

Critical Congenital Heart Disease (CCHD) Screening

Screening

The Florida Genetics and Newborn Screening Advisory Council recommended to the Department of Health that CCHD be added to the panel of disorders screened in Florida in January 2011. The United States Department of Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders for Newborns and Children (SDACHDNC) added CCHD to the Recommended Uniform Screening Panel (RUSP) in September 2010.

A panel consisting of members from both the Genetics and Newborn Screening Advisory Council and the Children's Medical Services Cardiac Technical Advisory Panel and other interested parties recommended the screening algorithm recommended by the CDC, AAP and SDACHDNC. Both the right hand and one extremity should be tested for oxygen saturation values. It is also recommended that the baby be over 24 hours of age to reduce the false positive rate.

According to the August 2013 Pediatrics article entitled "Factors Associated with Late Detection", the Florida Birth Defects Registry (FBDR) researchers found that 22.9% of infants born between 1998 and 2007 ultimately diagnosed with CCHD did not receive a CCHD diagnosis during their birth hospitalization.

Prenatal ultrasound examinations and physical examination findings detect most congenital heart defects, estimates are up to 75% of all CCHD cases. The addition of pulse oximetry screening to the assessment of the newborn will identify more cases but there will be some conditions that will not be detected using all three detection methods. Pulse oximetry screening is intended to identify infants with CCHD who appear healthy but have subclinical hypoxemia. It is important to note that a pass reading of a pulse oximetry device does not exclude the presence of a non-critical cardiac disorder because there is a low sensitivity for left to right shunting lesions.

The specificity of pulse oximetry screening is 99%. The sensitivity of pulse oximetry screening is 69.6%

More information about CCHD can be obtained at the following websites:

<http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/nominatecondition/reviews/cyanoticheart.pdf>

<http://www.aap.org/en-us/advocacy-and-policy/aap-health-initiatives/PEHDIC/Pages/Newborn-Screening-for-CCHD.aspx> (This link is especially helpful for primary care providers to navigate through the implementation process and provide effective, long-term medical homes for babies with CCHD. Topics include the rationale for screening, implementation, data collection and management, and screening in special settings such as home or high altitudes. This site also offers links to additional CCHD resources.)

<http://www.cdc.gov/ncbddd/pediatricgenetics/cchdscreening.html>

It is anticipated that the pulse oximetry testing will identify at least seven critical congenital heart defects that account for 17-31% of all congenital heart diseases. It is important to note that not all cases will be identified through pulse oximetry screening and there is lower sensitivity for left to right shunting cardiac lesions.

Hypoplastic Left Heart Syndrome (HLHS)

Pulmonary atresia, intact ventricular septum (PA)

Tetralogy of Fallot (TOF)

Total Anomalous Pulmonary Venous Return (TAPVR)

Transposition of the Great Arteries (TGA)

Tricuspid atresia (TA)

Truncus arteriosus (Truncus)

| <i>Heart Defect</i> | <i>Hypoxemia</i> | <i>Ductal dependent</i> | <i>Birth prevalence</i> | <i>Age at onset of symptoms</i> | <i>Untreated survival</i> |
|----------------------------------|------------------|-------------------------|--------------------------------------|--|--|
| HLHS | All | All | 1-5 to 6-7/10,000 live births | Immediately or within the first two months of life | Universally fatal without surgical intervention |
| Pulmonary atresia, intact septum | All | All | 3/10,000 live births | Immediately | When the ductus closes, the neonate becomes severely ill, leading to death if not urgently treated |
| TOF | Most | Variable | 3/10,000 | Neonatal period | Amount of pulmonary blood flow obstruction determines onset and severity of symptoms |
| TAPVR | All | None | 1-4% of all congenital heart defects | Immediately or within the first two months of life | Survival of the neonate is unlikely if the obstruction is left untreated |
| TGA | All | Variable | 2-3/10,000 live births | Immediately | Onset and severity of symptoms depend on anatomical and functional variants; if there is not adequate blood flow, the neonate will die |
| Tricuspid atresia | All | Variable | 1-3% of all congenital heart defects | Immediately or within the first month of life | Cyanotic neonates that are ductal-dependent are critically ill |
| Truncus arteriosus | All | None | 2-4% of all congenital heart defects | By two months of life | Fewer than 25% will survive past the first year of life without surgical intervention |

