The Turkish Journal of Pediatrics 34:47-53, 1992

ECHOCARDIOGRAPHIC FINDINGS IN ENDOMYOCARDIAL FIBROSIS*

*Muhsin Saraçlar MD***, Sema Özer MD***, Funda Öztunç MD****
*Alpay Çeliker MD****

SUMMARY: Saraçlar M, Özer S, Öztunç F, Çeliker A. (Department of Pediatric Cardiology, Hacettepe University Institute of Child Health, Ankara, Turkey). Echocardiographic findings in endomyocardial fibrosis. Turk J Pediatr 34:47-53, 1992.

An 18-month-old infant diagnosed as having endomyocardial fibrosis by echocardiography is presented. Most patients with endomyocardial fibrosis reported in the literature are either older children or adults. To our knowledge, our patient was the youngest ever to have been reported. Echocardiographic studies showed obliteration of the left ventricular apex and increased echo reflectance at the left ventricular endocardium and subendocardium. The left atrium and right ventricle were significantly enlarged. Doppler echocardiography showed minimal mitral, but significant tricuspid regurgitation. In regard to the contribution of echocardiography in the diagnosis, we recommend this method for suspected cases. Contrary to the other patients reported, there was no thickening of the atrioventricular valves. Mitral valve insufficiency was related to the restriction of the ventricular filling rather than to valve involvement occurring with the disease. *Key words: endomyocardial fibrosis, echocardiography.*

Restrictive cardiomyopathy may be arbitrarily divided into two major categories: 1) endomyocardial fibrosis including Löffler's fibroblastic endocarditis, and 2) infiltrative myocardial diseases, which include amyloid, sarcoid, hemochromatosis, and Pompe's and Fabry's diseases[1].

Endomyocardial fibrosis (EMF) was first described in Europe by Löffler in 1936. Later, a restrictive variant was reported from black Africa in 1948. Subsequent reports of the disease came essentially from tropical black Africa (Uganda, Nigeria). Since 1950, EMF has been observed in South America (Brazil, Venezuela, etc.)[2], but it has also been seen in temperate zones in the U.S.A., France and Japan. However epidemiological studies have shown the disease to be more prevalent in the tropics than in temperate areas[2,3].

* From the Department of Pediatric Cardiology, Hacettepe University Institute of Child Health, Ankara.

** Professor of Pediatrics and Pediatric Cardiologist, Hacettepe University Institute of Child Health, Ankara.

*** Associate Professor of Pediatrics and Pediatric Cardiologist, Hacettepe University Institute of Child Health.

**** Fellow in Pediatric Cardiology, Hacettepe University Institute of Child Health.

Until recently, procedures other than echocardiography have been used in the diagnosis of EMF. In this report, we describe an 18-month-old infant in whom a diagnosis of EMF was established using echocardiography. As to our knowledge, this is the youngest case of EMF ever to have been described in the literature and the first report from Turkey in the pediatric age-group.

Case Report

An 18-month-old boy was admitted to the hospital for evaluation of tiredness and malaise. Physical examination revealed an infant with a height of 74 cm and weight 9250 g. The arterial blood pressure was 110/70 mm Hg. At the xyphoid area there was a grade III/VI blowing holosystolic murmur. The second heart sound at the pulmonary area was single and loud. The liver edge was palpable 4 cm below the right costal margin, while the spleen was enlarged 2 cm. No cyanosis was noted.

The ECG showed a northwest QRS axis in the frontal plane and right ventricular hypertrophy. The telecardiogram revealed global cardiomegaly. The hemoglobin was 10.50 g/dl, hematocrit 33%, white blood cells 7800/mm^3, mean corpuscular volume 77fl, reticulocytes 0.2%, erythrocyte sedimentation rate 29 mm/h, ASO titer 625 units, CRP positive. The latex test was negative. Total eosinophil count 100 cells/mm^3, total protein/albumin 6.9/3.8 g/dl, alkaline phosphatase 98 BU, uric acid 4.5 mg/dl, AST (aspartate aminotransferase) 25 U, ALT (alanine aminotransferase) 8 U, and fasting blood glucose 83 mg/dl. Bone marrow and urinalysis were normal. There were no parasite ova or cysts in the stool. Echocardiographic studies were carried out using a Toshiba Sonolayer-S SSH 60 echocardiograph. A standard 2½-MHz as well as a 5-MHz transducer were utilized.

