CCHD Screening with Pulse Oximetry: A Success Story!

Nicole Spillane, MD
Associate Director of Neonatology, HackensackUMC
Hackensack Meridian Health
Objective

Recognize the contribution of pulse oximetry screening to the early detection of CCHD.
Definitions

Critical congenital heart disease

- Life threatening form of congenital heart disease that requires intervention in infancy.
Prenatal and Birth History

• Male infant born at 39 weeks 6 days gestation to a 32yo G1P0 woman.
• Pregnancy complicated by polyhydramnios.
• Maternal serology's: Blood type Opos, antibody neg, GBS neg, HepBSAg neg, HIV neg, RPR NR, and rubella immune
• Precipitous NSVD, ROM 5 hours of clear fluid, APGARS 9 & 9 at 1 and 5 min with routine resuscitation in the delivery room.
Hospital Course

• BW 3.68kg (7lbs 15oz); transitional respiratory distress that resolved shortly after birth.
• Roomed in with mother, breast feeding; voiding and stooling
• At 24 hours of age, routine CCHD screening performed with pulse oximetry in right foot of 91%, and 90-94% in right hand.
Hospital Course

- VS: HR 140’s; RR 60’s, pre/post ductal saturations now to mid 80’s, NC flow started without improvement.
- Infant transferred to NICU; placed on HFNC 3lpm FiO2 0.4, oxygen saturations 91-94%.
- FiO2 increased to 100% and saturations improved to 97%
- Electrolytes, CBC/diff, blood culture drawn
- Infant continued on ad lib feedings
- DOL 2: pre/post saturations in mid 90’s; RR 50-70; CXR: clear
- No obvious etiology for tachypnea or respiratory support
- Blood culture neg and screening serial CBC/diff reassuring
Hospital Course

- Echo: supra-diaphragmatic TAPVR- unobstructed & ASD –not ductal dependent
- Continued on supplemental O2 to maintain saturations in the 90’s.
- Transfer to cardiac center arranged.
Hospital Course

• DOL 3: Transferred to quatinary center NICU for Cardiac surgery
• DOL 6 underwent Repair of TAPVR, closure of ASD and PDA ligation.
• DOL 11 Discharged home on full ad lib feedings.
Why Screen for Critical Congenital Heart Defects?

- Congenital Heart Defects
  - 8-9/1,000 live births
  - Minimum of 32,000 - 40,000 infants affected each year in US
  - CHD is the leading cause of birth defect-associated infant illness and death.

- Critical Congenital Heart Defects (CCHD)
  - Approximately 25% of CHD or about 2 in 1,000 live births
  - About 8,000-10,000 infants born in the US every year

- Infants with undetected CCHD are at risk for death or significant disability

- Early diagnosis affords optimal treatment and better outcomes
New Jersey’s CCHD Screening Legislation

• NJ first state to implement a mandate for CCHD screening

• Legislation signed into law June 2, 2011

  Each birthing facility licensed by the Department of Health and Senior Services to perform a pulse oximetry screening for congenital birth defects (CHDs), a minimum of 24 hours after birth, on every newborn in its care.

  P.L.2011, CHAPTER 74, approved June 2, 2011
  Assembly, No. 3744

• Screening began August 31, 2011
AAP CCHD Screening Map: States’ Actions During 2016
Dylan’s Story

First-in-the-Nation New Jersey Newborn Heart Defect Screening Law Already Saving Lives

Wednesday, November 9, 2011 • Tags: Other

Governor Christie Travels to Sussex County to Meet Baby Dylan Who was Diagnosed and Treated as a Result of Landmark Law

“It is because of your law that our son’s life was saved, and my husband and I are very grateful to you...”

Letter to Governor Christie from the family of Dylan Gordon
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)
• Estimated 875 US infants annually will be detected with newborn CCHD screening (Ailes, et al, 2015)
• 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 hypoxemic 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
  - Hypoxic-ischemic encephalopathy

Mahle et al., 2009.
Antenatal Detection
Detection of CCHD

• Prenatal Ultrasound
  • Typically provides a four-chamber view of the heart
  • Quality varies, only half of all cases of CCHD are detected using this method
  • Less than 30% detection of lesions with two-ventricle anatomy
Physical Exam
Not As Pink As You Think...

