Adult Congenital Heart Disease

Douglas S. Moodie, MD, MS

Chairman, Department of Pediatrics, Ochsner Clinic Foundation, New Orleans, LA

One million people over the age of 20 suffer from congenital heart disease in the United States. These adult patients can slip through the cracks of our medical system; many are too old to be cared for in most pediatric institutions by pediatric cardiologists and, unfortunately, most adult cardiologists are not trained in congenital heart disease. Therefore, it is important to identify the common lesions in adult congenital heart disease and how they should be managed. Acyanotic congenital heart disease in the adult population primarily involves left-to-right shunts, such as atrial septal defect, patent ductus arteriosus, and obstructive lesions such as aortic coarctation of the aorta. The most common form of cyanotic congenital heart disease in adults is tetralogy of Fallot. Other complex conditions seen in adults include univentricular hearts, Ebstein's anomaly of the tricuspid valve, and corrected transposition of the great vessels. Most patients with congenital heart disease will need to undergo surgery, catheterization, or catheterization intervention. Results are excellent in the adult population. Long-term follow-up is needed for any adult congenital heart patient receiving care in institutions that are well organized and well equipped, as we learn more about the natural and unnatural history of these conditions.

Moodie DS. Adult congenital heart disease. The Ochsner Journal 2002;4:221-226.

One million people over the age of 20 suffer from congenital heart disease in the United States. In fact, the number of adult patients with congenital heart disease is now greater than those under 20. Adult patients fall into two categories: those patients who were operated on in childhood and are now living well into adulthood, and patients who are first diagnosed as adults.

Adult congenital heart disease can fall through the cracks of our medical system as these patients should receive their care is problematic. Many pediatric cardiologists in children's hospitals do not see patients over the age of 18, while many adult cardiology programs do not provide adequate training in the diagnosis and management of adult congenital heart disease. It is difficult for adults to obtain care in the settings where most pediatric cardiologists practice, namely children's hospitals. The special needs of adult congenital heart patients are also not well addressed by adult institutions, which are dominated by acquired heart disease, particularly coronary artery disease. It is important to have a good understanding of the congenital defects that present in adulthood and how to diagnose, treat, and follow these patients appropriately as adults.

ATRIAL SEPTAL DEFECT

If one excludes a bicuspid aortic valve and does not consider mitral valve prolapse a congenital lesion, atrial septal defect (ASD) is the most common cause of adult congenital heart disease. ASDs account for approximately 10% to 15% of all cardiac defects and are found more commonly in women than men with a ratio of 2:1. There are three major forms of atrial septal defect (Figure 1). The most common type, ostium secundum, makes up 66% to 75% of all ASDs; the ostium primum type (partial atrio-ventricular canal or partial endocardial cushion defect) makes up 15% to 20%. The sinus venosus type comprises 5% to 10% of all ASDs and is almost always associated with partial anomalous pulmonary venous drainage of the right upper pulmonary veins to the right atrium (Figure 1). In most

