Failure to detect Critical Congenital Heart Disease (CCHD) in newborns

Executive Summary Checklist

Congenital Heart Disease (CHD) is one of the most common types of birth defects. Critical Congenital Heart Disease (CCHD), including ductal-dependent lesions, represents 40% of death caused by CHD. CCHD is life threatening and typically takes place during the first year of infancy.

☐ Develop an action plan to immediately implement a universal pulse oximetry screening program.
  ○ Select technology proven to be effective for newborn screening. The technology must monitor and accurately read through during motion and low perfusion.
    ▪ e.g. Masimo SET pulse oximetry (until another technology is proven to be equivalent)
  ○ Determine the screening protocol
    ■ Age at screening: >24 hours or prior to discharge
    ■ Obtain pulse oximetry measurements from preductal (right hand) and postductal (either foot) sites
    ■ Screening results which will be considered positive and require further investigation
      ➢ \( \text{SpO}_2 \) <90% from any site; or
      ➢ \( \text{SpO}_2 \) <95% from the right hand or either foot
        ○ If initial \( \text{SpO}_2 \) measurement is <95%, proceed with up to two additional \( \text{SpO}_2 \) measurements.
        ○ If the second and third \( \text{SpO}_2 \) measurements read >95% the screening is negative.
        ○ If the second and third \( \text{SpO}_2 \) measurements are <95% the screening is positive.
        ➢ >3% difference in \( \text{SpO}_2 \) measurements between the right hand and either foot (repeat three times as described in the bullet above)
  ○ Additionally, if the Perfusion Index (PI) <0.7 that should increase the need for assessment of the baby (if <0.4 the baby should be immediately assessed)
  ○ Educate clinical staff on proper screening, strategies for family education and engagement, follow-up protocols for positive screens, and results reporting policy

☐ Develop a process for continuous improvement by educating and communicating with staff and implementing measures to improve processes in order to meet the universal newborn screening objective.
The Performance Gap

Congenital heart disease (CHD) is the most common birth defect, affecting approximately 8 in 1,000 live-born infants (Reller et al., 2008; Bernier et al., 2010). Nearly 40,000 infants are born with CHD per year in the US, and 1.35 million globally (Hoffman and Kaplan, 2002; van et al., 2011). Critical congenital heart disease (CCHD), including ductal dependent lesions, affects between one-quarter and one-third of these infants (Oster et al., 2013; Glidewell et al., 2015; Ailes et al., 2015). CCHD represents about 40% of the deaths from congenital anomalies and the majority of the deaths due to CHD that occur in the first year of life. (Hoffman 2002). In 2012, before newborn screening programs were introduced in the United States, it was estimated that between 70-100 infants died each year from late-diagnosed CCHD (Govindaswami, Jegatheesan and Song, 2012). It is now believed that the number of deaths is closer to 120 per year (Grosse et al., 2017).

Antenatal ultrasound and physician examination after birth improve detection and perinatal outcomes for certain forms of CCHD (Tworetzky et al., 2001; Bonnet et al., 1999). Evidence showed that prenatal detection increased every year (2006-2012); prenatal detection now occurs in 34% of patients (Quartermain et al., 2015). The benefit of a CCHD diagnosis before birth allows for counseling and coordination of delivery at an experienced cardiac center.

The gap in patient safety is that more than 30 percent of CCHD deaths have been attributed to late or missed diagnosis (Chang, Gurvitz and Rodriguez, 2008). It is estimated that 2,000 infants/year die or are undiagnosed in the US and some 300,000 infants/year die globally (Salvi, 2016). The burden of undiagnosed cases in the developing world is significant, with fewer than half of CHD cases diagnosed in the first week of life (Hoffman, 2013). The magnitude of the problem has been extensively documented (Singh et al., 2014; de-Wahl Granelli et al., 2014; Ewer, 2014; Ewer, 2014; Ewer, 2013; Ewer, 2013; Granelli et al., 2007).

Pulse oximetry noninvasively measures oxygen saturation (SpO₂) and pulse rate. In 2009, de-Wahl Granelli et al published a breakthrough cohort study in which 39,821 infants were screened for CCHD by identifying abnormal SpO₂ measurements from Signal Extraction Technology (SET) pulse oximetry. SET’s ability to measure through motion and low-perfusion is essential for accurate CCHD screening (de-Wahl Granelli et al., 2009). In a separate CCHD screening study of 20,055 asymptomatic newborns, Ewer et al, confirmed the importance of utilizing SET technology that can “produce accurate saturations that are stable in active neonates and in low perfusion states, making them suitable for use in the first few hours of a newborn baby’s life” (Ewer et al., 2012). In 2014, Zhao et al reported similarly positive results from a prospective study using SET in more than 100,000 newborns in China (Zhao et al., 2014).

