

# Large atrial septal defect misdiagnosed as arrhythmogenic right ventricular cardiomyopathy

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## ABSTRACT

*We report a case of a large ostium secundum atrial septal defect that was initially diagnosed as an arrhythmogenic right ventricular cardiomyopathy. We discuss the initial workup, diagnosis, and treatment of this patient's atrial septal defect and explore how the pathophysiology of arrhythmogenic right ventricular cardiomyopathy may mimic congenital heart disease when there is a large left-to-right shunt.*

**Keywords:** atrial septal defect, arrhythmogenic right ventricular cardiomyopathy, arrhythmogenic right ventricular dysplasia, congenital heart disease, left to right shunt

## INTRODUCTION

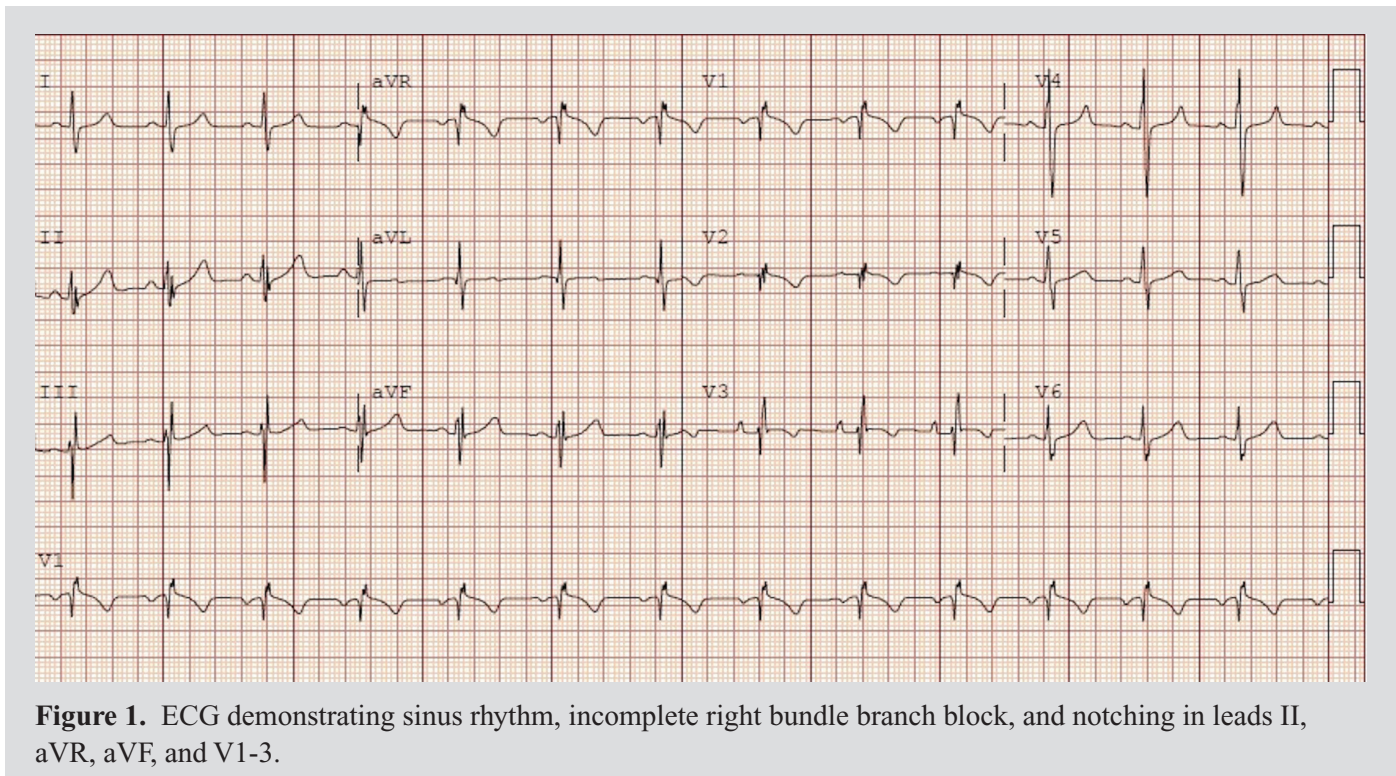
Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a rare form of inherited cardiomyopathy that typically presents in adulthood and is characterized by ventricular arrhythmias, right ventricular structural defects, and a high risk for sudden cardiac death.<sup>1</sup> It is estimated that ARVC affects approximately 1:2000-1:5000 people.<sup>2</sup> In contrast, atrial septal defects (ASDs) account for 25–30% of congenital heart defects diagnosed in adulthood.<sup>3</sup> Atrial septal defects often expand over time and may cause complications, like arrhythmias, paradoxical embolization, heart failure, pulmonary hypertension with Eisenmenger syndrome, and mortality. Large ASDs, measuring 25 mm or greater, may present a complex clinical picture. Early recognition and treatment of these conditions are necessary to prolong life and to avoid potential complications.

