

## Strategies for Implementing Screening for Critical Congenital Heart Disease

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**Abbreviations:** AAP (American Academy of Pediatrics), AHA (American Heart Association), ACCF (American College of Cardiology Foundation), CCHD (critical congenital heart disease), Centers for Disease Control and Prevention (CDC), CPT (Current Procedural Terminology), FDA (Food and Drug Administration), HHS (United States Department of Health and Human Services), HRSA (Health Resources and Services Administration), Maternal and Child Health (MCH), NICU (neonatal intensive care unit), NIH (National Institutes of Health), SACHDNC (Secretary's Advisory Committee on Heritable Disorders in Newborns and Children)

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## **Abstract**

**Background:** Although newborn screening for critical congenital heart disease (CCHD) was recommended by the United States Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) to promote early detection, it was deemed by the Secretary, HHS, not ready for adoption pending an implementation plan from HHS agencies.

**Objective:** To develop strategies for implementation of safe, effective, and efficient screening.

**Methods:** A workgroup was convened with members selected by the SACHDNC, the American Academy of Pediatrics, the American College of Cardiology Foundation, and the American Heart Association.

**Results:** Based on published and unpublished data, the workgroup made recommendations for a standardized approach to screening and diagnostic follow-up. Key issues for future research and evaluation were identified.

**Conclusions:** The workgroup members found sufficient evidence to begin screening for low blood oxygen saturation through the use of pulse oximetry monitoring to detect CCHD in well-baby and intermediate-care nurseries. Research is needed regarding screening in special populations (e.g., at high altitude) and to evaluate service infrastructure and delivery strategies (e.g., telemedicine) for nurseries without onsite echocardiography. Public health agencies will have an important role in quality assurance and surveillance. Central to the effectiveness of screening will be the development of a national technical assistance center to coordinate implementation and evaluation of newborn screening for CCHD.

## Introduction

Newborn screening has led to dramatic improvements in the morbidity and mortality for a variety of conditions.<sup>1</sup> Historically, newborn screening has been based on analysis of dried blood spots, and has operated as a partnership between healthcare providers, who obtain the samples and oversee medical follow-up, and state-based public health systems, which analyze the dried blood spots, assist healthcare providers and families in follow-up, and monitor the effectiveness of screening process through surveillance activities. The United States Health and Human Services (HHS) Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) was authorized by Congress to provide guidance to the Secretary, HHS, about which conditions should be included in newborn screening and how systems should be developed to assure appropriate screening and follow-up care.<sup>2,3</sup>

Prior to 2010, the only condition recommended for newborn screening that did not follow the dried blood spot paradigm was newborn hearing screening. Newborn hearing screening relies on in-hospital testing prior to discharge with subsequent outpatient audiology testing for those with abnormal results.<sup>4</sup> Unlike dried blood spot testing, individual hospitals and birthing centers had to invest in screening devices, maintain sufficient numbers of skilled staff to conduct the screening and interpret the results, and develop systems to track and communicate results of testing with public health departments, healthcare providers, and families. Because results of hearing screening originate in the hospitals and birthing centers, public health programs face significant challenges in assuring follow-up to assure the success of newborn hearing screening.<sup>5,6</sup>

In September 2010, the SACHDNC recommended that critical congenital cyanotic heart disease be added to the recommended uniform screening panel based on findings from a

comprehensive evidence review. The goal of this recommendation is to identify those newborns with structural heart defects usually associated with hypoxia in the newborn period that could have significant morbidity or mortality early in life with closing of the ductus arteriosus or other physiologic changes early in life. The SACHDNC considered seven specific lesions as primary targets for screening based on the advice of a technical expert panel: hypoplastic left heart syndrome, pulmonary atresia, tetralogy of Fallot, total anomalous pulmonary venous return, transposition of the great arteries, tricuspid atresia, and truncus arteriosus. This subset of lesions excludes those not usually associated with hypoxia (e.g., aortic valve stenosis).<sup>7</sup>

