

Newborn Screening ACT Sheet

[Hypoxemia]

Critical Congenital Heart Disease

Differential Diagnosis: Critical congenital heart disease (CCHD); congenital heart disease (CHD); patent ductus arteriosus (PDA); persistent pulmonary hypertension of the newborn (PPHN); pulmonary and infectious diseases; metabolic, endocrine, and hemoglobin disorders.

Condition Description: Critical congenital heart disease (CCHD) is a subset of congenital heart disease (CHD), comprising about 25% of CHD cases. CCHD includes common truncus arteriosus, hypoplastic left heart syndrome, pulmonary valvular atresia, Tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great vessels, and tricuspid valvular atresia. These are ductus dependent malformations in which the children appear well but when the ductus closes the patient can deteriorate rapidly. There are other types of CCHD (such as coarctation of the aorta) that, although less likely to cause arterial desaturation, may be detected by physical examination and/or pulse oximetry. CCHD may be associated with decreased levels of oxygen in the arterial blood (hypoxemia) and may bring a significant risk of morbidity and mortality if not diagnosed soon after birth.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Inform the family of the newborn screening result.
 - Evaluate the infant for cardiac and non-cardiac causes of hypoxemia.
 - If non-cardiac cause for hypoxemia has been excluded, consult with a pediatric cardiologist to arrange for urgent echocardiogram.
 - Transfer to appropriate referral center, if necessary.
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Diagnostic Evaluation: Echocardiogram and cardiology consultation to determine cardiac anatomy and function. Noncardiac causes of hypoxemia are to be addressed by studies appropriate to such etiologies as infectious or pulmonary disease.

Clinical Considerations: CCHD comprises ductus-dependent malformations in which the newborn may appear well, but when the ductus closes the patient can deteriorate rapidly. Signs of CCHD may include visible cyanosis, tachypnea, murmur, difficulty feeding, and failure to thrive. If untreated, CCHD may lead to significant morbidity or mortality. Health care providers will want to remember that the diagnosis of CCHD, even when a specific anatomical cardiac defect is identified, necessitates that a variety of etiologies, some of them heritable or genetic, be explored. While CCHD often occurs as an isolated finding, it may occur with other anatomic features in genetic syndromes of chromosomal origin (Down syndrome, Turner syndrome and 22q11.2 deletion syndrome.) Single gene mutations, specifically affecting cardiac development or resulting in recognized syndromes (Alagille syndrome and Noonan syndrome), have been identified as causing CCHD. Copy number variation may play an as yet undefined role in CCHD.

Additional Information:

[CDC Website](#)

[AHA/AAP Article](#)

[Genetics Home Reference](#)

Referral (local, state, regional and national):

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

Local Resource Site *(insert local and regional newborn screening website information)*

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

Additional Information:

CDC

www.cdc.gov/ncbddd/heartdefects/index.html

Referral *(local, state, regional and national):*

Find Genetic Services

<http://www.acmg.net/GIS/Disclaimer.aspx>

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