Pulse Oximetry Screening for Congenital Heart Disease: Toolkit



Elizabeth Goetz MD, MPH

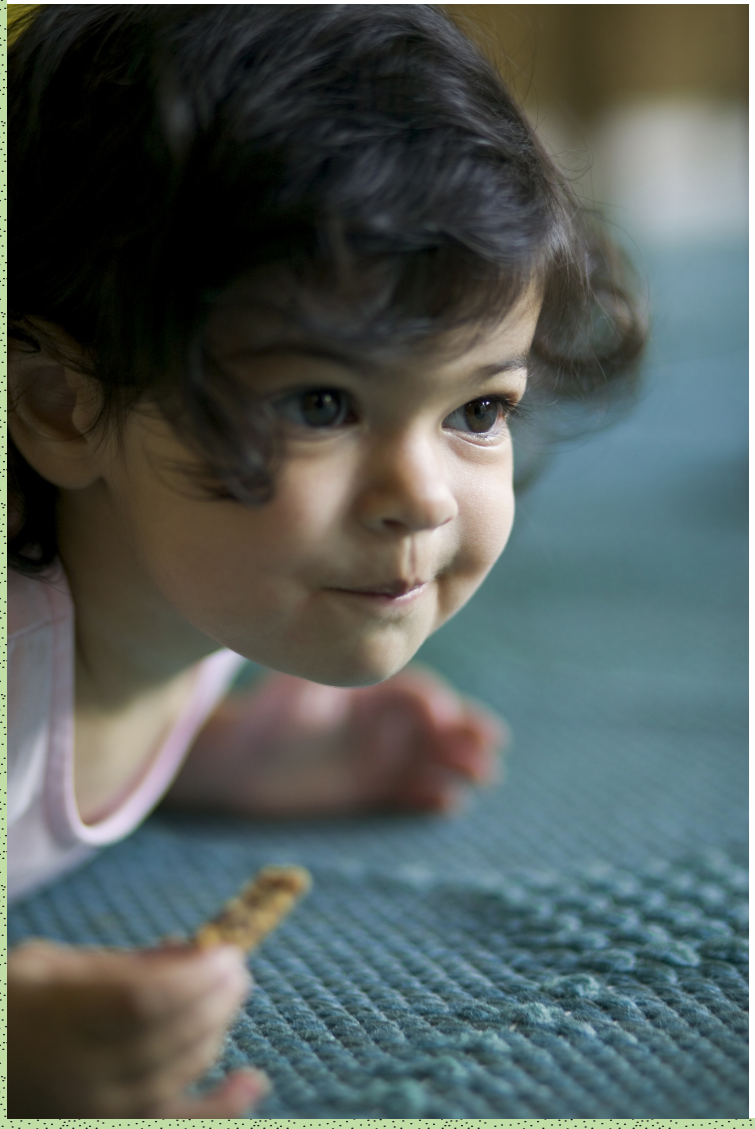
University of Wisconsin
Department of Pediatrics
Meriter Hospital Newborn Nursery

John Hokanson, MD

University of Wisconsin
Department of Pediatrics
Pediatric Cardiology

Table of Contents

1. Letter of Introduction
2. Ten Fast Facts about CCHD
3. Types of CCHD Identified by Pulse Oximetry Screening
4. Screening Recommendation
5. Sample Protocol
6. Provider Education
   - Sample Provider Letter
7. Staff Education
   - Power Point Presentation
8. Parent Education
   - Parent Brochure
9. Education Resources
10. Sample Screening Form
11. Quick Reference Card
12. References



Introduction

Every year 10/1000 babies in the United States are born with congenital heart disease (CHD). Of these babies, 25% will have critical congenital heart disease (CCHD), which can result in severe morbidity and mortality in the newborn period. While many infants with CCHD are identified by prenatal ultrasound, most cases of CCHD are discovered after birth. Unfortunately, changes in the structure and function of the newborn heart can lead to CCHD going unrecognized during the newborn hospital stay. Unrecognized CCHD may result in sudden deterioration and death.

While infants with CCHD can have a normal physical exam in the immediate newborn period with no heart murmur and no clinical cyanosis, most will have hypoxemia. Advances in pulse oximetry have improved the accuracy of this test in identifying hypoxemia in newborns and allowed it to be used as a screening test for CCHD. In asymptomatic infants, pulse oximetry can complement the clinical exam in the detection of CCHD by identifying clinically undetectable hypoxemia. Early detection of CCHD can significantly reduce morbidity and mortality in the newborn period.

In 2010 the United States Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) recommended that CCHD be added to the recommended uniform screening panel based on findings from a comprehensive evidence review. In 2011 the Secretary of Health and Human Services endorsed these recommendations, adding pulse oximetry to the recommended screening studies that all healthy newborns should have before being discharged home from the hospital.

Screening for CCHD with pulse oximetry has been shown to be cost effective, accurate, and easy to incorporate into the workflow of the normal newborn nursery. Included in this toolkit is information for nurseries to use in developing policies and procedures to implement pulse oximetry screening for CCHD.



Ten Fast Facts About Pulse Oximetry Screening for Critical Congenital Heart Disease in Neonates:

1. Congenital heart disease is the most common serious birth defect in humans and a potentially life threatening heart defect is present in approximately 1:400 births.

2. Less than half of babies with critical congenital heart disease will be diagnosed prenatally.

3. The diagnosis of critical congenital heart disease in newborns is missed prior to hospital discharge in anywhere from 1:3,500 to 1:25,000 births.

4. More than half of patients with a missed diagnosis of critical congenital heart disease will die at home or in an emergency room before their heart disease is recognized.

5. Each year between 100 and 200 babies die in the US due to unrecognized critical congenital heart disease.

6. In Wisconsin from 2002 to 2006, at least 9 neonates died at home or in an emergency room due to previously unrecognized critical congenital heart disease.

7. Those babies who survive a missed diagnosis of critical congenital heart disease have more complications and disabilities than patients who were diagnosed in a timely fashion.

8. Many babies with critical congenital heart disease will have a normal physical examination in the first days after birth. Although they will usually have abnormal oxygen saturations, our eyes can only detect extremely severe cyanosis. This clinical situation where the abnormal oxygen saturation level cannot be detected with our eyes is called the "Cyanotic Blind Spot."

9. Pulse oximetry has been shown to be a useful tool in the detection of previously unrecognized critical congenital heart disease.

10. In a large Swedish study, there were no deaths due to unrecognized critical congenital heart disease in the pulse oximetry screening population of 46,963 babies, but there were 5 deaths due to unrecognized critical congenital heart disease in the control population of 109,604 babies.

Will pulse oximetry detect all forms of serious congenital heart disease?

No. Pulse oximetry is most useful in detecting those forms of heart defects in which a baby can look entirely well a day after birth and become very ill within the next few days. The types of heart defects most likely to cause this are referred to as the "ductal dependent" and cyanotic defects.

Does pulse oximetry replace other methods of detecting critical congenital heart defects?

No. Approximately half of all babies with a critical congenital heart defect are detected by a prenatal ultrasound. If not detected prenatally, most babies with a critical congenital heart defect will be identified by an abnormality in their physical examination. Pulse oximetry serves as one additional method of catching the 1 or 2% of babies with critical congenital heart disease that aren't detected by prenatal ultrasound or newborn physical examination.

How often will a child with a normal heart fail the screening process (false positive result)?

This depends a lot on when the screening is performed and if the screening is rechecked before being considered positive. In large studies of pulse oximetry tests performed at least 24 hours after birth, the false positive rate ranges from about 1:600 to less than 1:1,000. One study showed a false positive rate of 1:10,000 (Boelke, unpublished data).

If a child fails the screening, what is the chance that they have a potentially life threatening heart condition (positive predictive value)?