This recommendation built upon the 2009 statement from the American Academy of Pediatrics (AAP) and the American Heart Association (AHA), which found compelling reasons for newborn screening but called for “...studies in larger populations and across a broad range of newborn delivery systems...” before pulse oximetry screening should be recommended.<sup>7</sup> The SACHDNC was especially persuaded by a prospective screening study of nearly 40,000 newborns in Sweden<sup>8</sup> and a separate study of nearly 40,000 newborns in Germany.<sup>9</sup> Comparing the accuracy of pulse oximetry monitoring for the seven defects specified by the SACHDNC to these other studies is somewhat challenging because of differences in the lesions that were targeted for detection by the screening. For example, the study in Sweden considered all ductal-dependent lesions. Their approach, for example, added critical aortic stenosis and coarctation of the aorta, but excluded tetralogy of Fallot. With this case definition, the study from Sweden found the sensitivity of pulse oximetry monitoring to be 62.1% and the specificity to be 99.8% and a false positive rate of 0.17%. In contrast, the AAP/AHA statement had a broader definition, including all lesions that would require surgery or catheter intervention in the first year of life.

The SACHDNC made the recommendation for screening with the understanding that specific activities would be undertaken, including having the Health Resources and Services Administration (HRSA) guide the development of screening standards and the infrastructure needed for the implementation of a public health approach to point-of-service screening, and develop education materials; having research conducted by the National Institutes of Health (NIH); and surveillance and tracking by the Centers for Disease Control and Prevention (CDC). However, the Secretary, HHS, did not endorse the recommendation from the SACHDNC to begin screening, in part because of questions about how to implement screening. Some states (e.g., Maryland, New Jersey) have legislation promoting newborn screening for CCHD, increasing the urgency for a draft implementation plan.

The SACHDNC, in collaboration with the AAP, the American College of Cardiology (ACCF), and the AHA, convened a workgroup to outline strategies to the SACHDNC for implementation, which is summarized in this report. It is important to recognize that many newborns with the targeted congenital heart defects do not develop clinically appreciable cyanosis until after nursery discharge and some lesions (e.g., hypoplastic left heart syndrome) may present with significant cardiovascular compromise without apparent cyanosis. Therefore the workgroup recommended renaming the target conditions *critical congenital heart disease* (CCHD), omitting the word “cyanotic.”

## Methods

A workgroup was convened for a two-day meeting in January 2011. Workgroup members (see Appendix) included: primary care providers; specialists, including pediatric cardiologists and neonatologists; nurses; representatives from the AAP, the ACCF, the AHA, the American College of Medical Genetics, the March of Dimes, the Association of Maternal and Child Health Programs, the Association of Public Health Laboratories, and the SACHDNC; parent screening advocates; state public health officials; and, representatives from the CDC, the Food and Drug Administration (FDA), HRSA, and NIH. Included were individuals who have implemented pulse oximetry monitoring for CCHD in newborn nurseries within Arkansas, California, Minnesota, New York, Washington, and Washington, DC. The workgroup was moderated by William T. Mahle, MD, a pediatric cardiologist who led the development of the 2009 AAP/AHA statement<sup>7</sup> and R. Rodney Howell, MD, chair of the SACHDNC. The workgroup was supported by other invited experts, including from the CDC and the FDA, and two who had conducted large-scale studies of screening in Europe. The workgroup meeting was open to the public.

The meeting focused on recommendations for pulse oximetry monitoring for CCHD, including recommendations for the service infrastructure needs for follow-up, and strategies for filling in important knowledge gaps. A smaller writing group prepared a summary report of the meeting, which was then iteratively revised with the workgroup until agreement was obtained. The report was subsequently reviewed by the AAP, the ACCF, and the AHA Council for Cardiovascular Disease in the Young, each of which endorsed this report.

