

REFERENCES

1. Pu J, Guardia CM, Keren-Kaplan T, Bonifacino JS. Mechanisms and functions of lysosome positioning. *J Cell Sci*. 2016;129(23):4329-4339.
2. Platt FM, Boland B, van der Spoel AC. The cell biology of disease: lysosomal storage disorders: the cellular impact of lysosomal dysfunction. *J Cell Biol*. 2012;199(5):723-734.
3. Wappner RS. Biochemical diagnosis of genetic diseases. *Pediatr Ann*. 1993;22(5):282-292, 295-287. doi: 10.3928/0090-4481-19930501-08.
4. Kingma SD, Bodamer OA, Wijburg FA. Epidemiology and diagnosis of lysosomal storage disorders; challenges of screening. *Best Pract Res Clin Endocrinol Metab*. 2015;29(2):145-157.
5. Scott CR, Elliott S, Buroker N, et al. Identification of infants at risk for developing Fabry, Pompe, or mucopolysaccharidosis-I from newborn blood spots by tandem mass spectrometry. *J Pediatr*. 2013;163(2):498-503.
6. Meikle PJ, Hopwood JJ, Clague AE, Carey WF. Prevalence of lysosomal storage disorders. *JAMA*. 1999;281(3):249-254.
7. National Organization for Rare Disorders Rare Disease Database. Mucopolysaccharidosis Type I. Available at <https://rarediseases.org/rare-diseases/mucopolysaccharidosis-type-i/>. Accessed March 19, 2021.
8. Imrie J. A guide to ASMD Niemann-Pick disease types A and B. 2010. Available at <https://nnpdf.org/files/2015/10/A-Guide-to-ASMD-Niemann-Pick-Disease-Types-A-and-B.pdf>. Accessed March 19, 2021.
9. Peters H, Ellaway C, Nicholls K, Reardon K, Szer J. Treatable lysosomal storage diseases in the advent of disease-specific therapy. *Intern Med J*. 2020;50 Suppl 4:5-27. doi.org/10.1111/imj.15100
10. Mehta A, Ricci R, Widmer U, et al. Fabry disease defined: baseline clinical manifestations of 366 patients in the Fabry Outcome Survey. *Eur J Clin Invest*. 2004;34(3):236-242.
11. Wenger DA, Coppola S, Liu SL. Insights into the diagnosis and treatment of lysosomal storage diseases. *Arch Neurol*. 2003;60(3):322-328.
12. Staretz-Chacham O, Lang TC, LaMarca ME, Krasnewich D, Sidransky E. Lysosomal storage disorders in the newborn. *Pediatrics*. 2009;123(4):1191-1207.
13. Marsden D, Levy H. Newborn screening of lysosomal storage disorders. *Clin Chem*. 2010;56(7):1071-1079.
14. Parenti G, Andria G, Ballabio A. Lysosomal storage diseases: from pathophysiology to therapy. *Annu Rev Med*. 2015;66:471-486.
15. Parenti G, Andria G, Valenzano KJ. Pharmacological chaperone therapy: Preclinical development, clinical translation, and prospects for the treatment of lysosomal storage disorders. *Mol Ther*. 2015;23(7):1138-1148.
16. Bley AE, Giannikopoulos OA, Hayden D, Kubilus K, Tiftt CJ, Eichler FS. Natural history of infantile G(M2) gangliosidosis. *Pediatrics*. 2011;128(5):e1233-1241.
17. McGovern MM, Dionisi-Vici C, Giugliani R, et al. Consensus recommendation for a diagnostic guideline for acid sphingomyelinase deficiency. *Genet Med*. 2017;19(9):967-974.
18. Beck M. Treatment strategies for lysosomal storage disorders. *Dev Med Child Neurol*. 2018;60(1):13-18.
19. Ballabio A, Gieselmann V. Lysosomal disorders: from storage to cellular damage. *Biochim Biophys Acta*. 2009;1793(4):684-696.
20. Cimaz R, Coppa GV, Kone-Paut I, et al. Joint contractures in the absence of inflammation may indicate mucopolysaccharidosis. *Pediatr Rheumatol Online J*. 2009;7:18.
21. Bruni S, Lavery C, Broomfield A. The diagnostic journey of patients with mucopolysaccharidosis I: A real-world survey of patient and physician experiences. *Mol Genet Metab Rep*. 2016;8:67-73.
22. Galimberti C, Madeo A, Di Rocco M, Fiumara A. Mucopolysaccharidoses: early diagnostic signs in infants and children. *Ital J Pediatr*. 2018;44(Suppl 2):133.
23. Clarke LA, Atherton AM, Burton BK, et al. Mucopolysaccharidosis type I newborn screening: Best practices for diagnosis and management. *J Pediatr*. 2017;182:363-370.
24. Patino-Escobar B, Solano MH, Zarabanda L, Casas CP, Castro C. Niemann-Pick disease: An approach for diagnosis in adulthood. *Cureus*. 2019;11(5):e4767.



