

Reference

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1. Doerfler P, Nayak S, Corti M, et al. Targeted approaches to induce immune tolerance for Pompe disease therapy. *Mol Ther Methods Clin Dev.* 2016;3:15053.
2. Micromedex DrugDex Compendium®. 2023. Alglucosidase alfa.
3. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Alglucosidase alfa.
4. MCG™ Care Guidelines, 22nd edition, 2018, Home Infusion Therapy, CMT: CMT-0009(SR).
5. Van der Meijden JC, Kruijshaar ME, Harlaar L, et al. Long-term follow-up of 17 patients with childhood Pompe disease treated with enzyme replacement therapy. *J Inherit Metab Dis.* 2018; 41(6):1205-14.
6. Byrne BJ, Fuller DD, Smith BK, et al. Pompe disease gene therapy: neural manifestations require consideration of CNS directed therapy. *Ann Transl Med.* 2019;(13):290-302.
7. Parini R, De Lorenzo P, Dardis A, et al. Long term clinical history of an Italian cohort of infantile onset Pompe disease treated with enzyme replacement therapy. *Orphanet J Rare Dis.* 2018; 13(1):32.
8. Confalonieri M, Vitacca M, Scala R, et al. Is early detection of late-onset Pompe disease a pneumologist's affair? A lesson from an Italian screening study. *Orphanet J Rare Dis.* 2019;14(1):62.
9. Alglucosidase alfa (Lumizyme) for injection, for intravenous use [package insert]. Genzyme Corporation. Cambridge, MA. Revised 03/2023.
10. Avalglucosidase alfa-ngpt (Nexviazyme) for injection, for intravenous use [package insert]. Genzyme Corporation. Cambridge, MA. Issued 04/2023.
11. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Avalglucosidase alfa.
12. Micromedex DrugDex Compendium®. 2023. Avalglucosidase alfa-ngpt.
13. Avalglucosidase Alfa-ngpt In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated August 30, 2021.
14. Pena LDM, Barohn RJ, Byrne BJ, et al. Safety, tolerability, pharmacokinetics, pharmacodynamics, and exploratory efficacy of the novel enzyme replacement therapy avalglucosidase alfa (neoGAA) in treatment-naïve and alglucosidase alfa-treated patients with late-onset Pompe disease: A phase 1, open-label, multicenter, multinational, ascending dose study. *Neuromuscul Disord.* 2019;29(3):167-186.
15. Alglucosidase In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated April 6, 2016.
16. Cao JD, Wiedemann A, Quinaux T, et al. 30 months follow-up of an early enzyme replacement therapy in a severe Morquio A patient: About one case. *Mol Genet Metab Rep.* 2016;42-45.
17. Regier DS, Tanpaiboon P. Role of elosulfase alfa in mucopolysaccharidosis IVA. *Appl Clin Genet.* 2016;9:67–74.
18. Finnigan N, Roberts J, Mercer J, et al.. Home infusion with elosulfase alpha (Vimizim) in a UK pediatric setting. *Mol Genet Metab Rep.* 2017;15–18.