## CASE

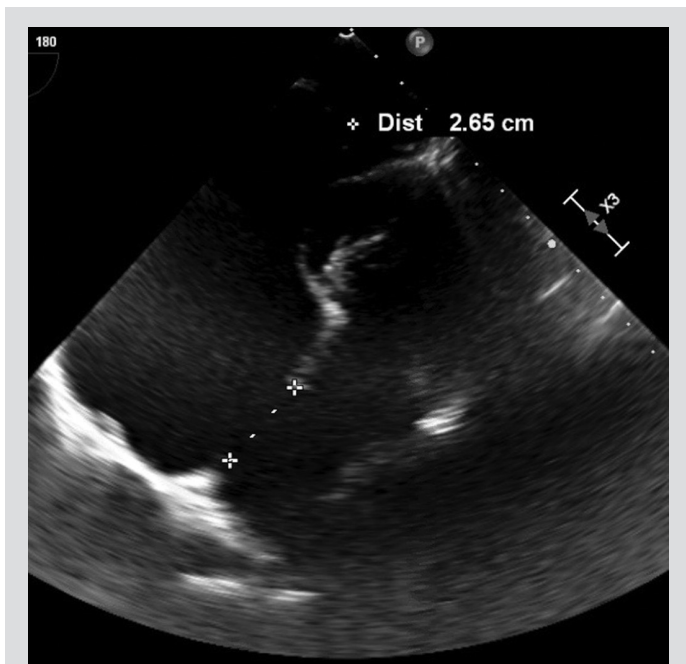
A healthy, athletic 28-year-old man presented to an outside facility with palpitations and exertional dyspnea. He had no known family history of cardiac disease. Workup from a referring provider included an abnormal EKG with possible epsilon waves, abnormal echocardiogram demonstrating severely dilated right atrium and right ventricle, and an abnormal Holter monitor demonstrating 7 beats of non-sustained ventricular tachycardia. He was initially diagnosed with ARVC.

The patient's workup in our facility included an ECG, transthoracic echocardiogram (TTE) followed by transesophageal echocardiogram (TEE). His ECG demonstrated sinus rhythm, incomplete right bundle branch block, and notching in leads II, aVR, aVF, and V1-3 (Figure 1). Transthoracic echocardiogram demonstrated a severely enlarged right atrium and right ventricle with moderately reduced systolic function, mildly elevated right ventricular systolic pressure, a severely enlarged pulmonary artery and branches. There was a dropout in the interatrial septum with Color Doppler suggestive of the large ostium secundum ASD. His TEE diagnosed the large ostium secundum ASD that measured approximately 26 mm in diameter (Figure 2).

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**DOI:** 10.12746/swrccc.v7i30.549



**Figure 1.** ECG demonstrating sinus rhythm, incomplete right bundle branch block, and notching in leads II, aVR, aVF, and V1-3.

