Session 9 – Point of Care for Critical Congenital Heart Disease (CCHD)

Wednesday, Oct. 29 – 4:00pm-5:30pm

Moderators – Julie Luedtke, BS, Nebraska Department of Health and Human Services and Brad Therrell, PhD, National Newborn Screening & Genetics Resource Center

Implementing Universal Pulse Oximetry Screening: From Pilot to Policy to Practice
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Abstract

When Critical Congenital Heart Disease (CCHD) was approved by Secretary Sebelius for addition to the Recommended Universal Screening Panel in 2011, state newborn screening programs struggled with the implementation of this unique screen. Though not the first point-of-care screen to be added to the newborn screening panel, the differences between CCHD screening and newborn hearing screening (EHDI) are vast; the timelines for follow-up, the potential for transfers of sick infants, and the lack of pediatric cardiology resources in many areas. Compounding these differences is the fact that most state EHDI programs are run separately from state newborn screening programs, so lessons learned from EHDI are often not available to screening programs implementing CCHD.

We will present Minnesota’s CCHD screening implementation planning. Key steps included: conducting a pilot study in a large health system; CCHD specific legislation; and full-state implementation. Utilizing this particular series of steps has allowed the State to control the pace of implementation and ensure preparedness by birth facilities and attendants.

We will discuss:
• Pilot project outcomes
• Expert workgroup decisions on utilizing a different protocol than the one recommended by the AAP
• Hospital survey outcomes and development of educational materials and training
• Electronic result reporting implementation and ongoing data collection (inclusive of first few months of data)
• Coordination with long term follow-up and birth defects registry
• The current status of CCHD screening implementation in Minnesota

The addition and implementation of new newborn screening conditions in different states often follow very different paths. Because different states face different challenges, and may enjoy different opportunities, sharing state experiences in implementation of new disorders is critical to improving the process nationwide and developing best practices.
CCHD Screening in Maryland - Year 1 Results
D. Badawi and J. Watson, Office for Genetics and People with Special Health Care Needs, Baltimore, MD

Abstract

Maryland’s newborn screening law, Health General Article 13-111, was amended in 2011 to mandate formation of an expert panel to produce a legislative report on the feasibility of implementing newborn screening for CCHD in Maryland. The law also stated that if CCHD screening was added to the national Recommended Uniform Screening Panel (RUSP), Maryland would begin screening. This presentation will outline the process of evaluation and implementation of CCHD screening in the state, and provide outcomes from the first year of the program. The legislative expert panel members formed an advisory committee to oversee establishment of newborn screening for Critical Congenital Heart Disease (CCHD) and mandated statewide screening began in Maryland on September 1, 2012. From September 1, 2012 to August 31, 2013, there were 69,609 births documented in the CCHD screening follow up system across 33 birth hospitals as of October 2013. The median screening rate as measured by documentation of screen results was 95.8%. There were 40 failed screens among these babies, and 4 of these infants were subsequently diagnosed with CCHD. Seven infants passed their pulse ox screen but were later diagnosed with CCHD. Newborn screening follow up programs, both blood spot and point of care, occur in the same office withing the Maryland Department of Health and Mental Hygiene. A web-based data collection system is used to monitor CCHD screening, and this system also includes birth defects reporting and infant hearing screening data. There have been several lessons learned that inform the continued evolution of the program. Logistically, the quality and comprehensiveness of data collection require ongoing quality improvement and education of nursery staff. Clinically, there is variation among hospitals in their approach to NICU babies, and questions remain regarding best practices. Regional and national data collection are needed to evaluate the issue of normal screens occurring in some infants with CCHD. The first year of CCHD screening in Maryland demonstrates the benefits of this screening, and provides an example of surveillance capabilities utilizing a database and an infrastructure that is integrated with other newborn screening programs.

