

UTILIZATION MANAGEMENT MEDICAL POLICY

POLICY: Pompe Disease – Enzyme Replacement Therapy – Pombiliti Utilization Management Medical Policy

- Pombiliti® (cipaglucosidase alfa-atga intravenous infusion – Amicus)

REVIEW DATE: 05/06/2026

OVERVIEW

Pombiliti, a hydrolytic lysosomal glycogen-specific recombinant human α -glucosidase enzyme, is indicated in combination with Opfolda® (miglustat capsules), an enzyme stabilizer, for **late-onset Pompe disease** (lysosomal acid α -glucosidase deficiency) in adults weighing ≥ 40 kg and who are not improving on their current enzyme replacement therapy.¹

Disease Overview

Pompe disease (glycogen storage disease type II, or acid maltase deficiency), is a rare lysosomal storage disorder characterized by a deficiency in acid α -glucosidase activity leading to the accumulation of glycogen, particularly in muscle.² In general, the condition is defined by the age of onset, organ involvement, severity and rate of progression. Late-onset Pompe disease has a more variable clinical course and can manifest any time after 12 months of age. Patients typically present with progressive muscle weakness which can progress to respiratory insufficiency. The diagnosis of Pompe disease is established by a deficiency of acid α -glucosidase activity or by identification of biallelic pathogenic variants by genetic testing. Enzyme replacement therapy has an important role in the management of Pompe disease.- Pombiliti plus Opfolda have a role in patients with late-onset Pompe disease who are not improving on their current enzyme replacement therapy regimen.

POLICY STATEMENT

Prior Authorization is recommended for medical benefit coverage of Pombiliti. Approval is recommended for those who meet the **Criteria** and **Dosing** for the listed indication. Extended approvals are allowed if the patient continues to meet the Criteria and Dosing. Requests for doses outside of the established dosing documented in this policy will be considered on a case-by-case basis by a clinician (i.e., Medical Director or Pharmacist). All approvals are provided for the duration noted below. Because of the specialized skills required for evaluation and diagnosis of patients treated with Pombiliti as well as the monitoring required for adverse events and long-term efficacy, approval requires Pombiliti to be prescribed by or in consultation with a physician who specializes in the condition being treated.

Automation: None.

RECOMMENDED AUTHORIZATION CRITERIA

Coverage of Pombiliti is recommended in those who meet the following criteria:

FDA-Approved Indication

- 1. Acid Alpha-Glucosidase Deficiency (Pompe Disease).** Approve for 1 year if the patient meets ALL of the following (A, B, C, D, E, and F):
 - A)** Patient is ≥ 18 year of age; AND
 - B)** Patient weighs ≥ 40 kg; AND

- C) The medication will be used in combination with Opfolda (miglustat capsules); AND
- D) Patient has not demonstrated an improvement in objective measures after receiving ONE of the following for at least one year (i or ii):
- Note: Examples of objective measures include forced vital capacity (FVC) and six-minute walk test (6MWT).
- i. Lumizyme (alglucosidase alfa intravenous infusion); OR
- ii. Nexviazyme (avalglucosidase alfa-ngpt intravenous infusion); AND
- E) Patient has late-onset acid alpha-glucosidase deficiency (late-onset Pompe disease) with diagnosis established by ONE of the following (i or ii):
- i. Patient has a laboratory test demonstrating deficient acid alpha-glucosidase activity in blood, fibroblasts, or muscle tissue; OR
- ii. Patient has a molecular genetic test demonstrating biallelic pathogenic or likely pathogenic acid alpha-glucosidase (GAA) gene variants; AND
- F) The medication is prescribed by or in consultation with a geneticist, neurologist, a metabolic disorder sub-specialist, or a physician who specializes in the treatment of lysosomal storage disorders.

Dosing. Each dose must not exceed 20 mg/kg administered intravenously no more frequently than once every 2 weeks.

CONDITIONS NOT RECOMMENDED FOR APPROVAL

Coverage of Pombiliti is not recommended in the following situations:

1. Coverage is not recommended for circumstances not listed in the Recommended Authorization Criteria. Criteria will be updated as new published data are available.

REFERENCES

1. Pombiliti® intravenous infusion [prescribing information]. Philadelphia, PA: Amicus; July 2024.
2. Sperry E, Leslie N, Berry L, et al. Pompe Disease. 2007 Aug 31 [Updated 2025 Aug 21]. In: Adam MP, Bick S, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2026. Available from: <https://www.ncbi.nlm.nih.gov/books/NBK1261/>. Accessed on May 4, 2026.

HISTORY

Type of Revision	Summary of Changes	Review Date
Early Annual Revision	Acid Alpha-Glucosidase Deficiency (Pompe Disease): Confirmation of a genetic mutation in the acid alpha-glucosidase gene was rephrased to more specifically state, “genetic test demonstrating biallelic pathogenic or likely pathogenic acid alpha-glucosidase gene variants”.	05/08/2024
Annual Revision	No criteria changes.	05/07/2025
Annual Revision	No criteria changes.	05/06/2026