• Some babies with CCHD can initially appear healthy
  • Not all present with murmurs or cyanosis
The Cyanotic “Blind Spot”

Hokanson, 2010.
The Cyanotic “Blind Spot”

• Cyanosis is detected when 3-5g/dl of deoxygenated hemoglobin is present
• Lets do some math...
The Cyanotic “Blind Spot”

• Hemoglobin of 18.5g/dl with saturation of 80%.
• 18.5g/dl x .80 = 14.8g/dl of hemoglobin is oxygenated and 18.5 - 14.8 = 3.7g/dl of deoxygenated hemoglobin is present
The Cyanotic “Blind Spot”

• How about the baby with Hemoglobin of 13.5 with saturation of 80%.

• $13.5\text{g/dl} \times 0.80 = 10.8\text{g/dl}$ of oxygenated hemoglobin. $13.5\text{g/dl} - 10.8\text{g/dl} = 2.7\text{g/dl}$ of deoxygenated hemoglobin.
The Cyanotic “Blind Spot”

Hokanson, 2010.
Detection of CCHD

• Physical Exam
  • Findings associated with CCHD may not be present before discharge
  • Exam results vary depending on timing, setting and clinician’s expertise
  • Low sensitivity (46%) but good specificity (99.8%)
  • Positive predictive value (PPV) around 60%
Detection of CCHD

<p>| | |</p>
<table>
<thead>
<tr>
<th></th>
<th></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>

<table>
<thead>
<tr>
<th></th>
<th>diagnostic gap</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>POS</td>
</tr>
<tr>
<td></td>
<td>clinical</td>
</tr>
<tr>
<td></td>
<td>prenatal</td>
</tr>
</tbody>
</table>

Detection of CCHD:
- **60%** Prenatal
- **20%** Clinical
- **15.6%** Pulse Ox
- **4.4%** Diagnostic Gap

Diagnostic Gap
CCHD Screening with Pulse Oximetry

- Indirectly monitors the oxygen saturation of the blood and variations in blood flow in the skin
- Can detect mild hypoxemia without apparent cyanosis
- Can provide continuous values
- Non-invasive
- Easy to use and widely available
- Cost-effective and extensively used
- Meets requirements for good screening test
Screening Cost

- CDC Study in 7 NJ Birthing Hospitals
  - Mean screening time per newborn was **9.1 min**
  - Total mean estimated cost per newborn screened was **$14.19**

Peterson et al., 2014.
Screening Cost per Infant

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

Excludes follow-up costs, such as further diagnostic testing, and administrative overhead.
Pulse Oximetry Screening Acceptance

- Over 90% of participants in study on perceptions of pulse oximetry screening supported CCHD screening.

Kumar et al, 2017
CCHD Screening 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
CCHD Screening Targets

• Can be just as severe but not detected as consistently:
  • Coarctation of the aorta
  • Double outlet right ventricle
  • Ebstein anomaly
  • Interrupted aortic arch
  • Single ventricle
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%
- 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 (0.05% vs 0.5%)

Thangaratinam et al., 2012.
Performing the Screen

- Right hand for pre-ductal reading
- Either foot for post-ductal reading
- Assure the sensor application site is clean and dry
- Use proper sensor for the device
- Screen while infant is awake and quiet
- Avoid screening when infant is crying, cold or in a deep sleep
- Have parent hold infant to calm
- Protect sensor from extraneous light
- Wait for steady wave or stable display on pulse oximeter
Screening Algorithm for Critical Congenital Heart Disease
Recommendations from the New Jersey Department of Health

All babies 24-48 hours of age or shortly before discharge if < 24 hours*

Perform and document pulse oximetry in both RIGHT HAND and either FOOT.

Is Pulse Oximetry reading < 90% in either the HAND or FOOT?

NO

Are both HAND and FOOT 95-100% ?

YES

Is the difference between the two measurements 3 or less?

YES

PASS

FAIL

Do not rescreen.

FAIL

Repeat the above pulse oximetry screening algorithm in one hour by obtaining new measurements from both right hand and either foot. If baby does not pass after a total of three screenings (initial screen and 2 repeat screens), notify responsible medical practitioner and follow recommendations in box below.

Notify responsible medical practitioner of the failed screen and refer for further evaluation.