This also depends on when the testing was performed. If done at least 24 hours after birth, the chance of a serious heart defect ranges from 21% to 26%. A low oxygen saturation may also be due to other problems that might not yet have become apparent such as lung disease, bacterial infection, or other disease.

Why does it matter when the screening is done?

Oxygen saturations gradually increase in normal babies over the first several hours of life. If pulse oximetry screening is done too early, some babies will fail the screening even if their hearts are normal. However, by 24 hours, the vast majority of normal babies will have an oxygen saturation greater than 95%. The average saturation for a 24 hour old baby is 97%.

What should be done if a baby fails the screening test?

The first step is to examine the baby closely for other medical problems that could result in a low oxygen saturation. If these are identified, the baby should receive the appropriate care for their particular problem. If there is no other cause for the low oxygen saturation, the baby should be evaluated for a critical congenital heart defect. That evaluation will usually involve an ultrasound test of the heart called an echocardiogram.

Are other tests helpful in excluding a critical congenital heart defect if a baby fails the screening and no other cause for the low oxygen saturation is identified?

Other cardiac testing may be useful in certain circumstances. A chest x-ray may be helpful in looking for problems with the lungs. An electrocardiogram may be helpful if a heart rhythm disorder is suspected. Laboratory tests may help to determine the presence of infection or a metabolic disease. As helpful as these tests may be, however, they probably aren't adequate to exclude the possibility of a critical congenital heart defect.

What if an echocardiogram can't be performed where the baby is born?

If a baby fails the pulse oximetry screening process and no other explanation for the low oxygen saturation is identified, an echocardiogram may be needed. In smaller communities and more rural hospitals, this may not be available. In this situation, the health care staff caring for the baby will often discuss the situation with a neonatologist or a pediatric cardiologist. The health care team caring for the baby may recommend that the baby go to another facility so that the echocardiogram can be performed. Depending on the circumstances, additional testing or observing the baby a little longer in the local hospital may be appropriate.



How pulse oximetry screening in newborns applies to different types of Congenital Heart Disease

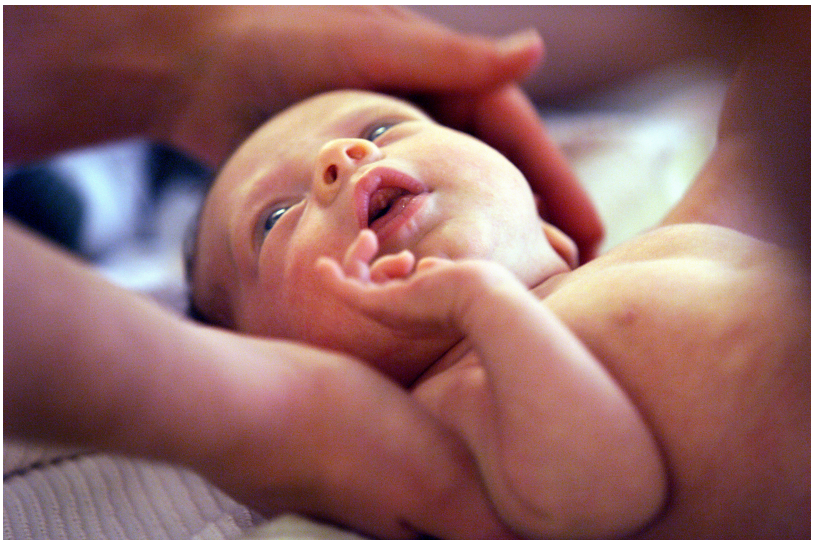
Pulse oximetry screening is used to detect low blood oxygen levels that cannot be recognized by the naked eye. In some newborns, this abnormal blood oxygen measurement may be the only clue that a critical congenital heart defect is present. Pulse oximetry screening is more likely to detect some forms of congenital heart disease than others. Fortunately, pulse oximetry is most effective in those babies with heart defects which become life threatening in the first days after birth.

Cyanotic Heart Defects:

These heart problems involve how the heart and lungs interact and may become apparent as soon as the lungs are put to use in the delivery room. These heart defects include Transposition of the Great Vessels, Tetralogy of Fallot, Tricuspid Atresia, Pulmonary Atresia, Truncus Arteriosus, Total Anomalous Pulmonary Venous Return, as well as many others. Just how cyanotic a child with these heart problems will be can vary quite significantly. The most severe variations of these problems will result in cyanosis which is immediately obvious to the naked eye. Pulse oximetry should detect nearly all of the children with less severe variations of these cyanotic heart defects.

Ductal Dependent Heart Defects:

Ductal dependent heart defects are typically those in which the right heart supports a left heart which was under developed or has a significant obstruction to blood flow. A structure called the ductus arteriosus connects the pulmonary artery and the aorta before birth and is programmed to close shortly after delivery. If there are significant problems with the left heart, the ductus arteriosus can provide and important "crutch" to let the right heart support the left heart. When the right heart helps to support the left heart, the baby's oxygen saturation will be abnormal. These babies may appear entirely well until the ductus arteriosus closes and the left heart is left on its own. The closure of the ductus usually occurs after a baby has gone home from the hospital. As these babies can become critically ill very quickly, they are one of the primary targets for pulse oximetry screening. Hypoplastic Left Heart Syndrome is the prototype of a heart defect in which a baby may appear entirely well at two days of age and become critically ill or die in the next few days. Other examples include Critical or Severe Aortic Stenosis, Coarctation of the Aorta, Interrupted Aortic Arch, and other defects in which there is an obstruction of blood flow to the body.



Left to Right Shunts

In these types of heart defects blood that should have gone to the body is sent to the lungs instead. This group includes some of the most common forms of congenital cardiac malformations. Babies with this type of heart defect will usually have entirely normal pulse oximetry measurements. Fortunately, it is very rare for these babies to become critically ill in the first few days after birth due to their heart defects. Usually, these babies will develop a heart murmur that will alert medical personnel to their heart defects before any symptoms develop. Examples of these types of heart defects include, but are not limited to Ventricular Septal Defects, Atrial Septal Defects, Atrioventricular Canal Defects, and Patent Ductus Arteriosus.

Isolated Valve Defects

Although these defects may be severe and may be associated with abnormal blood oxygen levels, there is usually a heart murmur that will alert the clinician to the presence of a heart problem. Pulse oximetry may be helpful in these babies, but most will have already been recognized on physical examination.

Single Ventricle Defects

There are many types of heart problems where one of the ventricles is too small or does not function adequately. Some of these babies will act more like cyanotic patients, with too little blood going to the lungs. Some will act more like babies with ductal dependent defects and will not have enough blood going to the body when the ductus arteriosus closes. Occasionally, these babies will have no symptoms immediately and behave more like a child with a left to right shunt defect. Pulse oximetry is very helpful in this setting as the blood oxygen level in these babies is almost never normal.

Screening Recommendations

Screening

- All babies in the well-baby nursery should be screened with pulse oximetry for CCHD.
- Babies in intermediate-care nurseries or other units in which discharge is common in the first week should be screened with pulse oximetry for CCHD.
- Screening should be based on the recommended screening algorithm.
- Pulse oximetry screening should not replace a complete history and physical exam.
- Screening should take place between 24 and 48 hours of life. If early discharge is planned, screening should occur as late as possible.
- Screening should be done in the right hand and one foot, in parallel or in sequence.
- Qualified personnel who have been educated in the use of the algorithm and trained in pulse oximetry monitoring of newborns should perform screening.
- Any abnormal screen requires complete clinical evaluation.
- Follow up of a positive screen should be initiated by the hospital or birth center prior to discharge.

Equipment

- Screening should be done with motion tolerant pulse oximeters that report functional oxygen saturation.
- Pulse oximeters can be used with either disposable or reusable probes.
- Manufacture-recommended pulse oximeter-probe combinations should be used.