19. Polinski JM, Kowal MK, Gagnon M, et al. Home infusion: safe clinically effective, patient preferred, and cost saving. *Healthcare*. 2016.
20. Micromedex DrugDex Compendium®. 2023. Elosulfase Alfa.
21. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Elosulfase alfa.
22. Elosulfase alfa (Vimizim™) for intravenous administration [package insert]. BioMarin Pharmaceuticals Inc. Novato, CA. Revised 12/2019.
23. Akyol MU, Alden TD, Amartino H, et al. Recommendations for the management of MPS IVA: Systematic evidence- and consensus-based guidance. *Orphanet J Rare Dis*. 2019;14(137).
24. Hendriksz CJ, Parini R, Al Sayed MD, et al. Long-term endurance and safety of elosulfase alfa enzyme replacement therapy in patients with Morquio A syndrome. *Mol Genet Metab*. 2016;119(1-2):131-43.
25. Ficicioglu C, Matalon DR, Luongo N, et al. Diagnostic journey and impact of enzyme replacement therapy for mucopolysaccharidosis IVA: a sibling control study. *Orphanet J Rare Dis*. 2020;15(1):336-46.
26. Barak S, Anikster Y, Sarouk I, Stern E, et al. Long-Term Outcomes of Early Enzyme Replacement Therapy for Mucopolysaccharidosis IV: Clinical Case Studies of Two Siblings. *Diagnostics*. 2020;10(2):108-19
27. Elosulfase alfa In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated June 2, 2016
28. Erazo-Narváez AF, Muñoz-Vidal JM, Rodríguez-Vélez GH, Acosta-Aragón MA. Clinical outcomes in elderly patients with Morquio a syndrome receiving enzyme replacement therapy - experience in a Colombian center. *Mol Genet Metab Rep*. 2020 2;25:1-8.
29. Naglazyme® (galsulfase), injection, for intravenous use [package insert]. BioMarin Pharmaceutical Inc. Novato, CA. Revised 12/2019.
30. Furujo M, Kosuga M, Okuyama T. Enzyme replacement therapy attenuates disease progression in two Japanese siblings with mucopolysaccharidosis type VI: 10-Year follow up. *Mol Genet Metab*. 2017;13:69-75.
31. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Galsulfase.
32. American Society of Health-System Pharmacists. ASHP guidelines on home infusion pharmacy services. *Am J Health-Syst Pharm*. 2014;71:325–41.
33. Polinski JM, Kowal MK, Gagnon M, et al. Home infusion: safe clinically effective, patient preferred, and cost saving. *Healthc(Amst)*. 2017;5(1-2):68-80.
34. American Society of Health-System Pharmacists. ASHP guidelines on evaluating and using home or alternate-site infusion service providers. *Am J Health-Syst Pharm*. 2016;73:922–6.
35. Quartel A, Harmatz PR, Lampe C, et al. Long-term galsulfase treatment associated with improved survival of patients with mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome) 15-year follow-up from the survey study. *J Inborn Errors Metab Screen*. 2018;6:1-6.
36. Akyol MU, Alden TD, Amartino H, et al. Recommendations for the management of MPS VI: systematic evidence-and consensus-based guidance. *Orphanet J Rare Dis*. 2019;14(1):118.

37. Garcia P, Phillips D, Johnson J, et al. Long-term outcomes of patients with mucopolysaccharidosis VI treated with galsulfase enzyme replacement therapy since infancy. *Mol Genet Metab.* 2021;133(1):100-108.
38. Micromedex DrugDex Compendium®. 2023. Galsulfase.
39. Galsulfase In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated January 1, 2006.
40. Eleprase® (Idursulfase) injection, for intravenous use [package insert]. Takeda Pharmaceuticals U.S.A., Inc. Lexington, MA. Revised 9/2021.
41. Pano A, Barbier A, Bielefeld B, et al. Immunogenicity of idursulfase and clinical outcomes in very young patients (16 months to 7.5 years) with mucopolysaccharidosis II (Hunter syndrome). *Orphanet J Rare Dis.* 2015;10:
42. Muenzer J, Hendriksz C, Fan Z, et al. A phase I/II study of intrathecal idursulfase-IT in children with severe mucopolysaccharidosis II. *Genet in Medicine.* 2015;36.
43. Sestito S, Ceravolo F, Grisolia M, et al. Profile of idursulfase for the treatment of Hunter syndrome. *Res Rep Endocr Disord.* 2015;5:79-90.
44. Guffon N, Heron B, Chabrol B, et al. Diagnosis, quality of life, and treatment of patients with Hunter syndrome in the French healthcare system: A retrospective observational study. *Orphanet J Rare Dis.* 2015;10:43.
45. Polinski JM, Kowal MK, Gagnon M, et al. Home infusion: safe clinically effective, patient preferred, and cost saving. *Healthcare.* 2016.
46. Micromedex DrugDex Compendium®. 2023. Idursulfase.
47. Idursulfase In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated September 1, 2010.
48. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Idursulfase.
49. Muenzer J, Jones SA, Tylki-Szymańska A, et al. Ten years of the Hunter Outcome Survey (HOS): insights, achievements, and lessons learned from a global patient registry. *Orphanet J Rare Dis.* 2017. 12(1):82.
50. Bradley L. A., Haddow H. R., & Palomaki G. E. Treatment of mucopolysaccharidosis type II (Hunter syndrome): results from a systematic evidence review. *Genet Medicine.* 2017. 19(11), 1187-1201.
51. Erwin AL. The role of sebelipase alfa in the treatment of lysosomal acid lipase deficiency. *Ther Adv Gastroenterol.* 2017;553-562.
52. Micromedex DrugDex Compendium®. 2023. Sebelipase alfa.
53. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Sebelipase alfa.
54. Kanuma (sebelipase alfa) for intravenous infusion [package insert]. Alexion Pharmaceuticals Inc. New Haven,CT. Revised 11/2021.

