JACC: CASE REPORTS VOL. 18, 2023

CASE REPORT

INTERMEDIATE

CLINICAL CASE

Right Ventricular Apical Hypertrophic Cardiomyopathy in Association With Infundibular, Valvular, and Supravalvular Pulmonary Stenosis

Aldo Cabello-Ganem, MD,[a] Mauricio Garcia-Cardenas, MD,[b] Gabriela Melendez-Ramirez, MD,[b] Aloha Meave-González, MD,[b] Santiago Saenz-Ancira, MD,[a] Sofia Molina-Castillo, MD,[a] Javier Serrano-Roman, MD,[c] Adrian Espejel-Guzman, MD,[d] Alexis D. Aparicio-Ortiz, MD,[a] Nilda Espinola-Zavaleta, MD, PhD[a,e]

ABSTRACT

A 31-year-old man was referred to our hospital because of exertional dyspnea and palpitations. Cardiac examination revealed a systolic murmur in the pulmonic area. Owing to the suspicion of pulmonary stenosis, cardiac magnetic resonance was notable for apical hypertrophy of the right ventricle and mixed pulmonary stenosis. (**Level of Difficulty: Intermediate.**) (J Am Coll Cardiol Case Rep 2023;18:101920) © 2023 The Authors. Published by Elsevier on behalf of the American College of Cardiology Foundation. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).

HISTORY OF PRESENTATION

A 31-year-old man with a 2-year history of exertional dyspnea (NYHA functional class II) and palpitations was referred to our hospital. Cardiac examination revealed a systolic murmur grade II/IV with fixed splitting of the second heart sound over the pulmonic area. Electrocardiogram showed sinus rhythm with signs of right ventricular hypertrophy (R in V_1 7 mm; R > S in DII and DIII; and T-wave inversion in V_1-V_3).

LEARNING OBJECTIVES

- To make a differential diagnosis of pulmonic valve disease using cardiac magnetic resonance.
- To discuss the management of unusual congenital heart defects in adult patients encountered by clinicians during everyday practice.

PAST MEDICAL HISTORY

The patient had a history of systemic hypertension, and the long-term therapy included irbesartan 150 mg/day and amlodipine 5 mg/day.

DIFFERENTIAL DIAGNOSIS

In patients with suspected pulmonic stenosis, conditions associated with increased pulmonary velocities, such as atrial septal defect and pulmonary regurgitation, must be investigated. In patients with signs

From the [a]Department of Nuclear Cardiology, National Institute of Cardiology Ignacio Chavez, Mexico City, Mexico; [b]Department of Cardiovascular Magnetic Resonance, National Institute of Cardiology Ignacio Chavez, Mexico City, Mexico; [c]Department of Internal Medicine, ABC Medical Center IAP, Mexico City, Mexico; [d]Department of Internal Medicine, Mexican Social Security Institute, Mexico City, Mexico; and the [e]Department of Echocardiography, ABC Medical Center IAP, Mexico City, Mexico.
The authors attest they are in compliance with human studies committees and animal welfare regulations of the authors' institutions and Food and Drug Administration guidelines, including patient consent where appropriate. For more information, visit the Author Center.

Manuscript received March 17, 2023; revised manuscript received April 21, 2023, accepted May 24, 2023.

ISSN 2666-0849 https://doi.org/10.1016/j.jaccas.2023.101920

ABBREVIATIONS AND ACRONYMS

CMR = cardiac magnetic resonance

EF = ejection fraction

LV = left ventricle

RV = right ventricle

and symptoms of right-sided heart failure, other causes should be considered, such as cor pulmonale or restrictive cardiomyopathy.

