Newborn Screening for Critical Congenital Heart Disease

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with thanks to
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Objectives

Discuss Common Critical Congenital Heart Diseases that present in the Newborn period

Discuss the rationale for use of pulse oximetry for screening of critical congenital heart disease

Discuss a standardized approach to pulse oximetry screening: procedures, management, & implications

* I have no conflicts of interest to disclose
**Congenital heart disease (CHD)**

- 10-12 / 1000 live births
- Most defects occur in otherwise healthy, well-developed, term infants
- Early recognition is significantly improved given the sophistication of ECHO, fetal diagnostic clinics
- Incidence is stable
- Inheritance is multifactorial
Multifactorial inheritance

- Combination of genetic and environmental factors
- If a family member is affected, the recurrence risk increases
- Risk of recurrence is proportional to the number of genes shared
- Recurrence risk increases if the affected parent is the mother
- Recurrence risk increases with the severity of malformation
Critical Congenital Heart Disease

25% of all congenital heart defects (3 to 4 /1000)

As many as 25% leave the newborn nursery undetected

Structural defects often associated with hypoxemia in neonates in the newborn period

Require some type of intervention early in life

Increased risk of morbidity and mortality

Bradshaw & Martin, Curr opinion Ped, 2012
Importance of Diagnosis

In the United States:

- Nearly 40,000 infants/year with congenital CHD
- 4,000 infants/year with Critical CHD
- Estimated 2,000 infants/year die or have missed diagnosis
- Median age of death for deaths in CA < 2 weeks
Why missed diagnosis?

**Circulatory adaptation**
- Low → high resistance
- Shunts, blood pressure

**Respiratory adaptation**
- alveolar recruitment
- oxygen exposure

**Fluid/water balance:**
- Insensible losses
- Transitional circulation

**Metabolic adaptation:**
- Thermal stresses
- Glucose metabolism
Review: normal anatomy

In-utero circulation
Review: ductus arteriosus

In healthy term babies:
   Closes between 15-96 hours of life

In CCHD
   Can be duct dependent systemic flow
   Can be duct dependent pulmonary flow
Ductal-dependent pulmonary flow

Tetralogy of Fallot
Tricuspid Atresia
Pulmonary atresia

Patent ductus provides pulmonary blood flow
Ductal-dependent pulmonary blood flow

Tetralogy of Fallot

Tricuspid Atresia

Pulmonary Atresia/IVS

with permission (Kerry Rosen, MD)
Tetralogy of Fallot (TOF)

Four features:
- RV outflow tract obstruction (pulmonary stenosis)
- VSD
- Over riding aorta
- Right ventricle hypertrophy

Right to left shunt occurs with crying

Exam:
- Cyanosis
- Systolic murmur
- “Boot shaped” heart on CXR
Pulmonary Atresia

Failure of pulmonary valve to develop
Main pulmonary artery (MPA) is also small
Tricuspid valve insufficiency
Ductal dependent for pulmonary blood flow
Cyanotic, Systolic murmur
Intact ventricular septum is critical → must provide mixing emergently
Challenge: differentiating obstructed pulmonary circulation from PPHN:

Especially difficult in infants with PPHN in absence of parenchymal lung disease.

In one study 9% of infants with presumed PPHN had congenital heart disease (after ECMO for PPHN).

CXR with ↓ pulm vascular mkgs (? black lungs), consider CHD with ↓ pulm blood flow

Penny DJ, Arch Dis Chld, 2001
Ductal-dependent systemic blood flow

HLHS
Ao Stenosis
Interr Ao Arch
Coarctation Ao

Patent ductus provides systemic blood flow
Ductal-dependent systemic blood flow

- Hypoplastic left heart
- Coarctation of Aorta
- Interrupted Aortic Arch
Hypoplastic Left Heart Syndrome (HLHS)

Hypoplastic LV & ascending aorta
Atretic or hypoplastic aortic & mitral valve
RV supports pulmonary & systemic circulation via PDA
Systemic perfusion is ductal dependent
Diminished pulses, pallor/cyanosis, poor perfusion
Challenge: differentiating critically obstructed systemic circulation from sepsis