Communication

- Results of the newborn CCHD screening should be communicated to the newborns primary care provider.
- Primary care providers will need to develop strategies for evaluating newborns who were not screened for CCHD.
- Healthcare providers must understand the rational for and limitations of pulse oximetry screening to detect CCHD.

Education

- Provide education materials to inform parents of the pulse oximetry screening program, including information about limitations of the screening program, the right to decline screening, and sources of more information.

Administration

- Each hospital or birthing center should develop a policy for pulse oximetry screening that includes screening procedures, documentation, and reporting of results.
- Each hospital or birthing center should establish a plan for management and evaluation of babies with positive screening results.
- Each hospital or birthing center should establish a procedure for parents who decline screening.
- Each hospital or birthing center should provide training in newborn pulse oximetry to individuals who will be involved in the screening process.

Pulse Oximetry Screening for Critical Congenital Heart Disease (CCHD) in Newborn Infants: Sample Protocol

I. PATIENT POPULATION AND/OR AREA AFFECTED

A. All infants admitted to normal newborn nursery.

II. GENERAL INFORMATION

A. Critical Congenital Heart Disease (CCHD) has an incidence of approximately 10/1000 births. Approximately 50% of infants who have CCHD are asymptomatic in the first few days of life. Clinical signs of CCHD are varied and nonspecific and can include tachypnea, bradycardia, tachycardia, hepatomegaly, decreased perfusion and decreased femoral pulses. Heart murmurs are often considered an important sign of CCHD, however, approximately 50% of infants with CCHD do not initially present with a heart murmur, and heart murmurs do not always signify structural heart disease. Cyanosis is also an important sign of CCHD. Unfortunately, cyanosis can be difficult to detect in newborns. Only severe decreases in oxygen saturation are visibly apparent in newborns.

B. In asymptomatic infants, pulse oximetry can complement the clinical exam in the detection of CCHD by identifying clinically undetectable hypoxemia. Specific lesions targeted by pulse oximetry screening include, but are not limited to, hypoplastic left heart syndrome, pulmonary atresia, tetralogy of fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriosus.

C. Presentation of CCHD can be sudden and catastrophic with the closing of the ductus arteriosus and physiologic changes in the heart after birth. Early recognition of CCHD can help to ensure timely intervention and treatment and reduce morbidity and mortality.

D. Prenatal ultrasound and fetal echocardiography have increased early detection of CCHD. However, due to test limitations, these studies cannot detect all cases of CCHD. Most infants with CCHD are not identified prenatally.

III. PROTOCOL

A. Pulse Oximetry screening should be done using motion-tolerant pulse oximeters that report functional oxygen saturation cleared by the FDA for use in newborns.
   A. Disposable or reusable probes may be used
   B. Use manufacture-recommended pulse oximeter-probe combinations

A. Perform pulse oximetry on infant between 24 and 48 hours of age.
   a. For infants discharged before 24 hours of age, perform pulse oximetry prior to discharge.
   b. Measure pulse oximetry on right upper extremity and either right or left foot. Measurements can happen at the same time, or one immediately after the other
   c. Ensure good pulse waveform.
   d. Perform test when baby is alert.

B. Interpret results according to algorithm
   a. *Screen is considered **negative** if pulse oximetry is ≥ 95% in right hand or either foot with ≤ 3% absolute difference in oxygen saturation between right hand and foot.*
      i. No further screening is needed
   b. *Screen is considered **positive** if pulse oximetry is < 90% in right hand or foot at any stage of screening.*
      i. Further evaluation needed
   c. *Screen is considered **positive** if pulse oximetry is < 95% on both extremities for three consecutive measures each separated by one hour*
      i. Further evaluation needed
   d. *Screen is considered **positive** if there is > 3% absolute difference in oxygen saturation between the right hand and foot on three consecutive measures each separated by one hour*
      i. Further evaluation needed

C. Algorithm
   a. Initial Screen: Measure pulse oximetry in right hand and either foot between 24 to 48 hours of age or shortly before discharge of < 24 hours of age
      i. Pulse oximetry < 90% in right hand or foot
         1. *Screen is **positive**. Infant has failed pulse oximetry screen.*
   a. Call MD for evaluation of infant
      ii. Pulse oximetry ≥ 95% in right hand or foot with ≤ 3% absolute difference in oxygen saturation between right hand and foot
         1. *Screen is **negative** and no further pulse oximetry follow-up needed.*
      iii. Pulse oximetry 90-<95% in right hand and foot or > 3% absolute difference in oxygen saturation between right hand and foot
         1. Continue Screening: repeat screen in one hour
   b. First Repeat Screen: Measure pulse oximetry in right hand and either foot one hour after initial screen if pulse oximetry 90-<95% in right hand and foot or > 3% absolute difference in oxygen saturation between right hand and foot on initial screen.
      i. Pulse oximetry < 90% in right hand or foot
         1. *Screen is **positive**. Infant has failed pulse oximetry screen.*

a. Call MD for evaluation of infant

ii. Pulse oximetry ≥ 95% in right hand or foot with ≤ 3% absolute difference in oxygen saturation between right hand and foot

1. *Screen is **negative** and no further pulse oximetry follow-up needed.*

iii. Pulse oximetry 90-<95% in right hand and foot or > 3% absolute difference in oxygen saturation between right hand and foot

1. Continue Screening: repeat screen in one hour

c. Second Repeat Screen: Measure pulse oximetry in right hand and either foot one hour after initial screen if pulse oximetry 90-<95% in right hand and foot or > 3% absolute difference in oxygen saturation between right hand and foot on first repeat screen.

i. Pulse oximetry < 90% in right hand or foot

1. *Screen is **positive**. Infant has failed pulse oximetry screen.*

a. Call MD for evaluation of infant

ii. Pulse oximetry ≥ 95% in right hand or foot with ≤ 3% absolute difference in oxygen saturation between right hand and foot

1. *Screen is **negative** and no further pulse oximetry follow-up needed.*

iii. Pulse oximetry 90-<95% in right hand and foot or > 3% absolute difference in oxygen saturation between right hand and foot

1. *Screen is **positive**. Infant has failed pulse oximetry screen.*

a. Call MD for evaluation of infant

D. Failed Pulse Oximetry Screen

a. Alert newborn care provider of positive pulse oximetry screen
b. Immediate clinical evaluation by licensed independent practitioner
c. If a non-cardiac explanation for hypoxemia is not identified, CCHD must be excluded

E. Diagnosis: Echocardiogram *(Neonatal Echocardiogram Available)*

a. Order echocardiogram
   i. Indication: Failed oximetry screening
b. Consult Pediatric Cardiology Group who will read Echo
c. Vital signs with pulse oximetry and clinical assessment every 4 hours until echocardiogram is complete. Report to newborn care provider
d. Do not discharge infant until echocardiogram results are called to attending physician and discharge order is given.

