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A CASE OF MUCOPOLYSACCHARIDOSES TYPE I WITH HEART INVOLVEMENT DURING INFANCY*

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Summary: Demirsoy S, Gücüyener K, Olguntürk R, Tunaoğlu S, Oğuz D. (Department of Pediatrics, Gazi University Faculty of Medicine, Ankara, Turkey). A case of mucopolysaccharidoses type I with heart involvement during infancy). Turk J Pediatr 32: 49-52, 1990.

We report a case of mucopolysaccharidoses I with severe cardiac involvement, which was diagnosed on the basis of clinical and laboratory findings even though, symptoms begin to occur in mucopolysaccharidoses after the first year of life. In our case cardiac failure occurred in the third month of life, which resulted in the patient's death after one month. *Key words: mucopolysaccharidoses, heart, infancy.*

The mucopolysaccharidoses (MPS) are a group of inherited disorders caused by incomplete degration and storage of acid mucopolysaccharides in various organs, including the heart[1].

We report a case of mucopolysaccharidoses type I with severe heart involvement during infancy, which resulted in death.

Case Report

A three-month-old girl was admitted to the Department of Pediatrics of Gazi University Faculty of Medicine with complaints of respiratory difficulty which had been present since birth, and cyanosis and vomiting which had been present for two days. Her parents were not related.

Physical examination on admission revealed an extremely agitated infant with pallor, peroral cyanosis and coarse facies. Her temperature was 38.3 °C, respiratory rate 60/min, pulse rate 200/min, and blood pressure 70/30 mm Hg. She had stridulous breathing and intercostal retractions. The liver was palpable four cm below the costal margin.

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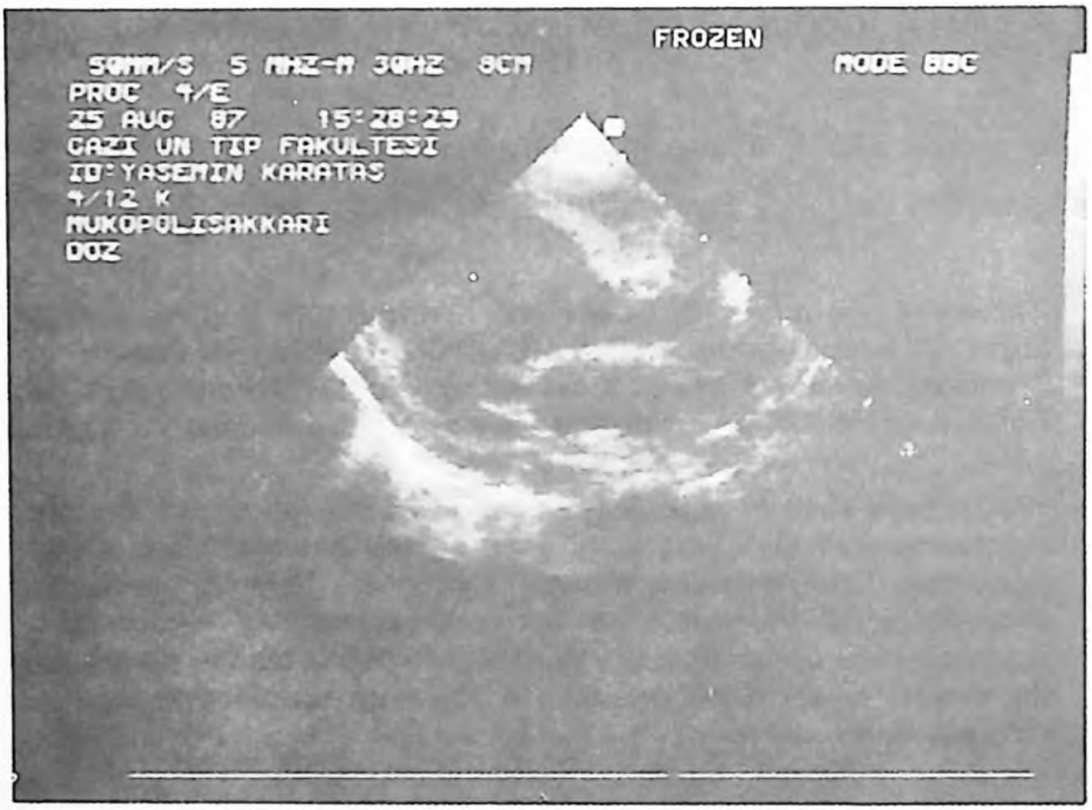

