Assessing the Current Communication Practices and Physicians' Perceptions Of Newborn Screening in Arizona

by

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ABSTRACT

Every year, hundreds of babies in Arizona are found to have a serious condition identified through newborn screening (NBS), and with current law requiring Arizona to include new conditions to be added to the recommended uniform screening panel (RUSP) within two years of addition to the RUSP, the number of identified babies can be expected to increase. It is essential that physicians are prepared to handle the results of NBS and discuss the implications, in a timely manner, with their patients in order to facilitate treatment. Purpose: To (1) evaluate the current practices and processes of communicating newborn screening results to parents; (2) assess the effectiveness and timeliness of the communication methods used for conveying NBS results; (3) identify potential barriers and challenges associated with the communication of NBS results. Methods: Approval for this study was obtained from the Arizona State University Institutional Review Board. A survey was generated through Qualtrics and Arizona physicians were contacted via email (n = 462). The email contained a link to the survey, or a scannable QR code was provided if the survey was to be accessed via handheld device.

Results: Seventy physicians responded (15% response rate). More than half of the participants often discuss NBS with families prior to conducting the initial screen. 40% of physicians do not feel confident in explaining the purpose and significance of the two newborn screens required by Arizona law. 54% of respondents are not satisfied with the training and resources provided to support them in counseling patients on newborn screening results. 51% of respondents involve a geneticist, other specialist(s), or other experts in the management of an abnormal result. Of the roughly half that do not involve

a geneticist or other specialist when an abnormal result requires follow-up, 50% of those physicians order further genetic testing in office.

Conclusions: Most physicians agree that their greatest barrier to delivering abnormal screening results is using patient friendly language and would prefer language friendly ACT sheets to improve their communication of abnormal screening results. Incorporating personnel skilled in communicating uncertain news, such as a genetic counselor (GC), into Arizona's NBS program could enhance physician communication skills and improve patient satisfaction, while also providing psychosocial support and facilitating follow-up care for patients.

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LIST OF ABBREVIATIONS

AAP ..... American Academy of Pediatrics

ACMG..... American College of Medical Genetics and Genomics

ACT..... ACTion

ADHS..... Arizona Department of Health Services

ASU..... Arizona State University

CDC ..... Centers for Disease Control and Prevention

CORN ..... Council of Regional Networks for Genetic Services

DBS..... Dried blood spot

GC ..... Genetic counselor

HRSA..... Health Resources and Services Administration

IRB..... Institutional Review Board

IOM..... Institute of Medicine

MCH ..... Maternal and child health

NBS..... Newborn Screening

NNSGRC ..... National Newborn Screening and Genetics Resource Center

NSGC..... National Society of Genetic Counselors

OASH..... Office of Disease Prevention and Health Promotion

PCH..... Phoenix Children's Hospital

PKU..... Phenylketonuria

POCT ..... Point-of-Care Testing

RUSP..... Recommended Uniform Screening Panel

CHAPTER 1

INTRODUCTION

1.1 Significance of Newborn Screening

Newborn screening (NBS) serves as a cornerstone of state public health programs, aiming to systematically screen all infants shortly after birth for a spectrum of serious genetic, metabolic, and endocrine disorders. The primary objective is to swiftly identify affected infants early in life, enabling prompt initiation of treatment and management protocols to mitigate morbidity and mortality risks ("Baby's First Test" 2015). However, NBS transcends the confines of a simple blood test; it embodies a sophisticated and integrated system encompassing various components. This comprehensive approach includes timely testing, diligent patient follow-up, outcome tracking, quality improvement measures spanning all facets of the process, and extensive education for healthcare providers, staff, and parents alike (McCandless & Wright, 2020). Historically, the expansion of NBS programs has been fueled by advancements in testing technology. Yet, in contemporary contexts, the evolution of NBS initiatives is increasingly influenced by the development of novel therapeutics and intensified political advocacy efforts. This synthesis of scientific innovation and policy advocacy underscores the dynamic nature of NBS programs and their pivotal role in safeguarding the health and welfare of newborns.

1.2 Physician Responsibilities in Newborn Screening

Physician communication plays a central role in the education of NBS and the delivery of results to parents. Physicians who receive training in pediatrics, family

medicine, or internal medicine are essential in effectively communicating NBS results, providing parental guidance, and educating families about the potential implications of the screening outcomes. The suggested role of primary care physicians extends beyond merely conveying results; they are tasked with assisting families in understanding the diagnosis, symptoms, and potential implications of the condition, as well as the availability of genetic counseling, family testing, and other support services.

