Challenges and Implications of Uncorrected Atrioventricular Septal Defect in Adults: A Complex Case Report

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The prevalence of congenital heart disease in the adult population has shown continuous growth over recent decades. This is primarily due to significant advances in the diagnosis and treatment of congenital heart defects during childhood, which have been promoted by pediatric cardiology and congenital cardiac surgery. As a result, most children with congenital heart disease, including the most complex forms, now survive into adulthood. Currently, it is estimated that approximately 90% of children diagnosed with severe congenital heart disease reach 18 years of age. The growing clinical relevance of the adult congenital heart disease (ACHD) population stands out in this context. This is a heterogeneous group, both anatomically and pathophysiologically, as well as in terms of the different types of corrective or palliative surgical interventions to which they have been subjected. Although the overall number of individuals with ACHD continues to rise, the number of patients with certain specific anomalies or who have undergone specific types of repairs may still be relatively small.[1]

Atrioventricular (AV) septal defect is morphologically characterized by a common AV junction, with absence of the membranous and muscular AV septa. As a result, on echocardiographic examination, the AV valves are seen situated within the defect.[2] Atrioventricular septal defects (AVSDs) can be classified as partial or complete. Partial AVSD occurs when there is an opening divided by the superior and inferior bridging leaflets, which are connected by a tissue structure known as the "connecting tongue," separating the valve into two distinct orifices — one on the left and one on the right. On the other hand, complete AVSD is characterized by a single, undivided orifice encompassing the valvular structures on both the left and right sides.[2]

Case report

Male patient, 18 years old, admitted to a tertiary care hospital presenting with fever, cough, and dyspnea for the past seven days. According to the patient's mother, he sought medical attention on the second day of symptoms at an urgent care clinic, where he was prescribed ibuprofen and azithromycin to take at home. Due to lack of improvement, he sought hospital care.

Medical history includes congenital heart disease, specifically a surgically uncorrected AVSD, as he had lost follow-up at the reference hospital since childhood. On physical exam, he appeared in fair condition, with a productive cough, rapid breathing, and cyanosis around the lips and extremities. Hemodynamically stable, with blood pressure of 125 × 86 mmHg, heart rate of 105 bpm in atrial fibrillation rhythm, and oxygen saturation of 69% on room air.

Laboratory tests revealed: hemoglobin 16.2 g/dL, leukocytes 8,960/μL, CRP values over different days: 69.82 – 65.88 – 152.55 mg/dL (reference < 5 mg/dL), urea 20 mg/dL, creatinine 0.62 mg/dL, sodium 137 mEq/L, troponins 12 and 11.9 ng/L (reference < 14), NT-proBNP 4,377 pg/mL, and D-dimer 11,656 ng/mL. Blood cultures, urine culture, and rapid COVID-19 test were negative. The sputum smear showed some gram-negative diplococci, but the sputum culture was negative. The Mycobacterium tuberculosis DNA test also came back negative. Electrocardiogram showed atrial fibrillation, and a chest X-ray was ordered for further evaluation (Figure 1 and 2).

The patient was admitted to the Intensive Care Unit (ICU), where antibiotic therapy was broadened with the addition of amoxicillin/clavulanate, in combination with azithromycin. Blood and sputum cultures were collected, and an arterial blood gas test was performed. A chest CT angiography was also requested, which ruled out pulmonary embolism but contributed to the elucidation of congenital heart disease (Figure 3).

