Critical Congenital Heart Disease

What is critical congenital heart disease?
Critical congenital heart disease (CCHD) is a name given to seven specific critical congenital heart defects. These defects or abnormalities in the structures of the heart have happened before birth. They may either block the blood flow or cause it to flow through the heart in an abnormal way. If left untreated it can lead to the infant’s death or serious developmental delay.

In the United States about 4800 babies are born with a critical heart defect each year. Heart defects are the most common birth defect and are the leading cause of death from birth defects.

How does critical congenital heart disease affect an infant?
Sometimes when an infant is born, parts of the fetal (before birth) blood-pumping system can continue to work hiding the signs and symptoms of CCHD. This makes it possible for an infant with CCHD to appear healthy and be discharged home without knowing they have a heart defect. Signs and symptoms are related to the type and severity of the heart defect. An infant may show shortness of breath, turn a grayish-blue color, have poor feeding habits, cold hands and feet, or be unusually sleepy or inactive.

What causes Congenital Heart Defects?
Despite much research in the majority of the cases heart defects occur without any known cause. However, since genetic and hormonal factors play a role in the development of the heart in the fetus, some defects may arise due to genetic “syndromes”. During pregnancy other health and environmental factors may contribute as well: maternal obesity, gestational diabetes, German measles, systemic lupus erythematosus, cigarette smoking, certain medications and alcohol and narcotic drug use.

Is there a test for critical congenital heart disease?
Yes. Some defects can be detected by your doctor prior to your infant being born. However, studies have shown that despite prenatal assessment and physical examinations, many infants are not diagnosed until after discharge from the hospital.

Now your baby will be tested using pulse oximetry as an additional part of the newborn screening at the hospital or center of birth prior to discharge home. Pulse oximetry is a simple test that measures the amount of oxygen in the blood. The test is painless and only takes a few minutes. Pulse oximetry has been proven to be successful in identifying some forms of CCHD. However, the pulse oximetry screening test does not replace a complete physical examination by the doctor. It is important to understand that even if an infant “passes” the pulse oximetry screening test he/she may still have a small chance of having a heart defect.

How can CCHD symptoms be treated?
In most cases, CCHD can only be treated immediately with medication. Once the diagnosis is confirmed, surgery may be necessary. Infants with CCHD may have lifelong medical problems even with surgical intervention.

DISCLAIMER: The information contained on these pages is not intended to replace the advice of a cardiac or genetic metabolic medical professional.
References:

- Kemper, A.R, Mahle, W.T., Martin, G.R., Cooley, W.C., Kumar, P., Morrow, R.W. et al. Strategies for Implementing Screening for Critical Congenital Heart Disease: Recommendations of the United States Health and Human Services Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children