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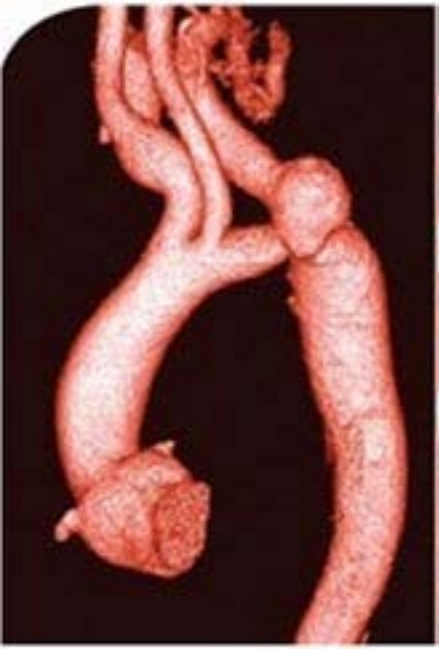
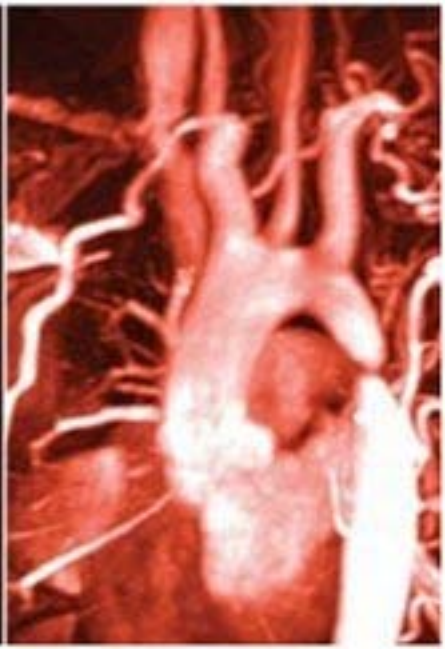
# Adult Congenital Heart Disease

Edited by Carole A. Warnes





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# The AHA Clinical Series

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## Adult Congenital Heart Disease

*Adult Congenital Heart Disease* Edited by Carole A. Warnes
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To Jane Somerville, who taught so many of us about congenital heart disease;
for inspiring me to follow a different path.

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The AHA Clinical Series

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# Adult Congenital Heart Disease

EDITED BY

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# Preface

The last fifty years have witnessed dramatic changes in the world of congenital heart disease; innovative cardiac surgeries, noninvasive imaging, and intensive care have all resulted in the successful survival of the majority of babies born with congenital heart disease. Now, there are approximately one million adults in North America with congenital heart disease, some of whom have had prior surgery and others who were surprised to learn as adults that they were born with heart disease. Although there are now more adults than children with congenital heart disease, the medical community has been ill-prepared to deal with their complex problems. The American College of Cardiology/American Heart Association have recently recognized the importance of this patient population by publishing guidelines to help medical practitioners manage some of their problems. The aim of this book is to offer further practical advice to physicians about common congenital anomalies and associated complications seen frequently in practice.

I believe our best learning experiences result from our clinical cases, and so each chapter of this book begins with a common clinical scenario related to each anomaly. This is followed by a description of the anatomy, features of the clinical diagnosis, a discussion of the imaging modalities, and appropriate treatment strategies. Each chapter then concludes with a discussion about the treatment used for each case and the outcome that resulted. Separate chapters on arrhythmias and imaging are also included.

The authors are an international group of experts in their field, and their contributions are very much appreciated. I hope the readers will benefit from the wealth of clinical experience included herein.

Carole Warnes, MD

# Foreword

The strategic driving force behind the American Heart Association's mission of reducing disability and death from cardiovascular diseases and stroke is to change practice by providing information and solutions to health care professionals. The pillars of this strategy are Knowledge Discovery, Knowledge Processing, and Knowledge Transfer. The books in the AHA Clinical Series, of which *Adult Congenital Heart Disease* is included, focus on high-interest, cutting-edge topics in cardiovascular medicine. This book series is a critical tool that supports the AHA mission of promoting healthy behavior and improved care of patients. Cardiology is a rapidly changing field and practitioners need data to guide their clinical decision-making. The AHA Clinical Series serves this need by providing the latest information on the physiology, diagnosis, and management of a broad spectrum of conditions encountered in daily practice.

