Suggested Readings

Unmet Needs Among Individuals With PD

Banugaria SG, Patel TT, Mackey J, et al. Persistence of high sustained antibodies to enzyme replacement therapy despite extensive immunomodulatory therapy in an infant with Pompe disease: need for agents to target antibody-secreting plasma cells. *Mol Genet Metab*. 2012;105:677-680. doi:10.1016/j.ymgme.2012.01.019

Burton BK, Hickey R, Hitchins L. Newborn screening for mucopolysaccharidosis type II in Illinois: an update. *Int J Neonatal Screen*. 2020;6:73. doi:10.3390/ijns6030073

Butterworth J, Droadhead DM. Diagnosis of Pompe's disease in cultured skin fibroblasts and primary amniotic fluid cells using 4-methylumbelliferyl-alpha-D-glucopyranoside as substrate. *Clin Chim Acta*. 1977;78:335-342. doi:10.1016/0009-8981(77)90325-4

Chien Y-H, Chiang S-C, Zhang XK, et al. Early detection of Pompe disease by newborn screening is feasible: results from the Taiwan screening program. *Pediatrics*. 2008;122:e39-e45. doi:10.1542/peds.2007-2222

Chien Y-H, et al. Newborn screening: Taiwanese experience. *Ann Transl Med*. 2019;7:281. doi:10.21037/atm.2019.05.47

de Filippi P, Ravaglia S, Bembi B, et al. The angiotensin-converting enzyme insertion/deletion polymorphism modifies the clinical outcome in patients with Pompe disease. *Genet Med*. 2010;12:206-211. doi:10.1097/GIM.0b013e3181d2900e

Ficicioglu C, Ahrens-Nicklas RC, Barch J, et al. Newborn screening for Pompe disease: Pennsylvania experience. *Int J Neonatal Screen*. 2020;6:89. doi:10.3390/ijns6040089

Fukuda T, Ewan L, Bauer M, et al. Dysfunction of endocytic and autophagic pathways in a lysosomal storage disease. *Ann Neurol*. 2006;59:700-708. doi:10.1002/ana.20807

Güngör D, de Vries JM, Hop WCJ, et al. Survival and associated factors in 268 adults with Pompe disease prior to treatment with enzyme replacement therapy. *Orphanet J Rare Dis*. 2011;6:34. doi:10.1186/1750-1172-6-34

Hagemans MLC, Winkel LPF, Van Doorn PA, et al. Clinical manifestation and natural course of late-onset Pompe's disease in 54 Dutch patients. *Brain*. 2005;128:671-677. doi:10.1093/brain/awh384

Hagemans MLC, Winkel LPF, Hop WCJ, et al. Disease severity in children and adults with Pompe disease related to age and disease duration. *Neurology*. 2005;64:2139-2141. doi:10.1212/01.WNL.0000165979.46537.56

Hamdan MA, El-Zoabi BA, Begam MA, et al. Antenatal diagnosis of Pompe disease by fetal echocardiography: impact on outcome after early initiation of enzyme replacement therapy. *J Inherit Metab Dis*. 2010;33(suppl 3):S333-S339. doi:10.1007/s10545-010-9179-2

Hopkins PV, Klug T, Vermette L, et al. Incidence of 4 lysosomal storage disorders from 4 years of newborn screening. *JAMA Pediatr*. 2018;172:696-697. doi:10.1001/jamapediatrics.2018.0263

Huggins E, Holland M, Case LE, et al. Early clinical phenotype of late onset Pompe disease: lessons learned from newborn screening. *Mol Genet Metab*. 2022;135:179-185. doi:10.1016/j.ymgme.2022.01.003

Kishnani PS, Howell RR. Pompe disease in infants and children. *J Pediatr*. 2004;144(5 suppl):S35-S43. doi:10.1016/j.jpeds.2004.01.053

Kishnani PS, Hwu W-L, Mandel H, et al. A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. *J Pediatr*. 2006;148:671-676. doi:10.1016/j.jpeds.2005.11.033

Kishnani PS, Steiner RD, Bali D, et al. Pompe disease diagnosis and management guideline. *Genet Med*. 2006;8:267-288. doi:10.1097/01.gim.0000218152.87434.f3

Kishnani PS, Goldenberg PC, DeArmey SL, et al. Cross-reactive immunologic material status affects treatment outcomes in Pompe disease infants. *Mol Genet Metab*. 2010;99:26-33. doi:10.1016/j.ymgme.2009.08.003

Kishnani PS, Beckemeyer AA, Mendelsohn NJ. The new era of Pompe disease: advances in the detection, understanding of the phenotypic spectrum, pathophysiology, and management. *Am J Med Genet Part C Semin Med Genet*. 2012;160C:1-7. doi:10.1002/ajmg.c.31324