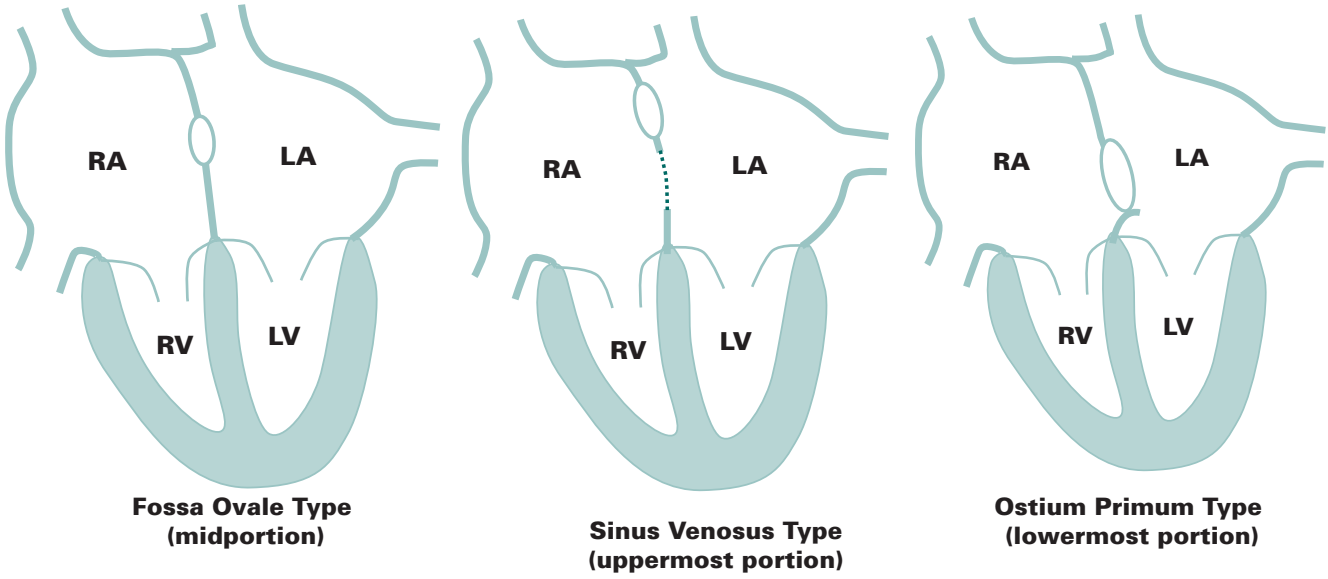

Figure 1. The three major forms of atrial septal defect. Reprinted with permission of the American College of Cardiology.
LV= left ventricle, LA= left atrium, RV= right ventricle, RA= right atrium.

adult series, however, sinus venosus defects make up a larger percentage than the common 5% to 10%. In a previous series, they made up as much as 20% to 25% of atrial defects in adults (1).

ASDs cause a volume-loaded right heart, so that the clinical findings are a dilated right heart with increased pulmonary blood flow. The most common presenting symptoms in adults are shortness of breath, easy fatigability, palpitations, and dyspnea (2,3). Twelve percent of affected adults experience preoperative atrial fibrillation.

The pathognomonic clinical finding in adults with ASD is a fixed split second heart sound, meaning that the second heart sound does not vary with respiration; it does not narrow or become single with a held expiration. Patients with ASD have grade 2 or 3/6 pulmonary flow murmurs, best heard at the left upper sternal border radiating to the back. They frequently also have a low-pitched diastolic flow rumble secondary to increased flow across the tricuspid valve, best auscultated at the right-mid/right-lower sternal border. Occasionally, they will have ejection clicks and may present with a prominent jugular V-wave in the neck.

The chest x-ray and electrocardiogram (ECG) show a volume-loaded right heart, with a dilated right atrium and right ventricle on the chest x-ray, and increased vascularity with prominent pulmonary arteries. The ECG in the secudum ASD shows sinus rhythm, right axis deviation, and right ventricular hypertrophy. In patients with the ostium primum defect, the classic ECG findings are left axis deviation of –20 to –80 degrees. In some adult patients with sinus venosus defects, the P axis is abnormal. Instead of the usual 30 to 60 degrees in the adult, the P axis can be 0 to –10 degrees with an inverted P wave in AVF.

All ASDs with significant left to right shunting should be closed. A recent study has indicated that as many as 20% to 30% of ASDs studied in children enlarged over time (4). In the past, closure meant suture or patch closure. Because of their location, sinus venosus defects almost always need to be closed by a patch directing pulmonary venous blood into the left atrium. Because ostium primum defects are large defects, they almost always undergo patch closure. Currently the procedure of choice to close secumdum ASDs in adults is with a device in the catheterization laboratory. Because of the risk for paradoxical embolus, there is an increased concern that even small atrial defects should be closed. The results from surgery are excellent with an operative mortality of less than 2%, even in adults over 60 years of age. However, despite low operative mortality and morbidity, there are late concerns in the follow-up of ASD patients.

Closing the ASD does not eliminate atrial arrhythmias. In fact, the right atriotomy may need to be modified to prevent reentrant tachycardia. Postoperative atrial fibrillation may occur in as many as 23% of patients. A risk factor for late atrial fibrillation is early atrial fibrillation in the immediate postoperative period. Thus, anticoagulation of all adult ASD patients from 6 weeks to 6 months is a preferred practice. In addition, ASD is the most common underlying congenital defect associated with post-pericardotomy syndrome (an immunologic reaction occurring more than 1 week after the opening of the pericardium

