

Isolated Right Ventricular Apical Hypoplasia

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Introduction

Isolated hypoplasia of the apical trabecular component of the right ventricle, also called isolated right ventricular hypoplasia, is a rare condition characterized by an abnormality of the trabecular portion of the right ventricle.¹ It has an apparent family influence.²⁻⁵ Clinical manifestations are nonspecific and physical examination is often normal. Diagnosis is often determined by echocardiography, hemodynamic test or by magnetic resonance imaging.^{2,3,6,7} Treatment can be based on early intervention with systemic-pulmonary anastomosis in cases that involve cyanosis and decreased pulmonary blood flow, as well as definitive correction surgeries, which include closure of atrial septal defect, when the right ventricle (RV) is of a reasonable size or univentricular correction is conducted by Glenn or Fontan surgery.^{2,6} This report describes the case of a male patient with heart failure and echocardiographic findings consistent with isolated right ventricular apical hypoplasia.

Case Report

A.C.S, male, 44, was admitted to the emergency service with progressive dyspnea, which currently appears at rest and gets worse with physical exertion. Associated with this condition, the patient reported cough with mucoid secretion and hemoptysis, and respiratory-dependent chest pain, paroxysmal nocturnal dyspnea, orthopnea, dizziness, sweating, drowsiness, facial, abdomen and leg swelling.

On physical examination, cardiac auscultation revealed irregular heart rhythm, normophonetic heart sounds with the presence of systolic murmur in the mitral area (+2/6+) radiating to the anterior axillary line. Physiological lung and abdominal physical examination. Edema of the lower limbs (+2/4+) with depressible edema (pitting).

The patient was admitted to hospital. Protocol of admission examinations was completed. Electrocardiogram showed atrial fibrillation only.

Keywords

Heart Defects, Congenital; Ventricular Dysfunction Right/surgery; Echocardiography, Doppler.

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Two-dimensional Doppler echocardiography revealed right atrium significantly increased (indexed volume: 55 mL/m²) and hypoplasia of the apical portion of the right ventricle. RV diameter was evaluated through an apical 4-chamber view focused on the right ventricle, obtained with lateral or medial transducer orientation. During the imaging procedure, the LV apex was at the center of the scanning area, displaying the biggest basal right ventricular diameter. In this study, the RV free wall was well defined.

RV base diameter: 55 mm; diameter of the middle portion in a section focused on the RV: 43 mm; longitudinal right ventricular diameter: 49.5 mm; hypoplastic apical portion and expansion of the subpulmonary outflow: 42.5 mm (Figures 1 and 2). Systolic right ventricular dysfunction was observed through the following echocardiographic criteria: TAPSE: 15 mm, FAC: 27% and pulsed Doppler S' wave smaller than 9.5 cm/s. Systolic pulmonary artery pressure underestimated by right ventricular dysfunction, global contractilities and LV segmental preserved at rest with an ejection fraction of 63%, minimal aortic regurgitation, moderate tricuspid regurgitation, absence of distal tricuspid valve septal leaflet displacement (Figure 3), absence of thrombi and/or intracardiac masses, patent foramen ovale with small left-right flow and mild pericardial effusion.

Discussion

The first study describing isolated right ventricular apical hypoplasia was published in 1959, with the case reported by Gasul et al.⁸ In 1971, Van der Hauwaert et al.³ published a report describing the case of two new patients and reviewing the 12 cases reported to date.^{1,2} However, when the study by Hauwaert was reviewed, many patients did not present angiography and many of them presented tricuspid valve defects associated.¹ In Brazil, the first study of this disease was published in 1996 by Amaral et al.² by USP, Ribeirão Preto, with the report of two new cases diagnosed.

