Section VIII
CONGENITAL HEART DISEASE

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Birth defects occur in approximately 2% of all births. Congenital heart disease comprises almost half of such defects, occurring in approximately 8 in 1000 newborn infants. Many classifications exist for congenital heart disease, and two variations based on a simple physiologic approach follow.

Congenital heart defects can be classified into those that result in cyanosis and those that do not. Acyanotic defects include those with a left-to-right shunt and increased pulmonary blood flow and obstructive defects without associated shunting. Left-to-right shunts occur at various anatomic levels: atrial (e.g., atrial septal defect), ventricular (e.g., ventricular septal defect [part of the complex defect depicted in Fig. 43-1]), or arterial (e.g., patent arterial duct). Obstructive lesions without any associated shunts include pulmonary stenosis, aortic stenosis, and coarctation of the aorta.

Cyanotic defects are generally characterized by a right-to-left shunt and may be classified into two broad categories. In the first group, with intracardiac defects and obstruction to pulmonary flow, cyanosis results from decreased pulmonary blood flow and the intracardiac mixing of oxygenated and desaturated blood. In the second group, cyanosis results from the admixture of pulmonary and systemic venous returns despite normal or increased pulmonary blood flow. In most cardiac malformations classified in this group, a single chamber receives the total systemic and pulmonary venous returns. The admixture lesion can occur at any intracardiac level: venous (e.g., total anomalous pulmonary venous connection), atrial (e.g., single atrium), ventricular (e.g., single ventricle), and great vessel (e.g., persistent truncus arteriosus). Near-uniform mixing of the venous returns usually occurs. Complete transposition of the great arteries (Fig. 43-2) can be included in this group, although only partial admixture of the two venous returns occurs, leading to severe hypoxemia.

**Clinical Indications for Medical or Surgical Intervention**

The interdisciplinary approach that is needed clinically to optimally care for children with congenital heart disease includes accurate assessment of anatomic defects and their physiologic consequences and effective communication of these findings. The consequences of altered blood flow induced by congenital heart disease and the effects of therapeutic interventions invariably influence the pulmonary circulation by increasing pulmonary blood flow (e.g., left-to-right shunting through intracardiac septal defects), decreasing pulmonary blood flow (e.g., right-sided obstructive heart lesions, such as tetralogy of Fallot) (Fig. 43-3), altering the pathway of pulmonary blood flow (e.g., Fontan-Kreutzer repair), or altering the hemodynamics to which pulmonary blood flow (e.g., pulmonary hypertension) is subjected. Successful management can often depend on the ability of the clinician to monitor pulmonary hemodynamics and assess pulmonary vascular impairment.

Critically important to an understanding of the physiologic consequences of these defects are the maturational differences that occur in cardiopulmonary function. For example, cardiac function is subject to maturational changes occurring at the cellular level in a variety of processes, including those in the neurocardiac functional unit: changes in neurotransmitter content, the receptor system, innervation, the effector/transducer systems, and the cellular components affected by autonomic stimulation (Fig. 43-4).
Interrupted Aortic Arch Complex

A. Pathophysiology

- Interrupted aortic arch
- Oxygenated blood flows to upper body
- Beginning closure of patent ductus arteriosus
- Deoxygenated blood flows to lower body (cyanosis)
- Differential cyanosis
- As ductus arteriosus closes, pulmonary volume overload occurs; increased left ventricular flow is shunted through ventricular septal defect thus compounding pulmonary volume overload.
- Left subclavian artery anastomosed to distal segment of aortic arch
- Pulmonary artery banded before ductus division and aortic arch reconstruction

B. Repair (palliative)

- Oxygenated blood flows to entire body through anastomosis.
- Ductus arteriosus divided and oversewn
- Ventricular septal defect
- Left-to-right shunt reduced
Regardless of the anatomic defects, the physiologic consequences necessitating medical intervention, surgical intervention, or both fall into three broad categories—heart failure, hypoxemia/hypoxia, and risk of pulmonary vascular disease—and represent a second approach to children with suspected congenital heart disease (by risk stratification).