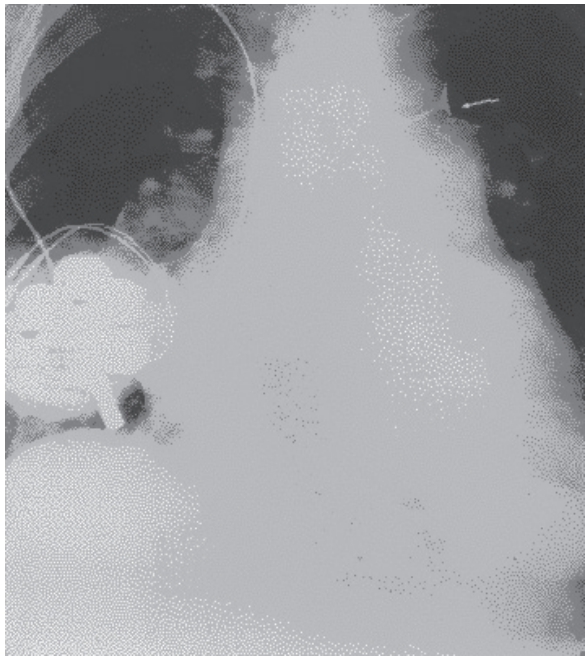

Figure 2. Chest x-ray showing calcification of the ductus. Reprinted with permission of the American College of Cardiology.

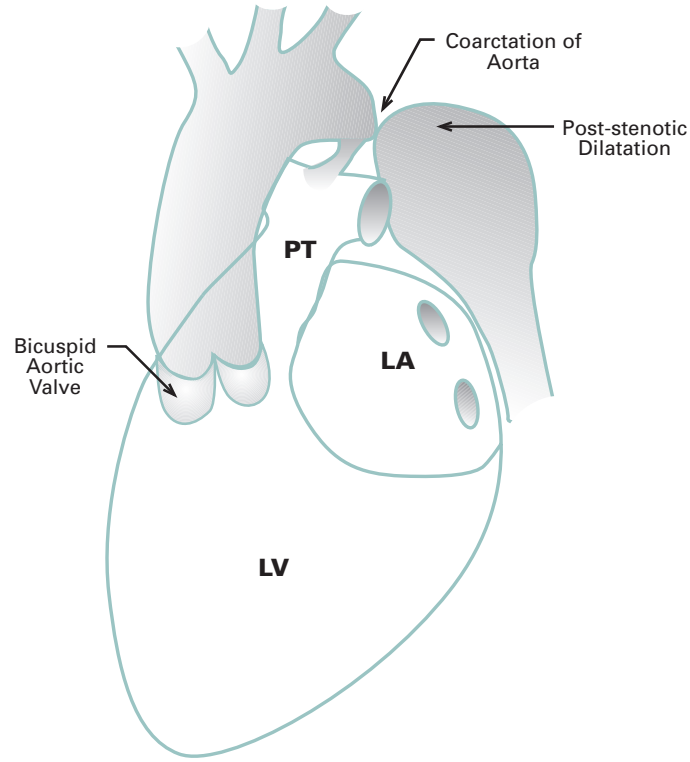

Figure 3. Coarctation of the aorta. LV= left ventricle, LA= left atrium, PT= pulmonary trunk.

characterized by fever, chest pain, and signs of pleural or pericardial inflammation). In addition, the longer we follow patients with ASD, the more concerned we are about late incidence of stroke, which appears not necessarily related to atrial fibrillation. Even though patients with ASD do well with surgical or catheterization laboratory closure of the defect, they demand lifelong follow-up and careful regular medical observation.

VENTRICULAR SEPTAL DEFECT

Ventricular septal defect (VSD) is the most common congenital cardiac defect in children and occurs in approximately 30% of pediatric patients with congenital heart disease. It is much less common in adult patients with congenital heart disease, affecting approximately 10%.

Unlike patients with an ASD, patients with a VSD present with a volume-loaded left heart with progressive enlargement of the left atrium and the left ventricle. They have increased vascularity, and a holosystolic murmur along the left-mid-/left-lower sternal border.

Also unlike patients with ASD, patients with VSD are subject to early pulmonary vascular disease with large shunts when the patient is over 3 years of age. Many small or medium size VSDs will close spontaneously, particularly muscular VSDs. Large VSDs are closed surgically, although there are ongoing trials studying the closure of VSDs in the catheterization laboratory.

PATENT DUCTUS ARTERIOSIS

Between 1951 and 1984 at the Cleveland Clinic, 117 patients (mean age 36 years) with a patent ductus arteriosis (PDA) were evaluated (5). Ductuses occur more frequently in females, similar to ASDs. As in VSD patients, the PDA presents a volume-load of the left heart. Unlike a VSD where there is a holosystolic murmur, however, patients with a PDA present with a continuous "machinery murmur" at the left upper sternal border, because aortic pressure is almost always greater than pulmonary pressure both in systole and diastole. Adult patients will show a dilated left atrium and left ventricle with increased vascularity and may have left ventricular hypertrophy on their ECG. They may also present with calcification of the ductus on chest x-ray (Figure 2).