Labrousse P, Chien Y-H, Pomponio RJ, et al. Genetic heterozygosity and pseudodeficiency in the Pompe disease newborn screening pilot program. *Mol Genet Metab*. 2010;99:379-383. doi:10.1016/j.ymgme.2009.12.014

Lee N-C, Chang K-L, In ‘t Groen SLM, et al. Outcome of later-onset Pompe disease identified through newborn screening. *J Pediatr*. 2022;244:139-147.e2. doi:10.1016/j.jpeds.2021.12.072

Leslie N, Bailey L. Pompe disease. In: Adam MP, Everman DB, Mirzaa GM, et al, eds. *GeneReviews*®. University of Washington, Seattle; 1993-2023. Accessed February 17, 2023. https://www.ncbi.nlm.nih.gov/books/NBK1261

Lim J-A, Li L, Raben N. Pompe disease: from pathophysiology to therapy and back again. *Front Aging Neurosci*. 2014;6:177. doi:10.3389/fnagi.2014.00177

Martiniuk F, Chen A, Mack A, et al. Carrier frequency for glycogen storage disease type II in New York and estimates of affected individuals born with the disease. *Am J Med Genet*. 1998;79:69-72. doi:10.1002/(SICI)1096-8628(19980827)79:1<69: :AID-AJMG16>3.0.CO;2-K

Peruzzo P, Pavan E, Dardis A. Molecular genetics of Pompe disease: a comprehensive overview. *Ann Transl Med*. 2019;7:278. doi:10.21037/atm.2019.04.13

Pinto R, Caseiro C, Lemos M, et al. Prevalence of lysosomal storage diseases in Portugal. *Eur J Hum Genet*. 2004;12:87-92. doi:10.1038/sj.ejhg.5201044

Prater SN, Patel TT, Buckley AF, et al. Skeletal muscle pathology of infantile Pompe disease during long-term enzyme replacement therapy. *Orphanet J Rare Dis*. 2013;8:90. doi:10.1186/1750-1172-8-90

Raben N, Danon M, Gilbert AL, et al. Enzyme replacement therapy in the mouse model of Pompe disease. *Mol Genet Metab*. 2003;80:159-169. doi:10.1016/j.ymgme.2003.08.022

Raben N, Fukuda T, Gilbert AL, et al. Replacing acid alpha-glucosidase in Pompe disease: recombinant and transgenic enzymes are equipotent, but neither completely clears glycogen from type II muscle fibers. *Mol Ther*. 2005;11:48-56. doi:10.1016/j.ymthe.2004.09.017

Raben N, Roberts A, Poltz PH. Role of autophagy in the pathogenesis of Pompe disease. *Acta Myol*. 2007;26:45-48.

Raben N, Wong A, Ralston E, et al. Autophagy and mitochondria in Pompe disease: nothing is so new as what has long been forgotten. *Am J Med Genet C Semin Med Genet*. 2012;160C:13-21. doi:10.1002/ajmg.c.31317

Reuser AJJ, van der Ploeg AT, Chien Y-H, et al. GAA variants and phenotypes among 1,079 patients with Pompe disease: data from the Pompe Registry. *Hum Mutat*. 2019;40:2146-2164. doi:10.1002/humu.23878

Sawada T, Kido J, Nakamura K. Newborn screening for Pompe disease. *Int J Neonatal Screen*. 2020;6:31. doi:10.3390/ijns6020031

Schoser B. Pompe disease: what are we missing? *Ann Transl Med*. 2019;7:292. doi:10.21037/atm.2019.05.29

Slonim AE, Bulone L, Ritz S, et al. Identification of two subtypes of infantile acid maltase deficiency. *J Pediatr*. 2000;137:283-285. doi:10.1067/mpd.2000.107112

Thurberg BL, Maloney CL, Vaccaro C, et al. Characterization of pre- and post-treatment pathology after enzyme replacement therapy for Pompe disease. *Lab Invest*. 2006;86:1208-1220. doi:10.1038/labinvest.3700484

van der Ploeg AT, Barohn R, Carlson L, et al. Open-label extension study following the Late-Onset Treatment Study (LOTS) of alglucosidase alfa. *Mol Genet Metab*. 2012;107:456-461. doi:10.1016/j.ymgme.2012.09.015

Yang C-F, Yang CC, Liao H-C, et al. Very early treatment for infantile-onset Pompe disease contributes to better outcomes. *J Pediatr*. 2016;169:174-180.e1. doi:10.1016/j.jpeds.2015.10.078

Bhengu L, Davidson A, du Toit P, et al. Diagnosis and management of Pompe disease. *S Afr Med J*. 2014;104:273-274. doi:10.7196/samj.7386

