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CASE REPORT

INTERMEDIATE

CLINICAL CASE

Partial Atrioventricular Canal Defect With an Anomalous Left Circumflex Coronary Artery in an Elderly Veteran

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ABSTRACT

This study reports a case of partial atrioventricular canal defect with an anomalous left circumflex coronary artery in an elderly veteran presenting with unexplained dyspnea on exertion. This is a rare finding in this population and emphasizes the importance of a broad differential diagnosis and meticulous evaluation when more common conditions have been excluded. (**Level of Difficulty: Intermediate.**) (J Am Coll Cardiol Case Rep 2019;1:291–6) 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/).

A 71-year-old man was referred for dyspnea of unclear etiology. He was well until 2 years prior, when he noted exertional dyspnea and a decline in his functional status. This was attributed to paroxysmal atrial fibrillation, and he was started on sotalol with improvement in atrial fibrillation burden. However, over the past year, his functional status and dyspnea continued to worsen. His review of systems was otherwise unremarkable. After an unrevealing initial work-up, including transthoracic echocardiogram, regadenoson ^{99m}Tc sestamibi stress test, and pulmonary function testing, he was referred to our institution for right heart catheterization.

LEARNING OBJECTIVES

- To be able to formulate a differential and diagnose the etiology of dyspnea, including less common causes such as undiagnosed ASD in the elderly.
- To understand some of the various forms of endocardial cushion defects and associated anomalies.
- In adults with unexplained dyspnea or atrial arrhythmias with right heart dilatation, an evaluation for ASDs must be considered, as these are commonly not diagnosed until adulthood, when such clinical manifestations occur.

On presentation, vitals included a heart rate of 48 beats/min, blood pressure 132/76 mm Hg, and an oxygen saturation of 97% at rest and on ambulation. Cardiac auscultation demonstrated a soft S1, split S2, and a 3/6 holosystolic murmur loudest at the apex, radiating to the left lower sternal border. He had elevated jugular venous pressures with prominent V waves. Lungs were clear on auscultation, and there was no lower extremity edema.

PAST MEDICAL HISTORY

He had a history of paroxysmal atrial fibrillation on sotalol, hypertension, hyperlipidemia, and obstructive sleep apnea.

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ABBREVIATIONS AND ACRONYMS

ASD = atrial septal defect

DIFFERENTIAL DIAGNOSIS

Initial differential diagnoses of ischemia, atrial arrhythmia, aortic valve disease, mitral valve disease, cardiomyopathy, or congenital heart disease such as an atrial septal defect (ASD).

INVESTIGATIONS

Initial electrocardiography demonstrated sinus bradycardia with first-degree atrioventricular block and normal QRS axis (**Figure 1**). Complete blood count, metabolic panel, and liver function were normal. Right heart catheterization showed normal filling pressures, cardiac output, and pulmonary vascular resistance. The superior vena cava oxygen saturation was 66%, and the pulmonary artery oxygen saturation was 82%, consistent with a step up in oxygen saturation and concerning for a predominantly left to right intracardiac shunt. The ratio of pulmonic to systemic blood flow (Qp:Qs) was calculated to be 1.94, consistent with a significant left-to-right shunt. Coronary angiogram revealed no obstructive disease but did reveal a long left main giving off only a left anterior descending artery (**Figure 2A**). On careful review of the angiogram images, we noted an anomalous left circumflex artery arising from the right coronary cusp (**Figure 2B**). Repeat transthoracic echocardiogram was performed and revealed normal left ventricular systolic function but mild-to-moderate right ventricular systolic dysfunction with severe dilatation, moderate mitral regurgitation, and both primum and secundum ASDs (**Figures 3A to 3C**). Bidirectional flow was noted on agitated saline contrast study (**Figures 3B and 3C**). A transesophageal echocardiogram confirmed a large ostium primum ASD (maximum diameter 1.8 cm) as well as a secundum ASD with a maximum diameter of 1 cm with bidirectional but predominantly left-to-right flow (**Figure 4**, Video 1). There was moderate tricuspid and mitral regurgitation with evidence of a cleft anterior mitral valve leaflet (Videos 2A, 2B, and 2C). The atrioventricular valves were equiplanar, consistent with a partial atrioventricular canal defect (**Figure 5**). He was discharged with an outpatient plan to continue assessment.

