Atrial Septal Defect

Rose Kalu Igwe, BS; Richard B. Towbin, MD; Jason N. Johnson, MD; Alexander J. Towbin, MD

Abstract

Atrial septal defects (ASDs) represent a significant congenital heart condition with the potential for severe long-term complications if left untreated. Imaging advances, particularly modalities such as transthoracic echocardiography, cardiac CT, and cardiac MRI, have greatly enhanced the early detection and precise characterization of ASD, allowing for more effective and personalized treatment strategies. These tools not only aid in distinguishing the type and severity of the defect but also provide invaluable guidance for surgical interventions, thereby improving patient outcomes.

Keywords: thorax, cardiac, congenital, shunt

Case Summary

A teenage female presented with worsening fatigue and shortness of breath with physical activity. Her physical exam showed a fixed split-second heart sound and a systolic ejection murmur of the left upper sternal border.

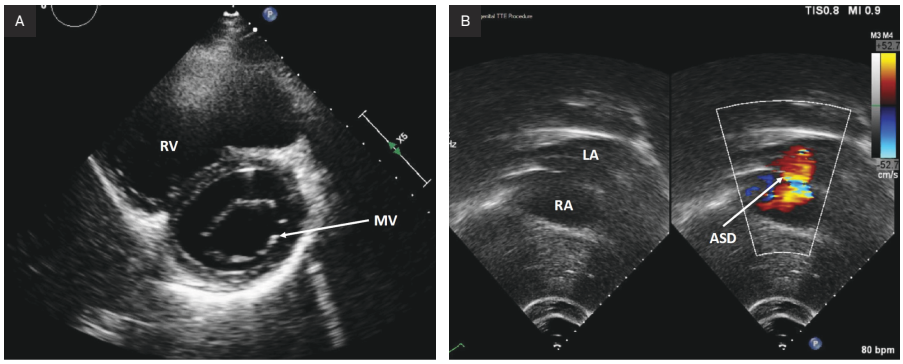
Imaging Findings

Transthoracic echocardiogram (Figure 1) showed a large secundum atrial septal defect (ASD) with left to right shunting and moderate right atrial and ventricular dilation. The cardiac MRI (Figure 2) confirmed the large secundum ASD, moderate right atrial and ventricular chamber dilation, normal pulmonary venous anatomy, and an increased Qp:Qs.

Diagnosis

Atrial septal defect.

Figure 1. (A) Parasternal short-axis transthoracic echocardiogram at end-diastole showing moderate right ventricular chamber dilation. MV, mitral valve; RV, right ventricle. (B) Subcostal coronal color compare at end-diastole showing a large secundum ASD. ASD, atrial septal defect; LA, left atrium; RA, right atrium.



The differential diagnosis for ASD in childhood includes atrioventricular septal defect, total anomalous pulmonary venous return, pulmonary stenosis, and ventricular septal defect.

Discussion

ASD is the second most common congenital heart defect in childhood after ventricular septal defect. ASD occurs in 0.2% of children or 1-2 per 1000 births.[1] In the past 60 years, the rate of ASDs has risen from below 0.5 cases per 1000 live births to around 1.6 per 1000.[1] This increase in prevalence is due to the improvement of imaging modalities and improved clinical detection due to educational and technical advancements of medical practitioners.[1] An ASD is defined as an abnormal opening in the interatrial septum, which leads to blood shunting between the atria.[1] Generally, small ASDs close on their own. However, larger ASDs cause cardiac and respiratory issues into adulthood.[1] ASD is more common in girls than boys, with a male-to-female ratio of 1:2, and is influenced by specific genetic conditions.[1]