After 48 hours in the ICU, the patient was transferred to the general ward and remained under the care of the cardiology team. A transthoracic echocardiogram was performed, confirming the diagnosis of partial AVSD, as described below: Situs: Left isomerism. Cardiac position: Levocardia. AV connection type: Via two valves located in the same plane. Ventriculo-arterial connection type: Concordant. Mode of ventriculo-arterial connection: With normally related great arteries. It showed Large single atrium due to absence of the interatrial septum. Moderate-to-severe left AV valve regurgitation (EROA: 0.22 cm^2, regurgitant volume 16 mL, vena contracta 0.45 cm). Moderate right AV valve regurgitation (EROA: 0.23 cm^2, regurgitant volume 21 mL, estimated peak jet velocity of 3.95 m/s, and pulmonary artery systolic pressure of 72 mmHg). Pulmonary hypertension (PH). Pulmonary artery significantly dilated, measuring approximately 5.0 cm. Left ventricle with concentric remodeling and preserved systolic function (61%). Right ventricle enlarged with reduced systolic function (FAC: 27.5%, S' wave 9.3 cm/s) (Figures 4 e 5, Video 1).

Keywords

Atrial Heart Septal Defects; Congenital Heart Defects; Adult.

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Manuscript received February 16, 2025, revised manuscript April 30, 2025, accepted June 25, 2025
Editor responsible for the review: Andrea Vilela

DOI: https://doi.org/10.36660/abcimg.20250014i

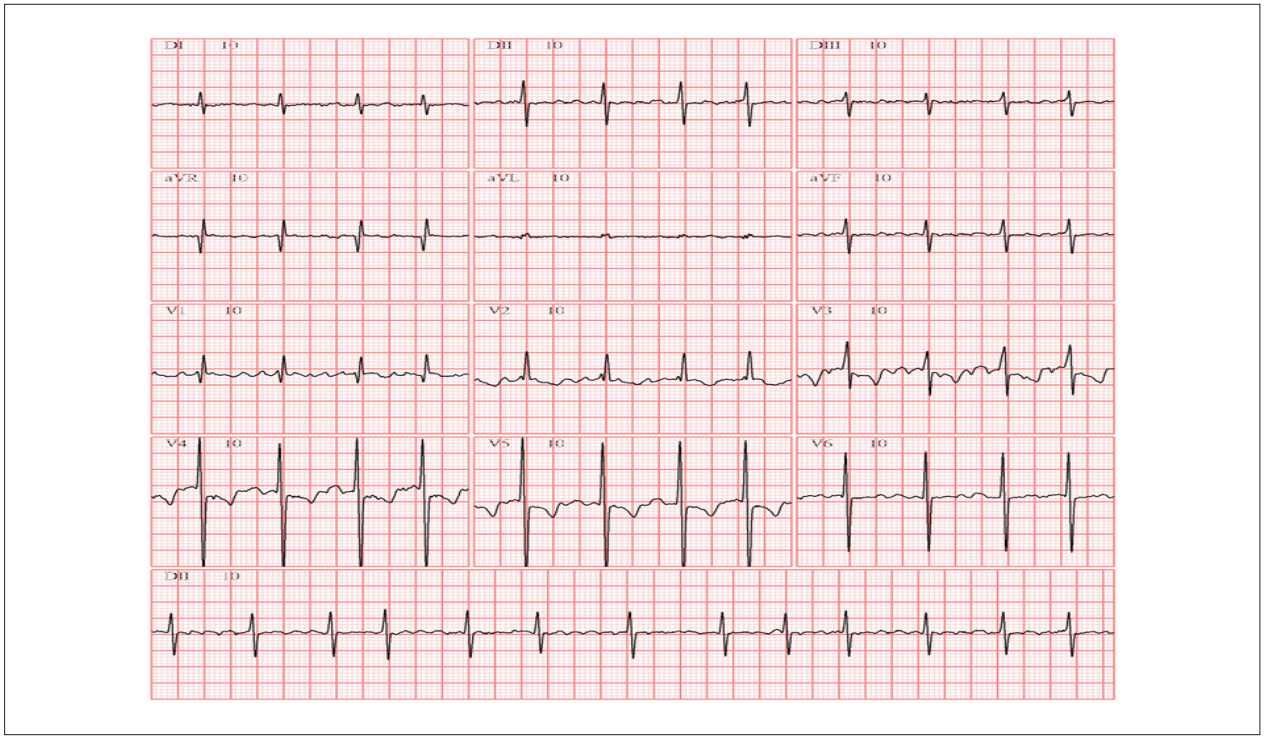


Figure 1 – *Resting Electrocardiogram: Atrial fibrillation rhythm, QRS axis +30°, secondary ventricular repolarization abnormalities.*

In this case report, a tissue tongue is observed connecting the two bridging leaflets, resulting in the separation of the AV valve into two distinct orifices — one right and one left. Additionally, the bridging leaflets are fused to the crest of the interventricular septum (Video 1).