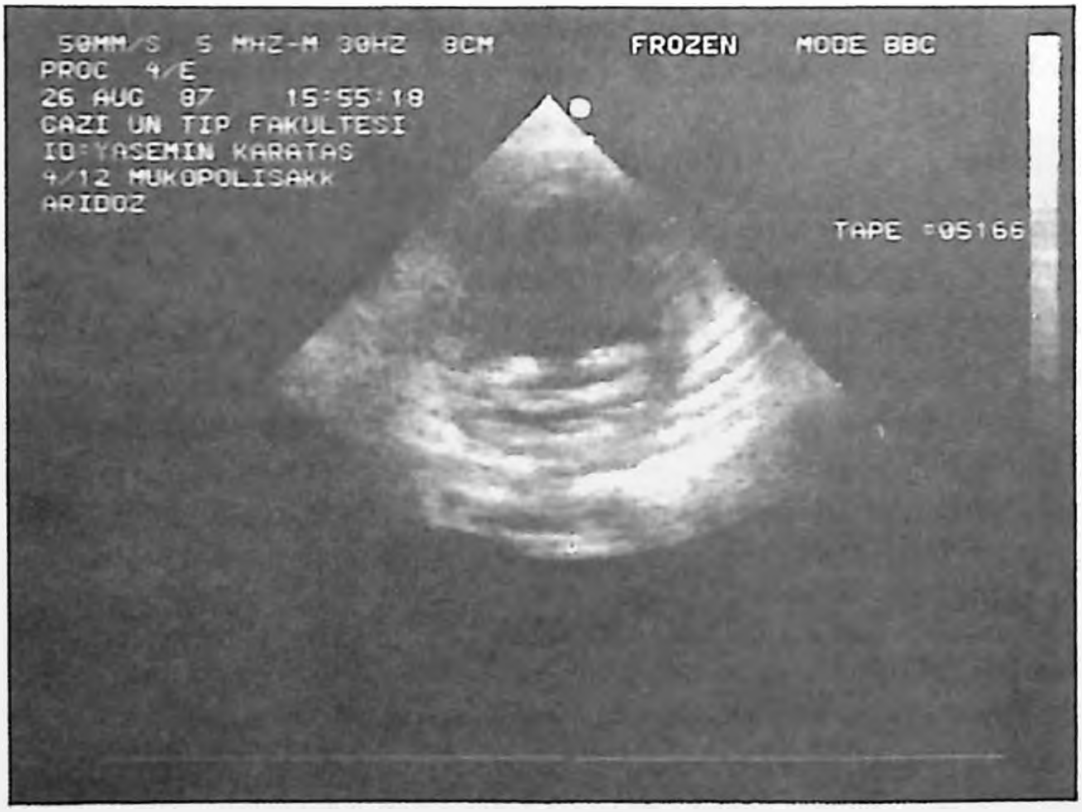

Fig. 1: Thickening in mitral valve leaflets (longitudinal A and horizontal B).