## **Results**

### *Screening Population and Targets*

The workgroup chose to focus initially on screening in the well-baby nursery because of the risk of missed cases of CCHD among healthy-appearing newborns. The workgroup recognized the importance of also considering screening within neonatal intensive care units (NICUs). However, developing a simple algorithm for the NICU setting is challenging because of the heterogeneity of underlying conditions (e.g., prematurity, meconium aspiration syndrome, sepsis). Unlike the well-baby nursery, many babies in the NICU undergo repeated medical evaluations, are monitored by pulse oximetry, and have longer lengths of stay. However, there was concern that screening only in well-baby nurseries would miss newborns with short stays in intermediate-care nurseries. The workgroup endorsed screening babies in intermediate-care nurseries or other units in which discharge is common in the first week, utilizing the workgroup protocol for screening in the well-baby nursery. The workgroup chose not to focus on out-of-hospital births, which raise challenging coordination-of-care issues, which will be addressed in the future.

One of the advantages of pulse oximetry monitoring is the ability to detect other hypoxic cardiac or non-cardiac associated conditions (e.g., persistent pulmonary hypertension), characterized by the SACHDNC as targets secondarily detected by the screening technology (“secondary targets”). Secondary targets are common to other newborn screening tests (e.g., identification of Hemoglobin H disease when screening for sickle cell anemia<sup>10</sup>). Although the primary goal of screening based on the SACHDNC recommendation is identification of the

seven specific lesions associated with CCHD, tracking rates of identification of important secondary targets could lead to modifications of the screening protocol.

### *Screening Technology*

The workgroup recommended that screening be done with motion-tolerant pulse oximeters<sup>11</sup> 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. Commercially available pulse oximeters often are labeled by manufacturers by generation of technology (e.g., “next generation”). However, generation designation is not standardized and may not be related to validity or reliability. Furthermore, no standards have been developed regarding motion tolerance. A new guidance document on the safety and effectiveness of pulse oximeters is being developed by the FDA.<sup>12</sup> When the guidance document is finalized, any pulse oximeter used for screening should meet FDA recommendations. Having specific FDA-cleared labeling and conformance to the relevant standard<sup>13</sup> will be an important strategy for assuring that appropriate devices are used for screening.

Pulse oximeters can be used with either disposable or reusable probes. Reusable probes can reduce the cost of screening, but must be appropriately cleaned between uses to minimize the risk of infection. Some probes have been developed that are partially reusable, which reduces the need to clean between uses and are less expensive than fully disposable probes. Probes with close coupling to skin (i.e., taped rather than clamped), provide better performance for oximetry monitoring in newborns. Pulse oximeters are validated only with the specific probes recommended by the manufacturer; therefore, to optimize valid screening, manufacture-recommended pulse oximeter-probe combinations should be used.

### *Screening Criteria*

The workgroup recommended that screening not begin until 24 hours of life or as late as possible if earlier discharge is planned, and be completed on the second day of life. Earlier screening can lead to false positive results because of the transition from fetal to neonatal circulation and stabilization of systemic oxygen saturation levels and later screening can miss an opportunity for intervention before closing of the ductus arteriosus. Screening was recommended in the right hand and one foot, either in parallel or in direct sequence. The pulse oximetry measure is complete once the waveform on the oximeter's plethysmograph is stable or there is other indication that the device is appropriately tracking the baby's pulse rate.

Selecting the threshold for a positive pulse oximetry monitoring is challenging because it must trade off the harm of missing CCHD against the harm of false positive screens. None of the studies reviewed by the SACHDNC included receiver operator characteristic curves developed from primary data, which would allow a direct evaluation of this tradeoff. However, based on new data from the large population-based screening activities in Sweden<sup>8</sup> and England (unpublished data from a National Institute for Health Research Health Technology Assessment for the United Kingdom<sup>14</sup>), the workgroup developed a recommendation for screening based on what has been shown to be effective in these studies.

The screening protocol is described in the figure. A screen would be considered positive screen when (1) any oxygen saturation measure is <90%; (2) oxygen saturation is <95% in both extremities on three measures each separated by one hour; or (3) there is a >3% absolute difference in oxygen saturation between the right hand and foot on three measures each separated by one hour. Any screening that is  $\geq 95\%$  in either extremity with  $\leq 3\%$  absolute difference in

oxygen saturation between the upper and lower extremity would be considered a pass and screening would end.

