ISSN: 2634 - 6796

Journal of Cardiology Research Reviews & Reports

SCIENTIFIC Research and Community

Case Report

Open Access

Apical Hypoplasia of the Left Ventricle A Very Rare Congenital Heart Disease

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ABSTRACT

Background: Apical hypoplasia of the left ventricle it is characterized by a spherical truncated left ventricle, with some degree of systolic and/or diastolic dysfunction, an elongated right ventricle, which surrounds the distal left ventricle, adipose tissue can be found infiltrating the apex of the left ventricle (LV)

Case: A 6-year old male patient with a history of Zollinger-Ellison syndrome, righ renal agenesis and systolic murmur in tricuspid focus, the transthoracic echocardiogram showed situs solitus, hypoplasia of the apical portion of the LV, interventricular septum with basal and medial dyskinesia, with restrictive systolic and diastolic dysfunction, severe pulmonary hypertension, the right ventricle is dilated and elongated surrounding the LV forming the cardiac apex, already with systolic and diastolic dysfunction of the wall free of global deformation of 9.3%.

Conclusion: there is currently very little knowledge about what appears to be a rare congenital disease. Some cases reported in the literature have responded to pharmacotherapy, although many of the affected patients may require invasive procedures and even heart transplantation.

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Received: March 10, 2023; **Accepted:** March 14, 2023; **Published:** March 20, 2023

Introduction

Left ventricular apical hypoplasia (LVAH) is an extremely rare disease, considered as congenital heart disease as an unusual type of cardiomyopathy that was first described in 2004 [1]. It is characterized by a spherical, truncated left ventricle with some degree of systolic dysfunction, an elongated right ventricle, which normally envelops the distal left ventricle, with adipose tissue infiltrating the apex of the left ventricle (LV) and origin of the papillary muscle in the flattened anterior apex [2].

This rare phenomenon frequently presents with different clinical manifestations depending on the age of detection, from the absence of symptoms in children to congestive heart failure.

Clinical Case

A 6-year-old male school patient with a history of duodenal atresia who received postnatal surgical treatment Zollinger-Ellison syndrome and right renal agenesis. due to multiple structural alterations, he began multidisciplinary management, sending him for evaluation by pediatric cardiology, referring him to the pediatric echocardiography service due to presenting pectus carinatum and grade III/VI tricuspid systolic murmur. The transthoracic echocardiogram showed situs solitus, levocardia with levoapex, normal pulmonary and systemic venous returns, concordant atrioventricular (AV) and ventriculoarterial (VA) connection, hypoplasia of the apical portion of the left ventricle (see Figure 1) interventricular septum. with basal and medial dyskinesia, bulging in diastole towards the right ventricle, hypokinesia of the lateral wall and lateral apical portion, systolic dysfunction estimating ejection fraction of 45% by Simpson's method and an overall longitudinal strain: -14.5%. Restrictive diastolic dysfunction of the left ventricle, with a left atrial volume of 74.5ml/M2Sc, left atrial reservoir strain of 1%, stiffness index of 6.4. (see Figure 2).

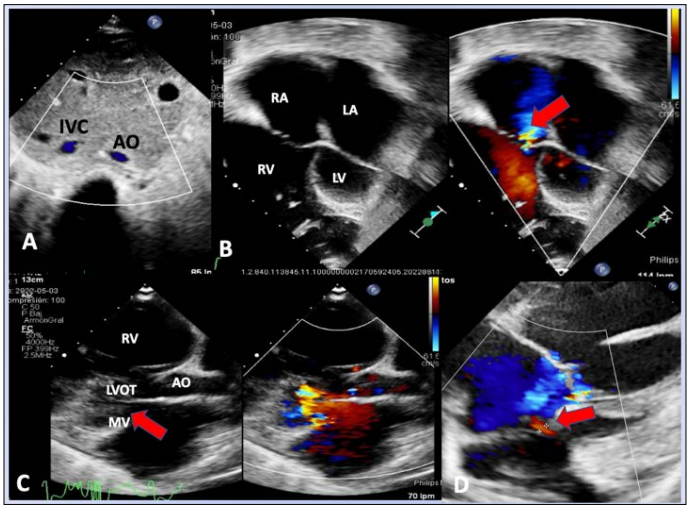

Figure 1: A) situs solitus. B) 4-chamber cut showing LV with absence of its apical portion, dilated RV, elongated apical portion surrounding the LV forming the apex, moderate tricuspid insufficiency. C) Long axis section showing mitral valve with moderate regurgitation. D) patent foramen ovale.