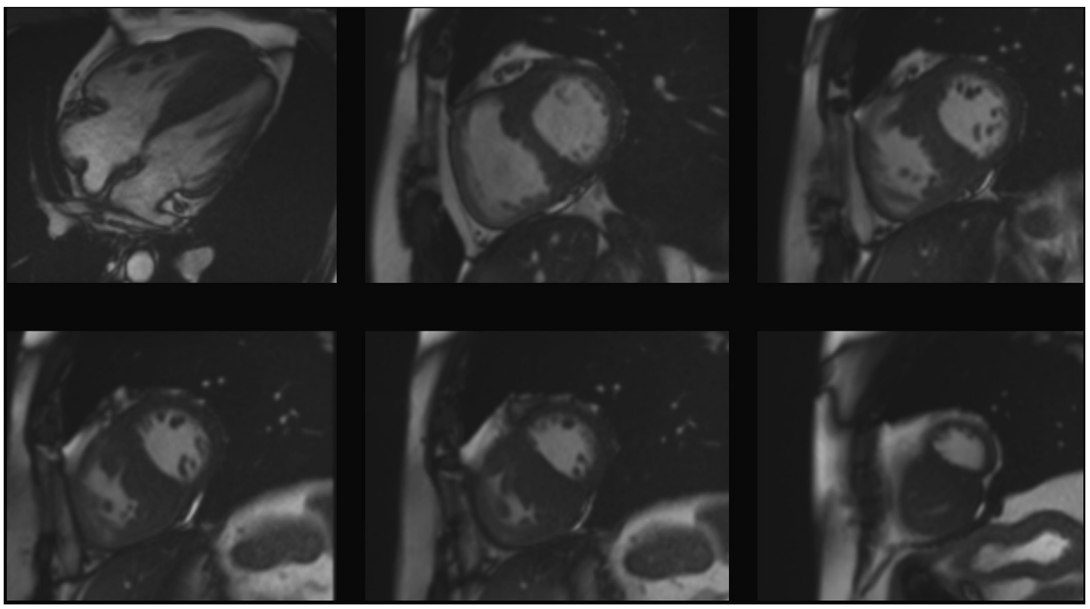
INVESTIGATIONS

Owing to the suspicion of pulmonary stenosis and a poor acoustic window during a transthoracic echocardiogram that was realized at another hospital that referred the patient to us. At our center, a transthoracic echocardiogram was performed, which revealed hypertrophic septal cardiomyopathy without obstruction of the left ventricle (LV) outflow tract; apical hypertrophy of the right ventricle (RV); conserved global systolic function of LV with a 2-dimensional ejection fraction (EF) of 75% (Simpson's method), and RV with an EF of 78% but compromised systolic circumferential function of the RV (2-dimensional fractional area change of 25%). Infundibular, valvular, and supravalvular stenoses of a tricuspid pulmonary valve with mild regurgitation, estimated pulmonic valve area of 0.95 cm^2 (continuity equation), thickened cusps, mean and peak Doppler gradient of 82 and 42 mm Hg, respectively, with a peak Doppler jet velocity of 4.52 m/s, classifying it as severe pulmonic stenosis.

Cardiac magnetic resonance (CMR) was performed with Siemens Magneton Avanto 1.5-T magnet to further evaluate intracardiac anatomy, biventricular function, and valvular function. CMR sequences 3-dimensional spoiled gradient echo and balanced steady-state free precession cine imaging were used for sequential segmental analysis. The balanced steady-state free precession cine imaging (**Figure 1**, Video 1) revealed atrial situs solitus, levocardia, and concordant atrioventricular and ventriculoarterial connections. The long-axis 4-chamber view demonstrated important hypertrophy of the septum and mid and apical portions of the RV free wall. The RV walls presented increased contractility with decreased end-systolic volume (29 mL) and an increased EF (79%). The right chambers were not dilated, with a right atrial indexed area of 14.2 cm^2/m^2. The LV also presented abnormalities, including a D-shaped configuration with hypertrophy in 5 of 17 segments involving the septum and anterior wall. It also presented with increased contractility, with an end-systolic volume of 27 mL and an EF of 75%.