Heart failure is defined as the inability of the heart to supply an adequate cardiac output...
CONGENITAL HEART DISEASE

AN APPROACH TO CHILDREN WITH SUSPECTED CHD

Figure 43-3

Tetralogy of Fallot

Pathophysiology

Right ventricular outflow obstruction

Right-to-left shunt through ventricular septal defect

Right ventricular hypertrophy

Intense cyanosis caused by high proportion of deoxygenated blood

Decreased pulmonary flow

Small pulmonary trunk

Aorta shifted to right and overrides defect

Ventricular septal defect

Blalock-Taussig Operation

(palliative)

Anastomosis of subclavian artery to pulmonary artery

Left-to-right shunt through anastomosis of subclavian artery to pulmonary artery

Cyanosis reduced or eliminated by increased proportion of oxygenated blood

Increased pulmonary flow distal to right ventricular outflow obstruction

Increased pulmonary pressure enlarges pulmonary arterial tree

Aorta shifted to right and overrides defect

Ventricular septal defect

(CO) to meet the aerobic metabolic demands of the body, including those incurred by growth; inefficiency of the heart to meet the metabolic demands can also be included in a more liberal definition of heart failure. An alteration in one or more physiologic determinants of ventricular
Figure 43-4

Innervation of Heart: Schema

Superior cervical sympathetic ganglion
Middle cervical sympathetic ganglion
Vertebral ganglion (variation)
Ans a subclavia
Cervicothoracic (stellate)ganglion
1st intercostal nerve
Inferior cervical (sympathetic) cardiac nerve
Thorac ocardiac branch of vagus nerve
2nd thoracic sympathetic ganglion
White ramus communicans
Gray ramus communicans
Thoracic (sympathetic) cardiac branches
4th thoracic sympathetic ganglion

Superior cervical (sympathetic) cardiac nerve
Middle cervical (sympathetic) cardiac nerve
Vagus nerves
Superior cervical (vagal) cardiac nerves
Inferior cervical (vagal) cardiac nerves
Ascending connections

Nucleus of solitary tract
Vagus nerves

Posterior nucleus of vagus nerve

Cardiac plexus
function—preload, afterload, contractility, and HR or rhythm—can adversely affect cardiac performance beyond the compensatory mechanisms, particularly in fetuses or newborn infants, where cardiac function occurs much higher (and hence less efficiently) on the Frank-Starling curve because of maturational aspects. As a physiologic consequence, fetuses and infants are more dependent on mechanisms that increase HR rather than those that increase stroke volume to increase CO in response to increased metabolic demands.

The etiology of hypoxemia (abnormal reduction in the arterial oxygen tension) must be established to determine whether therapeutic intervention is necessary immediately. Hypoxia (inadequate tissue perfusion) is always a medical emergency because high morbidity and mortality are associated with uncorrected metabolic acidosis. Hypoxemia is most often associated with defects characterized by right-to-left intracardiac shunting in which effective pulmonary blood flow is reduced. Pulmonary blood flow may be entirely dependent on the patency of the arterial duct. The arterial duct begins to close shortly after birth, at which time the hypoxemic (and hypoxic) consequences of ductal dependency manifest. Since the 1970s, pharmacologic manipulation of the arterial duct to maintain or reestablish patency by constant intravenous infusion of prostaglandin E1 or E2 has dramatically improved the care of affected children by diminishing hypoxia during transport to a center where diagnostic and therapeutic interventions can more safely take place.

Defining the pathophysiology of pulmonary vascular disease remains a fertile area for research. The primary approach is to study therapeutic interventions to eliminate the risk factors for pulmonary vascular disease (Fig. 43-5) in all children identified at high risk because knowledge of the pathogenesis of these arteriolar changes remains incomplete. Three principal risk factors should be characterized by noninvasive and invasive techniques described subsequently: increased pulmonary blood flow from left-to-right, intracardiac or extracardiac shunting or an abnormal cardiac connection (e.g., septal defect, patent arterial duct, arteriovenous fistula, transposition of the great arteries); increased pulmonary artery pressure from increased pulmonary blood flow or increased pulmonary venous pressure; and hyperviscosity as a consequence of hypoxemia from decreased pulmonary blood flow in right-sided obstructive heart lesions (e.g., tetralogy of Fallot, tricuspid atresia, pulmonary atresia) or hypoxemia from inadequate mixing (e.g., transposition of the great arteries).

