ORIGINAL RESEARCH PAPER

General Medicine

KEY WORDS: Atrial Septal defect, ASD, Sinus venosus, Pulmonary artery hypertension, PAH,

AN ACCIDENTALLY DIAGNOSED CASE OF SINUS VENOSUS TYPE OF ATRIAL SEPTAL DEFECT

Dr. Vishal Anadkat Junior Resident, General Medicine Dr. D.Y. Patil Hospital and Medical College, Nerul, Navi Mumbai.

Dr. Smita Patil Professor, General Medicine, Dr. D.Y. Patil Hospital and Medical College, Nerul, Navi Mumbai

Dr. Nivedita Maulick Professor, General Medicine, Dr. D.Y. Patil Hospital and Medical College, Nerul, Navi Mumbai.

Dr. Harsh Patel* Junior Resident, General Medicine, Dr. D.Y. Patil Hospital and Medical College, Nerul, Navi Mumbai.*Corresponding Author

INTRODUCTION

Atrial septal defect (ASD) is the most common congenital heart lesion in adults and is often asymptomatic until adulthood. Diagnosis is important, as timely ASD repair improves outcomes.

ASDs result from lack of sufficient tissue to completely septate the atria and are classified according to their location in the atrial septum. The location of the defect in relation to adjacent cardiac structures defines the anomalies associated with the ASD and impacts the natural history and requirements for repair.

Atrial septation begins as early as the fifth week of gestation. The septum primum arises from the superior portion of the common atrium and grows caudally to the endocardial cushions located between the atria and ventricles, eventually closing the orifice (ostium primum) between the atria. A second orifice (the ostium secundum) develops in the septum primum; this orifice is covered by another septum (the septum secundum) that arises on the right atrial side of the septum primum. The septum secundum grows caudally and covers the ostium secundum. However, the septum secundum does not completely divide the atria, but leaves an oval orifice (the foramen ovale) that is covered but not sealed on the left side by the flexible flap of the septum primum.

Flow through the foramen ovale is essential for fetal circulation. The foramen ovale closes spontaneously within the first two years of life in 70 percent of children. However, in a significant proportion (20 to 30 percent) of the population, the septa do not fuse, leading to a patent foramen ovale.

Sinus venosus defects account for 5 to 10 percent of ASDs and are located in the venoatrial portion of the atrial septum. Sinus venosus defects represent an abnormality in the insertion of the superior or inferior vena cava, which overrides the interatrial septum; the interatrial communication is then formed within the mouth of the overriding vein and is outside the area of the fossa ovalis. Thus, sinus venosus defects are technically not ASDs since the defect is within the sinus venosus septum. An anomalous connection involving one or more pulmonary veins is present in most patients with sinus venosus ASD (eg, 112 of 115 patients undergoing surgical repair). Sinus venosus defects are of two types:

1. Superior sinus venosus defects are located immediately below the orifice of the superior vena cava. The right upper lobe and middle lobe pulmonary veins often connect to the junction of the superior vena cava and right atrium or on the superior vena cava, resulting in a partial anomalous pulmonary venous connection.

Inferior sinus venosus defects, also known as inferior vena caval defects, are much less common. They are located immediately above the orifice of the inferior vena cava. These defects are also often associated with partial anomalous connection of the right pulmonary veins to the junction of the right atrium and inferior vena cava.

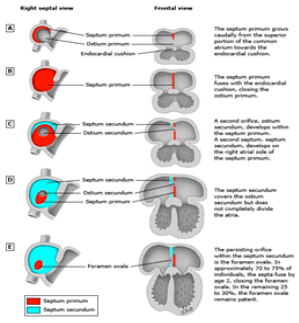

Figure 1: Atrial Septal Defects

Sources: © 2023 UpToDate, Inc. and/or its affiliates. All Rights Reserved.

Case report

A 54-year-old female patient came with the complaints of loose stools, vomiting, and fever of 3 days. On further questioning the patient had complaints of breathlessness. Initially only on exertion then started happening while doing household chores. On examination she was vitally stable, loud P2 was heard on cardiac auscultation. ECG was of normal sinus rhythm but with flat T waves. Chest X-ray revealed cardiomegaly. So 2D Echo was planned which was suggestive of LVEF 55%, Dilated RA/RV, Dilated MPA, Moderate TR, PASP 57 mm Hg, Severe PAH. Cardiac markers were sent, turned out to be BNP 182, Trop I 0.13, CKMB 6.5, D dimer 1005. CT pulmonary angiogram was done to rule out pulmonary thromboembolism. Patient was started on Inj Clexane, Sildenafil, and dual anti platelet therapy. 2D Echo with contrast/bubble test was done to find out the cause of PAH and dilated RA RV. Which was suggestive of some vascular malformation. CTPA senior opinion was taken that revealed the presence of partial anomalous pulmonary venous connection with sinus venosus type of atrial septal defect.

