CASE REPORT

Biventricular isolated apical hypoplasia: First-time description of a new cardiac abnormality by multimodality imaging

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ABSTRACT

Left ventricular isolated hypoplasia is a seldom-described cardiac abnormality. Right ventricular hypoplasia is usually associated with congenital anomalies of the pulmonary or the tricuspid valve, whereas biventricular isolated apical hypoplasia has never been described. We report the case of a 48-year-old man with no history of known cardiac disease who was found to have a complex cardiac abnormality characterized by: 1) Deficiency of the myocardium within the biventricular apex with adipose tissue infiltration; 2) Truncated right ventricle because of an absent trabecular portion of the inflow tract; 3) Truncated and spherical left ventricular apex; 4) Origin of the mitral papillary muscle in the flattened anterior left ventricular apex. Multimodality imaging was performed to delineate the morphological and functional characteristics of this cardiomyopathy fully. To the best of our knowledge, this is the first description of a new cardiac abnormality characterized by the hypoplasia of the apical region of both ventricles in the absence of valvular or coronary artery disease.

Key Words: Isolated apical hypoplasia, Biventricular, Echocardiography, Computed tomography, Cardiac magnetic resonance imaging

1. INTRODUCTION

Isolated left ventricular (LV) apical hypoplasia is an unusual cardiomyopathy that was firstly described by Fernandez-Valls et al. in 2004.[1] The clinical presentation varies from asymptomatic to symptoms ranging from a minor degree of fatigue to exertional dyspnea, chest pain, and supraventricular and ventricular arrhythmias.[1–11] One fatal case has been reported, but this patient had other relevant comorbidities, including severe pulmonary hypertension.[12] This condition is usually diagnosed at cardiac computed tomography (CT) and magnetic resonance imaging (MRI) and is characterized by: (1) a truncated and spherical LV configuration with rightward bulging of the interventricular septum, (2) deficiency of the myocardium within the LV apex with adipose tissue infiltrating the apex, (3) the origin of the papillary muscle in the flattened anterior apex, and (4) elongation of the right ventricle (RV) wrapping around the deficient LV apex.[1–10]

Isolated hypoplasia of the RV is a seldom reported congenital heart disease.[13–18] Two main features of isolated hypoplasia of RV are the absence of the trabecular portion of RV and the presence of normally developed tricuspid and pulmonary valves. The degree of hypoplasia has a significant effect

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on the variations in the clinical spectrum. Severe forms of hypoplasia were reported to be seen chiefly in infancy, usually associated with cyanosis in childhood. Less extreme conditions have been rarely reported in adults. To the best of our knowledge, no cases of biventricular isolated apical hypoplasia have been reported.

2. Case presentation

A 48-year-old man with no history of known cardiac disease was admitted with cough and chest pain. Familial history was not contributory. His functional class was NYHA I. His blood pressure was 130/80 mmHg, and his pulse was 80 bpm on a physical examination. ECG (see Figure 1) showed a normal sinus rhythm, right axis deviation, and low peripheral and precordial voltage with poor R-wave progression. Two years before admission, he had undergone echocardiography because of "abnormal ECG," but significant cardiac abnormalities had been excluded. On chest X-ray in the posteroanterior view (see Figure 2A) the third left arch was poorly represented, and the whole heart had an apparent mid-chest position. Pulmonary vascularity was normal. The lateral view (see Figure 2B) showed a mild pectus excavatum and flattened posterior cardiac contour. Transthoracic echocardiography (see Figure 3A) revealed a severely reduced RV cavity, a spherical LV with apparently foreshortened apex, and mildly reduced contractility. The injection of sonicated saline from a peripheral vein confirmed the small right ventricular cavity with an absent trabecular portion and excluded atrial shunts. The injection of contrast SonoVue® (sulfur hexafluoride 1 ml bolus) excluded a foreshortened left ventricular silhouette and confirmed a spherical truncated left ventricular apex (see Figure 3B). There was mild mitral regurgitation, whereas the aortic, pulmonary, and tricuspid valves were normal. A brightly echogenic mass protruding into the left atrial cavity was seen at the level of the mitral annulus (see Figure 3C). To better delineate this mass, a transesophageal echocardiogram was performed: an echolucent structure was seen infiltrating the so-called ligament of Marshal (a normally thin ridge between the left atrial appendage and the left superior pulmonary vein), which looked significantly thickened and echolucent (see Figure 3D).

