

Poster Abstract: Clinical

Laboratory and Clinical Findings Imitating Myotonic Dystrophy in the Patient with Late-Onset Pompe Disease

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INTRODUCTION

Late-onset Pompe disease is a progressive disease characterized by skeletal and respiratory muscle weakness and atrophy resulting in functional disability and reduced life span. The distinct phenotypic diversity is also characteristic of late-onset Pompe disease, and often the reason why many patients are initially misdiagnosed with other diseases. We report on a patient with clinical and laboratory findings which were formerly misdiagnosed as myotonic dystrophy.

CASE REPORT

A 51-year-old woman with 7 years history of slow muscle weakness development in the proximal limb muscles. The muscle weakness, mild myalgias, and muscle hypotrophies predominantly affected lower extremities. There was a mild elevation of serum creatine kinase. The needle EMG revealed (pseudo) myotonic discharges and myogenic changes of APMU. Bilateral cataracts were found at ophthalmologic investigation. The patient was supposed to have myotonic dystrophy. DNA analysis did not confirm myotonic dystrophy of either the DM1 or DM2 type. The DBS screening for Pompe disease was positive. Muscle biopsy, measurement of α -glucosidase

enzyme activity, and DNA analysis were performed, and the diagnosis of late-onset Pompe disease was confirmed. Enzymatic replacement therapy with recombinant GAA was administered in May 2012.

DISCUSSION

The spectrum of clinical symptoms in late-onset Pompe disease is broad and can imitate other muscle disorders (e.g. LGMD, FSHMD, chronic polymyositis). Screening of all risk individuals with muscle weakness, myalgias, elevated serum creatine kinase levels (including idiopathic hyperCKemia), muscle atrophies, respiratory insufficiency, and sleep apnea, using DBS has become a routine procedural method in clinical practice.

CONCLUSIONS

Our case report indicates that late-onset Pompe disease may also manifest itself with the phenotype of myotonic dystrophy. Early confirmation of diagnosis in patients with Pompe disease and early administration of enzymatic replacement therapy play the main role for patients' favourable outcome.

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