Two-dimensional echocardiography illustrated obliteration of the left ventricular apex in the apical four-chamber and parasternal long axis views (Figs. 1, 2). However, left ventricular functions were within normal limits (ejection fraction 53%, shortening fraction 26%). There was increased echo reflectance at the left ventricular endocardium and subendocardium, however, this could only be detected in some parts on the right ventricle. The structure of the mitral and tricuspid valves was normal echocardiographically. The left atrium and right ventricle were significantly enlarged. The left atrium was 20 mm. Pulmonary arteries and veins were dilated. Doppler echocardiography showed minimal mitral regurgitation, but significant tricuspid regurgitation. The systolic pressure gradiants were 16 mm Hg in the mitral and 80 mm Hg in the tricuspid valve.

Right and left heart catheterization and angiocardiography were performed from the right femoral region by the percutaneous technique. The catheter entered the right atrium, right ventricle, and pulmonary arteries via the inferior vena cava. The left heart catheter entered the left ventricle and atrium from the ascending aorta.

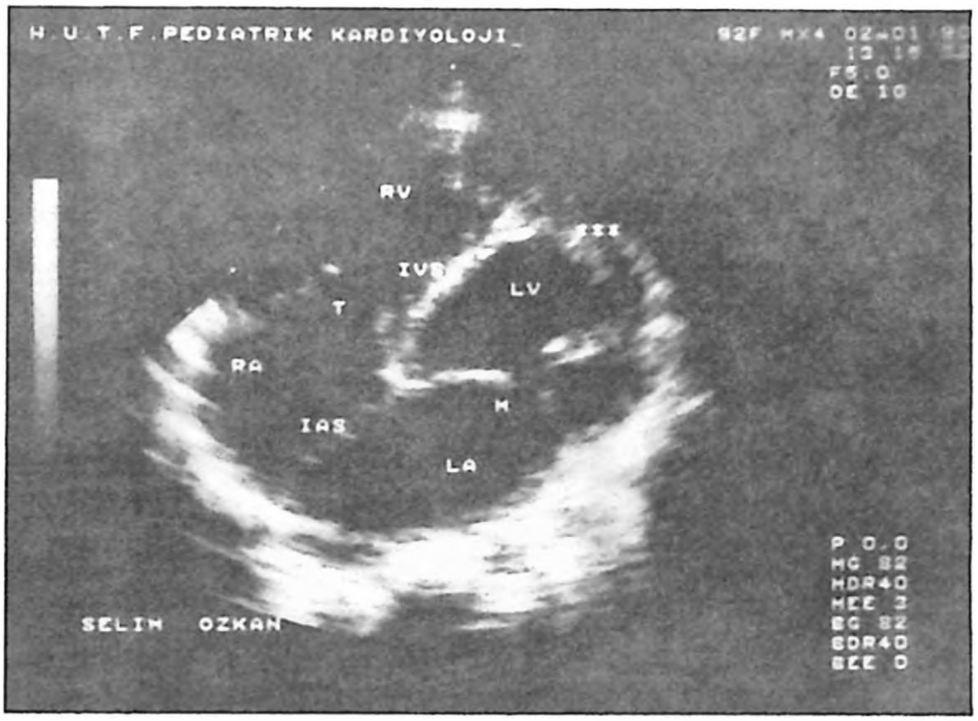

Fig. 1: Apical-four chamber view echocardiogram. Obliteration of left ventricular apex is seen (xxx). RV: right ventricle, T: tricuspid valve, RA: right atrium, LV: left ventricle, M: mitral valve, LA: left atrium, IVS: interventricular septum, IAS: interatrial septum.

