscle fibre

Though the genetic defect that causes Pompe Disease is present at birth, symptoms may show up at any time from infancy through adulthood and the age of onset generally correlates to the severity of the disease.

Most patients with infantile onset disease have a minimal to undetectable enzyme level, leading to massive glycogen accumulation and rapid, aggressive progression of the disease.

In contrast late onset patients tend to have a limited, but detectable, residual GAA activity and in these cases organ damage is less pronounced and disease progression is slower.

Progressive muscle weakness is the most common symptom of all forms of Pompe disease.

Incidence of Pompe Disease

It is estimated that Pompe disease occurs in approximately 1 in 40,000 live births. Higher rates may be found in some ethnic groups. For example, in the African-American population, the incidence of infantile onset form is thought to be as high as 1 in 14,000 in comparison to Caucasian infants who are estimated to be 1 in 100,000 births.

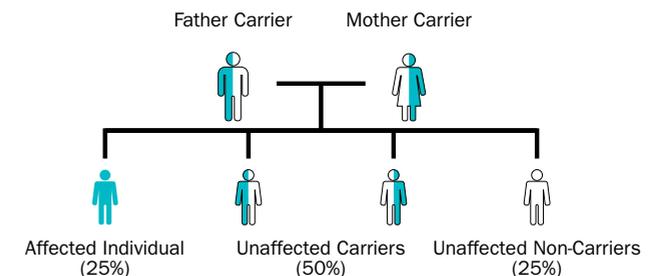
How is it inherited?

Pompe disease is inherited as an autosomal recessive trait.

This means that the disease only occurs when a person has 2 copies of a defective gene (one inherited from each parent).

Muscles of parents who carry one faulty gene function normally and present no symptoms of the disease.

When a baby is conceived, each parent passes on one copy of each of his or her gene to the baby.



If two carriers of the same faulty gene have a baby, there is a chance of passing on either the faulty gene or the working copy of the gene to the baby.

If the baby receives both copies of the affected gene, the baby will be affected by Pompe disease.

How is Pompe Disease diagnosed?

Due to the rarity and the relatively non-specific nature of early features, it can be a challenge for a diagnosis to be reached if there is no family history.

Often the disease is advanced and the patient very ill before Pompe disease is considered.

A clinical diagnosis is confirmed by the finding absence or virtual absence of the GAA enzyme in a blood sample from the patient.

Once a diagnosis is made, the blood will be analysed further to determine where in the gene the fault lies (DNA mutation analysis).

Genetic prenatal testing

Prenatal testing can be provided to pregnant women when there is a known risk to a baby.

Infantile Pompe Disease

Infantile Pompe disease is the most severe form of the disease. It is a result of a complete, or near complete, deficiency of the GAA enzyme.

Because there is no, or very little enzyme there is a fast build up of glycogen within muscle cells and this leads to an aggressive and rapidly progressive form of the disease.

Typically symptoms appear in the first few weeks or months of life, with feeding problems, poor weight gain, muscle weakness, floppiness (hypotonia) and respiratory problems. Affected infants often suffer from repeated respiratory infections and the heart becomes grossly enlarged from glycogen storage within the muscle wall.

Natural history studies report that most affected babies die before their first birthday from either cardiac or respiratory failure or a combination of both.

Early Symptoms

May include:

Delayed Motor Development

Motor milestones such as rolling over, crawling, sitting or standing are delayed, never achieved, or lost.

Babies demonstrate head lag when lifted from a supine position and have a frog-like posture of the legs typical of 'floppy baby syndrome' due to lack of muscle tone.



Feeding difficulties

Poor function of the muscles used in swallowing often leads to feeding difficulties. Cardiac failure can also cause breathlessness and interfere with normal feeding. Affected infants often present underweight with signs of failing to thrive.

Facial features

This can include:

- Open Mouth
- Macroglossia (enlarged tongue)
- Wide open eyes
- Poor facial muscle tone
- Nasal flaring because of respiratory difficulty



Disease Progression

Even in this form of the disease, severity can vary from one baby to another, however, it is expected that all will have some heart and lung involvement.

Cardiac / Heart

Marked Cardiomyopathy / Cardiomegaly (Enlarged Heart which leads to poor function).