Given this central role, it is imperative for physicians involved in NBS be well-informed about the conditions screened for on their state's panel and to recognize that NBS is not diagnostic by itself. False-positive results are inevitable to minimize missed cases, and a negative screening test result should not deter physicians from considering further diagnostic testing for infants displaying symptoms of a metabolic or genetic disorder. Waiting until the next regularly scheduled well check could be detrimental to a baby who has a condition that rapidly worsens, such as galactosemia or congenital adrenal hyperplasia. It is important to treat each result as though that baby is affected and to explain to parents why it is important to act quickly (Percenti, 2019). With each new condition added to the screening panel, the likelihood increases that individual physicians will encounter positive results, reinforcing the importance of ongoing education and awareness.

The pediatrician remains a primary source of education for parents and a valuable guide as families navigate the complex NBS systems. As with any genetic testing, consideration of patients' or parents' values, including cultural values, is essential to assist families in making informed decisions. Effective communication of positive NBS results is a nuanced task that requires careful thought, preparation, and evidence-based

approaches. Research indicates that parental distress can be more influenced by the perceived lack of knowledge of the communicator than by the actual screening result itself (Ulph et al., 2015). Poor or inappropriate communication strategies can have both short-term and long-term effects on parental and familial well-being (Ulph et al., 2015). Therefore, it is essential to handle the initial communication of positive NBS results sensitively, considering individual parent characteristics, to minimize distress and its consequences.

Moreover, parents argue that they have a right to know the risks associated with the heel prick process of obtaining a blood sample, including potential infection and bruising (van der Pal et al., 2022). Given that the standard of care in genetic testing includes pretest counseling, testing, and posttest results delivery with counseling, NBS should not be excluded from this process. Thus, ensuring comprehensive pretest counseling, informed consent, and posttest counseling is vital to uphold the standard of care and meet the expectations of parents regarding NBS.

1.3 Lawsuit

The investigative report by Ellen Gabler titled “The Price of Being Wrong” highlights the critical importance of proper knowledge and communication of NBS results by physicians by telling the story of Mel Russell. On April 8, 2010, just six days after Mel's birth, his pediatrician, Dr. Laurie Grunske, received a concerning result: Mel's newborn screening indicated a "POSSIBLE ABNORMAL" result. This blue piece of paper demanded immediate attention and action.

In adherence to the instructions provided by the Wisconsin State Lab, Dr. Grunske collected another blood sample from Mel for a retest. A week later, the new results arrived, this time on white paper with the comforting label: "NORMAL." However, a closer examination of the values revealed an elevated ratio used to screen for metabolic disorders. Alarmingly, the report lacked any recommendations or instructions for follow-up. Dr. Grunske, with her 25 years of experience as a pediatrician, interpreted "NORMAL" as a reassuring outcome. Little did she know, she was about to find herself at the center of a legal battle over a minute difference of less than two ten-thousandths of a point between Mel's actual result and the state lab's alert value. Had Mel's result remained rounded up, all six of his markers would have been flagged as abnormal, triggering an immediate referral to a metabolic specialist as per the state lab's protocol. However, due to the rounding protocol of the state lab, Mel's result fell just below the threshold, leading to a missed diagnosis and subsequent delays in treatment.

Gabler's report disclosed that at 18 months, Mel suffered a stroke, where further tests in the hospital showed he had propionic acidemia, a disorder where the body can't process certain fats or proteins. Mel likely never will be able to live independently or manage his own diet. If identified at birth, as it is supposed to be, the disorder can be treated with a regulated diet and a child can develop normally (2016).

This story serves as a stark reminder of the critical role that physicians play in the timely and accurate communication of NBS results. Proper knowledge, meticulous attention to detail, and clear communication are paramount to protecting families and preventing morbidity and mortality in infants. The need for comprehensive guidelines,

clear protocols, and effective communication strategies is evident to ensure the safety of newborns and prevent such tragic outcomes in the future.

