Echocardiographic characterization of left ventricular apical hypoplasia accompanied by a patent ductus arteriosus

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Left ventricular (LV) apical hypoplasia is an unusual, recently identified cardiomyopathy, whose clinical course is uncertain. In this report, we describe a case of this cardiomyopathy occurring in an asymptomatic 50-year-old male with a remote history of a surgically corrected patent ductus arteriosus (PDA), primarily using transthoracic echocardiography (TTE) to illustrate the imaging characteristics. This patient had been referred to our institution for an abnormal electrocardiogram, and TTE subsequently (Figure 1) revealed a dilated left ventricle with moderately to severely reduced function; LV ejection fraction was 30% by two- and three-dimensional quantification. The left ventricle had a spherical appearance with a thin-walled, truncated, and akinetic distal LV. The right ventricle appeared elongated and was noted to wrap around the distal left ventricle, but right ventricular systolic function was normal. There were no significant valvular abnormalities, and no evidence of residual PDA flow. Subsequent cardiac magnetic resonance (CMR) imaging confirmed these findings (Figure 1). The TTE and CMR findings seen in this patient are consistent with LV apical hypoplasia. Until now, this cardiomyopathy has been described only as an isolated congenital anomaly primarily using CMR and cardiac computed tomography. To our knowledge, this is the first reported case of LV apical hypoplasia in conjunction with another congenital cardiac abnormality, and the findings demonstrate that the distinctive appearance of this cardiomyopathy can be easily identified with echocardiography. As more cases are recognized and patients are followed over time, the natural history and optimal treatment for this cardiomyopathy may be further elucidated.

Keywords: PDA • Echocardiography • Cardiac imaging • Cardiomyopathy

Introduction

Left ventricular (LV) apical hypoplasia is a newly characterized, unusual cardiomyopathy that has been described up to this point as an isolated congenital cardiac abnormality.1–5 The typical imaging characteristics of this cardiomyopathy include a spherical, truncated left ventricle with some degree of systolic dysfunction and an elongated, normally functioning right ventricle (RV) that wraps around the distal left ventricle. This cardiomyopathy was initially described with cardiac magnetic resonance (CMR) imaging and cardiac computed tomography (CT)1–3 and has recently been described using echocardiography,4,5 occurring as an isolated cardiac anomaly. We present a case of this cardiomyopathy in an asymptomatic adult with a remote history of a successfully ligated patent ductus arteriosus (PDA), primarily using two-dimensional (2D) transthoracic echocardiography (TTE) to demonstrate the distinctive imaging characteristics. To our knowledge, this is the first reported case of LV apical hypoplasia in conjunction with another congenital cardiac abnormality, and the findings demonstrate that the distinctive appearance of this cardiomyopathy can be easily identified with echocardiography. As more cases are recognized and patients are followed over time, the natural history and optimal treatment for this cardiomyopathy may be further elucidated.

Case report

A 50-year-old asymptomatic man was referred for evaluation of an abnormal electrocardiogram that revealed a non-specific intraventricular conduction delay with lateral T-wave abnormalities. His only cardiac history included an uncomplicated, successful ligation of a PDA at 8 months of life at which time no cardiac imaging was performed. He had experienced a normal, healthy childhood without any developmental concerns, and at the time of evaluation, he was completely asymptomatic from a cardiac perspective.
Cardiac evaluation included a myocardial perfusion stress test demonstrating excellent exercise tolerance (the patient exercised for 11 min on a Bruce protocol) with normal perfusion imaging. A TTE (Figures 1 and 2 and Supplementary data online, Movie S1) revealed a mildly dilated left ventricle with moderately to severely reduced function; LV ejection fraction (LVEF) was 30% by two-dimensional (2D) and 3D quantification. The left ventricle had a spherical appearance with a thin-walled, truncated, and akinetic distal left ventricle. The RV appeared elongated and was noted to wrap around the distal left ventricle, but right ventricular systolic function was normal. There was trace mitral regurgitation with no other significant valvular abnormalities, and pulmonary artery systolic pressure was normal. Of note, there was no evidence of residual PDA flow. The patient then underwent CMR imaging (Figure 1 and Supplementary data online, Movie S2) that confirmed a spherical, mildly dilated left ventricle with apparent hypoplasia of the LV apex. The distal RV was again noted to wrap around the thin-walled, truncated, and akinetic distal left ventricle, while quantitative right ventricular ejection fraction (RVEF) was normal at 60%. LV end-diastolic volume was mildly increased, while LV systolic function was moderately to severely reduced. There was bulging of the interventricular septum towards the RV and anteroapical displacement of the papillary muscle insertion points. There was an area of focal fatty infiltration noted in the LV apical wall that appeared to be contiguous with the epicardial fat, but no evidence of late gadolinium enhancement in either the LV or RV myocardium.