F. Diagnosis: Echocardiogram *(Neonatal Echocardiogram Not Available)*

a. Consult Regional Pediatric Cardiology Group
b. Determine course of action based on consultation and resources available.
c. Vital signs with pulse oximetry and clinical assessment every 4 hours until echocardiogram is complete. Report to newborn care provider

Congenital Heart Disease (CHD) Assessment

09/11

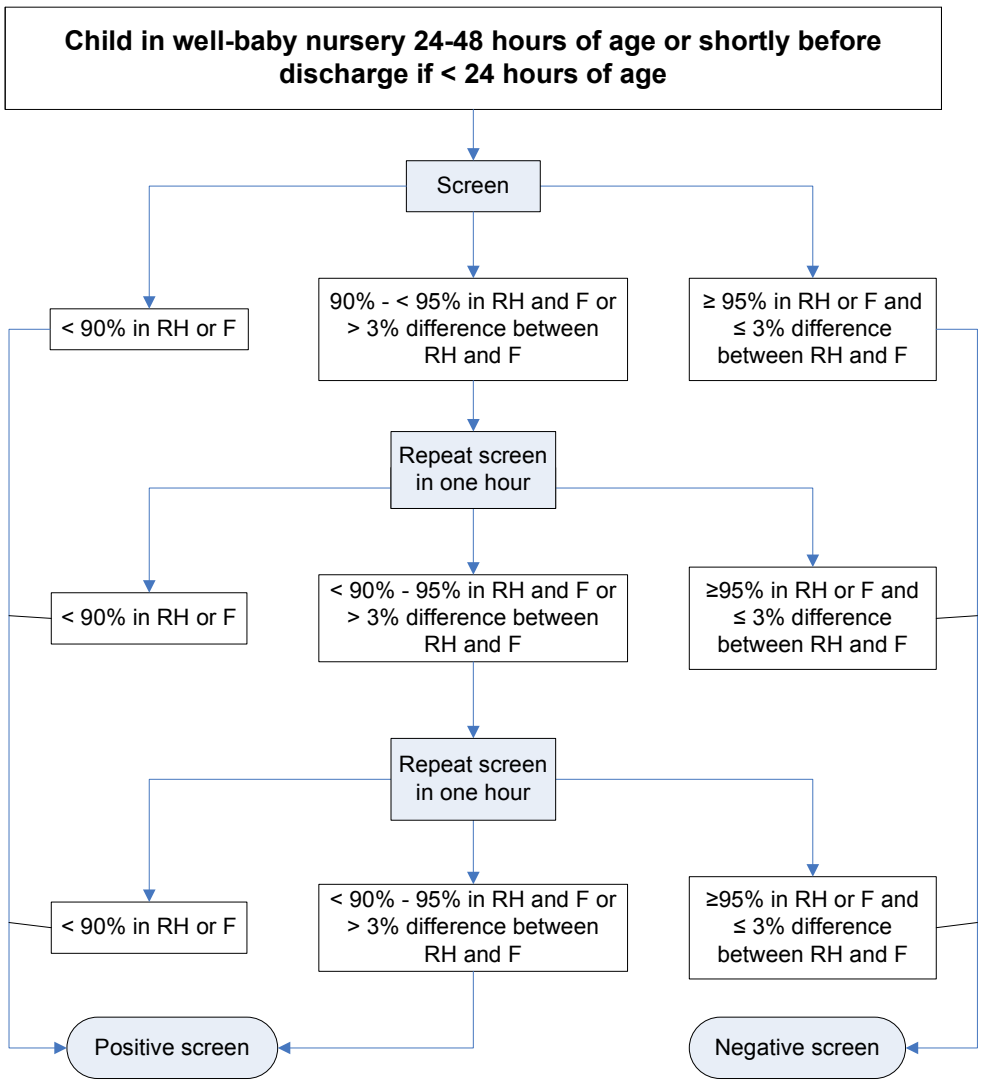


Figure. The proposed pulse oximetry monitoring protocol based on results from the right hand (RH) and either foot (F).

Provider Education: Sample Provider Letter

Dear Newborn Provider,

Pulse oximetry screening for critical congenital heart disease (CCHD) has been added to the newborn screening protocol in our newborn nursery. Although rare, CCHD may go undetected in the immediate newborn period and lead to significant morbidity and mortality in the weeks following birth. In September, 2011 the United States Secretary of Health and Human Services recommended adding pulse oximetry screening for CCHD to the Universal newborn screening panel.

Congenital Heart Disease (CHD) is a common birth defect affecting 10/1000 babies in the United States each year. Of these babies, 25% will have critical congenital heart disease (CCHD). While many infants with CCHD are identified by prenatal ultrasound, most cases of CCHD are discovered after birth. Unfortunately, changes in the structure and function of the newborn heart can lead to CCHD going unrecognized during the newborn hospital stay. While infants with CCHD can have a normal physical exam in the immediate newborn period with no heart murmur and no clinical cyanosis, most will have hypoxemia. Advances in pulse oximetry have improved the accuracy of this test in identifying hypoxemia in newborns and allowed it to be used as a screening test for CCHD. In asymptomatic infants, pulse oximetry can complement the clinical exam in the detection of CCHD by identifying clinically undetectable hypoxemia.

All babies cared for in the newborn nursery will be screened for CCHD between 24 and 48 hours of life using pulse oximetry to detect hypoxemia. The screening test can be thought of as a "two sites, three strikes" screen. Pulse oximetry is done at two sites, the right hand and either foot. An infant must fail three consecutive measurements spaced one hour apart, or have "three strikes," to have failed the screen (with the exception of an oxygen saturation <90% in either extremity which would be an immediate positive, or "failed" screen.)

- An infant will have a negative, or "passing" screen if the oxygen saturation is ≥95% in either the hand or the foot with ≤ 3% difference between extremities.
- An infant will have a positive or "failed" screen if the oxygen saturation is <95% in both the hand and the foot, or if there is a >3% difference between the extremities on three separate measures each separated by one hour.
- An infant will have a positive or "failed" screen in if the oxygen saturation is <90% in either the hand or the foot at any time.

Infants who have a positive screen will need further evaluation in the nursery prior to discharge. It is recommended that infants with a positive screen have an echocardiogram to rule out structural heart disease.

Thank you for your support in screening newborns for CCHD. Please contact us with any questions regarding the screening process.

Sincerely,

Pulse Oximetry Screening for Critical Congenital Heart Disease (CCHD)

Staff Education

Overview

- Scope of the Problem
- Review of congenital heart disease
- Challenges in the nursery
- Pulse oximetry screening
  - The who, what, how, when and where
- Follow up care

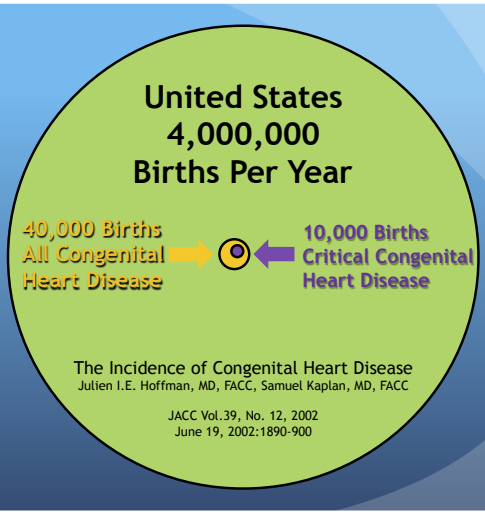
What we are trying to prevent

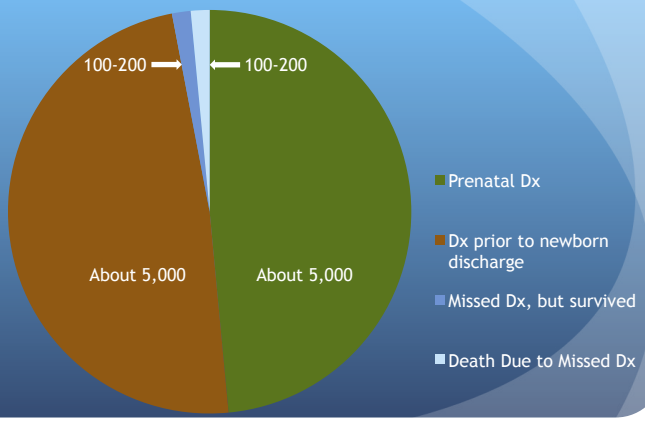
- Some infants with critical congenital heart disease will have no symptoms and have an entirely normal physical examination at the time they are sent home from the hospital after birth
- These children may become critically ill or die in the next few days of their congenital heart disease is not recognized

