How is alpha-mannosidosis treated?

There is currently no cure for alpha-mannosidosis. To date, management of the disease has focused on treating individual symptoms, improving quality of life, and preventing the development of further complications.

Patients with alpha-mannosidosis will usually be managed by a team of specialists working with a pediatrician to determine the patient's specific needs and suggest appropriate solutions. For example:

Ear infections	Otolaryngologist	Spinal deformities	Orthopedic specialist
Hearing loss	Hearing specialist	General orthopedic problems	Orthopedic specialist/ physiotherapist
Dental alterations	Dentist	Cognitive impairment	Psychiatrist/ psychologist/ neurologist
Eye problems	Ophthalmologist	Joint problems	Orthopedic specialist

Today, investigational treatments are in development to address the underlying cause of the disease.

Enzyme replacement therapy (ERT)

ERT is used in other lysosomal storage disorders, like Gaucher, Fabry, or Pompe disease. It is meant to replace the missing enzyme, reducing the amount of sugar build-up in the cells.

Bone marrow transplantation (BMT)

BMT uses donor cells to repopulate the patient's tissues and distribute the working enzyme.

Join the alpha-mannosidosis community

ISMRD

International Society for Mannosidosis and Related Diseases

A community for those with glycoprotein storage disorders, including alpha-mannosidosis.

Find out more at ismrd.org

For more information about alpha-mannosidosis, visit

discoveralphamannosidosis.com

References available upon request.

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Alpha-Mannosidosis

Alpha-mannosidosis:

A guide for parents and caregivers

Chiesi global rare diseases

What is alpha-mannosidosis?

Alpha-mannosidosis is a rare, inherited condition known as a **lysosomal storage disorder**. It occurs when the body is unable to make an enzyme that breaks down certain sugars in cells. This causes the sugars to build up, which can damage organs and tissues throughout the body.

The disease can generally be split into three types, depending on the severity of symptoms:

— MOST COMMON —

Type 1

Mild form

- Usually recognized after age 10 years
- Slow disease progression

Type 2

Moderate form

- Usually recognized before age 10 years
- Slow disease progression

Type 3

Severe form

- **High rates of mortality, with central nervous system** involvement or skeletal muscle involvement

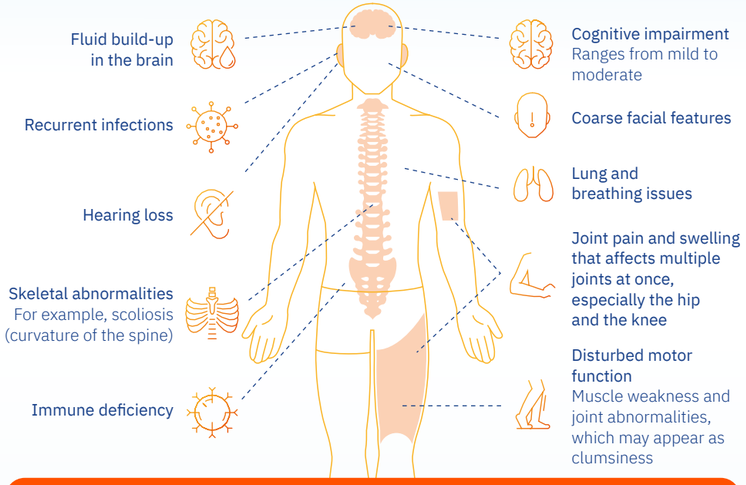
How common is alpha-mannosidosis?

Alpha-mannosidosis is extremely rare; it is thought to occur in around **1 in 500,000 to 1 in 1,000,000 births** worldwide.

1 in 500,000 to 1 in 1,000,000

What are the symptoms?

The clinical presentation of alpha-mannosidosis is variable, meaning there is a wide variety of symptoms patients may experience. Some of the possible symptoms and how they may change as patients get older are shown in the diagram below:



Childhood

- Hearing loss
- Delayed speech, motor functions, or mental functions
- Fluid build-up in the brain in the first year of life

2nd to 3rd decade of life

- Muscular weakness
- Ataxia (problems with coordination and balance)
- Skeletal abnormalities
- Joint problems
- Psychiatric symptoms

Adulthood

- Inability to achieve social independence
- Life expectancy unknown, although living to over 50 years is not uncommon

Living with alpha-mannosidosis

Alpha-mannosidosis can have a significant impact on patients and their families. The all-consuming nature of the condition means that patients require a high level of care.

The symptoms of alpha-mannosidosis mean that interventions such as hearing aids, glasses, or a wheelchair are often required.

Hearing aids for hearing loss

Glasses to correct vision

Use of a wheelchair

Because of the impact of alpha-mannosidosis, patients may need adaptations at home and school.

Home environment

- Wheelchair ramps may be needed
- For patients with visual impairment, it can help to ensure the home is well lit, remove trip hazards, and install non-slip flooring in the bathroom and kitchen

Educational intervention

- Patients are likely to need early educational intervention, such as help with development of social skills, speech therapy, and special education

Caring for someone with alpha-mannosidosis can be challenging. If you are caring for someone with the disease, keep in regular contact with their healthcare team, as they can offer specific advice depending on the patient's individual needs.