CARECONTINUUM™

UTILIZATION MANAGEMENT MEDICAL POLICY

POLICY: Pompe Disease – Enzyme Replacement Therapy – Lumizyme Utilization Management Medical Policy
- Lumizyme® (alglucosidase intravenous infusion – Genzyme)

REVIEW DATE: 05/07/2025

OVERVIEW

Lumizyme, a human hydrolytic lysosomal glycogen-specific enzyme (acid α-glucosidase), is indicated for patients with **Pompe disease** (acid α-glucosidase deficiency).[1] It is produced in a Chinese hamster ovary cell line via recombinant DNA technology. After administration of Lumizyme, it is internalized into cells and transported to lysosomes where it catalyzes the breakdown of glycogen to glucose.

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.[2,3] The onset, progression, and severity of Pompe disease is variable. Infantile-onset Pompe disease usually manifests in the first few months of life and death often occurs in the first year of life, if left untreated.[2] Clinical manifestations of infantile-onset Pompe disease includes hypotonia, difficulty feeding, and cardiopulmonary failure.[4] Late-onset Pompe disease a has more variable clinical course, can manifest any time after 12 months of age, and patients typically present with progressive muscle weakness which can progress to respiratory insufficiency.[3,4] The diagnosis of Pompe disease is established by demonstrating decreased acid α-glucosidase activity in blood, fibroblasts, or muscle tissue; or by genetic testing.[3,4] Definitive treatment of Pompe disease consists of enzyme replacement therapy with Lumizyme.[2-4]

POLICY STATEMENT

Prior Authorization is recommended for medical benefit coverage of Lumizyme. 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 Lumizyme as well as the monitoring required for adverse events and long-term efficacy, approval requires Lumizyme to be prescribed by or in consultation with a physician who specializes in the condition being treated.

Automation: None.

RECOMMENDED AUTHORIZATION CRITERIA

Coverage of Lumizyme 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 BOTH of the following (A and B):
   - **A)** The diagnosis is 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
   - **B)** Lumizyme 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 Lumizyme 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. Lumizyme® intravenous infusion [prescribing information]. Cambridge, MA: Genzyme; December 2024.
2. Chien YH, Hwu WL, Lee NC. Pompe disease: Early diagnosis and early treatment make a difference. *Pediatr Neonatol*. 2013;54:219-227.
3. Llerena Junior JC, Nascimento OJM, Oliveira ASB, et al. Guidelines for the diagnosis, treatment and clinical monitoring of patients with juvenile and adult Pompe disease. *Arq Neuropsiquiatr*. 2016;74:166-176.
4. Cupler EJ, Berger KI, Leshner RT, et al. Consensus treatment recommendations for late-onset Pompe disease. *Muscle Nerve*. 2012;45:319-333.

HISTORY

Type of Revision	Summary of Changes	Review Date
Annual Revision	No criteria changes.	04/12/2023
Update	10/04/2023: No criteria changes. Policy name changed from Enzyme Replacement Therapy – Lumizyme to Pompe Disease – Enzyme Replacement Therapy – Lumizyme.	NA
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