Diaz-Manera J, Kishnani PS, Kushlaf H, et al. Safety and efficacy of avalglucosidase alfa versus alglucosidase alfa in patients with late-onset Pompe disease (COMET): a phase 3, randomised, multicentre trial. *Lancet Neurol*. 2021;20:1012-1026. doi:10.1016/S1474-4422(21)00241-6

Do HV, Khana R, Gotschall R. Challenges in treating Pompe disease: an industry perspective. *Ann Transl Med*. 2019;7:291. doi:10.21037/atm.2019.04.15

Eggers M, Vannoy CH, Huang J, et al. Muscle-directed gene therapy corrects Pompe disease and uncovers species-specific GAA immunogenicity. *EMBO Mol Med*. 2022;14:e13968. doi:10.15252/emmm.202113968

Godefroy A, Daurat M, Da Silva A, et al. Mannose 6-phosphonate labelling: a key for processing the therapeutic enzyme in Pompe disease. *J Cell Mol Med*. 2019;23:6499-6503. doi:10.1111/jcmm.14516

Gutschmidt K, Musumeci O, Díaz-Manera J, et al. STIG study: real-world data of long-term outcomes of adults with Pompe disease under enzyme replacement therapy with alglucosidase alfa. *J Neurol.* 2021;268:2482-2492. doi:10.1007/s00415-021-10409-9

Kishnani PS, Corzo D, Leslie ND, et al. Early treatment with alglucosidase alpha prolongs long-term survival of infants with Pompe disease. *Pediatr Res*. 2009;66:329-335. doi:10.1203/PDR.0b013e3181b24e94

Kishnani P, Díaz-Manera J, Kushlaf H, et al. The avalglucosidase alfa phase 3 COMET trial in late-onset Pompe disease patients: efficacy and safety results after 97 weeks. Presented at: 18th Annual World*Symposium*; February 7-11, 2022; hybrid. Abstract 153.

Kronn D, Davison J, Brassier A, et al. Mini-COMET study: safety, biomarker, and efficacy data after avalglucosidase alfa dosing for ≥97 weeks in participants with infantile-onset Pompe disease (IOPD) previously
treated with alglucosidase alfa who had demonstrated clinical decline. 18th Annual World*Symposium*; February 7-11, 2022; hybrid. Abstract 156.

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;89:2365-2373. doi:10.1212/WNL.0000000000004711

Nuñez-Peralta C, Alonso-Pérez J, Llauger J, et al. Follow-up of late-onset Pompe disease patients with muscle magnetic resonance imaging reveals increase in fat replacement in skeletal muscles. *J Cachexia Sarcopenia Muscle*. 2020;11:1032-1046. doi:10.1002/jcsm.12555

Patel TT, Banugaria SG, Case LE, et al. The impact of antibodies in late-onset Pompe disease: a case series and literature review. *Mol Genet Metab*. 2012;106:301-309. doi:10.1016/j.ymgme.2012.04.027

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;29:167-186. doi:10.1016/j.nmd.2018.12.004

Puertollano R, Raben N. Pompe disease: how to solve many problems with one solution. *Ann Transl Med*. 2018;6:313. doi:10.21037/atm.2018.06.52

Regnery C, Kornblum C, Hanisch F, et al. 36 months observational clinical study of 38 adult Pompe disease patients under alglucosidase alfa enzyme replacement therapy. *J Inherit Metab Dis*. 2012;35:837-845. doi:10.1007/s10545-012-9451-8

Reuser AJJ, Kroos MA, Ponne NJ, et al. Uptake and stability of human and bovine acid alpha-glucosidase in cultured fibroblasts and skeletal muscle cells from glycogenosis type II patients. *Exp Cell Res*. 1984;155:178-189. doi:10.1016/0014-4827(84)90779-1

Ronzitti G, Collaud F, Laforet P, et al. Progress and challenges of gene therapy for Pompe disease. *Ann Transl Med*. 2019;7:287. doi:10.21037/atm.2019.04.67

Schoser B, Bilder DA, Dimmock D, et al. The humanistic burden of Pompe disease? Are there still unmet needs? A systematic review. *BMC Neurol*. 2017;17:202. doi:10.1186/s12883-017-0983-2

Schoser B, Stewart A, Kanters S, et al. Survival and long-term outcomes in late-onset Pompe disease following alglucosidase alfa treatment: a systematic review and meta-analysis. *J Neurol*. 2017;264:621-630. doi:10.1007/s00415-016-8219-8

Schoser B. Pompe disease: what are we missing? *Ann Transl Med*. 2019;7:292. doi:10.21037/atm.2019.05.29

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;20:1027-1037. doi:10.1016/S1474-4422(21)00331-8