Anecdotal reports suggest that false positives are decreased if the infant is alert, possibly by reducing the likelihood of low oxygen saturations due to hypoventilation in deep sleep. In addition, timing pulse oximetry monitoring around the time of the newborn hearing screening improves efficiency, assuming that the hearing screening is conducted after 24 hours or immediately prior to discharge. The particular screening strategy should reflect the conditions within each particular nursery and the needs of babies, families, and the healthcare providers.

The workgroup noted that performing a typical physical examination alone for CCHD led to almost ten times more false positive results compared to using similar screening protocols in Sweden and the United Kingdom.<sup>8,14</sup> Repeated pulse oximetry testing following an initial positive screen if oxygen saturation is <95% in both extremities or there is a >3% absolute difference in oxygen saturation between the right hand and foot, as illustrated in the protocol, lowers the likelihood of a false positive result compared to a single measurement. However, there is no need to repeat pulse oximetry testing if the oxygen saturation is <90% in any screen.

The workgroup emphasized the importance of not having pulse oximetry monitoring replace a complete history and physical examination, which can sometimes detect CCHD before the development of hypoxia. Pulse oximetry monitoring, therefore, should be used to complement the physical exam. While agreement was reached on the screening protocol, the workgroup was concerned that this screening protocol might lead to high rates of false positives in high elevation communities such as Denver, Colorado.<sup>15-17</sup> The criteria for a positive screen may need to be modified in these areas. Regardless of the specific screening thresholds, comprehensive training will be central to implementing safe and effective screening.

*Diagnostic Strategies*

Any newborn with a positive screen first requires a comprehensive evaluation for causes of hypoxemia. In the absence of other findings to explain hypoxemia, CCHD needs to be excluded based on a diagnostic echocardiogram, which would involve either an echocardiogram within the hospital or birthing center, transport to another institution, or through the use of telemedicine for remote evaluation. The workgroup also emphasized the need for high-quality echocardiograms with interpretation by a pediatric cardiologist because of the challenge of diagnosis in some cases (e.g., total anomalous pulmonary venous return). The workgroup recommended against replacing a diagnostic echocardiogram with other evaluations (e.g., chest x-ray, electrocardiogram, hyperoxia test), which can be inaccurate in diagnosing CCHD. When feasible, the workgroup endorsed consulting a pediatric cardiologist prior to obtaining the echocardiogram.

Because of the importance of quickly establishing the diagnosis of CCHD, the workgroup recommended that hospital or birthing centers should establish a protocol to assure timely evaluation, including echocardiograms and any necessary subsequent follow-up, prior to instituting a CCHD screening program. Future work will be needed to assure the quality of in-center and telemedicine approaches to echocardiography. The workgroup also recognized the importance of training an adequate number of pediatric cardiologists to assure that diagnostic services are available, either on-site, with short-distance transport, or through telemedicine. Similarly, pediatric cardiac surgery centers will have to be prepared to accept newborns with CCHD identified by pulse oximetry.

*Connection to the Medical Home*

The results of newborn CCHD screening should be communicated to newborns' primary care providers. During the first outpatient visit, primary care providers should assure that all newborns were appropriately screened and received any necessary follow-up. The workgroup recognized the importance of developing health information exchange systems to allow primary care providers, in addition to cardiology subspecialists to easily track this information. To facilitate this, standards for electronic reporting of pulse oximetry will need to be developed. Standards for electronic reporting would also help facilitate the development of quality measures.

