



# FROM CLUES TO CARE: IMPROVING RECOGNITION AND TREATMENT OF ALPHA-MANNOSIDOSIS

## Reference List

- Avenarius DF, Svendsen JS, Malm D. Proton nuclear magnetic resonance spectroscopic detection of oligomannosidic n glycans in alpha-mannosidosis: a method of monitoring treatment. *J Inherit Metab Dis*. 2011;34(5):1023-1027. doi:10.1007/s10545-011-9331-7
- Borgwardt L, Guffon N, Amraoui Y, et al. Efficacy and safety of velmanase alfa in the treatment of patients with alpha-mannosidosis: results from the core and extension phase analysis of a phase III multicentre, double-blind, randomised, placebo-controlled trial. *J Inherit Metab Dis*. 2018;41(6):1215-1223. doi:10.1007/s10545-018-0185-0
- Ceccarini MR, Codini M, Conte C, et al. Alpha-mannosidosis: Therapeutic strategies. *Int J Mol Sci*. 2018;19(5):1500. doi:10.3390/ijms19051500
- Ficicioglu C, Stepien KM. Alpha-mannosidosis. 2001 [updated 2024]. In: Adam MP, Bick S, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, eds. *GeneReviews [Internet]*. Seattle, WA: University of Washington, Seattle; 1993-2006.
- Guffon N, Borgwardt L, Tylki-Szymańska A, et al. Extended long-term efficacy and safety of velmanase alfa treatment up to 12 years in patients with alpha-mannosidosis. *J Inherit Metab Dis*. 2025;48(1):e12799. doi:10.1002/jimd.12799
- Guffon N, Burton BK, Ficicioglu C, et al. Monitoring and integrated care coordination of patients with alpha-mannosidosis: A global Delphi consensus study. *Mol Genet Metab*. 2024;142(4):108519. doi:10.1016/j.ymgme.2024.108519
- Guffon N, Tylki-Szymanska A, Borgwardt L, et al. Recognition of alpha-mannosidosis in paediatric and adult patients: Presentation of a diagnostic algorithm from an international working group. *Moll Genet Metab*. 2019;126(4):470-474. doi:10.1016/j.ymgme.2019.01.024
- Köse E, Kasapkara Ç S, İnci A, et al. Long-term clinical evaluation of patients with alpha-mannosidosis - A multicenter study. *Eur J Med Genet*. 2024;68:104927. doi:10.1016/j.ejmg.2024.104927
- Lamzede. Prescribing information. Chiesi USA, Inc., 2023. Accessed March 3, 2026. [https://www.accessdata.fda.gov/drugsatfda\\_docs/label/2023/761278s000lbl.pdf](https://www.accessdata.fda.gov/drugsatfda_docs/label/2023/761278s000lbl.pdf)
- Manickam K, McClain MR, Demmer LA, et al. Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2021;23(11):2029-2037. doi:10.1038/s41436-021-01242-6
- Mynarek M, Tolar J, Albert MH, et al. Allogeneic hematopoietic SCT for alpha-mannosidosis: an analysis of 17 patients. *Bone Marrow Transplant*. 2012;47(3):352-359. doi:10.1038/bmt.2011.99
- Šáhó R, Formánková R, Eisengart JB, et al. Outcome of haemopoietic stem cell transplantation in 21 patients with alpha-mannosidosis. *J Inherit Metab Dis*. 2025;48(4):e70047. doi:10.1002/jimd.70047
- Santoro L, Cefalo G, Canalini F, Rossi S, Scarpa M. Diagnosis of alpha-Mannosidosis: Practical approaches to reducing diagnostic delays in this ultra-rare disease. *Mol Genet Metab*. 2024;142(1):108444. doi:10.1016/j.ymgme.2024.108444
- Santoro L, Monachesi C, Zampini L, et al. First experience of combined enzyme replacement therapy and hematopoietic stem cell transplantation in alpha-mannosidosis. *Am J Med Genet A*. 2023;191(7):1948-1952. doi:10.1002/ajmg.a63210
- Stepien KM, Thomas S, Hennermann JB, et al. Evolution of mobility, pain/discomfort, self-care, and mental health in patients with alpha-mannosidosis: an international caregiver and patient survey. *Orphanet J Rare Dis*. 2025;20(1):217. doi:10.1186/s13023-025-03694-4