Pathology

A combination of the previously described defects results in interatrial and interventricular shunts, LV-to-RA shunt, and atrioventricular regurgitation. Because these defects include deficiency of the inlet portion of the ventricular septum, the LV outflow tract is lengthened and may be narrowed, producing the characteristic goose-neck deformity.

The natural history for patients with complete AVSD is characterized by the early development of pulmonary vascular disease, leading to irreversible damage that often occurs by 1 year of age, particularly for patients with Down syndrome. Surgery needs to be undertaken early if it is to be successful. Patients who are diagnosed in adulthood can be categorized in two groups: those with Eisenmenger's syndrome and those who had their defects closed in childhood.

Clinical Presentation

On physical examination, most previously repaired patients are cardiovascularly normal. However, patients with significant left atrioventricular (AV) valve regurgitation have a grade 3 or 4 (of 6) holosystolic regurgitant murmur at the apex. For the rare patient with subaortic stenosis, a grade 2 or 3 systolic murmur can be detected at the left midsternal border and radiating to the neck. The physical examination findings for patients with Eisenmenger's syndrome are similar to those for patients with unoperated VSDs.

Diagnosis

On the ECG, first-degree heart block is a common finding for patients with AVSD. All patients have a superior, leftward QRS axis. For those with Eisenmenger's syndrome, the chest radiograph demonstrates cardiomegaly, large proximal pulmonary arteries, and small peripheral pulmonary arteries (i.e., peripheral pruning). Patients who underwent previous repair and have significant systemic left AV valve regurgitation have cardiomegaly with increased vascular markings.

Treatment

Patients who underwent previous repair with significant left AV valve regurgitation causing symptoms, atrial arrhythmias, or deterioration in ventricular function should undergo elective repair or replacement. Previously repaired patients who develop significant subaortic stenosis (i.e., peak cardiac catheterization or echo gradient of ≥50 mm Hg) should undergo surgical repair.

Prognosis

Overall, for patients who underwent early repair before the development of pulmonary vascular disease, the long-term prognosis is good. The most common long-term complication is left AV valve regurgitation, with approximately 5% to 10% of patients requiring surgical revision for left AV valve repair or replacement during follow-up. The second most common long-term complication for this group is subaortic stenosis, occurring in up to 5% of patients after repair. Other long-term complications include residual atrial- or ventricular-level shunts, complete heart block, atrial and ventricular arrhythmias, and endocarditis.

Patients with Eisenmenger's syndrome are symptomatic with exertional dyspnea, fatigue, palpitations, edema, and syncope. Survival is similar to that for other forms of Eisenmenger's syndrome, with a mean age at death of 37 years. In retrospective studies, strong predictors for death included syncope, age at presentation of symptoms, poor functional class, low oxygen saturation (≤85%), increased serum creatinine and serum uric acid concentrations, and Down syndrome.

Coarctation of the Aorta

Definition

Coarctation of the aorta is an abnormal narrowing of the aortic lumen. It constitutes 5% of congenital heart defects. Coarctation of the aorta may occur anywhere along the descending aorta, even below the diaphragm, but in more than 95% of cases, the narrowing is just below the takeoff of the left subclavian artery. In 50% to 85% of cases, there is an associated bicuspid aortic valve. Other associated lesions include VSDs, subaortic stenosis, and mitral valve stenosis.

Pathology

Coarctation of the aorta is an aortopathy of the entire aorta rather than a localized abnormality. In the young, significant coarctation can decrease blood flow to the kidneys, gut, and lower extremities, resulting in severe acidosis and shock requiring immediate treatment. Unrepaired coarctation of the aorta can be seen in adults, but it is rare. Affected individuals develop extensive arterial collateralization to maintain distal perfusion. Most patients seen in adulthood are patients who have had previous coarctation of the aorta repair using a variety of different techniques.

Even after successful repair to relieve the obstruction, multiple studies have demonstrated that patients have persistent abnormalities in the media of the aorta proximal and distal to the coarctation repair site. The stiff aortic wall is characterized by decreased distensibility and endothelial and vascular dysfunction. Examples include resting and exercise-induced hypertension, increased carotid intimal thickness, and abnormal peripheral arterial responses to augmented blood flow and nitroglycerin. Patients with coarctation of the aorta are at increased risk for other left-sided obstructive lesions, particularly a bicuspid aortic valve, which occurs in 50% of cases.

Clinical Presentation

The clinical presentation of coarctation of the aorta depends on the severity of obstruction and the associated anomalies. Unrepaired coarctation of the aorta typically manifests with symptoms before adulthood. Symptoms include headaches related to hypertension, leg fatigue or cramps, exercise intolerance, and systemic hypertension. Untreated patients surviving to adulthood typically have only mild coarctation of the aorta.

Cardinal clinical features in the setting of a significant coarctation of the aorta include upper body hypertension, weak and delayed femoral pulses, and a blood pressure gradient between the right arm and right leg determined by blood pressure cuff. On auscultation, the aortic valve closure sound is usually loud; in the setting of a bicuspid aortic valve, an ejection click, often with a