Due to the clinical presentation, the working diagnosis upon admission was community-acquired pneumonia. Following antibiotic therapy, his fever and general condition improved, though he continued to experience exertional shortness of breath and maintained a resting oxygen saturation of 87%. He was discharged on rivaroxaban, sildenafil, carvedilol, enalapril, spironolactone, digoxin, and furosemide, and scheduled to start bosentan to improve functional class and quality of life.

While a recent right heart catheterization was not performed, echocardiographic findings strongly suggest Eisenmenger syndrome, indicating elevated pulmonary vascular resistance. As such, surgical correction of the AV septal defect is contraindicated. A combined heart-lung transplant may be considered for patients with complex cases, poor prognosis, and unresponsiveness to medical therapy.

Discussion

From a management standpoint, most adults with AVSD will have undergone surgical repair during childhood. In cases of complete AVSD — involving both a large Atrial Septal Defect (ASD) e Ventricular Septal Defect (VSD) —, failure to perform early surgical correction — typically before six months of age — can lead to irreversible pulmonary vascular disease and, eventually, Eisenmenger physiology, which rules out the possibility of complete repair.[3]

A 2014 study found that the overall prevalence of PH in adults with congenital heart disease was 3.2%, being more prevalent among older patients.[4] Among patients who previously underwent correction for simple congenital defects, such as atrial or ventricular septal defects, or patent ductus arteriosus, the prevalence of PH was found to be 3%. The risk increased significantly with older age at the time of surgical intervention.[5]

PH in ACHD presents a wide range of clinical manifestations. In this population, management encompasses both non-pharmacological strategies (such as patient education, dental care, vaccination, and psychosocial support) and pharmacological therapies. In recent years, treatment options have expanded with the advent of pulmonary vasodilators targeting various pathways, leading to significant improvements in quality of life.[6] Outcomes for those with PH have also improved, thanks to broader access to targeted PH therapies, advances in perioperative care, and the adoption of multidisciplinary management approaches.[7]

For patients who have undergone surgical repair, long-term follow-up is essential to monitor for potential complications such as left AV valve regurgitation or stenosis, left ventricular outflow tract (LVOT) obstruction due to anatomical distortion, and tachyarrhythmias or bradyarrhythmias. Left AV valve regurgitation is the most