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Chapter 1

# Secundum atrial septal defect

Sabrina D. Phillips

*A 20-year-old woman presented for evaluation of palpitations and one episode of near syncope after exertion. Her past medical history was remarkable only for a diagnosis of exercise-induced asthma. She was involved in competitive sports at her University, participating on the track team. She denied any chest discomfort, dyspnea at rest, or perceived exercise limitation. She had never had lower extremity edema, orthopnea, or paroxysmal nocturnal dyspnea. There was no history of cardiac or pulmonary disease in her family. Physical exam revealed a normal jugular venous pressure. The carotid upstroke was normal. The lungs were clear to auscultation and percussion bilaterally. The cardiac exam was notable for a 1+ right ventricular impulse with a normal left ventricular impulse. The first heart sound (S1) was normal, but the second heart sound (S2) was persistently split, with no variation with respiration. The pulmonary component of S2 was mildly accentuated. A systolic crescendo–decrescendo murmur, grade I/VI, was heard at the upper left sternal margin. No diastolic murmur or added heart sounds were heard. There was no evidence of cyanosis, clubbing, or edema of the extremities.*

*Evaluation included a chest x-ray (Fig. 1.1) that demonstrated increased pulmonary vascular markings, enlarged central pulmonary arteries, and cardiac enlargement involving the right heart chambers. An electrocardiogram revealed normal sinus rhythm with right axis deviation and right bundle branch block. The P-R interval was normal. Two-dimensional (2-D) echocardiogram was notable for moderate–severe right heart enlargement, mild tricuspid valve regurgitation, and a secundum atrial septal defect (ASD) measuring 22 mm. The estimated pulmonary artery systolic pressure was 40 mm Hg. The pulmonary valve was normal. A Holter monitor revealed no atrial or ventricular arrhythmias.*

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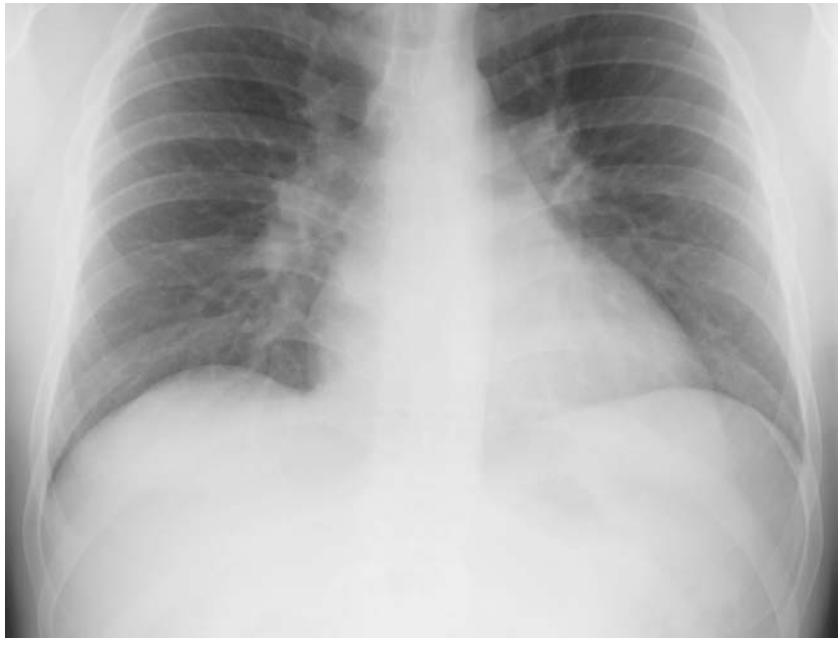

**Fig. 1.1** Chest x-ray demonstrating enlarged cardiac silhouette, enlarged central pulmonary arteries, and increased pulmonary vascularity.

## Embryology and anatomy

Atrial septation is a complex embryological event that occurs in the first 60 days after conception. Throughout the septation process, a channel for blood flow must be maintained between the left and right atria so that placental blood (oxygenated) that is entering the right atrium can be shunted to the left atrium and into the systemic circulation. As endocardial cushion tissue closes the ostium primum, the ostium secundum forms via fenestrations in the anterosuperior position of the septum primum. The septum secundum then develops and eventually provides partial closure of the ostium secundum. At the completion of atrial septation, the limbus of the fossa ovalis is the septum secundum and the septum primum is the valve of the fossa ovalis. Secundum ASDs occur when there is inadequate septum primum. The underlying cause of this malformation is usually multifactorial, although there are a few recognized genetic defects that result in secundum ASD, such as Holt-Oram syndrome.