Summary

On September 1, 2012 Maryland was the third state in the nation to begin mandated newborn screening for Critical Congenital Heart Disease (CCHD). The State Advisory Council on Hereditary and Congenital Disorders (the Council) convened an Expert Panel mandated by amendments to Health General Article 13-111 to evaluate the feasibility of implementing newborn screening for CCHD in Maryland. This panel consisted of newborn screening experts, pediatric cardiologists, neonatal nurses, neonatologists, and community advocates. Three subcommittees addressed the issues of Clinical Feasibility, Education, and
Quality Assurance. Once this screening was added to the RUSP, the Expert Panel focused on implementation and approved the CCHD screening algorithm that was endorsed by the AAP and the AHA.

A significant issue national issue in implementing CCHD screening has been to define the role of public health in surveillance for CCHD screening. For point of care screening, quality assurance cannot take place in real time, but can consist of monitoring performance of birth facilities and offering technical assistance where needed. Maryland has had an electronic system for reporting infant hearing screening results, and in September of 2012 was planning to add a module for birth defects reporting. CCHD screening documentation was added to this new module, to include whether or not a child was screened, and a pass/fail result. This reporting system was later upgraded to include the date and time of the screen as well space for the optional documentation of actual pulse oximetry readings.

Community stakeholders were notified of the implementation of screening. Hospital CEO’s received a letter and providers, including pediatricians, family practice physicians and OB/GYNs were notified by email blasts and newsletter articles. Nurses were contacted through the Maternal Child Health Directors group in Maryland as well as the Perinatal Collaborative in the state. Birth hospitals were asked to identify a contact person for CCHD screening and two live webinars were presented, with an archived version made available as well. A website was created with resources for families and providers (http://phpa.dhmh.maryland.gov/genetics/SitePages/CCHD_Program.aspx), including the algorithm for screening, background articles and a chart to indicate pass/fail pulse ox values. Screening was widely accepted by birth facilities, including midwives, however reporting of results presented some challenges. Reporting was limited and often delayed initially, with frequent calls and emails for technical assistance, either regarding the screening protocol or documentation. However, after the first quarter of implementation, the majority of hospitals were documenting a screening rate of over 90%.

From September 1, 2012 through December 31, 2013, there were 92,728 births documented in the CCHD screening follow up system. Of these infants, 9,347 were admitted to the Neonatal Intensive Care Unit (NICU), and 83,381 were screened in the Well Baby nursery. Results will be discussed separately for infants in the Well Baby nursery versus the NICU and then compared. For well babies, there were 74 infant deaths over this time period, and of the remaining infants, 94% had a documented CCHD screen. Twenty-eight infants failed their screen, and of these, 1 primary condition (tricuspid atresia) and 9 secondary conditions (ASD, PFO, Pulmonary artery stenosis, septal aneurysm, pneumonia) were identified. There were 11 infants who presented with primary or secondary cardiac conditions after passing their CCHD screen (Tetralogy of Fallot, TAPVR, coarctation, surgical VSD). This results in a false positive rate of 0.017% and a false negative rate of 80% for primary target conditions.

For infants in the NICU, there were 267 deaths over this time period, and of the remaining infants, 77% were screened. Twenty-two infants failed their CCHD screen, and of these, 13 were diagnosed with a primary (interrupted aortic arch, TAPVR) or secondary (PPHN, PFO, tricuspid regurgitation, AV canal) condition. There were 6 infants later diagnosed with a primary (Tetralogy of Fallot) or secondary (coarctation, pulmonary stenosis, VSD) cardiac condition after a negative CCHD screen. This results in a false positive rate of 0.24% and a false negative rate of 25% for NICU babies. It is worth noting that 148 infants in the NICU had clinical signs prompting an echocardiogram; 13 of these infants had a primary target condition identified, and 23 had secondary cardiac conditions identified.
Several lessons were learned from the first 16 months of CCHD screening in Maryland. First and foremost, diligent and dedicated staff is essential to the collection of follow up data on infants who fail their CCHD screen, and to ascertain children identified with CCHD after their birth hospitalization. While electronic documentation is an excellent tool for surveillance and quality assurance, it leaves the follow up to the state program. Physician override provides a way for nursery staff to indicate that an infant does not clinically require screening, for example due to prenatal diagnosis or clinical symptoms necessitating evaluation before screening. However, without further specification of the reason for physician override, this category was often used incorrectly. Anecdotally, most birth facilities are using the recommended AHA/AAP protocol, however we have not been able to confirm this, and do not have details of NICU screening protocols.