Scope of the Problem

Reference	Years	Incidence of CCHD	Missed or Delayed Diagnosis	Deaths due to Missed Diagnosis	Deaths per live birth	Location
Aamir	1999-2004	1:971	1:14,261	Not reported		New Jersey
Ng	2002-2006	Not reported	1:24,684	1:38,397	9/345,672	Wisconsin
Wren	1985-2004	1:1032	1:3486	1:23,007	30/690215	Northern Health Region, UK
Mellander	1993-2001	1:1135	1:6899	Not Reported		Sweden
De-Wahl	2004-2007	1:853	1:3878	1:21,721	5/108,604	Sweden

Pulse Oximetry Screening for Unrecognized Congenital Heart Disease in Neonates. Neonatology Today 5(12):2010





Scope of the Problem in Wisconsin

- Missed Congenital Heart Disease in Neonates. Benton Ng, MD and John Hokanson, MD. Congenit Heart Dis. 2010
  - Wisconsin 2002-2006
  - Babies discharged as normal newborns who were hospitalized or died due to unrecognized critical congenital heart disease in the first two weeks after birth
  - Death or hospitalization
    - 1 in 24,684 births **3 per year in Wisconsin**
  - Death
    - 1 in 38,397 births **2 per year in Wisconsin**

Deaths due to Unrecognized CCHD

Cause	Estimated Number of Deaths per Year in the United States
Unrecognized critical congenital heart disease in newborns	100-200
Sudden deaths in young athletes	<100
Sudden death associated with stimulant medication use	<10

Pulse Oximetry Screening for Unrecognized Congenital Heart Disease in Neonates. Neonatology Today 5(12):2010

Congenital Heart Disease

- Structural or functional defects in the heart present at the time of birth
- Range from benign problems that resolve with growth and development of the heart to life threatening in the delivery room
- May or may not be associated with cyanosis
- Most concerning are:
  - Left sided obstruction
  - Cyanotic heart defects

Left Sided Obstructive Lesions

- Critical Coarctation of the Aorta
- Critical Aortic Stenosis
- Hypoplastic Left Heart Syndrome and variants
- Total Anomalous Pulmonary Venous Return (TAPVR)
- Interrupted Aortic Arch

Cyanotic Heart Defects

- Truncus Arteriosus
- Transposition of the Great Arteries
- Tricuspid Atresia
- Tetrology of Fallot
- TAPVR
- Pulmonary Atresia
- Ebsteins Anomaly

Cyanotic Heart Defects

- Usually become apparent very shortly after birth
- Never resolve spontaneously
- Not associated with normal life expectancy without surgery

Presentation of CCHD

- Survival of fetus depends on patent Ductus Arteriosus
  - Infants with CHD may depend on the ductus arteriosus being open for systemic and pulmonary circulation
- After delivery the ductus arteriosus closes in normal newborns over the course of minutes to several days
- Infants with structural heart defects may rely on ductus arteriosus being open to maintain pulmonary or systemic circulation

What does "Ductal Dependent" Mean?

- Heart depends on patent ductus for adequate function
  - Left sided obstructive lesions
    - Some or all of systemic cardiac output is supplied from the right heart via the ductus arteriosus
    - Closure of the ductus arteriosus results in obstruction to systemic output
  - Cyanotic lesions
    - May become MORE cyanotic at ductal closure if some or all of pulmonary blood flow has been supplied from the left heart via the ductus

Diagnosis of CCHD

- Prenatal ultrasound
- Postnatal physical exam
- Screening pulse oximetry

Prenatal Diagnosis

- Prenatal Ultrasound
  - Advantages:
    - Assess cardiac and non-cardiac anatomy
    - Widely available
  - Disadvantages
    - Highly variable in ability to detect CHD
    - A four chamber view alone will miss most cases of CHD

Prenatal Diagnosis

- Sensitivity of Prenatal Ultrasound
  - 2007 Acherman (Las Vegas)
    - Prenatal detection rate 36%
  - 2006 Tegnander et al (Norway)
    - Prenatal detection rate 43%
  - 2004 Acharaya et al (Norway)
    - Prenatal detection rate 24%
  - 1999 Bull (UK)
    - Prenatal detection rate ranged 0-80%
      - Center specific rates of detection

Prenatal Diagnosis

- Fetal Echocardiography
  - Advantages
    - Extremely sensitive and specific
  - Disadvantages
    - Not widely available
    - Time consuming

Neonatal Physical Exam

- Advantages
  - Widely available
  - Very useful for detecting many types of congenital heart disease
  - Very useful for detecting non-cardiac malformations
- Disadvantages
  - Physician skill level dependent
  - Only severe cyanosis can be detected visually
  - Not very useful for some of the most critical heart defects

Neonatal Physical Exam

- Findings?
  - Heart murmur
    - 50% of infants with CCHD will not have a murmur
  - Feeding difficulties
    - Many newborns have difficulty with feeding in first days of life
  - Cyanosis
    - Most newborns with CCHD have hypoxemia
    - Cyanosis is not readily detectable in newborn period

Clinical recognition of cyanosis

- Transition from blue to pink in the delivery room, as assessed by NICU staff who were blinded to the pulse oximeter reading
- **Average saturation considered "pink" 69%**

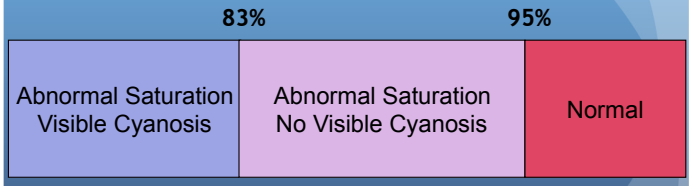
O'Donnell et al. Clinical assessment of infant color at delivery. Arch Dis Child Fetal Neonatal Ed. 2007

The Cyanotic "Blind Spot"

- Cyanosis visible with 3 g/dL of deoxygenated hemoglobin
  - Hemoglobin of 17.5 g/dL (50th percentile)
    - Visible cyanosis at <83%
    - No visible cyanosis at abnormal oxygen saturations 83-95%
  - Hemoglobin of 15 g/dL
    - Visible cyanosis at <80%
    - No visible cyanosis at abnormal oxygen saturations 80-95%
  - Hemoglobin of 12 g/dL
    - Visible cyanosis at <75%
    - No visible cyanosis at abnormal oxygen saturations 75-95%

The Cyanotic Blind Spot

Example: Hemoglobin of 17.5 g/dL (50th percentile)



Challenges in the nursery

- Prenatal diagnosis is not adequate to detect all cases of CHD
- Due to changing physiology, CCHD may not be detectable on physical exam in the immediate newborn period
- Average hospital stays for healthy newborns are 24-48 hours

Pulse Oximetry

- Intended to be IN ADDITION TO, not INSTEAD OF newborn physical exam
- Identify infants with CCHD who appear healthy
  - Detect subclinical hypoxemia
  - Catch babies in the "blind spot"
- The use of routine pulse oximetry may provide a valuable safety net to catch those babies with critical congenital heart disease who are not diagnosed by prenatal or postnatal assessments prior to discharge from the hospital

Pulse Oximetry Screening

- Advantages
  - Widely Available
  - Inexpensive
  - Outstanding specificity
  - Outstanding sensitivity for the lesions you care about the most
- Disadvantages
  - Low sensitivity for left to right shunting lesions
  - Any false positive will delay discharge inappropriately

Pulse Oximetry Screening

- Timing
  - Saturation increases through the first day
  - By 24 hours the mean oxygen saturation of a term baby is 97.2%
  - **Screening is recommended between 24-48 hours of life**
    - Measurement of pulse oximetry before 24 hours leads to more false positives
- Location
  - Post-ductal (foot) measurement will detect most critical lesions
  - **Preductal (right arm) and postductal (foot) has greatest sensitivity and is recommended**

Pulse Oximetry

- Right hand and either foot
  - At the same time or sequentially
- Try to measure when baby is in quiet awake state
- Wait long enough to establish a good wave form
  - Recommend waiting 1 full minute with good wave form before recording measurement
- Newer generation oximeters with motion correction software will be easiest to use, give the best results, and require the least nursing time
- Reusable probes will decrease the cost of screening

