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Authors

Evan Withrow, MS
Pediatric Genomics and Early Hearing Epidemiologist, Maternal and Child Health Section,
Division of Lifecourse Epidemiology and Genomics

Mary Kleyn, MSc
Manager, Newborn Screening Follow-up Section,
Division of Lifecourse Epidemiology and Genomics

Kristen Thompson, MPH
Coordinator, Newborn Screening Follow-up Section,
Division of Lifecourse Epidemiology and Genomics

Karen Andruszewski, BS
Quality Assurance Coordinator, Newborn Screening Follow-up Section,
Division of Lifecourse Epidemiology and Genomics
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Executive Summary

The critical congenital heart disease (CCHD) annual report provides an overview of the pulse oximetry screening portion of the State of Michigan Newborn Screening (NBS) Program, including screening methods, metrics, quality assurance information, and implications.

The Michigan NBS Program began in 1965 with one disorder and has grown since, adding many new disorders to the screening panel. Pulse oximetry screening for CCHD became a mandatory component of the screening panel on April 1, 2014. Since the screening mandate was implemented, around 500,000 infants in Michigan have been screened for CCHD and approximately 20 cases of CCHD have been detected through screening.

Of the 109,740 infants born in Michigan in 2017 with blood spot screens submitted, 71 failed their pulse oximetry screen, and six were diagnosed with a CCHD. In 2017, seven infants out of every 10,000 screened failed their pulse oximetry screen and one out of every 16,873 infants born in Michigan who had a valid pulse oximetry screen was identified with a CCHD through pulse oximetry screening.

Developments occurring in 2017:

Michigan continued to disseminate findings to committees, providers, and partners.

- The findings from different studies and analyses related to pulse oximetry screening were presented at the following meetings:
  - Michigan Epidemiology Conference in Grand Rapids, Michigan
  - Newborn Screening and Genetic Testing Symposium in New Orleans, Louisiana

Michigan continued to conduct trainings with hospitals.

- Information on pulse oximetry screening was presented at trainings for hospital personnel in Lansing and Detroit. These meetings were attended in-person and via webinar by approximately 120 health professionals representing 44 birthing hospitals across Michigan.

NBS personnel continued serving on national work groups, including:

- CCHD Technical Assistance Workgroup
- Education and Training workgroup for the Advisory Committee on Heritable Disorders in Newborns and Children

NBS personnel presented or participated as an exhibitor at numerous education events, including:

- WIC Conference
- Beaumont Baby Fair
- American Academy of Pediatrics, Michigan Chapter, Novi
- March of Dimes Health Walk, University of Michigan, Ann Arbor
Listing of Figures & Tables

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## Acronym Key

<table>
<thead>
<tr>
<th>Acronym</th>
<th>Name</th>
</tr>
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<tbody>
<tr>
<td>CCHD</td>
<td>Critical Congenital Heart Disease</td>
</tr>
<tr>
<td>CHD</td>
<td>Congenital Heart Disease</td>
</tr>
<tr>
<td>MDHHS</td>
<td>Michigan Department of Health and Human Services</td>
</tr>
<tr>
<td>NBS</td>
<td>Newborn Screening</td>
</tr>
<tr>
<td>NICU</td>
<td>Neonatal Intensive Care Unit</td>
</tr>
<tr>
<td>PCP</td>
<td>Primary Care Physician</td>
</tr>
<tr>
<td>QA</td>
<td>Quality Assurance</td>
</tr>
<tr>
<td>SCN</td>
<td>Special Care Nursery</td>
</tr>
</tbody>
</table>
I. Introduction

This report provides an overview of Michigan’s pulse oximetry screening program for critical congenital heart disease (CCHD), which is a component of Michigan’s Newborn Screening (NBS) Program. The report includes methods for screening and submission of results, screening performance metrics, and quality assurance information.

This report is intended to provide:

- An introduction to the pulse oximetry screening process and the history behind it
- Pulse oximetry screening performance metrics
- Quality assurance information

What is newborn screening?