Almost all PDAs at any age are closed today in the catheterization laboratory and do not require surgery. It has been particularly helpful to be able to close the calcified ductus in the adult because in the past these patients have required cardiopulmonary bypass and a median sternotomy.

COARCTATION OF THE AORTA

In adults, coarctation of the aorta (Figure 3) almost always presents with systolic hypertension and 50% of the time with a heart murmur. The hallmark of coarctation in the adult is absent or diminished

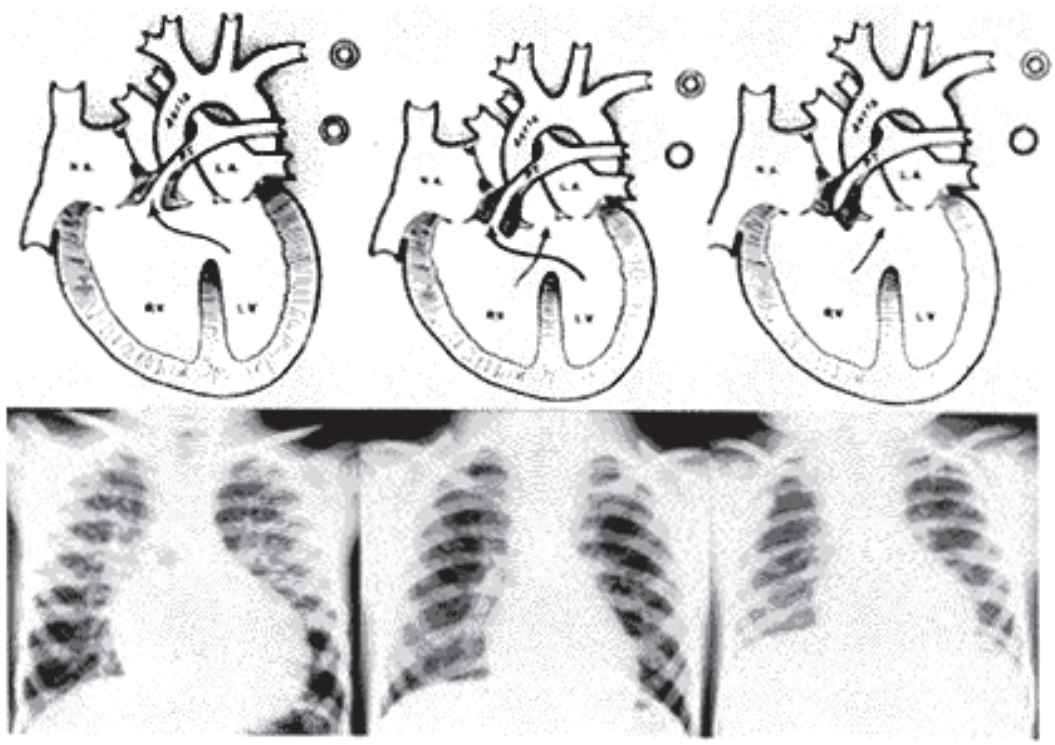

Figure 4. The relationship between the degree of right ventricular outflow track obstruction and chest x-ray findings in Tetrallogy of Fallot. Reprinted with permission of the American College of Cardiology.

femoral pulses with dramatically higher blood pressure in the arms than in the legs. Blood pressures must be taken in both upper extremities and one lower extremity because of the high incidence of anomalous origin of the right subclavian artery, which originates below the coarctation.

Patients with coarctation can have multiple murmurs. They may have a murmur of blood flow across the coarctation at the left upper sternal border. They may also have collateral or continuous murmurs over the anterior and posterior chest, secondary to the increased vessel development of the thyrocervical or intercostal vessels. Sixty to eighty percent of patients with coarctation will also have a bicuspid aortic valve and may present with a murmur of aortic stenosis or aortic regurgitation with an ejection click.

The chest x-ray may show the reverse three sign, and there may be rib notching secondary to erosion of the under-surface of the posterior third to fifth rib by large intercostal vessels. Coarctation can be diagnosed with ECG and MRI.