The right ventricle has three different components described by Goor and Lillehei:⁹ the inflow tract, including the atrioventricular valve apparatus, the trabecular (or apical) portion and the outflow tract. Right ventricular hypoplasia is characterized when one of these three components is affected. It may be associated with many different abnormalities in addition to reduced chamber size, such as pulmonary valve atresia, tricuspid valve atresia and other congenital defects, such as interatrial or interventricular septal defect.¹

The trabecular portion is responsible for the normal ventricular morphology. When an abnormality affects that portion, the right ventricle presents a reduced size. However, the tricuspid and pulmonary valves are preserved, as they originate from the other two components. An abnormality

Case Report

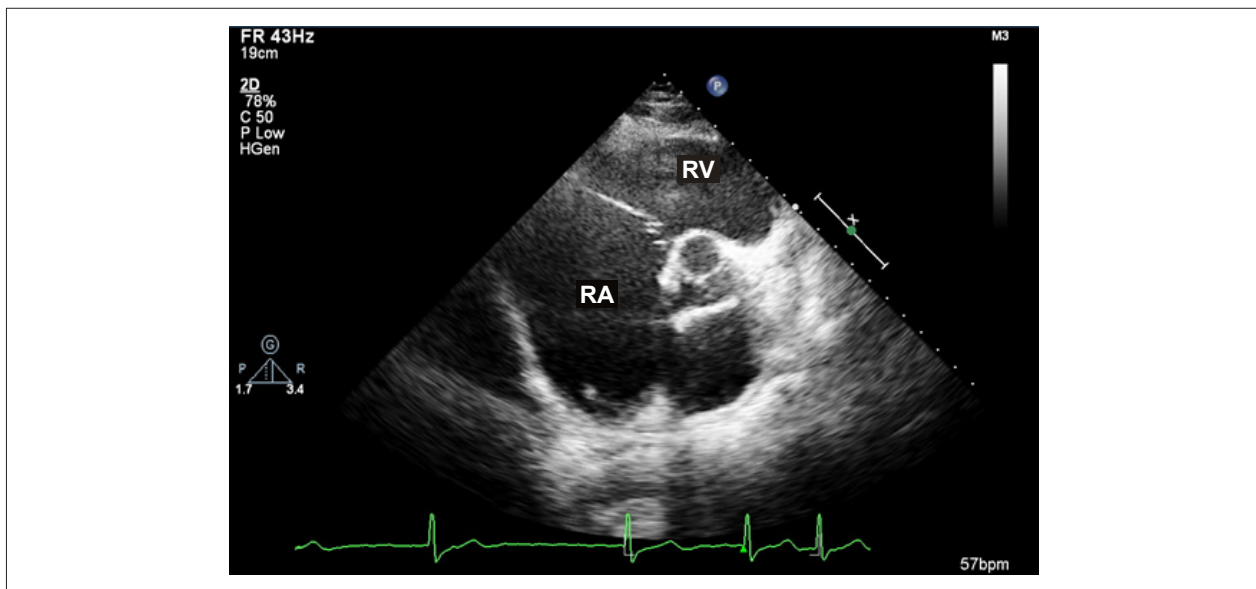


Figure 1 – Transverse parasternal view - RV apical hypoplasia. RV: right ventricle; RA:right atrium.

