

# THE POMPE DISEASE

## *Suggested Readings by Topic*

### Introduction

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Bernstein DL, et al. Pompe disease: dramatic improvement in gastrointestinal function following enzyme replacement therapy. A report of three later-onset patients. *Mol Genet Metab*. 2010;101:130-133. <https://pubmed.ncbi.nlm.nih.gov/20638881/>

Burrow TA, et al. Acute progression of neuromuscular findings in infantile Pompe disease. *Pediatr Neurol*. 2010;42:455-458. <https://pubmed.ncbi.nlm.nih.gov/20472203/>

Hagemans MLC, et al. Clinical manifestation and natural course of late-onset Pompe's disease in 54 Dutch patients. *Brain*. 2005;128:671-677. <https://pubmed.ncbi.nlm.nih.gov/15659425/>

Hagemans MLC, et al. Disease severity in children and adults with Pompe disease related to age and disease duration. *Neurology*. 2005;64:2139-2141. <https://pubmed.ncbi.nlm.nih.gov/15985590/>

Hahn SH, et al. Efficacy, safety profile, and immunogenicity of alglucosidase alfa produced at the 4,000-liter scale in US children and adolescents with Pompe disease: ADVANCE, a phase IV, open-label, prospective study. *Genet Med*. 2018;20:1284-1294. <https://pubmed.ncbi.nlm.nih.gov/29565424/>

Hamdan MA, et al. Antenatal diagnosis of pompe disease by fetal echocardiography: impact on outcome after early initiation of enzyme replacement therapy. *J Inherit Metab Dis*. 2010;33(suppl 3):S333-S339. <https://pubmed.ncbi.nlm.nih.gov/20821053/>

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Kishnani PS, et al. Pompe disease diagnosis and management guideline. *Genet Med*. 2006;8:267-288. <https://pubmed.ncbi.nlm.nih.gov/16702877/>

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Kishnani PS, et al. The new era of Pompe disease: advances in the detection, understanding of the phenotypic spectrum, pathophysiology, and management. *Am J Med Genet Part C Semin Med Genet*. 2012;160C:1-7. <https://pubmed.ncbi.nlm.nih.gov/22253049/>

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## Improving Disease Progression Outcomes with Advancing Therapeutics

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Kronn D, et al. Response to omalizumab in a 5 yr old patient with infantile onset Pompe disease (IOPD) with anaphylactoid reactions. 14th Annual World Symposium. February 5-9, 2018. Poster 198.

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Kronn D, et al. Mini-COMET study: safety, biomarker, and efficacy data after avalglucosidase alfa dosing for  $\geq 97$  weeks in participants with infantile-onset Pompe disease (IOPD) previously treated with alglucosidase alfa who had demonstrated clinical decline. Presented at: 14th Annual WorldSymposium; February 5-9, 2018; Abstract 156.

Leslie N, Bailey L. Pompe disease. In: Adam MP, et al (eds). *GeneReviews [Internet]*. University of Washington, Seattle; 1993-2022. <https://www.ncbi.nlm.nih.gov/books/NBK1261>

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Young SP, et al. Assessing disease severity in Pompe disease: the roles of a urinary glucose tetrasaccharide biomarker and imaging techniques. *Am J Med Genet C Semin Med Genet.* 2012;160C:50-58.

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## Clinical Case Review and Discussion: Implementing New Therapies into Practice

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Kazi ZB, et al. An immune tolerance approach using transient low-dose methotrexate in the ERT-naïve setting of patients treated with a therapeutic protein: experience in infantile-onset Pompe disease. *Genet Med*. 2019;21:887-895. doi:10.1038/s41436-018-0270-7

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Kronn DF, et al. Management of confirmed newborn-screened patients with Pompe disease across the disease spectrum. *Pediatrics*. 2017;140(suppl 1):S24-S45. [https://publications.aap.org/pediatrics/article/140/Supplement\\_1/S24/179262/Management-of-Confirmed-Newborn-Screened-Patients](https://publications.aap.org/pediatrics/article/140/Supplement_1/S24/179262/Management-of-Confirmed-Newborn-Screened-Patients)

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