NBS is the process of identification of adverse health conditions and implementation of treatment in newborns before the onset of disease processes in the individual. Screening is conducted shortly after birth, confirmatory testing is used when necessary, and treatment of the disorder is administered in a timely fashion. Proper use of NBS minimizes the risk associated with disease, reduces the possibility of long-term sequelae, and aims to increase the quality of life of any non-treatable diseases. Potential outcomes of disorders on the NBS panel include neurological impairment and damage, intellectual disability, organ damage including the liver, eyes, or spleen, and even death if not detected early.

Three different screens are administered to the newborn to reduce the likelihood of these outcomes occurring. Blood spots are collected from infants in the first days of life to screen for metabolic disorders, hemoglobinopathies, endocrine disorders, and other genetic conditions. Hearing screening is conducted to detect hearing loss in the infant, so that treatment and intervention may be started promptly. Pulse oximetry screening is used to evaluate the oxygen saturation of blood in the extremities of the newborn to detect potential heart defects, specifically critical congenital heart defects.

What are CCHDs?

Approximately 1 in 100 babies is affected by a congenital heart defect (CHD), making CHDs rank among the most prevalent birth defects. Pulse oximetry screening has been shown to detect some forms of CHDs based on low oxygen saturation and, more specifically, has been able to detect more serious abnormalities categorized as critical congenital heart disease (CCHD). It is imperative that CCHDs be detected as early as possible to reduce the risk of circulatory collapse and death.

What is pulse oximetry screening?

Pulse oximetry screening sends red light and infrared light through the hand and foot of the infant and measures the amount of each type of light absorbed to detect the amount of oxygen in the blood. Low oxygen saturation in the blood signifies that there may be a problem with the heart and circulatory system. Pulse oximetry screening targets identification of 12 specific CCHDs (Table 1).
Table 1: Primary Targets for Pulse Oximetry Screening

<table>
<thead>
<tr>
<th>Critical Congenital Heart Disease</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Coarctation of the Aorta</td>
<td>Tetralogy of Fallot</td>
</tr>
<tr>
<td>Double-Outlet Right Ventricle</td>
<td>Total Anomalous Pulmonary Venous Return</td>
</tr>
<tr>
<td>Ebstein’s Anomaly</td>
<td>D-Transposition of the Great Arteries</td>
</tr>
<tr>
<td>Hypoplastic Left Heart Syndrome</td>
<td>Tricuspid Atresia</td>
</tr>
<tr>
<td>Interrupted Aortic Arch</td>
<td>Truncus Arteriosus</td>
</tr>
<tr>
<td>Single Ventricle</td>
<td>Pulmonary Atresia</td>
</tr>
</tbody>
</table>

Pulse oximetry screening background:

Pulse oximetry screening was officially recommended by the U.S. Department of Health and Human Services Secretary in 2011 as an important tool for detecting CCHDs in asymptomatic newborns. Studies have shown the benefit of pulse oximetry screening in improving CCHD detection rates. The State of Michigan NBS Program added pulse oximetry screening to the mandated screening panel effective April 1, 2014. Michigan’s NBS Program recommends the pulse oximetry algorithm endorsed by the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children in 2011 (Appendix).

Hospitals:

In 2017, Michigan had 83 hospitals with birthing units. Each hospital has a designated NBS coordinator who helps facilitate the screening process and submission of results. Pulse oximetry screening quality assurance reports were developed at the end of 2017 and were sent to hospitals during the first quarter of 2018. The goal of these quality assurance reports is to help monitor reporting rates, screening metrics, and areas of improvement for each hospital. Periodic site visits are also made by the NBS Nurse Consultant to evaluate screening processes and make recommendations for further improvement.