The addition of pulse oximetry screening to antenatal ultrasound and physical examination may increase detection rates for CCHD to over 90%. Furthermore, the detection of non-critical CHDs and significant non-cardiac neonatal conditions, such as respiratory problems or early-onset sepsis, is an additional benefit. However, clinicians need to be aware that, although combining pulse oximetry screening with other screening methods will reduce this diagnostic gap, some babies will still be missed. The Journal of Pediatrics has published a study estimating the number of infants with CCHDs potentially detected or missed through universal screening for critical CHDs using pulse oximetry (Frank et al., 2013). CDC researchers estimated that about 1,755 infants with critical CHDs would be diagnosed late (meaning on or after the third day after birth). Of these, about half (875 infants) with a critical CHD would be detected through newborn screening using pulse oximetry, but an equal number (880 infants) might still be missed each year in the United States.

Most studies report that the lesions most often missed are those causing obstruction to aortic outflow (e.g. coarctation and interrupted arch), which may not necessarily be detected in antenatal ultrasound, physical examination, or by abnormal SpO₂ values from pulse oximetry. However, an additional SET pulse oximetry measurement may increase detection of CCHD with obstructions to aortic outflow. This measurement is called perfusion index (PI), which is an assessment of strength of perfusion at the monitored site. In a 2007 study, Granelli showed that adding abnormal PI to pulse oximetry screening may increase sensitivity to identifying CCHD with an obstruction to the aortic outflow. The authors of this study also noted that adding PI to the screening criteria may also result in an increase in false positives (Granelli, 2007).

In 2011, the federal CCHD workgroup, with members selected by the US Health and Human Services Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children, the American Academy of Pediatrics, the American College of Cardiology Foundation, the Newborn Foundation, the March of Dimes, and the American Heart Association, developed a report: Strategies for Implementing Screening for Critical Congenital Heart Disease (Kemper et al., 2011). After a thorough review, the workgroup relied upon a thorough body of evidence and independent published studies to recommend that “screening be performed with motion tolerant pulse oximeters that report functional oxygen saturation, have been
validated in low-perfusion conditions, have been cleared by the FDA for use in newborns, and have a 2% root mean-square accuracy.”

Several domestic and international studies have shown parents are predominantly satisfied with pulse oximetry screening and those whose babies had a false positive result were no more anxious than those with true negative tests (Ewer 2012). Parents generally perceived it as an important and valuable test to detect ill babies. Additionally, all staff groups (healthcare assistants, midwives, nurses and doctors) were predominantly positive about the testing procedure and perceived the test as important.

Screening for CCHD not only reduces pain and suffering of infants and families but can also reduce costs associated with severe cardiovascular and other organ or neurological compromise upon delayed admission to a cardiac unit – and has been tied to significantly reduced mortality, fewer poor surgical outcomes, and lower incidence of prolonged ventilation and potential developmental issues (Peterson et al., 2013).

Relative to the developing world, the prevalence of certain heart lesions varies significantly on the global map, as does the burden of hypoxemia-related conditions such as neonatal pneumonia, sepsis, necrotizing enterocolitis (NEC), and PPHN (Hoffman 2013). Every year nearly 41% of all under-five child deaths are among newborn infants, babies in their first 28 days of life or the neonatal period (WHO, 2012). Three-quarters of all newborn deaths occur in the first week of life, and 1/3 of these newborn deaths are from infection, such as pneumonia, tetanus, and sepsis. Each of these conditions are likely to manifest with below normal oxygen saturation. Some are preventable deaths in that when diagnosed in a timely fashion, a course of antibiotics and/or supplemental oxygen therapy can save a life or improve an outcome.

Considerations regarding algorithms for screening

A recent review describes the experience of CCHD screening in the United States in reference to optimizing the algorithm for screening, educating all stakeholders and performing screening using the proper equipment (Oster et al., 2016). There are many factors to consider when determining the optimal screening algorithm, including the balance of sensitivity and specificity, resource utilization, cost, high altitude and timing of screening. For this reason, other screening protocols have been evaluated in the United States and in other countries (Ewer and Martin, 2016; Ewer, 2016). For this reason, other screening protocols have been evaluated in the United States and in other countries. For example, infants at high altitude may have a lower oxygen saturation than those at sea level with potential implications at elevations over 6,800 feet. Therefore, to identify the optimal algorithm in particular settings, it may be necessary to modify the screening protocol described in this document, including the saturation cutoff points and the timing of screening.