AN INTERACTIVE GUIDE
TO UNDERDIAGNOSED AND
UNDERESTIMATED BUT TREATABLE
METABOLIC DISORDERS

25. Sanofi : Positive topline results demonstrated by olipudase alfa, first and only investigational therapy in late-stage development for acid sphingomyelinase deficiency. Available at <https://www.sanofi.com/en/media-room/press-releases/2020/2020-01-30-07-00-00>. Accessed March 19, 2021.
26. Fabry Disease Testing Algorithm. Available at https://www.mayocliniclabs.com/it-mmfiles/Fabry_Disease_Testing_Algorithm.pdf. Accessed March 19, 2021.
27. Zar-Kessler C, Karaa A, Sims KB, Clarke V, Kuo B. Understanding the gastrointestinal manifestations of Fabry disease: promoting prompt diagnosis. *Therap Adv Gastroenterol*. 2016;9(4):626-634.
28. Bekri S, Enica A, Ghafari T, et al. Fabry disease in patients with end-stage renal failure: the potential benefits of screening. *Nephron Clin Pract*. 2005;101(1):c33-38.
29. Mehta A, Hughes DA. GeneReviews. Pagon RA, et al, eds. 1993 University of Washington. Lidove O, et al. In: Mehta A, et al. Oxford PharmaGenesis; 2006. Chapter 24. Available from: www.ncbi.nlm.nih.gov/books/NBK11605/. Accessed October 4, 2020.
30. Patil RB, Joglekar VK. Teenager male with burning pain in extremities--suspect Fabry disease, 2 case reports. *J Assoc Physicians India*. 2014;62(1):69-71.
31. Di Rocco M, Andria G, Deodato F, Giona F, Micalizzi C, Pession A. Early diagnosis of Gaucher disease in pediatric patients: proposal for a diagnostic algorithm. *Pediatr Blood Cancer*. 2014;61(11):1905-1909.
32. Mistry PK, Cappellini MD, Lukina E, et al. A reappraisal of Gaucher disease-diagnosis and disease management algorithms. *Am J Hematol*. 2011;86(1):110-115.
33. Linari S, Castaman G. Clinical manifestations and management of Gaucher disease. *Clin Cases Miner Bone Metab*. 2015;12(2):157-164.
34. Ortega-Rosales A, Burneo-Rosales C, Romero-Ulloa G, Burneo-Rosales G. Case Report: Pancytopenia as an indicator for lysosomal storage disease (Gaucher's Disease) [version 2; peer review: 1 approved, 1 approved with reservations]. *F1000Research* 2019, 8:755.
35. American College of Medical Genetics and Genomics. Alpha-glucosidase deficiency (Pompe disease). Available at <https://www.ncbi.nlm.nih.gov/books/NBK55827/bin/Pompe-Algorithm.pdf>. Accessed March 20, 2021. .
36. Disease AWGoMoP, Kishnani PS, Steiner RD, et al. Pompe disease diagnosis and management guideline. *Genet Med*. 2006;8(5):267-288.
37. Dasouki M, Jawdat O, Almadhoun O, et al. Pompe disease: literature review and case series. *Neurol Clin*. 2014;32(3):751-776, ix.
38. Toscano A, Montagnese F, Musumeci O. Early is better? A new algorithm for early diagnosis in late onset Pompe disease (LOPD). *Acta Myol*. 2013;32(2):78-81.
39. Baby's first test: Conditions by state. Available at <https://www.babysfirsttest.org/newborn-screening/rusp-conditions#mucopolysaccharidosis-type-i>. Accessed March 20, 2021.
40. Parenti G, Medina DL, Ballabio A. The rapidly evolving view of lysosomal storage diseases. *EMBO Mol Med*. 2021;13(2):e12836.
41. Benjamin ER, Della Valle MC, Wu X, et al. The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. *Genet Med*. 2017;19(4):430-438.
42. Newborn Screening ACT Sheets and Algorithms. 2001. <https://www.ncbi.nlm.nih.gov/books/NBK55827/>
43. Kuperus E, Kruijshaar ME, Wens SCA, et al. Long-term benefit of enzyme replacement therapy in Pompe disease: A 5-year prospective study. *Neurology*. 2017;89(23):2365-2373.
44. de Vries JM, van der Beek NA, Hop WC, et al. Effect of enzyme therapy and prognostic factors in 69 adults with Pompe disease: an open-label single-center study. *Orphanet J Rare Dis*. 2012;7:73.
45. Güngör D, Kruijshaar ME, Plug I, et al. Impact of enzyme replacement therapy on survival in adults with Pompe disease: results from a prospective international observational study. *Orphanet J Rare Dis*. 2013;8:49.
46. Toscano A, Schoser B. Enzyme replacement therapy in late-onset Pompe disease: a systematic literature review. *J Neurol*. 2013;260(4):951-959.
47. Amalfitano A, Bengur AR, Morse RP, et al. Recombinant human acid alpha-glucosidase enzyme therapy for infantile glycogen storage disease type II: results of a phase I/II clinical trial. *Genet Med*. 2001;3(2):132-138.