1.4 Purpose of Study

The study aims to investigate the existing communication practices and physician perceptions regarding NBS in Arizona, given the absence of a national standard and the state-based nature of NBS programs. Historically, there has been limited uniformity between state NBS programs, leading to variations in policies and procedures across states. Physicians involved in NBS are responsible for the timely collection, transport and follow-up of NBS specimens, but there are no standardized guidelines on when or how they should report results to parents. The lack of transparency regarding reporting timelines and the non-mailing of normal results by the Arizona Department of Health Services (ADHS) can lead to parental concerns and potential missed cases.

The recommended uniform screening panel (RUSP) includes two point-of-care (POCT) conditions for NBS: hearing loss and critical congenital heart disease (Grosse et al., 2017). POCT is clinical testing conducted close to the site of patient care where care or treatment is provided (Larkins & Thombare, 2023). This allows parents to be informed of the results immediately before leaving the hospital postpartum. On the other hand, dried blood spot (DBS) screening, which requires time for sample analysis and subsequent communication of results by the physician. This study specifically concentrates on DBS screening and the manner in which physicians communicate these results to parents.

With the expansion of the newborn DBS screening panel and the anticipated increase in demand for genetic services, the study emphasizes the need to assess and improve communication practices of screening results to parents and caretakers. Arizona, with its diverse population and unique healthcare landscape, serves as an intriguing case study for this research, providing valuable insights into potential variations in communication practices across different healthcare settings and regions within the state. Arizona's NBS program has undergone significant changes and improvements, driven by comprehensive needs assessments and protocol revisions. However, the next natural progression in enhancing NBS outcomes lies in evaluating how physicians obtain and communicate NBS results to families.

Therefore, the primary objectives of this research are to:

1) Assess the current practices and processes of communicating NBS results to parents
2) Evaluate the effectiveness and timeliness of communication methods utilized for conveying these results, and
3) Identify potential barriers and challenges associated with the communication of NBS results.

By addressing these objectives, this research aims to provide perspectives that can inform the development of strategies to optimize communication practices and ensure timely and effective dissemination of NBS results, ultimately improving health outcomes for newborns and their families in Arizona.

To understand the importance of this study, a foundational understanding of the history and evolution of NBS in the United States is important, and therefore, will be

discussed. In addition, a focused exploration of the specific context within Arizona will be provided, highlighting the unique challenges and advancements in NBS practices within the state. This comprehensive background aims to set the stage, provide perspective and context that inform the rationale and significance of my study within the broader landscape of NBS initiatives.

CHAPTER 2

BACKGROUND

2.1 History of Newborn Screening in the United States

The genesis of NBS programs traces back to the early 1960s, catalyzed by the pioneering work of Dr. Robert Guthrie. Dr. Guthrie's groundbreaking development of a screening test for phenylketonuria (PKU) and an innovative system for blood sample collection and transportation on filter paper laid the foundation for NBS as we know it today. Massachusetts led the charge by launching a voluntary newborn PKU screening program in 1962, demonstrating the feasibility and potential of mass genetic screening (El-Hattab et al., 2018). This success spurred a wave of advocacy, leading to the passage of laws mandating NBS for PKU in most states during the early 1960s. By 1973, forty-three states had formal statutes in place, with state health departments, particularly their maternal and child health (MCH) programs, assuming a central role in implementation. However, the 1970s brought a recognition of the need to enhance quality assurance in NBS. A proficiency-testing study conducted by the Centers for Disease Control and Prevention (CDC) revealed significant variability among health department laboratories, prompting the establishment of the Newborn Screening Quality Assurance Program at the CDC, with additional funding from the Health Resources and Services Administration (HRSA). Federal legislation in 1976 further supported screening for genetic diseases, with 34 state genetic service programs receiving federal funding in fiscal years 1979 and 1980 (Sepe et al., 1982). This concerted effort at both the state and federal levels marked a vital moment in the evolution of NBS, laying the groundwork for the comprehensive

programs in place today, dedicated to ensuring the early detection and intervention of genetic disorders in newborns.