55. Jones SA, Rojas-Caro S, Quinn AG, Friedman M, Marulkar S, Ezgu F, et al. Survival in infants treated with sebelipase alfa for lysosomal acid lipase deficiency: an open-label, multicenter, dose-escalation study. *Orphanet J Rare Dis.* 2017; 12(25):1-12.
56. Jones SA, Valayannopoulos V, Schneider E, et al. Rapid progression and mortality of lysosomal acid lipase deficiency presenting in infants. *Genet Med.* 2016 18(5), 452-458.
57. Patterson C, So S, DeAngelis M, Ghent E, Southmayd D, and Carpenter C. Physical activity experiences in children post-liver transplant: Developing a foundation for rehabilitation interventions. *Pediatr Transplant.*2018. 22(4), e13179.
58. Sebelipase Alfa In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated March 22, 2017.
59. Potter JE, Petts G, Ghosh A. et al. Enzyme replacement therapy and hematopoietic stem cell transplant: a new paradigm of treatment in Wolman disease. *Orphanet J Rare Dis.* 2021;16(235): 1-14.
60. Vijay S, Brassier A, Ghosh A. et al. Long-term survival with sebelipase alfa enzyme replacement therapy in infants with rapidly progressive lysosomal acid lipase deficiency: final results from 2 open-label studies. *Orphanet J Rare Dis.* 2021;16(13): 1-16.
61. Clinical Pharmacology™ Compendium. 2023.Tampa FL: Gold Standard, Inc..Vestronidase alfa.
62. Mepsevii™ (vestronidase alfa-vjbc), for intravenous infusion [package insert]. Ultragenyx Pharmaceutical, Inc. Novato, CA. Revised 12/2020.
63. Montano A, Lock-Hock N, Steiner R, et al. Clinical course of sly syndrome (Mucopolysaccharidosis Type VII). *J Med Genet.* 2016;53(6):403-418.
64. Qi Y, McKeever K, Taylor J, et al. Pharmacokinetic and pharmacodynamic modeling to optimize the dose of vestronidase alfa, an enzyme replacement therapy for treatment of patients with mucopolysaccharidosis type VII: results from three trials. *Clin Pharmacokinet.* 2019;58(5):673-83.
65. Harmatz P, Whitley CB, Wang RY, et al. A novel bind start study design to investigate vestronidase alfa for mucopolysaccharidosis VII, an ultra-rare genetic disease. *Mol Genet Metab.* 2018;123(4):488-94.
66. Morrison A, Oussoren E, Friedel T, et al. Pathway to diagnosis and burden of illness in mucopolysaccharidosis type VII—a European caregiver survey. *Orphanet J Rare Dis.* 2019;14(1):254.
67. McCafferty EH, Scott LJ. Vestronidase alfa: a review in mucopolysaccharidosis VII. *BioDrugs.* 2019;11;33(2):233-240.
68. Wang R, da Silva Franco JF, Lopez-Valdez J, et al. The long-term safety and efficacy of vestronidase alfa, rhGUS enzyme replacement therapy, in subjects with mucopolysaccharidosis VII. *Mol Genet Metab.* 2020;129(3):219-227.
69. Vestronidase alfa-vjbc In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated May 20, 2019.
70. Micromedex DrugDex Compendium®. 2023. Vestronidase Alfa-vjbc.
71. Aldurazyme® (laronidase) injection, for intravenous use [package insert]. BioMarin Pharmaceutical, Inc.; Novato, CA, Revised 03/2023.