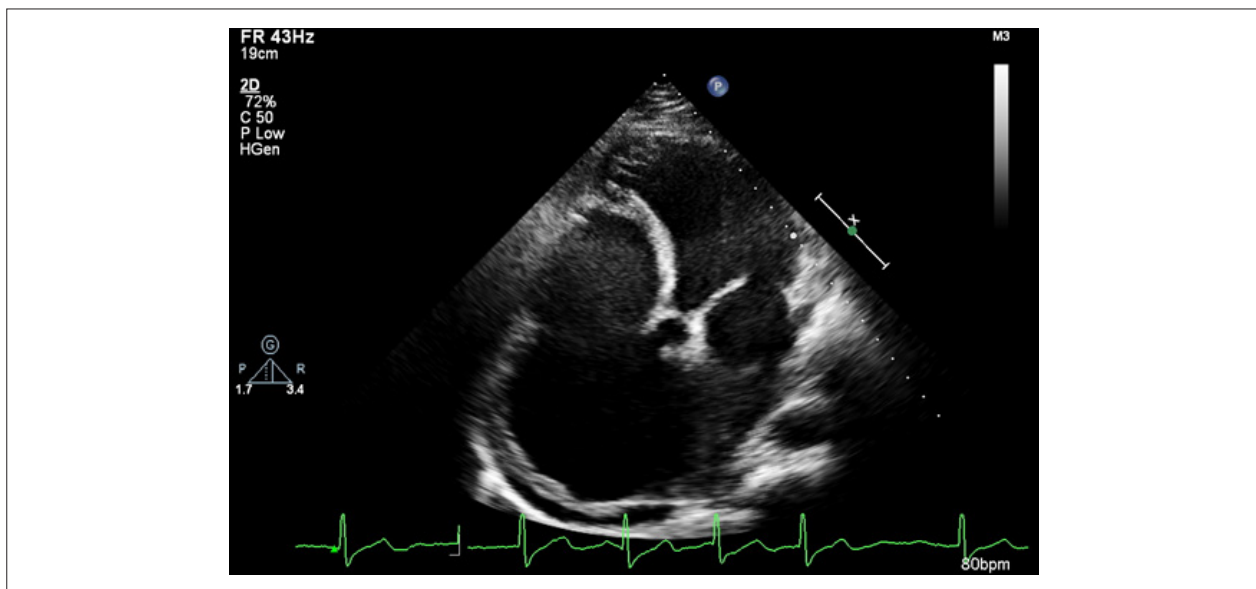


Figure 2 – Apical four-chamber view - RV apical hypoplasia.

only affecting the trabecular portion characterizes isolated right ventricular hypoplasia, a rare disease, with few cases described.¹ This structural abnormality promotes increased resistance to the entry of blood flow into the right ventricle, increasing the pressure in this heart chamber at the end of diastole and right atrium, thus requiring more work.³

This clinical entity appears to have family influence, since this correlation was presented in several cases described in the literature.²⁻⁵ There is no degree of predominance between the sexes and, because of a rare nature, there is no reliable estimate of its prevalence.^{2,3,5}

Clinical presentation of patients with isolated RV hypoplasia is highly variable and may appear less severely and not easily recognizable and more severely, requiring early palliative surgery.^{2,3} Congestive heart failure and cyanosis can be found in childhood in severe cases.^{3,6} In less serious cases, symptoms may appear later, with dyspnea, cyanosis and digital clubbing.^{3,6} Interatrial septal defect and patent foramen ovale are described in some cases. The onset and severity of symptoms depend on the degree of hypoplasia.³

Given the combination of right atrial enlargement, axis deviation to the left and left ventricular hypertrophy, an

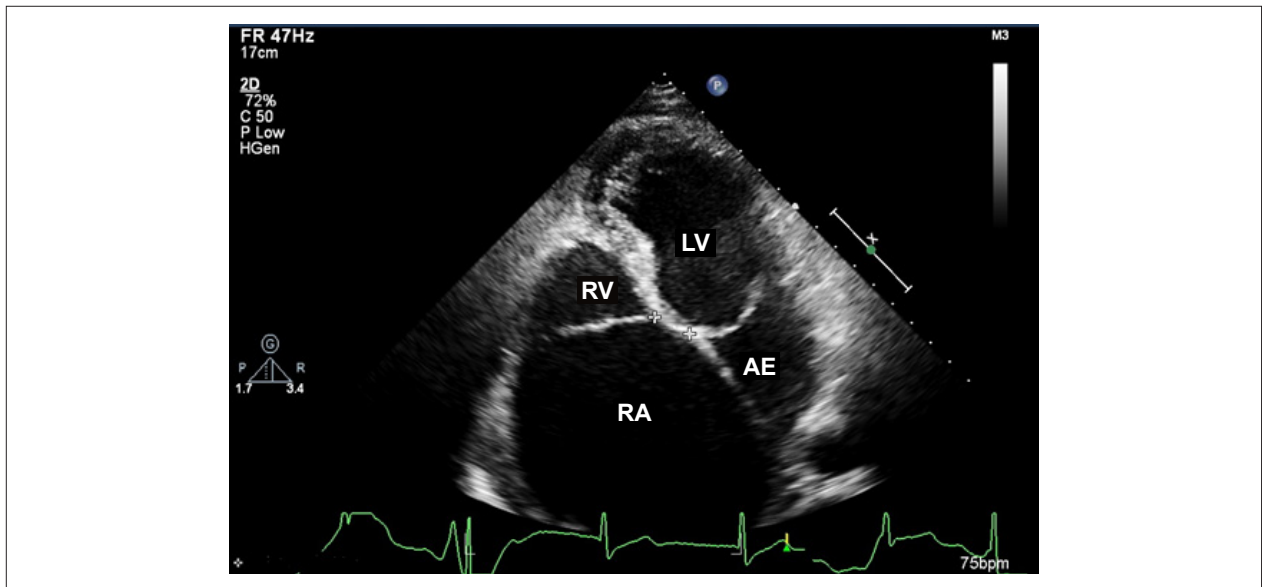


Figure 3 – Absence of tricuspid valve septal leaflet distal displacement. RV: right ventricle; LV: left ventricle; RA: right atrium; LA: left atrium.