Pulmonary / Lung

Progressive respiratory involvement due to muscle weakness.

Heart and Lung involvement may lead to:

- Increased respiratory rate (due to progressive cardiac failure or respiratory infection)
- Increased work of breathing
- Frequent chest infections
- Cool and clammy, sweaty, extremities due to progressive heart failure
- Weak cough and difficulty in clearing secretions

Musculoskeletal

Profound muscle weakness, decreased spontaneous movement.

Problems with bone density (osteoporosis or thinning of the bones secondary to immobility).

Gastrointestinal

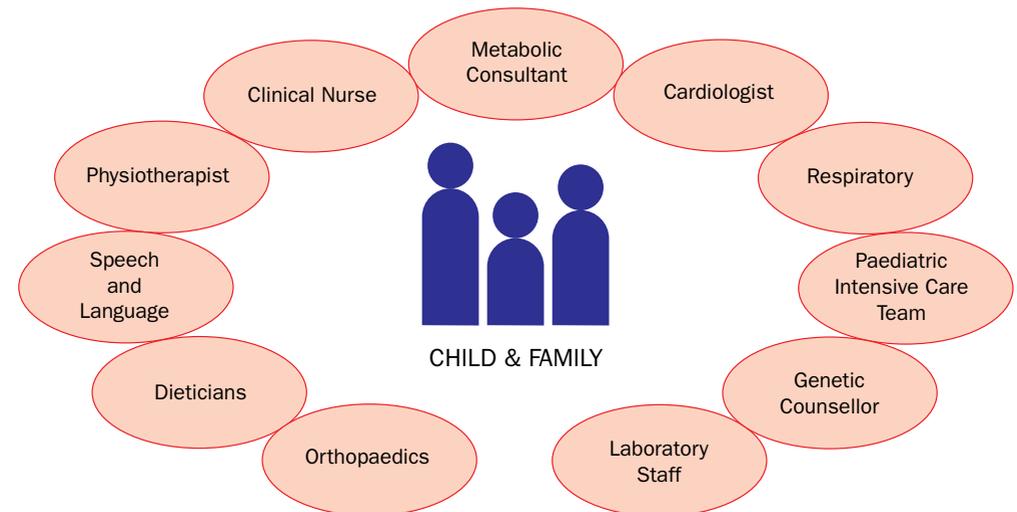
Difficulty in feeding, gastro-oesophageal reflux. Tube feeding is often required to avoid pulmonary aspiration (food entering the lungs).

Hepatomegaly (enlarged liver).

Because of the severity and complexity of Infantile Pompe Disease it is important that patients are managed at specialist centres, where extensive medical input from experts in various medical specialties is available.

Usually the Metabolic Disease team will provide the key lead in care, however it is likely that many other professionals will be involved.

Multi disciplinary care





Enzyme Replacement Therapy

Enzyme replacement therapy (ERT) is a treatment aimed at replacing the enzyme that is missing in children with Pompe disease. Myozyme, recombinant acid alpha-glucosidase, (Genzyme Corp, Cambridge, MA, USA) is currently the only licensed treatment for Pompe disease and has been commercially available since 2006.

The aim of treatment with ERT is to improve cardiac and skeletal muscle function. It is a life-long treatment given via an intravenous infusion. The infusion time is approximately four hours and the frequency of infusions is two-weekly, although this may change depending on the patient's condition.

Initially infusions are always given at a specialist centre, but it may be possible for infusions to be given at the local hospital, or even at home if considered safe for the patient and the home is suitable.

Reactions to the enzyme have been reported in some children and these may include skin rash, temperature (pyrexia), breathing difficulties, vomiting and pallor.

Some children may require simple pre-medication prior to the administration of ERT to help prevent reactions from occurring.

Blood and urine samples will usually be obtained at regular intervals to monitor the effectiveness of ERT and to also look for the development of antibodies to the drug.

How it is given

The infusions are given directly through a vein or via a central venous device. There are two central venous devices which are commonly used. These are peripherally inserted central catheters (PICC) and an implanted port. PICC lines are advantageous as they can be inserted using a local anaesthetic avoiding the need for general anaesthesia. It does require weekly dressing changes and has the potential to become dislodged. Implanted ports do not require a dressing as the needle is removed following the treatment and activities of daily living are not restricted when the device is not accessed. The disadvantage of a port is that this may require a general anaesthetic for insertion.