MANAGEMENT

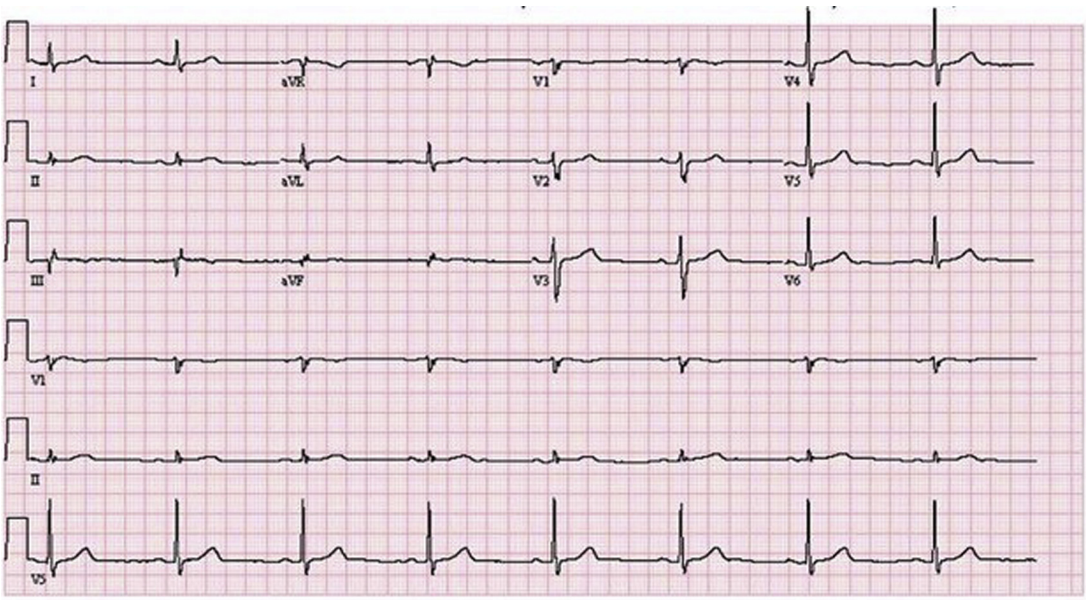
One month after presentation, he underwent surgical repair of his primum ASD, repair of his secundum ASD with a bovine pericardial patch, and tricuspid annuloplasty. A mitral valve replacement with a bioprosthesis (Carpentier-Edwards Magna 27 mm, Edwards Lifesciences, Irvine, California) was performed, as the mitral valve was not amenable to repair.

DISCUSSION

ASDs are one of the most common congenital cardiac lesions and are often not diagnosed until adulthood (1,2). The primitive atria are divided into the left and right atria by the septum primum during development, which grows from the roof to the atrioventricular canal septum (also known as the endocardial cushion). Ultimately, the endocardial cushion forms the inferior portion of the atrial septum, the superior portion of the ventricular septum, and portions of the anterior mitral leaflet, as well as the septal leaflet of the tricuspid valve. Developmental deficiencies in this process result in the formation of a primum ASD (3). A primum ASD is a variant of a common atrioventricular canal defect, leading to an interatrial communication located between the anterior-inferior margin of the fossa ovalis and the atrioventricular valves (4). In addition to the septal defect, the atrioventricular valves are almost always abnormal, including a cleft in the anterior mitral leaflet. The direction and amount of flow through an ASD is determined by its size and by the relative atrial pressures; this relates to the compliances of the left and right ventricles (5). In 2018, the American Heart Association and the American College of Cardiology published new guidelines for the management of adults with congenital heart disease (6). The guidelines recommend surgical repair in adults with primum ASDs causing impaired functional capacity, right atrial or right ventricular enlargement, and net left-to-right shunt sufficiently large to cause physiological sequelae (i.e., ratio of pulmonic to systemic blood flow [Qp:Qs] $\geq$1.5:1) without cyanosis at rest or during exercise, as long as systolic pulmonary artery pressures are lower than 50% of systemic pressures and pulmonary vascular resistance is less than one-third of systemic vascular resistance (6).

