Folia Cardiologica 2018
tom 13, nr 6, strony 556–560
DOI: 10.5603/FC.a2018.0121

ISSN 2353–7752

Unexplained heart failure in a 14-month baby

Niewyjaśniona niewydolność serca u 14-miesięcznego dziecka

Santosh Kumar Sinha, Puneet Aggarwal, Lokendra Rekwal, Sunil Tripathi, Nishant Kumar Abhishekh

Department of Cardiology, LPS Institute of Cardiology, G.S.V.M Medical College, Kanpur, India

Abstract

Endocardial fibroelastosis is a cardiomyopathy rarely seen in the present age. Here, we report a case of 14-months old baby who presented with failure to thrive, sweating while feeding, a dilated left ventricle, and left ventricular dysfunction. Chest X-ray revealed cardiomegaly with pulmonary venous hypertension. Electrocardiogram showed left ventricular hypertrophy with strain pattern. Echocardiogram revealed dilated left ventricle with severe systolic dysfunction which was labelled as endocardial fibroelastosis. As there were no associated lesions, it was labelled as primary endocardial fibroelastosis (pEFE). The baby was managed conservatively. Here, we would like to highlight the fact that primary endocardial fibroelastosis (pEFE) can masquerade as idiopathic dilated cardiomyopathy. Progressive ventricular dilation and ventricular dysfunction portends poor prognosis, though few of them may be managed with conservative treatment.

Key words: cardiomyopathy, primary endocardial fibroelastosis, systolic dysfunction, dilated cardiomyopathy

Folia Cardiologica 2018; 13, 6: 556–560

Introduction

Endocardial fibroelastosis (EFE) is characterized by diffuse thickening of the ventricular endocardium and by associated myocardial dysfunction. It can be primary (idiopathic) or secondary to structural heart diseases [1]. In infancy or early childhood, this progressive process usually results in heart failure. It is primarily a disease of infancy and early childhood, with rare occurrences in young adulthood. On the basis of left ventricular size, primary EFE can present in the more common dilated form or the rarer contracted form (with restrictive physiology).

Case report

A 14-months old baby presented with tachypnea, excessive sweating, irritability and failure to thrive. His pulse rate was 148/min, regular, normovolemic and all pulses were palpable without radio-radial or radio-femoral delay. His weight was 3.8 kg. Tachypnea while feeding and grunting respiration with subcostal and intercostal retractions were also noted. He had received all vaccines as per immunization schedule. Developmental milestones were normal as per his age. On palpation, cardiac impulse was hypodynamic. On auscultation, S1 and S2 were faint with loud P2 component, a gallop rhythm with an audible third heart sound were audible. Routine haemogram was normal except mild anaemia. Chest X-ray showed cardiomegaly with congested pulmonary vascularity (Figure 1). Electrocardiogram showed normal axis, tall R-waves, deep Q-waves, and T-wave inversion in inferior and left precordial leads, suggestive of left ventricular hypertrophy with strain pattern (Figure 2). Two-dimensional transrthoracic echocardiography showed spherical left ventricular enlargement, generalized hypokinesia with systolic dysfunction having an ejection fraction of 30%. Septal and free wall were

Address for correspondence: Santosh Kumar Sinha MD, FAESC, Asst. Professor, Department of Cardiology, LPS Institute of Cardiology, G.S.V.M. Medical College, G.T. Road, Kanpur, Uttar Pradesh 208002, India, fax +91 0512 255 61 99/255 65 21, e-mail: fionasan@rediffmail.com

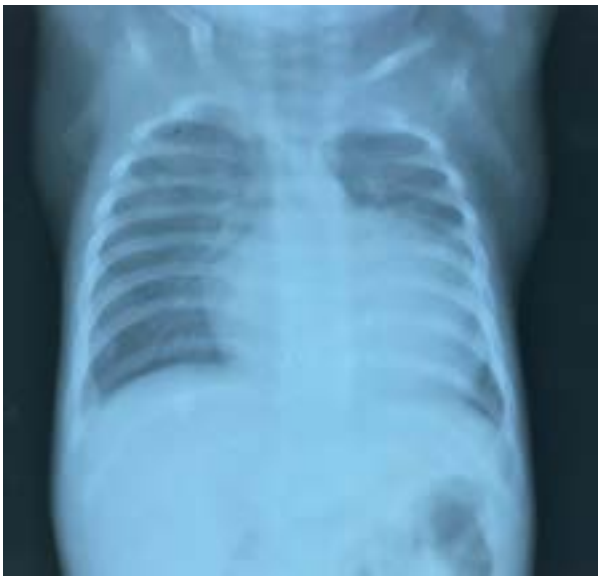

Figure 1. Chest radiograph, anteroposterior (AP) view, showing cardiomegaly and pulmonary venous congestion in a 14-month old baby with primary endocardial fibroelastosis (pEFE)