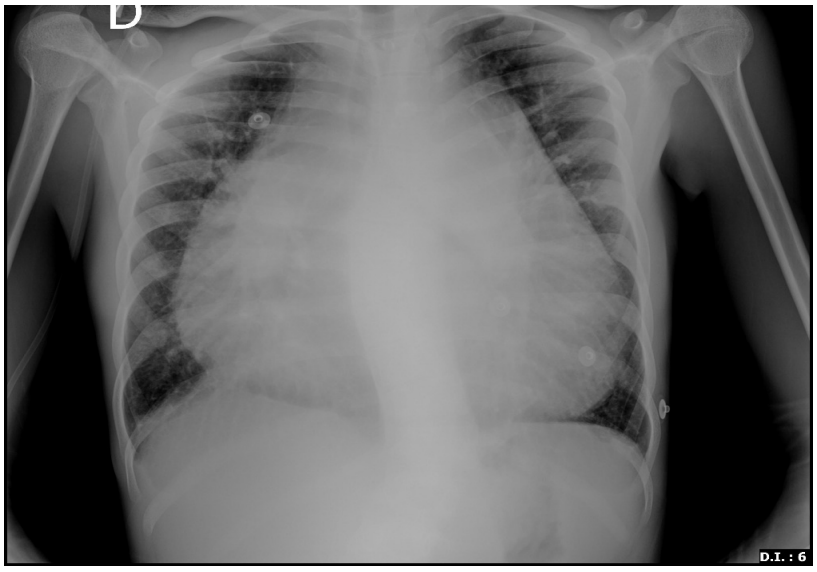


Figure 2 – *Posteroanterior Chest X-ray: Hospital admission. Significant global cardiomegaly.*

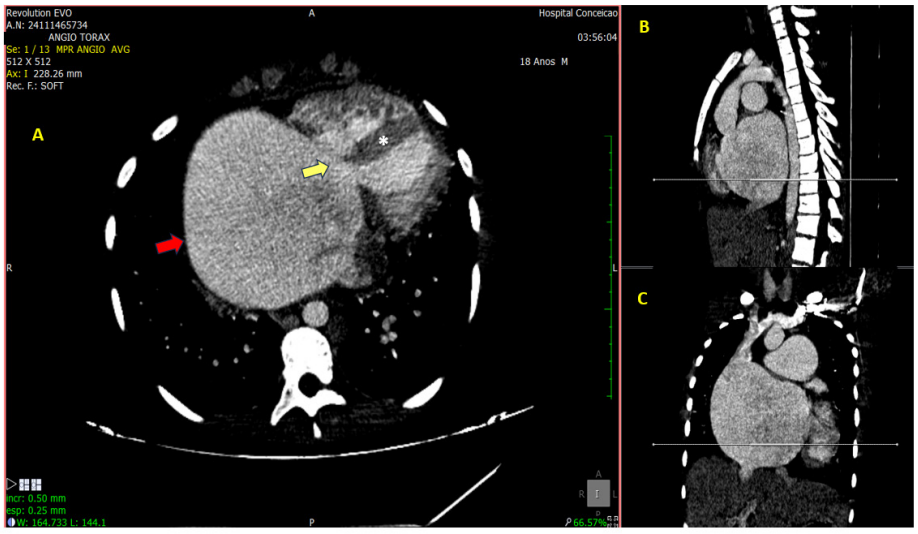


Figure 3 – *Chest CT with multiplanar reconstruction: A) Transverse view showing significant cardiomegaly and presence of AV defect. The red arrow identifies a single atrium with marked enlargement. The yellow arrow shows the AVSD. The asterisk identifies an intact interventricular septum. B) Sagittal view showing dilation of the pulmonary artery. C) Coronal view.*

common cause of late surgical reintervention. There are few long-term follow-up studies of patients after childhood AVSD repair; therefore, the most effective and efficient timing and method of monitoring are still under investigation.[8]

In this case, most AVSD features were successfully identified via 2D echocardiography. However, when surgical repair is a viable option, 3D echocardiography can provide a more detailed view of the AV valve anatomy, enhancing preoperative planning for valve reconstruction.[8,9]

Adults with uncorrected congenital heart disease represent a growing population, characterized by complex clinical scenarios that demand highly individualized therapeutic approaches. In this context, two-dimensional echocardiography becomes an essential tool, not only for accurate diagnosis but also for supporting the development of patient-specific therapeutic strategies.