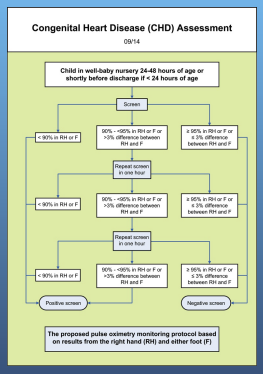
Screening Pulse Oximetry

- Positive Screen
  - <90% in either right hand or foot at any time
  - 3 consecutive measurements spaced 1 hour apart with
    - 90-94% saturation in right hand or foot OR
    - >3% difference in oxygen saturation between right hand and foot
- Negative Screen
  - ≥ 95% in right hand or foot AND
  - ≤ 3% difference between right hand and foot

Two Sites Three Strikes

- Two Sites
  - Right arm
  - Either foot
- Three Strikes
  - Baby has three chances to pass screen
    - Three measurements spaced one hour apart
    - Baby has to fail all 3 in order to have failed the screen
      - UNLESS pulse oximetry <90% in either hand or foot at any time – this would be an immediate failed screen

Screening Algorithm



Negative Screen

- No further screening necessary
- No screening test is perfect! Does not exclude the possibility of critical congenital heart disease
- Normal newborn care

Positive Screen

- Comprehensive evaluation for causes of hypoxemia
- If no other findings to explain hypoxemia
  - Critical congenital heart disease needs to be excluded
  - Know your available resources
    - Diagnostic echocardiogram in hospital
    - Transfer to institution with diagnostic echocardiography
    - Consider extended period of observation
  - **Consult with pediatric cardiology early to determine best options for baby and family**
  - There will be babies who fail the screening tests even though the do not have significant heart disease

Screening Safety Net

- Hospital / Birthing Centers / Home births
  - Have policy for routine pulse oximetry screening
  - Have mechanism for communicating screening results to primary care provider
  - Follow up of positive screens
- Primary Care Providers
  - Strategy for screening newborns who missed screening
  - Assure that all newborns were screened
  - Long-term follow up and care coordination

Facts about Critical Congenital Heart Disease (CCHD)

- CCHD occurs when the heart does not develop normally before birth
- Critical Congenital Heart Disease (CCHD) is a leading cause of newborn and infant death
- Some babies with CCHD are discovered by prenatal ultrasound, but many babies with CCHD are diagnosed after they are born
- CCHD affects 2-3 of every 10,000 births
- Not all kinds of congenital heart disease are life threatening

Local Resources





Newborn Screening for Heart Defects



Critical Congenital Heart Disease (CCHD)

Congratulations on the birth of your new baby! While you are in the hospital we want to do everything we can to make sure we are sending you home with a healthy baby. One of the things that will be done before you are sent home is a measurement of the level of oxygen saturation in your baby's blood. Low levels of oxygen saturation can mean that your baby has a problem with his or her heart called Critical Congenital Heart Disease (CCHD). Although it is very rare, some babies can have serious heart problems but look normal in the first few days after birth.

Screening for CCHD

- Pulse oximetry screening is meant to identify babies with severe heart defects known as critical congenital heart disease (CCHD). CCHD may lead to death in the newborn period.
- Pulse Oximetry uses a sensor with a light to measure blood oxygen levels to look for low levels of oxygen in the blood
- The sensor will be placed on your baby's right hand and one foot
- The test is fast and will not cause your baby any discomfort or pain
- Pulse oximetry screening will be done between 24 and 48 hours after your baby is born
- Pulse oximetry complements prenatal ultrasound a complete newborn physical exam

What if the Pulse Oximetry Screening Test is Positive?

- Pulse oximetry screening is considered positive if the oxygen saturation level is less than 90% or less that 95% three times in a row.
- Pulse oximetry screening is also considered positive if there is more than 3% difference between the right hand and the foot.
- If the test is positive, your baby will have further testing to look at the structure and function of your baby's heart.
- Some babies who fail pulse oximetry screening are found to have normal hearts.
- An echocardiogram is an ultrasound test that uses a probe to look at your baby's heart through the skin. This test is painless and similar to the prenatal ultrasound you may have had during pregnancy.

Will screening find all types of congenital heart disease?

Pulse oximetry screening is not a perfect test. This test may miss CCHD that is not associated with low oxygen levels in the blood.

What if an abnormality in my baby's heart is found?

If your baby is found to have CCHD you will meet with a pediatric cardiologist to discuss treatment options.

Can every hospital do echocardiograms?

Not every hospital has the resources to do echocardiograms on babies. If your baby has a positive screen, you may need to go to a larger hospital where your baby can be properly evaluated.

Pulse Oximetry Screening

Pulse Oximetry is a painless and non-invasive way to measure oxygenation of the blood. Blood that is carrying a lot of oxygen has a bright red color, while blood that has low levels of oxygen appears more maroon or blue. Pulse Oximetry uses the absorption of red light by a probe to determine how "red" the blood is, which correlates to the percent of hemoglobin that is bound to oxygen. Signal Extraction Technology (SET) allows the pulse oximeter to give accurate readings when the patient is moving around or has low perfusion

Education Resources

University of Wisconsin Pulse Oximetry Screening Program
http://www.pediatrics.wisc.edu/research/research-groups/hokanson/

American Heart Association
www.heart.org

Children's Heart Foundation
http://www.childrensheartfoundation.org/

Congenital Heart Information Network
www.tchin.org

Mended Little Hearts
www.mendedlittlehearts.org

Children's National Medical Center Congenital Heart Disease Screening Program
http://www.childrensnational.org/PulseOx/

Wisconsin Association for Perinatal Care (WAPC)
www.perinatalweb.org

American Academy of Pediatrics (AAP)
www.aap.org

Congenital Heart Public Health Consortium
www.chphc.org



Sample Screening Form
Pulse Oximetry Screen for Critical Congenital Heart Disease

Patient Identification

Name	MR	Date

Screening

Pass

- If pulse ox is > 95% in hand or foot with ≤ 3% difference between hand and foot mark pass and screening is complete

Fail

- If pulse ox is <90% at any time mark fail and contact newborn health care provider immediately
- If pulse ox is <95% in both the hand and the foot or there is a >3% difference between hand and foot mark fail.
- If infant fails three times with each measurement separated by one hour, contact health care provider immediately.

First Screen

Age at First Screen: Time:

Pulse Ox Right Hand	
Pulse Ox Foot	
Percent difference between right hand and foot	
❑ Pass	If pass screening is complete. No need to do second and third screen.
❑ Fail	If fail re-screen in one hour

Second Screen

Age at First Screen: Time:

Pulse Ox Right Hand	
Pulse Ox Foot	
Percent difference between right hand and foot	
❑ Pass	If pass screening is complete. No need to do second and third screen.
❑ Fail	If fail re-screen in one hour