Overall, CCHD screening has successfully identified infants with both primary and secondary cardiac conditions. Given the high false negative rates, particularly for well babies with secondary cardiac conditions, parents and providers must be reminded that a normal CCHD screen does not rule out the possibility of congenital heart disease.

Exploring False Negative Pulse Oximetry Screens to Improve Critical Congenital Heart Disease Detection
L. Hom and G. Martin, Children’s National Health System, Washington, DC

Abstract

Background: Screening for Critical Congenital Heart Disease (CCHD) was endorsed by the United States Secretary of Health and Human Services as part of the recommended uniform screening panel for newborns in 2011. Stakeholder meetings led to a recommended protocol as well as a minimum data set for reporting. Accurate and complete reporting is considered essential to analysis and systematic improvement. Sensitivity in the largest European study using pulse oximetry alone for identifying CCHD was only 62%. Explanations for false negative studies (FNS) have not been explored. The purpose of this study was to examine screening results of infants and provide possible explanations for FNS.

Methods: Children’s National serves as a referral center for an area performing screening for CCHD. Newborn records of infants less than two months admitted for CCHD were reviewed for CCHD screening results and oxygen saturations in screening were compared with those obtained on admission at time of diagnosis. The CCHD target lesions included HLHS, TOF, PA/IVS, TAPVD, d-TGA, tricuspid atresia and truncus arteriosus. Secondary targets included coarctation of aorta and all other forms of CCHD as well as other infectious or pulmonary causes of low pulse ox readings.

Results: During the time period June, 2013 to October, 2013 we identified 5 false negative cases of CCHD. The newborns had undergone screening around 24 hours of age (n=4) or did not receive screening (n=1). There were no protocol violations in interpretation of the results of oxygen saturations. During this time period there were 2 true positive cases of CCHD identified through screening referred to Children’s National.

Conclusions: Screening for CCHD with the current protocol has false negatives. The most common cause appears to be anatomic and physiologic variables that result in normal saturations at the timing of screening in the majority of cases. Failure to screen was a factor in 1 case but protocol violations did not appear to be a significant problem in this limited data set. Further data on oxygen saturations in false
negatives may improve screening efforts or increase our knowledge of the transitional circulation of babies with CCHD.

**Presenter:** Lisa Hom, RN, ESQ, Children’s National Health System, Heart Institute/Cardiology, Washington, DC, Phone: 202.476.5063, Email: lhom@cnmc.org

**Summary**

**Background:**
Screening for Critical Congenital Heart Disease (CCHD) was endorsed by the United States Secretary of Health and Human Services as part of the recommended uniform screening panel for newborns in 2011. Stakeholder meetings led to a recommended protocol as well as a minimum data set for reporting. Accurate and complete reporting is considered essential to analysis and systematic improvement. Sensitivity in the largest European study using pulse oximetry alone for identifying CCHD was only 62%. When paired with physical exam, sensitivity increases to 87%. A significant number of types of cardiac congenital anomalies do not result in cyanosis and are therefore not detectable through pulse oximetry screening. An added benefit of pulse oximetry screening is that false positives for CCHD have a significant likelihood of identifying a non-cardiac issue such as pneumonia or sepsis in which there is significant value in detecting.

Improving the detection of false negatives in newborn screening is an important public health priority. Sub-specialty clinics working closely with public health departments can provide valuable insight into why false negatives are occurring. It is important to understand whether babies are missed due to biologic factors, process errors (including algorithm misinterpretations) or whether they were simply not screened. Explanations for CCHD false negative studies (FNS) have not yet been explored. The purpose of this study was to examine screening results of infants and provide possible explanations for FNS.