During the 1980s, significant advancements in NBS systems occurred at both the state and regional levels, in a joint effort to enhance coordination and efficiency. Public health agencies spearheaded the establishment of NBS systems, aiming to streamline communication and collaboration among key stakeholders. These systems were designed to facilitate seamless coordination between hospitals, public health laboratories, pediatricians or primary care health professionals receiving positive results, and pediatric subspecialists responsible for diagnosis and treatment. In response to the growing need for organized efforts, the Council of Regional Networks for Genetic Services (CORN) emerged in 1985 (Therrell et al., 1992). CORN played a noteworthy role in facilitating state genetic program initiatives by fostering coordination and implementing special initiatives. The publication of NBS system guidelines by CORN marked a climatic milestone, outlining an 8-part framework (APPENDIX E) of program-wide mutual agreement. The areas of importance include: organization and administration; selection and evaluation of disorders for screening; communication; quality assurance; funding; diagnosis, management, treatment and counseling; program evaluation; and liability (Therrell et al., 1992). While NBS programs operate on a state-based model, resulting in variations in policies and procedures across states, the guidelines provided by CORN served as a foundational blueprint for public health agencies, offering a structured approach to NBS systems. This era witnessed a shift towards greater standardization and coordination, laying the groundwork for more cohesive and effective NBS programs nationwide. In 1999, CORN was disbanded, and HRSA established the National

Newborn Screening and Genetics Resource Center (NNSGRC) (Ojodu et al., 2017). Since the conclusion of CORN, there has been a lack of a federal oversight body to provide and review guidelines for NBS programs. Instead, the frameworks established by CORN have been incorporated into the foundation of these programs, leading to variations in how state programs operate without national standardization or oversight.

2.2 Complexities of Informed Consent for Newborn Screening

The informed consent process for NBS has been a subject of careful consideration and recommendation by expert committees. The 1994 Institute of Medicine (IOM) Committee on Assessing Genetic Risks emphasized three key criteria for NBS: clear benefit to the newborn, availability of confirmatory diagnosis, and accessibility of treatment and follow-up (IOM, 1994). While the committee acknowledged the appropriateness of mandatory offering of established tests like PKU and congenital hypothyroidism, they put a spotlight on the importance of informed consent to educate parents about the screening process. The IOM report also highlighted that timing of treatment may or may not be crucial, and thus, informed consent should be a part of the decision-making process (IOM, 1994).

Similarly, the 1999 Final Report of the Task Force on Genetic Testing reiterated the importance of NBS benefiting the identified infant and stressed the necessity for analytical and clinical validity and utility of the tests. However, the Task Force differed from the IOM in its stance on informed consent, suggesting that it could be waived if the validity and utility of the tests were established (Holtzman & Watson, 1999). This

nuanced approach to informed consent in NBS reflects the complex considerations surrounding the balance between public health benefits and individual autonomy.

Whether a parent decides to consent or dissent to NBS, it is crucial that the decision-making process involves a comprehensive discussion. This discussion should cover the benefits and risks of screening, potential outcomes of the screening results, available treatments or interventions, and the implications of the decision to either consent or dissent. Ensuring that parents have a clear understanding of these elements is essential for informed decision-making and supports parents in making decisions that align with their values and preferences. The New York Task Force of Life and Law (Task Force) describes this balancing of minimal risk of the test procedure and the significant medical consequences of a missed case suggests "that the autonomy of the parent to make health care decisions for their minor children must give way to the state's role in protecting children from harm" (Task Force, 2000).

2.3 Arizona's History of Newborn Screening

Arizona, despite its early foray into NBS in 1979 alongside Colorado, lagged behind other states in formalizing its program (*Arizona Newborn Screening Panel Guidelines*, 2011). It was not until 1993 that Arizona made momentous strides in this arena. Prior to this milestone, the *Arizona Newborn Screening Panel Guidelines* reports that Arizona relied on the expertise of the Colorado State Laboratory by sending samples across state lines, which conducted screenings for six disorders including sickle cell, PKU, congenital hypothyroidism, homocystinuria, maple syrup urine disease, and galactosemia (2011).

A pivotal moment for Arizona came in 1993 when ADHS established the Office of Newborn Screening, supported by legislative backing, to institute a formal program. This initiative birthed the Arizona Newborn Screening Program, housed within the Bureau of State Laboratory Services under the Division of Public Health Services (*Arizona Newborn Screening Panel Guidelines, 2011)*. With funding sourced from screening fees, the program initially targeted the same six disorders with the addition of biotinidase deficiency that was incorporated into the testing panel in 1989. The Arizona Newborn Screening Program also provided follow-ups for abnormal results. Although a second screening was recommended, it was not mandatory at that time. Arizona's journey to becoming the 47th state to enact NBS reflects a deliberate progression towards safeguarding the health of its youngest citizens.

