

Let's talk about alpha-mannosidosis with your healthcare team



Preparing for appointments with healthcare professionals who may not regularly see patients with alpha-mannosidosis

This leaflet has been funded by Chiesi Global Rare Diseases and developed by Rare Disease Research Partners in collaboration with MPS Society UK, ISMRD, specialist clinicians, a metabolic nurse, and reviewed by people with alpha-mannosidosis (AM) and their caregivers. Everyone involved received a fee for their consultancy advice, except for people with AM and their caregivers, who kindly shared their invaluable time and expertise voluntarily.

How can I use this leaflet?

Alpha-mannosidosis is very rare and many healthcare professionals may not regularly see patients with this condition. **You can use this leaflet to prepare for appointments with healthcare professionals who are not familiar with alpha-mannosidosis.** You can complete this leaflet as a patient or on behalf of a patient with alpha-mannosidosis.



It includes tips to help you:

- Prepare for the appointment
- Effectively communicate concerns
- Share what is important to you
- Ask for any support needed

This appointment is with:

Date:

About the person with alpha-mannosidosis:

My main alpha-mannosidosis specialist:

Hospital:

Name:

E-mail:

Phone number:

Date of diagnosis:

Treatment history:

.....
.....
.....
.....

Other healthcare professionals involved in my care (e.g. name and role):

.....
.....
.....
.....
.....

Current medications:

.....
.....
.....
.....

The most important things I want from this appointment:

- 1)
- 2)
- 3)

Disclaimer: This information serves as a guide to help patients with alpha-mannosidosis and/or their caregivers prepare for medical appointments with healthcare professionals who do not regularly treat these patients. This leaflet does not provide medical advice; always consult a healthcare professional if you have any medical questions or concerns.

You can print a copy of these pages to take to your appointment or download an editable PDF to your phone, tablet or laptop from:

If you live in the UK: MPS Society



If you live in the US: ISMRD



If you are unable to scan the QR codes, links are provided on page 5

What healthcare professionals should know about me

Daily living needs

- Help with hearing, communication**
- Help with walking or moving around (e.g. use crutches, wheelchair, rely on another person)
- Help with washing/dressing
- Help with eating/drinking
- Breathing problems
- Emotional or mental health (e.g. behavioural problems, social anxiety, psychosis)
- Other:

.....

.....

.....

.....

.....

.....

Let the healthcare professional know at the start of the appointment if you need help with communication



- Speak slowly and clearly to me - it may take me a bit longer to process what you are saying
- Face me when speaking - I may not hear you
- I may get anxious, for example, if the appointment overruns
- I need someone familiar with me
- Other needs:

.....

.....

.....

.....

.....

.....



Bring someone with you, like a family member or friend for support during the appointment, if you like.

People I would like in the appointment with me:

.....

.....



Watch out: let the healthcare professional know if any of these apply

Risks the healthcare professional should be aware of:

- Back, joint or muscle issues
- May have issues with anaesthesia, cannulation, scans, x-rays
- Airway issues (e.g. breathing, enlarged tonsils, sleep apnoea - breathing may stop during sleep)
- Seizures or episodes of confusion
- Heart problems (e.g. heart valve disease, heart beats abnormally)
- Immune issues (e.g. infection risk, vaccines may not work well)
- Feeding tube

General/medication allergies:

.....

.....

Other (e.g. surgeries or procedures):


.....

.....

.....

.....

.....

 **Current symptoms or concerns checklist**

Issues to discuss during the appointment

(e.g. when it started, is it getting worse or remaining the same, how it affects the person with alpha-mannosidosis)

Brain or nerves

- Fatigue
- Headache/migraine
- Pain/ache or discomfort in the body (where?)
- Seizures/confusion
- Issues with sleep

Digestive system

- Stomach, bowel, digestion

Ears

- Hearing issues/loss

Eyes

- Vision

Immune system

- Infection (e.g. ear, chest)
- Allergy

Lungs and airways

- Breathing, coughing

Muscle, bones and joints

- Muscle or joint pain, stiffness
- Movement, coordination, balance

Issues with...

- Behaviour or learning difficulties (behavioural problems, anxiety, psychosis)
- Mood
- School, work, day centre

- Other issues or things you have noticed**

✔ If you are not sure how to start the conversation you could say something like:

TAKE YOUR TIME TO THINK



? Ask any questions you have. Ask for an explanation if you do not understand.

“My child has alpha-mannosidosis. It’s a lifelong inherited condition that affects the brain and multiple body systems including hearing, learning, movement, immunity, and bones. It’s progressive and needs regular specialist care. Their alpha-mannosidosis is managed by **[name of specialist centre]** and under the care of **[name specialists]**. We’re here today because we need **[care e.g. a scan, referral, have an infection, are in pain - list of symptoms from checklist]**.”

You can also give your doctor the matching leaflet called ‘*Let’s talk about alpha-mannosidosis for healthcare professionals*’, which includes detailed medical information to guide care and will help explain the condition, if needed.

You can print or download the leaflet for healthcare professionals here:

! If you are unable to scan the QR codes, links are provided on page 5

If you live in the UK:



MPS Society

If you live in the US:



ISMRD

? **Example questions to ask your healthcare professional:**

About symptoms/care

- Is there anything I can do to manage the symptom/s better?
 - Any changes in diet or lifestyle I can make to alleviate the symptoms/s?
- Do I need to see a specialist for this?
 - Which specialist?
 - Are you able to make the referral?
 - Is seeing the specialist covered by my insurance? (if applicable in your country)
- Are there other related signs or symptoms I need to look out for?
- What are the risks to my other children or family members to be affected by alpha-mannosidosis?
- What are my treatment options?

Other questions you may have

.....

.....

.....

.....

.....

.....

.....

About tests

- What are the tests for? What will these tests tell us?
- What do these tests involve?
- Will the test be painful or uncomfortable? How long will the test take?
- Are there any risks or side effects?
- Do I need to prepare for it (e.g. fasting)?
- How/when will I get the results? Who will explain them to me?
- Who do I contact if I do not get the results?
- Is this test covered by my insurance? (if applicable in your country)
- Can you provide support for school Individual Educational Plans/Educational Support Plans?

About follow-up

- Who should I contact if I am worried or symptoms change?
 - Who should be my first point of contact after today?
 - How do I contact them?
 - Do I need to come back and see you? If so, when?
- Will you share updates with my specialist team?
- Where can I go for more information?
- Is there anyone I can talk to for support in the meantime?
- Can I have a summary report with actions from the appointment?