**Methods:**
Children’s National serves as a referral center for an area performing screening for CCHD and works closely with the state public health departments of Virginia, Maryland and D.C. in CCHD education and implementation. Newborn records of infants less than two months admitted for CCHD were reviewed for CCHD screening results and oxygen saturations in screening were compared with those obtained on admission at time of diagnosis. The CCHD target lesions included hypoplastic left heart syndrome (HLHS), tetralogy of Fallot (TOF), pulmonary atresia with intact septum (PA/IVS), total anomalous pulmonary venous drainage (TAPVD), transposition of the great arteries (d-TGA), tricuspid atresia and truncus arteriosus. Secondary targets included coarctation of aorta (CoA) and all other forms of CCHD as well as other infectious or pulmonary causes of low pulse oximetry readings.

**Results and Discussion:**
During the time period June, 2013 to October, 2013 we identified 5 FNS and two true positive cases of CCHD. Of the 5 FNS, 3 were primary targets, 2 cases of TOF and one case of TAPVD. The remaining 2 FNS consisted of CoA, a secondary target for CCHD screening and one of the most common types of left ventricular outflow tract defects. The two true positive cases of CCHD were HLHS and TAPVD, both identified through screening referred to Children’s National.
The newborns had undergone screening using the nationally endorsed 2 limb approach with screening occurring around 24 hours of age (n=6) or did not receive screening (n=1). There were no protocol violations in interpretation of the results of oxygen saturations.

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Conclusions:
Screening for CCHD with the current national protocol has false negatives. The most common cause appears to be anatomic and physiologic variables that result in normal saturations at the timing of screening. Failure to screen was a factor in 1 case but protocol violations did not appear to be a significant problem in this limited data set. Further data on oxygen saturations in false negatives may improve screening efforts or increase our knowledge of the transitional circulation of babies with CCHD. CCHD screening combined with pre-natal ultrasound and newborn assessment provides the most effective detection method. Further innovative research on peripheral perfusion index (PPI) and refinements in algorithm methodology may lead to effectively closing the remaining gap in CCHD screening.

Modification of Critical Congenital Heart Disease Screening Practices at Moderate Altitude
C. Rausch\(^1\), M. Kohn\(^2\), L. Russell\(^3\), J. Miller\(^3\), E. Leuth\(^1\); \(^1\)Children’s Hospital Colorado, Aurora, CO, \(^2\)University of Colorado Hospital, Aurora, CO, \(^3\)Colorado School of Public Health, Aurora, CO

Abstract

Introduction: Consensus guidelines have recommended pulse oximetry screening of newborns for critical congenital heart disease (CCHD). Previous studies at our center at moderate altitude (Aurora, CO; 1620 meters) have demonstrated feasibility of a sea level screening protocol, but with higher screening failure rates (~1% of all infants screen) when compared with sea level (~0.2%) centers. We therefore
modified the screening protocol in an attempt to reduce the number of screening failures in children with normal hearts born at moderate altitude.

**Methods:** CCHD NBS was implemented as standard of care in the well-baby nursery at a large academic hospital. Infants with known congenital heart disease or conditions known to predispose to hypoxia were excluded. Pulse oximetry screenings were performed in accordance with national recommendations with the exception that infants who failed the initial three step screening process were observed closely with repeat screening performed every four hours while waiting for echocardiography.

**Results:** 1968 infants were screened between 9/1/12 and 7/31/13 (females: 934, males 1032, unk: 2). Median gestational age was 39 weeks (IQR 38-40 weeks) and mean birthweight was 3200 grams (STD DEV 516 grams). In infants who failed initial screens, repeat screenings were performed after the initial series of screening was complete if the infant failed. Additional screens were implemented until an echo was performed or the screen was passed. No infants with CCHDs were identified.

**Conclusions:** The initial screening failure rate in the well-baby nursery of healthy infants remains higher when compared to sea level algorithms and is comparable to our previously published findings. Screening failure using the modified algorithm with delayed repeat screens reduced the failure rate and may provide a reasonable alternative to the sea-level algorithm to reduce the number of false positive screening results for infants at moderate altitude.