Increased pulmonary blood flow can be distinguished physiologically with the concept of independent or obligatory flow, where dependency is defined relative to pulmonary vascular resistance (or impedance). For example, in children with unrestricted ventricular septal defects, the magnitude of the left-to-right shunting, and therefore pulmonary blood flow, depends on the relative difference between pulmonary and systemic vascular resistances (or impedances). As physiologic influences change this relative difference, the ratio of pulmonary to systemic flow changes proportionally. Therefore, this type of shunting depends on the status of the pulmonary vascular bed. In contrast, in children with atrioventricular (AV) septal defects with unrestricted left ventricular (LV)–right atrial (RA) shunting via the abnormal left AV valve, a significant difference in the resistances determining this flow (e.g., LV systolic pressure compared with simultaneous RA pressure) always exists. Therefore, increased flow occurs across the tricuspid and pulmonary valves, independent of the pulmonary vascular resistance. The magnitude of such a shunt is modulated more by ventricular function. Commonly in this clinical setting, pulmonary hemodynamics are further impaired by pulmonary hypertension, increasing the burden on ventricular function and subjecting the child to higher risks of heart failure and accelerated development of pulmonary vascular disease.

Timing for medical or surgical intervention becomes more evident by examining the actuarial consequences of these three risk factors for pulmonary vascular disease: increased pulmonary blood flow, pulmonary hypertension, and hyperviscosity. Increased pulmonary blood flow alone contributes to the risk of development of pulmonary vascular disease, but the time course for irreversible pulmonary vascular
changes is measured in years. In contrast, pulmonary hypertension is a more significant risk, with irreversible changes observed in months to 1 to 2 years. Severe hyperviscosity states and pulmonary hypertension in children with cyanotic heart disease contribute to an extremely high risk of irreversible changes as early as 3 months of age. An optimal time for intervention to decrease the risk associated with the natural history can be determined by overlaying the actuarial experience for specific medical and surgical interventions.

INITIAL NONINVASIVE ASSESSMENT OF CHILDREN WITH CONGENITAL HEART DISEASE

History
The history is critically important for children with suspected congenital heart disease. Because congenital heart disease is most often diagnosed in early infancy, a chronologic approach is simple but effective. The history of pregnancy, labor, and delivery is often helpful (e.g., perinatal asphyxia) with age and developmentally appropriate attention to expected activity. For example, inquiry into the feeding history may be disproportionately important in infants, whereas inappropriate fatigue or exercise tolerance may be important in older children. One issue that cannot be overemphasized in the pediatric age group is growth. Growth is a cardiovascular stress, and absence of growth may be the only manifestation of heart failure.

The family history is often benign but may alert the clinician to relevant issues, such as the incidence of and the genetic predisposition to congenital heart disease. Information about gene-specific etiologies of specific defects (or risk of such expression) will increase the importance of family history and inquiry into genetic predisposition in the near future.

Physical Examination
The physiologic features associated with altered pulmonary artery hemodynamics that are discernible by physical examination can be generally ascribed to features associated with pulmonary hypertension and decreased or increased pulmonary blood flow. Cardiac situs must first be established by means of palpation.

Children with decreased pulmonary blood flow secondary to congenital heart disease present clinically with cyanosis. Cyanosis necessitates approximately 5 g of circulating deoxygenated hemoglobin; therefore, in children with relative anemia, cyanosis may not be as obvious as expected, even in cyanotic congenital heart disease. Despite cyanosis, children with congenital heart disease often seem comfortable, without evidence of respiratory distress—an important distinction to differentiate hypoxemia as a consequence of a parenchymal disorder (leading
to a ventilation/perfusion defect of perfused but underventilated portions of the lungs. Children with congenital heart disease who are cyanotic because of obstruction to pulmonary blood flow have alterations in the second heart sound with a diminished or absent pulmonary component resulting from diminished or absent flow across the pulmonary valve. Despite the most astute clinical efforts, the diagnosis of specific congenital heart defects by means of physical examination is often disappointing and can only be regarded as an initial screening procedure.

The physical diagnosis of pulmonary hypertension is rarely difficult. The cardiac examination predictably consists of a prominent right ventricular (RV) impulse that is either visible or easily palpable at the lower left sternal border or in the subxiphoid area (when present with normal cardiac situs). On auscultation, a single, loud or narrowly split second heart sound with a loud pulmonary component is present. Pulmonary systolic ejection clicks are also common in severe pulmonary hypertension, arising from a dilated, hypertensive proximal main pulmonary artery. Systolic murmurs at the lower left sternal border consistent with tricuspid insufficiency are sometimes present, although tricuspid insufficiency is common and usually presents without a murmur being noted on auscultation. In severe, long-standing pulmonary hypertension, a decrescendo, high-pitched, early diastolic murmur of pulmonary insufficiency may be present along the mid left sternal border. When pulmonary hypertension is accompanied by RV failure, findings of systemic venous engorgement are present, including hepatosplenomegaly and peripheral edema. Abnormal v and a waves may be found during examination of the neck veins.

