

Isolated left ventricular hypoplasia - A singularity

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ABSTRACT

Isolated left ventricular hypoplasia is an entity with uncertain natural history and etiology. The presentation could vary from being asymptomatic to sudden death. This form of cardiomyopathy has been reported in infants as well as in adults. This case report aims to alert physicians to this diagnosis and the undeniable advantage of cardiac magnetic resonance.

Keywords: Cardiomyopathy, fatty left ventricular apex, isolated left ventricular hypoplasia, septal bowing

INTRODUCTION

We present a case of an under-recognized form of cardiomyopathy with unique features on cardiac magnetic resonance imaging (MRI). A PubMed search of this entity reveals just about 20 reports so far. We describe this disease in a child who continues to be on our follow-up.

CASE REPORT

Our patient was a 2-year-old male child who presented with dyspnea. The child had no previous significant medical or surgical history. The child was first born with no family history of cardiac disease. Clinical examination was unremarkable. There was no evidence of any arrhythmia. Complete blood count was within normal limits. An echocardiography was done which revealed trabeculated left ventricular (LV) walls and septal bowing toward the right ventricle (RV). The situs and atrioventricular and ventriculoarterial concordance was normal. The systemic and pulmonary venous returns were also normal. The biventricular contractility was

normal. With uncertainty of the diagnosis, a cardiac MRI was performed under sedation.

Right and left atria showed normal morphology. The LV had abnormal morphology with a rounded, blunted apex with the RV forming a “sleeve-like” projection around the LV apex. Papillary muscles appeared small and abnormal in morphology with origin from the flattened anterior part of the LV apex. Interventricular septum appeared to be bowed to the right [Figures 1 and; Video 1. Half-Fourier single-shot turbo spin-echo (HASTE) images revealed hyperintense signal in the LV apical wall, suggesting fatty infiltration [Figure 3 and Video 2]. The child was managed conservatively with enalapril, carvedilol and low dose furosemide and is now on regular follow up.

DISCUSSION

A recently recognized type of cardiomyopathy – isolated left ventricular apical hypoplasia – was first identified in 2004 by Fernandez-Valls *et al.* in a case series.^[1] In 2007, the diagnosis was reported in a child.^[2] According to the cases fitting the criteria of isolated left ventricular

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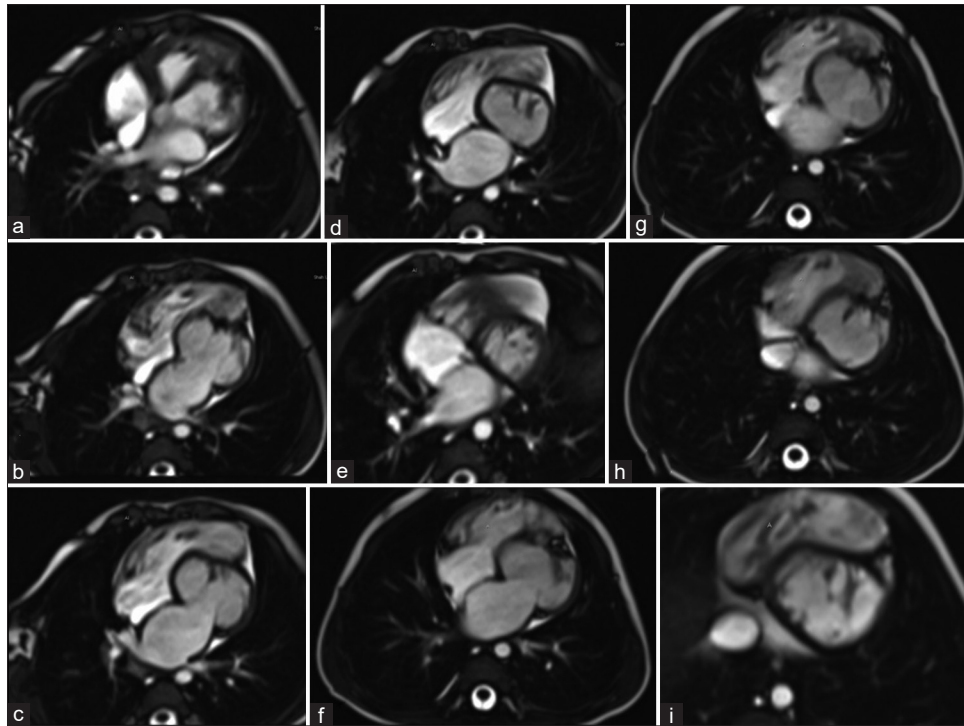


Figure 1: Four-chamber Cardiac Magnetic Resonance (CMR) still images stack of one cardiac cycle (a-i) revealing a truncated left ventricular apex with spherical configuration of left ventricular chamber (b-d). Elongated right ventricle with extension anteriorly along the left ventricular apex (d-g). Stunted papillary muscle originating from the blunt left ventricular apex (c and d). Bulging interventricular septum toward right ventricle (b-d)

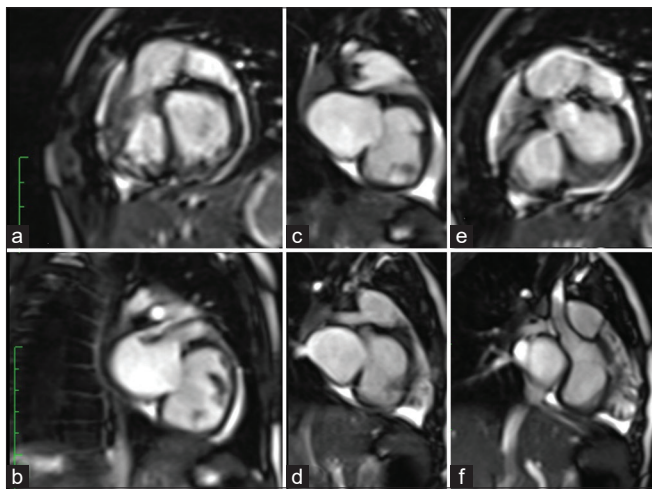


Figure 2: Left ventricular outlet and left ventricular long-axis views (a-f) depicting a spherical left ventricle (b and c) with right ventricular extending in a sleeve-like fashion along apex (d and f)

hypoplasia (ILVH) reported so far in the literature, the minimum age was 3 months while the maximum was 66 years. A literature review reported the mean age to be 29 years with no specific gender predilection.^[3]

