Objectives

- Recognize the importance of screening for Critical Congenital Heart Defects (CCHD) using pulse oximetry
- Apply the New Jersey recommended CCHD screening protocol
- Identify the role of the primary care physician in the detection of and referral of CCHD

Disclosure

- Dr. Koppel has no conflict of interest to disclose.
- The New Jersey Department of Health does not endorse or promote a specific brand or vendor for pulse oximetry supplies and equipment. Equipment and/or supplies presented in the education are for informational purposes only.

The Law

“[The Commissioner of Health and Senior Services shall require] each birthing facility licensed by the Department of Health and Senior Services to perform a pulse oximetry screening, a minimum of 24 hours after birth, on every newborn in its care.”

- NJ first state to implement a mandate for pulse oximetry screening
- Legislation signed into law June 2, 2011
- Implementation date August 31, 2011
- P.L. 2011, Chapter 74

Congenital Heart Defects

- 8-9/1,000 live births
- Minimum of 32,000 -40,000 infants affected each year in US
- Approximately 25% of these are critical congenital heart defects (CCHD) or about 2 in 1,000 live births

Dylan’s Story

Dylan’s Story:

On September 1, a day after the law mandating inclusion of pulse oximetry testing on newborns became effective, a hospital pediatrician informed Lisa and Bill Gordon of Newton that the test performed on their baby was abnormal and he had a murmur.

Dylan was rushed to Morristown Medical Center, where it was determined he needed specialized pediatric cardiac heart surgery.

Dylan was transferred to Columbia University Medical Center, and several days later had the life-saving surgery correcting the abnormality discovered from the newly mandated newborn testing.

**Rationale for Pulse Ox Screening**

- An estimated 25-30% of newborns with CCHD could be missed at the time of hospital discharge (Mahle et. al., 2009).
- About 1,200 more newborns with CCHD could be identified at birth hospitals using pulse oximetry (Peterson et al., 2013).
- Approximately 200 newborns have died each year from missed CCHD and numerous others have significant morbidity from delayed diagnoses (Hokanson, 2010).
  - Compare to an average 66 young athletes each year who die suddenly of undiagnosed cardiac defects (Maron et al., 2009).

**Morbidity Due to Delayed Diagnosis**

- Shock - global hypoxic injury with multi-organ dysfunction
  - Hypotension
  - Poor ventricular function
  - Myocardial ischemia
  - Pulmonary hypertension
  - Renal dysfunction
  - Hepatic dysfunction
  - Decreased intestinal blood flow - NEC
  - DIC
  - Metabolic: hypoglycemia, hypocalcemia, myoglobinuria, hypoxic-ischemic encephalopathy

**Detection of CCHD Continuum**

- Prenatal Ultrasound
- Physical exam

**Detection of CCHD**

<table>
<thead>
<tr>
<th>Percentage</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>60%</td>
<td>Prenatal</td>
</tr>
<tr>
<td>20%</td>
<td>Clinical</td>
</tr>
<tr>
<td>15.6%</td>
<td>Pulse Ox</td>
</tr>
<tr>
<td>4.4%</td>
<td>Diagnostic Gap</td>
</tr>
</tbody>
</table>

**Not As Pink As You Think...**

- Some babies can initially appear healthy
  - Some babies do not have murmurs or cyanosis
  - Physical exam alone failed to identify half of CHDs that were not detected by a prenatal ultrasound
  - It's estimated that 30% of infant deaths from CCHD occur prior to diagnosis

**The Cyanotic “Blind Spot”**

- Hemoglobin of 17.5 g/dL, 83% Abnormal Saturation
  - Visible Cyanosis
  - No Visible Cyanosis
- Hemoglobin of 13.5 g/dL, 95% Normal

Hokanson, 2010.
CCHD Screening with Pulse Oximetry

- Indirectly monitors the oxygen saturation of a patient’s blood and variations in blood flow in the skin
- Can detect mild hypoxemia without apparent cyanosis
- Can provide continuous and direct values
- Non-invasive
- Easy to use and widely available
- Cost-effective and extensively used

The Texas Pulse Oximetry Project, 2013.

Newborn Screening - New York – 1960’s

Robert Guthrie, MD  Virginia Apgar, MD


Screening Cost

- CDC Study in 7 NJ Birthing facilities
  - Mean screening time per newborn was 9.1 minutes (standard deviation 3.4 minutes)
  - Hospitals’ total mean estimated cost per newborn screened was $14.19 (in 2011 U.S. dollars), consisting of $7.36 in labor costs and $6.83 in equipment and supply costs

Peterson et al., 2014.