AN INTERACTIVE GUIDE
TO UNDERDIAGNOSED AND
UNDERESTIMATED BUT TREATABLE
METABOLIC DISORDERS

48. Kishnani PS, Corzo D, Nicolino M, et al. Recombinant human acid [alpha]-glucosidase: major clinical benefits in infantile-onset Pompe disease. *Neurology*. 2007;68(2):99-109.
49. Klinge L, Straub V, Neudorf U, et al. Safety and efficacy of recombinant acid alpha-glucosidase (rhGAA) in patients with classical infantile Pompe disease: results of a phase II clinical trial. *Neuromuscul Disord*. 2005;15(1):24-31.
50. van der Ploeg AT, Kruijshaar ME, Toscano A, et al. European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10-year experience. *Eur J Neurol*. 2017;24(6):768-e731.
51. Imiglucerase [package insert]. Cambridge, MA. Genzyme Corporation. 2020.
52. Velaglucerase [package insert]. Shire Human Genetic Therapies, Inc. Lexington, MA. 2021.
53. Taliglucerase alfa [package insert]. Pfizer, Inc. New York, NY. 2020.
54. El-Beshlawy A, Tylki-Szymanska A, Vellodi A, et al. Long-term hematological, visceral, and growth outcomes in children with Gaucher disease type 3 treated with imiglucerase in the International Collaborative Gaucher Group Gaucher Registry. *Mol Genet Metab*. 2017;120(1-2):47-56.
55. Agalsidase beta [package insert]. Genzyme Corp. Cambridge, MA. 2021.
56. Ortiz A, Abiose A, Bichet DG, et al. Time to treatment benefit for adult patients with Fabry disease receiving agalsidase beta: data from the Fabry Registry. *J Med Genet*. 2016;53(7):495-502.
57. Hopkin RJ, Jefferies JL, Laney DA, et al. The management and treatment of children with Fabry disease: A United States-based perspective. *Mol Genet Metab*. 2016;117(2):104-113.
58. Laronidase [package insert]. Cambridge, MA. Genzyme Corporation. 2019.
59. Felis A, Whitlow M, Kraus A, Warnock DG, Wallace E. Current and investigational therapeutics for Fabry disease. *Kidney Int Rep*. 2020;5(4):407-413.
60. Beck M, Whybra C, Wendrich K, Gal A, Ries M. Anderson-Fabry disease in children and adolescents. *Contrib Nephrol*. 2001(136):251-255.
61. Ortiz A, Germain DP, Desnick RJ, et al. Fabry disease revisited: Management and treatment recommendations for adult patients. *Mol Genet Metab*. 2018;123(4):416-427.
62. Migalastat [package insert]. Amicus Therapeutics, LLC. Philadelphia, PA. 2021.
