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ACID MALTASE DEFICIENCY IN ADOLESCENCE: REPORT OF AN UNUSUAL CASE*

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Introduction

Classical type IIa glycogenosis or Pompe's disease is a hereditary condition characterized by muscular hypotony, weakness, hepatomegaly, cardiomegaly and frequently cardiac decompensation. These patients usually die within the first year of life. However, exceptional cases have been reported where the condition developed for the first time in childhood[1-3], adolescence or adulthood[4,5], which are probably glycogenosis type IIb disease.

In this report a patient with onset of the disease in adolescence who survived beyond the age of 16 is described. In contrast to the adult form of the disease the symptoms found were cardiac enlargement without organomegaly and skeletal muscle weakness. Histological and biochemical studies on the muscle biopsy led to the diagnosis of acid maltase deficiency.

Case Report

A.Ç., a male patient, was born at term following a normal pregnancy and delivery. Both parents were in good health; there was no consanguinity. A male sibling, who was probably affected with a similar conditon, was known to have died of cardiac failure at 12 years of age. The patient was first admitted to the hospital at 15 years of age with complaints of weakness and dyspnea that developed after an upper

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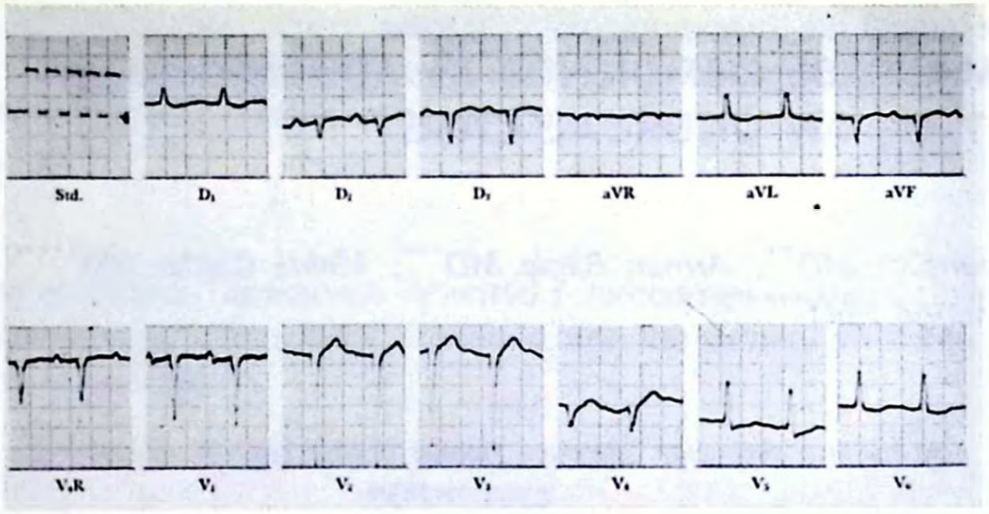

Figure 1.

Electrocardiogram showing left axis deviation, prolongation of the PR interval, biatrial enlargement, and incomplete left bundle branch block.

respiratory tract infection. His development and intelligence were within normal limits. He weighed 48 kg and was 170 cm tall. His blood pressure was 100/60 mmHg. There were no cardiac murmurs; the rhythm was tachycardic. The liver and spleen were nonpalpable. There was no muscular atrophy, and the deep tendon reflexes were normo-active. The rest of the physical examination was unremarkable. An electrocardiogram (Figure 1) revealed left axis deviation, prolongation of the PR interval, biatrial enlargement, and an incomplete left bundle branch block. On the telecardiogram a global cardiomegaly was noted (Figure 2). The echocardio-

