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Case Report

Late-Onset Pompe Disease in a Middle-Aged Patient: A Rare Entity in Clinical Practice

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ABSTRACT

Pompe disease (PD), or glycogen storage disease type II, is a rare genetic condition caused by the deficiency of the lysosomal enzyme acid alpha-glucosidase, resulting in glycogen accumulation in skeletal, cardiac, and respiratory muscles. Its late-onset form (LOPD) presents primarily with progressive proximal muscle weakness and respiratory insufficiency. This report describes the case of a 48-year-old man who developed severe neuromuscular symptoms over a decade, culminating in the diagnosis of PD associated with chronic inflammatory demyelinating polyneuropathy (CIDP) and deep vein thrombosis (DVT). Diagnostic evaluation included electromyography, genetic testing, and metabolic investigations. Treatment involved enzyme replacement therapy (ERT) with alglucosidase alfa, intravenous immunoglobulin, and full anticoagulation, resulting in significant clinical improvement. This case highlights the importance of considering genetic and autoimmune etiologies in patients with progressive weakness and underscores the relevance of early diagnosis and a multidisciplinary therapeutic approach to improve functional and respiratory prognosis.

Keywords: Pompe disease; Glycogen storage disease type II; CIDP; Enzyme replacement therapy; Muscle weakness

Introduction

Pompe disease (PD), also known as glycogen storage disease type II, is an autosomal recessive inherited metabolic disorder first described in 1932 by Johannes Pompe. It is characterized by the deficiency of the lysosomal enzyme acid alpha-glucosidase (GAA), responsible for intracellular glycogen degradation. Enzyme failure leads to progressive glycogen accumulation, primarily affecting skeletal, cardiac, and respiratory muscles[1-5]. Clinically, it presents in two main forms: the classic infantile form, associated with severe cardiomyopathy and early death, and the late-onset form (LOPD), with predominant muscular symptoms such as proximal weakness and alveolar hypoventilation. The heterogeneity of LOPD symptoms often delays diagnosis, and it is frequently mistaken for inflammatory myopathies or peripheral neuropathies. Recently, mass spectrometry techniques, newborn screening, and genetic testing have become essential tools for early diagnosis. Enzyme replacement therapy (ERT) with alglucosidase alfa is the approved specific treatment, though clinical response is variable. In rare cases, PD may be associated with other autoimmune neurological conditions, such as chronic inflammatory demyelinating polyneuropathy (CIDP), adding complexity to diagnosis and treatment. This report presents a clinical case of late-onset PD in a middle-aged patient with concurrent CIDP and DVT, highlighting the clinical challenges, therapeutic strategies, and the importance of a multidisciplinary approach[6-10].

Objectives

This study aims to report a clinical case of a middle-aged patient diagnosed with late-onset Pompe disease.

Materials and Methods

A retrospective case report was prepared through the analysis of electronic medical records, complemented by a brief literature review.

Case Report

A 48-year-old retired male patient, with no history of alcohol or tobacco use, began experiencing diffuse myalgia and progressive proximal weakness in the shoulder and pelvic girdles in 2012, along with urinary and fecal incontinence and increasing dependence on basic daily activities such as dressing. Initial investigations were inconclusive. In 2019, he was hospitalized for respiratory failure, which progressed to respiratory arrest requiring orotracheal intubation and mechanical ventilation, attributed to an acute infectious event. Subsequently, he developed axial paralysis and difficulty ambulating. In 2021, he was referred to a specialized neurology service due to ascending paresthesia and weakness in the limbs, neuropathic pain, and mild exertional dyspnea; he was already using BIPAP during sleep. Electroneuromyography revealed a symmetric demyelinating polyneuropathy, confirming CIDP. A color Doppler ultrasound showed deep vein thrombosis (DVT) in the left femoral vein. Metabolic and genetic testing confirmed biallelic mutations in the GAA gene, consistent with late-onset PD. In December 2023, he was started on hospital-based alglucosidase alfa (20 mg/kg IV every 14 days), reporting improvement in dyspnea and upper limb strength. During the current admission, ERT was maintained along with intravenous immunoglobulin (2 g/kg over five days) for CIDP, full anticoagulation with low molecular weight heparin, and daily motor and respiratory physiotherapy. After 12 days, he was discharged with referral to the Lysosomal Diseases Reference Center in Curitiba and follow-up with neurology.

Conclusion

This case illustrates an atypical presentation of late-onset Pompe disease with CIDP and DVT. Early recognition and the combination of ERT, immunomodulation, and anticoagulation contributed to significant motor and respiratory recovery. In the face of progressive weakness of unknown origin, combining genetic testing with neurophysiological studies is essential for accurate diagnosis and targeted therapy, impacting prognosis and quality of life.

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