Cardiac CT confirmed that the apex of both ventricles was truncated (see Figure 4A); this aspect was better seen at volume-rendered imaging (see Figure 4B). In addition, fatty material was present in the apical pericardial sac. Neoplastic lesions infiltrating the heart were excluded.

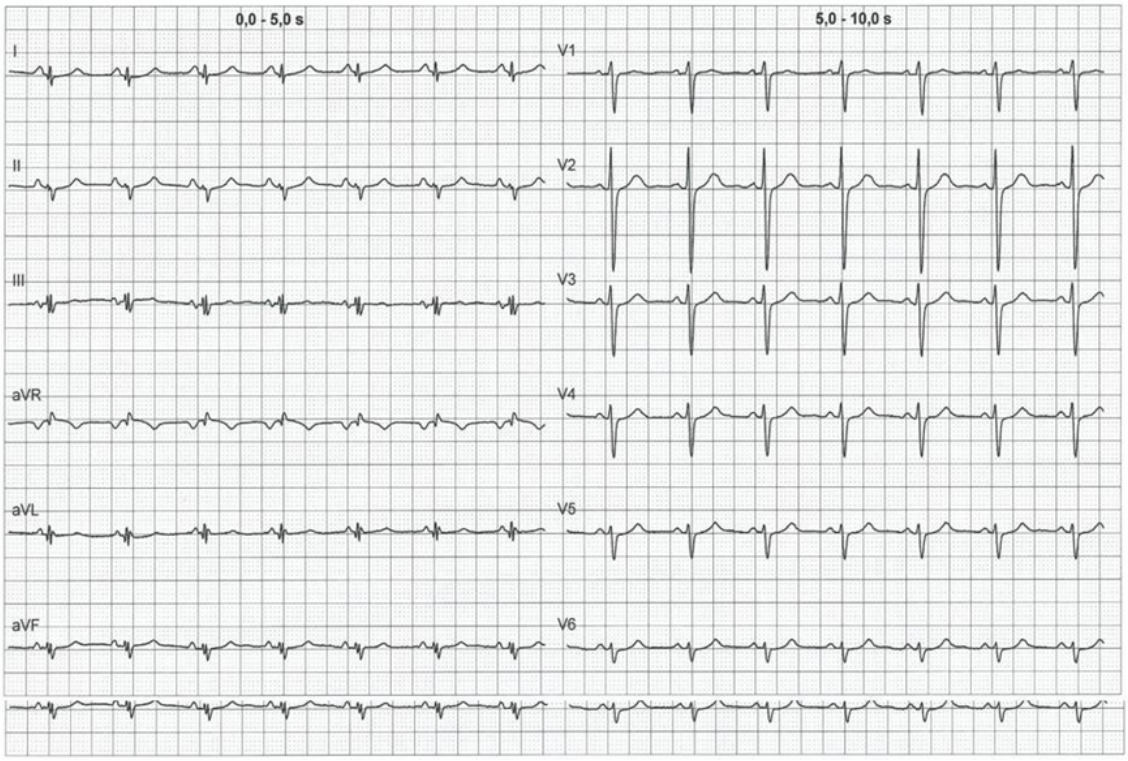

Figure 1. Presenting ECG showing low voltages and poor R wave progression

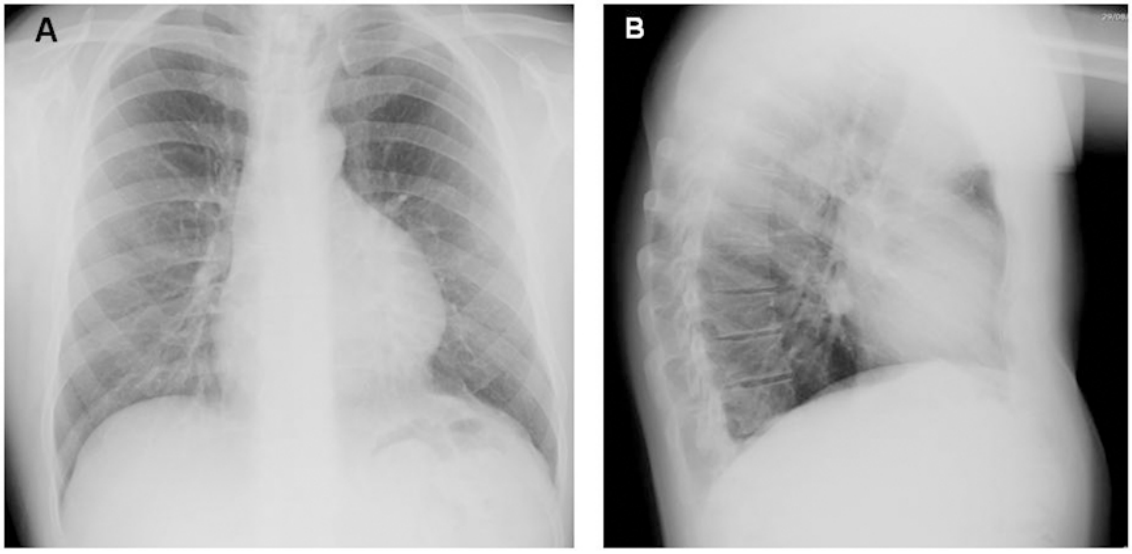

Figure 2. Chest X-ray in the postero-anterior (A) and lateral (B) view

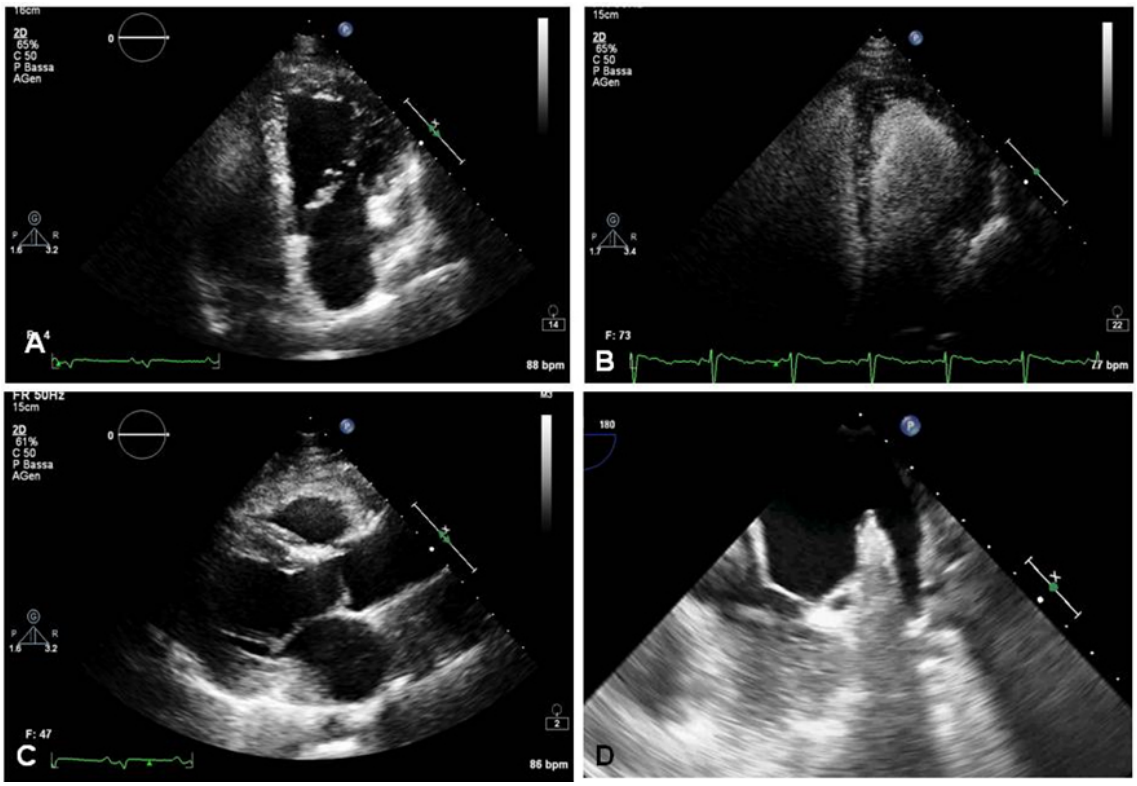

Figure 3. Echocardiography

Four-chamber view at baseline (A) and after (B) the injection of contrast showing a severely reduced right ventricular cavity with absent trabecular portion. The left ventricular apex is truncated. In the parasternal long-axis view (C) an echolucent mass is visible at the mitral annulus. Transesophageal echocardiography (D) identified this mass as a thickened and echolucent ligament of Marshall

Cardiac MRI with steady-state free precession sequences in a four-chamber view showed a severely reduced right ventricular cavity with an absent trabecular portion of the inflow tract and a truncated appearance of the left ventricular apex (see Figure 5A). Cine cardiac MRI demonstrated mild biventricular hypokinesis. A two-chamber view of the left ventricle confirmed a defective apical region, reduced ventricular cavity, and displaced papillary muscles arising from

the anterolateral area (see Figure 5B).

