

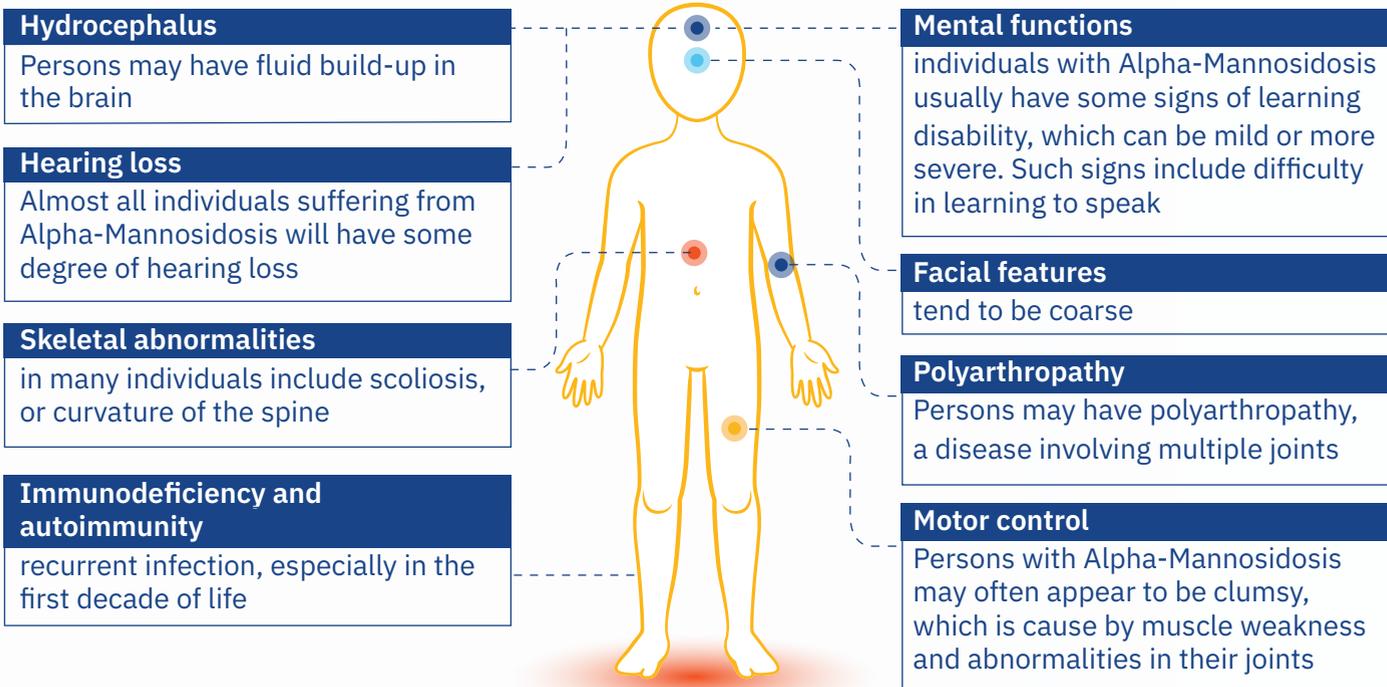
DISCOVER™

Alpha-Mannosidosis

What is Alpha-Mannosidosis?

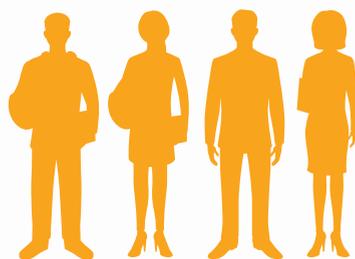
Alpha-Mannosidosis is a rare inherited disease that can cause children and adults to suffer from skeletal deformities, coarse facial features, hearing loss, cognitive disabilities, problems with the immune system (which may mean, for example, you catch infections more readily), mental health and behavioural issues.¹

Signs of Alpha-Mannosidosis²



First presentation

Recurrent ear infections associated with hearing loss.¹ Progressive impairment of mental functions, speech and motor control.¹ Hydrocephalus can be present in the first year of life.¹



2nd-3rd decade of life

Muscular weakness and ataxia, skeletal abnormalities and destructive poly-arthropathy.¹

Neuro-cognitive development is usually arrested and psychiatric disorders may also manifest.¹



In adulthood

Patients are unable to achieve complete social independence.¹

There are very limited data regarding life expectancy in adults.