Midwives and Home Births:

There are approximately 93 midwives registered with the NBS Program in Michigan. Guidance and individual assistance are provided to midwives to facilitate meeting program standards. The NBS Program also conducts a program that allows for midwives to loan pulse oximeters from the NBS Program. The goal for this program is to alleviate a cost barrier for midwives while elevating pulse oximetry screening rates in the out-of-hospital birth population.
II. Methods

This section describes the process in which infants are screened and the methods to calculate a) the total number of newborns eligible for screening, b) the demographic characteristics of the newborns, c) total number of infants who failed their pulse oximetry screen, d) screening performance metrics, and e) quality assurance indicators.

Screening reporting methods:

For each birth, hospital staff have three options for submitting the pulse oximetry screening results to the NBS Program. They may enter the individual-level screening results into a web-based reporting system (eReports®, Perkin Elmer Life Sciences, Inc.). Hospitals also have the option to use a secure file transfer service (FTS) to send a file of screening results to the NBS Program. The final option is to upload information using Health Level Seven (HL7), which is an instantaneous information upload system. Midwives have the option of submitting results electronically via eReports® or by sending the results on paper forms to the NBS Program.

Pulse oximetry screening results for total number of newborns eligible for screening:

Vital records statistics data collected by the Division for Vital Records and Health Statistics at MDHHS were used to determine the total number of live births eligible for screening.

Demographic characteristics:

The demographic characteristics are presented for Michigan residents who received a pulse oximetry screen in Michigan. This report focuses on this population, as out-of-state infants, who were born at a Michigan hospital, are often followed-up and diagnosed elsewhere. Screening information, including demographic information and screening outcomes, was obtained from NBS records.

Total number of newborns with CCHD identified by pulse oximetry screening:

The MDHHS laboratory information management system (PerkinElmer Life Sciences, Inc.) was used to identify individuals who failed their pulse oximetry screens. CCHD cases referred to in this report must be a) identified by NBS through pulse oximetry screening and b) Michigan residents.
Screening metrics

Two different screening metrics that can help evaluate a screening test are positive predictive value and false positive rate. The positive predictive value is the number of infants confirmed with CCHD divided by the number of infants who failed the pulse oximetry screen, expressed as a percentage. The false positive rate is defined as the number of infants with false positive screens divided by the number of infants screened expressed as a percent. Ideal screening tests have a high positive predictive value (perfect = 100%) and a low false positive rate (perfect = 0%). This ideal screening test would correctly identify all cases of a disorder with no false positives.

Quality assurance:

Quality assurance (QA) data were obtained from the laboratory information management system. The QA indicators focus on time of birth to pulse oximetry screen, time between pulse oximetry screen and reporting of results to the NBS Program, compliance with the NBS pulse oximetry screening algorithm, and whether screens were missed. Table 4 describes each of the QA metrics and how the metrics are calculated. Other QA indicators look at reasons for a missed screen, including prenatal CCHD diagnosis, postnatal CCHD diagnosis, infant distress, infant transfer to a different hospital, refusal of screening procedures, infant death, and being referred for further examination to a practitioner.
III. Screening Results

Demographic characteristics of screened newborns

This section describes the population of screened infants born in 2017 in terms of race, birth weight, gestational age, and birth place (hospital nursery, NICU/SCN, or non-hospital). These data are helpful in understanding the epidemiology (distribution of disease among the population in Michigan) of CCHD covered in the subsequent sections of the report.

The Michigan NBS Program received blood spot cards for more than 99% of the 110,519 live births occurring in Michigan in 2017. These blood spot cards were screened for more than 50 different disorders. Pulse oximetry screening results were reported for 92.3% of those with a blood spot submitted (data not shown). Approximately 7.9% (n=8,501 without valid results, n=13 followed incorrect algorithm, n=124 did not receive a proper rescreen) of newborns with blood spot screens submitted in 2017 did not have valid pulse oximetry screening results.