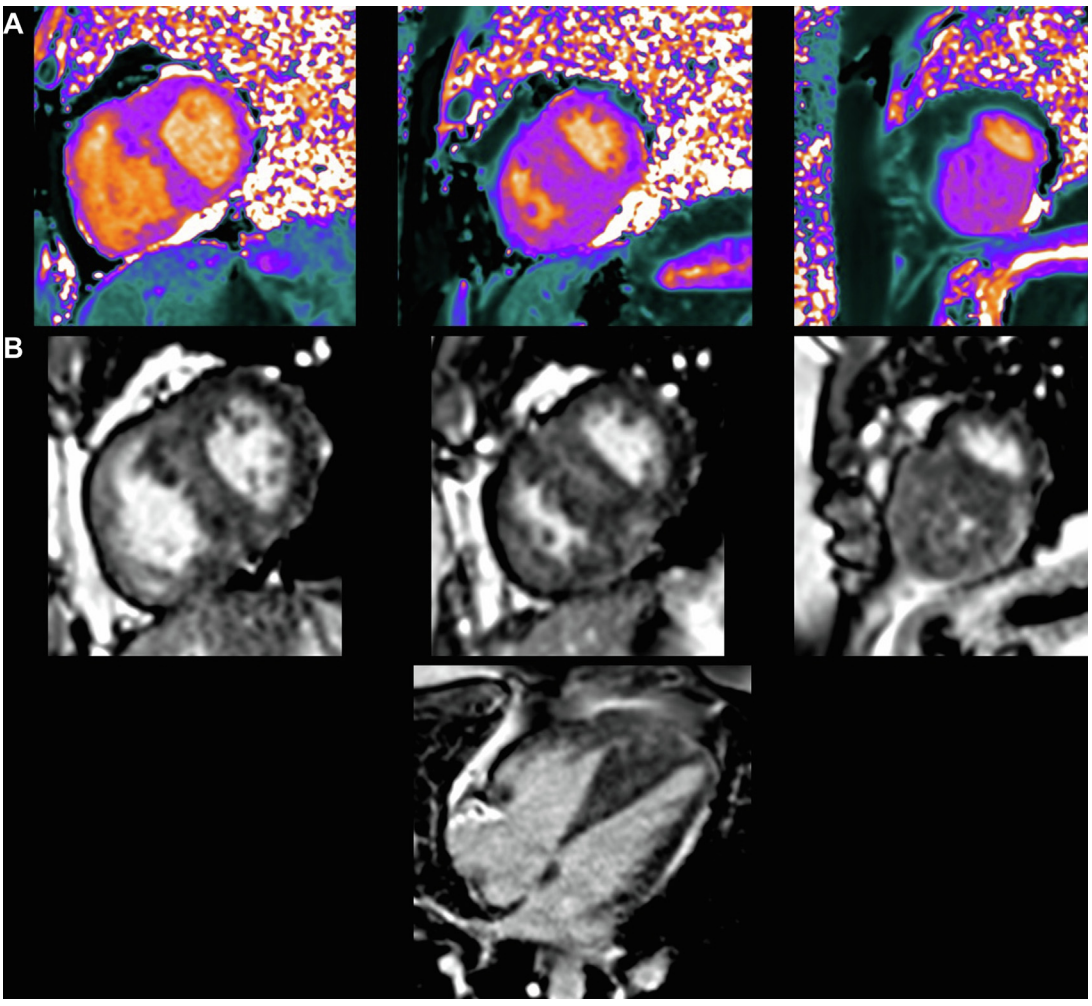
T1 and T2 parametric maps (**Figure 2**) obtained with modified Look-Locker inversion recovery were notable for elevation of T1 and T2 inversion times (1,072 ms and 62 ms, respectively) in the segments

FIGURE 1 Cardiac Magnetic Resonance



Balanced steady-state free precession cine imaging was implemented, revealing significant hypertrophy of the septum and mid and apical portions of right ventricular free wall.

FIGURE 2 **T1 Parametric Map Relaxation Time**

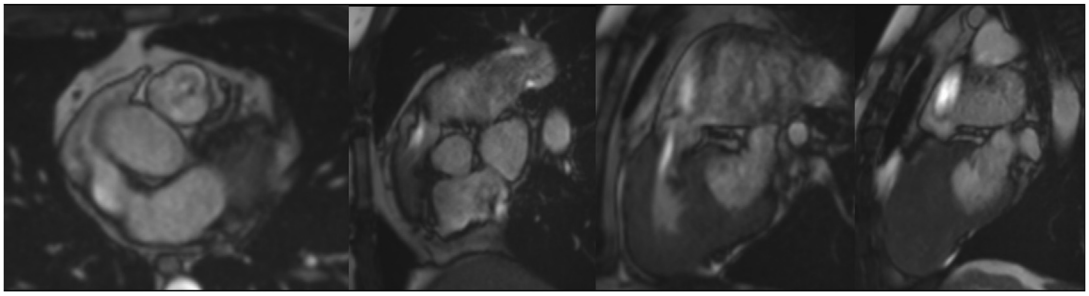


The first row **(A)** shows the T1 parametric map obtained via modified Look-Locker inversion recovery sequence demonstrating elevation of relaxation times in the segments with the most severe hypertrophy both in T1 (1,072 ms) and T2 (63 ms); the extracellular volume was increased (33%). Second row **(B)**, sequences after contrast demonstrate late gadolinium enhancement with a diffuse and patchy pattern in the septum and right ventricle.

with the most severe hypertrophy and increased extracellular volume (33%), a finding observed in both ischemic and nonischemic cardiomyopathies. Inversion recovery sequences after contrast administration demonstrated late gadolinium enhancement with a diffuse and patchy pattern in the septum and RV. A patchy pattern is seen in nonischemic cardiomyopathies such as hypertrophic cardiomyopathy, suggesting that RV hypertrophy is not secondary to pulmonary valve disease.

The RV outflow tract showed an increased jet acceleration in the infundibular, valvular, and supravalvular portions estimated by phase contrast imaging techniques (Figure 3). Infundibular acceleration was secondary to the infundibular muscular ring. The pulmonary valve was tricuspid and presented with stenosis with an area of 1.9 cm^2. Supravalvular stenosis was secondary to a muscular membrane 18 mm from the pulmonary annulus, which decreased the functional diameter of the

FIGURE 3 Mixed Pulmonary Stenosis



The axial plane of the pulmonary valve shows a tricuspid valve with stenosis. Sagittal views of the right ventricular outflow tract showed infundibular, valvular, and supravalvular jet acceleration. Infundibular acceleration was secondary to the infundibular muscular ring. The pulmonary valve was tricuspid and presented stenosis with an area of 1.9 cm^2. Supravalvular stenosis was secondary to a muscular membrane 18 mm from the pulmonary annulus, which decreased the functional diameter of the pulmonary trunk to 7 mm.

pulmonary trunk to 7 mm. Mild regurgitation was also observed. In addition, we observed post-stenotic dilation of the main pulmonary artery (39 mm), right pulmonary artery of 22 mm, and left pulmonary artery of 37 mm.

