palpable thrill at the base of the heart. On auscultation, these patients have an ejection click followed by a crescendo-decrescendo systolic murmur, which is heard best at the left midsternal border and radiating to the right upper sternal border and the neck. Correlation between the degree of stenosis and the intensity of the murmur is not good. However, it is rare for a murmur of 2/6 or less to be associated with severe stenosis. Some patients with aortic stenosis also have aortic regurgitation, in which case a decrescendo diastolic murmur at the left midsternal border that radiates to the apex is detected at presentation.

Diagnosis

Many patients with significant aortic stenosis have LV hypertrophy identified on the ECG. However, the correlation between the severity of stenosis and the finding of LV hypertrophy on the ECG is unreliable. On chest radiography, most patients with severe aortic stenosis have a normal heart size unless there is concurrent aortic regurgitation. Post-stenotic dilation of the ascending aorta is common irrespective of degree of stenosis, and ascending aorta dilation is a common finding. It appears on the chest radiograph as a widened mediastinum.

Echocardiography is the gold standard for evaluation of the severity of aortic valve stenosis and the anatomic morphology of the aortic valve. Cardiac catheterization is primarily indicated to evaluate coronary artery disease before surgical intervention, because approximately one half of adults with symptomatic aortic valve stenosis have concurrent coronary artery disease.

Treatment

Patients with severe aortic stenosis and symptoms or asymptomatic patients with severe aortic valve stenosis and reduced LV systolic function (<50%) should be considered for intervention. Treatment involves manipulating the valve to reduce stenosis. This can be accomplished by transvenous balloon dilation of the valve, open surgical valvotomy, or surgical or catheter-based valve replacement. In absence of significant aortic regurgitation, most centers favor balloon dilation or surgical valvotomy for children and young adults who have pliable valves with fusion of the commissures. In older adults, aortic valve replacement is the treatment of choice.

Prognosis

The natural history of aortic valve stenosis in adults varies but is characterized by progressive stenosis over time. By 45 years of age, approximately 50% of bicuspid aortic valves have some degree of stenosis. Most patients requiring surgical valvotomy to relieve the stenosis before adulthood do well. However, by the 25-year follow-up, up to 40% of patients required a second operation for residual stenosis or regurgitation.

CYANOTIC HEART DISEASE

Tetralogy of Fallot

Definition and Epidemiology

Tetralogy of Fallot (TOF) is the most common cyanotic heart disease seen in adulthood, and it represents 10% of congenital heart defects. It consists of a large VSD, pulmonary stenosis (which may be valvular, subvalvular, and or supravalvular), an aorta that overrides the VSD, and RV hypertrophy.

Pathology

Newborns with TOF are cyanotic because of the right-to-left shunt through the VSD and decreased pulmonary blood flow. The amount of pulmonary blood flow depends on the severity of the obstruction through the RV outflow tract. By the time TOF patients reach adulthood, most have had complete repair or palliative surgery.

Many adults with repaired TOF have had a transannular patch (i.e., synthetic patch across the pulmonary annulus) placed to relieve the RV outflow tract obstruction. This patch causes obligatory free pulmonary regurgitation. Free pulmonary regurgitation can be well tolerated by the right ventricle for many years, but usually in the third or fourth decades, the right ventricle begins to dilate, and it may become dysfunctional. Significant RV dilation and dysfunction can lead to LV dysfunction, significant tricuspid regurgitation, and atrial or ventricular arrhythmias. Almost 29% of adults with repaired TOF also have a dilated ascending aorta due to increased blood flow through the aorta before repair.

Clinical Presentation

Patients with repaired TOF typically have normal oxygen saturation levels. On palpation, there often is an RV lift at the left lower sternal border. On auscultation, there typically is a widely split second heart sound with a to-and-fro murmur in the pulmonary area due to significant pulmonary regurgitation or, less commonly, aortic regurgitation. A holosystolic murmur due to tricuspid regurgitation may be heard at the left lower sternal border. Symptoms in the adult with repaired TOF may include exertional dyspnea, palpitations, syncope, and sudden cardiac death.

Diagnosis

The ECG almost universally reveals a right bundle branch block pattern in patients who underwent repair of TOF. The QRS duration from the standard surface ECG correlates with the degree of RV dilation and dysfunction. A maximum QRS duration of 180 milliseconds or more is a highly sensitive and relatively specific marker for sustained ventricular tachycardia and sudden cardiac death. Patients with significant pulmonary regurgitation often have cardiomegaly with dilated central pulmonary arteries identified on the chest radiograph. A right aortic arch occurs in 25% of cases, and it can be detected by close observation of the chest radiograph. An echocardiogram is useful for evaluating the RV outflow tract (e.g., pulmonary regurgitation, residual stenosis), biventricular size and function, tricuspid valve function, and ascending aortic size. MRI is the gold standard for assessing RV size and function (Fig. 6-2). It can also give an accurate assessment of the degree of pulmonary insufficiency and branch pulmonary artery anatomy.

Treatment

Treatment for TOF is surgical repair. Repair is typically performed between 3 to 12 months of age and consists of patch closure of the VSD and relief of the pulmonary outflow tract obstruction by patch augmentation of the RV outflow tract or