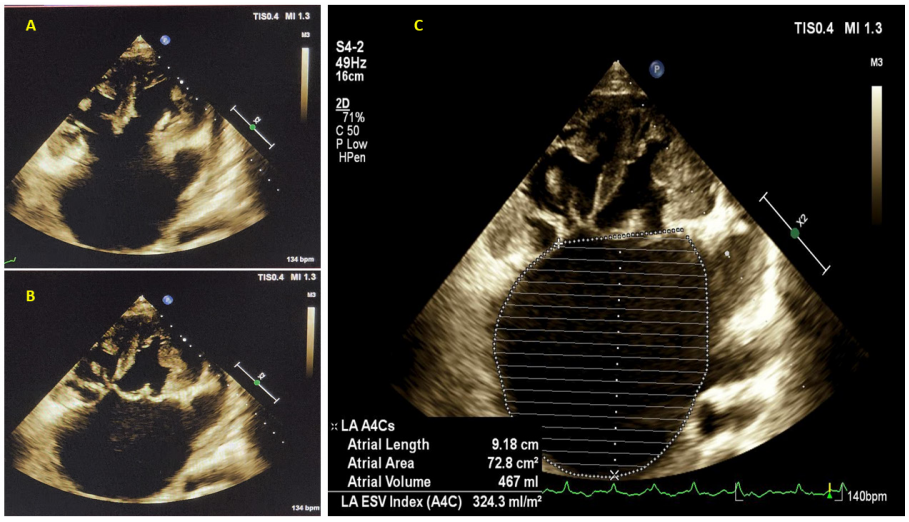


Figure 4 – *Apical 4-chamber (A4C) transthoracic echocardiogram: A) Left and right AV valves are open and aligned in the same plane. B) Left and right AV valves are closed. C) Image shows the single atrium volume.*