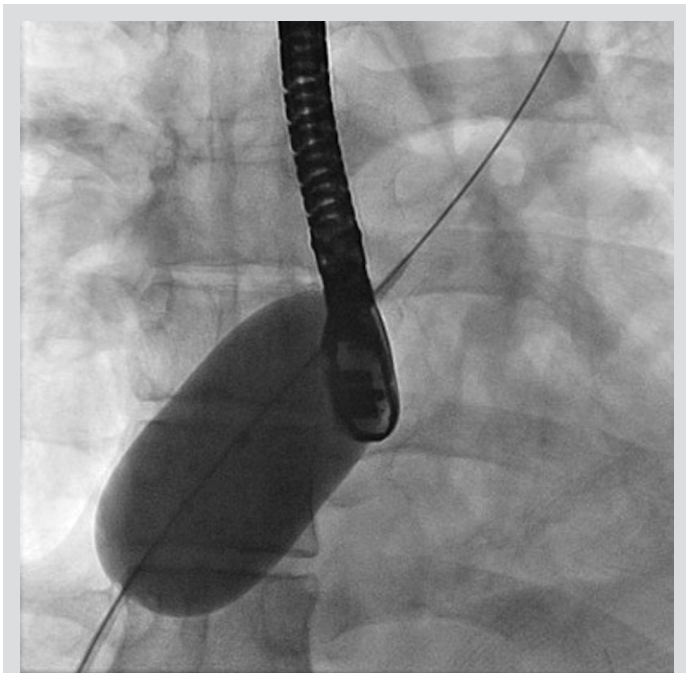


**Figure 2.** Transesophageal echo demonstrating large ostium secundum ASD measuring 26.5 mm.

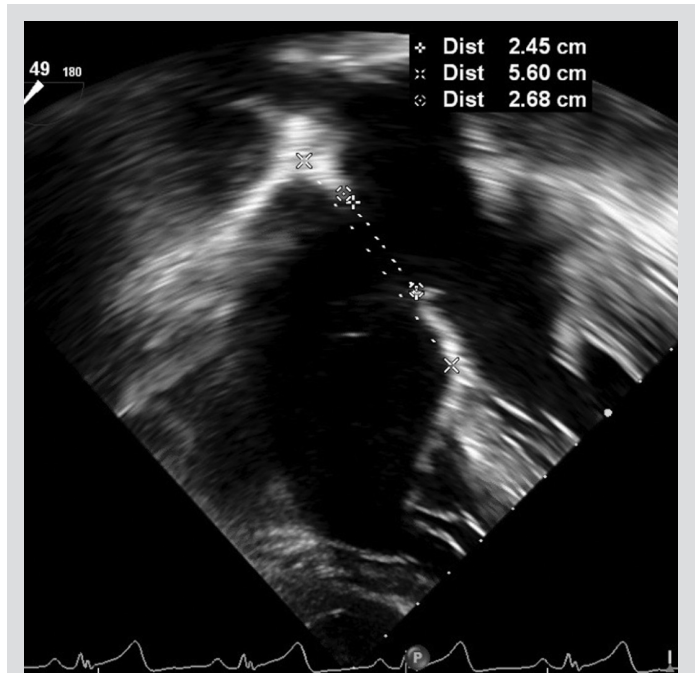
After discussion of management for this large ASD with the patient, percutaneous transcatheter closure was attempted with an Amplatzer 38 mm Atrial Septal Occluder (ASO) (Figures 3-7). This was unsuccessful due to the large size of the defect with deficient rims, and the procedure was aborted. The patient underwent elective surgical ASD closure (Figures 8-9).