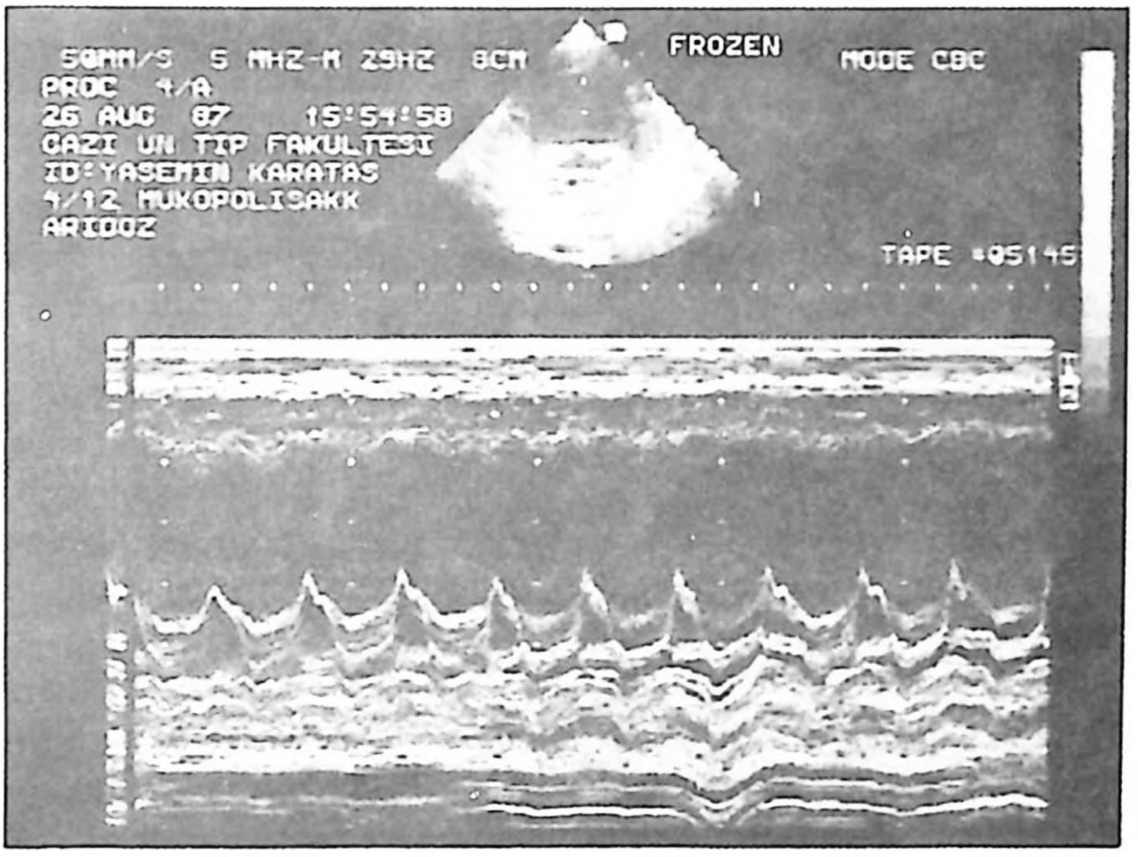

Fig. 2: Echocardiogram shows mitral valve prolapse.

Laboratory studies, including a complete blood count, urinalysis, and liver and kidney function tests were within normal limits. Moderate cardiomegaly was detected on the telecardiogram, and the L 1 vertebral body appeared more rounded and immature than usual in the dorsolumbar graph. Electrocardiography revealed left ventricular hypertrophy.

The echocardiogram was recorded using a Hewlett Packard Ultrasonoscope (Model 77 020 A) with a 3.5 MHz transducer. Utilizing the echocardiographic measurements and Bi-plane Ellipse Method, ventricular functions (ejection fraction and fractional shorting) were found to be decreased. There was a profuse thickening and restriction in the mitral valve leaflets during diastole (Figs. 1 a, b). A prolapse in the anterior leaflet of the mitral valve was observed (Fig. 2). The left atrial dimensions were slightly increased. The aortic, pulmonary and tricuspid valves were normal.

Because of the presence of coarse facies, cardiomegaly, hepatomegaly and noisy breathing, the diagnosis of mucopolysaccharidoses was suspected and urinary excretion of mucopolysaccharides was investigated. Total mucopolysaccharides were 97 mg/dl (normal: up to 85 mg/dl) with 20 % heparan sulfate (normal: 0), 63 % dermatan sulfate (normal: 0-20 %), and 17 % hyaluronic acid (normal: 10-20 %). Institution of appropriate therapy resulted in moderate clinical improvement, and the patient remained in stable condition for the next month. On her

second hospital admission, physical examination revealed an infant in poor health with symptoms of severe cardiac failure. She died five hours after admission.

Discussion

Deficiency of ten specific lysosomal enzymes has been demonstrated in various mucopolysaccharidoses[1]. The clinical manifestations of mucopolysaccharidoses have some common symptoms. Generally, the clinical course in all the mucopolysaccharidoses is one of apparent normal development in the first years of life, and progressive deterioration beginning at some time thereafter[1,2]. Death usually occurs before the age of ten in the prototype disorder, Hurler's syndrome, which carries the gravest prognosis among all the mucopolysaccharidoses[3]. On the other hand, patients with the Scheie and Hunter syndromes have survived way into their sixties, and even longer. No specific treatment is available for any of these disorders[1].

In our case, stridulous breathing, coarse facies, cardiomyopathy, hepatomegaly, immature vertebrae and excessive amounts of dermatan sulfate and heparan sulfate in the urine were the criteria used for the diagnosis of mucopolysaccharidoses[1]. The interesting aspect of our case was the early severe cardiac involvement which resulted in the patients's death due to cardiac failure. Cardiac involvement occurs in at least eighty percent of patients but only half show clinical incidence of cardiovascular problems. Cardiomegaly may be detected without evidence of organic murmur, as in our case. Thickening of the mitral annulus and the leaflets may be due to infiltration of mucopolysaccharides, as previously described[4]. Fifty percent of patients die either from cardiac failure or suddenly. Early death is the usual outcome[5].

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