

## Poster Abstract: Clinical

### Late-Onset Pompe Disease (Juvenile-Onset Form) with Elevated CK, Fatigue, and Muscular Pain After Exercise, Without Weakness

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#### INTRODUCTION

The evolution provided by enzyme replace treatment (ERT) for Pompe disease has encouraged the quest of physicians to diagnose these patients, especially patients with late-onset disease. This effort to find a diagnosis has led physicians to look for clinical characteristics other than muscular weakness and respiratory difficulty.

#### CASE REPORT

An 11-year-old Caucasian female, the first child born to healthy unrelated parents after a normal gestation and parturition, had adequate neuropsychomotor development. At the age of 10 years she was diagnosed with mononucleosis. Laboratory investigations demonstrated elevated AST/ALT. These investigations were repeatedly carried out, and AST/ALT remained elevated. The girl complained of body pain and fatigue after non-strenuous exercise. Her CK was investigated and found to be twice that of normal. Metabolic myopathy was suspected. Ischemic and fasting tests were negative. EMG was normal. She had muscular pain after light running, but no muscular weakness. A dry blood spot (DBS) test and muscular biopsy were then carried out. The muscle biopsy showed vacuolar myopathy with some vacuoles inside the fibers containing increase glycogen (PAS) and increased lysosomal activity on acid phosphatase reaction. DBS revealed severely reduced GAA activity (0.4  $\mu\text{mol/l/h}$  of protein – normal value:  $>3 \mu\text{mol/l/h}$ ). Gene sequencing: GAA showed two heterozygous

mutations (c.-32-13T>G and c.655G>A p.G219R, confirming late-onset Pompe disease of the juvenile-onset form. After discussing the case with her family, we decided to begin with enzyme replace treatment (ERT), despite her lack of muscular weakness (face, tongue, limbs, paravertebral and abdominal musculature strength was preserved).

#### CONCLUSION

This report illustrates an atypical case of a patient with late-onset Pompe disease of the juvenile-onset form, without muscular weakness, but with fatigue, muscular pain after exercise and elevated CK. Therefore, it is important, when patients with these symptoms are seen in clinical practice to investigate for Pompe disease. The decision to start ERT was made because, despite no clinical muscular weakness, muscle was affected (vacuolar myopathy on muscular biopsy) and by natural history in late-onset Pompe disease. Furthermore, as Hagemans et al. wrote “With every additional year since diagnosis, the odds for wheelchair use increased by 13% and the odds for respiratory support by 8% (both  $p<0.001$ )”.<sup>1</sup> Considering this, the possibility of a worsening clinical picture is real, and an early start of ERT would probably be better for the patient’s health.

#### REFERENCE

- [1] Hagemans ML, Winkel LP, Hop WC, Reuser AJ, Van Doorn PA, Van der Ploeg AT. Disease severity in children and adults with Pompe disease related to age and disease duration. *Neurology* 2005;64:2139–41.

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