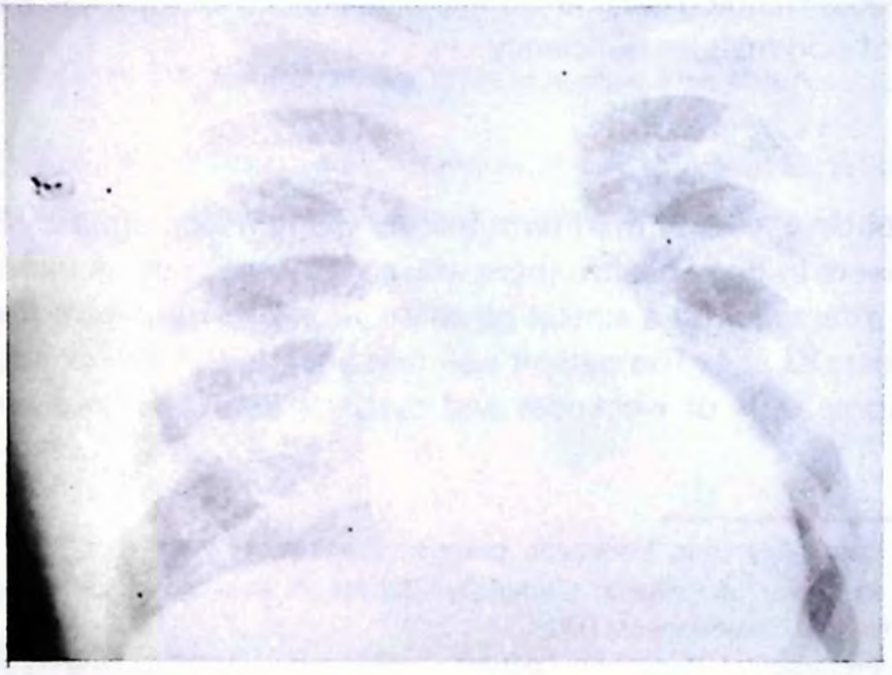

Figure 2.

Telecardiogram showing global cardiomegaly.

graphic examination showed paradoxical septal motion; the end-diastolic dimension was 6 cm (N = 3.3 -4.7), ejection time was 240 msec, interventricular septal thickness was 0.6 cm, and the left ventricular posterior wall thickness was 0.9 cm. DE slope was 400 mm/sec (Figure 3). In right heart catheterization, mean right atrial pressure was 20 mmHg, right ventricular pressure was 40/10-17 mmHg, pulmonary artery pressure was 40/20 mmHg. Atrial oxygen saturation was 96%, venous saturation was 70%, cardiac index was 2 l/min/m^2. A cineangiocardiogram revealed a huge right ventricular cavity with a weak contraction; during venous return the left ventricular cavity was also large and the contractions were feeble; the emptying of the ventricle was late.

Hemogram, urinalysis and blood electrolyte concentrations were normal.

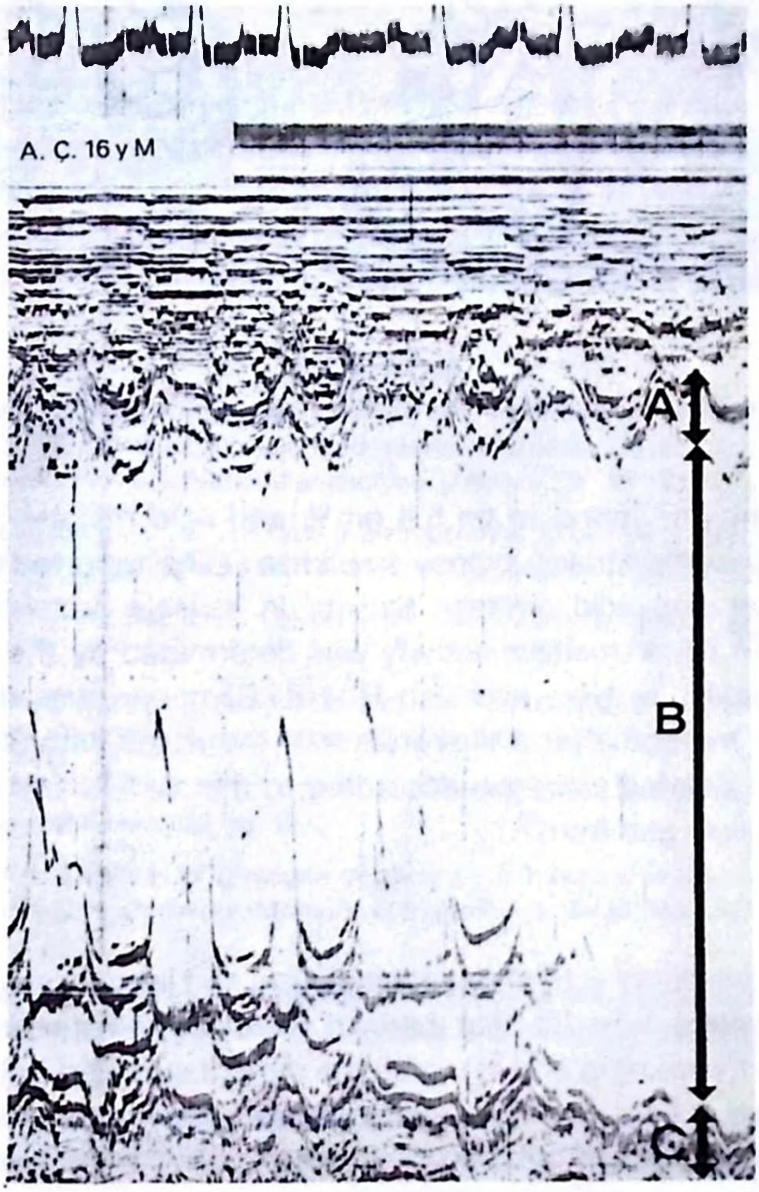

Figure 3.