DISCUSSION

RV hypertrophy can be secondary to pulmonary disease or left heart disease.[1] Isolated RV hypertrophy is often secondary to pulmonary disease (cor pulmonale) and presents as global hypertrophy, with focal hypertrophy being uncommon.[2] It is important to note that the predominance of hypertrophy in the basal and apical segments suggests that the patient is on the spectrum of Yamaguchi syndrome, a diagnosis that is often missed owing to a characteristically poor acoustic window on echocardiography, highlighting the importance of CMR in this disease. Yamaguchi syndrome is a subtype of hypertrophic cardiomyopathy that presents with isolated apical hypertrophy; however, it usually involves the LV.[3]

For patients with symptoms (otherwise unexplained symptoms of heart failure, cyanosis of right-to-left communication between the arteries, and/or intolerance to exercise) with moderate or severe pulmonary stenosis and less than moderate pulmonary valve regurgitation, balloon valve valvotomy is recommended.[4] The surgical and medical management of the patient is still being evaluated owing to the complexity of the congenital heart defect the patient presented. Therefore, we mainly considered beta-blockers as part of medical management in place of the patient's current medical management (irbesartan and amlodipine), because these have been used in patients with tetralogy of Fallot as they cause relaxation of the RV outflow tract with improved pulmonary flow.[5,6]

Finally, it is notable that significant fibrosis measured in the patient may indicate a poor prognosis. Maron et al[7] reported that fibrosis of >20% of the LV mass increased the risk of sudden cardiac death, with current guidelines establishing 15% as intermediate risk. However, these cutoff points have yet to be extensively studied in subtypes, such as the present case, with predominantly RV apical hypertrophy.

CONCLUSIONS

It is essential to note the importance of CMR in diagnosing this patient; the apical subtype of hypertrophic cardiomyopathy usually has a characteristically poor acoustic window, and echocardiography cannot evaluate fibrosis as extensively as CMR. Finally, a complete evaluation of mixed pulmonary stenosis may only have been possible with the detailed intracardiac anatomy and function provided by CMR.

FUNDING SUPPORT AND AUTHOR DISCLOSURES

The authors have reported that there are no relationships relevant to the contents of this paper to disclose.

ADDRESS FOR CORRESPONDENCE: Dr Nilda Espinola-Zavaleta, Department of Nuclear Cardiology, National Institute of Cardiology Ignacio Chavez, Juan Badiano 1, 14080, Colonia Seccion XVI, Tlalpan, Mexico City, Mexico. E-mail: niesza2001@hotmail.com. Twitter: @nilda_zavaleta.

REFERENCES

1. Chatterjee S, Narayan P. Idiopathic isolated right ventricular apical hypertrophy. *Acta Cardiologica Sinica*. 2018;34:288-290.

2. Galderisi M, Severino S, Caso P, et al. Right ventricular myocardial diastolic dysfunction in different kinds of cardiac hypertrophy: analysis by pulsed Doppler tissue imaging. *Ital Heart J*. 2001;2:912-920.

3. Hughes RK, Knott KD, Malcolmson J, et al. Apical hypertrophic cardiomyopathy: the variant less known. *J Am Heart Assoc*. 2020;9:e015294.

4. Stout KK, Daniels CJ, Aboulhosn JA, et al. 2018 AHA/ACC guideline for the management of adults with congenital heart disease: A report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. *J Am Coll Cardiol*. 2019;73:e81-e192.

5. Fanous E, Mogyorósy G. Does the prophylactic and therapeutic use of beta-blockers in preoperative patients with tetralogy of Fallot significantly prevent and treat the occurrence of cyanotic spells? *Interact Cardiovasc Thorac Surg*. 2017;25:647-650.

6. Villarreal E, Farias J, Tweddell J, Loomba R, Flores S. Beta-blocker use after complete repair of tetralogy of Fallot: an analysis of a national database. *Cardiology Young*. 2021;32:584-588.

7. Maron BJ. Hypertrophic cardiomyopathy: a systematic review. *JAMA*. 2002;287, 1308-1020.

KEY WORDS **cardiac magnetic resonance, mixed pulmonary stenosis, right ventricular apical hypertrophic cardiomyopathy**

APPENDIX **For a supplemental video, please see the online version of this paper.**