Results from ERT

Historically patients with infantile Pompe disease died before their first birthday, most commonly due to cardio-respiratory failure (Heart or Lung failure, or a combination of both).

Clinical trials, carried out between 2003 to 2006 demonstrated that ERT could reduce the mortality caused by this rapidly progressive, life limiting disorder. Since 2006 this has become a recognised treatment in many countries including the UK.

In several cases timely intervention with ERT has halted disease progression and improved cardiomyopathy, reversing the heart to near normal size and function. Some children that have started treatment early in the disease course are now surviving with very little sign of the disease.

However, some babies have not responded as could be expected, and the disease has continued to progress despite commencing on treatment. As treatment is still relatively new and the number of patients treated still small, data on long term outcome is limited.

A questionnaire-based study was performed between April and June 2009 and involved all treating centres in the UK with the aim of evaluating the outcome of all patients with infantile Pompe disease treated in the United Kingdom since the availability of recombinant enzyme.

This survey concluded that the long term outcome of enzyme-treated patients with infantile Pompe disease was highly variable and that no available baseline investigations would appear to be able to predict the outcome at the time of commencement of ERT.

CRIM status

A number of studies have suggested that the formation of antibodies to the recombinant enzyme drastically reduces the efficacy of enzyme replacement therapy.

Antibodies are a type of protein that are found in the blood and are part of the body's defence mechanism (immune system). They are usually produced to identify and neutralise or eliminate foreign proteins such as bacteria and viruses. It is estimated that 40 % of patients with infantile Pompe disease are CRIM (Cross Reacting Immune Material) negative. This means that these infants produce zero, or near to zero, enzyme protein within their cells.

Other babies with Pompe disease produce a very small amount of enzyme and these are classed as CRIM positive.

In CRIM negative babies when enzyme replacement is given, the cell fails to recognise it and produces large amounts of antibody in an attempt to destroy the foreign protein. This renders ERT ineffective in these patients.

In infants that produce a very small amount enzyme (CRIM positive), when ERT is given the cell recognises the enzyme and does not produce the same level of antibody response as in CRIM negative patients and therefore the treatment is likely to be much more effective.

It has been suggested that if you give medicines that reduce the body's ability to produce these antibodies it could improve the outcomes in these CRIM negative babies. This is known as 'Immune modulation'.

Once an antibody response is generated it is very difficult to reverse, therefore to be most effective, immune modulation should be commenced prior to ERT.

Previously CRIM testing took up to 12 weeks and babies suspected of being CRIM negative were immune suppressed routinely, before knowing the CRIM status.

CRIM testing is now available within days of the diagnosis, so only babies who are known to be CRIM negative receive immune modulation.

Risk from immuno-suppression

There is a potential risk when trying to stop antibodies that destroy the enzyme, that this also reduces some of the antibodies that we need to protect the body from infection with bacteria or viruses.

Minimising the risk

There are several different types of antibodies and research has been carried out to identify the most suitable medication that can be given to target the particular antibodies that are thought to destroy the enzyme product, but leaving the many other antibodies intact to help the body's immune system fight off infection.

These medications are given at a low dose when compared to many other forms of immune suppression therapies.

These drugs can induce a long-lived immune suppression so are only used for a short period of time, and then the immune system will return to normal.

Cardiac involvement

Children diagnosed with Pompe disease develop abnormalities in the heart muscle caused by a build up of glycogen due to their enzyme deficiency. The heart muscle becomes very thick because of this material and this decreases the heart's ability to pump blood around the body effectively.

Nearly all patients with infantile Pompe disease are affected to varying degrees and the heart can be examined regularly using an ultrasound scan to diagnose the problem and also to assess the progress of the thickening. This technique is called "echocardiography" and is a painless and quick test often done in association with a tracing of the heart's electrical activity – an electrocardiogram or ECG.

When enzyme replacement therapy is used to treat infantile Pompe disease, the heart muscle becomes thinner in most affected children, but this can take a considerable period of time and the infant can remain very unstable until the enzyme has had a positive effect on the heart muscle.