Timely diagnosis of Alpha-Mannosidosis is important as it can influence how well the individual does in the long-term as therapies should be initiated as soon as possible.³

Your doctor can use a number of very simple tests to diagnose Alpha-Mannosidosis, including:



Urine sample¹

This will measure oligosaccharide levels in urine. High levels will be suggestive of Alpha-Mannosidosis.



Blood sample¹

A small sample of blood is taken to measure the presence of mannosidase enzyme in blood cells. This can be used to confirm a diagnosis.

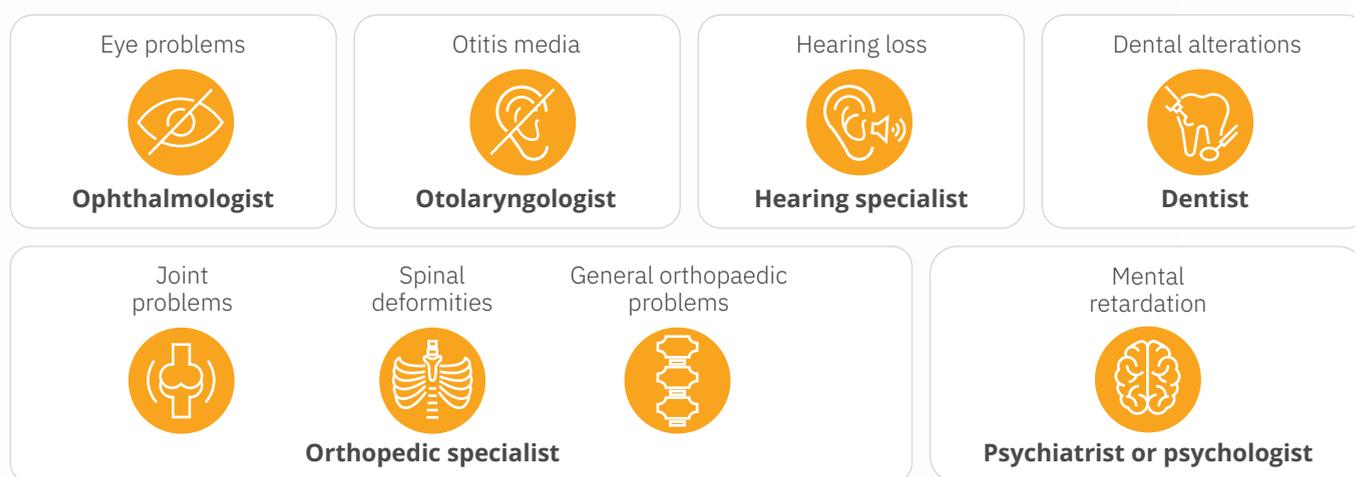


Genetic testing¹

A diagnosis of Alpha-Mannosidosis can also be confirmed through genetic testing, which can detect the mutation in the gene that causes the disorder.

Caring for persons with Alpha-Mannosidosis

Ideally, the individual with alpha-mannosidosis will be followed by a team of medical specialists who can help determine the specific needs of each person and help suggest the most appropriate solutions.¹ For examples:



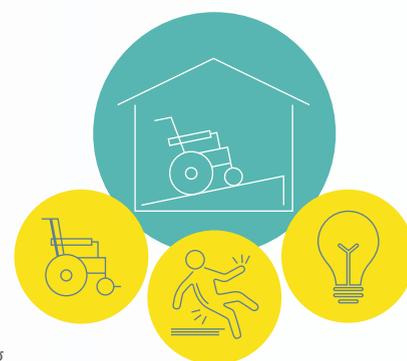
Tips for helping those with Alpha-Mannosidosis²

The home environment should be adequately prepared to meet these needs, such as

- The use of ramps to allow wheelchair access
- Fixing any hazards for walking and install non-slip flooring in the bathroom and kitchen
- Increasing the amount of light

Moreover, individuals will need early educational intervention for development of social skills, speech therapy and special education to maximise learning.

Thus, caring for someone with Alpha-Mannosidosis can be extremely challenging. If you are caring for someone with the disease, always keep in close contact with the specialist who is managing the person, as they can provide specific advice depending on the individual needs.



References:

1. Malm D & Nilssen Ø. Alpha-mannosidosis. Orphanet J Rare Dis 2008;3:21 -
2. Malm D, Nilssen O. Alpha-Mannosidosis. NCBI. Available at: <https://www.ncbi.nlm.nih.gov/books/NBK1396/> -
3. Guffon N, et al. Mol Gen Metab. 2019;126:470.