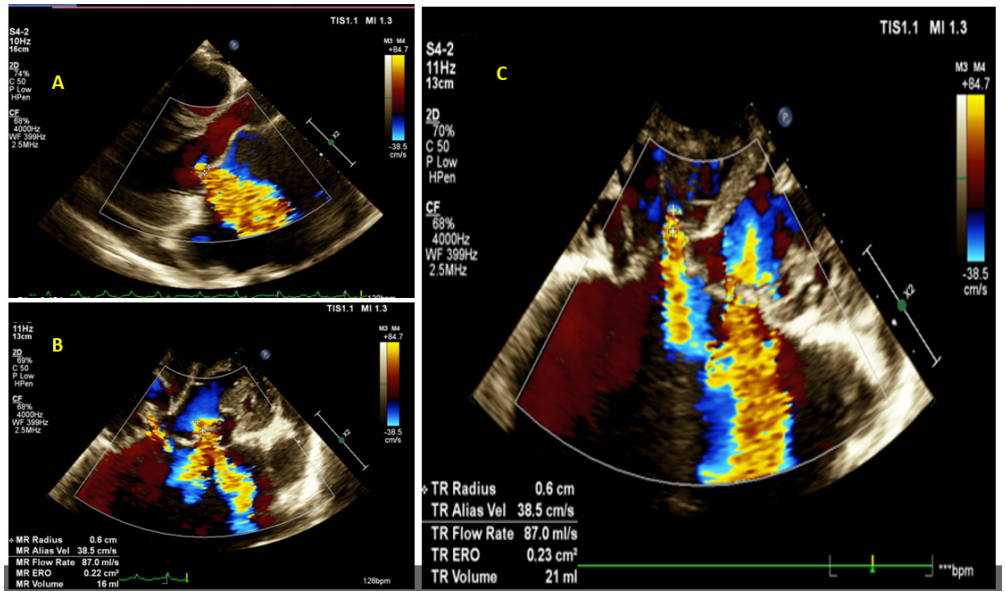


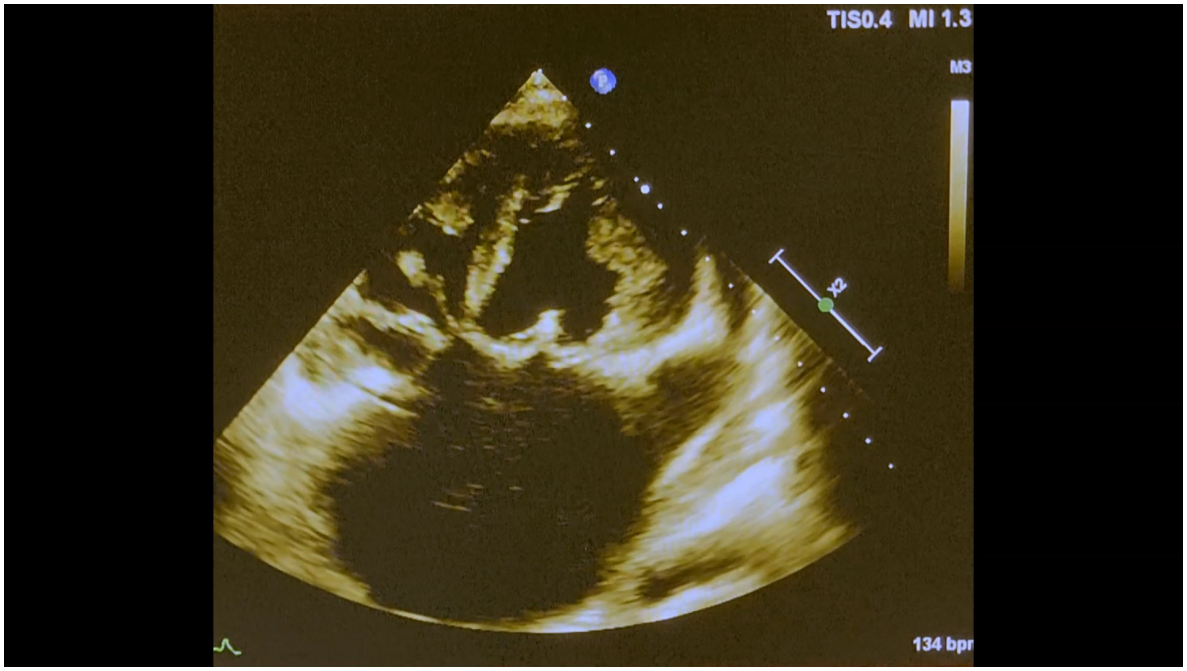
Figure 5 – *Transthoracic echocardiogram: A) Left AV valve in parasternal long-axis view with vena contracta measurement. B) Left AV valve in A4C view with measurements of jet radius, EROA, and regurgitant volume. C) A4C view showing single atrium and regurgitant jets from both AV valves, with right valve jet measurements including radius, EROA, and regurgitant volume.*

Author Contributions

Conception and design of the research: Cunha SR, Barros FC, Pandolfo F, Barcellos PT, Pianca EG; acquisition of data: Cunha SR, Minotto RF, Barros FC, Mazzuca ACM, Pianca EG; analysis and interpretation of the data: Cunha SR, Minotto RF, Barros FC, Pandolfo F, Mazzuca ACM, Barcellos PT, Pianca EG; writing of the manuscript: Cunha SR, Pianca EG; critical revision of the manuscript for intellectual content: Pandolfo F, Mazzuca ACM, Barcellos PT, Pianca EG.

Potential Conflict of Interest

No potential conflict of interest relevant to this article was reported.



Video 1 – *Ecocardiograma transtorácico mostrando valvas AVs esquerda e direita no mesmo plano com ampla comunicação interatrial. Link: http://abcimaging.org/supplementary-material/2025/3801/2025-0014_video_1.mp4*

Sources of Funding

There were no external funding sources for this study.

Study Association

This article is part of the scientific output of the Medical Residency Program in Echocardiography, developed by the residents and preceptors of Hospital Nossa Senhora da Conceição.

Ethics Approval and Consent to Participate

This study was approved by the Ethics Committee of the Hospital Nossa Senhora da Conceição Sá under the protocol number CAAE: 91751125.8.0000.5530, Technical opinion: 7.974.789. All the procedures in this study were in accordance with the 1975 Helsinki Declaration, updated in 2013. Informed consent was obtained from all participants included in the study.

Use of Artificial Intelligence

The authors did not use any artificial intelligence tools in the development of this work.

Availability of Research Data

The underlying content of the research text is contained within the manuscript.

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