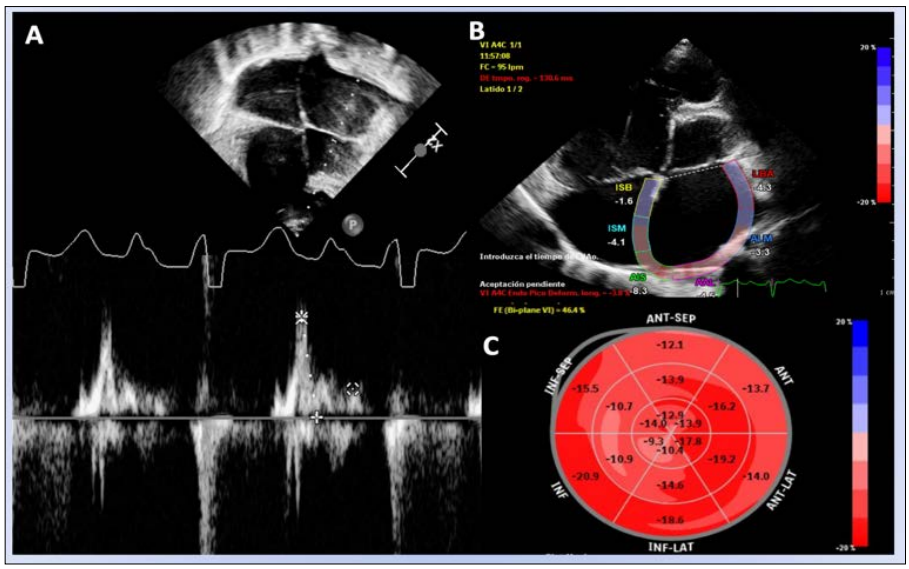

Figure 2: A) Diastolic dysfunction of the restrictive type deceleration time 50 m/s. B) left atrial strain. C) global left ventricular longitudinal strain of -14.5%.

The mitral valve with moderate insufficiency and severe pulmonary hypertension, systolic pressure of 92 mmHg and mean pressure of 63mmHg. The right ventricle is observed dilated and elongated surrounding the left ventricle forming the cardiac apex, already with systolic and diastolic dysfunction secondary to left pathology with a shortening fraction of 22%, TAPSE 8.9 mm, global strain of the free wall of -9.3% (See Figure 3) As an associated defect, a patent foramen ovale measuring 3.8 mm x 2.7 mm with a shunt left to right.