Children who are unwell related to poor heart function are often treated with medicine to try and improve this by removing excess fluid and slowing the heart rate to allow it to fill up better. This aspect of the child's health will be reviewed regularly by paediatric cardiologists, specialists in heart problems for children. It is likely that the infant will require a prolonged period of inpatient care until signs of cardiac stabilisation have been seen.



Visibly enlarged heart at diagnosis

Reduced 3 months following treatment

12 months following treatment , heart within normal limits.

Anaesthesia

Due to poor cardiac function, caution is required if these babies require anaesthesia for any reason. The affected heart muscle is prone to episodes of abnormal beating (arrhythmia) and this risk increases with some of the drugs used for anaesthesia.

The need for any surgery in infants with Pompe disease should be carefully weighed up against these significant risks and the anaesthetic should always be given by an anaesthetist experienced in this disease.

Respiratory Involvement and Breathing Support

The majority of babies with Infantile Pompe Disease experience some form of respiratory compromise and regular respiratory monitoring is essential for these patients.

If respiratory support is required because of either the underlying Pompe disease or an acute illness such as a chest infection, management of this complication may involve:

- Feeding modifications to reduce aspiration risks
- Supplemental Oxygen
- Airway secretion clearance through assisted cough and other techniques
- Various forms of mechanical ventilation used to assist the weakened breathing muscles

Non-invasive ventilation

A mask over the nose or mouth can deliver air or oxygen to the lungs.

Ventilatory support can be delivered by one of two different methods depending on the patient's needs.

Continuous positive airway pressure (CPAP).

Bi-level positive airway pressure (BiPAP).

Invasive ventilation

Involves a tube inserted into the nose or mouth or directly into the trachea via a tracheostomy (an opening on the neck connected directly to the main airway).

The tube is connected to a ventilator to provide intensive breathing support; this will require the baby to be admitted to the Paediatric Intensive Care Unit.



The need for ventilation

Often a baby with Pompe disease will have been developing problems for quite some time before a diagnosis is made and by the time treatment is started the heart and breathing muscles can be severely affected.

Both ventilatory (breathing) and cardiac (heart) support may be required to enable the baby to overcome an acute period of illness or to give the heart time to improve in response to ERT.

Ventilation may be a short-term treatment to overcome this acute illness, but in some patients the weakness does not improve enough to allow them to become independent of mechanical ventilation. If a child requires long term ventilation, this will have a significant impact in terms of quality of life for both the child, parents and other family members such as siblings.

The effective management of this situation is a great challenge and requires a lot of experience. There is often no right or wrong answer in terms of the level of intervention that should be applied to an individual patient and each case should be treated on its merits.

If a decision is taken to ventilate a child, the implications of this will be fully discussed with the parents in conjunction with a team of experts in this field.

When a baby is first ventilated it will be through a tube from either the mouth or the nose (endotracheal or nasotracheal tube).

This is a flexible plastic tube that is put into the mouth or nose and down into the trachea (airway). This is connected to a ventilator machine and permits air to pass freely to and from the lungs.

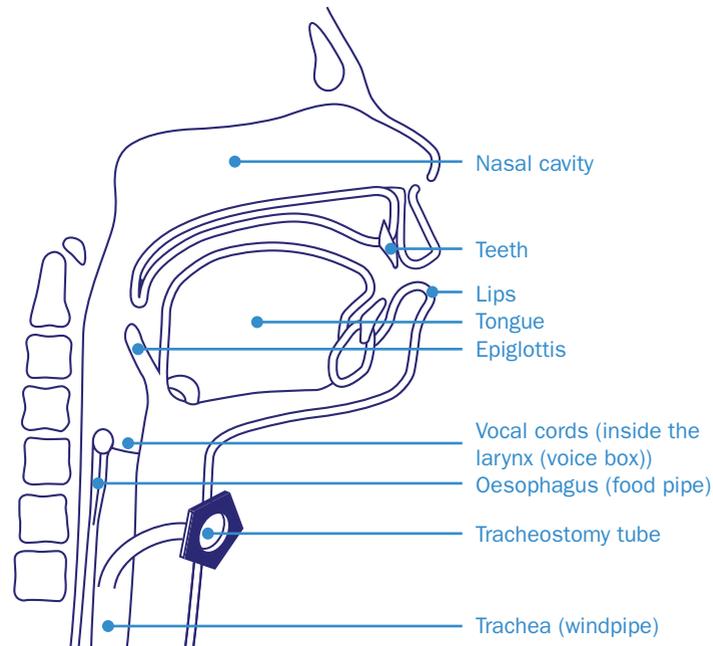
This is usually a temporary measure, as prolonged endo or nasotracheal intubation carries a high risk of damage to the soft tissue of the mouth, nose, pharynx and trachea.

