

Poster Abstract: Diagnostic

A Case of Late-Onset Pompe Disease Occurring with a Muscle Weakness Pattern Similar to that of Facioscapulohumeral Muscular Dystrophy

Lucia Ruggiero, Fiore Manganelli, Floriana Vitale, Rosa Iodice, Chiara Pisciotta, Raffaele Dubbioso, Antonietta Topa and Lucio Santoro*

Department of Neurosciences, Reproductive and Odontostomatological Sciences, University Federico II of Naples, Naples, Italy

BACKGROUND

Pompe or glycogen storage disease type II is an autosomal recessive disorder, caused by an accumulation of glycogen in the lysosome. The clinical spectrum ranges from the classic form with early onset and severe phenotype to the non-classic form with later onset and different phenotype. In fact, the clinical presentation of Pompe disease can resemble that of many musculoskeletal disorders. According to the clinical variability of the disease, we report a 48-year-old man with the adult form of acid maltase deficiency showing many clinical similarities to facioscapulohumeral muscular dystrophy (FSHD).

MATERIALS AND METHODS

The evaluation of the patient included neurological examination, blood tests, electrophysiological study, cardiac and respiratory assessments, and muscle biopsy.

RESULTS

Our patient reported winged scapula in his third decade, difficulty in climbing stairs, and hyperlordotic posture which began 5–6 years before our evaluation.

Over the years, the symptoms had slowly progressed. At the last neurological examination, the patient had a hyperlordotic posture, Beevor's sign, bilateral scapular winging, and neck flexor weakness. Moreover, muscle involvement was markedly asymmetrical and serum creatine kinase level was slightly elevated. However, the absence of clear facial muscle weakness and inheritance patterns was in contrast to the hypothesis of FSHD. Moreover the electromyography (EMG) showed myopathic changes with a short duration of motor unit action potentials and widespread fibrillation, especially in the paravertebral muscles. Muscle biopsy was not clarifying as showing variability of fiber morphology, a mild increase in connective tissue, and rare vacuoles with glycogen accumulation. In addition, respiratory evaluation revealed a restrictive pattern. Overall, we considered late-onset Pompe disease as a possible diagnosis. Thus acid maltase activity was evaluated on dried blood spot and lymphocytes and was very low.

CONCLUSION

We recommend that Pompe disease is considered in the differential diagnosis of FSHD, especially if there is early respiratory involvement and fibrillation potentials at EMG are present without clear facial muscles involvement.

*Correspondence to: Lucio Santoro, Department of Neurosciences, Reproductive and Odontostomatological Sciences, University Federico II of Naples, Naples, Italy. Phone: +39 817463103; fax: +39 817462667. E-mail: lusantor@unina.it.