thickened due to left ventricular hypertrophy. Distinctive bright echoes originating from ventricular endocardium were also noted. Valvular echoes were normal and shunt lesions were ruled out. There was no left ventricular outflow obstruction (Figure 3). He was therefore diagnosed as primary endocardial fibroelastosis (pEFE). He was discharged with digoxin, diuretics, and hydralazine (3 mg/kg/day in four divided doses) in stable condition with appropriate follow up advice.

Discussion

The term endocardial fibroelastosis (EFE) was introduced by Weinberg and Himmelfarb in 1943 [1]. It is characterized by an opaque, pearly-white thickening due to proliferation of collagen and elastic fibres resulting into a pronounced, diffuse thickening of the ventricular endocardium resulting into myocardial dysfunction. Isolated endocardial fibroelastosis resides in the endocardium of a dilated hypertrophied left ventricle, which is also known as **primary endocardial fibroelastosis of the dilated type**. It can be secondary to various congenital heart diseases, most notably hypoplastic left heart syndrome, aortic stenosis, or atresia, patent ductus arteriosus, coarctation of aorta, and anomalous left coronary artery from pulmonary artery [2–5]. Among the two pathological types of primary endocardial fibroelastosis *i.e.* dilated, and restrictive, former is more common. Once regarded as a common cause of unexplained heart failure, endocardial fibroelastosis is now exceedingly rare.

Fibroelastosis per se is a response to a variety of endocardial stimuli, with intrauterine endocardial injury as the common denominator. It may occur in infants and adults after myocardial infarction which underscores endocardial response to injury. The endocardial thickening is believed to be caused by persistent and increased wall tension in the ventricles, possibly secondary to damaged myocardium, mitral regurgitation, or both. However, endocardial fibroelastosis changes are progressive with age. Primary EFE is the type that occurs in infants. Beyond infancy, it is patchy and associated with myocardial fibrosis. The disease is usually sporadic, but familial cases have been reported (10%).

Dilated endocardial fibroelastosis is characterized by a markedly enlarged globular heart, mainly involving the left ventricle (LV) and left atrium (LA). The normal thickness of the E of the LV is said to be 0.02 mm at the outflow tract and 0.01 mm at the inflow tract [6]. The LV endocardium is opaque, glistening, milky white, and diffusely thickened to about 1–2 mm. The thickening is most marked in the outflow tract. Papillary muscles and trabeculae carneae are flattened and partially incorporated in the fibrotic process, giving a smooth appearance to the lining of the cavity. They exert an undesirable lateral traction on the chordae tendinae and mitral cusps, leading to faulty leaflet opposition