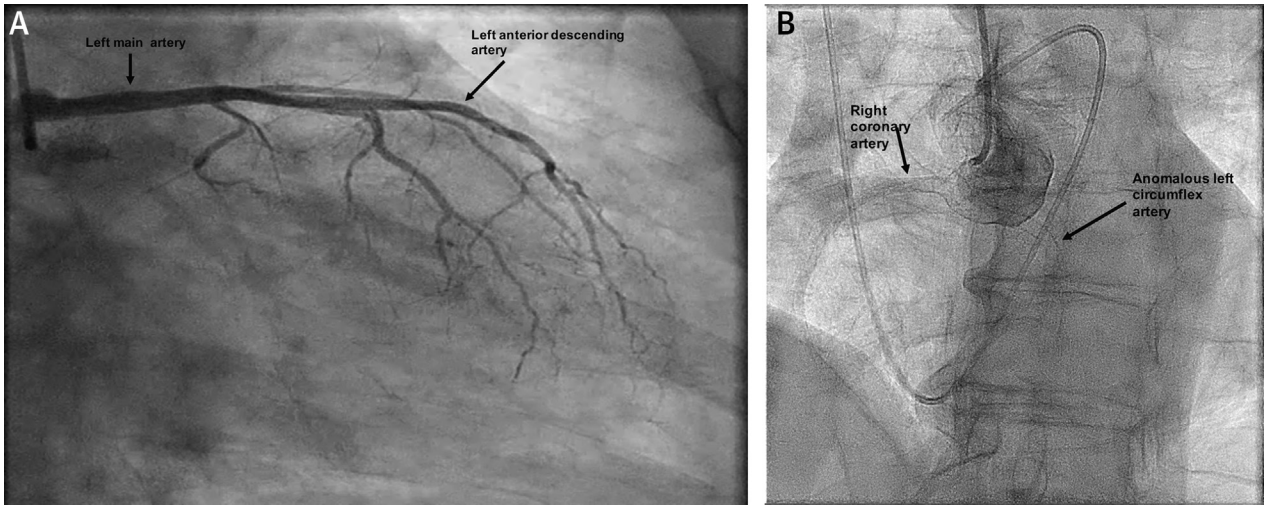
This case is unusual in many aspects. First, despite having a partial atrioventricular canal defect, the patient had a very subtle and late presentation. This likely can be explained by the age-related decrease in left ventricular compliance, progressive atrial remodeling, associated atrial arrhythmia, consequent increase in left-to-right shunting, and the absence of