Affiliations: A.T Still University- Kirksville College of Osteopathic Medicine, Kirksville, Missouri (Igwe); Department of Radiology, Phoenix Children's Hospital, Phoenix, Arizona (RB Towbin); Heart Institute, Le Bonheur Children's Hospital, Memphis, Tennessee (Johnson); Department of Radiology, Cincinnati Children's Hospital and University of Cincinnati College of Medicine, Cincinnati, Ohio (AJ Towbin).
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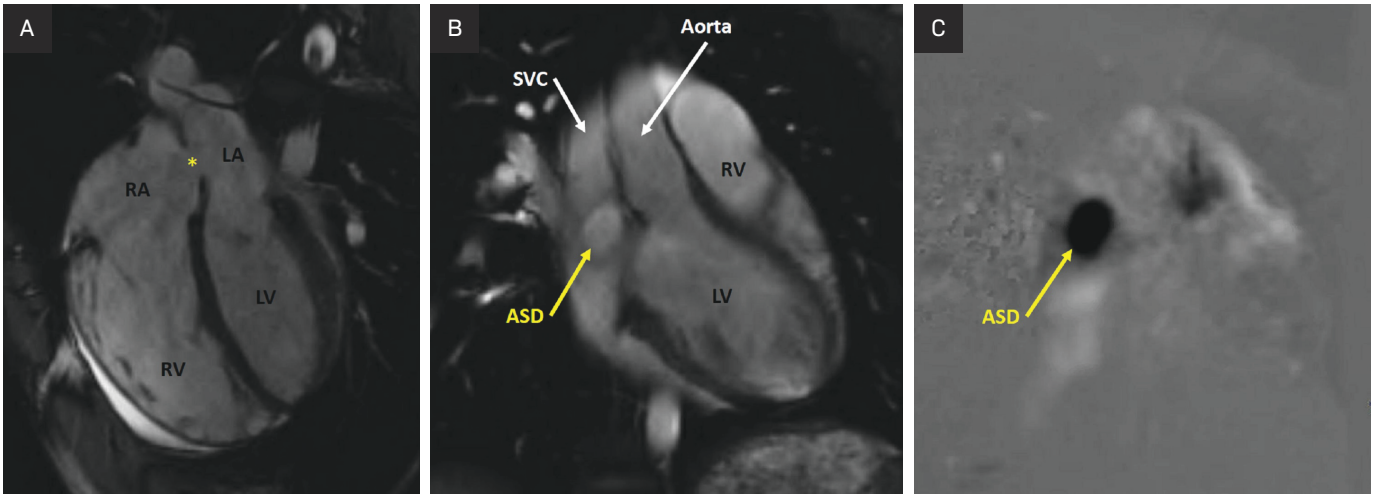
Figure 2. (A) A 4-chamber CMR cine steady state free precession (SSFP) at end-diastole showing a large secundum ASD (yellow asterisk), moderately dilated right atrium and right ventricle, and a small anterior pericardial effusion. The right ventricular end-diastolic volume was 120 mL/m^2 (normal 60-100 mL/m^2). LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle. (B) ASD en face view (off-axis coronal view) CMR cine SSFP at end-diastole showing a large secundum ASD (yellow arrow). ASD, atrial septal defect; LV, left ventricle; RV, right ventricle; SVC, superior vena cava. (C). ASD en face view CMR phase contrast through plane at end-diastole showing the shunting through the ASD (yellow arrow). The flow through the ASD was 4.59 L/minute. The flow across the aortic valve was 3 L/minute, and the pulmonary valve was 7.60 L/minute. The calculated Qp:Qs was 2.5:1. ASD, atrial septal defect.



A variety of syndromes are associated with ASDs as well as other congenital cardiac defects. Holt-Oram syndrome, caused by mutations in the TBX5 gene, affects heart and upper limb development.[1,2] Ellis-van Creveld syndrome, due to mutations in the EVC or EVC2 genes, is strongly associated with dysrhythmias and secundum ASDs and more common in certain Amish and Indigenous Australian communities.[1] In addition to ASDs, patients with this syndrome commonly present with a single atrium.[1] Some families also show a pattern of ASDs linked to mutations in the NKX2.5 and GATA4 genes, emphasizing the complex genetic factors that can contribute to this condition.[1] These associations are heavily influenced by Mendelian inheritance and maternal exposure to alcohol, drugs like cocaine, and rubella.[1]

