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Review

Pompe Disease: A Review of the Article

Ian Caldeira Ruppen[1]*, Haloany Maola Chitolina[1], Lara Beatriz Dallaqua Bitiati[1], Clovis Cavalcanti de Albuquerque Neto[2], Rafaela Beatriz Siqueira[1], Leandro Hideki Otani[3], Felicia Satie Ibuki Otani[3], Gabriel Petermann[4], Maria Clara Palácio Fachina[1], Maria Eduarda Dias Stuani[1], Paschoal Carlos Figueiredo Morelli[1], Mariane Zancanaro Gallina[1], Patrícia de Vilhena Pimenta Neves[1], Marianna de Vilhena Pimenta Neves[1], Geórgia Verona Cruz[3], Júlia Alvares Dal' lago[5], Amanda Larissa Zotarelli Pasquali[1], Isabela Viana Veronez[1], Alana Reigota da Costa Rosa[1], José Henrique Damas Garcia[1], Maria Vitória Mendonça[1] and Luana Padovani[1]

[1]Centro Universitário Ingá - Uningá, Maringá, Paraná, Brazil

[2]Faculdade Cesumar - Unicesumar Maringá, Paraná, Brazil

[3]Instituto Maringá de Imagem, Maringá, PR, Brazil

[4]Universidade Anhanguera Uniderp, Brazil

[5]Ulbra- Universidade Luterana do Brasil, Brazil

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***Corresponding author:** Ian Caldeira Ruppen, Centro Universitário Ingá - Uningá, Brazil, E-mail: Ian2ruppen@gmail.com

ABSTRACT

Pompe Disease or glycogen storage disease type II, is a rare genetic disorder caused by a deficiency of the enzyme acid alpha-glucosidase (GAA), which is responsible for glycogen degradation in lysosomes. The accumulation of glycogen in tissues, particularly cardiac and skeletal muscles, leads to varied clinical manifestations, ranging from severe infantile forms to late-onset variants. Treatment includes enzyme replacement therapy (ERT), which improves patient survival and quality of life. However, challenges persist, such as immune responses to ERT, limited efficacy in different tissues and difficulties in early diagnosis. This article reviews clinical, diagnostic and therapeutic aspects of Pompe Disease based on recent literature.

Keywords: Glycogen; Enzymes; Immunological; Acid Alpha-Glucosidase

Introduction

Pompe Disease, first described in 1932 by Dutch pathologist Johannes Pompe, is a lysosomal glycogen storage disorder with autosomal recessive inheritance. Its prevalence ranges from 1:40,000 to 1:300,000 live births[1], depending on the population[2]. The disease manifests in two main clinical spectrums: the classic infantile form, characterized by cardiomegaly and early respiratory insufficiency and the late-onset form (LOPD), which predominantly affects skeletal muscles, leading to progressive weakness[3]. Diagnosing Pompe Disease is often challenging due to its clinical heterogeneity[4]. Advances such as enzyme activity testing via mass spectrometry and neonatal screening have contributed to earlier diagnoses[5]. Despite the benefits of enzyme replacement therapy (ERT), challenges like immune response and the need for more targeted treatments remain[6].

Objectives

To review and synthesize information on Pompe Disease, focusing on pathophysiology, clinical manifestations, diagnostic strategies, therapeutic advancements and long-term management challenges.

Materials and Methods

A narrative literature review was conducted using the PubMed, Scopus and Web of Science databases. Inclusion criteria considered articles published between 2010 and 2024 in English, Portuguese or Spanish addressing clinical, molecular, diagnostic and therapeutic aspects of Pompe Disease.

Discussion

Enzyme replacement therapy has revolutionized the management of Pompe Disease, particularly in the infantile form[7], where early treatment can prevent cardiomyopathy and improve survival[8]. However, its efficacy in skeletal muscle, especially in late-onset cases, remains limited due to barriers in enzyme transport to these tissues[9]. Other approaches, such as gene therapy and the use of pharmacological chaperones[10], are being explored to overcome these limitations[11]. Recent studies show the potential of these therapies to correct specific mutations and improve GAA biodistribution[12]. Furthermore, efforts to enhance early diagnosis, such as neonatal screenings, allow interventions before irreversible symptoms appear[13].

On the other hand, challenges such as high variability in disease progression and the elevated costs of treatments highlight the need for more studies to personalize therapeutic approaches and make them accessible[14].

Conclusion

Pompe Disease remains a model for rare diseases benefiting from advancements in specific therapies. Despite significant progress with ERT, clinical and economic challenges persist. Integrating novel therapies, such as gene therapy, alongside early diagnosis through universal screenings, could revolutionize disease management in the coming years. Future research should focus on personalized approaches and strategies to mitigate ERT's adverse effects, aiming to improve clinical outcomes and patient quality of life.

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