Echocardiogram showing congestive type myopathy.
A. Interventricular septum, B. Left ventricular end-diastolic dimension, C. Left ventricular wall.

Muscle biopsy specimens were obtained from the gastrocnemius muscle. The formalin-fixed and hematoxylin-eosin stained tissue revealed large vacuoles in the muscle fibers which suggested storage of glycogen (Figure 4).

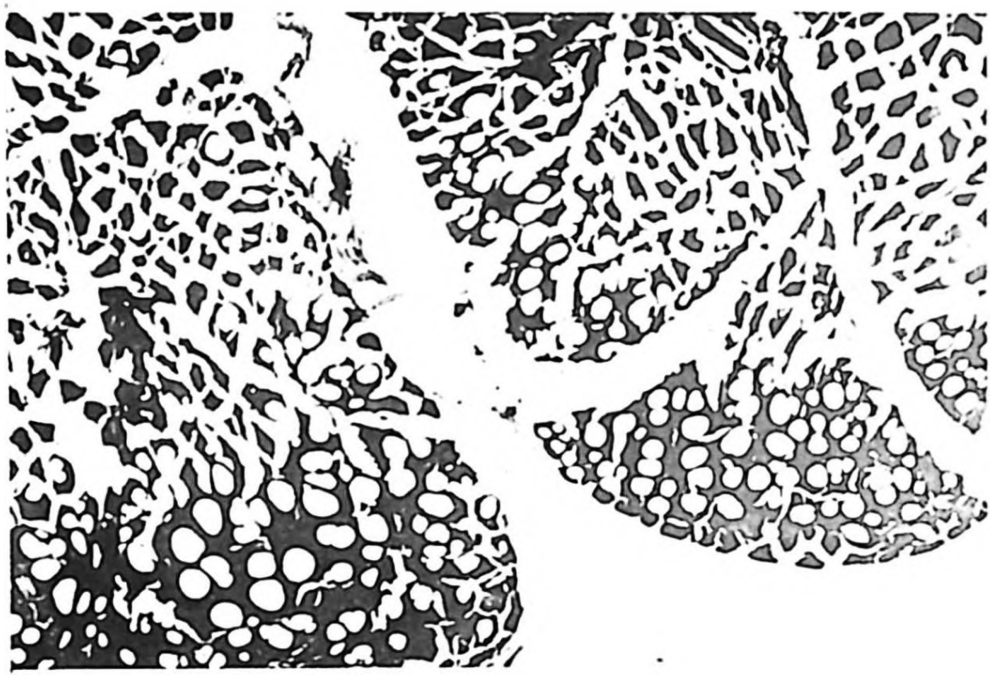

Figure 4.

Skeletal muscle showing vacuoles in the muscle fibers which suggest storage of glycogen.

Glycogen content was found to be 5.4 gm% and acid maltase (pH = 5) activity 4 μmol/min/gm in the muscle biopsy specimen. (The reported normal values for glycogen content and acid maltase activity in muscle are 1.5 gm% and 8.22 μmol/min/gm [2,6].) Acid maltase activity was determined by the rate of hydrolysis of maltase according to Hers and Van Hoof[6]. Glycogen was determined by the glucose oxydase method after disintegration of the tissue with 30% KOH and acid hydrolysis of the isolated glycogen according to the methods of Hassid and Abraham[7] and Bergmeyer and Berrt[8].

Discussion

The purpose of this paper is to present the clinical and laboratory data of an unusual case of glycogenosis type IIb that had an onset of the disease in adolescence without muscular weakness and in which the patient survived beyond sixteen years of age. Glycogen excess and acid maltase deficiency in the skeletal muscle and histopathological changes in the muscle correlate well with the biochemical findings. However the presence of cardiomegaly, and the rapid progress of the symptoms leading to cardiac decompensation may characterize this case as an intermediate clinical type when compared with those reported in the literature[1-5], since car-

diomegaly without skeletal myopathy is thought to be less involved in cases whose symptoms became evident at later ages.

The basic reason for the variability of organ or tissue involvement at different ages in acid maltase deficiency is unknown. Therefore, systemic study of these cases may help in better understanding the pathogenesis and biochemical processes underlying acid maltase deficiency. Compensatory factors responsible for the milder expression of the syndrome in children and adults have not yet been identified.

Therapeutic measures are not available; only symptomatic care is given to the patient. Thus the recognition of the disease at all ages may be important for genetic counselling.

Summary

A sixteen-year-old male patient with acid maltase deficiency is reported. The case is unusual because of the age of onset of the disease and the symptom of cardiac enlargement without muscular weakness, in contrast to the adult type IIb glycogenosis. The importance of recognizing the disease at all ages for genetic counselling is discussed.

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