

DIAGNOSING RARE DISEASES BRINGS
NEW TREATMENTS TO PATIENTS

EDUCATION FOR
ADVANCED CARE PRACTITIONERS

RESOURCES

Generic diagnostic algorithms for recognizing LSDs

Staretz-Chacham O, Lang TC, LaMarca ME, et al. Lysosomal storage disorders in the newborn. *Pediatrics* 2009;123:1191-1207. [\[LINK\]](#)

Diagnostic algorithms for individual disorders

Fabry disease diagnostic algorithm

Mayo Clinic Laboratories [\[LINK\]](#)

Gaucher disease 1 diagnostic algorithms

2011 testing and treatment consensus conference: Mistry PK, Cappellini MD, Lukina E, et al. A reappraisal of Gaucher disease-diagnosis and disease management algorithms. *Am J Hematol*. 2011;86:110-5. [\[LINK\]](#)

Pediatric patients: Di Rocco M, Andria G, Deodato F, Gilona F, Micalizzi C, Pession A. Early diagnosis of Gaucher disease in pediatric patients: Proposal for a diagnostic algorithm. *Pediatr Blood Cancer*. 2014;61:1905-1909.

[\[LINK\]](#)

Mucopolysaccharidosis I diagnostic algorithm

Cimaz R, Coppa GV, Koné-Paut I, et al. Joint contractures in the absence of inflammation may indicate mucopolysaccharidosis. *Pediatr Rheumatol*. 2009;7:18. [\[LINK\]](#)

Niemann-Pick disease diagnostic algorithms

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. doi:10.1038/gim.2017.7 [\[LINK\]](#)

Pompe disease diagnostic algorithms

Alpha-glucosidase deficiency (Pompe disease). American College of Medical Genetic and Genomics:

[\[DOWNLOAD\]](#)

Infantile and late-onset Pompe disease: Kishnani PS, Steiner RD, Bali D, et al. Pompe disease diagnosis and management guideline. *Genet Med*. 2006;8(5):267-288. [\[LINK\]](#)

Key publications for LSD treatments

[α-galactosidase A](#): Eng CM, Guffon N, Wilcox WR, et al. Safety and efficacy of recombinant human α-galactosidase A replacement therapy in Fabry's disease. *N Engl J Med*. 2001;345(1):9-16. doi:10.1056/NEJM200107053450102 [\[LINK\]](#)

[α-galactosidase A](#): Schiffmann R, Kopp JB, Austin HA III, et al. Enzyme replacement therapy in Fabry disease: a randomized controlled trial. *JAMA*. 2001;285(21):2743-2749. doi:10.1001/jama.285.21.2743 [\[LINK\]](#)

[Alglucosidase](#): Nicolino M, Byrne B, Wraith J, et al. Clinical outcomes after long-term treatment with alglucosidase alfa in infants and children with advanced Pompe disease. *Genet Med*. 11, 210–219 (2009). <https://doi.org/10.1097/GIM.0b013e31819d0996> [\[LINK\]](#)

[Alglucerase, imiglucerase, miglustat reviewed](#): Martins AM, Valadares ER, Porta G, et al. Recommendations on diagnosis, treatment, and monitoring for Gaucher disease. *J Pediatr*. 2009;155(4 Suppl):S10-S18. doi:10.1016/j.jpeds.2009.07.004 [\[LINK\]](#)

Elosulfase alfa: Hendriksz CJ, Burton B, Fleming TR, et al. Efficacy and safety of enzyme replacement therapy with BMN 110 (elosulfase alfa) for Morquio A syndrome (mucopolysaccharidosis IVA): a phase 3 randomised placebo-controlled study. *J Inherit Metab Dis*. 2014;37(6):979-990. doi:10.1007/s10545-014-9715-6 [\[LINK\]](#)

Galsulfase: Harmatz PR, Garcia P, Guffon N, et al. Galsulfase (Naglazyme®) therapy in infants with mucopolysaccharidosis VI. *J Inherit Metab Dis*. 2014;37(2):277-287. doi:10.1007/s10545-013-9654-7 [\[LINK\]](#)

Idursulfase: Muenzer J, Beck M, Eng CM, et al. Long-term, open-labeled extension study of idursulfase in the treatment of Hunter syndrome. *Genet Med*. 2011;13(2):95-101. doi:10.1097/GIM.0b013e3181fea459 [\[LINK\]](#)

Laronidase: Wraith JE. The first 5 years of clinical experience with laronidase enzyme replacement therapy for mucopolysaccharidosis I. *Expert Opin Pharmacother*. 2005;6(3):489-506. doi:10.1517/14656566.6.3.489 [\[LINK\]](#)

Migalastat: Germain DP, Hughes DA, Nicholls K, et al. Treatment of Fabry's Disease with the pharmacologic chaperone migalastat. *N Engl J Med*. 2016;375(6):545-555. doi:10.1056/NEJMoa1510198 [\[LINK\]](#)

Taliglucerase alfa: Zimran A, Brill-Almon E, Chertkoff R, et al. Pivotal trial with plant cell-expressed recombinant glucocerebrosidase, taliglucerase alfa, a novel enzyme replacement therapy for Gaucher disease [published correction appears in *Blood*. 2012 May 10;119(19):4577]. *Blood*. 2011;118(22):5767-5773. doi:10.1182/blood-2011-07-366955 [\[LINK\]](#)

Review of treatments for MPS disorders: Parini R, Deodato F. Intravenous enzyme replacement therapy in mucopolysaccharidoses: Clinical effectiveness and limitations. *Int J Mol Sci*. 2020, 21, 2975. [\[LINK\]](#)

Sebelipase alfa: Burton BK, Balwani M, Feillet F, et al. A phase 3 trial of sebelipase alfa in lysosomal acid lipase deficiency. *N Engl J Med*. 2015;373(11):1010-1020. doi:10.1056/NEJMoa1501365. [\[LINK\]](#)

Velaglucerase alfa: Gonzalez DE, Turkia HB, Lukina EA, et al. Enzyme replacement therapy with velaglucerase alfa in Gaucher disease: Results from a randomized, double-blind, multinational, phase 3 study. *Am J Hematol*. 2013;88(3):166-171. doi:10.1002/ajh.23381 [\[LINK\]](#)