

Alpha-mannosidosis virtual community meeting

Jointly hosted by:



MPS Society
transforming lives through
support, research & awareness



Summary report

Date: 13 January 2026

Facilitator: Sophie Thomas

Attendees: Parents, carers, individuals living with alpha-mannosidosis, clinicians, researchers and representatives from national MPS societies (UK, Germany, Spain).

Purpose of the meeting

This first international alpha-mannosidosis community meeting was created to bring people together and strengthen connections across countries. Our aims were to:

- Create a shared space for families, individuals living with alpha-mannosidosis, and clinicians to meet and learn from one another.
- Share helpful updates on medical care, research progress, and new resources being developed to support the community.
- Talk openly about the challenges families face and identify what support is most needed going forward.
- Explore the best ways to stay connected, including regular online gatherings and the possibility of future in person meetings.

Our goal was to make the information easy to understand, reduce technical language, and ensure everyone, regardless of background, could take part fully.



We gratefully acknowledge the financial support provided by Chiesi Global Rare Diseases through an independent educational grant. The sponsor had no role in the planning, content development, or execution of this meeting. A high-level summary of the key outcomes will be provided to them post-event for transparency.

Presentation: understanding clinical and support needs in alpha-mannosidosis – Dr Christina Lampe

Key points

Alpha-mannosidosis is a very rare condition that affects many parts of the body. Because symptoms can be so different from person to person, it can be difficult for doctors to diagnose, monitor, and predict how the condition may change over time.

Unmet clinical needs

Families and clinicians face several challenges, including:

- **Limited treatment options.** Current treatments can help some people, but none are a cure, and some options come with significant risks.
- **No newborn screening in most countries,** meaning many children are diagnosed later than ideal.
- **Not enough long term information** about how the condition changes as people grow older.
- **Symptoms vary widely between individuals,** making it harder to create standard care plans.
- **Different countries use different monitoring approaches,** leading to inconsistent follow up and care.

Unmet support needs

Families also experience important practical and emotional challenges:

- Not enough access to mental health and psychological support.
- High demands on caregivers, including physical, emotional, and financial pressures.
- Feeling isolated, with limited opportunities for support or respite.
- Barriers in education, employment, and independent living, especially as needs change over time.

What families prioritise

Families shared what matters most to them:

- Keeping the condition stable, even if improvements are not possible.
- Better quality of life, including improved mobility and independence.
- Clear communication and being involved in decisions about care.
- A holistic approach, meaning doctors look at the whole person, not just individual symptoms.

What research shows

- Many people receiving enzyme replacement therapy (ERT) experience stable or slightly improved mobility, reduced pain, or better ability to manage daily activities.
- Outcomes after a stem cell transplant (HSCT) can be very different from person to person, success often depends on age, disease severity, and how well the transplant works.

Discussion points and questions

- Parents/carers expressed that monitoring is not consistent between different clinicians or countries, which can be confusing and stressful.
- One parent asked about how well HSCT works, especially in the UK. Dr Lampe explained that results vary widely and that collecting good long term data through registries is essential.
- Another question focused on why pain results differ between people who had HSCT and those on ERT. Dr Lampe clarified that these differences come from caregiver reported experiences, not from a known biological cause.

Presentation overview of research – Dr Mercedes Gil Campos

Dr Gil-Campos shared an overview of what recent research is showing about alpha-mannosidosis and current treatments. Rather than focusing on technical trial data, she explained the practical takeaways for individuals and families.

Key points

- Many people taking enzyme replacement therapy (ERT) have seen their energy levels and physical stamina stay steady or improve, such as being able to walk a bit further or manage stairs more easily.
- Some studies show that the immune system may work better in certain treated individuals, meaning fewer infections or improved blood markers.
- In adults, treatment has sometimes helped with day to day activities, making it easier to manage tasks like moving around or looking after themselves.
- Pain and discomfort often improved for many people on treatment, although experiences differ widely from person to person.
- Researchers have now published the first study that compares three groups together: people who received no treatment, those on ERT, and those who had a stem cell transplant (HSCT). This gives a clearer picture of how different approaches may help over time.
- A major challenge is the small number of patients worldwide, which makes studies harder to interpret and means results can vary from one person to another.