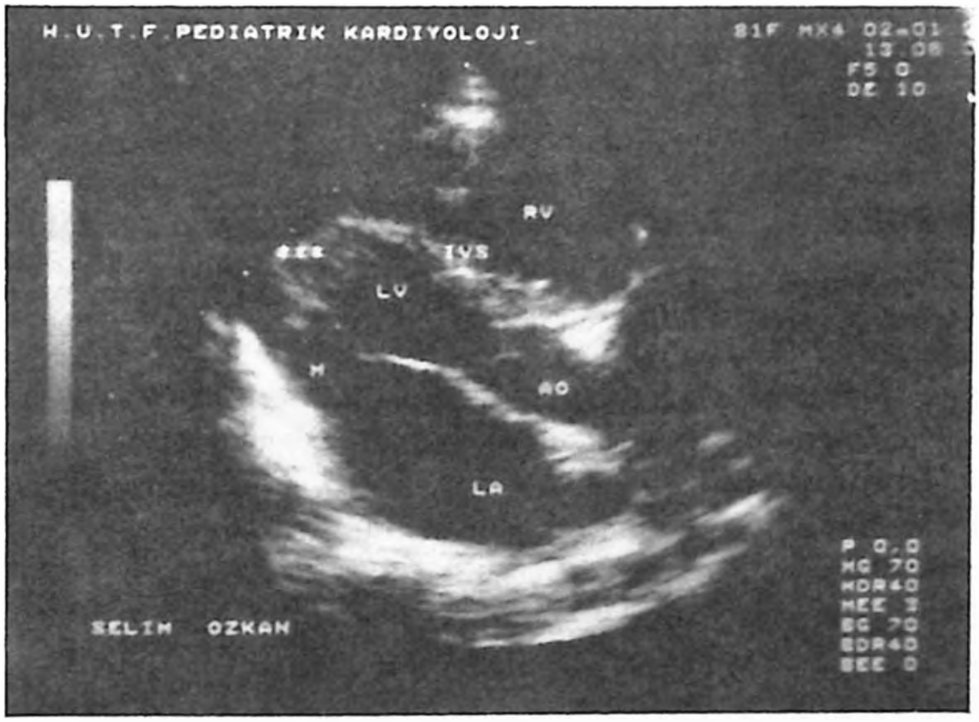

Fig. 2: Parasternal long axis view echocardiogram. Obliteration of left ventricular apex is seen (xxx).

There was no significant change in oxygen saturation. Pressures were pulmonary wedge, 20 mm Hg (mean); main pulmonary artery, 80/25 mm Hg (mean 64); right ventricle, 80/2-13 mm Hg; right atrium, 10 mm Hg (mean); left atrium, 22 mm Hg (mean); left ventricle, 90/3-16 mm Hg, and aorta, 90/54 mm Hg (mean 75). The

right ventricular angiocardiogram showed a dilated right ventricle, large main pulmonary artery and tricuspid insufficiency. Left ventricular angiocardiograms showed severe amputation of the left ventricular cavity resembling an "apple stalk" (Fig. 3). Moderate mitral insufficiency was observed. Left atrial angiography revealed that the left atrium and pulmonary veins were enlarged.