Screening Cost per Infant

- Hospital – Based Hearing Screening $36 - $39
- Laboratory Metabolic Screening $20.00
- Pulse Ox Screening $14.19

These cost estimates exclude follow-up costs, such as further diagnostic testing, as well as non-reimbursable overhead costs.

Screening Case Discussion

- Brandon- TGA- failed screen; early detection
- HLHS – not screened; late detection
- Coarctation of aorta- passed screen (false negative); late detection

Case Presentation

- 40 weeks gestation, C/S, 3600 grams
- Discharged home on day 3
- Day 5: returned to ED for poor feeding and decreased activity
- SpO₂: 80%
- ABG: pH 6.8
- Echo: HLHS
- Pre-op stabilization X 5 days
- Norwood stage I
- Post-op ECMO X 8 days
Case Presentation

- 39 weeks, NSVD, Apgar 9/9
- Discharged home on Day 2
  - Oximetry screening - post-ductal SpO2 100%
- Day 3
  - Lethargy
  - Decreased PO intake
  - Dry diapers
  - Tachypnea
  - Evaluated by pediatrician

- Referral to ED for respiratory distress
  - grunting
  - retracting
  - unable to measure SpO2
- Intubated
- Umbilical arterial and venous catheters inserted
- ABG: 7.09/17/199/8/-23.3
- Chemistry: 143/8/6/63/5.98
- Echo: coarctation, ductus arteriosus closed
  - (history of normal fetal echo)
- Prostaglandin infusion
- Dialysis prior to repair of coarctation

CCHD Screening

- Seven Primary Targets (17-31% of all CHDs):
  - Hypoplastic left heart syndrome
  - Pulmonary atresia (with intact ventricular septum)
  - Tetralogy of Fallot
  - Total anomalous pulmonary venous return
  - Transposition of the great arteries
  - Tricuspid atresia
  - Truncus arteriosus

- Five Secondary Targets:
  - Coarctation of the aorta
  - Double outlet right ventricle
  - Ebstein anomaly
  - Interrupted aortic arch
  - Single ventricle

Effect of CCHD on O2 Saturations

Modified diagrams courtesy of the Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities. www.cdc.gov/ncbddd/heartdefects
Reliability of Pulse Ox Screening for CCHD

- Meta-analysis of 13 eligible studies with data for 229,421 newborn babies
  - Overall sensitivity of pulse oximetry for detection of critical congenital heart defects was 76.5% (95% CI 67.7 – 83.6)
  - Specificity was 99.9% (95% CI 99.7 – 99.9)
  - False-positive rate of 0.14% (95% CI 0.06 – 0.33)
  - Low false positive rate when pulse ox done after 24 hours from birth compared to before 24 hours

Thangaratnam et al., 2012.

CCHD Screening 2011

- Maryland Law Enacted
- Indiana Law Enacted
- New Jersey Law Enacted
- 1st Newborn screened positive after mandated screening
- CCHD added to the RUSP
- AAP Endorses POxS

AAP CCHD Screening Map: 2011 State Actions on CCHD

- 3 states enacted legislation

AAP CCHD Screening Map: States’ Actions During 2014 (As of January 29, 2015)

Performing the Screen

- Best results when infant is at least 24 hours of age
- Use proper sensor for the device
- Conduct screening in quiet area and, if possible, with parent present to soothe and comfort the infant
- Conduct screening while infant awake and quiet
- Avoid screening when infant is crying, cold or in a deep sleep

Reich et al., 2008.

Technical Factors

- False positive and negative readings
  - Poor perfusion
  - Motion artifact
  - Ambient light
  - Partial probe detachment
  - Differences between manufacturers

Reich et al., 2008.
NJ Recommended Algorithm: Abridged Version

**PASS**
- 95%-100% in both extremities AND a difference of 3% or less between the readings.

**RESCREEN IN 1 HOUR**
- 90%-94% in either extremity OR a difference of 4% or more between the readings. Rescreen the infant up to 2 times, for a total of 3 screens.