Difficult b/c both are associated with:

- Poor pulses, poor color, acidosis…

Incidence of sepsis & obstructed heart lesions ~ same

All infants with suspected congenital heart disease should be on antibiotics

Penny DJ, Arch Dis Chld, 2001
Other critical lesions:

TGA- parallel circulations: no mixing
TAPVR- all oxygenated blood $\rightarrow$ R side (3 types)
Truncus- 2 ventricles $\rightarrow$ single trunk for pulm & systemic flow
Double outlet RV- functionally 1 ventricle, with 2 outflow tracts
Transposition of the Great Arteries

Aorta arises from RV

Pulmonary artery arises from LV

Parallel circuits

Need ASD or PDA to allow mixing of oxygenated and un-oxygenated blood

Atrial septostomy

Cyanosis
Truncus Arteriosus

Single, large great vessel arises from both ventricles, overriding a VSD

Common trunk supplies pulmonary & systemic circulation

Pulmonary over circulation occurs once PVR falls (in absence of PS)

Bounding pulses, chest heave
Critical congenital heart disease: importance of obtaining a Diagnosis

- Most common birth defect in the US*
- CHD accounts for 8 in 1000 live births
- 4 in 1000 live births need surgical intervention in 1\textsuperscript{st} year of life.
- Nearly 40\% of deaths from congenital anomalies
- Mostly in 1\textsuperscript{st} year of life

*March of Dimes Perinatal Stats
In the past....

- We looked for Cyanosis
- Auscultated for murmurs
- Obtained 4 extremity BP’s/Palpated pulses

Difficulty with clinical diagnosis,

- many CCHD lesions are not accompanied by cyanosis
- many CCHD are not accompanied by a murmur, and
- pulses are subjective, and commonly present during transition
- Unknown rates of diagnosis with these methods!

Penny DJ, Arch Dis Chld, 2001
• Cyanosis is not a reliable indicator of critical congenital heart disease (CCHD)
METHODS: A work group was convened with members selected by the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children, the American Academy of Pediatrics, the American College of Cardiology Foundation, and the American Heart Association.

RESULTS: On the basis of published and unpublished data, the work group made recommendations for a standardized approach to screening and diagnostic follow-up. Key issues for future research and evaluation were identified.

CONCLUSIONS: The work-group members found sufficient evidence to begin screening for low blood oxygen saturation through the use of pulse-oximetry monitoring to detect CCHD in well-infant and intermediate care nurseries. Research is needed regarding screening in special populations (e.g., at high altitude) and to evaluate service infrastructure and delivery strategies (e.g., telemedicine) for nurseries without on-site echocardiography. Public health agencies will have an important role in quality assurance and surveillance. Central to the effectiveness of screening will be the development of a national technical assistance center to coordinate implementation and evaluation of newborn screening for CCHD. Pediatrics 2011;128:e1259–e1267

Kemper AR, et al, Pediatrics 2011
Pulse Oximetry for Screening

- Measures % of Hemoglobin saturated with the oxygen molecule
- Non-invasive
- Painless
- Cost effective
Pulse oximetry:

• developed in the early 1970s

• oxygenated and deoxygenated hemoglobin absorb light in different bands / wavelengths

• the ratio of light absorbance at these 2 wavelengths correlates with the saturation of Hgb in capillary bed
What is normal pulse oximetry (SpO2) reading?

• >92%? … >95%? … >98%? …
• generally accepted that > 97% is normal SpO2
• first 24 hours: can have SpO2<95% secondary to transitional circulation (lungs/heart)…screen at ≥ 24 hours
• newborn saturations decrease with sleep, feeding or crying (therefore, screen when awake)
Equipment / personnel needed

- Motion-tolerant Pulse Oximeter (cleared by FDA)
- Infant Disposable or Reusable Pulse Oximeter Sensor/Probe
- Rolling Cart for supplies
- Data collection form for collecting results, etc…
- Dedicated individual(s) trained to perform (and report) screening results
- Red Heart Shaped Stickers
- Blankets for warming the infant and to block extraneous light
- Parents for comforting infant
Helpful Hints

• Pair screening with other standard care screenings (audiology, NBS) after 24 hours of age, before D/C.
• Conduct screenings in a quiet area and with parents present, if possible.
• Ideally done when infant is quiet and alert
• Do not do while infant is cold or crying
Screening Procedure

1. Wrap probe around outside of infant’s right hand (pre ductal). Place the light emitter on the top of the hand, with the photo detector directly opposite on the bottom of the hand.