72. Micromedex DrugDex Compendium®. 2023. Laronidase.
73. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Laronidase.
74. Dornelles A, Artigas O, Anjos da Silva A, et al. Efficacy and safety of intravenous laronidase for mucopolysaccharidosis type I: A systematic review and meta-analysis. *PLoS ONE*. 2017; 12(8): 1-18.
75. Laronidase In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated December 1, 2003.
76. Jameson E, Jones S, Remington T. Enzyme replacement therapy with laronidase (Aldurazyme®) for treating mucopolysaccharidosis type I. *Cochrane Database of Systematic Reviews*. 2019;6:1-24.
77. Fabrazyme® (agalsidase beta) for injection, for intravenous use. [package insert]. Genzyme Corp. Cambridge, MA. Revised 03/2023.
78. Micromedex DrugDex Compendium®. 2023. Agalsidase beta.
79. Clinical Pharmacology™ Compendia. 2023. Tampa FL: Gold Standard, Inc. Agalsidase beta.
80. Ortiz A, Kanters S, Hamed A, et al. Agalsidase beta treatment slows estimated glomerular filtration rate loss in classic Fabry disease patients: results from an individual patient data meta-analysis. *Clin Kidney J*. 2020;1-11.
81. Agalsidase beta In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated January 1, 2006.
82. Xenpozyme™ (olipudase alfa-rpcp) for injection, for intravenous use. [package insert]. Genzyme Corp. Cambridge, MA. Revised 08/2022.
83. Wasserstein M, Lachmann R, Hollak C, et al. A randomized, placebo-controlled clinical trial evaluating olipudase alfa enzyme replacement therapy for chronic acid sphingomyelinase deficiency (ASMD) in adults: One-year results. *Genet Med*. 2022;24(7):1425-1436.
84. Diaz GA, Jones SA, Scarpa M, Met al. One-year results of a clinical trial of olipudase alfa enzyme replacement therapy in pediatric patients with acid sphingomyelinase deficiency. *Genet Med*. 2021 Aug;23(8):1543-1550.
85. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Generic Drug Olipudase alfa.
86. Olipudase Alfa-rpcp In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated September 12, 2022.
87. Elfabrio® (pegunigalsidase alfa-iwxj) for injection, for intravenous use. [package insert]. Cheisi Inc., Cary NC. Revised 05/2023.
88. Schiffmann R, Goker-Alpan O, Holida M, et al. Pegunigalsidase alfa, a novel PEGylated enzyme replacement therapy for Fabry disease, provides sustained plasma concentrations and favorable pharmacodynamics: A 1-year Phase 1/2 clinical trial. *J Inher Metab Dis*. 2019;42(3):534-544.
89. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Generic Drug Pegunigalsidase alfa.
90. Micromedex DrugDex Compendium®. 2023. Pegunigalsidase Alfa-iwxj.

91. Micromedex DrugDex Compendium®. 2023. CipaglucoSIDase Alfa-atga.
92. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Generic Drug Pegunigalsidase alfa.
93. Pombiliti™ (cipaglucoSIDase alfa-atga) injection for intravenous injection [package insert]. Amicus Therapeutics LLC, Philadelphia, PA. Revised 09/2023.
94. Schoser B, Roberts M, Byrne BJ, et al. Safety and efficacy of cipaglucoSIDase alfa plus miglustat versus alglucoSIDase alfa plus placebo in late-onset Pompe disease (PROPEL): an international, randomised, double-blind, parallel-group, phase 3 trial. *Lancet Neurol*. 2021;20(12):1027-1037.
95. Lamzede® (velmanase alfa-tycv) injection, for intravenous use [package insert]. Cheisi USA, Inc., Cary, NC. Revised 02/2023.
96. Harmatz P, Cattaneo F, Ardigò D, et al. Enzyme replacement therapy with velmanase alfa (human recombinant alpha-mannosidase): Novel global treatment response model and outcomes in patients with alpha-mannosidosis. *Mol Genet Metab*. 2018;124(2):152-160.
97. 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.
98. Micromedex DrugDex Compendium®. 2023. Velmanase Alfa-tycv.
99. Clinical Pharmacology™ Compendium. 2023. Tampa FL: Gold Standard, Inc. Velmanase Alfa-tycv.
100. Velmanase Alfa In: AHFS Drug Information Online Electronic Medical Library. Bethesda, MD: American Society of Health-System Pharmacists. Updated March 27, 2023.