## Pathophysiology

A secundum ASD allows blood to cross the atrial septum. The amount of shunt is determined by the end-diastolic pressures of the ventricles, the size of the

defect, and the status of the atrioventricular valves. Usually, the right ventricular end-diastolic pressure is lower than the left ventricular end-diastolic pressure, creating a left-to-right shunt. Right-to-left shunting can occur when there is significant tricuspid valve disease or when the right ventricular end-diastolic pressure is elevated by pulmonary valve disease, abnormal compliance of the right ventricle, or pulmonary hypertension.

A left-to-right shunt at the atrial level results in volume overload of the right atrium and right ventricle. Volume overload leads to dilatation of these chambers, which can eventually result in right ventricular systolic dysfunction. Tricuspid valve regurgitation can also progress as a result of right ventricular enlargement and subsequent annular dilatation. Pulmonary artery systolic pressure is proportional to the pulmonary vascular resistance multiplied by the pulmonary arterial flow (PAP $\approx$ PVR $\times$ Qp). Therefore, mildly elevated pulmonary artery pressures are not unexpected, even when the pulmonary vascular resistance is normal, since the pulmonary arterial blood flow is increased secondary to the left-to-right shunt. However, increased blood flow through the pulmonary arteries increases shear stress on the arterial wall and can lead to changes in the pulmonary vasculature that result in increased pulmonary vascular resistance and pulmonary hypertension. In the setting of severe pulmonary hypertension, a right-to-left shunt at the atrial level results in systemic desaturation that is unresponsive to supplemental oxygen. This right-to-left shunt in the setting of pulmonary vascular disease is called Eisenmenger Complex and occurs in approximately 5% of secundum ASDs, most commonly in women. Left atrial enlargement will occur, but left ventricular enlargement is unusual given the compliance characteristics of the left ventricular myocardium.

## Natural history

The presentation of secundum ASD depends on the size of the shunt and the associated cardiac status. Defects associated with very large shunts may present in infancy with failure to thrive, but it is not uncommon for large defects to be diagnosed for the first time in adulthood. Left ventricular compliance tends to diminish with age, often in association with the onset of hypertension or coronary artery disease. The stiffer left ventricle then increases the left-to-right shunt through the ASD, causing progressive right ventricular and right atrial enlargement. Often patients present for the first time in their 40s and 50s with atrial fibrillation. Other symptoms include exercise intolerance, frequent upper respiratory tract infection, and palpitations.

## Diagnosis

The physical exam may demonstrate a pronounced right ventricular impulse, but the left ventricular impulse is usually normal. The second heart sound

(S2) may be widely split, with no variation with respiration. The pulmonary component of the second heart sound (P2) is variably accentuated, depending on the pulmonary artery pressures. A systolic murmur from increased flow through the pulmonary valve is heard at the upper left sternal margin. If the left-to-right shunt is significant (Qp:Qs > 2.5:1), a diastolic murmur representing increased flow across the tricuspid valve can be heard.

The chest x-ray findings of secundum ASD include cardiomegaly related to right-sided cardiac enlargement, central pulmonary artery enlargement, and prominent pulmonary vascularity secondary to pulmonary overcirculation.

The electrocardiogram will usually demonstrate a right bundle branch block with right axis deviation. Crochetage (Fig. 1.2 ), a notch seen in the QRS in lead II and III, has also been reported in secundum ASD [1].

Transthoracic echocardiography is invaluable in the diagnosis of secundum ASD. Even if the defect itself cannot be visualized, the hemodynamic consequence of the shunt can be assessed by an evaluation of the right heart size and function. Identification of the defect by surface echocardiogram is influenced by