63. Hughes DA, Nicholls K, Shankar SP, et al. Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. *J Med Genet*. 2017;54(4):288-296.
64. Chimenti C, Nencini P, Pieruzzi F, et al. The GALA project: practical recommendations for the use of migalastat in clinical practice on the basis of a structured survey among Italian experts. *Orphanet J Rare Dis*. 2020;15(1):86.
65. Belmatoug N, Di Rocco M, Fraga C, et al. Management and monitoring recommendations for the use of eliglustat in adults with type 1 Gaucher disease in Europe. *Eur J Intern Med*. 2017;37:25-32.
66. Cox TM, Amato D, Hollak CE, et al. Evaluation of miglustat as maintenance therapy after enzyme therapy in adults with stable type 1 Gaucher disease: a prospective, open-label non-inferiority study. *Orphanet J Rare Dis*. 2012;7:102.
67. Pastores GM, Elstein D, Hrebicek M, Zimran A. Effect of miglustat on bone disease in adults with type 1 Gaucher disease: a pooled analysis of three multinational, open-label studies. *Clin Ther*. 2007;29(8):1645-1654.
68. Mistry PK, Lukina E, Ben Turkia H, et al. Effect of oral eliglustat on splenomegaly in patients with Gaucher disease type 1: the ENGAGE randomized clinical trial. *JAMA*. 2015;313(7):695-706.
69. Cox TM, Drelichman G, Cravo R, et al. Eliglustat compared with imiglucerase in patients with Gaucher's disease type 1 stabilised on enzyme replacement therapy: a phase 3, randomised, open-label, non-inferiority trial. *Lancet*. 2015;385(9985):2355-2362.
70. Eliglustat [package insert]. Genzyme Corporation. Cambridge, MA. 2018.
71. Estrada-Veras JI, Cabrera-Pena GA, Perez Estrella de Ferran C. Medical genetics and genomic medicine in the Dominican Republic: Challenges and opportunities. *Mol Gen Gen Med*. 2016;4:243-256. [doi:10.1002/mgg3.224](https://doi.org/10.1002/mgg3.224)
72. Muenzer J. Early initiation of enzyme replacement therapy for the mucopolysaccharidoses. *Mol Genet Metab*. 2014;111(2):63-72.



AN INTERACTIVE GUIDE
TO **UNDERDIAGNOSED** AND
UNDERESTIMATED BUT **TREATABLE**
METABOLIC DISORDERS

73. Simpson WL, Jr, Mendelson D, Wasserstein MP, McGovern MM. Imaging manifestations of Niemann-Pick disease type B. *AJR Am J Roentgenol*. 2010;194(1):W12-19.
74. Florida Newborn Screening. Available at <https://floridanewbornscreening.com/conditions/core-secondary-conditions/>. Accessed March 31, 2021.