If the infant cannot be weaned from respiratory support a decision will be made to continue with Long Term Ventilation via a Tracheostomy.

Tracheostomy

A tracheostomy is a small opening in the windpipe which is used to open up the airway. A tube is inserted into the hole to assist breathing.

A tracheostomy is considered to be a safer more stable way of delivering long term ventilation.



It can be used to promote the process of weaning from intubation. In some cases, even when a child requires long-term ventilation, it may not be required 24 hours a day. Some infants will require support at night time only or when they are asleep.

Physiotherapists Rôle / Physiotherapy Management

The physiotherapist's intervention is to optimise and preserve motor function within the limits of the disease.

All children should have a basic assessment that looks at the range of movement, muscle tone, posture and alignment, hip stability and motor development. There are a number of assessments that can be used including standardised measures and it is often helpful to video the assessments so that an easy comparison can be made over time.

Due to the variability of severity, not all children will experience all the following problems but the key issues commonly encountered are:

- Muscle weakness
- Delayed motor development
- Muscle contractures
- Spinal deformity
- Respiratory compromise
- Osteopenia (thinness or weakness of bone) and fractures

Muscle Weakness

In patients with later onset disease, or infants following treatment, the weakness in the muscles tends to be symmetrical. The proximal muscles (e.g. muscles around the shoulders and hips) are weaker than the distal muscles (e.g. the muscles around the wrists and ankles). The muscles in the trunk are also weak.

Some children tend to adopt certain postures due to these weaknesses, such as standing with an increased curve in the lower spine, walking with a waddling pattern and difficulty moving against gravity e.g. getting up from the floor to standing, or going up and down stairs.

Currently there are no national guidelines for strengthening programmes for children with Pompe disease.

It is felt that exercise should be aerobic and sub-maximal to avoid over fatigue and damage to the muscles.

The best activities are those that are functional and avoid excessive resistance. Active assisted exercises and facilitated movements to encourage head and trunk control, weight shift and sequencing between positions in sitting, crawling and standing should be encouraged.

Hydrotherapy can be beneficial but used only after expert advice on disease stability and the effects of prolonged exposure to heat are taken into account.

Regular rest periods should be incorporated into any exercise programme to allow recovery and to avoid over fatigue.

A physiotherapist will be able to give you specific advice regarding the types of exercise for individual children.

Specialist buggies and wheelchairs need to be considered for long term use if the child is severely affected. Some children may only need a buggy for long distances. There may be a need for home adaptations to be done to allow wheelchair access. An Occupational Therapist can give advice on the need for adaptations, specialist seating, bathing and toileting devices.

Delayed motor development



Children acquire skills by the development of postural control and co-ordination against gravity. It is important to give the child as much opportunity as possible to learn by exploring their environment. This can be done by facilitating and assisting movement to practice and promote developmental skills. Maximising the building blocks of movement such as head and trunk control, and transition between positions will help this.

It is important to use adapted positions to encourage normal movement patterns. For example, it takes more effort for an infant to move its hands into the midline to play with toys whilst lying on its back compared to the effort needed when on its side as the effects of gravity are neutralised allowing the child to reach independently.

In their early development children may show:

- Head lag on pull to sit
- Antigravity movements are slow to develop e.g. poor reaching for toys, decreased kicking
- Poor head and trunk control
- Delayed rolling, sitting balance,
- Decreased ability to lift head whilst on tummy
- Positive Gower's manoeuvre (difficulty getting up from the floor)
- Waddling gait

Muscle contractures

Often due to the muscle weakness the joints and muscles are unable to move through their full range independently. The unopposed force of gravity will pull the limb into a particular posture and if this is not corrected muscles will shorten and become tight. When it becomes fixed this is called a contracture.