Features associated with increased pulmonary artery flow are typically related to auscultatory findings from excessive flow crossing normal heart valves (Fig. 43-6). Because the semilunar valves have approximately one half the cross-sectional area of the AV valves, early diastolic murmurs associated with increased flow across the AV valves require more flow than midsystolic flow murmurs associated with flow across the semilunar valves. This point can be a distinguishing feature in quantifying a left-to-right shunt with normal ventricular function because flow across the AV valves must be approximately doubled to auscultate such diastolic murmurs.

**Chest Radiography**

Although more sophisticated imaging modalities exist to provide anatomic and physiologic information regarding the pulmonary circulation, chest radiography is still used routinely as a screening method to determine the status of the pulmonary vasculature, pulmonary parenchyma, and cardiac situs, size, and morphology. Although its role in cardiopulmonary assessment when compared with cross-sectional echocardiographic techniques is challenged, its availability, speed, and usefulness in providing information about pulmonary features suggest that its future as an imaging modality remains secure.

Evaluation of pulmonary hemodynamics by chest radiography includes assessment of pulmonary ventilation and perfusion. Evaluation of perfusion by pulmonary vasculature assessment in chest radiographs is useful to distinguish the pathophysiology of altered pulmonary hemodynamics in children with congenital heart disease. For example, specific diagnostic entities can be considered by evaluating the pulmonary vascularity. Pulmonary vascularity on a posteroanterior or chest radiograph can be classified as normal, increased (Fig. 43-7), decreased (Fig. 43-8), or abnormally redistributed and for which each lung field must be compared with the other fields. For pulmonary arterial vasculature to be identified as increased by chest radiography, an increase in pulmonary blood flow of approximately 100% is required. This helps to evaluate children with left-to-right shunting and correlate physical examination findings. An increase in CO of a similar amount (approximately 100%) is necessary to auscultate an early diastolic ventricular filling murmur across either AV valve.

Diminished pulmonary vasculature typically represents obstruction of blood flow to the lungs and is an ominous radiographic finding in newborns. Central dilation and peripheral pruning of pulmonary arterial vessels is noted in more advanced pulmonary vascular disease and is found with evidence for RV hypertrophy as defined by retrosternal filling on the lateral chest radiograph with the cardiac silhouette.
Infant with respiratory distress (including orthopnea and tachypnea) caused by pulmonary volume overload

Perspiration and tense, anxious facies

Flared nostrils

Sternal retraction

Intercostal retractions

Pulmonary edema presents a more distinctive pattern of haziness in the lung fields that warrants immediate investigation about etiology because significantly increased morbidity and mortality are associated with this finding. Specific assessment of the size of the main pulmonary artery is possible by means of the chest radiograph. Because the pulmonary artery is thin walled, it dilates readily when exposed to increased flow or pressure. Dilation of the main pulmonary artery is readily visible on the chest radiograph, and differentiating radiographic features are then sought to determine the physiologic etiology.
Evaluation of lung ventilation by assessment of conducting airways and lung parenchyma, including lobar and lung volumes, provides information about pulmonary physiology. Evaluation of the cardiac situs and chamber enlargement by cardiac contour evaluation can greatly aid in the assessment of altered pulmonary hemodynamics. Because the right ventricle is affected by altered pulmonary hemodynamics, attention must be given to changes in shape and
size. However, defining changes in RV function by means of chest radiography is less sensitive and specific than by means of evaluation of pulmonary perfusion.

Additional noninvasive assessment of children with congenital heart disease includes application of echocardiographic techniques (chapter 44). Less frequently, an invasive approach, involving cardiac catheterization, is required (chapter 45).

FUTURE DIRECTIONS

Clinical emphasis has focused on optimizing diagnosis and treatment of children with congenital heart disease and including fetuses as patients. In the future, the clinical focus will include the prevention of congenital heart disease through a more complete understanding of the influence of cardiac development. Completion of the initial mapping phase of the Human Genome Project in 2003 will result in accelerated investigations into the control and modulation of gene expression in the development of the human heart. This expanded understanding of cardiac development may allow interventions to augment specific structural and functional deficiencies and to prevent maldevelopment of the human heart.

REFERENCES