Primary care providers will also need to develop strategies for screening those newborns who missed screening. As with other newborn screening tests, primary care providers play a central role in assuring long-term follow-up for those babies diagnosed with CCHD through newborn screening and coordinating their care with a pediatric cardiologist.<sup>2</sup>

*Public Health, Quality Assurance, and Surveillance*

Follow up of a positive screen should be managed by the hospital or birth center prior to discharge; therefore, the role of public health agencies in CCHD screening is different than in the case of newborn dried blood spot screening or newborn hearing screening. However, public health agencies can play a central role in quality assurance and surveillance. There are several challenges to public health agencies' involvement with CCHD screening, including: inability to collect real-time screening data through health information exchange systems, absence of a direct presence of public health personnel in hospitals and birthing centers, and financial and staffing limitations required for development of a new public health program.

surveillance and prevention programs should have a role in surveillance and evaluation of CCHD screening. These programs already conduct public education and outreach; train providers; and support genetic services, newborn screening programs, and services for children with special healthcare needs. Although state birth defects programs could assist with CCHD surveillance, there are differences across states in resources for such activities and the approaches to case ascertainment. As of February 2011, there were forty birth defects surveillance programs in the United States and six more in development. With adequate resources, some of these programs could potentially collect and track data on populations screened, not screened, or with false negative screening results. Data could also be collected on whether a diagnosed CCHD was detected through prenatal ultrasound or newborn pulse oximetry monitoring. Collecting data to understand the factors associated with false positive pulse oximetry monitoring could also help refine the recommended screening activities. Although there is currently no capacity in birth defects programs to undertake real-time follow-up of CCHD positive screens including short-term follow-up, the infrastructure is in place in many states for birth defects surveillance programs to play a critical role in conducting long-term surveillance and evaluation.

#### *Healthcare Costs*

The main costs of a screening program for CCHD are related to staff time for screening, tracking results, and communicating with parents, the purchase and maintenance of screening equipment, consumables associated with screening (e.g., probes, adhesive wraps, cleaning supplies), the costs associated with verifying a positive screen, and the costs associated with treatment. The cost of conducting pulse oximetry examination and follow-up is quite low in absolute terms, with published estimates of \$5 or less per infant,<sup>7,8</sup> up to \$10 per infant,

depending on the protocol.<sup>14</sup> Although screening can sometimes be completed in less than one minute, other studies estimate that the process takes five minutes of staff time, including communication with parents.<sup>14</sup> The cost estimate compares quite favorably with cost estimates for newborn hearing screening (\$30 or more per infant, with an average reimbursement by private health plans in 2004 of \$84 if billed separately<sup>18</sup>). Moreover, the cost of pulse oximetry is significantly offset by avoided costs of care. The report from Sweden calculated that the savings in healthcare costs from the prevention of one case of complications of circulatory collapse resulting from an undiagnosed CCHD may exceed the cost of screening two thousand newborns.<sup>8</sup>

Another potentially important cost is related to delayed discharge because of the need to repeat screening or to obtain diagnostic evaluation, leading to extra hospital days which may not be reimbursed by insurance carriers. Echocardiography is typically well-reimbursed. However, the cost of transport can be high with variable insurance reimbursement. Although telemedicine for remote echocardiography could be important for hospitals and birthing centers without ready access, it is unclear who would pay to develop and maintain the infrastructure.

At present, there is no clear way to bill for pulse oximetry monitoring, since the currently available Current Procedural Terminology (CPT) codes for pulse oximetry are only appropriate when accompanied by a diagnostic code for a pulmonary disease associated with hypoxia.<sup>19</sup> The AAP, AHA, and ACC should work with the American Medical Association, which develops CPT codes, to develop the appropriate CPT codes for pulse oximetry monitoring as well as with public and private payers to assure appropriate reimbursement. However, newborn hospital-based screening services such as hearing screening are commonly not reimbursed separately if conducted by regular hospital nursery staff, even with appropriate CPT codes available. Because

the cost of conducting pulse oximetry monitoring is quite low, the cost to hospitals and birthing centers should not be a major barrier. In Switzerland, for example, most birthing centers have adopted pulse oximetry monitoring, with an estimated 85% of infants being screened, despite no mandate for either screening or insurance reimbursement for screening.<sup>20</sup>

The workgroup recognized the concerns about limited healthcare resources, and emphasized the need to weigh the costs of pulse oximetry against the potential benefits of early diagnosis of CCHD, including the costs saved by decreasing the morbidity associated with later diagnosis. Cost data should be compared to the screening outcomes data, such as those collected by public health agencies, to inform policy makers and to develop new interventions to improve the efficiency of screening.