FIGURE 1 Initial Electrocardiography



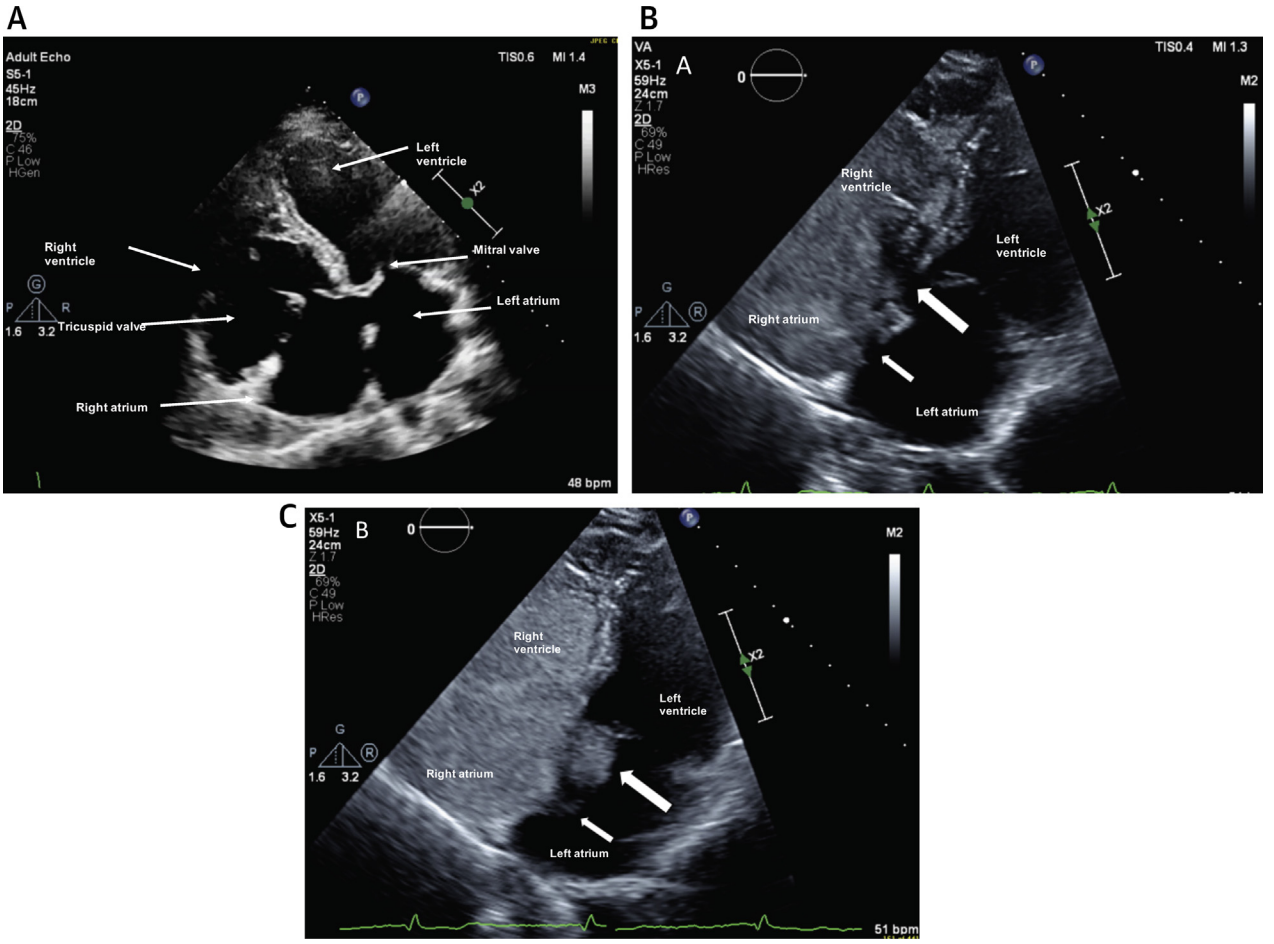
Normal sinus rhythm with first-degree atrioventricular block.

FIGURE 2 Anomalous Left Circumflex Coronary Arising From Right Coronary Cusp



(A) Coronary angiogram in right anterior oblique caudal view showing a long left main artery that only gives off a left anterior descending artery. **(B)** Coronary angiogram in left anterior oblique cranial view with an anomalous left circumflex artery.