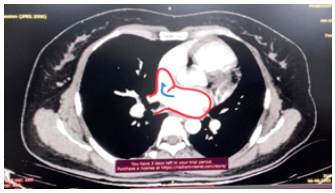

Figure 2: CT Pulmonary Angiogram showing atrial septal defect

Sources: Patient records, Harsh Patel, Dr. DY Patil Scholl of Medicine

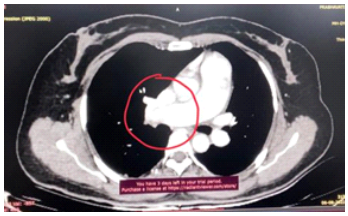

Figure 3: CT Pulmonary Angiogram showing partial anomalous pulmonary venous connection

Sources: Patient records, Harsh Patel, Dr. DY Patil Scholl of Medicine

CONCLUSIONS

An atrial septal defect (ASD) is a defect in the atrial septum and is classified by type based upon location including secundum ASD, primum ASD, superior sinus venosus defect, inferior sinus venosus defect, and unroofed coronary sinus.

ASDs are associated with left-to-right shunt which causes volume overload of the right heart chambers, which is generally tolerated well for years. The development of progressive pulmonary vascular disease and pulmonary hypertension is highly variable and depends not only on the size and duration of the shunt but also the presence of other left-to-right shunts such as partial anomalous pulmonary venous connection.

Most patients with an ASD and significant shunt flow (ie, pulmonary to systemic flow ≥1.5:1) will be symptomatic (with atrial arrhythmias, exercise intolerance, fatigue, dyspnoea, and heart failure) and require closure by the age of 40. However, some patients do not become symptomatic until 60 years of age or older.

The characteristic physical findings with an ASD with large left-to-right shunt and normal pulmonary artery pressures are a right ventricular heave, wide, fixed splitting of the second sound (S2), and pulmonary outflow murmur due to increased volume of flow across the pulmonary valve.

Patients with a systolic murmur at the left sternal border associated with fixed splitting of the second heart sounds, unexplained right ventricular volume overload, or pulmonary hypertension should be referred for evaluation of possible ASD and/or partial anomalous pulmonary venous connection.

The diagnosis and evaluation of an ASD includes demonstration of the presence, location, size, and direction of the shunt as well as evaluation of any associated cardiac lesions and complications including right ventricle (RV) volume overload and pulmonary hypertension.

Echocardiography is the initial imaging modality of choice for the diagnosis of ASD. Transthoracic echocardiography (TTE) can be definitive for ostium secundum and primum defects. Transoesophageal echocardiography (TEE) aids in the diagnosis of sinus venosus defects of superior or inferior type, and the assessment of associated anomalies such as anomalous pulmonary venous connection. TEE is superior to TTE for the sizing and anatomic evaluation of defects for potential device closure.

If TTE and TEE results are inconclusive for the presence and cause of right ventricular volume overload, cardiovascular magnetic resonance (CMR) or computed tomography (CT) imaging may be helpful, particularly for identification of sinus venosus defect or assessment of anomalous pulmonary venous connection.

Cardiac catheterization is not generally required for diagnosis and assessment of ASDs but is most commonly performed for assessment of equivocal shunts, severity and reversibility of pulmonary hypertension and left ventricular diastolic dysfunction, and for device closure of secundum ASD.

REFERENCES:

1. 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. Circulation 2019; 139:e698.
2. Baumgartner H, Bonhoeffer P, De Groot NM, et al. ESC Guidelines for the management of grown-up congenital heart disease (new version 2010). Eur Heart J 2010; 31:2915.
3. Gutgesell HP, Huhta JC. Cardiac septation in atrioventricular canal defect. J Am Coll Cardiol 1986; 8:1421.
4. al Zaghal AM, Li J, Anderson RH, et al. Anatomical criteria for the diagnosis of sinus venosus defects. Heart 1997; 78:298.
5. Attenhofer Jost CH, Connolly HM, Danielson GK, et al. Sinus venosus atrial septal defect: long-term postoperative outcome for 115 patients. Circulation 2005; 112:1953.
6. Van Praagh S, Geva T, Lock JE, et al. Biatrial or left atrial drainage of the right superior vena cava: anatomic, morphogenetic, and surgical considerations--report of three new cases and literature review. Pediatr Cardiol 2003; 24:350.
7. Posch MG, Perrot A, Berger F, Ozcelik C. Molecular genetics of congenital atrial septal defects. Clin Res Cardiol 2010; 99:137.