Evaluate for other causes of low oxygen saturation (e.g., persistent pulmonary hypertension, pneumonia, infection, etc.).

In the absence of a clear cause of hypoxemia, obtain a diagnostic echocardiogram by an expert in the interpretation of fetal echocardiograms and review the report prior to discharge home. This may require transfer to another institution or use of telemedicine.

If saturation is < 90% in either the hand or foot, the baby should have immediate clinical assessment and immediate referral to pediatric cardiology. In this case, do not wait and rescreen.

A pass on the screen does not exclude the existence of a cardiac disorder.

If cardiac evaluation is otherwise indicated (e.g., clinical signs, prenatal diagnosis of critical congenital heart disease, dysmorphic features, etc.), proceed with cardiac evaluation even if baby receives a pass on the pulse oximetry screen.

Optimal results are obtained by using a motion-tolerant pulse oximetry that reports functional oxygen saturation, has been validated in low perfusion conditions, has been cleared by the FDA for use in newborns, and has a 2% root-mean-square accuracy.

Document results in medical record.

Screen in the right hand and one foot, either in parallel or direct sequence.

Apply probe to lateral aspect of right hand and foot in areas that are clean and dry. The two sensors (light emitter and detector) should be placed directly opposite each other.

Administration of supplemental oxygen may alter the interpretation of the screening result. For infants requiring supplemental oxygen, delay this screening algorithm until infant is stable in room air. For infants being discharged home on supplemental oxygen, perform screen prior to discharge and review results with responsible medical practitioner.

Symptomatic babies require clinical evaluation.

This screening algorithm should not take the place of clinical judgment or customary clinical practice.

* In the NICU, screening should be performed at 24-48 hours of age or as soon as medically appropriate after 24 hours of age. Screening must be performed prior to transfer out of the hospital at ≥ 24 hours of age. In all cases, screening should be performed prior to discharge to home.

Adapted from the Secretary's Advisory Committee on Heritable Disease in Infants and Children (SAAC-HDI) Expert Panel: Guidelines for Prenatal Recommendations, Jan 2011. C-9356
# NJ Recommended Algorithm: Abridged Version

<table>
<thead>
<tr>
<th>PASS</th>
<th>95%-100% in both extremities AND a difference of 3% or less between the readings.</th>
</tr>
</thead>
<tbody>
<tr>
<td>RESCREEN IN 1 HOUR</td>
<td>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.</td>
</tr>
<tr>
<td>FAIL</td>
<td>90%-94% in either extremity OR a difference of 4% or more between the readings after three screens.</td>
</tr>
<tr>
<td>IMMEDIATE FAIL</td>
<td>A reading of 89% or less in either extremity.</td>
</tr>
</tbody>
</table>
Evaluation for Failed Screen

- Any signs or symptoms of congenital heart defect should prompt rapid evaluation, including potential urgent transfer to a center with advanced care capabilities.
- Exclude infectious or pulmonary pathology
- Echocardiogram by provider with pediatric echo training
- Pediatric cardiology referral
Parent Education

http://nj.gov/health/fhs/nbs/cchd_resources.shtml

Available in English, Spanish, Arabic, Hindi, Korean and Polish.
Communication of Screening Results

- Include screening results in discharge summary
- Include in the hand-off report to the receiving hospital if infant was transferred
New Jersey Pulse Ox Screening Results
New Jersey Data Reporting

• Pulse ox screening results for all newborns entered into the electronic birth record.
• Birthing facilities are required to report all failed pulse ox screens 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.
Failed Screens Registered to NJ BDR

Total failures
N=332

At least 1 of 3 pre-identified factors*
N=187 (56.3%)

None of 3 pre-identified factors
N=145 (43.7%)

CCHD
N=26

CHD
N=21

Other significant medical conditions
N=13

*Factors include:
1. Prenatal diagnosis of CHD,
2. Signs or symptoms at the time of the screen,
3. Cardiac consult or echocardiogram prior to the screen
26 Infants with CCHD Were Detected

- Total anomalous pulmonary venous return (10)
- Coarctation of the aorta (7)
- d-Transposition of the great arteries (3)
- TGA and double outlet right ventricle (2)
- Ebstein anomaly (1)
- Interrupted aortic arch (1)
- Tetrology of Fallot (1)
- Tricuspid atresia (1)
Other Conditions Detected