Table 2 reports the demographic and perinatal characteristics of Michigan residents born in 2017 with a pulse oximetry screen reported. As indicated in Table 2, most in-state infants screened were white, born in a well birth nursery of a hospital, full term (≥37 weeks gestational age), and normal birth weight (>2,500 g). Overall, 8.6% of infants were admitted to a NICU or SCN, 8.7% were low birth weight, and 8.2% were born preterm (<37 weeks). Black infants were overrepresented in the NICU/SCN population, in the low birthweight population, and the preterm population. White infants were overrepresented in the out-of-hospital birth population.

Table 2: Demographic and Perinatal Characteristics of Infants born in Michigan with a Valid Pulse Oximetry Screen, 2017

<table>
<thead>
<tr>
<th>Race</th>
<th>Column Total</th>
<th>Nursery Type</th>
<th>Birth Weight (g)</th>
<th>Gestational Age (wks.)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Regular Hospital</td>
<td>NICU/SCN</td>
<td>Non-Hospital</td>
</tr>
<tr>
<td></td>
<td>N</td>
<td>%</td>
<td>N</td>
<td>%</td>
</tr>
<tr>
<td>White</td>
<td>63,037</td>
<td>62.4</td>
<td>57,864</td>
<td>91.8</td>
</tr>
<tr>
<td>Black</td>
<td>18,335</td>
<td>18.1</td>
<td>15,759</td>
<td>86.0</td>
</tr>
<tr>
<td>Multi-Racial</td>
<td>6,254</td>
<td>6.2</td>
<td>5,779</td>
<td>92.4</td>
</tr>
<tr>
<td>Other</td>
<td>7,854</td>
<td>7.8</td>
<td>7,337</td>
<td>93.4</td>
</tr>
<tr>
<td>Missing</td>
<td>5,622</td>
<td>5.6</td>
<td>5,222</td>
<td>92.9</td>
</tr>
<tr>
<td>Total</td>
<td>101,102*</td>
<td>100.0</td>
<td>91,961</td>
<td>91.0</td>
</tr>
</tbody>
</table>

*8,501 newborns were missing valid screening results, 13 did not follow the Michigan Algorithm while screening, and 124 newborns were missing proper rescreens.
Pulse oximetry screening outcome information

Figure 2 shows the breakdown of screening outcomes in Michigan in 2017. In total, of the 101,102 infants who had valid pulse oximetry screening results submitted, 71 infants, or 0.1%, failed their pulse oximetry screen, while 101,031 infants passed their screens (Table 3). A total of 124 infants were missing rescreens and 13 infants had screens that did not follow Michigan’s screening algorithm. These 137 infants were excluded from Table 3. All racial groups had at least 99% of babies with a valid outcome from the pulse oximetry screen pass their screen, with a total of 101,031 infants, or 99.9%, passing their screens (Table 3).

Table 3: Pulse Oximetry Screening Outcomes of Infants Born in Michigan, by Race, 2017

<table>
<thead>
<tr>
<th>Race</th>
<th>Column Total</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Column Total</td>
<td>Pass</td>
</tr>
<tr>
<td></td>
<td>N</td>
<td>%</td>
</tr>
<tr>
<td>White</td>
<td>63,037</td>
<td>62.4</td>
</tr>
<tr>
<td>Black</td>
<td>18,335</td>
<td>18.1</td>
</tr>
<tr>
<td>Multi-Racial</td>
<td>6,254</td>
<td>6.2</td>
</tr>
<tr>
<td>Other</td>
<td>7,854</td>
<td>7.8</td>
</tr>
<tr>
<td>Missing</td>
<td>5,622</td>
<td>5.6</td>
</tr>
<tr>
<td><strong>Column Total:</strong></td>
<td><strong>101,102</strong>*</td>
<td><strong>100.0</strong></td>
</tr>
</tbody>
</table>

*8,501 newborns were missing valid screening results, 13 did not follow the Michigan Algorithm while screening, and 124 newborns were missing proper rescreens.
Reasons for missed screens

Of the 8,501 infants with no pulse oximetry screening values, 6,812 had no pulse oximetry screening information reported, while 1,689 infants had results reported as missing. The most prevalent reason for missing a screen was receiving an echocardiogram (n=563), followed by missing a screen without reason (n=522), and the infant being transferred between facilities (n=311). A total of 79 infants had a prenatal diagnosis of a CCHD and 60 infants had a postnatal diagnosis of a CCHD. Of the 1,689 infants who had a screen reported as missing, 65 infants were reported as being in distress, 41 were referred for further testing (including five who received an echocardiogram), 17 had parents who would not permit screening, and 31 infants expired before screening could occur.