It is important to use positioning, stretching and exercise programmes to counteract these forces. Wraps, pillows, seating systems, standing frames, splints etc can be used to provide adequate support for all joints in all positions. Daily therapy programmes should become part of the everyday routine to maximise range of movement but also to highlight any early signs of muscle tightness. Areas particularly at risk are muscles around the hips, knees, ankles and elbows.

Provision of specialist footwear / boots /insoles may be appropriate to enable the children to have a stable base of support.



Spinal deformity

Spinal deformity can develop due to the lack of muscular support for the bony structure of the spine and the effect of gravity on the body. An awareness of this as a possible complication is important as it may have an impact on respiratory functioning.

Spinal braces may need to be considered early to prevent the development of a spinal curvature (scoliosis), they would also support the child in a sitting position if trunk weakness was preventing independent sitting.

These need to be light and easy to use and an orthotist will be asked to assess the child in conjunction with the physiotherapist and doctors.

Respiratory compromise

Physiotherapists may advise parents how to manage their child's chest, possibly performing daily chest treatment to prevent complications arising, and to monitor for signs of deterioration such as increased work of breathing or thicker secretions which would require medical intervention and treatment by a specialist physiotherapist.

Osteoporosis and fractures

If bone is not stimulated to develop through movement osteoporosis (thinning of the bone) can occur. Bones become fragile and can break easily (fracture). Physiotherapists will try to prevent this condition by encouraging early weight bearing and general activity programmes.

Low vitamin D levels can also contribute to bone weakness. Regular blood tests will be carried out to monitor the vitamin D levels and supplements will be prescribed if required.

Provision of equipment

Other equipment may be needed to help your child with their movement skills including standing and walking frames, tricycles etc. The physiotherapist and occupational therapist will be able to provide you with advice regarding the need for any of these aids.

Speech & Language Therapy

A Speech and Language Therapist (SALT) may be involved with a child who has difficulties with feeding / swallowing or with communication.

Feeding and swallowing

Affected children can have feeding difficulties and are at significant risk of aspiration (the entry of food or drink into the lungs). This is not always obvious as children may not always cough or show other signs of distress during feeding. The effects of aspiration, particularly on the respiratory system, can be very serious e.g. repeated chest infections.

Initial assessment

On initial assessment the SALT will assess the safety of oral feeding with both fluids and solids where appropriate. The SALT will assess both the oral stage (sucking at bottle, control of food in mouth) and pharyngeal stage (swallow stage).

Most patients will go on to have a videofluoroscopy.

Videofluoroscopy

Also known as 'video swallow / modified barium swallow'. This is an x-ray that enables the SALT to define the anatomy and physiology of the swallow. It detects aspiration and enables range of therapeutic options to be tried.

The x-ray is recorded during the procedure. The SALT will guide you through the examination and with the Consultant Radiologist will make recommendations regarding the safety of oral feeding

Managing feeding difficulties

The primary aim of the SALT will be to ensure safe and effective feeding.

The SALT may consider any of the following:

- **Texture modification:** changes to the texture or consistency of food and drink can be beneficial. For example, this may involve making drinks thicker or choosing foods which require less chewing.
- **Positioning:** changes to the child's position for feeding can help support the swallow mechanism as well as making feeding easier for the person feeding the child. For example, special seating.
- **Volume and speed of presentation of food or fluids.**
- **Activities to promote developmental feeding skills:** For example, mouthing and messy play.

Due to the progressive nature of Pompe disease, the swallow is likely to change over time and therefore the way in which difficulties are managed will also change. Therefore regular assessments by SALT will be required.

When swallowing is unsafe



Tube-feeding is used as an alternative to oral feeding. A Nasogastric (NG) or Nasojejunal (NJ) tube is a tube which is passed down through the nose into the stomach or through the stomach into the small intestine (jejunum). Gastrostomy feeding requires a short operation in which a tube is passed directly into the stomach through a small hole in the abdomen and is fixed in place.

Some children will have a combination of tube and oral feeding. The SALT will provide recommendations on what the child can eat and drink safely.

Sensory feeding difficulties

As well as changes to the mechanics of swallowing, some children may also experience difficulties with heightened oral sensitivity. The SALT can provide advice and activity to help reduce this in order to develop the child's acceptance of certain textures or oral feeding in general.