Patients may be asymptomatic or have arrhythmias, murmurs, dyspnea, or heart failure.^[3] In the first report of a child with ILVH, the symptoms were fatigue, shortness of breath, and chest discomfort.^[2] However, patients may have a fulminant presentation as well.

A 19-year-old patient who presented with arrhythmia, fulminant heart failure, and refractory pulmonary hypertension was diagnosed as ILVH and died shortly after presentation.^[4]

This anomaly may isolated or have associations such as Patent Ductus Arteriosus (PDA).^[3]

The morphologic criteria of ILVH have been described as:

- A truncated and spherical LV with abnormal diastolic or systolic function, with exaggerated rightward bulging of the interventricular septum during diastole
- Invagination of fatty material into the myocardium of the defective LV apex
- Abnormal origin of a complex papillary network from the flattened apical LV
- An elongated RV wrapping around the deficient apex.^[1,3]

ILVH has a wide differential diagnosis including both congenital and acquired conditions. The prime differential is hypoplastic left heart syndrome. The etiology of hypoplastic left heart syndrome (HLHS) has been ascribed to genetic influences or secondary to diminished flow to the developing LV. As a result, there is interrupted growth of the LV, aorta, and aortic valve. Also are associated dysplastic cardiac valves.^[1,5] The limitation of involvement to LV apex and sparing of aorta and aortic valve helps establish the diagnosis of ILVH.^[6]

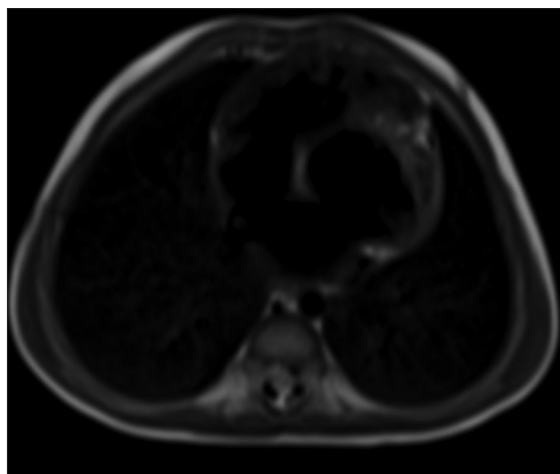


Figure 3: Axial HASTE image showing hyperintensity involving left ventricular apex suggesting adipose deposition

Another close differential is LV noncompaction cardiomyopathy. It is a result of defective morphology of the endomyocardium, whereby the ventricle is dilated with marked trabeculations.^[1] A ratio of noncompacted to compacted myocardium of more than 2.3 at cardiac MRI is diagnostic of noncompaction cardiomyopathy (CMP).^[6] ILVH may have a heavily trabeculated LV wall. Ancillary findings such as apical truncation and an elongated RV free wall that wraps around the LV point toward ILVH as in our case [Figure 1]. In addition, LV apical myocardium may be replaced with fat with contiguity of epicardial fat.^[1,4,7]

Ventricular diverticulum and ventricular aneurysms are usually severe, mostly fatal conditions. Patients present early in life due to associated intracardiac and extracardiac defects.^[1,5] A mirroring pathology is isolated RV hypoplasia characterized by RV under-development with the absence of its trabeculated apex with diastolic dysfunction.^[1] Progressive fibrofatty replacement of myocytes is pathognomonic of arrhythmogenic RV dysplasia. Although typically involves the RV, LV involvement is also known. However, only few cases of isolated LV involvement have been reported. The fibrofatty proliferation in patients with ARVD is not limited to the apex, however.^[6] An acquired differential includes Chagas disease which may cause fibrosis and LV wall thinning with the development of apical aneurysms and generalized ventricular dilatation.^[1]

Theories have been proposed about potential mechanisms of this anomaly. Presumably congenital, it may be a result of abnormal septation of the ventricles during the 5th week. The LV remains spherical due to inadequate dilatation.^[1,3] However, there is possibility of an acquired etiology like *in utero* infection. A case describing mid wall fibrosis in ILVH raises this prospect.^[7]

Management is controversial. Most of the cases have been managed conservatively with the treatment of arrhythmia or heart failure. Clinical follow-up in reported cases is limited with the longest follow-up been described

for 5 years.^[3] The potential use of heart or heart and lung transplantation has been suggested in severe cases complicated by pulmonary hypertension.^[6] Perhaps, patients with ILVH may not present till adulthood. However, children with the diagnosis need to be monitored for progressive heart failure, pulmonary hypertension, and arrhythmias, with a proposed lifetime follow-up.^[2-4]

CONCLUSION

ILVH is a rare entity with limited data. Clinicians and radiologists need to be aware of this presumably congenital diagnosis as the only way to understand the natural history of this disease is prompt recognition. A truncated, spherical LV apex, elongated RV, and septal bulging on echocardiography should cue cardiac MRI.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient's parents have given his consent for his images and other clinical information to be reported in the journal. The patient's parents understand that his names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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