

## Poster Abstract: Clinical

---

### Infantile Pompe Disease: Atypical Variant

Thaís Rodrigues<sup>1,\*</sup> and Carlo D. Marrone<sup>2</sup>

<sup>1</sup>*Department of Neurology, GHC – Grupo Hospitalar Conceição, Porto Alegre, Brazil*

<sup>2</sup>*Clinica Marrone, Porto Alegre, Brazil*

#### INTRODUCTION

Pompe disease (glycogen storage disease type II, glycogenosis II, or acid maltase deficiency) is a lysosomal storage disorder in which an alpha-glucosidase (GAA) deficiency causes intralysosomal accumulation of glycogen in all tissues, notably skeletal muscles. Pompe disease is transmitted as an autosomal recessive trait and is caused by mutations in the gene encoding the GAA, located on chromosome 17q25.2-q25.3. The different disease phenotypes are related to the levels of residual GAA activity in muscles. Less than 3% of normal enzyme activity is found in severe infantile cases, and residual levels ranging from 3% to 30% of normal are found in less severe late onset forms. Pompe disease clinically, presents a wide spectrum of phenotypes, ranging from the severe and rapidly progressive infantile onset form, which incorporates patients who display symptoms before 1 year of age, and the heterogeneous and more slowly progressive late onset form, which develops symptoms after 1 year of age and includes the childhood, juvenile and adult onset groups. This type typically presents respiratory insufficiency and no cardiac manifestations. The infantile onset is classified as classic infantile (presence of cardiomyopathy) and atypical or muscular variant.

#### CASE REPORT

We report a case of a boy who started with respiratory insufficiency at birth, but the diagnosis of Pompe disease was only made at 1 year and 5 months of age.

He had oropharyngeal dysphagia and multiple respiratory infections. At 1 year and 3 months, he was transferred at UTI of HMIPV because of the complication of a respiratory infection and the necessity of mechanical ventilation. Neurologic examination showed global hypotonia and areflexia. He was submitted to electromyography (which showed a myopathic pattern), a muscular biopsy (showing glycogen accumulation and vacuolated fibers containing acid phosphatase-positive material) and an echocardiography (demonstrating no cardiomyopathy, which distinguishes the classic infantile form from the atypical infantile and late onset forms). Dried blood spot and DNA evaluation confirmed the diagnosis. He started treatment with enzyme replace treatment (ERT) at 1 year and 9 months and a partial clinical improvement was observed.

#### DISCUSSION

This case illustrates an atypical pattern of the infantile onset form in a child with symptoms at birth, and a partial response to ERT.

#### CONCLUSION

Early diagnosis and early treatment are both important for patients with the infantile onset form, especially in patients with the atypical form, and ERT has been associated with improved motor capability and stabilized pulmonary function. Therefore, Pompe disease is the first neuromuscular disorder with an FDA-approved treatment. The early respiratory involvement and the characteristic laboratory abnormalities in a myopathic patient should include GAA deficiency in the differential diagnosis.

---

\*Correspondence to: Thaís Rodrigues, Rua Professor Cristiano Fischer 312/ 302 CEP, 91410-000 Porto Alegre – RS, Brazil.  
E-mail: tharr@ig.com.br.