## DISCUSSION

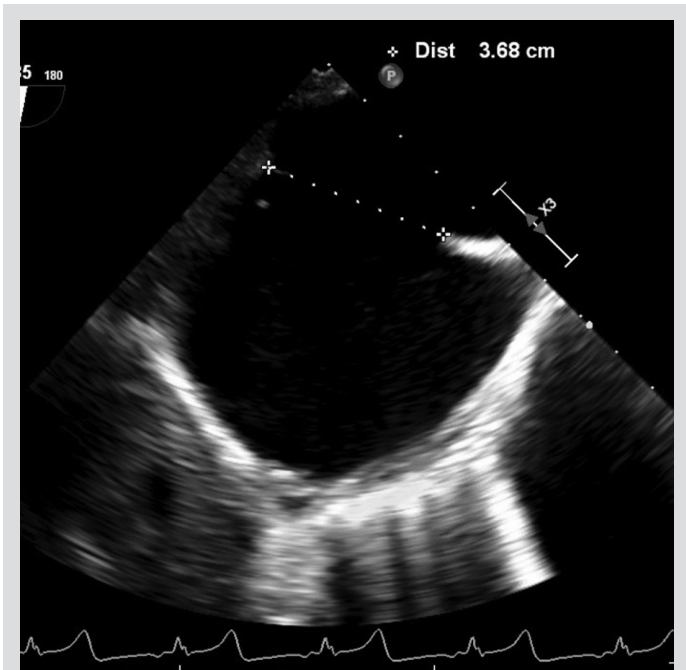
This case illustrates a large ASD that was initially misdiagnosed as ARVC. The clinical history of a young man with palpitations, an atypical ECG, abnormal Holter monitor study, and right ventricular structural changes on echocardiogram led to the initial diagnosis of ARVC. Through further workup, definitive diagnosis of ASD was made. The commonalities between ASD and ARVC stem from the differing pathophysiologies that lead to right ventricular dysfunction.<sup>4</sup> Case studies in pediatric patients have demonstrated that congenital heart disease can duplicate the structural and electrophysiologic



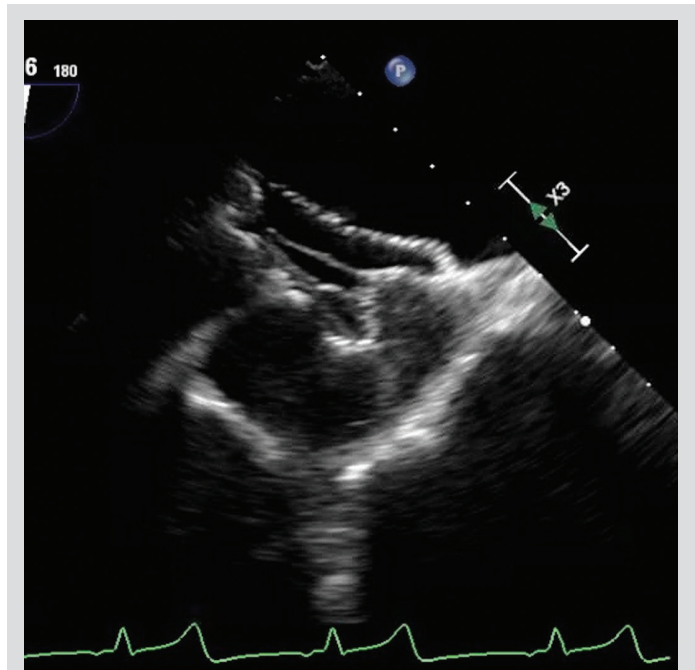
**Figure 3.** 34 mm Amplatzer Sizing Balloon, estimated size of deficit to be 37 mm.