FIGURE 3 Ostium Primum on Transthoracic Echocardiogram

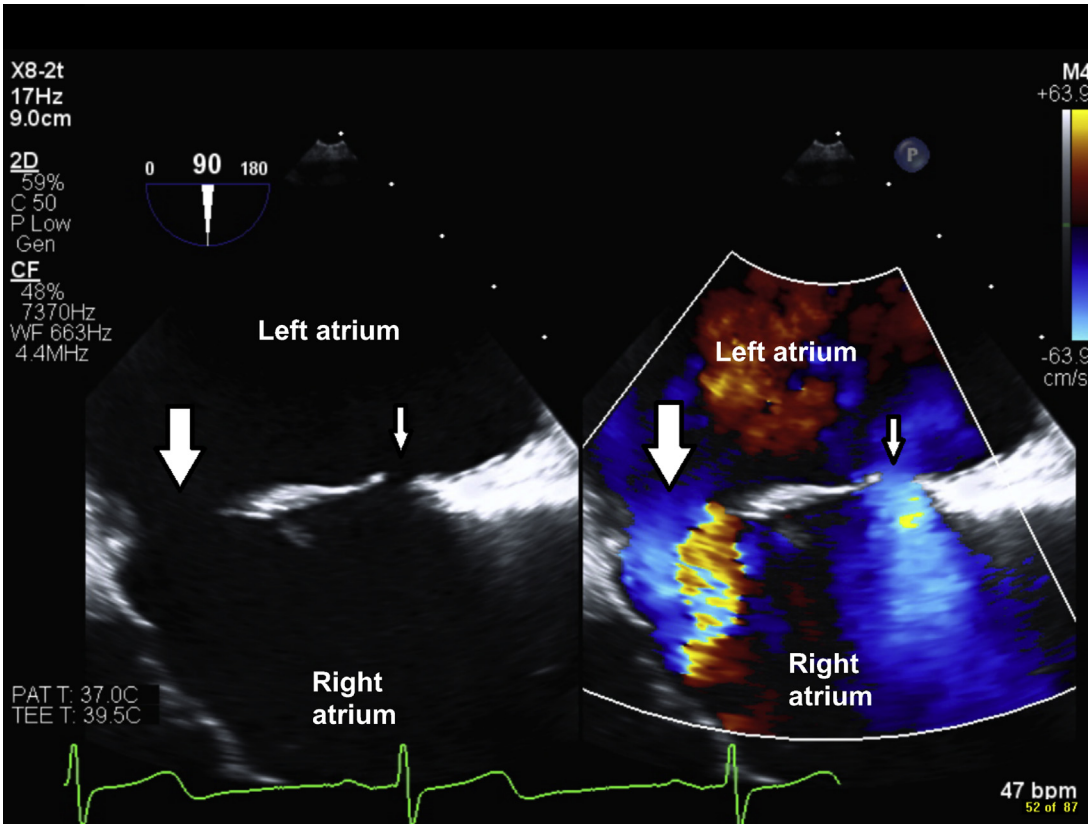


Transthoracic echocardiogram showing **(A)** severe right ventricular dilatation and equiplanar atrioventricular valves and **(B)** large ostium primum **(large arrow)** and smaller ostium secundum **(small arrow)** atrial septal defects, as evidenced by negative contrast on bubble study consistent with predominantly left-to-right shunting on apical 4-chamber view. **(C)** Agitated saline extravasation through ostium primum **(large arrow)** and ostium secundum **(small arrow)** atrial septal defects suggestive of bidirectional shunting.

severe mitral regurgitation. Second, unlike most primum ASDs, he had no left axis deviation or an incomplete right bundle branch block on electrocardiography, which is normally found in primum ASD due to the anomalous development of the Purkinje conduction system (specifically the left bundle branch fascicles) (7). This may be explained by the patient's unique phenotype and the degree of abnormal development of the endocardial cushions. Third, the anomalous left circumflex coronary artery arising from the right coronary cups is a rare finding as well. It is now accepted that the coronary arteries develop within the epicardial atrioventricular groove close to the endocardial cushion, with their proximal segments growing into the aortic sinuses. This may explain how the congenital anomalies in the patient occurred together (8). The role of this anomaly in his presentation remains unclear, but given his clinical improvement, its contribution to his initial presentation is likely trivial.

