Congenital Heart Defects: Cyanotic and Acyanotic

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Congenital heart disease is the most common congenital abnormality at birth, occurring in approximately 0.8% of live births. The majority of patients with a severe problem will manifest this during the first year of life, usually during the first month. Apart from the high incidence of patent ductus arteriosus in premature babies, the incidence of congenital heart disease has not changed during the past five decades and is similar in different countries and cultures. There is approximately a fivefold increase in the incidence of congenital heart disease in children and siblings of patients with congenital heart disease. This familial contribution to heart disease probably will increase as more patients with congenital heart disease live to childbearing age because of advances in interventional techniques and surgery.

Practically all congenital heart abnormalities are now amenable to palliative or definitive repair with acceptable morbidity and mortality. Therefore, appropriate and timely referral is vital in the optimal management. Most congenital abnormalities are due to abnormal “plumbing” and not an abnormal or diminished pump force. In contrast to most adult heart disease, only a few congenital lesions are primarily due to abnormal pump function. This review suggests that the clinical manifestations of congenital heart disease are best approached and interpreted by keeping in mind the segmental anatomy of the heart, the relationship of any clinical findings to a specific phase of the cardiac cycle, and to hemodynamic events and their results.

EDUCATIONAL OBJECTIVES
1. Present an approach to the understanding, diagnosis, and management of patients with congenital heart disease.
2. Introduce a simplified segmental classification of congenital heart disease.
3. Review common clinical manifestations of congenital heart disease as they present to the general pediatrician.

CLASSIFICATION
The various types of congenital heart disease and their consequences can be classified into the following basic categories. This classification is based on the...
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abnormal origin of the coronary artery by echocardiography or angiography, as surgical correction is possible and lifesaving. Other conditions, such as myocarditis, dilated cardiomyopathy, or metabolic disorders, occasionally may recover naturally, respond to correction of the metabolic problem and treatment of heart failure, or require cardiac transplantation.

Left to Right Shunt With an Obstruction. Right-Sided Obstruction. This results in right-sided venous congestion and manifestation of right heart failure. The shunt, however, compensates for the reduced flow. For example, in pulmonary stenosis and associated patent ductus arteriosus, the pulmonary blood flow obstruction is compensated by the ductal flow.

Distal Left-Sided Obstruction. An example is a patient with a ventricular septal defect and coarctation of the aorta. This combination has a larger left to right shunt and more severe congestive cardiac failure than either lesion alone.

Proximal Left-Sided Obstruction. In this type of problem, the shunt may partially compensate for the stenosis if pulmonary hypertension develops. Thus, a baby with aortie stenosis and patent ductus arteriosus may have better systemic cardiac output due to pulmonary to aortic shunt at the ductus level.

Cyanotic Congenital Heart Disease

Abnormal Communication Proximal to Right-Sided Obstruction. This results in a right to left shunt when the right- and left-sided pressures in the proximal chambers equalize. A common example of this is tetralogy of Fallot and tricuspid atresia with atrial septal defect. The clinical presentation and prognosis depend on the degree and rate of progress of the right to left shunt, the resultant cyanosis, polycythemia, and their complications.

Abnormalities of Alignment Between Various Segments. The most common examples of this group are D-transposition of the great arteries, truncus arteriosus, and anomalous pulmonary venous return. These lesions result in cyanosis with or without increased or decreased pulmonary blood flow. Polycythemia and its consequences then will ensue.

CLINICAL PRESENTATION

History and physical examination are still the most important means of diagnosis and timely management of congenital heart defects. Congenital heart defects can present in neonates, infants, or children.2 The
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Important step is to realize which patients need referral and whether it should be urgent or elective.

Neonates and Infants
The majority of patients who have severe congenital heart disease present during the first year of life, especially during the neonatal period. Early presentation of congenital heart disease in neonates and infants is usually due to severity or complexity of the disease.

Cyanosis. 
Cyanosis is caused by deoxygenated blood in the systemic capillaries with at least 5 g of reduced hemoglobin in the blood. Cyanosis may be central or peripheral depending on whether desaturation involves only peripheral capillaries or arterial blood as well. Peripheral cyanosis usually is due to sluggish capillary circulation with increased extraction of oxygen from the capillary bed due to low systemic cardiac output, congestive cardiac failure, peripheral constriction in cold weather, anxiety, or peripheral vascular disorders. Central cyanosis may be due to cardiac or pulmonary causes or rarely disorders of hemoglobin. Cardiac causes are usually due to right to left shunting or severe irreversible pulmonary hypertension. Differential cyanosis indicates cyanosis that is more pronounced in the lower extremities than the upper limbs or vice versa. The usual pathologic cause is reversal of blood flow through a large ductus arteriosus so that the lower half of the body receives desaturated pulmonary arterial blood. In transposition of the great arteries with pulmonary hypertension and a large ductus, the upper half of the body has a lower saturation than the lower half.