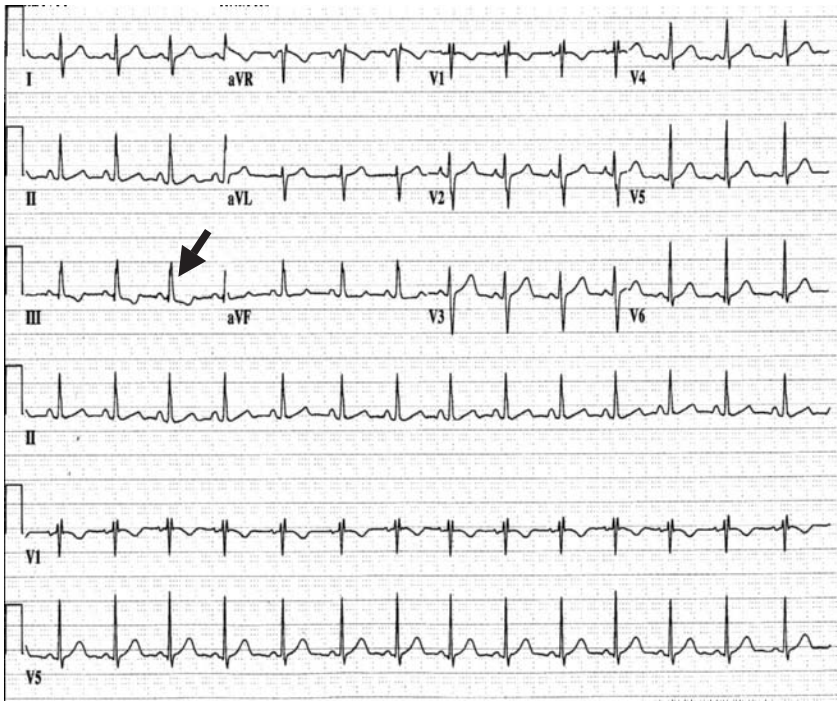


**Fig. 1.2** Crochetage. Note the notch at the peak of the QRS complex in leads II, III, and aVF.

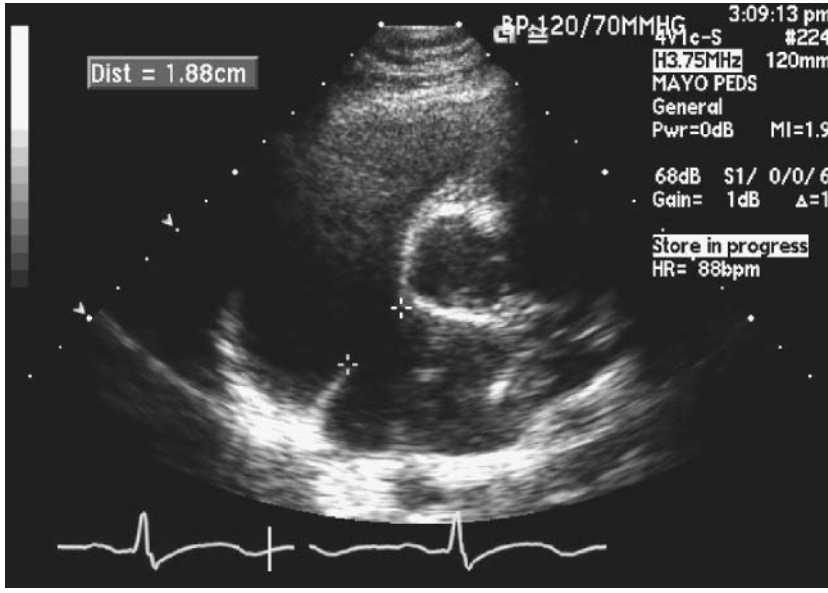


**Fig. 1.3** Transthoracic echocardiogram, parasternal short axis view. The atrial septal defect measures 1.88 cm.

the size of the defect. The subcostal window provides a good look at the atrial septum and should be used. The parasternal short axis view at the base of the heart may also demonstrate the defect (Fig. 1.3). The apical four-chamber view can be misleading; when the atrial septum is parallel to the echocardiographic signal, drop out may occur. Tilting the apical view off-axis will align the atrial septum at an angle and allow for better 2-D and color interrogation. The degree of tricuspid valve regurgitation should be assessed, as this may influence the mode of repair. The pulmonary artery pressures should be estimated via the modified Bernoulli equation ($\Delta P = 4v^2$), with the systolic pressure calculated from the tricuspid valve regurgitation velocity (assuming there is no pulmonary stenosis) and the diastolic pressure estimated from the pulmonary regurgitation end-diastolic velocity. There is very little usefulness in calculating the ratio of pulmonary blood flow to systemic blood flow (Qp:Qs) by echocardiography, as the measurements are often inaccurate.

Transesophageal echocardiography allows improved visualization of the atrial septum and therefore enhanced diagnostic accuracy. Transesophageal echocardiography should be employed if the diagnosis is in question or if there is a concern about the adequacy of the residual atrial septal tissue when device closure is being considered. Transesophageal echocardiography also allows improved visualization of the pulmonary venous return and can be used to

rule out anomalous pulmonary venous connection, which is an important differential diagnosis if a patient is found to have right ventricular enlargement and no ASD.

Cardiac catheterization provides accurate pressure measurements. Flow measurements can be obtained through various methods. These measurements can then be used to calculate pulmonary vascular resistance and quantitate the shunt volume. However, in the modern era, cardiac catheterization is unnecessary unless coronary angiography is being performed or the patient has important pulmonary hypertension. Cardiac catheterization should be employed when there is a question regarding pulmonary artery pressure and vascular resistance before committing to defect closure. Currently, the main role for cardiac catheterization in the patient with isolated secundum ASD is therapeutic.