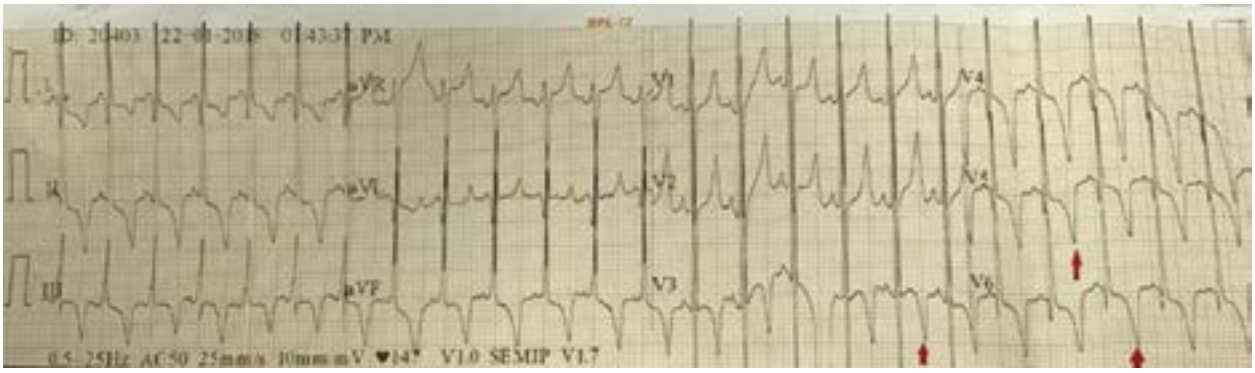

Figure 2. Electrocardiogram showed tall R-waves, deep Q-waves, and T-wave inversion in inferior and left precordial leads, suggestive of left ventricular hypertrophy with strain pattern

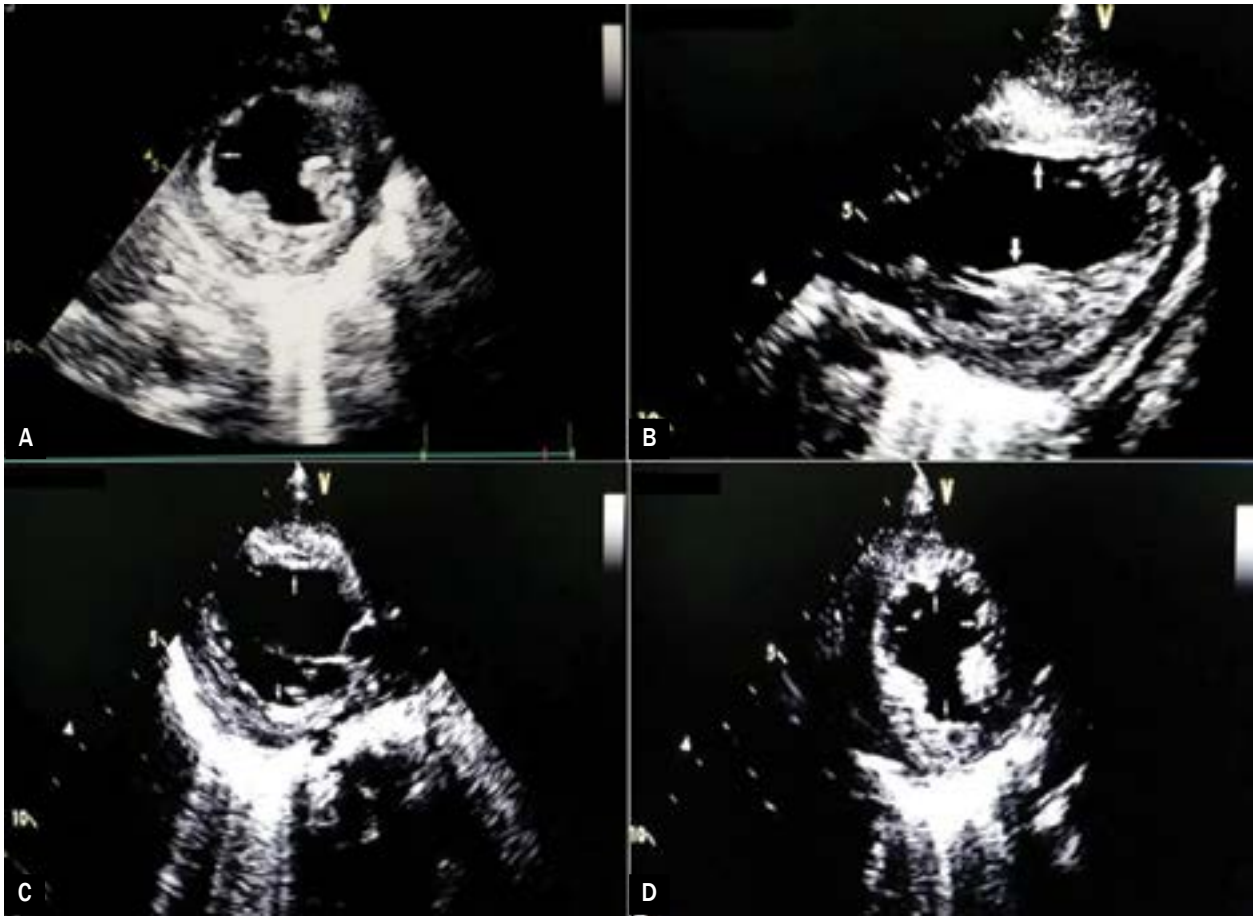

Figure 3A–D. Spherical left ventricular enlargement, with thickened septal and free wall. Distinctive bright echoes originating from ventricular endocardium were also noted (white arrow)

