Pompe Disease With Heterogeneous Presentations Within A Family

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Pompe disease is an acid maltase deficiency being part of glycogen storage diseases that affects all age groups. In both childhood and adult forms, the classic clinical picture is that of a progressive myopathy. Respiratory muscle involvement is common, may occur early in the course of the disease, and is the most frequent cause of mortality from acid maltase deficiency. Its association with rhabdomyolysis is rare and with a fatal prognosis. We describe the cases of a family with Pompe disease with a clinical spectrum extending throughout different ages of onset, degrees of organ involvement, and rates of progression. The twin patients with adult form of Pompe disease presented episodes of acute renal failure and respiratory insufficiency with a good outcome.

Key words: Acid maltase deficiency; Acute renal failure; Pompe disease; Respiratory insufficiency.

1. INTRODUCTION
Pompe disease is a rare, progressive muscle disease that can often be fatal (1, 2, 3). It occurs worldwide and may present at any age from infancy to late adulthood (4). A broad classification of Pompe patients divides them in two categories: infantile-onset (including classical and non-classical presentations) and late-onset (juvenile and adult) (5, 6). We describe the cases of a family with Pompe disease with a clinical spectrum extending throughout different ages of onset, degrees of organ involvement, and rates of progression.

2. CASE 1
A 21-year-old man was admitted in department of infectious diseases for diarrhea, muscle pain, weakness, limpness and darkly colored urine and oligoanuria. The physical examination revealed muscles hypertrophy of inferior extremities. Laboratory examinations revealed high seric levels of creatinine, creatine phosphokinase (CP) and lactat-dehydrogenasis (LDH) (Table 1). Because of acute respiratory failure (ARF), the mechanical ventilation of patient was needed. After intensive treatment the situation was completely improved, and all laboratory parameters were normalized. Since he experienced in childhood episodes of muscle pain, followed by darkly colored urine after mild exercise, a muscle biopsy for our patient was performed, and fibroblast culture was made, which indicated a deficient enzyme (acid alpha-glucosidase, “GAA”) and was classified as glycogen storage disease type II–Pompe disease. The same situation was repeated five years later, with acute kidney injury (AKI) (creatininemia: 5 mg/dl), ARF requiring mechanical ventilation for two weeks, and elevated muscular enzymes (CP: 9800 U/l, LDH: 1100 U/l). After intensive therapy he was discharged in good general condition making a complete recovery of renal and pulmonary function. In two episodes was not required hemodialysis.

3. CASE 2
The patient’s twin brother 26-year-old was presented at the Service of Nephrology with oliguria, darkly colored urine and high seric levels of creatinine, CP and LDH, making the diagnosis of rhabdomyolysis, and hemodialysis was required. Two days later he was complicated with respiratory muscle involvement, but not requiring mechanical ventilation. The situation was completely improved after the intensive treatment. In the family history, the twin’s sons of first patient and a patients’ brother died some days after partum from cardiac problems.

4. DISCUSSION
Acid maltase deficiency is a glycogen storage disease with heterogeneous...
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Internal Propedeutics with Communication Skills in Medicine

Authors: Izet Hozo at all, Publisher: Society of Gastroenterology of Croatia, 2013, hard cover, B5, 368p, ISBN 978-953-97541-3-4

In Split was recently promoted textbook by Prof. dr. Izet Hozo “Internal propedeutics with communication skills in medicine,” which filled the void that existed until the release of this book. If it is compared with similar books that are in use in the study of medicine, it is difficult to contemplate the work that could be used for comparison. Specifically, this comprehensive, yet easily applicable textbook that deals with clinical examination skills and communication in the Croatian language has not been written. Terminology and units of measurement are consistent with existing regulations, book is written concisely, clearly, in linguistic and conceptual terms.

The new textbook “Internal propedeutics with communication skills in medicine,” by Izet Hozo and his associates from the University Hospital Split; an interesting venture, which will not only fill a significant void in the textbook, but also contribute to a systematic theoretical approach to medical teaching, which is the day give quite a new and unusual dimension medical profession speaks during the study.

The fact that three family members died during the first days of the life from cardiac problems, is compatible with the diagnosis of Pompe disease. Genotyping is important in genetic counseling and may be of supportive diagnostic value. It provides genetic counseling and may be of supplementary diagnostic value.

In conclusion, we think that the present cases corresponds to a severe and rare form of type II glycogenosis, Pompe disease, with a clinical spectrum extending throughout different ages of onset, degrees of organ involvement and rates of progression. The twin patients with adult form of Pompe disease presented episodes of acute renal failure and respiratory insufficiency with a good outcome. We consulted these cases with genetics that provided guidance on genetic issues, such as family planning, carrier testing, and prenatal screening for other offsprings patients.

REFERENCES


BOOK REVIEW

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