**FAIL**
- 90%-94% in either extremity OR a difference of 4% or more between the readings after three screens.

**IMMEDIATE FAIL**
- A reading of 89% or less in either extremity.

Evaluation for Failed Screen

- Clinical assessment
- Exclude infectious or pulmonary pathology
- Complete echocardiogram
- Pediatric cardiology referral as specified

Managing the Failed Screen

- Unless a non-cardiac cause can be identified for a failed screen, an infant who fails the screen should have a diagnostic echocardiogram done before being discharged
- This could involve an echocardiogram within the hospital or birthing center, transport to another institution for the procedure, or the use of telemedicine for remote evaluation

Parent Education

Available in English and Spanish

http://nj.gov/health/fhs/nbs/cchd_resources.shtml
Communication of Screening Results

- Include screening results in discharge summary
- Include in the hand-off report to the receiving hospital if infant was transferred

First Office Visit

- Pediatrician should have access to all screening results from hospital (Metabolic, CCHD, Hearing)
  - Metabolic
  - CCHD
  - Hearing

- This should be a “clean hand-off” between the hospital and pediatrician
- If patient not appropriately screened at birth facility, develop strategies for screening and evaluation

If Your Patient Failed CCHD Screening

- Confirm that the infant had a diagnostic echocardiogram
  - Make sure that the patient receives appropriate follow-up, such as being seen by a cardiologist
  - Facilitate long-term follow-up for patients diagnosed with CCHDs

Signs & Symptoms of CHD

- Cyanosis
- Tachypnea (often with diaphoresis during feeding)
- Lethargy
- Feeding difficulty
- Poor weight gain

New Jersey Pulse Ox Screening Experience
New Jersey POxS Experience

- Data Reporting
  - Birthing facilities are required to report all failed POxS to the NJ Birth Defects Registry (BDR)
  - Health care professionals are required to report infants with CCHD (and other congenital defects) who are New Jersey residents to the BDR
  - Pulse ox core team and BDR staff investigate all POxS fails and registered cases of CCHD
  - New electronic birth record (Vital Information Platform) includes prenatal CCHD detection and POxS data

Failed Screens Registered to NJBDR: August 31, 2011- September 30, 2014

- Total fails N= 208
- Diagnostic evaluation attributable to POxS:
  - No (One of 3 criteria) N= 111
  - Yes (None of 3 criteria) N= 97

One of 3 Criteria
- Prenatal diagnosis of CCHD
- Signs/symptoms at the time of the screen
- Cardiac consult or echocardiogram planned prior to the screen

Results of NJ Screening

- August 31, 2011- September 30, 2014
- Number of live-births: 313,005
- Number of live-births eligible to be screened: 304,211
- Number of live-births screened: 304,211
- Proportion of eligible live-births screened: 99.6%

*Excludes expirations, <24 hours or not medically appropriate at end of period

Failed Screens Registered to NJBDR

- Diagnostic evaluation attributable to POxS (n=97)
  - 14 - CCHD
  - 12 - CHD
  - 9 - Other significant non-cardiac conditions
  - 27 - PDA or PFO as only finding
  - 35 – No documented reason for failed POxS
    - 23 (66%) did not follow protocol

Characteristics of Failed Screens: CCHD

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<tr>
<th>Case</th>
<th>Days</th>
<th>POxS</th>
<th>Cardiac</th>
<th>Screen</th>
<th>Followed Protocol</th>
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<td>Y</td>
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</table>

14 Infants with CCHD Were Detected

- Coarctation of the aorta (5)
- Ebstein anomaly (1)
- D-Transposition of the great arteries (2)
- Tricuspid atresia (1)
- Total anomalous pulmonary venous return (5)
Other Conditions Detected

- 12 CHDs:
  - Atrial septal aneurysm
  - ASD
  - VSD
  - Pulmonary artery/pulmonary artery branch stenosis

- 9 Other significant non-CHD medical conditions:
  - Sepsis
  - Pneumonia
  - Persistent pulmonary hypertension
  - Pulmonary bulla

Screening Success

- Since implementation, 99.6% of eligible infants were screened in New Jersey
- POxS in NJ led to the detection of:
  - 14 infants with critical congenital heart defects (CCHD)
  - 12 infants with other congenital heart disease (CHD)
  - 9 infants with serious non-cardiac conditions

Acknowledgements

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CCHD Screening information:
New Jersey Department of Health
http://www.state.nj.us/health/fhs/nbs/cchd.shtml

American Academy of Pediatrics

New Jersey Chapter, American Academy of Pediatrics