ASDs involve a true septal deficiency that promotes communication of blood between the atria of the heart.[1] In a normal heart, the interatrial septum separates the right and left atria, preventing oxygenated and deoxygenated blood from mixing.[3] Structurally, the interatrial septum includes the septum primum and septum secundum, which develop during embryogenesis to fully divide the atria.[3] The septum primum begins as a thin membrane growing from the upper part of the common atrium downward, closing the initial opening between the left and right sides.[3] The septum secundum then forms on the right side, partially covering a secondary opening and creating the fossa ovalis, which allows prenatal blood flow between the atria.[3] Proper septal formation allows for appropriate separation of the circulation and oxygen-poor blood to flow into the right atrium and oxygen-rich blood to enter the left atrium.[3] ASDs disrupt this structure by creating openings in the septum that permit abnormal shunting of blood between the atria.[3] Pathologically, these defects typically lead to increased blood flow in the right atrium and ventricle in patients without pulmonary hypertension and normal right ventricular compliance because they create a left-to-right shunt, allowing oxygen-rich blood from the left atrium to flow back into the right atrium.[3] This added volume causes the right heart to enlarge as it accommodates the extra workload.[3] Additionally, the increased blood flow to the lungs raises both blood volume and pressure in the pulmonary circulation, further stressing the right side of the heart and pulmonary vessels.[3]

There are 4 types of ASD: including a secundum ASD, primum ASD, sinus venosus, and unroofed coronary sinus.[3] Secundum ASD makes up 75% of ASD cases and is characterized by a large fossa ovalis or deficient septum primum. It is usually located in the central part of the septum due to increased reabsorption of the septum primum or the septum secundum fails to occlude the ostium secundum.[2] Primum ASD makes up 15-20% of ASD cases and is located at the lower region of the septum because they arise from improper formation of the septum primum and endocardial cushions, which are essential for dividing the atria and ventricles.[1] Primum ASD is often a part of an atrioventricular canal defect and is usually associated with trisomy 21.[2] Sinus venous defect makes up <10% of ASD cases and is usually associated with atypical drainage of the right superior or inferior pulmonary vein into the superior vena cava or inferior vena cava (partial anomalous pulmonary venous return).[1] Lastly, an unroofed coronary sinus, otherwise known as coronary sinus ASD, arises due to maldevelopment in

the wall separating the coronary sinus and left atrium.[3] A notable mention is patent foramen ovale.[3] It occurs when the opening between the right and left atria that is normally present in fetal circulation (fossa ovalis) fails to close after birth.[3] The patent foramen ovale typically closes shortly after birth and is not considered an ASD because no septal tissue is missing.[3]

Clinical appearances of ASDs in children depend on their age, the size of the shunt, and the presence or absence of mitral incompetence.[1] Small ASDs, usually <6 mm in size, are likely to close spontaneously, with closure occurring in 89% of cases for 4-mm defects and 79% for 5-6-mm defects, often within the first year of life.[4] In contrast, defects larger than 8 mm have only a 4% chance of spontaneous closure, with 91% requiring surgical or catheter-based intervention.[2,4] Without intervention, larger ASDs lead to a left-to-right shunt that increases blood flow to the right heart and lungs as long as the pulmonary vascular resistance (PVR) remains low.[4] This overload can cause cardiomegaly, pulmonary plethora, and right ventricular dilation and ultimately hypertrophy, as well as elevated pressures in the pulmonary arteries, leading to pulmonary hypertension.[4] Over time, these effects can also dilate the proximal pulmonary arteries, underscoring the need for timely treatment.[1] In patients with long-standing large ASDs without treatment that develop pulmonary hypertension, the shunting across the ASD becomes right to left and patients develop desaturations.

