

**Detection of Critical Congenital Heart Disease in the Newborn with Pulse Oximetry**

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**Detection of CCHD With Pulse Oximetry**

I have no relevant financial relationships with the manufacturer of any commercial product discussed here.

I do not intend to discuss an unapproved / investigative use of a commercial product / device in my presentation.

Thank you Robert Morrow, MD

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**Objectives**

- Why talk about early detection of CCHD?
- Why screen for CCHD with pulse oximetry?
- What are we screening for?
- Examine recent data on use of pulse oximetry for "screening" for congenital heart disease with emphasis on cyanotic congenital heart disease
- Review guidelines for screening from the SACHDNC Work Group Secretary's Advisory Committee on Heritable Dis. Newborn

The terms Cyanotic Congenital Heart Disease and Critical Congenital Heart Disease will be used interchangeably.

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**A Missed Diagnosis**

A six day old full term infant presents to a local emergency room with history of 24 hrs of tachypnea, poor feeding, and lethargy. A quick assessment reveals an ashen, moribund baby.

Pulse 180, Resp 100, Temp 35r, and O2 sat 79%. Oxygen is started and while preparing for intravenous access and intubation a CXR is obtained....

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**The CXR Shows.....**



Bertram H et al. Circulation 2008;117:e319-e321

American Heart Association

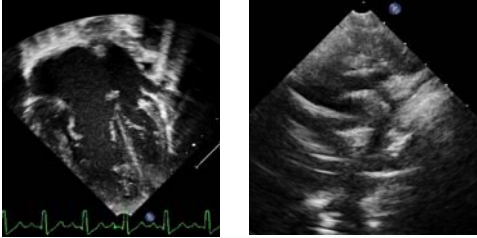
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**What next!**

A stat call goes to the cardiologist for evaluation and an echocardiogram is obtained.

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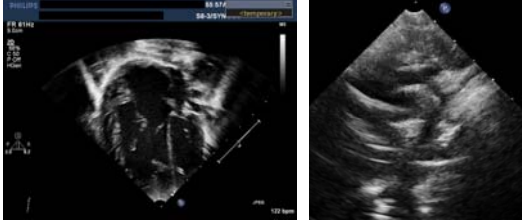
### The echocardiogram shows.....



Hypoplastic Left Heart      Hypoplastic Aortic Arch

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### The echocardiogram shows.....



Hypoplastic Left Heart      Hypoplastic Aortic Arch

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### Detection of CCHD

A diagnosis of hypoplastic left heart syndrome is made. The baby is intubated, placed on prostaglandin infusion, and an initial ABG is obtained showing **pH=6.91**, PaCO<sub>2</sub>=28, PaO<sub>2</sub>=45 and BE= -15.

After explaining the nature of the defect and the dire straits of the baby the distraught family asks...

**Why wasn't this picked up in the nursery?**

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### Why Talk About Detection of CCHD?

- CHD most common birth defect, 8/1000 newborns
- CCHD occurs in 1.8/1000 accounts for 6-10% of infant deaths
- Prenatal dx is made in 23% pregnancies, 11% live births
- 30% of CCHD (13-48%) may leave nursery without dx
- 1.4/10,000 infants die from CCHD before recognition

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### Why Talk About Detection of CCHD?

- Of infants with CHD who die, 15% die as a result of sequelae of delayed diagnosis. 89% would have had cyanosis.  
Death in infancy from unrecognized congenital heart disease  
 Abu-Harb et al. Archives of Disease in Childhood
- Liske et al estimated that 7 infants with critical left heart obstructive disease will be missed per year in Tennessee.  
Report of the Tennessee Task Force on Screening Newborn Infants for Critical Congenital Heart Disease: Pediatrics 2006;118:e1250

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### Why Talk About Detection of CCHD?

- Failing to detect CCHD in the nursery may lead to serious events such as cardiogenic shock or death
- Survivors presenting late are at greater risk for neurologic injury and subsequent developmental delay or other organ damage due to hypoxia

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## Why Talk About Detection of CCHD?

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Consequences of missing CCHD in the newborn are very serious, often lethal.

We have the ability to identify CCHD and treat before clinical deterioration.