important differential diagnosis is tricuspid stenosis, which was suspected in many of the cases described in the literature.^{2,3} Besides, other possible differential diagnoses are tricuspid atresia, pulmonary atresia and Ebstein's anomaly.³ It is extremely important to diagnose and, if necessary, treat these patients early. Neonatal follow-up potentially stands out in the suspicion and diagnostic confirmation of isolated hypoplasia of the apical portion of the right ventricle.

Complementary tests are extremely important. ECG usually shows signs of right atrial or biatrial hypertrophy, cardiac axis deviation to the left, and atrioventricular conduction disorders.²⁻⁴ Chest X-ray contributes little to the diagnosis, as it may show normal cardiac silhouette, cardiomegaly and/or normal or decreased pulmonary blood flow.²⁻⁵ Diagnosis is often determined by echocardiography, hemodynamic tests or magnetic resonance imaging.

Echocardiography demonstrates significant reduction in right ventricular size, especially the trabecular portion. The tricuspid and pulmonary valves are normal. The apical portion presents hypoplasia. Besides, the patent foramen ovale may be present and interatrial septal defect may appear as compensatory components.¹

Hemodynamic test by complete cardiac catheterization usually presents an increase in the right atrial pressure. An increase in initial and final diastolic pressures demonstrates reduced ventricular filling capacity. Moreover, right-left or bidirectional shunts can be found, as well as an oxygen saturation of 66% to 90%.³

On angiography, the right ventricle on anteroposterior view is small and located medially in a trabecular area with hypoplasia or even absent. The infundibulum is normal. Right ventricular contraction is ineffective, with reduced systolic and diastolic volumes.³

Magnetic resonance imaging is an excellent diagnostic method. It does not use ionizing radiation or potentially nephrotoxic means of contrast and provides more comprehensive cardiac evaluation.⁷ It provides an analysis of cardiac and vascular anatomy, of ventricular function, myocardial perfusion, as well as accurate tissue characterization.⁷ Therefore, due to diagnostic versatility and accuracy, magnetic resonance imaging has become a complementary imaging method widely used in the suspicion and evaluation of aortic diseases and other conditions affecting the vascular beds, in addition to acquired and congenital heart defects, such as isolated hypoplasia of the apical portion of the right ventricle.⁷

Pathological studies corroborate all the findings mentioned above, demonstrate histological examination of the normal right ventricular cavity and differentiate isolated right ventricular hypoplasia from the Uhl's anomaly, another rare disease characterized by the complete absence of the right ventricular parietal wall with the epicardial and endocardial layers without any fat interposition.^{3,10}

Major progress in pediatric cardiac surgery and improvement of techniques have recently contributed to increased patient survival.^{11,12} Treatment may be based on early intervention with systemic-pulmonary anastomosis for cases associated with cyanosis and decreased pulmonary blood flow, as well as definitive repair surgeries, which include interatrial septal defect closure, when the RV is of reasonable size, or univentricular repair by Glenn or Fontan surgery.⁶

Conclusion

This case demonstrates the importance of echocardiography combined with clinical history and physical examination, etiologic diagnosis of heart failure from the most common causes to the most rare ones and isolated apical right ventricular hypoplasia.

Case Report

Authors' contributions

Research creation and design: Lima CJM; Data acquisition: Alcantara ACB; Data analysis and interpretation: Freire ES; Manuscript drafting: Cavalcante FP; Critical revision of the manuscript as for important intellectual content: Martins CA, Evangelista NL.

Potential Conflicts of Interest

There are no relevant conflicts of interest.

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Academic Association

This study is not associated with any graduate program.

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