Imaging plays a major role in the diagnosis, size determination, and course of treatment of ASD.[5] The gold standard for diagnostic imaging for children and adults is the echocardiogram.[5] The transthoracic echocardiography (TTE) is the preferred imaging method for diagnosing ASDs in children due to its noninvasive nature, accessibility, and clear visualization of cardiac structures.[5] It examines the blood flow direction via color Doppler imaging, pressure of the pulmonary artery (PA), size of opening, systemic and pulmonary flow ratio, and to uncover other associated abnormalities.[1] It effectively evaluates the defect's size, location, and any impacts on the heart, especially in younger children who typically provide good imaging windows.[5] However, as children grow older or if more detailed imaging is required for procedures like transcatheter closure, transesophageal echocardiography (TEE) becomes more advantageous.[5] TEE offers superior detail of the septal region and surrounding structures, making it valuable for complex cases or older patients.[5] Thus, age plays a role in choosing the most suitable imaging technique. Other imaging tools that could be used are cardiac CT scan, MRI, catheter angiography, and chest x-ray.

A CT scan provides detailed visualization of the wall thickness and chamber diameter.[1] It could also be used to measure the aortopulmonary ratio of the patient. Cardiac MRI allows for the quantification of the pulmonary blood flow (Qp) to systemic blood flow (Qs) ratio (Qp:Qs), stroke volume, right ventricular volume, and atrial level shunt.[1] Its high spatial resolution allows for precise measurement of the ASD, a critical factor in guiding clinical decision-making and optimizing therapeutic interventions for pediatric patients.[1] Cardiac MRI can be used to calculate the flow across the ASD to help confirm the amount of shunting through the defect, and the flow across the ASD plus the flow across the aortic valve will equal the flow across the pulmonary valve (Qs + QASD = Qp). Catheter angiography is typically employed when assessing older patients or when pressure measures are needed to assess for evidence of pulmonary hypertension.[1] Chest x-rays can also be used to observe clinical status via the identification of enlargement in the heart and PA.[2]

In pediatric patients with ASD, treatment options primarily include transcatheter closure and surgical repair, chosen based on defect characteristics and size, patient age, and overall health. Transcatheter closure is often the preferred method for secundum ASDs in children due to its minimally invasive nature and high success rate in defects up to 35 mm in diameter.[6] To minimize procedural risks, transcatheter closure is usually recommended for patients weighing over 15 kg, with thresholds such as a systolic PA pressure <50% systemic and a PVR below one-third of systemic vascular resistance.[6] This approach has shown success rates exceeding 95%, with benefits including shorter recovery times and reduced hospital stays.[6] However, risks in younger patients may include vascular complications and rare device embolization, mitigated with meticulous pre-procedure imaging.[6]

Surgical repair is typically reserved for larger, complex ASDs or non-secundum defects, such as primum or sinus venosus types, which present anatomical challenges unsuitable for device closure. This method, which also requires careful consideration of PA pressure and PVR thresholds, carries the inherent risks associated with cardiopulmonary bypass, such as infection and bleeding, though it remains effective with low recurrence rates.[6] Given the diversity of device options for transcatheter closure, including the Amplatzer Septal Occluder (Abbott Laboratories, Chicago, Illinois, USA) and Gore Cardioform Occluder (Gore, Newark, Delaware, USA), echocardiography is used to assess defect size and positioning, aiding in device selection and minimizing procedural complications.[6] In pediatric ASD management, both treatment approaches yield high efficacy with careful threshold-based risk management, ensuring successful outcomes with personalized care for each child's specific needs.[6] For patients with ASD who are symptomatic or not immediately eligible for surgery, medical management is essential. Anticoagulants help reduce the risk of clot formation, beta-blockers manage irregular heart

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rhythms, and diuretics help alleviate fluid accumulation.[7] In pediatrics, early detection through prenatal care and advanced imaging, like TTE, plays a critical role in identifying ASDs early, allowing for timely intervention.[7] Together, these medical strategies and diagnostic tools improve outcomes by addressing symptoms and preventing complications until or if surgery becomes viable.

Conclusion

ASDs represent a significant congenital heart condition with the potential for severe long-term complications if left untreated. Imaging advances, particularly modalities such as TTE, cardiac CT, and cardiac MRI have greatly enhanced the early detection and precise characterization of ASD, allowing for more effective and personalized treatment strategies. These tools not only aid in distinguishing the type and severity of the defect but also provide invaluable guidance for surgical interventions, thereby improving patient outcomes.

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