A 12-year-old girl of poor socioeconomic status from the northern part of India presented with insidious onset facial puffiness, pedal edema, and increasing abdominal girth of 3-year duration. During the past 6 months, she had 3 episodes of spontaneous onset fast irregular palpitations associated with transient loss of consciousness. Physical examination of the grossly malnourished child revealed severe pallor, bilateral pitting pedal edema up to the ankle, and the absence of cyanosis or clubbing. Cardiovascular system examination revealed elevated jugular venous pulse with prominent A wave, prominent precordial pulsations in the left parasternal area in the third and the fourth intercostal space, and apical impulse in the left fifth intercostal space 11 cm lateral to midsternal line with normal size and contour. Right heart border could be percussed in right parasternal area 9 cm lateral to sternal border; auscultation was unremarkable except for the presence of right ventricular (RV) third heart sound and short systolic murmur in tricuspid area. Abdominal examination revealed massive ascites with fluid thrill and hepatomegaly. To summarize, clinical impression was of a cachexic child with chronic right heart failure, massive ascites, hepatomegaly, cardiac enlargement, and ectopic pulsations in RV outflow tract area (Figure 1A). Electrocardiogram (Figure 1B) showed sinus rhythm, right-axis deviation, and qr in V1 suggesting right atrial enlargement and nonspecific ST/T changes. Chest x-ray (Figure 1C) showed massive cardiomegaly with grossly enlarged right heart border. Blood investigation showed severe anemia (hemoglobin, 6.8 g %) and eosinophilia (absolute eosinophil count of 531 per mm³). Transthoracic echocardiographic examination revealed congested inferior vena cava with partial inspiratory collapse, obliteration of apex, and most part of inflow tract of RV by dense fibrous tissue with echobright endocardial border and apical dimple, leaving behind tiny residual RV inflow cavity (Figure 2A–2C; Movie I in the Data Supplement). In parasternal short-axis view (Figure 2D; Movie II in the Data Supplement), huge right atrium, dilated RV outflow tract, and relatively small pulmonary arteries could be related to body, head, and trunk of elephant, respectively (elephant appearance). Right atrium was humongously dilated with dense spontaneous echo contrast and large adherent thrombus (Figure 3A; Movie III in the Data Supplement). Pericardial effusion was present posterior to RV. Color Doppler examination (Figure 3B; Movie IV in the Data Supplement) revealed low-velocity significant tricuspid regurgitation. Restrictive filling pattern was noted across tricuspid valve with tricuspid E/A ratio >2, deceleration time <140 ms, and reduced septal and lateral E’ on tissue Doppler imaging (Figure 4D; Figure I in the Data Supplement). Diastolic forward flow into pulmonary artery, driven by atrial systolic contraction, was demonstrated on M-mode color Doppler echocardiography and continuous-wave Doppler imaging (Figure 4A and 4B; Movie V in the Data Supplement); similarly, diastolic pulmonary valve opening was seen on pulmonary valve M-mode echocardiography (Figure 4C). Left atrium and ventricle were spared with normal left ventricular dimension, ejection fraction, diastolic function, and no mitral regurgitation. Cardiac magnetic resonance (Figure 5A and 5B) demonstrated similar findings with characteristic obliteration of RV apex and inflow tract with apical dimpling and sparing of left ventricle. On the basis of classical clinical and echocardiographic findings, diagnosis of isolated RV endomyocardial fibrosis was confirmed, and the child was managed with vigorous decongestive treatment and anticoagulation. In the absence of nearly any functional RV cavity, heart transplantation was the only distant curative option left for the patient. Palliative surgical options and high surgical risk of death and complication were discussed with parents, and they choose to continue medical management.