Black-blood T1-weighted sequences with and without fat saturation demonstrated an extended substitution of the biventricular apex by fatty material and significant fatty infiltration of both the right and left atrioventricular groove and the atrial septum (see Figure 5C); the thickened ligament of Marshall seen on echo was therefore attributed to fatty infiltration.

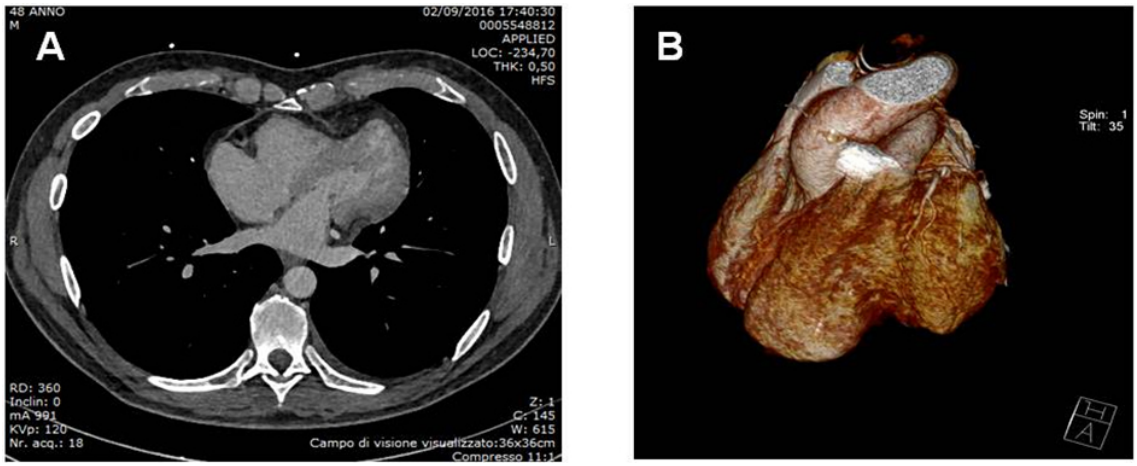

Figure 4. Computed tomography (A) demonstrating the absent right ventricular trabecular apical portion and the truncated left ventricular apex. Both anomalies are better seen in a volume-rendered image (B).