The approach to coarctation has been surgical, with excellent results obtained in adults. Alternative therapy with balloon dilation and stenting is another approach gaining wider acceptance; however, most institutions would still operate on naive coarctation and reserve catheterization procedures for recoarctation where there is significant scar tissue.

In a long-term study of 69 patients, there was no operative mortality in correcting coarctation in adults (6). However, there was a 22% late mortality in patients followed almost 30 years. This mortality was almost always secondary to complications related to the ascending aorta, such as aortic rupture or dissection and early coronary artery disease. In this study, many of the adults operated on (48% at a mean age of 35 to 45 years) were hypertensive 25 years later.

TETRALOGY OF FALLOT

Tetralogy of Fallot is the most common form of cyanotic congenital heart disease in children, adolescents, and adults. Transposition of the great vessels is the most common form of cyanotic heart disease in infants and newborns. Figure 4 demonstrates the relationship between the degree of right ventricular outflow track obstruction and the chest x-ray findings. In adult patients with minimal obstruction, the shunt may be left to right, and the patient presents as a "pink tetralogy" with cardiomegaly increased vascularity, although they do have right ventricular hypertrophy on the ECG. The key to diagnosis is a decreased second heart sound and right ventricular hypertrophy in a patient that looks like they have a VSD.

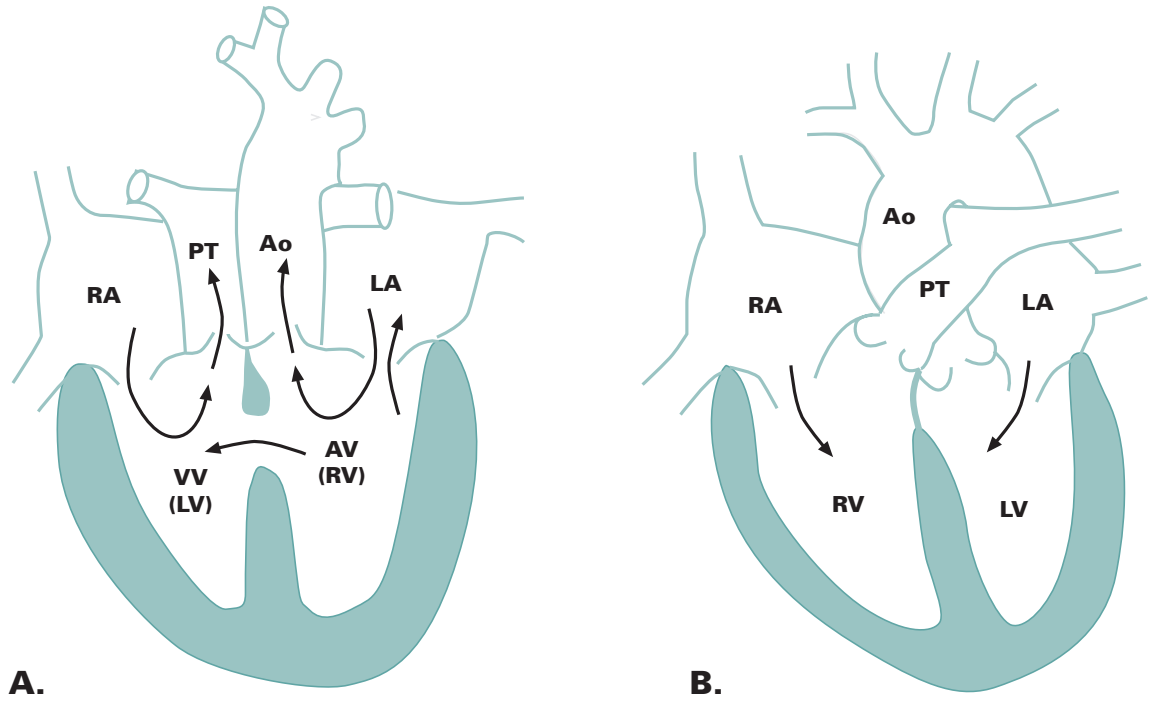

Figure 5. Correcting transposition in Tetrallogy of Fallot. **A.** Corrected transposition. **B.** Normal. LV= left ventricle, LA= left atrium, PT= pulmonary trunk, RV= right ventricle, RA= right atrium, Ao= aorta, VV= venous ventricle, AV= arterial ventricle. Adapted with permission of the American College of Cardiology.