## Why Talk About Detection of CCHD?

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Early detection of CCHD can potentially improve the prognosis and decrease the mortality and morbidity rate of affected infants



## Why Screening for CCHD?

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- Growing public awareness of options for palliation and repair of cyanotic congenital heart disease (arterial switch, Norwood)
- There is growing and sufficient evidence that supports the use of pulse oximetry to detect CCHD in well appearing infants

## Why Screening for CCHD?

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Statement by AAP and AHA (Mahle et al, 2009)

- “the association of delayed diagnosis of CCHD with mortality, morbidity, and disability provides a rationale for strategies such as pulse oximetry assessment to improve early detection”.
- Called for “studies in larger populations and across a broad range of newborn delivery systems” before Pox screening should be recommended.

## Why Screening for CCHD?

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- Large scale studies in Sweden (deWahl-Grenelli) and Germany (Riede)
- Work group of the SACHDNC convened January 2011 to consider the evidence and make recommendations for screening methodology
  - AAP, AHA, ACCF, NIH, CDC, FDA, other agencies and parents

## Why Screening for CCHD?

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Large scale studies in Sweden (deWahl-Grenelli) and Germany (Riede) 2009-2010

Study	N	NI Sat	Sens %	Spec %	False Pos	PPV	NPV
Sendelbach	15,233	≥ 96	50	94	4.5	0.1	99
Reide	41,445	≥ 96	60	99	0.1	75	100
De Wahl-Grannelli	38,429	≥ 95	62	99	0.2	12	100

## Why Screening for CCHD?




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In January, 2011 the *Health Resource Service Administration's Advisory Council on Heritable Diseases in Newborns and Children*, hosted a workshop to discuss implementation recommendations surrounding screening.

AAP, AHA, ACCF, NIH, CDC, FDA, other agencies and parents

The outcome of this meeting included a screening protocol based on the most current evidence.

This protocol is reflected in the recommendations which follow.








# PEDIATRICS

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

**Strategies for Implementing Screening for Critical Congenital Heart Disease**  
 Alan E. Koenig, William T. Mahle, Gerald R. Martin, W. Carl Cowley, Praveen Koush, W. Robert Moore, Kellie Krebs, Omid D. Pourson, Jill Chiodo-vill, Scott D. Greene and E. Rodney Howell  
*Pediatrics* 2011;128(4):1219, originally published online October 10, 2011;  
 DOI: 10.1542/peds.2011-1317

**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.

## Why Screening for CCHD?




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Health and Human Services Secretary Kathleen Sebelius endorses the inclusion of screening for critical CHD in the recommended uniform screening panel.

Aug, 2011

The American Heart Association, American Academy of Pediatrics and American College of Cardiology also support newborn pulse oximetry screening for critical CHD.

Policy Statement: *Pediatrics* 2012;129:190









## Pulse Oximetry Screening for Critical Congenital Heart Disease

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Legislation Passed / Considered

- Maryland
- New Jersey
- Mississippi
- Tennessee
- Virginia
- West Virginia










## Seven CCHDs can be detected by pulse oximetry: Primary targets

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- Hypoplastic left heart syndrome
- Pulmonary atresia (with intact septum)
- Tetralogy of Fallot
- Total anomalous pulmonary venous return
- Transposition of the great arteries
- Tricuspid atresia
- Truncus arteriosus

These represent about 17-31% of all congenital heart disease








## Can all CHD be detected by Pulse Oximetry?

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No

- Primary targets
- Secondary targets

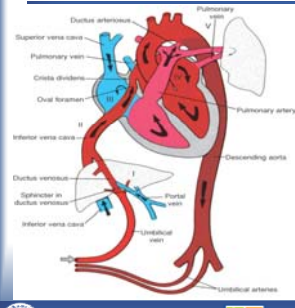




### Secondary targets (may not be consistently picked up by Oximetry)


- Aortic arch atresia or hypoplasia, interrupted aortic arch, coarctation of the aorta,
- Double-outlet right ventricle,
- Ebstein's anomaly, pulmonary stenosis, atrioventricular septal defect,
- Single ventricle defects (other than hypoplastic left heart syndrome and tricuspid atresia).



