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:

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

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Current medications:

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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:

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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:

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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:

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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:

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- [ ] Other (e.g. surgeries or procedures):

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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:

"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]***."

TAKE YOUR TIME TO THINK

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

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

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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?

- Use this page to write down what you discussed during the appointment and what happens next. Keep your notes.
- Follow up with your main alpha-mannosidosis specialist if needed.
- Keep copies of referral letters and test results. You can ask to see copies of letters written about the person with alpha-mannosidosis.

Remember, you can request a summary report from your healthcare professional with actions from the appointment.

Before you leave check that you:

- Covered everything on your most important things list that you wanted to discuss
- Understood what your doctor has told you and asked if you were not sure
- Know what should happen next and when

Date of appointment: .....

I had this conversation with: .....

My appointment notes:

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Next steps:

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QR code links

If you live in the UK: www.mpssociety.org.uk/conditions/research/alpha-mannosidosis-patient-and-caregiver-resources
If you live in the US: www.ismrd.org/glycoprotein-diseases/alpha-mannosidosis/#patientresources

The MPS Society (www.mpssociety.org.uk), the Society for Mucopolysaccharide Diseases, is the only registered charity providing professional support to individuals and families affected by MPS, Fabry and related lysosomal conditions in the UK. We are committed to transforming lives through specialist knowledge, support and research, making sure anyone affected by these conditions gets to live the life they want.

ISMRD (www.ismrd.org), the International Society for Mannosidosis and Related Diseases, is an internationally focused not-for-profit organization whose mission is to advocate for families and patients. We are The International Advocate for Glycoprotein Storage Diseases. Our mission, through partnerships built with medicine, science and industry, is to seek, detect and cure these diseases while providing a global network of support and information.

Rare Disease Research Partners (www.rd-rp.com) is a wholly owned, not for profit subsidiary of the MPS Society. Its social objectives are to reinvest any surplus to support the mission of the MPS Society to transform the lives of patients through specialist knowledge, support, advocacy and research.

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