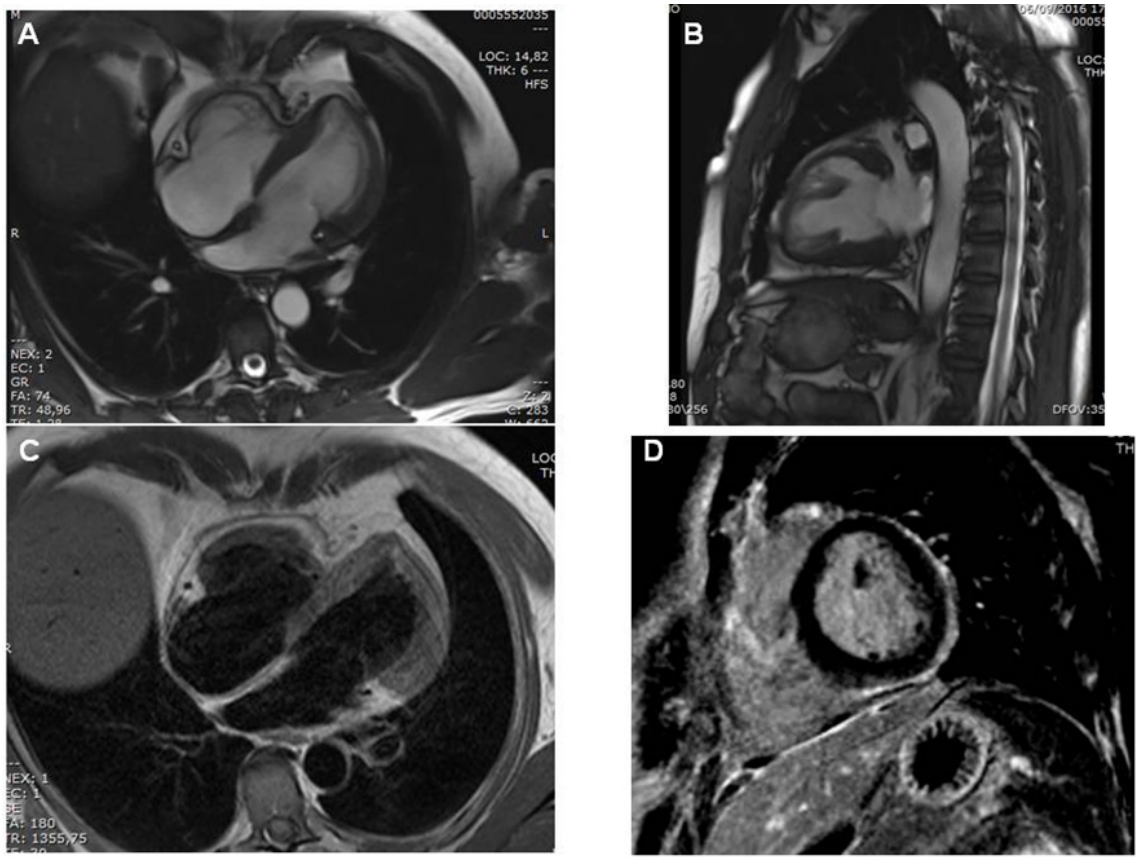

Figure 5. Magnetic resonance imaging four-chamber view (A) demonstrating the truncated biventricular apical segments, two-chamber view (B) showing the hypoplastic left ventricular apex with abnormal insertion of the papillary muscles. T1 weighted four chamber view (C) shows the massive fatty infiltration and post-contrast view (D) excludes LV myocardial fibrosis.

Finally, late gadolinium enhancement imaging was performed using a fast gradient-echo sequence and a phase-sensitive inversion recovery protocol: no areas of delayed enhancement were seen within the myocardium (see Figure 5D).

At cardiac catheterization, the filling pressure of both ventricles and pulmonary pressures were normal. No significant stenosis was evident on selective coronary angiogram. The patient was discharged with standard therapy for heart failure.

3. DISCUSSION

This is the first case of isolated biventricular apical hypoplasia reported in the literature to the best of our knowledge. Unlikely isolated left ventricular apical hypoplasia wrapping of the left ventricle by the RV and rightwards systolic bulging of the interventricular septum were not present because of the concomitant right ventricular hypoplasia. The basal and mid-portion of the ventricles were developed sufficiently to warrant normal ventricular filling pressure and cardiac output, providing a normal quality of life and were likely responsible for the missed echocardiographic diagnosis the patient had received before admission, probably because of a foreshortened apical view. Both on CT and MRI, there were prominent adipose tissue infiltrates of the apex, the atrioventricular groove, and the atrial septum. Unlikely arrhythmogenic cardiomyopathy, the fatty tissue did not infiltrate the myocardium, and areas of fibrosis were not seen on MRI. Left ventricular apical hypoplasia is postulated to be a congenital abnormality due to inadequate dilatation of chambers during partitioning.[1] In our cases, the concomitant right ventricular hypoplasia required an alternative pathogenetic mechanism. Underdevelopment of some cardiac segments and substitutive infiltration by fatty tissue seems the presumptive embryological mechanism underlying this condition; however, the primitive cause remains completely unclear. Left ventricular hypoplasia should be differentiated from dilated cardiomyopathy (diagnosed by a left or biventricular dilatation with reduced systolic function, in the absence of severe coronary artery disease or abnormal loading condition), arrhythmogenic cardiomyopathy (RV and/or LV segmental dilation and wall motion abnormalities, fibrous-fatty infiltration of the myocardium, complex ventricular arrhythmias), hypoplastic left heart syndrome (caused by aortic or mitral atresia), endocardial fibroelastosis (apically thickened myocardium, superimposed over a combination of inflammation and thrombi), apical hypertrophic cardiomyopathy (discrete hypertrophy or obliteration of the LV apex, negative giant T waves on ECG and frequent apical fibrosis on MRI). Differential diagnoses of RV hypoplasia included atretic tricuspid or pulmonic valve and right ventricular arrhythmogenic cardiomyopathy.

In conclusion, this is the first description of a new cardiac abnormality characterized by biventricular apical hypoplasia without valvular or coronary artery disease. Multimodality imaging precisely delineated the morphologic and functional characteristics of this unique cardiac abnormality. As more cases are reported, and patients followed up over time, the natural history and adequate treatment for this cardiac condition may be further delineated.

CONFLICTS OF INTEREST DISCLOSURE

The authors declare they have no conflicts of interest.

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