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Poster Abstract: Clinical

Pompe Disease: A Case Presentation

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Pompe disease is a metabolic inherited myopathy due to acid maltase (1,4-glucosidase) deficiency with glycogen storage.

Our case is a 30-year-old female patient who came to our clinic for enzyme replacement therapy. The first symptoms had occurred insidiously in 2005 with fatigability, standing and walking impairment, especially stair climbing, frequent falls with a slowly progressive course and increasing difficulties.

Clinical examination established: underweight patient, weak connective tissue, walking impairment with bilateral assistance, proximal weakness, and generalized muscular atrophies, more important at the shoulder and pelvic girdle muscles, globally abolished deep tendon reflexes.

In 2006, the EMG revealed a myopathic disorder with progressive muscular dystrophy, and a muscle biopsy was recommended. The muscle biopsy showed changes compatible with glycogen metabolic myopathy.

In 2012, during the course of the disease, respiratory failure due to respiratory muscle weakness set in. The patient underwent pulmonary function testing and the pneumologist recommended BIPAP non-invasive ventilation, 8 hours daily during sleep.

In February 2013, the DNA PCR detected two heterozygous mutations in the GAA gene, the first was located in intron 1 and the second in exon 15, and the likely diagnosis was adult-onset Pompe disease.

Since May 2013, the patient has started replacement therapy with recombinant acid α-glucosidase injected intravenously every 2 weeks with respiratory and fatigability improvement, and non-progressive motor weakness.

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