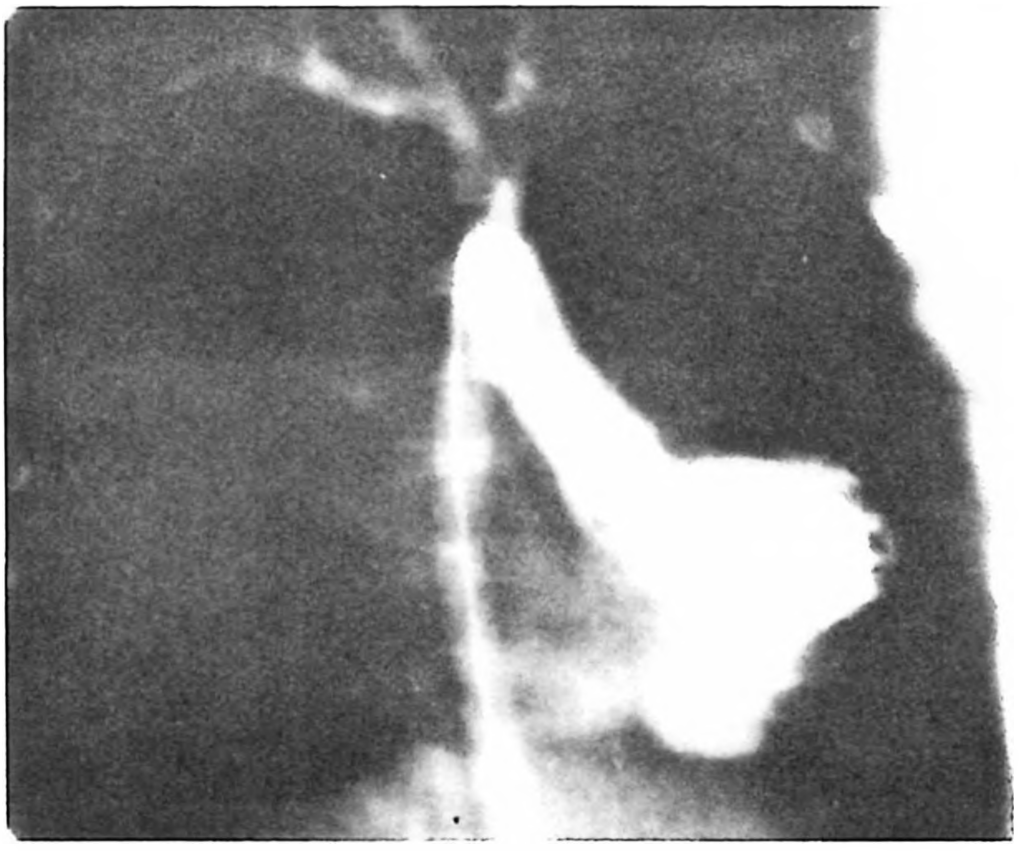

Fig. 3: Left ventricular cineangiogram in the right anterior-oblique projection: severe amputation of the left ventricular cavity (arrows) and mitral insufficiency are illustrated.

Bioptome was inserted through the right femoral vein and four specimens were taken from the right ventricular outflow tract, which were studied by light microscopy. Endomyocardial biopsy material showed moderate thickening of the endocardium and hypertrophy in some heart muscle fibers along with edema of the connective tissue of the myocardium.

After studying the clinical and laboratory findings, the patient was diagnosed as having EMF. He will be closely followed and measures will be taken to prevent congestive heart failure, in addition to his being considered a candidate for endocardiectomy in the future.

Discussion

Most patients with EMF reported in the literature are either older children or adults. Metras et al[4] reported 20 patients whose mean age was 13.3 years, the youngest being six years old. Gottdiener et al[5] reported 21 patients between the

ages of 16 and 61 while Vijayaraghavan et al[6] reported the ages to be between 5 and 48. Martines et al[7] studied 15 consecutive patients with an age-range of 11 to 56 years. In a larger series, Mady et al[8], reported 110 patients with EMF, the youngest, aged five. The patients studied by Acquatella et al[9] were all adults. None of the patients described by these workers was as young as our patient, 18 months of age.

There was a male predominance in the patients reported in the literature. Our patient was also a male.

EMF may present with or without eosinophilia. However, pathologic series suggest that both forms represent a spectrum of a single entity[9]. There was no eosinophilia observed in our patient.

In the past, this disease was mainly diagnosed in endemic areas by its clinical picture, by the characteristic obliteration of the ventricular apex or at necropsy[9]. However, recent reports indicate that echocardiography alone is sufficient to arrive at a reliable diagnosis of EMF. At the same time, this technique may be used to assess the extent and progression of the disease. During life, early isolation of the activity of the disease may also be evaluated by echocardiography[6]. In our patient, we were able to diagnose the disease first by echocardiography. Then, we confirmed the diagnosis with angiocardiography and biopsy.

EMF may only involve the right or left ventricle. However, in some patients both ventricles are affected[2]. In the majority of EMF patients, sheets of fibrous tissue of varying thickness develop in the subendocardium, initially involving the apices of the ventricles and extending into the inflow tracts of the right and the left ventricles. In addition an intracavitary thrombus appears as a filling defect in the apices of one or both ventricles[1]. In the case presented, echocardiographic findings showed that the left ventricular apex was involved (Figs. 1, 2). However the right ventricular apex was not apparently involved. The left ventricular apical intracavitary amputation was also detected by angiocardiography (Fig. 3).

The distribution of endocardial fibrosis may be either patchy or confluent. St John Sutton[1] reported that increased echo reflectance of the subendocardial surface is consistent with fibrous replacement. The spectrum of left ventricular geometry in restrictive cardiomyopathy is more varied than in either hypertrophic cardiomyopathy or dilated cardiomyopathy. Although left ventricular chamber size is almost always normal, wall thickness may be decreased, normal or increased, and these changes may occur throughout ventricles or be localized to specific regions within the ventricles[1]. In our patient, the left ventricular endocardium was involved extensively and the left ventricular functions determined were within normal limits echocardiographically. However, the right ventricle showed increased echo

reflectance of the subendocardial surface in some areas, which suggested patchy infiltration of fibrous tissue.