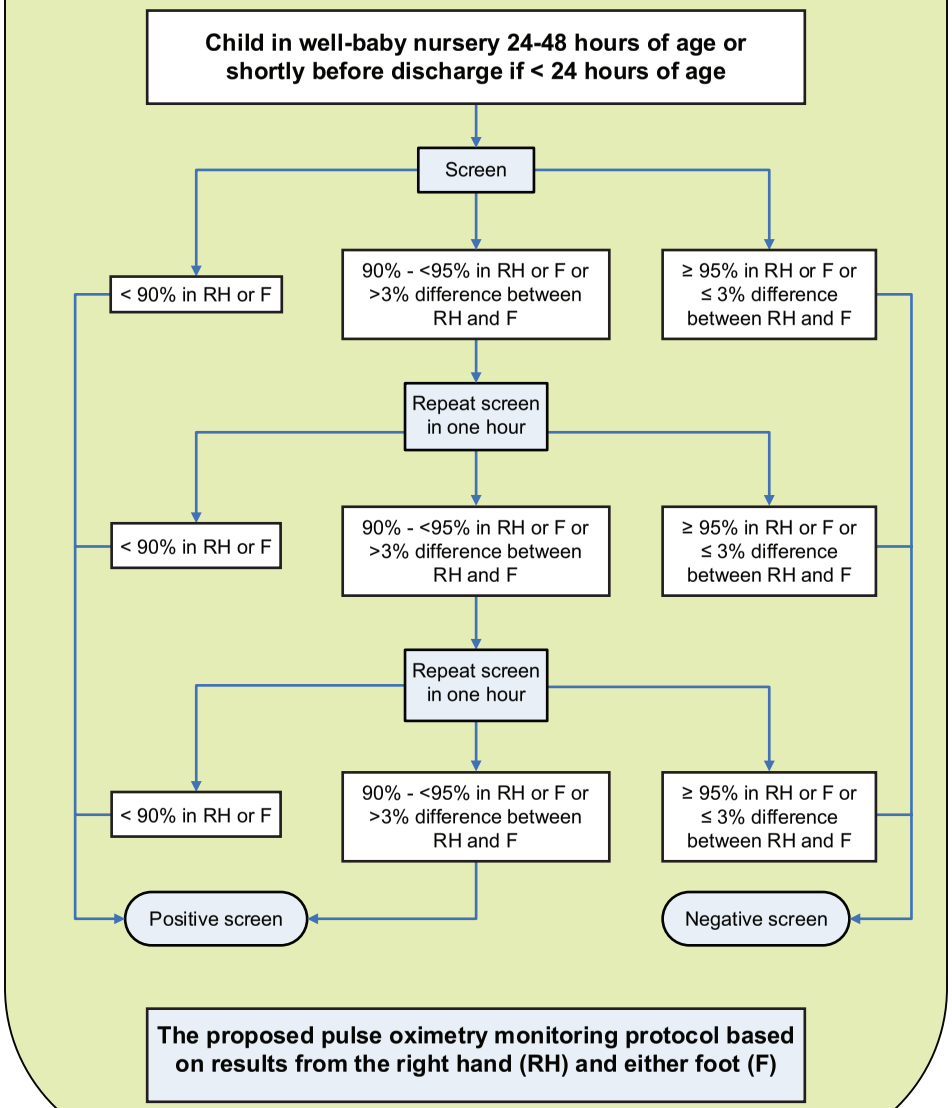
Third Screen

Age at First Screen: Time:

Pulse Ox Right Hand	
Pulse Ox Foot	
Percent difference between right hand and foot	
❑ Pass	If pass screening is complete. No need to do second and third screen.
❑ Fail	If fail re-screen in one hour

Congenital Heart Disease (CHD) Assessment

09/14



Positive Screen

- Complete clinical evaluation by baby's health care provider
- If no other findings to explain low oxygen saturation, an echocardiogram interpreted by a pediatric cardiologist is needed to rule out critical congenital heart disease
- Contact regional Pediatric Cardiology Center to arrange consult and echocardiogram

Local Cardiology Referral Contact Information

A Limited Annotated Bibliography of Materials Relating to the Use of Pulse Oximetry for the Detection of Critical Congenital Heart Disease (CCHD) in Asymptomatic Newborns

November 2011

The Incidence and Natural History of Critical Congenital Heart Disease

Only US articles are reported here, as the incidence of missed CCHD tends to be lower in US reports than in those from Europe.

Chang, et al: A cohort study of missed and delay diagnosis of critical congenital heart disease in California from 1989-2004. They report up to 30 infant deaths per year in California due to missed or late diagnosis of critical congenital heart disease. This is frequently used to estimate the yearly incidence of missed critical congenital heart disease in the US.[1]

Ng, et al: This survey of Wisconsin from 2002 to 2006 outlined an incidence of death due to unrecognized CCHD of approximately 1:40,000. There were also a number of near misses, leading to a rate of missed diagnosis of 1:25,000.[2]

Aamir, et al: This study of New Jersey medical records is also frequently quoted in the estimation of missed CCHD. The incidence of missed or delayed diagnosis of CCHD was more like 1:15,000.[3]

Kuehl, et al: This report from the Baltimore-Washington Infant Study (1981-1989) looked at the failure to diagnose congenital heart disease before death. 10% of the deaths due to congenital heart disease occurred before diagnosis, although it isn't clear that the congenital heart disease was always the cause. The two most common heart defects associated with death due to unrecognized CCHD after 48 hours of age were coarctation of the aorta and hypoplastic left heart syndrome.[4]

Physical Examination of the Newborn/Visible Cyanosis

Levesque, et al: This study reviews the normal values for pulse oximetry in newborns.[5]

O'Donnel, et al: This study of babies in transition from blue to pink in the delivery room demonstrates just how low a baby's saturation has to be in order for there to be visible cyanosis.[6] The average transition from "blue" to "pink" occurred at an oxygen saturation of 69%, suggesting that visual assessment detects only the most severe cyanosis.

Summaries of Pulse Oximetry Studies

Hokanson: This is a summary of the state of pulse oximetry as a screening tool as of December 2010. It is fairly brief and covers oximetry as it stood prior to Ewer's paper and the SACHDNC recommendations.[7] It outlines the concept of the "Cyanotic Blind Spot."

Clinical Trials of Pulse Oximetry in the Detection of Critical Congenital Heart Disease

The 15 most influential clinical studies are presented here in order of publication. Those studies which tend to support the use of pulse oximetry for CCHD tend to be larger, from Europe, and use later screening ages (typically >24 hours). Those studies which do not support the use of pulse oximetry for CCHD tend to be smaller and from the US.

2002

Richmond, et al: A study of 5,600 babies screened for CCHD both at 2 hours, and if failing the initial screening, at discharge. This was the first study to of this concept. This was a post-ductal only protocol.[8] This study was from the UK.

Hoke, et al.: This Florida study of just under 3,000 babies looked at both pre-and post-ductal saturation measurements. It also assessed the effect of early (3-6 hour) vs. later screening.[9]

2003

Koppel, et al: This was a study of 11,000 patients from New York and reported three patients with CCHD detected by post-ductal screening. They reported only one false positive.[10]

Reich, et al: Just over 2,000 Florida babies were screened with post-ductal measurements. Three echocardiograms were ordered due to failed screening, none were deemed to be normal.[11]

2004

Bakr, et al: More than 5,000 Egyptian babies were screened with post-ductal measurement. Five babies had echocardiograms because of failed screening. All but one of these had significant congenital heart disease. [12]

2005

Rosati,et al: This study of more than 5,000 Italian newborns showed one false positive, one false negative (coarctation), and two true positives (TAPVR, coarctation).[13]

2006

Arlettaz, et al: 3,000 Swiss newborns were screened with early (6-12 hour) post-ductal screening. No infant with CCHD was missed by the screening. A total of 24 babies had echocardiography because of low oxygen saturations, only one of which was felt to be entirely normal.[14]

2008

Reich, et al: The same group which published their Florida results in 2003 published a larger study of nearly 8,000 newborns and focused primarily on the reliability of the screening based on the training of the person performing the screening. Of the 12 patients with CCHD, none were detected by pulse oximetry. The oximeter used was a Nellcor Oximax N-595, which was available in 2003 and is no longer in production. This was the first major study that suggested flaws in pulse oximetry screening.[15]

Sendelbach, et al: At the time, this was the largest study of pulse oximetry for CCHD, including over 15,000 newborns from a single hospital in Dallas. Post-ductal screening was performed initially at 4 hours of age and repeated before discharge if abnormal. The false positive rate was very low at 1:15,000, but no cases of CCHD were detected by oximetry alone. This also raised questions about the need for pulse oximetry as an additional screening for CCHD.[16]

