ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

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Replacement of
2013 CCHD ACT Sheet

ACT Sheet

ACMG website: bit.ly/3ZgZTEg

Newborn Screening ACT Sheet

[Abnormal oxygen saturation on pulse oximetry]

Critical Congenital Heart Disease (CCHD)

Differential Diagnosis: Types of CCHD include coarctation of the aorta, double outlet right ventricle, Ebstein's anomaly, hypoplastic left heart syndrome, interrupted aortic arch, pulmonary atresia, single ventricle, tetralogy of Fallot, total anomalous pulmonary venous return, d-transposition of the great arteries, tricuspid atresia, truncus arteriosus, other cardiac lesions. Non-CCHD disorders that also may be detected include hemoglobinopathies, sepsis, pulmonary disease (acquired or congenital), hypothermia, non-critical cardiac lesions, persistent pulmonary hypertension.

Condition Description: Cardiac malformations are the most common birth defects in newborns. Critical congenital heart disease comprises about 25% of congenital heart disease and is defined by requiring surgical or catheter-based intervention in the first year of life. Many are ductal dependent lesions in which the newborn appears well but may deteriorate rapidly when the ductus closes; most are associated with hypoxemia. Critical congenital heart disease may present as an isolated finding or be associated with other congenital abnormalities. A significant proportion have a genetic etiology which can cause both isolated heart defects and syndromic disease.

You Should Take the Following IMMEDIATE Actions:

- Inform the family of the newborn screening result.
- Consult with pediatric cardiologist.
- Evaluate the newborn (cyanosis, tachypnea, hypotension, hypoxemia, cardiac murmur; simultaneously test for infectious, pulmonary, and hematologic etiologies). If any of these signs are present, or if the newborn is ill, transport for further treatment in consultation with a neonatologist and/or pediatric cardiologist.
- Provide family with basic information about the specific type of CCHD and its management in consultation with a pediatric cardiologist.

- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: Echocardiogram: is required for the diagnosis of CCHD and should be performed while evaluation for other etiologies is ongoing. Molecular genetic testing: may be required to identify the cause and inform management of CCHD.

Clinical Considerations: CCHD comprises a subset of congenital heart disease in which the newborn may appear well but can deteriorate rapidly. Without intervention, CCHD can lead to significant morbidity or mortality. Pulse oximetry will not identify all types of cardiac disorders and not all forms of CCHD present with hypoxemia. Screening accuracy depends upon testing a newborn that is awake and not agitated. Some newborns who screen positive with pulse oximetry will not have CCHD but may have other types of congenital heart disease or other non-cardiac conditions including sepsis, hypothermia, lung disease, persistent pulmonary hypertension, or a hemoglobinopathy. Treatment and management of CCHD is based upon the specific lesion.

Additional Information:

- How to Communicate Newborn Screening Results
- Medline Plus
- American Academy of Pediatrics CCHD Resources
- AAP Patient Resources

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

- Find a Genetics Clinic Directory
- Genetic Testing Registry