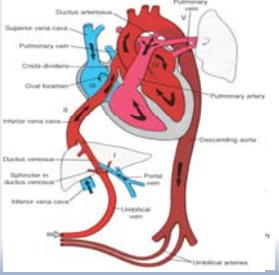
### Fetal "Parallel" Circulation




- Foramen bypasses right ventricle to perfuse upper body
- Ductus bypasses lungs to perfuse lower body and placenta
- Both may provide majority of flow to upper and lower body in the case of valve atresia, stenosis, or chamber hypoplasia ("single ventricle")



### Transitions at Birth: A Perfect Storm




- Ductus closes and lungs receive entire output of RV
- Pulmonary resistance drops and pulmonary venous return to LA facilitates closure of foramen
- Series circulation is established

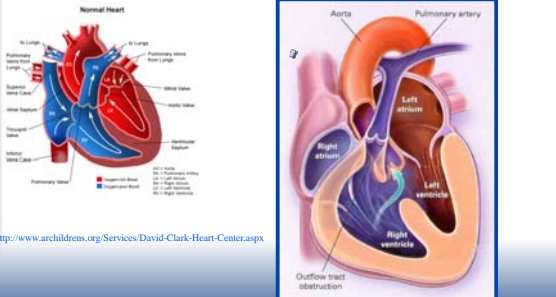


### CCHD


- **Ductal Dependent Pulmonary Circulation**  
Tetralogy of Fallot / Pulmonary atresia  
Tricuspid atresia
- **Ductal Dependent Systemic circulation**  
Hypoplastic left heart syndrome  
Interrupted aortic arch
- Transposition of the great arteries
- Total anomalous pulmonary venous return
- Truncus arteriosus



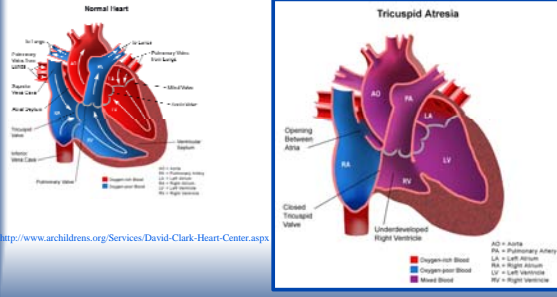
### Tetralogy of Fallot and Pulmonary Atresia Ductal Dependent Pulm Circ




<http://www.archildrens.org/Services/David-Clark-Heart-Center.aspx>



### Tricuspid Atresia Ductal Dependent Pulm Circ



<http://www.archildrens.org/Services/David-Clark-Heart-Center.aspx>



### Hypoplastic Left Heart Ductal Dependent Systemic Circ

**Normal Heart**

**Hypoplastic Left Heart Syndrome**

Very Small Aorta, Vessel Connecting Aorta and Pulmonary Artery, Opening Between Aorta, Underdeveloped Left Ventricle.

AD = Aorta, PA = Pulmonary Artery, LA = Left Atrium, RA = Right Atrium, LV = Left Ventricle, RV = Right Ventricle.

Legend: Red = Oxygen-rich Blood, Blue = Oxygen-poor Blood, Purple = Mixed Blood.

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### Transposition of the Great Arteries Parallel Circulation

**Normal Heart**

**Transposition of Great Arteries**

Vessel Connecting Aorta and Pulmonary Artery, Opening Between Aorta, Vessels Switched.

AD = Aorta, PA = Pulmonary Artery, LA = Left Atrium, RA = Right Atrium, LV = Left Ventricle, RV = Right Ventricle.

Legend: Red = Oxygen-rich Blood, Blue = Oxygen-poor Blood, Purple = Mixed Blood.

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### Why Screening for CCHD?

Mellander and Sunnegardh, Acta Paediatr 2006;95:407

**By 2 days of age.....**

- 25% of CCHD with ductus dependent pulmonary circulation undiagnosed
- 60% with ductus dependent systemic circulation or other right to left shunt
- 75% of coarctation, 45% of interrupted aortic arch, 40% with aortic stenosis, and 25% with HLHS

**In US nurseries these babies would be long gone.....**

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### Detection of CCHD

Is this baby cyanotic?