**Presenter:** Marci Sontag, PhD, Colorado School of Public Health, Aurora, CO & NewSTEPs, APHL, Silver Spring, MD, Phone: 303.724.4430, Email: marci.sontag@ucdenver.edu

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**Critical Congenital Heart Disease Resource Center**  
T. Wood¹, S. Singh¹, J. Ojodu¹, M. Sontag²; ¹NewSTEPs, Association of Public Health Laboratories, Silver Spring, MD, ²Colorado School of Public Health, University of Colorado Denver, Aurora, CO

**Abstract**

**Background:** Critical Congenital Heart Disease (CCHD) was added to the Recommended Uniform Screening Panel (RUSP) in September 2011. State newborn screening (NBS) programs are working to define the role of Public Health in CCHD screening, while developing implementation and data collection systems.

**Objective:** NewSTEPs provides technical assistance through monthly webinars and in-person meetings (inaugural in-person meeting held February 2014) to develop tools for guiding CCHD implementation, and tracking implementation challenges and successes across the U.S.

**Methods:** NewSTEPs CCHD technical assistance activities are guided by a workgroup of expert stakeholders. Monthly national webinars are held on topics suggested by the workgroup and the CCHD community. NewSTEPs hosted an in-person meeting in February 2014 of 80 representatives including state public health professionals from 38 states, pediatric cardiologists, database vendors, and Federal partners. Topics included education; quality control/quality improvement; data interpretation; data collection; implementation, legislation, and approval; Neonatal Intensive Care (NICU) protocols; and challenges in remote hospitals/home births/telemedicine. Attendees reviewed these topical areas in small groups and then presented their ideas broadly.

Results: All CCHD webinars are recorded, transcribed, and archived within the CCHD Resource Center. The in-person CCHD meeting provided a forum to assemble information for use in the NewSTEPs Technical Resource section of the website. The ideas discussed, including an analysis of challenges and successes, are available on the NewSTEPs website in a CCHD Resource Center. Two subsequent monthly CCHD webinars focused on the take home messages in each of the topical areas and were specifically intended for state program staff unable to attend the in-person meeting. The CCHD Resource Center will be updated in a dynamic manner to continue to provide a current and useful resource for state programs on the broad spectrum of CCHD implementation, data collection, and tracking.

Conclusion: NewSTEPs has developed a Resource Center to facilitate the addition of CCHD to screening and data surveillance activities. The CCHD Resource Center will serve as a model for additional conditions as they are added to the RUSP.

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Summary

The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs) provides resources and technical assistance to state newborn screening programs. This resource center was designed for the Critical Congenital Heart Disease (CCHD) programs in particular. The website and webinars are developed to help programs in the areas of implementation, data collection, and education.

CCHD was added to the Recommended Uniform Screening Panel (RUSP) in September 2011. NewSTEPs assists state programs by hosting webinar calls and held an in-person meeting to develop tools that would assist states as they progressed with CCHD implementation. The in-person meeting provided information and resources now shared on the CCHD section of the NewSTEPs website. This information includes challenges and potential solutions in several areas including education, data collection, Neonatal Intensive Care, legislation, and Quality Control among others. All of the presentations from the in-person meeting are available on the website along with the slides produced as a result of the breakout groups. The webinars are recorded, transcribed, and archived on the website as well.

A CCHD Technical Assistance work group was formed to meet on a monthly basis and determine topics and speakers for the bi-monthly (formerly monthly) webinars. The work group consists of pediatric cardiologists, Federal partners, and state CCHD program staff.

Additional resources and educational materials are welcome and states are requested to submit them to NewSTEPs staff for inclusion on the website. States are also encouraged to send pertinent publications and any upcoming events or meetings concerning CCHD.

The purpose of the CCHD resource center is to provide current and relative information for state programs as they continue to make improvements in their mission to provide a comprehensive newborn screening system which includes point of care screening.