• **21 CHDs:**
  • Atrial septal aneurysm
  • ASD
  • VSD
  • Peripheral pulmonary artery stenosis
  • Prolonged QT

• **13 Other significant non-cardiac medical conditions:**
  • Sepsis
  • Pneumonia
  • Persistent pulmonary hypertension
  • Pulmonary bulla
### Screen results: CCHD

<table>
<thead>
<tr>
<th>CASE</th>
<th>PRE-DUCTAL</th>
<th>POST-DUCTAL</th>
<th>FINAL DIAGNOSIS</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>43</td>
<td>39</td>
<td>d-TGA</td>
</tr>
<tr>
<td>2</td>
<td>59</td>
<td>64</td>
<td>d-TGA</td>
</tr>
<tr>
<td>3</td>
<td>60</td>
<td>70</td>
<td>d-TGA</td>
</tr>
<tr>
<td>4</td>
<td>85</td>
<td>85</td>
<td>DORV, d-TGA, pulmonary atresia, VSD</td>
</tr>
<tr>
<td>5</td>
<td>81</td>
<td>81</td>
<td>DORV, d-TGA</td>
</tr>
<tr>
<td>6</td>
<td>87</td>
<td>84</td>
<td>TAPVR</td>
</tr>
<tr>
<td>7</td>
<td>92</td>
<td>93</td>
<td>TAPVR</td>
</tr>
<tr>
<td>8</td>
<td>75</td>
<td>75</td>
<td>TAPVR</td>
</tr>
<tr>
<td>9</td>
<td>89</td>
<td>93</td>
<td>TAPVR</td>
</tr>
<tr>
<td>10</td>
<td>85</td>
<td>88</td>
<td>TAPVR</td>
</tr>
<tr>
<td>11</td>
<td>77</td>
<td>78</td>
<td>TAPVR</td>
</tr>
<tr>
<td>12</td>
<td>91</td>
<td>92</td>
<td>TAPVR</td>
</tr>
<tr>
<td>13</td>
<td>82</td>
<td>85</td>
<td>TAPVR</td>
</tr>
<tr>
<td>14</td>
<td>71</td>
<td>80</td>
<td>TAPVR</td>
</tr>
<tr>
<td>15</td>
<td>88</td>
<td>92</td>
<td>TAPVR</td>
</tr>
</tbody>
</table>
## Screen results: CCHD

<table>
<thead>
<tr>
<th>CASE</th>
<th>PRE-DUCTAL</th>
<th>POST-DUCTAL</th>
<th>FINAL DIAGNOSIS</th>
</tr>
</thead>
<tbody>
<tr>
<td>16</td>
<td>89</td>
<td>88</td>
<td>TETRALOGY OF FALLOT</td>
</tr>
<tr>
<td>17</td>
<td>91</td>
<td>92</td>
<td>TRICUSPID ATRESIA</td>
</tr>
<tr>
<td>18</td>
<td>97</td>
<td>84</td>
<td>COARCTATION OF AORTA</td>
</tr>
<tr>
<td>19</td>
<td>94</td>
<td>86</td>
<td>COARCTATION OF AORTA</td>
</tr>
<tr>
<td>20</td>
<td>99</td>
<td>88</td>
<td>COARCTATION OF AORTA Out of state resident</td>
</tr>
<tr>
<td>21</td>
<td>99</td>
<td>90</td>
<td>COARCTATION OF AORTA</td>
</tr>
<tr>
<td>22</td>
<td>92</td>
<td>97</td>
<td>COARCTATION OF AORTA- NICU</td>
</tr>
<tr>
<td>23</td>
<td>99</td>
<td>68</td>
<td>COARCTATION OF AORTA</td>
</tr>
<tr>
<td>24</td>
<td>99</td>
<td>80</td>
<td>COARCTATION OF AORTA</td>
</tr>
<tr>
<td>25</td>
<td>96</td>
<td>91</td>
<td>INTERRUPTED AORTIC ARCH</td>
</tr>
<tr>
<td>26</td>
<td>95</td>
<td>92</td>
<td>EBSTEIN ANOMALY-Passed national protocol</td>
</tr>
</tbody>
</table>
We’re just the beginning.......