A certain degree of controversy still remains, and debate continues regarding the most appropriate time to screen, the most effective screening pathway, what saturations are acceptable, which conditions are we trying to identify and screening outside the well-baby nursery.

When evaluating algorithms, it is important to consider sensitivity, specificity, and false-positive and false-negative rates. It is also vital that screening leads to timely diagnosis (ie, before presentation with acute collapse).

- The screening should be pre-and post-ductal as analysis of raw saturation data from infants who had both limb measurements shows that some infants with CCHD would be missed by post ductal testing alone.
- False-positive rate is significantly higher with earlier testing (<24 hours). This led to recommendations that screening be performed after 24 hours of age.
- However, analysis of recent studies show that many false-positive tests (30%–80%) have alternative non-cardiac conditions (eg, congenital pneumonia, early-onset sepsis, or pulmonary hypertension), which may be equally as life threatening as CCHD if diagnosed late.
- In published studies that adopted earlier screening (< 24 hours) the false-positive rate was higher, but more non-cardiac disease was identified.
- In some countries, mothers and infants are discharged from the hospital within 24 hours after birth, and an increasing proportion is born at home. In these circumstances, screening in hospital > 24 hours is not practical.
- Additionally, infants at high altitude may have a lower oxygen saturation than those at sea level with potential implications for screening for CCHD at elevations over 6,800 feet. Therefore, to identify the optimal algorithm in particular settings, it may be necessary to modify the screening protocol described in this document, including the saturation cutoff points and the timing of screening.
Although usually reserved for former premature infants going to high altitude, any infant who fails high altitude stress testing (HAST) also merits special consideration and may require an echocardiogram to confirm normal anatomy.

Be all this as it may, if SpO\textsubscript{2} is < 90% in either limb the infant needs to be assessed immediately. If SpO\textsubscript{2} is between 90-94% in one or both limbs and the infant does not look completely healthy, clinical assessment is mandatory without delays for repeated measurements. If infant is completely healthy, measurement should be repeated as described.

In summary, the lack of a systematic approach to prevent failure to rescue in CCHD significantly affects patient safety, quality, and cost of care. Universal newborn screening with pulse oximetry technology has been shown to increase the detection of CCHD by identifying potential abnormalities that are not apparent in prenatal or postnatal examinations. Closing the performance gap with CCHD will require hospitals, healthcare systems and all members of the neonatal healthcare team (RN’s, RT’s and MD’s) to commit to action in the form of specific leadership, practice, and technology plans for all newborn infants.

Leadership Plan

- Implement a plan that includes fundamentals of change outlined in the National Quality Forum safe practices, including awareness, accountability, and action.
- Hospital governance and senior administrative and medical and nursing leadership commit to becoming aware of this major performance gap in their own healthcare system.
- Hospital governance, senior administrative leadership, and clinical/safety leadership close their own performance gap by implementing a comprehensive approach to addressing the performance gap
- Set a goal date to implement the plan to address the gap with measurable quality indicators.
- Allocate a budget for the plan to be evaluated by governance boards and senior administrative leaders.
- Clinical/safety leadership endorse the plan and drive implementation across all providers and systems.
- Conduct data collection and analysis to be used for implementation and assessment of outcomes.

Practice Plan

- Evaluate guidelines and reviews and choose a screening strategy that, when implemented, is in compliance with processes described and technology used in well-designed, large published studies.
- Develop an action plan with a timeline with concrete milestones to implement universal newborn screening.
  - Select technology proven to be effective for newborn screening
  - Use SET pulse oximetry screening strategy
  - Determine the screening protocol
    - Age at screening: >24 hours or prior to discharge
    - Obtain pulse oximetry measurements from preductal (right hand) and postductal (either foot) sites
    - Screening results which will be considered positive and require further investigation
      - SpO\textsubscript{2} <90% from any site; or
      - SpO\textsubscript{2} <95% from the right hand or either foot
        - If initial SpO\textsubscript{2} measurement is <95%, proceed with up to two additional SpO\textsubscript{2} measurements.
        - If the second and third SpO\textsubscript{2} measurements read >95% the screening is negative.
        - If the second and third SpO\textsubscript{2} measurements are <95% the screening is positive.
          - >3% difference in SpO\textsubscript{2} measurements between the right hand and either foot (repeat three times as described in the bullet above)
        - Additionally, if Perfusion Index (PI) <0.7 that should increase the need for assessment of the baby (if <0.4 the baby should be immediately assessed)
  - Implement interdisciplinary strategies and educational activities for all members of the neonatal healthcare team
    - Proper screening methods
    - Strategies for family education and engagement
    - Follow-up investigation protocols for positive screens
    - Public health results reporting policy
  - Implement optimization and workflow guidelines to ensure performance of adequate screening.
○ As a quality indicator, each week randomly assess the number of babies that have should have been screened but were not. Communicate with staff and, based on results, implement measures to improve processes in order to meet the goal of screening all newborns. Utilize clinical decision support tools and software whenever available to avoid misinterpretation or screening results or faulty data entry
  