#### *Healthcare Provider and Family Education*

Both healthcare providers and families must understand the rationale for and limitations of pulse oximetry monitoring to detect CCHD, including the important understanding that negative screening does not exclude the possibility of CCHD or other congenital heart disease. Similarly, education is needed to minimize the harm that may be generated by false positive screens. Implementation of other newborn screening tests has been improved through the development of simple clinical decision support tools for healthcare providers which explain the screening and what should be done in the event of a positive screen (e.g., the HRSA-funded ACTION sheets) and simple fact sheets for families.<sup>21</sup> Similar materials need to be developed for pulse oximetry monitoring, and should be available in print and through electronic media in English, Spanish, and other local languages. Implementation toolkits to help hospitals and birthing centers assess their degree of readiness for screening, to develop algorithms for screening, and to evaluate their ongoing activities are also important.

*Coordination of Implementation Activities*

The workgroup endorsed the development of a national clearinghouse and technical assistance center similar to the National Resource Center for Newborn Hearing Screening (<http://www.infanthearing.org>), the National Newborn Screening and Genetics Resource Center (<http://genes-r-us.uthsca.edu>), or the Emergency Medical Services for Children National Resource Center (<http://www.childrensnational.org/EMSC>). Each of these provides examples of ways to coordinate service delivery between healthcare providers and state public health agencies. Replicating this approach through partnership with State Title V MCH programs would allow implementation that takes into account specific local factors such as the availability of diagnostic services.

## Discussion

A significant body of evidence suggests that early detection of CCHD through pulse oximetry monitoring is an effective strategy for reducing morbidity and mortality in young children. The workgroup identified strategies for hospitals and birthing centers to implement pulse oximetry monitoring for CCHD , including the following specific recommendations:

- Screening should be conducted using motion-tolerant pulse oximeters that report functional oxygen saturation cleared by the FDA for use in newborns.
- Screening should be based on the recommended screening algorithm, and be performed by qualified personnel (e.g., nurses, allied health technicians) who have been educated in the use of the algorithm and trained in pulse oximetry monitoring of newborns.
- The algorithm cut-offs may need to be adjusted in high-altitude nurseries.
- Any abnormal pattern of low blood oxygen saturation requires a complete clinical evaluation by a licensed, independent practitioner. In the absence of other findings to explain hypoxemia, CCHD needs to be excluded, based on a comprehensive echocardiogram interpreted by a pediatric cardiologist before discharge to home. If an echocardiogram cannot be performed in the hospital or birthing center and diagnosis by telemedicine is not possible, strong consideration should be made for transfer to another medical center for diagnosis. Before implementing screening, protocols for arranging diagnostic follow-up should be established.
- Hospitals and birthing centers should establish partnerships with local and state public health agencies to develop strategies for quality assurance and to monitor the impact of screening.

- Primary care providers should assure that newborns in their practice were appropriately screened and should work to facilitate long-term follow-up for those diagnosed with CCHD.
- Standards should be developed for electronic reporting of pulse oximetry monitoring and diagnostic outcomes.

The workgroup recognized the challenges of implementing a new screening program. To assure that screening is implemented in a safe and effective manner, the workgroup strongly endorsed the development and funding of a national technical assistance center to disseminate best practices; to partner with public health agencies to monitor the impact of screening; to evaluate and make recommendations regarding workforce and related infrastructure needs; and, to coordinate research to help answer the important unanswered questions regarding screening thresholds and optimal strategies for diagnosis and follow-up. The Secretary, HHS, has directed an interagency workgroup to develop a plan to address these critical gaps prior to recommending that CCHD be a part of the recommended uniform screening panel.

