

Endocardial fibroelastosis presenting with congenital complete heart block at birth

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Introduction

Endocardia! fibroelastosis (EFE) is a rare disorder with a poor prognosis¹. It is characterised by abnormal thickening of the endocardium of one or both ventricles², often associated with other cardiac abnormalities³. It can either present with fetal bradycardia, absent heart sounds in utero or absent heart sounds in the newborn⁴. Both the obstetrician and the paediatrician should be familiar with this condition.

Case Report

A 27-year-old primipara was admitted to the Teaching Hospital, Colombo South, from the antenatal clinic (ANC) six days after her expected date of delivery. She had failed to attend her last two clinic appointments at the ANC and had been apparently normal during her previous visits.

However, on this occasion, her blood pressure was 130/90 and the fetal heart sounds were inaudible, but she insisted that she could feel regular fetal movements. An ultrasound examination of the abdomen was carried out soon after admission. This confirmed the presence of fetal movements. The fetal heart was found to be enlarged and no other abnormality was reported. There was no evidence of effusion in any of the serous cavities.

A live non-asphyxiated baby boy weighing 2.87 kg was delivered by Caesarean section. The baby was pink and active at birth, but the heart sounds were inaudible. The rest of the neonatal examination was unremarkable except for absent peripheral pulses. The Apgar score at 1 and 10 minutes were 5 and 7 respectively. This was low owing to the inaudible heart sounds. ECG showed evidence of complete heart block with a rate of 35 beats per minute. Teleradiogram of the chest showed marked cardiomegaly. There were no effusions in the pleural

or pericardial cavities. A 2D echocardiogram done 16 hours after birth revealed gross cardiomegaly with dilatation of the left ventricle. The ductus was patent and the left ventricular ejection fraction was 45%.

The haemoglobin (Hb) concentration was 17.2 g/dl, the total white cell count was $16.2 \times 10^9/L$ with N-70%, L-28% and M-2%, ESR was 4mm 1st hour, PCV, platelets, reticulocyte count, and the serum bilirubin were within the normal range for age. The TORCH screens of both the mother and the baby were normal.

The baby was treated with salbutamol, frusemide and spironolactone at 24 hours of age. Thirty-six hours after starting the above treatment, the heart sounds became audible and the heart rate rose to 50 beats per minute. He fed well and the subsequent weight gain was satisfactory. He was discharged on the same medication.

He remained well on salbutamol and diuretics but continued to have persistent heart block. The subsequent growth was rather slow with both the height and the weight being between 10-25th centiles. He had two severe chest infections complicated by cardiac failure during the next five months and on one such occasion required ventilator support. On the 3rd occasion, in addition to the chest infection complicated by heart failure, paroxysms of pallor with few jerky movements of hands and feet were also noticed. These coincided with multiple ventricular ectopics and ventricular tachyarrhythmia seen on the ECG monitor. His condition deteriorated despite vigorous treatment and he succumbed 3 days later.

Autopsy revealed the persistence of ductus with a grossly dilated heart. The endocardium of both ventricles was markedly thickened with the involvement of the papillary muscles and the chordae tendinae. The myocardium was normal. The microscopic sections done from several areas of both the right and the left ventricle showed a thick layer of collagenous fibrous tissue and elastin fibres confirmed by Vangisen Elastin stain.

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Discussion

Although no single aetiological factor is directly responsible, EFE has been shown to be associated with tissue carnitine deficiency, twin-to-twin transfusion, carbamazepine therapy and mumps or enteroviral infections in the mother during pregnancy^{5,6,7,8}. However, no obvious aetiological factor was found in this mother. Both constricted and dilated forms have been described but many authorities believe that all EFEs are secondary to a reactive process set off by diverse stresses on the myocardium^{1,9}. Our patient had the dilated form of the disease. It is also known to be associated with other structural abnormalities such as hypoplastic left heart syndrome and abnormalities of the aortic valve including stenosis³. This baby did not have any of the above abnormalities but was found to have a persistent ductus, which was not an abnormality in other reported cases of EFE. Other recognised association described with EFE is a new syndrome of hydrocephalus, cataracts, hydrops fetalis and congenital heart block^{10,11}.

This baby who was born with congenital complete heart block complicating EFE, did not show any evidence of hydrops fetalis which is among one of many recognized causes of absent heart sounds at birth⁴.

Conclusions

Emphasis should be placed on the importance of considering the possibility of congenital complete heart block in cases of fetal bradycardia or absent heart sounds and the necessity to do an ultrasound examination, if the overall clinical picture does not suggest fetal distress or fetal death.

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