*Source: SDCHDNC Report dated 9/3/2010

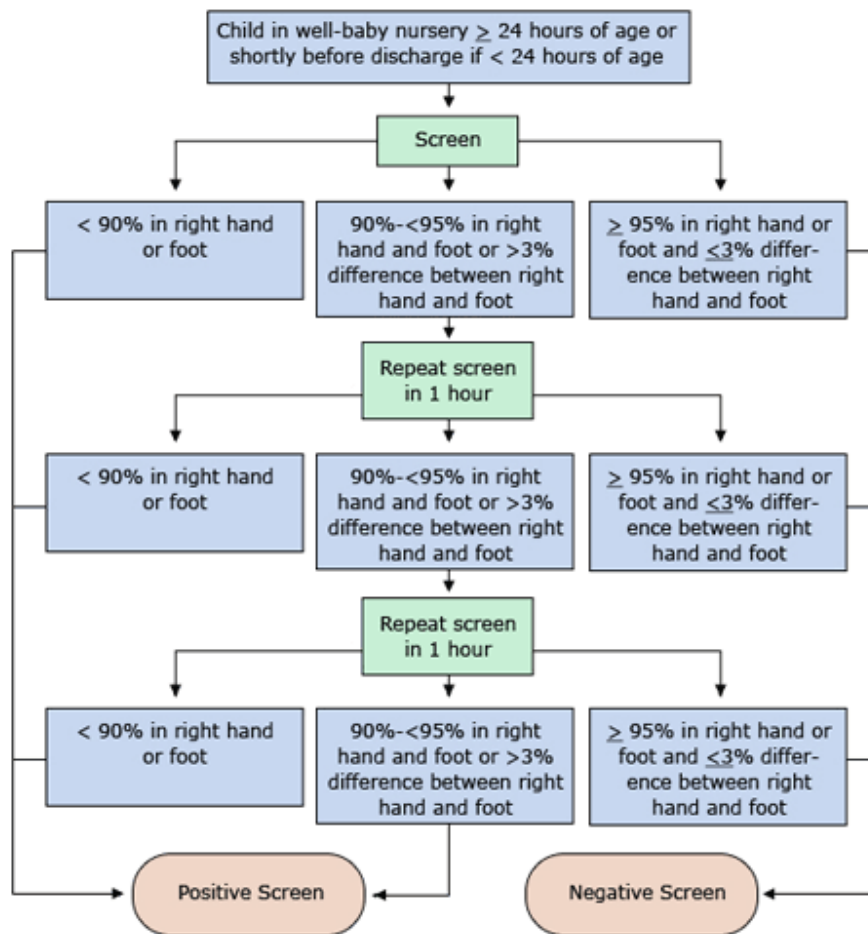
Pulse Oximetry (POX) Screening Algorithm for Normal, Term, or Well-Baby Nursery Infants
(and NICU infants who have been off oxygen for 24 hours who are not discharged on oxygen)

Pulse oximetry screening is intended to be in addition to, not instead of the newborn screening physical examination.

Although the screening algorithm was developed for term and well babies, it is recommended that all babies receive a pulse oximetry test before hospital discharge. Babies that have been admitted to the NICU (Neonatal Intensive Care Unit) should be off oxygen for 24 hours before the screening test is administered. The test should not be administered to a baby who is scheduled to be discharged on oxygen.

A pass on CCHD screening does NOT exclude the presence of a non-critical cardiac disorder. If a cardiac evaluation is indicated (e.g. clinical signs or prenatal diagnosis of congenital heart disease), proceed with evaluation even if infant achieves pass on CCHD screening.

The American Academy of Pediatrics and the Centers for Disease Control and Prevention recommend the following algorithm for pulse oximetry screening:



Reporting POX Screening Results on the Newborn Screening Specimen Card

If the infant passes or fails the pulse oximetry test on the first, second or third attempt, put the last oxygen saturation reading, bubble in “Pass” or Fail” and include the date of the last reading on the newborn screening specimen card.

| | | | | | | | |
|------------------------------|----------------------------|------|---|---|---|---|---|
| PULSE OXIMETRY RESULT | | | | | | | |
| RH(%) _____ | LE(%) _____ | M | M | D | D | Y | Y |
| <input type="radio"/> PASS | <input type="radio"/> FAIL | DATE | | | | | |

The specimen card should not be delayed if the hearing or pulse oximetry screening results are not available at the time that the specimen is ready for transport to the Newborn Screening Laboratory

Reporting POX Screening Results Through Web-based Portal (eReports)

Currently under development. More information will be provided at a later time.