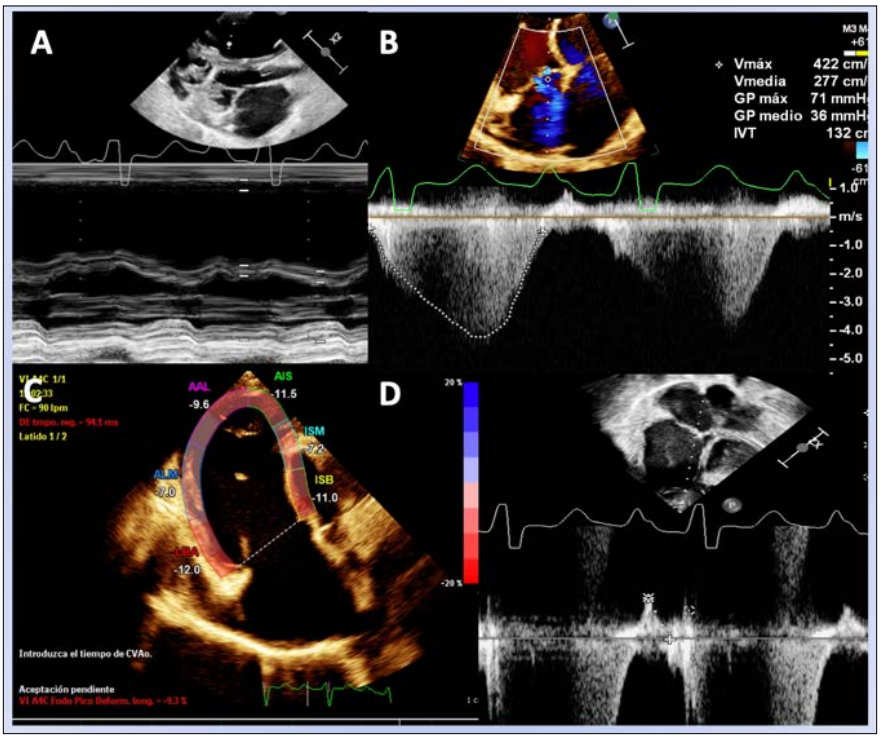

Figure 3: A) M-mode long axis showing septal dyskinesia. B) Tricuspid regurgitation gradient estimating PASP 91 mmHg and PMAP 56 mmHg. C) RV free strain with systolic dysfunction. D) Diastolic dysfunction with restrictive pattern.

Magnetic resonance imaging was performed as an extension study, reporting the left ventricle with the absence of its apical portion and dilation of its basal and middle third, without the presence of fibrosis and with normal systolic function (LVEF 55%), dilated right ventricle, elongated apical portion that surrounds the left ventricle forming the apex and severe systolic dysfunction (FEVD 13%), moderate mitral regurgitation, dilation of the left atrium.

He is currently being monitored under treatment with furosemide, spironolactone, digoxin, captopril, carvedilol, and acetylsalicylic acid, however, due to a poor prognosis and insidious evolution, he is accepted for heart transplantation pending treatment.

Discussion

There is currently very little knowledge about what appears to be a rare congenital disease. The cause is unknown, but it may be

related to inadequate LV-RV dilation, and identified a mutation of the lamin A/C gene, a gene known to be associated with other forms of cardiomyopathy, there is a clinical trial in progress to identify the genetic basis of this alteration since until now it is not associated with any race specifically [3].

Most of the reported cases of isolated LV apical hypoplasia have not been accompanied by other abnormalities. A case series of five young patients with the condition; describe cyanosis in two of the patients, although these patients had associated patent ductus arteriosus and severe pulmonary hypertension [4].

Various modalities have been introduced to diagnose LV apical hypoplasia with variable diagnostic accuracy. However, two-dimensional echocardiography and magnetic resonance imaging seem to be the most effective tools for the diagnosis of LV apical hypoplasia.

Definitive diagnostic criteria have not yet been established. The characteristic features on imaging studies are: truncated, spherical, and deteriorated LV with bulging of the interventricular septum toward the RV, apical LV fatty material, papillary/trabecular muscle abnormalities, and RV elongation involving the deficient LV apex [5].

A review of the literature published in the last ten years showed that most cases of LV apical hypoplasia have been reported among European and Caucasian populations, and more rarely in Spanish, Mexican, and Oriental populations. Clinical manifestations of LV apical hypoplasia were also found. disease was absent in young patients with adult-onset symptoms including fatigue, shortness of breath, sharp chest pain, and palpitations; and, in some cases with more progressive manifestations, abnormal electrocardiography, progressive dyspnea, pulmonary edema and even sudden death. One of the pathophysiological mechanisms proposed for the appearance of symptoms is the dilation of Vi, which results in a spherical shape [5,6].

Some cases reported in the literature have responded to the recommended first-line treatment for heart failure, although in patients like our case they can be controlled with a conservative approach, many affected patients may need invasive procedures and even heart transplantation.

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