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### Screening Physical Exam?

- Screening exam depends on detection of symptoms and signs (tachypnea, heart murmurs, overt cyanosis, diminished pulses)
- Cyanosis (visible desaturation) hard to recognize in infants (especially in more pigmented).

(4-5 gms of deoxygenated Hb to recognize desaturation. For newborn with Hb of 20, an 80% saturation is needed to see cyanosis).

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## Screening Physical Exam?

- Pulses may be present in severe LV obstructive disease or coarctation when the ductus remains patent.
- Changes occur at ductal closure; early discharge (24hrs) creates limitations for PE
- Physical exam 10 times more likely to yield false positives (murmur) leading to evaluation  
Wahl-Grenelli, Riede



## Screening ECG, CXR, Echo?

- ECG and CXR lack sensitivity but can provide diagnostic information
- Echocardiography is highly sensitive and, in trained hands, highly specific.
  - Limited availability for mass screening
  - False positives and negatives in untrained hands
  - Echo's obtained prior to ductal closure can lead to false reassurance
  - Expensive !!!!



## Detection of CCHD Fetal Echocardiogram

- Obstetric screening ultrasound based on demonstration of four chamber heart-
  - will miss 4 chamber CCHD
- Limited training of obstetric personnel
- Detects fewer than 50% of cases
- Fewer than 50% single ventricle referrals for surgery had prenatal detection and less than 30% of four chamber CCHD had prenatal diagnosis.



## Screening for CCHD Is Oximetry a Viable Screening Tool?

- Pulse oximetry is based on the differential absorption of light by oxygenated and deoxygenated Hb
- Does not require calibration
- Correlates well with PaO2
- Reliable normal values exist at 20-24hrs.



## Pox Screening for CCHD Sensitivity, Specificity, PPV, NPV:2009 and after

Study	N	NI Sat	Sens %	Spec %	False Pos	PPV	NPV
Sendelbach	15,233	≥ 96	50	94	4.5	0.1	99
Reide	41,445	≥ 96	60	99	0.1	75	100
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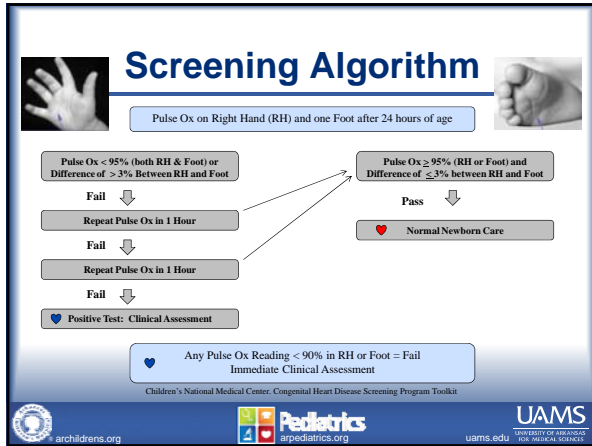


## Then How Do You Screen Recommended Screening Algorithm

*Kemper et. al. Pediatrics, November, 2011*

- Screen at 24hrs or later, if at all possible after 12 hours
- Determine value for right arm and one leg
- < 90% sat= positive screen
- <95% in right arm and leg or >3% between arm and leg= initial positive.
- Repeat hourly times two (total 3 screens)
- Negative if ≥ 95% and ≤ 3% between arm and leg. If all 3 screens positive then positive screen





## POx Screening for CCHD

### What is role of primary care physician?

- Results should be communicated to physician who should be directing further evaluation and follow-up.
- Physician should be involved in arranging screening for babies who did not get screened.
- Physician receive results of further testing.
- Central role in ensuring long-term follow-up and care for those infants with CCHD

## Positive Screen?

### What do you do?

- "Comprehensive evaluation for causes of hypoxemia"
- "In absence of other findings to explain hypoxemia, CCHD needs to be excluded on the basis of a diagnostic echocardiogram."
- Echo must be performed by tech experienced with neonatal echo and congenital heart disease and interpreted by pediatric echocardiographer.
- Consult pediatric cardiologist, when possible, before obtaining the echocardiogram.