Meberg, et al (Journal of Pediatrics): This report of 50,000 Norwegian babies evaluated the use of pulse oximetry alone as well as pulse oximetry in addition to clinical examination. Post-ductal saturations were measured at less than 6 hours of age with a Massimo RAD-5 Oximeter was used in this study and the false positive rate was 147/50,008.[17]

2009

De-Wahl Granelli, et al: This large Swedish study of nearly 40,000 babies screened with both pre- and post-ductal saturations at a mean of 38 hours after birth has become the model for the recent recommendations from the US Secretary of Health and Human Services recent recommendations. This study compared the screening group to a larger unscreened population and demonstrated a marked improvement in the detection of CCHD (72% vs 92%) and a marked decrease in the rate of death due to unrecognized CCHD (5% vs 0%).[18]

Meberg, et al (Acta Paediatrics): The same Norwegian group published a related article comparing the 50,000 screened newborns to the rest of the Norwegian population. Excluding the patients with a prenatal diagnosis of CCHD, the detection rate of CCHD was 88% with pulse oximetry and 73% without pulse oximetry. [19]

2010

Reide, et al: In this German study, over 40,000 babies were screened with post-ductal oximetry. The false positive rate was 1:1,000 if all non CCHD was included, but only 1:3,400 if sepsis and pulmonary disease were excluded. The false negative rate was 1:10,000, suggesting that very few children with CCHD slipped through the cracks. In this study, nearly as many babies were detected to have CCHD by oximetry alone as were detected by physical examination and symptoms.[20]

Walsh, et al: A study of 14,500 babies in multiple settings in Tennessee did not support the use of pulse oximetry as the false positive rate was high (1:130). However, there were several flaws with this study. No training was provided to the personnel performing the screening and the screening was a single pass-fail test. No repeat testing of abnormal measurements was performed and of the 113 children failing the screening, only 3 had echocardiography performed.[21]

2011

Ewer, et al: This was the last of the large studies to be considered before the Secretary of Health and Human Services made her recommendation to include screening for CCHD to the core panel of neonatal screening tests. This was a study of 20,000 British newborns screened with both pre- and post-ductal oximetry. Babies were screened at a mean age of 12 hours after birth, although some were screened at less than three hours. [22]

Governmental and Organizational Publications and Statements

AHA/AAP Scientific Statement on the Role of Pulse Oximetry in Examining Newborns for Congenital Heart Disease: This consensus statement primarily stated that there just wasn't enough evidence to go ahead with routine screening of US newborns. This document was being prepared after the release of the Sendelbach and Reich papers and around the same time the Meberg, DeWahl-Granelli, and Reide articles were being published. Had the document been prepared six months later, the conclusions might have been very different.

2011

August 22, 2011: The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) formally recommends that pulse oximetry screening be added to the core panel of newborn screening tests.

http://pediatrics.aappublications.org/site/misc/2011-1317.preprint.pdf

September 21, 2011: The US Secretary of Health and Human Services officially recommends that pulse oximetry screening be adopted.

http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendations/correspondence/cyanoticheartsecre09212011.pdf

1. Chang RK, Gurvitz M, Rodriguez S. Missed diagnosis of critical congenital heart disease. *Arch Pediatr Adolesc Med* 2008;162(10):969-74.
2. Ng B, Hokanson J. Missed congenital heart disease in neonates. *Congenit Heart Dis* 2010;5(3):292-6.
3. Aamir T, Kruse L, Ezeakudo O. Delayed diagnosis of critical congenital cardiovascular malformations (CCVM) and pulse oximetry screening of newborns. *Acta Paediatr* 2007;96(8):1146-9.
4. Kuehl KS, Loffredo CA, Ferencz C. Failure to diagnose congenital heart disease in infancy. *Pediatrics* 1999;103(4 Pt 1):743-7.
5. Levesque BM, Pollack P, Griffin BE, Nielsen HC. Pulse oximetry: what's normal in the newborn nursery? *Pediatr Pulmonol* 2000;30(5):406-12.
6. O'Donnell CP, Kamlin CO, Davis PG, Carlin JB, Morley CJ. Clinical assessment of infant colour at delivery. *Arch Dis Child Fetal Neonatal Ed* 2007;92(6):F465-7.
7. Hokanson J. Pulse Oximetry Screening for Unrecognized Congenital Heart Disease in Neonates. *Neonatology Today* 2010;5(12):1-6.
8. Richmond S, Reay G, Abu Harb M. Routine pulse oximetry in the asymptomatic newborn. *Arch Dis Child Fetal Neonatal Ed* 2002;87(2):F83-8.

9. Hoke TR, Donohue PK, Bawa PK, Mitchell RD, Pathak A, Rowe PC, et al. Oxygen saturation as a screening test for critical congenital heart disease: a preliminary study. *Pediatr Cardiol* 2002;23(4):403-9.

10. Koppel RI, Druschel CM, Carter T, Goldberg BE, Mehta PN, Talwar R, et al. Effectiveness of pulse oximetry screening for congenital heart disease in asymptomatic newborns. *Pediatrics* 2003;111(3):451-5.

11. Reich JD, Miller S, Brogdon B, Casatelli J, Gompf TC, Huhta JC, et al. The use of pulse oximetry to detect congenital heart disease. *J Pediatr* 2003;142(3):268-72.

12. Bakr AF, Habib HS. Combining pulse oximetry and clinical examination in screening for congenital heart disease. *Pediatr Cardiol* 2005;26(6):832-5.

13. Rosati E, Chitano G, Dipaola L, De Felice C, Latini G. Indications and limitations for a neonatal pulse oximetry screening of critical congenital heart disease. *J Perinat Med* 2005;33(5):455-7.

14. Arlettaz R, Bauschatz AS, Monkhoff M, Essers B, Bauersfeld U. The contribution of pulse oximetry to the early detection of congenital heart disease in newborns. *Eur J Pediatr* 2006;165(2):94-8.

15. Reich JD, Connolly B, Bradley G, Littman S, Koeppel W, Lewycky P, et al. Reliability of a single pulse oximetry reading as a screening test for congenital heart disease in otherwise asymptomatic newborn infants: the importance of human factors. *Pediatric cardiology* 2008;29(2):371-6.

16. Sendelbach DM, Jackson GL, Lai SS, Fixler DE, Stehel EK, Engle WD. Pulse oximetry screening at 4 hours of age to detect critical congenital heart defects. *Pediatrics* 2008;122(4):e815-20.

17. Meberg A, Brugmann-Pieper S, Due R, Jr., Eskedal L, Fagerli I, Farstad T, et al. First day of life pulse oximetry screening to detect congenital heart defects. *The Journal of pediatrics* 2008;152(6):761-5.

18. de-Wahl Granelli A, Wennergren M, Sandberg K, Mellander M, Bejlum C, Inganas L, et al. Impact of pulse oximetry screening on the detection of duct dependent congenital heart disease: a Swedish prospective screening study in 39,821 newborns. *BMJ* 2009;338:a3037.

19. Meberg A, Andreassen A, Brunvand L, Markestad T, Moster D, Nietsch L, et al. Pulse oximetry screening as a complementary strategy to detect critical congenital heart defects. *Acta Paediatr* 2009;98(4):682-6.

20. Riede FT, Worner C, Dahnert I, Mockel A, Kostelka M, Schneider P. Effectiveness of neonatal pulse oximetry screening for detection of critical congenital heart disease in daily clinical routine-results from a prospective multicenter study. *Eur J Pediatr* 2010;169(8):975-81.

21. Walsh W. Evaluation of pulse oximetry screening in Middle Tennessee: cases for consideration before universal screening. *J Perinatol* 2010.

22. Ewer AK, Middleton LJ, Furmston AT, Bhoyar A, Daniels JP, Thangaratinam S, et al. Pulse oximetry screening for congenital heart defects in newborn infants (PulseOx): a test accuracy study. *Lancet* 2011;378(9793):785-94.