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Review on Pompe Disease

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Abstract: *Pompe disease, also known as glycogen storage disease type II, is a rare genetic disorder caused by the deficiency of the enzyme acid alpha-glucosidase. This enzyme is essential for breaking down glycogen, a stored form of glucose, into glucose that the body can use for energy. In individuals with Pompe disease, the lack of this enzyme leads to the accumulation of glycogen in various tissues, particularly in the heart, skeletal muscles, and liver.The disease can manifest in different forms, primarily categorized into infantile-onset and late-onset types. Infantile-onset Pompe disease typically presents within the first few months of life, characterized by severe muscle weakness, respiratory difficulties, and heart enlargement (cardiomyopathy). If left untreated, it can lead to significant morbidity and mortality within the first year of life. Late-onset Pompe disease, which can occur from childhood to adulthood, tends to have a milder course, with symptoms including progressive muscle weakness and respiratory issues, but typically without the severe cardiac involvement seen in the infantile form.Diagnosis of Pompe disease is usually confirmed through enzyme assay testing and genetic testing to identify mutations in the GAA gene responsible for the enzyme deficiency. Treatment options include enzyme replacement therapy (ERT) with alglucosidase alfa, which can help improve muscle function and overall health in affected individuals. Early diagnosis and intervention are crucial for better outcomes in managing Pompe disease. Pompe disease is a serious genetic condition that affects glycogen metabolism, leading to significant health challenges, particularly in muscle and heart function, but can be managed with appropriate treatment strategies.*

Keywords: Pompe disease; GSD-II; GAA deficiency; Lysosomal disorder; Infantile-onset; Late-onset; Enzyme replacement therapy; Alglucosidase alfa; Gene therapy; AAV vectors; CRISPR; Glycogen accumulation; Muscle weakness; Cardiomyopathy; New treatments

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