What To Do If Infant “FAILS” the Pulse Oximetry Screening Test

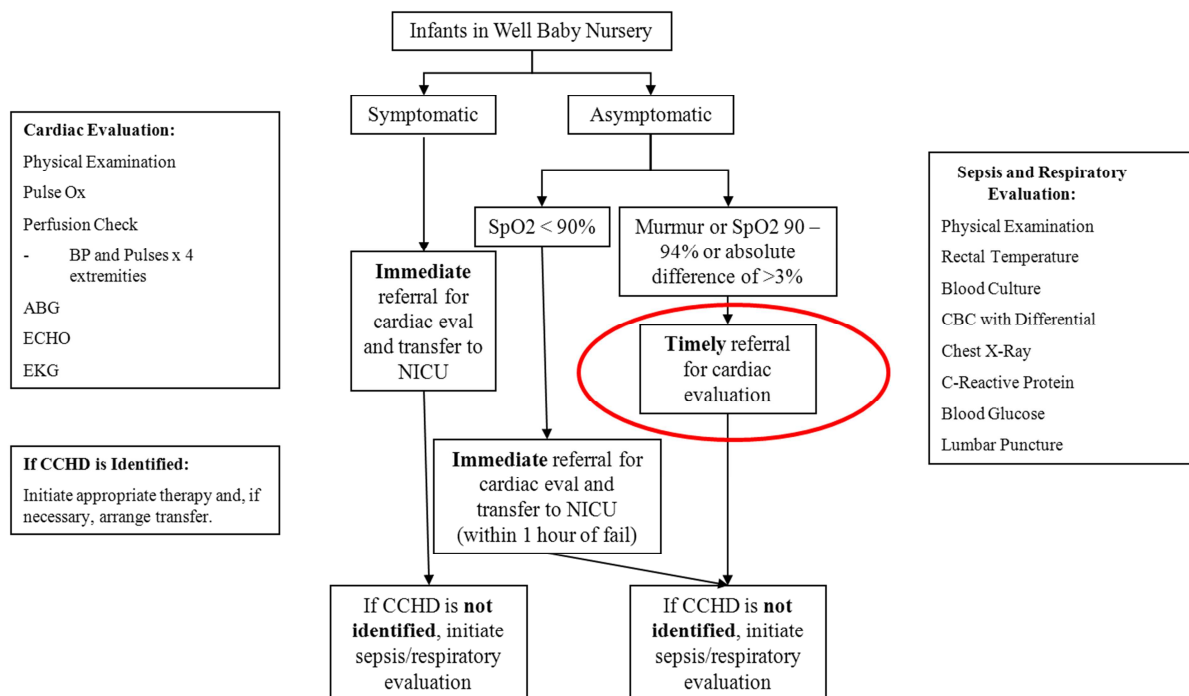
The screener should promptly NOTIFY the responsible medical practitioner of failed screen and the need for further evaluation.

Failure of this test is a potential emergency requires immediate evaluation with an echocardiogram and immediate referral to a NICU with cardiac care availability.

In the absence of a clear non-cardiac cause of hypoxemia, the medical practitioner should OBTAIN an echocardiogram and CONSULT neonatology IMMEDIATELY.

If the echocardiogram shows a normal heart structure, the primary care physician should rule out other reasons for hypoxemia, such as, infection or respiratory issues. Some conditions that could cause hypoxemia include group B streptococcal pneumonia, congenital pneumonia, early onset sepsis and pulmonary hypertension.

If the echocardiogram is abnormal, the newborn should be referred to a Pediatric Cardiologist for interpretation of test results, determination of the need for Prostaglandin, and further evaluation. To prevent an adverse event should the baby become cyanotic or decompensate rapidly, the baby should be transported to the services of a neonatologist for immediate cardiac evaluation that includes an echocardiogram. The neonatologist will make the referral to a pediatric cardiologist as appropriate.



An asymptomatic child with a murmur needs a timely referral to a cardiologist if SpO2 90-94%

Transfer Protocol

Follow the birthing facility's transfer policy for newborns that must be transported to another facility that can adequately treat a potential cardiac condition.

If the hospital that performed the pulse oximetry test does not have a neonatal intensive care unit, the infant should be transferred to a facility that can provide cardiac evaluation services.

Cardiac Centers available for consult:

The following CMS Pediatric Cardiac Facilities, through program evaluation and review, have been determined to meet established minimum standards, as set forth in 64C-4.003, F.A.C.:

| | |
|---|----------------|
| All Children's Hospital, Johns Hopkins Medicine | (727) 767-6666 |
| Arnold Palmer Hospital for Children | (407) 649-6907 |
| Jackson Memorial Hospital | (305) 585-6683 |
| Joe DiMaggio Children's Hospital | (954) 265-3437 |
| Miami Children's Hospital | (305) 669-6500 |
| The Congenital Heart Center at UF Healthcare | (352) 265-0111 |
| St. Joseph's Children's Hospital | (727) 767-6666 |
| Wolfson Children's Hospital | (904) 633-4110 |

Please visit [https://www.flrules.org/gateway/RuleNo.asp?title=PERSONNEL AND FACILITIES STANDARDS&ID=64C-4.003](https://www.flrules.org/gateway/RuleNo.asp?title=PERSONNEL%20AND%20FACILITIES%20STANDARDS&ID=64C-4.003) for the CMS Pediatric Cardiac Facility Standards

Non-CMS Pediatric Cardiac Facilities

| | |
|---|----------------|
| Florida Hospital for Children Orlando | (407) 303-3692 |
| Palm Beach Children's Hospital at St. Mary's Medical Center | (561) 844-6300 |

Reporting Diagnostic Information

Physicians and/or Cardiac Centers who care for newborns that have **failed** the newborn screening pulse oximetry (POX) test must report the final outcome information to the Florida Newborn Screening Program. See attached file with CCHD Case Report form.