2. Obtain reading
Screening Procedure

3. Wrap probe around the outside of infant’s foot (post ductal). Place light emitter on top of foot, with photo detector directly opposite on the bottom of the foot.

4. Obtain reading

5. Remove probes when finished.
   - Disposable probes – dispose of properly
   - Reusable probes – dispose of disposable wrap and clean reusable probe
Screening Procedure

5. Remove probes when finished.
   Disposable probes – dispose of properly
   Reusable probes – dispose of disposable wrap and clean reusable probe

6. Document procedure and both pre and post ductal readings.

7. Follow Algorithm for repeating if necessary based on readings.

8. Follow Nursery protocol for positive readings
Pearls:

• Place probe on infant before turning machine on
• Cover the extremity with a blanket to protect it from ambient light (may interfere with signal)
• To assure a good reading remember:
  • Heart rate display should be consistent with a newborn heart rate – 100 to 160 bpm
  • Arterial wave form should be stable with no motion artifact
Pulse Oximeter Arterial Waves

The effect of artifact on a pulsatile signal from an oximetry sensor

With permission,
Kerry Rosen, MD
Who: All infants

All infants will be screened before discharge unless they have had an ECHO

When: at or after 24 hours of life.

Screen infants after 24 hours of age and before discharge, or when medically appropriate in infants born premature or critically ill.

How: Right hand (pre-ductal), any one foot

Place sensor on baby’s RH and either foot. The two readings may be done either in parallel or in sequence. Screening when infant is quiet and alert reduces the likelihood of a false positive screen.
Why right hand and a foot?
pre-ductal = right hand
post-ductal = foot
Kemper et al, 
*Pediatrics*. 2011; 128: e1259-1267

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Strategies for Implementing Screening for Critical Congenital Heart Disease

- **Pulse Ox on Right Hand (RH) and One Foot After 24 Hours of Age**

  - **FAIL**
    - Pulse Ox <95 Percentage Points (both RH & foot) or Difference of >3 Percentage Points Between RH and Foot
    - Repeat Pulse Ox in 1 Hour
    - **FAIL**
      - Repeat Pulse Ox in 1 Hour
      - **FAIL**
        - Clinical Assessment

  - **PASS**
    - Pulse Ox ≥95 Percentage Points (RH or Foot) and Difference of ≤3 Percentage Points Between RH and Foot
    - Normal Newborn Care

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RH Application Site

Foot Application Site

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NATIONWIDE CHILDREN'S
When your child needs a hospital, everything matters."
Pass (negative screen)

If: SpO2 in right hand and lower extremity is $\geq 95\%$
and the absolute difference between the right hand and either lower extremity is $\leq 3\%$

The screening is negative (passed screen) and no additional screening is needed
Fail (positive screen)

SpO2 < 90% (RH or either foot) = definite FAIL

Next step:
- consult cardiology or order ECHO

If SpO2 90% - 94% in RH, or if the difference b/t RH and either foot >3% is inconclusive, possible fail

Next step:
- repeat in one hour (2nd); may repeat (3rd) 1 hr later
- If still 90% - 94% in RH <or>
- If > 3% difference b/t RH and foot = FAIL

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**Strategies for Implementing Screening for Critical Congenital Heart Disease**

**Pulse Ox on Right Hand (RH) and One Foot After 24 Hours of Age**

- **FAIL:** Repeat Pulse Ox in 1 Hour
  - **FAIL:** Repeat Pulse Ox in 1 Hour
  - **FAIL:** Clinical Assessment

- **PASS:** Normal Newborn Care
  - Pulse Ox ≥95 Percentage Points (RH or Foot) and Difference of ≤3 Percentage Points Between RH and Foot