Feeding and / swallowing in children with tracheostomies

If a child has a tracheostomy this may restrict the normal movements of the larynx (voice box) needed for safe swallowing, and ventilation may disrupt the child's ability to coordinate swallowing and breathing. Both of these can exacerbate any pre-existing difficulties.

The SALT will be able to consider assessing the child with a tracheostomy for placement of a one way valve.

Communication development

Speech

Children may have speech which is difficult to understand. This can take the form of one or more of the following;

- Dysarthria (slurred speech) as a result of muscle weakness.
- Articulation or phonological difficulties (speech sound errors).

Speech and language therapy can be effective in improving speech and sound production and in providing advice on compensatory strategies for use at home and school.

Hypernasality (nasal speech) appears to be particularly common in children with infantile Pompe disease. This may be caused by weakness of the palate, or form part of a more general speech sound error pattern. Referral to a specialist centre is advised in order to determine appropriate management options.

Communication for children with tracheostomies

If a child has a tracheostomy the type and size of tracheostomy tube can affect voice production. Following a tracheostomy the best option for communication will be decided by the family and SALT.

Options for communicating with a tracheostomy include:

Leak Speech: Some air may bypass the tracheostomy tube to pass up through the vocal cords (voice box). This voice might be quiet or only last for a few syllables at a time especially so if a child has weak breathing when off ventilation.

One way valve: A valve fitted onto the tracheostomy tube that allows the child to breathe in through the tracheostomy but closes on exhalation to redirect air up through the vocal cords. This is not suitable for all, and a speech therapy assessment is needed.

Alternative/Augmentative communication (AAC)

Where a child experiences periods with no voice, or has speech which is very difficult to understand, it is important to establish an alternative method of communication. This can range from a picture communication book, pen and paper, or signing to high-tech devices for children who need them long term. The right method will depend on each child's age and needs and a speech therapist input.

The effects of Pompe disease varies widely from person to person.

Not every child will face the same health problems at the same degree, therefore care and treatment plans must be individualised to each patient's needs.



Specialist centres in England

Birmingham

Birmingham Children's Hospital
Steelhouse Lane
B4 6NH
Tel 0121 333 9907/8
www.bch.nhs.uk/department.htm

London

Great Ormond Street Hospital
Great Ormond Street
London WC1N 3JH
Tel 020 7405 9200 ext 0366
www.gosh.nhs.uk

Manchester

Manchester University Children's Hospital
Department of Genetic Medicine,
St. Mary's Hospital, Oxford Road,
Manchester M13 9WL
Tel 0161 701 2137/8
www.cmft.nhs.uk/childrens-hospitals/home.aspx

Patient and Family Support

Infantile Pompe disease can be a devastating condition that has an emotional impact on the entire family. Apart from therapies and interventions provided by medical teams, families and patients often require informed advice, education and emotional support.

There are a number of patient support groups in the UK that you may want to contact, the most relevant to Pompe disease are listed here:

AGSD-UK

The Association for Glycogen Storage Disease (UK) provides support for children, adults and their families affected by any of the glycogen storage diseases, including Pompe disease. It provides information (print and internet), puts people in contact with each other for mutual support, issues regular newsletters and holds patient conferences and workshops. A full-time patient support nurse is available for families and patients living with Pompe disease.

Office Address:
Old Hambledon Racecourse Centre,
Sheardley Lane, Droxford, SO32 3QY
Telephone 0300 123 2790
(9.00 – 5.00, Monday to Thursday)
0300 123 2799 (Out of hours)
Email: info@agsd.org.uk
Web: www.pompe.org.uk
and
www.agsd.org.uk

IPA

The International Pompe Association is a federation of Pompe disease patient support groups world-wide. It seeks to coordinate activities and share experience and knowledge between the national groups. The IPA is an incorporated society (registered in the Netherlands), led by a Board consisting of international volunteers. The IPA can be contacted through the AGSD-UK who are founder members and have a position on the board. The IPA website holds a wealth of useful information including information leaflets, Pompe Connections, and a large collection of patient stories.

Web: www.worldpompe.org

MDC

The Muscular Dystrophy Campaign is a UK charity supporting all muscular dystrophies and related muscle diseases. The MDC provides practical care and support. It campaigns for increased government spending on medical research and for improved statutory services for neuromuscular conditions.

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