Screening metrics

Six confirmed cases of CCHD were detected after infants failed their pulse oximetry screen. Pulse oximetry screening, in Michigan, in 2017, had a positive predictive value of 8.5% and a false positive rate of 0.06%. In addition to the six infants who failed their screen and were diagnosed with a CCHD, two infants who were incidentally diagnosed with a secondary condition. Secondary conditions include hypothermia, lung disease, infection, non-critical congenital heart defects, persistent pulmonary hypertension, and other hypoxic conditions not otherwise specified.
IV. Quality Assurance Information

This section includes quality assurance (QA) information about pulse oximetry screening. These indicators are included in quarterly reports distributed to hospitals.

Table 4: Indicators and Performance Goals for Pulse Oximetry Screening, Michigan, 2017

<table>
<thead>
<tr>
<th>Metric</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Percent Screened</td>
<td>Calculated by dividing the number of newborns with a right hand and foot pulse oximetry screen results reported to the NBS Program by the total number of newborns with a bloodspot screen. Target = 90%.</td>
</tr>
<tr>
<td>Percent Reported on Time</td>
<td>Calculated by dividing the number of newborns with a right hand and foot pulse oximetry screen reported to the NBS Program within 10 days of the screen date by the total number of newborns with a bloodspot screen. Target = 90%.</td>
</tr>
<tr>
<td>Percent Timely Screened</td>
<td>Calculated by dividing the number of newborns with a right hand and foot pulse oximetry screen conducted between 20 and 28 hours after birth by the total number of newborns with a bloodspot screen. Target = 90%.</td>
</tr>
</tbody>
</table>

Performance indicators

Performance indicators were calculated for the entire 2017 birth year. NICU or SCN births are excluded from these calculations.

Table 5: Performance Indicators for Pulse Oximetry Screening in Michigan, Well Baby Nurseries, 2017

<table>
<thead>
<tr>
<th></th>
<th>Total Blood Spots (N)</th>
<th>Reported Screens (n)</th>
<th>Percent Reported (%)</th>
<th>Screens Reported On-Time (n)</th>
<th>Percent Reported On-Time (%)</th>
<th>Timely Screens (n)</th>
<th>Percent Timely Screens (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>STATE OF MICHIGAN</td>
<td>98,729</td>
<td>93,617</td>
<td>94.8</td>
<td>52,474</td>
<td>53.1</td>
<td>36,443</td>
<td>36.9</td>
</tr>
</tbody>
</table>

Overall, 94.8% of babies born in well birth nurseries in 2017 had pulse oximetry values reported to the NBS Program. Just over half (53.1%) of the babies had their first screen reported to the NBS Program within 10 days and 36.9% had the pulse oximetry screen conducted between 20-28 hours of life.
V. Conclusion

NBS is a critical public health program that protects the lives of newborns in our state. One crucial piece of this program is pulse oximetry screening for CCHD. Of the 101,239 infants screened for CCHD in 2017, 71 failed their pulse oximetry screen and six confirmed with a CCHD after a failed screen. Since the pulse oximetry screening mandate was implemented on April 1, 2014, approximately 250 newborns have failed their pulse oximetry screen, leading to 20 CCHD diagnoses. The NBS Program is continually expanding and growing, allow for screening procedures to be refined to better protect the health of Michigan infants.
VI. References


Michigan Algorithm for Pulse Oximetry Screening