resulting into mitral incompetence [6]. Although the endocardium is thickened, the ventricular wall (myocardium) thickness is within the reference range. Endocardial fibrosis acts as a substrate for mural thrombosis, setting a stage for systemic emboli. In approximately 50% of patients, the mitral and aortic valves are involved, often producing marked deformity and either valvular regurgitation or stenosis, although aortic incompetence is very rare. The less common contracted type of primary endocardial fibroelastosis is associated with a relatively hypoplastic or normal LV size. The right and left atria and the right ventricle are markedly enlarged and hypertrophied, with minimal or no endocardial sclerosis.

Infants who present with acute failure almost always die from the acute episode unless they receive a transplant. Patients with a chronic presentation have a 30–40% mortality rate due to resistant heart failure. Contracted endocardial fibroelastosis has a grave prognosis and is generally fatal. Clinical diagnosis of pEFE is difficult because clinical symptoms, electrocardiographic findings, and chest X-ray are usually nonspecific. Echocardiogram by showing characteristic endocardial enhancement may be diagnostic, although may not be always seen [8]. The ability of myocardial delayed-enhancement (MDE) by cardiac magnetic resonance imaging (MRI) technique to image EFE has been reported [8, 9]. Maredia et al. [9] performed MDE cardiac MRI on a 17-year old patient with the diagnosis of primary EFE where they demonstrated high-signal intensity in ventricular endocardium. In patients with secondary EFE, cMRI demonstrated hyper enhancement involving the endocardial surface of the LV and a hypo intense layer at the endocardial surface in the perfusion sequence as shown by Stranziger et al. [8] This enhancement is thought to be because EFE is avascular; hence, it will appear as a low signal in the perfusion sequence [8]. Death usually results from heart failure within weeks, usually within the first 6 months of life and those who manage to survive, more chronic course is common as in our case. Such patients respond to medications used to treat congestive heart failure. Remissions may rarely be seen with intensive medical management. Occasionally, cardiac arrhythmias in form of electrical storm may be seen [3]. Early diagnosis and prompt persistent administration of digitalis may result in clinical improvement and reversion of the cardiac

enlargement (CE) to normal. Early and prolonged treatment with digoxin is suggested. Continue therapy for several years after the symptoms disappear; cessation of drug administration may result in acute cardiac failure, even when heart size has returned to normal. Other measures for acute failure and exacerbations of failure may be required, and precipitating factors, such as infection and anaemia, require attention. Anticoagulation may be required in the presence of thromboembolic complications.

Recently, surgical endocardial resection has been performed with left ventricular rehabilitation for patients with borderline hypoplastic left heart syndrome and secondary EFE [8–12]. The thick endocardial layer in EFE is thought to cause mechanical impairment of fluid–structure interaction and prevent LV growth. Of note, endocardial resection improved LV systolic and diastolic performance and relieved the mechanical impairment of myocardial function. Recurrence of EFE within the previous resection fields did not occur. If endocardial stripping can result in improved function in these structurally malformed ventricles, it is reasonable to speculate that the same therapeutic benefit might occur in the patients with primary EFE without a hypoplastic ventricle. Besides potential therapeutic interventions, making the diagnosis of EFE clinically would lead to a better understanding of the genetics and natural history of this underappreciated entity.

Our case is unusual as pEFE in an older child is rarely seen in the era of widespread vaccination. Cases of EFE from mumps infection in-utero have become so rare, due to the almost universal use of the MMR (measles, mumps, and rubella) vaccine, that lack of familiarity and very low probability virtually exclude this diagnosis from consideration. Also, cardiac echocardiogram is a sensitive test for EFE. Though pEFE is rare, one need to be cognizant that it can present as an idiopathic dilated cardiomyopathy.

Conflict(s) of interest

The authors declare no conflict of interest.