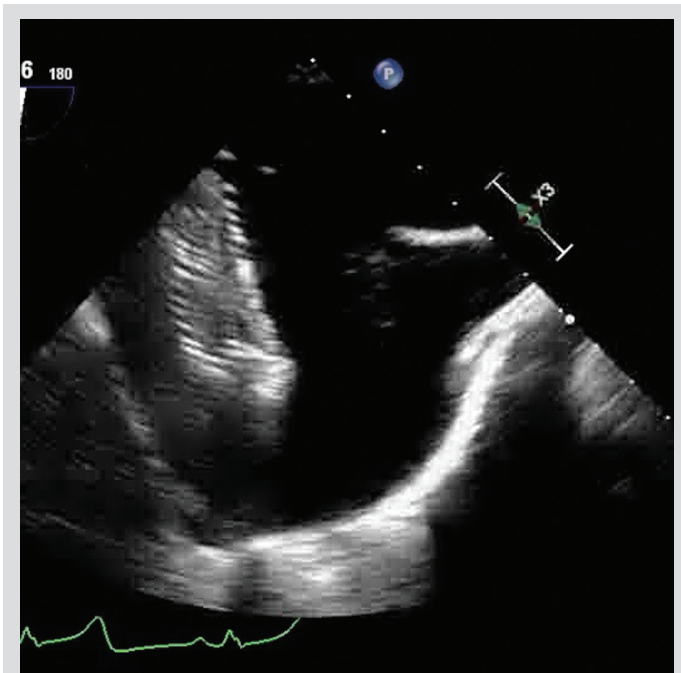
**Figure 5.** Various measurements of ASD.



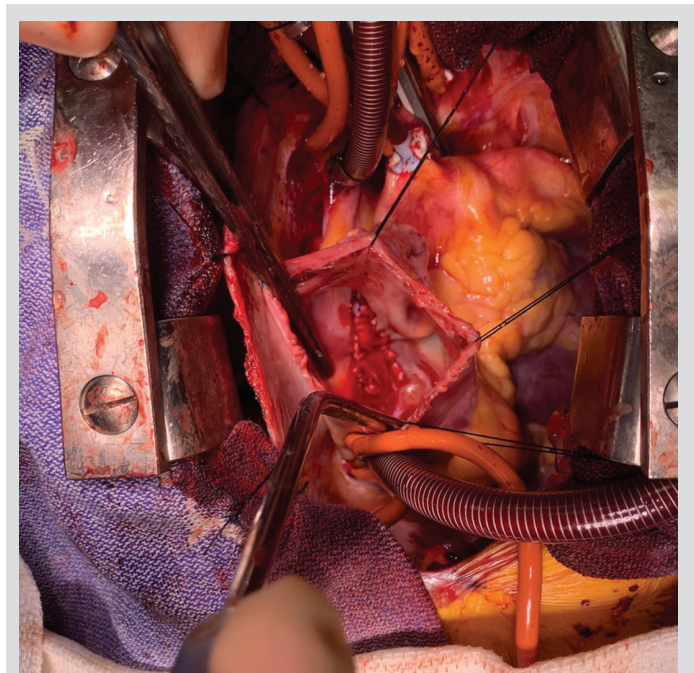
**Figure 4.** Diameter of defect at largest point across found to be 36.8 mm.



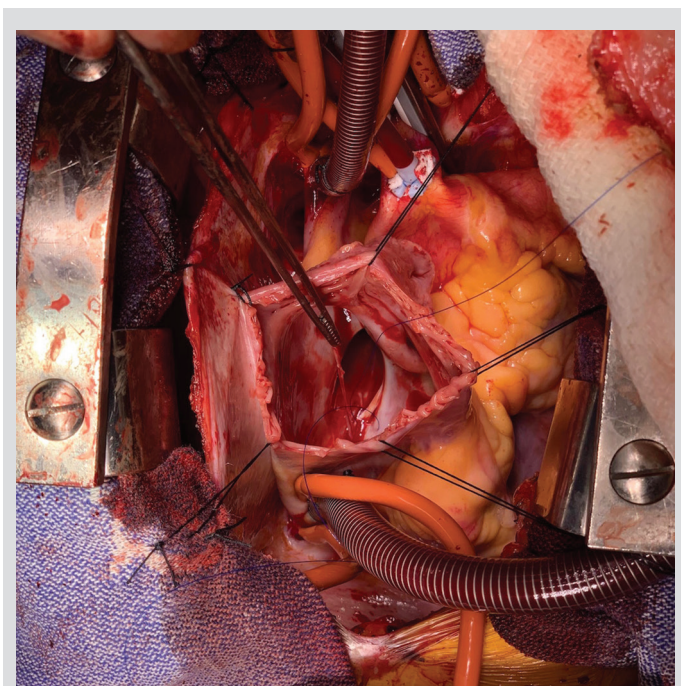
**Figure 6.** Amplatzer ASDO device in place with the delivery cable unreleased.



