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Nov 19, 2024 · Pompe disease is a rare genetic disorder marked by the buildup inside cells of the sugar molecule glycogen, which damages organs and tissues. What is Pompe disease? Pompe disease is an inherited (genetic) condition that prevents the body from processing sugars properly. Pompe disease is named for the first doctor to describe the condition. ... Apr 13, 2026 · In the movie "Extraordinary Measures," Dr. Stonehill invented a treatment for Pompe disease. This is the true story of how John Crowley helped save his children's lives. Sep 27, 2023 · Pompe disease is a rare genetic condition that causes muscle weakness that can lead to life-threatening complications. Learn more about the symptoms and treatment of this disorder. Pompe Disease is a lysosomal disease caused by changes in

the acid alpha-glucosidase (GAA) gene. It is an autosomal recessive inheritance, meaning both parents are genetic carriers of the disease but ... Pompe's disease, hereditary defect in the body's ability to metabolize glycogen, resulting in a muscle disorder that is usually fatal during the first year of life. The defect responsible, absence of the ... Jan 18, 2024 · Learn about Pompe Disease, including symptoms, causes, and treatments. If you or a loved one is affected by this condition, visit NORD to find resources and Glycogen storage disease type II (GSD-II), also called Pompe disease, and formerly known as GSD-IIa or Limb-girdle muscular dystrophy 2V, is an autosomal recessive metabolic disorder [1] which ... Diagnosis of Pompe Disease Evaluation and Management of Infants With a Positive NBS for Pompe

Disease1-3 Developed with the expertise of Barbara K. Burton, MD, Northwestern University ... Pompe disease is characterized by a single enzyme deficiency, acid alpha-glucosidase (GAA), resulting in progressive glycogen accumulation primarily in the lysosomes of muscle cells1 Signs and ...

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