## POx Screening for CCHD

### Secondary Targets- positive screen, not CCHD

**50% of false positives have significant neonatal disease**

- Neonatal pneumonia
- Asymptomatic transitional circulation
- Persistent pulmonary hypertension
- Sepsis ( most common)
- Polycythemia

## POx Screening for CCHD

### What are Limitations of POx?

False positives

- Bad test- older equipment, technician, probe
- Patient motion, crying
- Transient hypoventilation
- Hyperbilirubinemia
- Hemoglobinopathy

## POx Screening for CCHD

### What are Limitations of POx?

False negatives

- "Acyanotic cyanotics"
- Other acyanotic critical CHD
- Ambient light, probe malfunction, etc.




## POx Screening for CCHD Is Therapy Available for CCHD

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Is therapy available for critical CHD detected by screening?


Yes, effective short and long term treatment is available.



Congenital Heart Defects  
Repairs and Outcomes in 2012

Renee, A. Bornemeier, MD  
Professor of Pediatrics  
Division of Cardiology  
UAMS and ACH








## A Few Essentials

### What equipment? Who performs test?

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- New generation pulse oximeter required!
- Minimal training (technician, LPN, RN)
- May use re-usable probes; clean according to guidelines.
- Use tape on probes, not clip on
- Probe should be recommended probe by manufacturer of pulse oximeter
- Baby should be supine, not prone




## POx Screening for CCHD

### Some things are obvious!

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"The ringing in your ears—I think I can help."

# QUESTIONS ?










## POx Screening for CCHD

### In Arkansas

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- Collaboration established between Arkansas Department of Health, Arkansas Newborn Screening Program, ACH, UAMS Neonatology, and others
- Currently not mandated and will require legislation to mandate
- UAMS is implementing screening
- Applying for grant funding from HRSA
- If interested in establishing a screening program at your hospital call Diana McDaniel, Senior Project Director, ACH,
- See guidelines for screening in Kemper, et al Pediatrics, Nov. 2011, e-pub

## What can pediatricians do to help facilitate screening?

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- Work with your hospital or delivery center.
- Engage your AAP Chapter leadership.
- Advocate with state health department to support statewide adoption of screening.
- Where legislation is required engage legislators and policy makers, educate, and advocate for screening.
- Some states have adopted (Maryland, New Jersey, Mississippi, others)
- Advocate for evidence based screening based on guidelines of the working group (Kemper, et al Pediatrics, Nov. 2011)

## POx Screening for CCHD

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Is the cost benefit ratio favorable for pulse oximetry?

## POx Screening for CCHD

### What is the cost?

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- Cost of POx machine and related equipment
- Time cost of tech, LPN, RN (takes 1-5 minutes per screen if screening arm and leg)
- Estimate cost is \$10/baby (compare hearing screen at \$30/baby)
- Estimated 2.3 echo's per CCHD as result of positive screen
- Savings from one case identified without circulatory complications exceed cost of screening 2000 babies.

## POx Screening for CCHD

### Costs for Screening for CCHD

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- Analysis by Bill Mahle (personal communication) estimated \$0.99 cost per infant screened and \$3,000 to \$6,000 cost per life saved
- Assumptions and lack of data prevent accurate cost analysis for U.S.

## POx Screening for CCHD

### What is the cost?

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- Cost of telemedicine infrastructure?
- Cost of transport?
- Emotional cost of false positive and negative diagnoses?
- Who pays? (hearing screen reimbursed by most payers)

## POx Screening for CCHD

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Is the cost benefit ratio favorable for pulse oximetry?

Maybe, but more data needed.

## Detection of CCHD

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<http://www.archildrens.org/Services/David-Clark-Heart-Center/CCHD-Newborn-Screening.aspx>

**Or contact:**  
 Diana McDaniel, MPA CMPE  
 501-364-7800 [DMcDaniel@archildrens.org](mailto:DMcDaniel@archildrens.org)

<http://www.childrensnational.org/pulseox/>

## Detection of CCHD in the Newborn



“If you save one life it is as if you have saved an entire world.”

Talmud