Discussion

Since endomyocardial fibrosis was first reported by Jack N.P. Davies in Uganda in 1948, it has been known to be prevalent in tropical countries with a geographical distribution within 15° on either side of the equator, in underserved areas of...
sub-Saharan Africa, Latin America, and south India, particularly, the coastal belts of Kerala. However, India has recently witnessed considerable decline in incidence of endomyocardial fibrosis, probably because of decline in childhood malnutrition, infections, and parasite-associated eosinophilia. Although the origin of this restrictive cardiomyopathy is mystery, it is more common in populations of low socioeconomic status with protein malnutrition, worm infestation, and filarial infection with eosinophilia. Recently, the role of toxins, such as serotonin in plantain, cerium in tapioca, and magnesium deficiency, has been evaluated. Pathologically, sub-endocardial fibrosis affects the apices and the inflow tract of the RV or left ventricle, or both, papillary muscles tethering causing atrioventricular valve regurgitation. Outflow tracts are unaffected, and atria are grossly dilated with contained thrombi denoting stagnation and atrial fibrillation. Davies described 3 phases of the disease; first is an acute carditis phase that often goes unrecognized, followed by subacute and final chronic burnt out phase. The characteristic clinical feature of RV endomyocardial fibrosis is triad of raised jugular venous pressure, ascites, and hepatomegaly. Ascites is often out of proportion to pedal edema because of concomitant protein loosing enteropathy. Progressive atrial dilation leads to atrial arrhythmia, commonly atrial fibrillation. Atrial contraction augments the forward flow in pulmonary arteries; however, once atrial fibrillation sets in, right-sided chambers act as passive conduit, and pulmonary artery flow is based on principles of Fontan physiology that is elevation of mean systemic venous pressures above the pulmonary artery diastolic pressures. Chest x-ray usually shows cardiomegaly because of RA enlargement or pericardial effusion; rarely, endocardial calcification may be seen. ECG usually demonstrates low-voltage QRS complex, QR in V3R or V1, suggesting RA enlargement, nonspecific ST-T changes, and occasionally, atrial fibrillation. An echocardiography-based criterion has been developed for community screening and also for assessing the severity of endomyocardial fibrosis. Expensive and invasive investigations, such as magnetic resonance imaging and cardiac catheterization for hemodynamic studies, are reserved for research purposes only. Palliative surgical correction (RV endocardectomy and tricuspid valve repair/replacement) is indicated in all symptomatic patients and carries high-operative mortality and morbidity. The disease overall has poor prognosis with high mortality and morbidity related to refractory heart failure, cardiac cirrhosis and hepatic failure, thromboembolism, and arrhythmias. To conclude, isolated severe RV endomyocardial fibrosis is rare in childhood. Bedside clinical evaluation along with echocardiography remains the confirmatory diagnostic tool; however, overall prognosis of the disease remains dismal despite advancement in medical and surgical care.

Disclosures

None.

References


Key Words: echocardiography ▪ endomyocardial fibrosis ▪ heart atria ▪ pulmonary artery ▪ thrombosis
Figure 1. A. Clinical photograph showing cachexic child with massive ascites, which was out of proportion to the other signs of systemic venous congestion such as pedal edema. B. Chest x-ray pulmonary artery view showing massive cardiomegaly with hugely enlarged right atrium. C. Twelve-channel ECG showing sinus rhythm, right-axis deviation, qr in V1, and nonspecific ST/T changes.
Figure 2. A, Apical 4-chamber view showing humongous right atrium (RA), near complete obliteration of right ventricular (RV) apex and inflow tract, and spared left atrium (LA) and left ventricle (LV) with pericardial effusion (PE). B, M-mode echocardiography in parasternal long-axis view showing dilated RV outflow tract (RVOT) and normal LV dimensions. C, Parasternal long-axis view showing dilated RVOT with spared LA and LV. D, Parasternal short-axis view showing huge RA, dilated RVOT, and relatively small pulmonary artery (PA) giving appearance of elephants (trunk and head).

Figure 3. A, Echocardiography showing large adherent thrombus in right atrium. B, Color Doppler showing significant tricuspid valve regurgitation.
Figure 4. A, Color M-mode across pulmonary artery showing diastolic forward flow into pulmonary artery (PA; arrowheads). B, Continuous-wave Doppler imaging across PA showing diastolic forward flow into PA (arrowheads). C, Pulmonary valve M-mode echocardiography showing diastolic pulmonary valve opening (arrowheads). D, Pulsed wave Doppler across tricuspid valve showing restrictive filling pattern with E/A ratio >2 and deceleration time <140 ms.
Figure 5. A, Cardiac magnetic resonance imaging (MRI) steady state free precession sequence 4-chamber view shows hugely dilated right atrium (RA) and near complete obliteration of right ventricular (RV) cavity with apical dimple and pericardial effusion. B, Cardiac MRI (sagittal view) showing dilated RV outflow tract (RVOT). LA indicates left atrium; LV, left ventricle; PA, pulmonary artery; and PE, pericardial effusion.
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Supplementary image showing reduced tricuspid annulus lateral E’ on tissue Doppler imaging.

**Movie Legends**

Movie 1: Apical four chamber view showing humangous RA, near complete obliteration of RV apex and inflow tract, spared LA and LV with pericardial effusion.

Movie 2: Parasternal short axis view showing huge RA, dilated RVOT and relatively small PA giving appearance of “elephants” trunk and head.

Movie 3: Parasternal short axis view showing dense spontaneous echo contrast and large adherent thrombus in RA.

Movie 4: Colour Doppler showing significant low velocity tricuspid valve regurgitation.

Movie 5: Parasternal short axis view showing diastolic forward flow into PA.