**Figure 7.** Prolapse of the ASD into the RA.



**Figure 9.** Final ASD closure.



**Figure 8.** Atriotomy revealing atrial septal wall defect.

findings of ARVC.<sup>4</sup> To make the diagnosis of ARVC, it is necessary to rule out right ventricular overload caused by left-to-right shunting.<sup>4</sup>

In patients with ARVC, the structural changes in the heart are caused by gene mutations in desmosome proteins, the intracellular adhesion complexes that serve as the mechanical connection between cardiac myocytes.<sup>1</sup> While these gene mutations are present at birth, the clinical symptoms of ARVC develop over time, and individuals are diagnosed in their adult years.<sup>1</sup> Impaired desmosomes lead to cell detachment and increased mechanical stress, which leads to fibro-fatty replacement of the myocardium and inflammation.<sup>1,5</sup> It is postulated that the scar tissue slows depolarization of the muscle and creates ECG changes, including symmetrically inverted T waves and epsilon waves [reproducible low-amplitude signals between the end of QRS complex to the onset of the T wave in the right precordial leads (V1-V3)] in approximately 30% of patients.<sup>2,6</sup> Arrhythmias are common and

vary; the most common is monomorphic ventricular tachycardia.<sup>2</sup>

Left-to-right shunts across an ASD create right ventricular volume overload and pulmonary over-circulation, which can cause fatigue and exercise intolerance.<sup>7,8</sup> Pulmonary artery and branches are typically dilated in patients with ASD, which is not present in patients with ARVC. Pulmonary hypertension commonly develops in patients with left-to-right shunts, usually later in life.<sup>8</sup> Longstanding right-sided heart volume and pressure overload create stretching of the atria and may cause atrial arrhythmias.<sup>8</sup> Additional problems that accompany left-to-right shunts include paradoxical embolisms and congestive right heart failure.<sup>8</sup> ECG findings characteristic of large secundum ASDs will show right-axis deviation, right atrial enlargement, and incomplete right bundle-branch block.<sup>8</sup> In this patient, TTE and TEE demonstrated that there was a large ASD, which explains the patient's clinical symptoms, findings on ECG, and severely enlarged right heart chambers and pulmonary artery.

Management differs greatly between the pathologies, and an ASD is a more favorable diagnosis than ARVC. Large ASDs may require percutaneous transcatheter closure or surgical repair.<sup>7,8</sup> Arrhythmogenic right ventricular cardiomyopathy will ultimately require cardiac transplantation.<sup>1,2</sup> Treatment of ARVC seeks to prevent sudden cardiac death through medical management of arrhythmias, ICD, and exercise restriction.<sup>1,2</sup> Heart disease presenting in early adulthood with severe symptoms requires a thorough workup and immediate treatment to reduce mortality. When creating a differential, it is important to consider how unique pathologies can create similar findings to ensure proper diagnosis and treatment.

## CONCLUSION

Large ostium secundum ASD can mimic ARVC. Cardiac imaging, including TTE, TEE, or cardiac MRI, can differentiate these diseases, which are treated differently.

**Article citation:** The Southwest Respiratory and Critical Care Chronicles 2019;7(30):58–62

**From:** Rocky Vista University College of Osteopathic Medicine, UC Health Heart Center Memorial Hospital Central, Colorado Springs, CO.

**Submitted:** 5/9/2019

**Accepted:** 7/1/2019

**Reviewers:** Scott Shurmur MD; Aliakbar Arvandi MD

**Conflicts of interest:** none

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