Stepien KM, Hendriksz CJ, Roberts M, et al. Observational clinical study of 22 adult-onset Pompe disease patients undergoing enzyme replacement therapy over 5 years. *Mol Genet Metab*. 2016;117:413-418. doi:10.1016/j.ymgme.2016.01.013

Thurberg BL, Maloney CL, Vaccaro C, et al. Characterization of pre- and post-treatment pathology after enzyme replacement therapy for Pompe disease. *Lab Invest*. 2006;86:1208-1220. doi:10.1038/labinvest.3700484

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;41:1205-1214. doi:10.1007/s10545-018-0166-3

Wu Z, Sun J, Zhang T, et al. Optimization of self-complementary AAV vectors for liver-directed expression results in sustained correction of hemophilia B at low vector dose. *Mol Ther*. 2008;16:280-289. doi:10.1038/sj.mt.6300355

Xu S, Lun Y, Frascella M, et al. Improved efficacy of a next-generation ERT in murine Pompe disease. *JCI Insight*. 2019;4:e125358. doi:10.1172/jci.insight.125358

Zhou Q, Stefano JE, Harrahy J, et al. Strategies for Neoglycan conjugation to human acid α-glucosidase. *Bioconjug Chem*. 2011;22:741-751. doi:10.1021/bc1005416

Zhu Y, Jiang J-L, Gumlaw NK, et al. Glycoengineered acid alpha-glucosidase with improved efficacy at correcting the metabolic aberrations and motor function deficits in a mouse model of Pompe disease. *Mol Ther*. 2009;17:954-963. doi:10.1038/mt.2009.37

Tailoring Therapy to Disease Progression

Chan J, Desai AK, Kazi ZB, et al. The emerging phenotype of late-onset Pompe disease: a systematic literature review. *Mol Genet Metab*. 2017;120:163-172. doi:10.1016/j.ymgme.2016.12.004

Chien Y-H, Hwu W-L, Lee N-C. Pompe disease: early diagnosis and early treatment make a difference. *Pediatr Neonatol*. 2013;54:219-227. doi:10.1016/j.pedneo.2013.03.009

Chien Y-H, Goldstein JL, Hwu W-L, et al. Baseline urinary glucose tetrasaccharide concentrations in patients with infantile- and late-onset Pompe disease identified by newborn screening. *JIMD Rep*. 2015;19:67-73. doi:10.1007/8904_2014_366

Kishnani PS, Steiner RD, Bali D, et al. Pompe disease diagnosis and management guideline. *Genet Med*. 2006;8:267-288. doi:10.1097/01.gim.0000218152.87434.f3

Kishnani PS, Beckemeyer AA. New therapeutic approaches for Pompe disease: enzyme replacement therapy and beyond. *Pediatr Endocrinol Rev.* 2014;12(suppl 1):114-124.

Matern D, Gavrilov D, Oglesbee D, et al. Newborn screening for lysosomal storage disorders. *Semin Perinatol*. 2015;39:206-216. doi:10.1053/j.semperi.2015.03.005

Montagnese F, Barca E, Musumeci O, et al. Clinical and molecular aspects of 30 patients with late-onset Pompe disease (LOPD): unusual features and response to treatment. *J Neurol*. 2015;262:968-978. doi:10.1007/s00415-015-7664-0

Parisi D, Musumeci O, Mondello S, et al. Vacuolated PAS-positive lymphocytes on blood smear: an easy screening tool and a possible biomarker for monitoring therapeutic responses in late onset Pompe disease (LOPD). *Front Neurol*. 2018;9:880. doi:10.3389/fneur.2018.00880

Ruggeri P, Monaco LL, Musumeci O, et al. Ultrasound assessment of diaphragm function in patients with late-onset Pompe disease. *Neurol Sci*. 2020;41:2175-2184. doi:10.1007/s10072-020-04316-6

Toscano A, Rodolico C, Musumeci O. Multisystem late onset Pompe disease (LOPD): an update on clinical aspects. *Ann Transl Med*. 2019;7:284. doi:10.21037/atm.2019.07.24

Young SP, Piraud M, Goldstein JL, et al. Assessing disease severity in Pompe disease: the roles of a urinary glucose tetrasaccharide biomarker and imaging techniques. *Am J Med Genet C Semin Med Genet*. 2012;160C:50-58. doi:10.1002/ajmg.c.31320

van der Beek NAME, Hagemans MLC, Reuser AJJ, et al. Rate of disease progression during long-term follow-up of patients with late-onset Pompe disease. *Neuromuscul Disord*. 2009;19:113-117. doi:10.1016/j.nmd.2008.11.007

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;24:768-e31. doi:10.1111/ene.13285