**Figure 1** Apical four-chamber view demonstrating the truncated, spherical left ventricle with the distal right ventricle wrapping around the deficient apical area (A). The corresponding cardiac magnetic resonance (CMR) view is shown for correlation (B). LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle.

**Figure 2** Contrast-enhanced apical four-chamber views showing the elongated, contrast-filled right ventricle wrapping around the apical area (A). The spherical left ventricle with a thin-walled distal segment is visualized (B).
There was no evidence of a residual PDA structure or other intracardiac shunt.

**Discussion**

The echocardiographic and CMR findings seen in this patient are consistent with LV apical hypoplasia, a cardiomyopathy that was first described by Fernandez-Valls et al. in 2004. To our knowledge, only eight cases of this cardiomyopathy have been described up to this point. The majority of patients identified in the literature have been adults, ranging from 19 to 46 years of age, and there have been two reports of children (3 months, 11 years) with this cardiomyopathy. With the exception of three cases (one fatal presentation of ventricular fibrillation with subsequent haemodynamic deterioration and death and two cases presenting with peripartum pulmonary oedema with subsequent atrial and ventricular arrhythmias), many of these patients have been asymptomatic or have had non-specific, mild cardiac symptoms. Given the small number of cases reported, the natural history and optimal treatment of this cardiomyopathy are unknown. Furthermore, the previously reported cases were all isolated anomalies. To our knowledge, this is the first reported case involving another congenital cardiac abnormality in the form of a PDA. In this case, there was no evidence to suggest that the PDA was still present or a contributing factor to the patient’s current condition.

The imaging characteristics of this cardiomyopathy were initially described with CMR and cardiac CT with more recent case reports illustrating 2D echocardiographic characteristics. The typical appearance is one of a truncated, spherical left ventricle with varying degrees of LV dysfunction. Fatty infiltration (on CMR or CT) of the thinned and akinetic apical wall that is contiguous with the epicardial fat has been noted in some but not all cases. Patients have been noted to demonstrate abnormal origin of the papillary muscles from the anteroapical region, an interventricular septum that bulges towards the right, restrictive haemodynamics, and varying degrees of mitral regurgitation. The distal RV wraps around the deficient LV apex, and in the majority of patients, right ventricular systolic function is normal.

Although the pathogenesis of this cardiomyopathy is unknown, it has been proposed that it may be due to deficient LV dilation in utero.

By echocardiographic and CMR quantification, our patient’s LV function was moderately to severely reduced. The patient was treated with standard therapy for a non-ischaemic-dilated cardiomyopathy and was initiated on angiotensin converting enzyme inhibitor and beta-blocker therapy, with a plan for close follow-up.

**Conclusions**

LV apical hypoplasia is an unusual, newly characterized cardiomyopathy whose clinical course is unknown. This cardiomyopathy can occur in isolation or in conjunction with other congenital lesions, as we have demonstrated with this case. Furthermore, LV apical hypoplasia has a distinct appearance that can be easily identified on echocardiography. As more cases are recognized and patients are followed over time, the natural history and optimal treatment for this cardiomyopathy may be further elucidated.

**Supplementary data**

Supplementary data are available at *European Journal of Echocardiography* online.

**Conflict of interest:** none declared.

**References**