Clubbing. Significant and prolonged cyanosis eventually causes clubbing of the nails. This occurs in all untreated cyanotic congenital heart disease.

Hypocyanotic Spells in Tetralogy of Fallot.
These spells need emergency referral because of potential risk of death. The spells are caused by hypoxemia associated with low pulmonary output. Severe spells may be hard to differentiate from seizures and colic in infants, or temper tantrums in early childhood. The typical clinical sequence is severe cyanosis and shallow, rapid breathing associated with tachycardia, irritability, pallor, and marked respiratory distress. This happens due to reduced pulmonary blood flow. Extreme hypoxemia may cause loss of consciousness or a seizure. During the attack, the pulmonary systolic murmur disappears or decreases in duration and intensity. Knee-chest position, squatting, oxygen, and intravenous morphine may abort the attack. This vicious cycle can reverse because the patient relaxes, breathing is improved, and oxygen demand is decreased. Daily use of oral propranolol may prevent further episodes.

Tachypnea and Dyspnea. Tachypnea in a neonate is a rapid respiratory rate of more than 60 breaths/minute. Severe dyspnea in an infant often presents with grunting and substernal retractions (respiratory distress). There is often air trapping and wheezing due to edema of the bronchi. Exercise and stress accentuate dyspnea. In infants, this happens with crying, feeding, and defecation. In cyanotic conditions, cyanosis is intensified by activity and physical exertion. In severe obstructive lesions and severe pulmonary hypertension, exertion results in low cardiac output, pallor, and even syncope. Infants with large left to right shunt or with congestive cardiac failure may have tachypnea, respiratory distress, sweating during feeding, and failure to thrive.

Older Children
In addition to symptoms of congestive cardiac failure, children with congenital heart defects may present with the following manifestations that would indicate referral for further cardiac evaluation.

Exercise Intolerance. Most children may not have apparent symptoms despite a structurally important congenital heart disease. They frequently participate in all normal activities. However, symptoms may occur with stress and strenuous activity. An exercise test or Holter monitor may elucidate this.

Chest Pain. This is a fairly common complaint in otherwise healthy children. A detailed history and examination is the best evaluation tool. The exact cause of chest pain remains uncertain in 40% to 50% of patients. Significant chest pain can occur in the following situations:

- patients with structural abnormalities, left ventricular outflow obstruction, severe pulmonary hypertension, mitral valve prolapse, or congenital coronary anomalies,
- patients with acquired myopericardial disease, pericarditis, coronary artery disease, or Kawasaki disease, and
- patients with arrhythmias.

Dysrhythmias. The lethal primary dysrhythmias include prolonged Q-T interval syndrome, catecholamine-sensitive ventricular tachycardias, atrial flutter with low AV block, atrial fibrillation associated with Wolff-Parkinson-White syndrome, and congenital complete AV block. A history of resting- or exercise-induced dizziness or syncope in patients with one of these arrhythmias is an indication for referral.

Syncope and Sudden Death. Sudden death may occur in association with cardiovascular disease in children. Of the patients who die suddenly, approxi-
continuously 10% are engaged in active sports and a history of syncope occurs in 16%. Unoperated severe aortic stenosis is the most common lesion in syncope. Exercise-induced dizziness or syncope should be regarded seriously and investigated further.

HELPFUL PRIMARY TESTS FOR DIAGNOSIS

Electrocardiogram
The electrocardiogram is an effective tool in diagnosis of dysrhythmias, assessment of chamber enlargement, hypertrophy, and myocardial ischemia.

Chest Radiograph
The chest radiograph demonstrates the pulmonary vascular pattern, cardiac size and contour, and associated lung or musculoskeletal abnormalities.

Echocardiogram
An echocardiogram provides detailed information about cardiac anatomy and hemodynamics. The M-mode is used for chamber and vessel measurements, and quantitative assessment of systolic and diastolic functions. The two-dimensional or real-time echo outlines the anatomy and dynamics of the heart and its exact abnormalities. The Doppler echo demonstrates normal and abnormal cardiac hemodynamics including qualitative and quantitative data on shunts, cardiac output, valvar gradients, and regurgitation.

Holter Monitor
This determines whether dysrhythmia or ischemia is responsible for symptoms such as chest pain, dizziness, syncope, and dyspnea.

Exercise Testing
Exercise testing unravels latent dysrhythmia, ischemia, significant cardiac decompensation, and inadequate cardiac output or reserve by imposing additional demands on the patient through graded exercise. This is done by documenting heart rate and rhythm, blood pressure, ST-T changes, duration of exercise tolerance, and symptoms, by exercising the patient to 80% of his or her maximal heart rate, development of symptoms and EKG abnormality.

REFERENCES