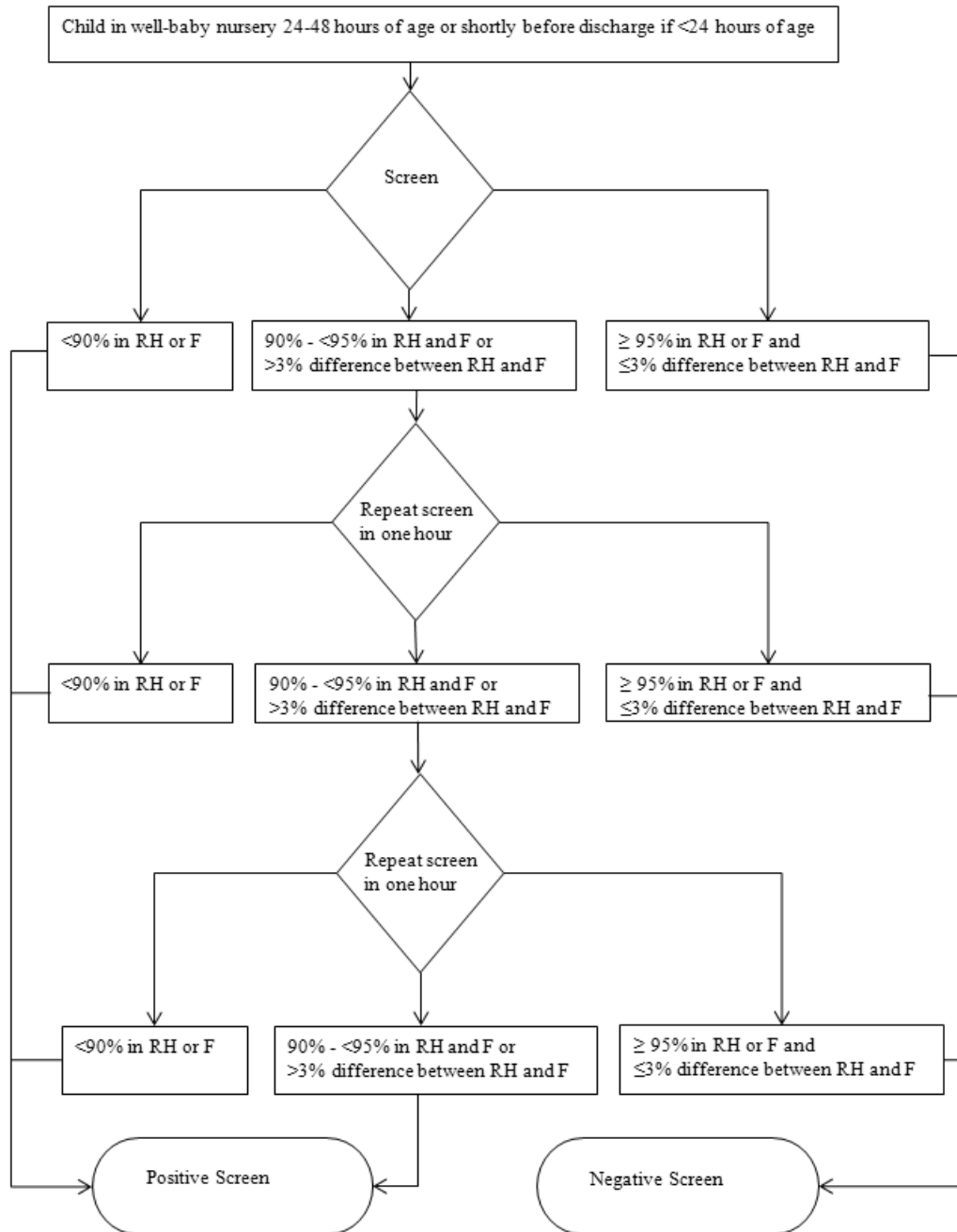


Figure. The proposed pulse oximetry monitoring protocol based on results from the right hand (RH) and either foot (F).

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## **Appendix: Workgroup Members**

The following is a list of workgroup members and the agencies or organizations they represented at the meeting. Being listed as a workgroup member does not imply that the individuals or the organization that they represent endorses all aspects of this report.

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