In the patients in the middle panel, the shunt is now bi-directional. The patient may be acyanotic or cyanotic, the heart is smaller, and the vasculary tends to be normal. In the severe form of Tetralogy of Fallot, there is a complete right-to-left shunt because of severe right ventricle outflow tract obstruction. These patients tend to be dyspneic and to squat. They also may present with ventricular arrhythmias, particularly premature ventricular contractions.

The approach to Tetralogy of Fallot is surgical with relief of the right ventricular tract obstruction and closure of the VSD. In most centers today, operative mortality in adults is less than 2%. Again, however, these patients need to be followed up for a lifetime because of a late 1% incidence of sudden death related to the development of ventricular tachycardia. Postoperatively, patients need periodic stress tests and Holter monitor studies.

CORRECTED TRANSPOSITION

Corrected transposition (Figure 5) is ventricular inversion with aorta anterior to the left; thus the blood enters a morphologic right atrium, passes a morphologic mitral valve into the right-sided anatomic left ventricle (7). The blood then passes to the pulmonary artery that is posterior and to the right, but returns from the lungs into a morphologic left atrium, and crosses a morphologic tricuspid valve into a left-sided anatomic right ventricle, which is connected to the aorta. The aorta is anterior to the left instead of posterior to the right. Other frequent cardiac findings in corrected transposition are VSDs, pulmonary valve and subvalvular stenosis, and an Ebsteinian malformation of the left atrio-ventricular valve with left atrio-ventricular valve regurgitation. Heart block will spontaneously develop in 1% of patients per year with corrected transposition.

The approach to these patients is almost always surgical with good results. However, the concern about right ventricular dysfunction and congestive heart failure as well as tricuspid regurgitation has led to earlier and more aggressive surgery.

EBSTEIN'S ANOMALY

Ebstein's anomaly is a downward displacement of the tricuspid valve atrializing a portion of the right ventricle. Some adult patients with Ebstein's anomaly present with severe tricuspid regurgitation. Others will present with cyanosis with exercise secondary to a right-to-left shunted atrial level. Others may present with atrial fibrillation or findings of Wolf-Parkinson-White Syndrome.

Most patients with Ebstein's anomaly will eventually require surgery, usually in early adulthood or childhood. Patients with Ebstein's need a lifetime of vigilance because of a late incidence of sudden death.

CONCLUSION

It is important to understand the congenital heart defects that present in adulthood. Patients with adult congenital heart disease fall through the cracks of our medical system and are often undiagnosed or misdiagnosed as adults. Almost all forms of adult congenital heart disease can be treated with surgery or with catheterization interventional procedures. The results are excellent, but long-term follow-up is mandatory.

REFERENCES

1. Moodie DS. Diagnosis and management of congenital heart disease in the adult. Cardiol Rev 2001; 9:276-281.
2. Marelli AJ, Moodie DS. Adult congenital heart disease. In: Textbook of Cardiovascular Medicine 2nd Edition. Eric Topol, ed. Philadelphia: Lippincott, Williams & Wilkins, 2002, 709-733.
3. Moodie DS, Sterba R. Long-term outcomes excellent for atrial septal defect repair in adults. Cleve Clin J Med 2000; 67:591-597.
4. Brassard M, Fouron JC, van Doesburg NH, et al. Outcome of children with atrial septal defect considered too small for surgical closure. Am J Cardiol 1999; 83:1552-1555.
5. Fisher R, Moodie DS, Sterba R, et al. Patent ductus arteriosus in adults--long-term follow-up: nonsurgical versus surgical treatment. J Am Coll Cardiol 1986; 8:280-284.
6. Matthew P, Moodie DS, Blechman G, et al. Long-term follow-up of aortic coarctation in infants, children, adults. Cardiology in the Young 1993; 3:20-26.
7. Graham TP Jr, Bernard YD, Mellen BG, et al. Long-term outcome in congenitally corrected transposition of the great arteries: A multi-institutional study. J Am Coll Cardiol 2000; 36:255-261.

Dr. Moodie joined the Cleveland Clinic in 1978 as the first Pediatric Cardiologist and headed the Pediatric Cardiology Section. He has been a member of the Best 100 Doctors in America since 1996. He has published 176 papers, written 50 chapters, has had 201 abstracts at national and international meetings, and 51 posters and displays. He was the only Pediatric Cardiologist involved with the Adult Clinical Cardiology Self Assessment Program of the American College of Cardiology in 2000 and 2002 and has spoken at all the major medical institutions in North America, Europe, the Middle East, and the Far East. Dr. Douglas Moodie joined the Ochsner Clinic as the new Chairman of the Department of Pediatrics in February 2002.