

Abetalipoproteinemia Presenting with Congestive Cardiac Failure

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Abetalipoproteinemia is an uncommon autosomal recessive disease in which affected individuals are incapable of normally synthesising betalipoprotein. This disorder has five basic features, abetalipoproteinemia, malabsorption of fat, acanthocytosis, retinitis pigmentosa and ataxic neuropathy(1). Though only a few cases have been published in the literature(2), abetalipoproteinemia has attracted attention of a variety of specialists including neurologists, gastroenterologists and ophthalmologists. We report a case of this rare disorder, which presented with unusual manifestations.

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Case Report

A 6-year-old child was admitted with complaints of breathlessness on exertion and edema over face and lower limbs. These complaints were of one year duration and were slowly progressive. He had history of night blindness since 1 year and passing stools soon after food since the last 4 years. He used to pass few drops of blood in the stool since 3 months and he used to pass 3 to 4 pale bulky stools upto the age of 3 years. The boy was born of a nonconsanguineous marriage; he was normal at birth and attained developmental milestones normally.

Physical examination of the child revealed, pallor, marked clubbing and bilateral pedal edema. His skin was dry and both eyes showed Bitot's spots. The height was 107 cm, weight 18.5 kg, pulse rate 120/min, BP 100/70 mm Hg and respiratory rate 40 per minute. On cardiac examination precordial bulge was present, apical impulse was palpable on the left 5th intercostal space 2 cm outside the midclavicular line and was forcible in nature. Parasternal heave was present. Heart sounds were normally heard. Grade II ejection systolic murmur was heard in the 4th intercostal space at the left sternal border. The child had generalized hypotonia but all the deep tendon reflexes and superficial reflexes were normal. The fundus examination showed pigmentary changes similar to retinitis punctata albescens. On abdominal examination liver was palpable 2 cm below right costal margin, soft with rounded borders and spleen was palpable 1 cm.

Blood examination showed hemoglobin 7 g/dl, ESR 45 mm at 1 hour, total leucocyte count of 5,400 cells/cmm. The erythrocytes showed marked anisocytosis with

good number of macrocytes, microcytes and 20% acanthocytes. No immature cells or parasites were seen. The packed cell volume was 14%. The routine urine examination was normal. The blood level of cholesterol was 60 mg/dl, triglycerides 112 mg/dl, HDL cholesterol 30 mg/dl, and LDL cholesterol 32 mg/dl. The serum creatinine was 1 mg/dl and urea was 27 mg/dl. The serum glutamic oxalate transaminase was 87 IU/L, serum glutamine pyruvic transaminase was 23 IU/L and serum bilirubin was 0.4 mg/dl.

The chest radiograph showed cardiomegaly with normal lung parenchyma and the cardiothoracic ratio was 0.56. ECG showed deep Q waves in Leads II and III with left ventricular hypertrophy. On echocardiography, the following parameters were noted: aorta 22 mm, left atrium 32 mm, left ventricular internal diameter in diastole 44 mm, left ventricular internal diameter in systole 26 mm and right ventricular internal diameter in diastole 12 mm. The mitral, tricuspid, aortic and pulmonary valves were normal. The 2 D-echo showed mild dilatation of the left ventricular and left atrial chambers.

The right atrium and ventricular chambers were normal in size. The left ventricle showed global hypokinesia. The left ventricular end diastolic volume was 60.85 ml and left ventricular end systolic volume was 38.36 ml, giving a systolic volume of 22.5 ml and left ventricular ejection fraction of 37%. The interatrial and interventricular septa were intact, valves were normal, and there was no evidence of pericardial effusion, vegetation or clots. On colour flow mapping a mild Grade I mitral regurgitation was detected.

The child was given tablet frusemide 20

mg twice daily for 5 days to control his cardiac failure. He was treated with oral iron 100 mg, folic acid 0.5 mg and cyanocobalamin 5 microgram all given thrice daily. After four weeks of this therapy, the hemoglobin rose from 7 to 11 g/dl. At this stage the echocardiogram was repeated which showed a picture of dilated left ventricle with global hypokinesia and reduced left ventricular systolic function. The left ventricular diastolic volume was 68.25 ml and left ventricular systolic volume was 46.45 ml giving a left ventricle ejection fraction of 32%, showing thereby that the child had cardiomyopathy.

Discussion

Abetalipoproteinemia presents with failure to thrive and fat malabsorption in infancy with subsequent development of spinocerebellar ataxia and retinitis pigmentosa(3). Marked clubbing with no evidence of pulmonary pathology and a past history of bulky stools and presence of night blindness made us consider a malabsorption syndrome in the present patient. Approximately 50 cases of abetalipoproteinemia have been reported upto 1992(4).

Vision and retinal appearances are normal in early childhood. A decrease in visual acuity, night blindness and loss of dark adaptation become first evident and occasionally progress to blindness(2). Fundoscopic changes of progressive retinitis resembling retinitis punctata albescens occur in about 50% of patients(4). This child had night blindness and retinal changes resembling retinitis punctata albescens. With progression of the disease, optic atrophy, bilateral scotomata and limitation of convergence are reported(5).

Cardiac changes in abetalipoproteinemia have been described in a review by

Kayden(6) who has reported 6 early deaths. Cardiomyopathy with arrhythmias and cardiomegaly similar to that of Friedreich's ataxia, is described in patients with abetalipoproteinemia(1). Cardiac abnormalities including irregularities of rhythm are common(1). Cardiac failure as the cause of death has been reported by Sobrevilla *et al.*(7) and by Dische and Porro(8) in one case each. However, one patient who was reported to be alive at 38 years of age(5) had no symptoms or signs of cardiac disease and had a normal electrocardiogram.

Abetalipoproteinemia should be considered in any patient having acanthocytosis, malabsorption, retinitis pigmentosa or unexplained neurological abnormalities resembling Friedreich's ataxia. The experience with our case indicated that abetalipoproteinemia should be considered in patients presenting with congestive cardiac failure and visual disturbance as they may not be isolated entities.

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