Streszczenie

Fibroelastoza wsierdzia jest kardiomiopatią rzadko występującą w XXI wieku. Przedstawiono przypadek 14-miesięcznego dziecka, u którego stwierdzono upośledzenie rozwoju, pocenie się podczas karmienia, powiększenie i dysfunkcję lewej komory serca. Badanie radiologiczne klatki piersiowej ujawniło kardiomegalię z tętniczym nadciśnieniem płucnym. W elektrokardiogramie stwierdzono zmiany typowe dla przerostu lewej komory serca. Badanie echokardiograficzne uwidoczniło poszerzoną lewą komorę z ciężką dysfunkcją skurczową, co wskazywało na fibroelastozę wsierdzia. W związku z tym, że nie stwierdzono innych zmian, uznano, że jest to pierwotna (idiopatyczna) fibroelastoza wsierdzia (pEFE). Zastosowano leczenie zachowawcze. Należy podkreślić, że pEFE może imitować idiopatyczną kardiomiopatię rozstrzeniową. Postępujące powiększanie się lewej komory i nasilanie się jej dysfunkcji wiążą się ze złym rokowaniem, choć niektóre takie przypadki poddają się leczeniu zachowawczemu.

Słowa kluczowe: kardiomiopatia, pierwotna fibroelastoza wsierdzia, dysfunkcja skurczowa, kardiomiopatia rozstrzeniowa

Folia Cardiologica 2018; 13, 6: 559–560

References

1. Thomas WA, Randall RV, Bland EF, et al. Endocardial fibroelastosis; a factor in heart disease of obscure etiology: a study of 20 autopsied cases in children and adults. N Engl J Med. 1954; 251(9): 327–338, doi: 10.1056/NEJM195408262510902, indexed in Pubmed: 13185424.
2. Ursell PC, Neill CA, Anderson RH, et al. Endocardial fibroelastosis and hypoplasia of the left ventricle in neonates without significant aortic stenosis. Br Heart J. 1984; 51(5): 492–497, indexed in Pubmed: 6721945.
3. Revel A, Ariel I, Rein A, et al. Fetal endocardial fibroelastosis. J Clin Ultrasound. 1994; 22(5): 355–356, doi: 10.1002/jcu.1870220513.
4. Sharland GK, Chita SK, Fagg NL, et al. Left ventricular dysfunction in the fetus: relation to aortic valve anomalies and endocardial fibroelastosis. Br Heart J. 1991; 66(6): 419–424, indexed in Pubmed: 1837727.
5. Lurie PR. Endocardial fibroelastosis is not a disease. Am J Cardiol. 1988; 62(7): 468–470, doi: 10.1016/0002-9149(88)90981-2.
6. Okada R. Clinicopathological study on the thickening of parietal endocardium in the adult heart. Jpn Heart J. 1961; 2: 220–255, indexed in Pubmed: 13730566.
7. Moller JH, Lucas RV, Adams P, et al. Endocardial fibroelastosis. Circulation. 1964; 30(5): 759–782, doi: 10.1161/01.cir.30.5.759.

8. Stranzinger E, Ensing GJ, Hernandez RJ. MR findings of endocardial fibroelastosis in children. Pediatr Radiol. 2008; 38(3): 292–296, doi: 10.1007/s00247-007-0707-7, indexed in Pubmed: 18172637.
9. Maredia N, English K, Greenwood J. Assessment of endocardial fibroelastosis by cardiac MRI. Can J Cardiol. 2008; 24(5): e33, indexed in Pubmed: 18464952.
10. van Son JA, van Son JA, Falk V, et al. Ross-Konno operation with resection of endocardial fibroelastosis for critical aortic stenosis with borderline-sized left ventricle in neonates. Ann Thorac Surg. 1997; 63(1): 112–116, indexed in Pubmed: 8993251.
11. Emani SM, Bacha EA, McElhinney DB, et al. Primary left ventricular rehabilitation is effective in maintaining two-ventricle physiology in the borderline left heart. J Thorac Cardiovasc Surg. 2009; 138(6): 1276–1282, doi: 10.1016/j.jtcvs.2009.08.009, indexed in Pubmed: 19819471.
12. Czosek RJ, Atallah J, Emani S, et al. Electrical dyssynchrony and endocardial fibroelastosis resection in the rehabilitation of hypoplastic left cardiac syndrome. Cardiol Young. 2010; 20(5): 516–521, doi: 10.1017/S1047951110000600, indexed in Pubmed: 20529397.