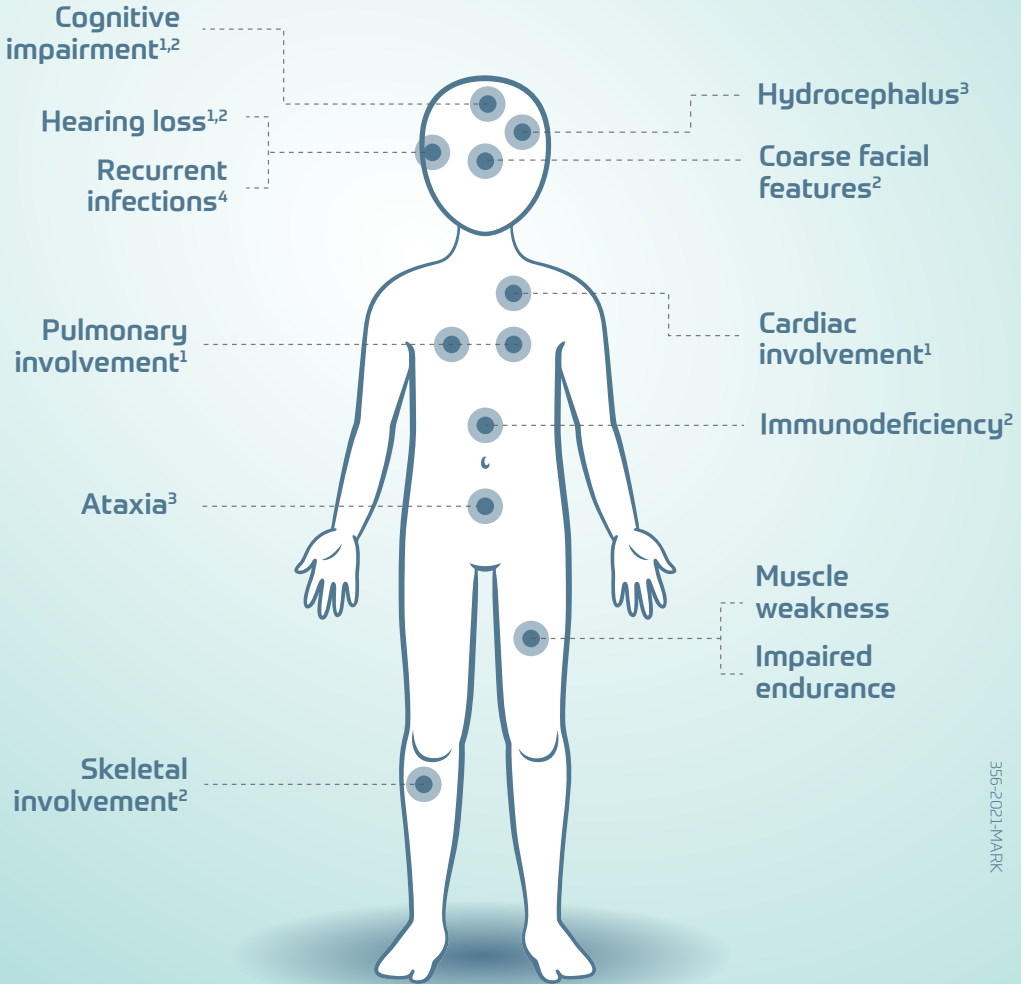


Could it be Alpha-mannosidosis ?



356-2021-MARK

Diagnosing alpha-mannosidosis

Patients ≤ 10 years⁴

1. Hearing impairment and/or speech delay

↓ Yes

2. Plus at least two manifestations among:

- Cognitive delay
- Motor disturbances/impaired balance
- Facial features (can be milder than MPS)

↓ No

↓ Yes

3. Keep monitoring if alpha-mannosidosis signs in box 2 appear

Yes



4. Refer to an expert metabolic centre

Patients > 10 years⁴

1. Mental retardation and motor impairment regression and/or psychiatric manifestations*

↓ Yes

2. History of at least two among:

- Hearing impairment
- Intellectual disability
- Motor disturbances/ataxia
- Skeletal disorders/joints disease

↓ No

↓ Yes

3. Keep monitoring if alpha-mannosidosis signs in box 2 appear

Yes



4. Refer to an expert metabolic centre

Adapted from Guffon et al. 2019⁴

References

1. Beck M et al. *Orphanet J Rare Dis* 2013;8:88.
2. Borgwardt L et al. *Orphanet J Rare Dis* 2015;10:70.
3. Malm D and Nilssen Ø. *Orphanet J Rare Dis* 2008;3:21.
4. Guffon N et al. *Mol Genet Metab* 2019;126(4):470–474.