Although the combination of primum and secundum ASD is not novel, to the authors' knowledge, this

FIGURE 4 Ostium Primum and Ostium Secundum on Transesophageal Echocardiogram



Transesophageal echocardiogram showing ostium primum **(large arrow)** and ostium secundum **(small arrow)** atrial septal defects with and without color flow Doppler (Video 1).

is the first case report that also included an associated anomalous left circumflex coronary artery. In addition, the advanced age of the patient at time of diagnosis highlights that congenital heart disease is not a diagnosis reserved for just the young. This case raises many questions about the approach to patients with unexplained dyspnea and the importance of considering a broad differential diagnosis, including underlying congenital heart disease.

FOLLOW-UP

His post-operative course was uncomplicated and his symptoms have improved dramatically.

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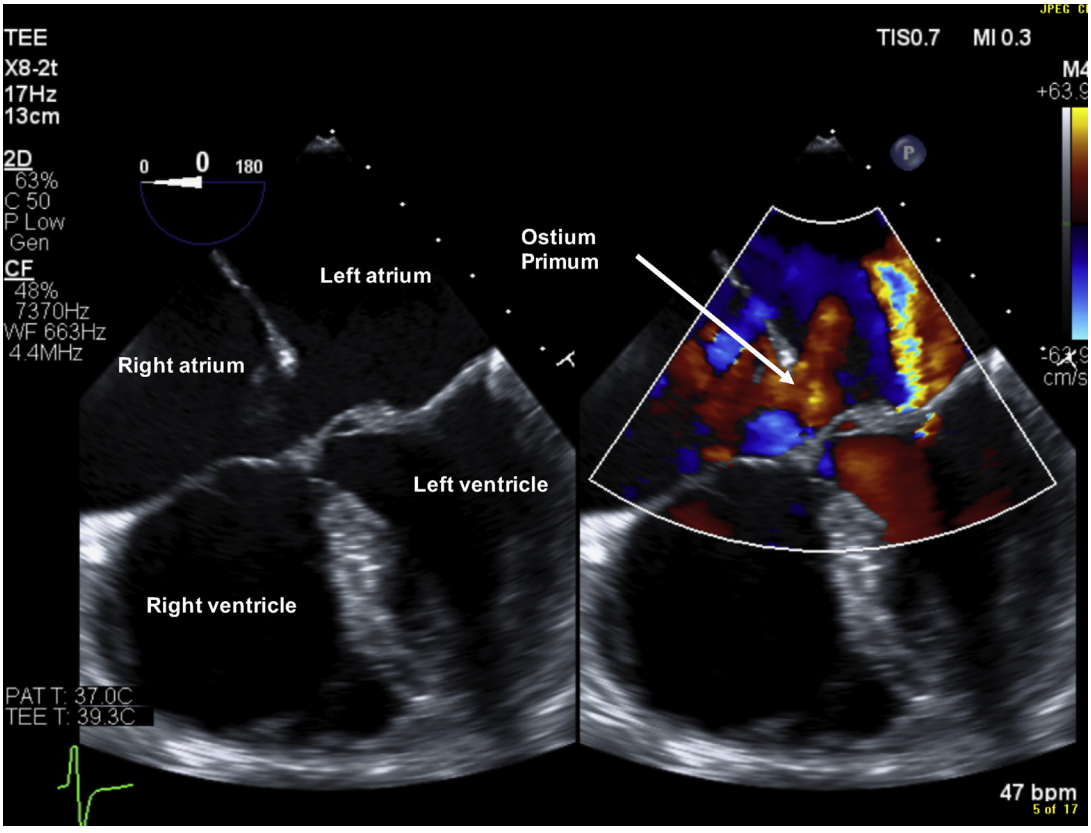
FIGURE 5 Ostium Primum and Equiplanar Atrioventricular Valves on Transesophageal Echocardiogram



Transesophageal echocardiogram showing equiplanar atrioventricular valves with an ostium primum on apical 4-chamber window **(left)** and left-to-right flow across ostium primum on color Doppler **(right)**. See Videos 2A, 2B, and 2C.

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KEY WORDS adults with congenital heart disease, anomalous coronary artery, atrial septal defect, echocardiography, Veteran

APPENDIX For supplemental videos, please see the online version of this paper.