“Participants highlighted the importance of international patient registries...which help bring information together from around the world”



Download the plain English summary

Discussion points and questions

- Families asked for a clearer explanation of who took part in the research studies and how people were chosen.
- There were questions about how many people in Europe are currently known to have alpha-mannosidosis.
- Participants highlighted the importance of international patient registries, such as SPARKLE, which help bring information together from around the world. These registries make it easier for researchers and clinicians to understand the condition better and improve future care.

Presentation development of patient resources – Eva Raebel, Rare Disease Research Partners (RDRP)

Resources being developed

Eva Raebel from RDRP shared an update on several new resources designed to support people living with alpha-mannosidosis and their families. These materials aim to be easy to understand, practical, and helpful in day to day life.

Understanding alpha-mannosidosis guide

A simple, clear booklet explaining what the condition is, common symptoms, how it is managed, and what tests people may need. It also includes an easy to follow explanation of genetics.

Healthcare professional pack

A short guide for doctors who may not be familiar with alpha-mannosidosis. It includes symptom checklists, useful questions to ask during appointments, and guidance on when to refer someone to a specialist.

Appointment preparation toolkit

Templates families can use before medical visits to track symptoms, write down questions, and note any care needs or concerns. This helps make appointments more focused and productive.

Education and school support pack

Information for both parents and teachers.

- **For parents:** how to prepare for school meetings, understanding educational rights, and planning support needs.
- **For teachers:** what alpha-mannosidosis can look like in the classroom, suggested teaching approaches, and strategies to help with learning and participation.

Patient diary

A simple daily or weekly diary for tracking symptoms, mood, appointments, treatments, and anything else families want to monitor.

Future publication in 2026

- A Wellbeing Toolkit with strategies and resources to support mental and emotional health.
- A Transition to Adulthood Guide to help young people and families prepare for the shift from paediatric services to adult care, as well as growing independence.

Discussion points and questions

- Families and participants were very positive about the resources being developed and felt they would be extremely useful. They also suggested some additional tools that could benefit the community, including:
- A quick access emergency passport for use during hospital visits.
- Employer guidance to help adults in work receive appropriate adjustments and understanding.
- Support materials for siblings to help them understand the condition and express their feelings.
- Clear mental health signposting so families know where to find emotional and psychological support.

Group discussion – support, future meetings and community needs

During our group discussion, families shared what matters most to them and what would help the alpha-mannosidosis community stay connected and supported.

Key themes

- Everyone placed strong value on staying connected, learning from one another, and knowing they are not alone.
- Families highlighted the need for better psychological and emotional support, both for individuals living with the condition and for caregivers.
- There was a clear interest in improving international connections, so families from different countries can learn from each other's experiences.
- Participants also expressed the importance of having quality of life tools that work well for ultra rare conditions, helping families and clinicians understand daily challenges more clearly.

How often should we meet online?

Most people preferred holding community meetings twice a year and suggested once in March/April and once in September/October. These timings help ensure that updates from major conferences can be shared in a simple and accessible way.

MPS Society UK
mpssociety.org.uk

MPS Germany
mps-ev.de

MPS Spain
mpslisosomales.org

Thoughts on an in person meeting

There was strong interest in meeting face to face in the future. Families felt this would help build deeper connections and create a sense of community. Several important points were raised:

- The meeting could potentially be linked to a MPS Society event, as some people already attend these.
- Suggested locations included
 - Germany (Würzburg)
 - Spain (Barcelona)
 - UK (Manchester)
- A hybrid option (in person and online) is important so those unable to travel can still take part.
- Funding support, such as help with travel, accommodation or childcare would make it easier for families to attend.

Conclusion

Thank you to everyone who was part of this first international alpha-mannosidosis community meeting. The experiences you shared, whether as parents, carers, clinicians, researchers or as someone living with the condition, helped bring into focus the priorities that matter most to our community.

Although alpha-mannosidosis is rare, this meeting showed that none of us are facing it alone. By staying connected, learning from one another and continuing to meet, we can build a stronger and more supportive community.

We are grateful for your time, honesty and contributions, and we look forward to continuing this journey together.

With all our thanks

MPS UK, MPS Germany and MPS Spain