- **Pulse Ox <95 Percentage Points (both RH & foot) or Difference of >3 Percentage Points Between RH and Foot**

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**RH Application Site**

**Foot Application Site**
Failed Screen → now what?

• assess for pulmonary or infectious process
• echocardiogram locally (performed/interpreted)
• echocardiogram locally, remote interpretation (telemedicine or “tele-echo”)
• transfer to tertiary care center
• discharge from nursery with plans for office evaluation with pediatric cardiologist (certain cases; use caution)
Failed Screen = needs high quality ECHO

- sonographer training
- appropriate machine, probes, pediatric settings
- read by pediatric cardiologist
- accuracy of pediatric echocardiograms interpreted by adult cardiologists is low
Which CCHD will we catch?
7 main diseases (+1)

- HLHS
- pulm atresia/IVS
- TGA
- truncus arteriosus
- tricuspid atresia
- tetralogy of Fallot
- TAPVR
- +1: persistent pulm HTN
Secondary Targets for CCHD Screening

- Coarctation of the aorta
- Double outlet right ventricle
- Ebstein anomaly
- Interrupted aortic arch
- Single Ventricle
Pulse Ox Screening will NOT detect

- most aortic stenosis
- most pulmonary stenosis
- ASDs, VSDs…
- some single ventricles lesions with normal saturations early in life…
- parental education is important
What about False Positives?

- Incidence is Low (0.07% - 0.17%)
- Cyanotic infants should be transferred immediately

With permission, Kerry Rosen MD
Ohio... the latest information

- Senate Bill 4 signed into law by Governor Kasich, summer 2013 for mandatory Pulse Oximetry Screening of newborns at 24 hours of life, before hospital discharge
- “Rules” have been written, and are soon to be finalized.
- Will be developed for implementation of law
www.nationwidechildrens.org

search: pulse oximetry

Congenital Heart Disease Screening Form

Last name: ___________________________ Medical record #: ___________________________
First name: ___________________________
Date of birth: _________________________
Age at initial screening: ____________ hours

Initial Screening

Time: ___________________________
Pulse oximetry on right hand %: ___________________________
Pulse oximetry on left hand %: ___________________________
Difference (right hand – left hand) %: ___________________________

Second Screening (4 hour following initial screening, if initial screening failed)

Time: ___________________________
Pulse oximetry on right hand %: ___________________________
Pulse oximetry on left hand %: ___________________________
Difference (right hand – left hand) %: ___________________________

Third Screening (1 hour following second screening, if second screening failed)

Time: ___________________________
Pulse oximetry on right hand %: ___________________________
Pulse oximetry on left hand %: ___________________________
Difference (right hand – left hand) %: ___________________________

What is pulse oximetry?
Pulse oximetry (also called Spo2) is a simple and painless test that measures how much oxygen is in the blood. Another term for pulse oximetry is “pulse ox” or “sat.”

How is the pulse oximetry test performed?
A small clip, like a bedazzling, with a small red light (or pulse) is placed on the baby’s hand or foot. The probe is attached to a wire, which is attached to a special monitor that shows the pulse oximetry reading. The pulse oximetry test takes just a few minutes to perform while the baby is still, quiet and warm. If a baby is crying, squirming or cold, it may take longer or not be possible. You can help ensure your baby is still, quiet and warm.

Why is pulse oximetry conducted?
The pulse oximetry test is routinely used to monitor an infant’s oxygen level during a procedure or treatment. It can also help to identify babies with serious heart conditions that may have low levels of oxygen in their blood.

A doctor or nurse practitioner may ask for more testing, such as an ultrasound of the heart or echocardiogram, when a low pulse oximetry reading is identified. The echocardiogram will screen for a serious problem in the structure or blood flow of the heart. The pulse oximetry test can identify a baby with serious congenital heart disease (CHD) before he or she leaves the newborn nursery.

Nationwide Children’s
When your child needs a hospital, everything matters.
Thank you for your attention!

Special thanks to:

• Gail Bagwell, RN, MSN
• Kerry Rosen, MD, MBA

Questions?