Supporting shared decision-making in Pompe disease:

Making complex treatment evidence accessible for patients

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Background

Pompe disease (PD) is a rare lysosomal disorder for which three enzyme replacement therapies (ERT) are currently available. Clinicians and patient advocates in the UK identified a need for accessible information on PD and its management, including the ERT treatment options.

The care and treatment of patients with PD in the UK is managed at specialised paediatric and adult centres. Clinical guidelines[1] are in place to manage the impact on health services and ensure treatment is targeted to those most likely to benefit from it. These guidelines include both start and stop criteria for ERT and routine monitoring and follow-up of patients.

Project Aims

- Provide accessible information for patients and their families on the care and treatment they will receive from their specialist centre.
- Help patients and families to understand the clinical guideline framework within which ERT is offered and how it relates to their situation.
- Support informed discussions with their healthcare team and decisions about treatment.

Methods

Patient-facing materials were developed using a collaborative approach of co-creation involving patient advocacy professionals, specialist clinicians and nurses and the patient community.

To develop shared decision-making materials, input was gathered from treatment recipients and providers via separate question sets: one for meetings with providers and another for meetings with advocates and patients. These meetings were audio recorded and summary notes created to guide the development and content of resources.

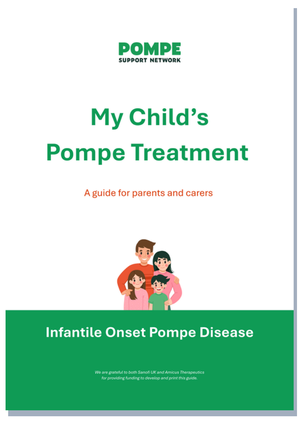
Examples of questions posed to patients:

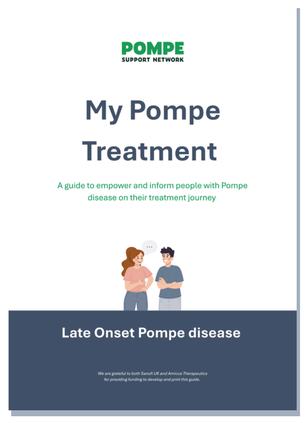
- What type of information do you wish you were told by your healthcare professionals when you first started ERT?
- What would you like other patients to know about starting ERT?

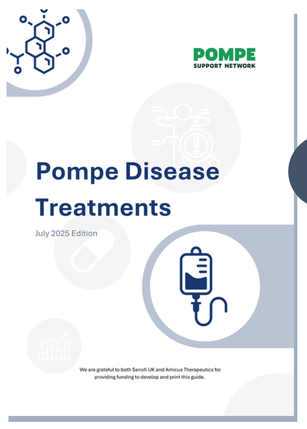
Examples of questions posed to clinicians:

- Are there specific concerns or misconceptions you encounter when discussing ERT with patients?
- What lifestyle changes do you recommend to help patients effectively manage their symptoms?
- Which symptoms do you find most useful for your patients to monitor and discuss in order to assess their response to ERT and overall wellbeing?

Information was also drawn from the academic literature and European[2] and UK management guidelines.[1] Careful attention was paid to the language used to make the materials accessible for a lay audience. Technical terms were explained, and signposts to additional resources and sources of help from patient organisations were included.







Find the full booklets at:
www.pompe.uk

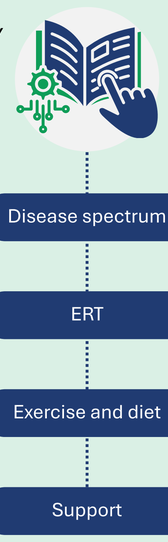
Poster presented at WORLD Symposium February 2026, San Diego

Results

Patient and caregiver insight

Patients and parents wanted a resource that would '*make them part of the multidisciplinary team*,' giving them the confidence to discuss their or their child's care and treatment with their clinicians. Information that would support this included:

- The spectrum of Pompe disease
- Expected outcomes of ERT
- How ERT works
- The practicalities of ERT infusions
- Exercise and diet
- Psychological support



The symptoms that impacted them most included muscle weakness, fatigue, respiratory issues, pain and mental health. They felt it was important that conversations with their clinicians included impacts on their daily activities and for caregivers to raise other challenges e.g. access to services.

Specialist insight

Specialist clinicians and nurses felt it was important to manage expectations of ERT and make it clear that it is a life-long treatment. Specific information on the following was requested:

- All treatment options - in a form that can be handed or e-mailed to patients as clinicians are not able to share the Summary of Product Characteristics before a patient goes on treatment
- Early treatment of IOPD
- Possible allergic reactions to ERT and what to do if they occur
- How missed infusions or going on holiday can be managed
- Switching between treatments
- Comparison of ERTs in terms of administration, clinical trials
- New treatment research and clinical trials
- Exercise, diet, mobility aids, home adaptations and social support
- Monitoring

They found it useful for patients to monitor and discuss their ability to undertake activities of daily living, exercise endurance, breathing, hours on ventilation, fatigue, mobility, sleep, pain, infections and mental health.

The resources

Five resources were developed, including disease reference booklets for Infantile Onset Pompe Disease (IOPD) and Late Onset Pompe Disease (LOPD), and appointment sheets for IOPD and LOPD designed to help patients and their families prepare for appointments with their specialists. A separate treatment booklet insert was developed to enable regular updating as more treatments or treatment studies become available. This booklet provides basic information about the current disease-modifying treatments and studies that have compared these treatments to each other.

Design features

Layout and text on each page is designed to be patient friendly, with key information highlighted, medical terms explained, and signposting to further information.

For those starting treatment

The eligibility criteria are clearly explained and a list of questions that patients may wish to ask about ERT are included.

Content

The individual booklets for IOPD and LOPD provide an overview of the disease and explain the care provided by the National Health Service in line with national guidelines. Descriptions of the specialists they may encounter in their multidisciplinary team, how their disease is monitored, and how ERT works, are included. The criteria for starting treatment and conditions where stopping treatment may be discussed are clearly explained. At the end of each booklet, readers are directed to sources of further support and information.

In addition, to support patients beyond the initial treatment discussions, appointment aids were developed for recording symptoms and concerns, to discuss when visiting their specialists. These printable sheets provide prompts for recording the things a patient wishes to discuss with their specialist, sections for recording changes in symptoms, and space for notes. The specific symptoms included are based on those that patients told us had the greatest impact on their daily lives.

The treatment booklet gives information about the three ERTs currently available, including the licensed indication, dosing and administration. A guide to understanding the results of clinical trials preceeds summaries of the comparative trials.

Conclusions

In rare disease with complex treatment evidence and eligibility guidelines, accessible patient information is vital to support patients and clinicians in shared decision-making, disease management and conversations about treatment access and continuation. We created a package of resources that can act as a disease, care and treatment reference guide. It can be easily updated as new treatments and evidence arise and support patients and their families in interactions with their healthcare providers and to participate in shared decision-making. It is essential to develop such resources in collaboration with patients, caregivers and their clinicians to ensure their utility.

Acknowledgements

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References

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