A secundum ASD that is associated with right ventricular volume loading should be considered for closure. The demonstration of a Qp:Qs greater than 1.5:1 is not required. Magnetic resonance imaging (MRI) can detect ASDs and can provide right ventricular volume measurements, as well as an evaluation of the pulmonary venous return [2,3]. Shunt calculations can be also be determined [4]. MRI is not a first-line test in the evaluation of secundum ASD because of cost, time, and availability constraints, but it should be considered as an excellent alternative in patients who cannot undergo transesophageal imaging.

## Treatment

Hemodynamically significant ASDs (those that have resulted in right heart enlargement) should be closed to prevent the complications of right heart failure, atrial dysrhythmia, and pulmonary hypertension. Closure of important secundum ASDs before the age of 25 years results in an excellent long-term prognosis. Closure after the age of 40 years reduces the complications related to right heart failure, but the increased risk of atrial dysrhythmia remains [5].

Closure of a secundum ASD can be accomplished surgically or percutaneously. Surgical closure has been performed successfully since 1953 [6]. The surgical approach is via midline sternotomy or right thoracotomy. Minimally invasive technique through a right thoracotomy is currently being used in some centers. At the time of surgery, the ASD can be suture closed or patched, depending on the size. Patch closure may involve autologous material, bovine pericardium, or artificial material. The mortality for surgical closure of ASD is reported as 0.3% in the STS database [7] for procedures performed between 1998 and 2002. Complications include incomplete closure, obliteration of the inferior caval orifice, heart block, and atrial arrhythmias.

Percutaneous closure can now be accomplished. There are several devices available for use. Percutaneous closure can be considered for defects up to 38 mm in stretched diameter, but closure becomes more difficult for defects greater than 30 mm. Adequate septal tissue rims must be present to anchor the

device, but patients with deficient retro-aortic rims have undergone successful percutaneous closure. The success rate of percutaneous closure is greater than 90%, with a complication rate of about 7%. Reduction in right ventricular size is seen in the majority of patients. Complications include atrial fibrillation, cardiac perforation, device migration, and access site complications [8]. Infection and thrombosis of the device have been reported after successful closure.

The mechanism of closure should be determined based on anatomic characteristics and patient preference. Patients with other cardiac anomalies that need treatment, including anomalous pulmonary venous return, should undergo surgical repair. Patients with more than moderate tricuspid valve regurgitation may need to be considered for combined surgical ASD closure and tricuspid valve repair. Patients with a history of atrial fibrillation may benefit from a surgical MAZE procedure, although catheter-based arrhythmia management can be considered in conjunction with device closure. Any catheter-based arrhythmia procedure must be performed prior to device closure, as access to the left atrium will be difficult after device implantation.

## Patient follow-up

*The patient elected to proceed with percutaneous closure of the defect. The closure was successful without residual shunt. On a follow-up visit, 1 year after the procedure, the patient was asymptomatic and had improved her 200-meter race times dramatically. Echocardiogram was notable for normal right ventricular size and function, mild tricuspid valve regurgitation, and an estimated pulmonary artery systolic pressure of 28 mm Hg.*

## References

1. Heller J, Hagege AA, Besse B, et al. "Crochetage" (notch) on R wave in inferior limb leads: a new independent electrocardiographic sign of atrial septal defect. *J Am Coll Cardiol* 1996;27:877–82.
2. Chin SP, Kiam OT, Rapaee A, et al. Use of non-invasive phase contrast magnetic resonance imaging for estimation of atrial septal defect size and morphology: a comparison with transesophageal echo. *Cardiovasc Intervent Radiol* 2006;29:230–4.
3. Greil GF, Powell AJ, Gildein HP, Geva T. Gadolinium-enhanced three dimensional magnetic resonance angiography of pulmonary and systemic venous anomalies. *J Am Coll Cardiol* 2002;39:335–41.
4. Beerbaum P, Korperich H, Barth P, et al. Noninvasive quantification of left-to-right shunt in pediatric patients: phase-contrast cine magnetic resonance imaging compared with invasive oximetry. *Circulation* 2001;103:2476–82.
5. Murphy JG, Gersh BJ, McGoon MD, et al. Long term outcome after surgical repair of isolated atrial septal defect. Follow-up at 27 to 32 years. *N Engl J Med* 1990;323(24):1645–50.