Critical Congenital Heart Disease

Jean Ballweg, M.D.
Division of Pediatric Cardiology
Le Bonheur Children's Medical Center

Outcome without screening:

Critical congenital heart disease involves lesions that can result in severe cyanosis and tissue hypoxia and/or profound low cardiac output. If a child has critical congenital heart disease that goes unrecognized the end result at worse is death. Many neonates who are discharged with unrecognized critical congenital heart disease will present with severe cyanosis and oxygen deprivation to the tissues or circulatory shock with acidosis from inadequate cardiac output to the systemic circulation. This can result in injury to the brain, kidneys, liver, adrenal glands, and intestine that are severe and irreversible. Additionally, multiple studies have demonstrated a relationship between poor preoperative condition and poor operative outcome with respect to both morbidities and mortality.

Incidence:

Congenital heart disease occurs in 7 to 9 of 1000 live births in the United States. One fourth of these infants have critical congenital heart disease, which is defined as severe and life-threatening disease requiring surgical or catheter intervention in the first month of life.

Outcome with screening:

The goal of screening is to identify neonates at risk for critical congenital heart disease. It will not ensure that every infant with critical congenital heart disease is identified prior to discharge but should aid in the assessment of those infants who may otherwise look well but have a slight abnormality in the systemic saturation which may allow for identification of heart disease because of further investigation. Some cardiac lesions that are critical and will require early intervention will not be able to be identified with pulse oximetry screening. Additionally, there will be a small percentage of infants who will have a false negative screen and still have critical congenital heart disease. With the early recognition of critical congenital heart disease, the goal is to prevent the significant changes in tissue oxygenation and end organ damage by immediate intervention with medications, catheter based interventions and/or surgical interventions.

Causes of Congenital Heart Disease:

In the majority of cases, the cause of congenital heart disease is unknown. However, genetic and environmental factors do appear to play roles in the abnormal cardiac development. There are multiple genetic mutations that have been isolated and associated with specific congenital heart lesions. These lesions can present alone or as part of a wider pattern of birth defects or inherited disorders. These patterns include, but are not limited to Trisomy 21, Noonan syndrome, Turner syndrome, Alagille syndrome, Holt-Oram syndrome and velocardiofacial syndrome.

Environmental factors including use of certain medications, chronic illnesses of the mother and acute viral illnesses during the first trimester of pregnancy can increase the risk of abnormal cardiac development. Additionally, some studies have suggested that the consumption of alcohol and use of cocaine in pregnancy may increase the risk of abnormal cardiac development.

Screening Test and Confirmation:

The screening test will be pulse oximetry as described in the flow diagram provided by the Tennessee Genetics Advisory Committee. With a failed screen, the infant will be referred for further evaluation by a pediatric cardiologist. A physical examination of the infant and pediatric echocardiogram will be performed to evaluate the intracardiac anatomy. Additional testing to rule out pulmonary or infectious etiology to the failed test may also be performed simultaneously. A positive screen predicts the presence of CCHD in 13-20% of the cases. Many babies with critical congenital heart disease (primarily coarctation) will pass the screen and still have the potential to present very ill.

Treatment:

Treatment of critical congenital cardiac disease will be dictated by the type of cardiac lesion identified. Initially, many infants will require stabilization with prostaglandin infusion to maintain patency of the ductus arteriosus. Additionally, many will require cardiac catheterization and surgical procedures to palliate or correct the abnormality.

These infants can be medically fragile. Many will require medications for short or long term following catheter based or surgical interventions. Some infants will require additional surgical procedures and cardiac catheterizations, depending upon the diagnosis. These infants will require long term follow up with pediatric cardiology specialists.