  ● Report screening results per state and federal requirements

Technology Plan

*Suggested practices and technologies are limited to those proven to show benefit or are the only known technologies with a particular capability. As other options may exist, please send information on any additional technologies, along with appropriate evidence, to info@patientsafetymovement.org*

  ● Select pulse oximetry technologies have proven to be effective in helping clinicians screen for CCHD.
    ○ SET measure-through motion and low perfusion pulse oximetry (until another technology is proven to be equivalent)
    ○ SET pulse oximetry is available in:
      ■ Standalone monitors (Rad-5, Rad-57, Radical-7, Rad-87)
      ■ Integrated in over 100 devices from over 50 companies including Atom, Drager, Fukuda Denshi, GE, Mindray, Nihon Koden, Philips, Spacelabs, and Welch Allyn.
  
  ● Consider utilizing a device that reduces operator induced-variability and improves efficiency by automating the screening steps, measurement selection, application of the measurements to the screening criteria chosen by the hospital, and categorization of the test as a positive or negative screen
    ○ Eve® app on the Radical-7 (note this device CE Marked but has not received U.S. FDA 510k)
  
  ● Consider utilizing public health reporting systems for newborn screening
    ○ Such as Oz® Systems newborn screening or automated reporting with Oz® BabyBundle

Metrics

Topic:

**Critical Congenital Heart Defects (CCHD)** is the number of patients identified with CCHD through technology-enabled pulse oximetry newborn screening. The rate is the reflection of the number of patients diagnosed with CCHD over the total number of infants screened.

**Outcome Measure Formula:**

Numerator: Number of newborns identified with CCHD  
Denominator: Number of patients screened  
*Measure typically displayed as a percentage: Numerator/Denominator *100

Metric Recommendations:

**Indirect Impact:**

All newborns that received technology-enabled newborn screening of CCHD via pulse oximetry

**Direct Impact:**

Number of asymptomatic infants identified with CCHD through pulse oximetry and received successful clinical intervention.

**Lives Spared Harm:**

Number of asymptomatic infants identified with CCHD through pulse oximetry or echocardiogram and received successful clinical intervention.

Years of Potential Life Saved
Those infants saved multiplied by average life expectancy.

**Live Saved:**

\[ \text{Lives Saved} = \text{Lives Spared Harm} \times 0.825 \]

**Data Collection:**

Both the numerator and denominator data could be collected from the medical record.

**Mortality (will be calculated by the Patient Safety Movement Foundation):**

Prenatal detection of CCHD has been shown to improve surgical outcome reducing neonatal morbidity and mortality, supporting findings of neonatal studies that early detection has impact on the clinical results and morbidity and mortality rates. The Patient Safety Movement Foundation will use the mortality rates associated with the findings published in 2013 by Oster. The Oster study found that the one-year survival of newborns screened by pulse oximetry after 24 hours of age was 82.5%. Based on these data the mortality rate of 17.5% will be used.
Workgroup

Chair:
*Augusto Sola (Masimo)

Members:
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Mitchell Goldstein, MD (Loma Linda University)
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Conflicts of Interest Disclosure

The Patient Safety Movement Foundation partners with as many stakeholders as possible to focus on how to address patient safety challenges. The recommendations in the APSS are developed by workgroups that may include patient safety experts, healthcare technology professionals, hospital leaders, patient advocates, and medical technology industry volunteers. Some of the APSS recommend technologies offered by companies involved in the Patient Safety Movement Foundation that the workgroups have concluded, based on available evidence, are beneficial in addressing the patient safety issues addressed in the APSS. Workgroup members are required to disclose any potential conflicts of interest.

*This Workgroup member has reported a financial interest in an organization that provides a medical product or technology recommended in the Technology Plan for this APSS.

References