Screening Tips for Pulse Oximetry Screening (POX)

The pulse oximetry screening test should be administered to all infants who are at least 24 hours of age or shortly before discharge if younger than 24 hours of age.

The infant should be awake and quiet.

It is helpful if the parent is present to soothe and comfort the infant.

The infant should not be actively crying or cold during the screening test.

The infant's skin should be clean and dry. Skin color and jaundice do not affect the pulse oximetry reading.

The red light, or emitter, should always go on the top of the hand or foot. If you are using a disposable probe, there should be a star or other symbol to indicate which side of the probe goes on the top of the foot/palm. If you are using a reusable probe there may be a bar or star to specify which side of the probe should be placed on top of the hand or foot. Two helpful little sayings that you can use to remember this are "Stars to the sky" and "Raise the bar".

The side of the probe that is the photo detector should be placed directly opposite the red light emitter. To accurately obtain the saturation reading the two sides of the probe need to be directly opposite of one another.

It is suggested that the pulse oximetry screening is paired with other standard of care newborn screening at the same time. A suggested pattern is:

- Undress and weigh baby
- Swaddle baby
- Place pulse oximetry probe on right hand
- Place foot warmer on foot
- Check transcutaneous bilirubin
- Check pulse oximetry value on right hand; if passing value, move to either foot
- Check pulse ox, if passing, you are finished
- Perform the heel stick for newborn screening specimen sample and collect serum bilirubin if needed

Use only pulse oximetry screening devices developed for newborns. Never use an adult pulse oximetry clip on a newborn.

Do not perform pulse oximetry test on limb with blood pressure cuff.

Do not perform pulse oximetry test in bright or infrared lighted area. Cover sensor with a blanket to ensure accuracy of reading.

Do not use tape or use your own hand to apply the pulse oximetry sensor to the infant's skin.

If using equipment with only one probe/sensor, test one extremity, then move probe/sensor to other extremity.

Online tool and free app for smartphones developed by Children's Healthcare of Atlanta:

<http://www.pulseoxtool.com/>

Table for interpreting results developed by Cynthia Mueller (permission to use was granted on 9/16/2013):

Virginia Department of Health Mueller CCHD Screening Table

Green = Negative Screen (PASS)

Yellow = Rescreen in 1 hour

Yellow for 3 consecutive screens = Positive Screen (FAIL)

* Red = Automatic Positive Screen (FAIL)

| Right Hand | Foot | | | | | | | | | | | <90 |
|------------|------|----|----|----|----|----|----|----|----|----|----|-----|
| 100 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 99 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 98 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 97 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 96 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 95 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 94 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 93 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 92 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 91 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| 90 | 100 | 99 | 98 | 97 | 96 | 95 | 94 | 93 | 92 | 91 | 90 | * |
| <90 | * | * | * | * | * | * | * | * | * | * | * | <90 |

Created by Cynthia Mueller BSN, RN - Anne Arundel Medical Center



Screen all babies after 24 hours, before discharge.

Check how you would interpret the following examples of POX results.

| <i>1st screen</i> | <i>2nd screen</i> | <i>3rd screen</i> | <i>Required Action</i> |
|--|--|--|---|
| Example #1 RH Sat - 96% LE Sat - 94% PASS RETEST FAIL | | | No further action required. |
| Example #2 RH Sat - 89% LE Sat - 87% PASS RETEST FAIL | | | Do not repeat screening test. Follow protocol for management of failed CCHD screening test. |
| Example #3 RH Sat - 100% LE Sat - 96% PASS RETEST FAIL | RH Sat – 99% LE Sat – 98% PASS RETEST FAIL | | No further action required. |
| Example #4 RH Sat - 100% LE Sat - 96% PASS RETEST FAIL | RH Sat - 92% LE Sat - 94% PASS RETEST FAIL | RH Sat – 99% LE Sat – 98% PASS RETEST FAIL | No further action required. |
| Example #4 RH Sat - 92% LE Sat - 96% PASS RETEST FAIL | RH Sat - 92% LE Sat - 94% PASS RETEST FAIL | RH Sat - 92% LE Sat - 92% PASS PASS FAIL | Follow protocol for management of failed CCHD screening test. |

RH = Right Hand; LE = Lower Extremity (either foot)