Since the left ventricular cavity was decreased in size, the left ventricular diastolic pressure was elevated. This results in left atrial enlargement[1]. In our patient, we detected only minimal mitral regurgitation but significant enlargement of the left atrium, demonstrated by Doppler, two-dimensional echocardiography or by angiocardiography. Left atrial mean pressure and pulmonary artery wedge pressure were elevated because of the situation described above. This results in pulmonary hypertension and elevation of right ventricular pressure, as detected in our patient. We related significant tricuspid regurgitation to the elevation of right ventricular pressure, as described by St John Sutton[1].

It has been reported that in most cases evaluated by echocardiography, thickening of the mitral and/or tricuspid posterior valves is one of the characteristic findings of EMF[6,9]. However, in our patient, the leaflets of the atrioventricular valves did not illustrate echocardiographic thickening. Therefore, we related atrioventricular valve incompetence to restriction of ventricular filling rather than to valve involvement, as suggested by Cherian et al[10] and Metras et al[4].

Strong echoes were recorded from the left ventricular endocardium, which were interpreted as fibrotic changes in the endocardium or subendocardium. At the same time, the left ventricular apex was obliterated and reflected dense echoes. This occurred because of endomyocardial scarring of the left ventricular apex. We could not detect any significant evidence of fibrosis in the right ventricle, but there were increased echoes in some parts of the endocardial surface of the right ventricle. Schmaltz et al[11] has demonstrated that an endomyocardial biopsy performed during childhood was a diagnostic tool which could add useful information to the etiology or pathogenesis of an underlying myocardial disease. They found that biopsies were diagnostic in 11.7 percent of patients, helpful in 71.7 percent of patients, and were of no help in 16.6 percent of patients. In our patient, the biopsy taken from the right ventricle supported the diagnosis. If the biopsy had been obtained from the left ventricle, where the lesion was very significant, it would have probably been diagnostic.

Surgical treatment of. EMF consists basically of endomyocardiectomy and valvular replacement[4,7,8,10]. Therefore, we decided to follow up our patient and to perform an endocardiectomy in the future.

REFERENCES

1. St John Sutton M. Cardiomyopathy. In St John Sutton M, Oldershaw PJ (eds). *Textbook of Adult and Pediatric Echocardiography and Doppler* Boston: Blackwell Sci, pp 339-379, 1989.
2. Metras BD. Paediatric cardiology in the tropics. In Anderson RH, Macartney FJ, Shinebourne EA, Tynan M (eds). *Paediatric Cardiology* Vol 2. New York: Churchill, pp 1321-1341, 1987.
3. Gupta PN, Valiathan MS, Balakrishnan KG, et al. Clinical course of endomyocardial fibrosis. *Br Heart J* 62:450, 1989.
4. Metras D, Coulibaly AO, Ouattara K, et al. Endomyocardial fibrosis: early and late results of surgery in 20 patients. *J Thorac Cardiovasc Surg* 83:52, 1982.
5. Gottdiener JS, Maron BJ, Schooley RT, et al. Two-dimensional echocardiographic assessment of the idiopathic hypereosinophilic syndrome. Anatomic basis of mitral regurgitation and peripheral embolization. *Circulation* 67:572, 1983.
6. Vijayaraghavan G, Davies J, Sadanandan S, et al Echocardiographic features of tropical endomyocardial disease in South India. *Br Heart J* 50:450, 1983.
7. Martinez EE, Venturi M, Buffolo E, et al. Operative results in endomyocardial fibrosis. *Am J Cardiol* 63:627, 1989.
8. Mady C, Pereira Barretto AC, de Oliveira SA, et al. Effectiveness of operative and nonoperative therapy in endomyocardial fibrosis. *Am J Cardiol* 63:1281, 1989.
9. Acquatella H, Schiller NB, Puigbó JJ, et al. Value of two-dimensional echocardiography in endomyocardial disease with and without eosinophilia. A clinical and pathologic study. *Circulation* 67:1219, 1983.
10. Cherian G, Vijayaraghavan G, Krishnaswami S, et al. Endomyocardial fibrosis: report on the hemodynamic data in 29 patients and review of the results of surgery. *Am Heart J* 105:659, 1983.
11. Schmaltz AA, Apitz J, Hort W, Maisch B. Endomyocardial biopsy in infants and children: experience in 60 patients. *Pediatr Cardiol* 11:15, 1990.