Arizona's NBS program underwent significant enhancements and expansions in subsequent years. Momentum continued through the early 2000's with legislative mandate of the second screen and the program extended its scope to incorporate follow-up procedures for hearing screening, emphasizing a holistic approach to newborn care. The integration of tandem mass spectrometry testing facilitated the expansion of the panel to encompass the 29 disorders outlined in the RUSP (*Arizona Newborn Screening Panel Guidelines,* 2011*)*. This expansion signaled Arizona's dedication to aligning its screening program with national standards and ensuring comprehensive coverage for newborns. Conditions added to the panel through the decades marked a culmination of efforts to provide thorough and comprehensive screening for Arizona's newborn population. The latest legislation from January 2022 mandates that congenital disorders added to the core and secondary conditions list of the RUSP must be incorporated into

Arizona's NBS panel within two years of their inclusion in the RUSP (S.B. 1680). This addition highlighted Arizona's ongoing dedication to remaining at the forefront of advancements in newborn healthcare, ensuring the early detection and intervention of critical disorders for the state's youngest residents.

2.4 Challenges faced by Arizona's Newborn Screening Program

Throughout 2012-2013, Arizona's NBS program was facing a number of financial challenges brought about by increasing reagent costs, a call for resources to replace aging tandem mass spectrometers, staff turnover, and stakeholders urging us to increase the number of disorders we test for in our NBS panel (Martz et al., 2015).

In November 2013, the Milwaukee Journal Sentinel shook the nation with its investigative report titled "Deadly Delays". The report shed light on a systemic issue plaguing NBS programs across the United States. It revealed that over 160,000 blood samples from newborns were arriving late at labs nationwide, prompting concerns about the timely detection of potentially life-threatening conditions. The report called out the state explicitly saying "Arizona has one of the worst track records in the country, with 17% of NBS samples arriving at the state lab five or more days after collection in 2012" (Gabler, 2013). Some Arizona hospitals even ranked among the worst in the nation for delays.

Following the publication of "Deadly Delays," Will Humble, Director of ADHS, took decisive action. Recognizing the urgency of the issue, he made the NBS program a top priority and assembled a task force to address the problem head-on. Thus, the Transit Time Project was born, with a bold goal: to ensure that 95% of NBS DBS reached the

Arizona State Public Health Laboratory within three days of collection by July 1, 2014 (Transit Time Project, 2014). To achieve this ambitious target, the project team embarked on a comprehensive assessment of hospital policies and procedures statewide, identifying areas for improvement in specimen transport. Synergizing with key stakeholders like the Arizona Perinatal Trust, the Arizona Hospital and Healthcare Association, and the March of Dimes, they disseminated baseline performance data to hospitals, fostering accountability and transparency. Extensive outreach and education efforts followed, equipping hospital staff with the knowledge and resources needed to adapt to the changes introduced by the Transit Time Project.

Critical to the project's success was the establishment of a robust statewide courier system, ensuring timely delivery of specimens to the state laboratory six days a week. Previously, hospitals would often batch samples and send them intermittently instead of every day (Sarah Cox, Certified Genetic Counselor, personal communication, 2024). State laboratory staff also extended their working hours to accommodate the increased volume of tests, including receipt of samples on Saturdays and testing on holidays that create a three day weekend (Fran Altmaier, NBS Follow-up Coordinator, personal communication, 2024). Moreover, efforts were made to align state regulations governing healthcare institutions with NBS rules, streamlining operations and enhancing efficiency. Through coordinated efforts and unwavering dedication, Arizona's Transit Time Project aimed not only to mitigate delays in NBS but also to preserve the health and prosperity of the state's youngest residents. As a result of the collaborative work taken on by stakeholders representing public health, hospitals, interest groups, and private actors, in 5 months, 99% of initial NBS bloodspot specimens collected at birth hospitals were

received at the Arizona State Public Health Laboratory within 1 day of collection (Martz et al., 2015), and 100% were sending samples within four days of collection (Lee, 2014).

2.5 Arizona's Newborn Screening Awards and Recognition

Arizona's NBS program has garnered well-deserved recognition and accolades for its remarkable achievements in enhancing the wellness of newborns statewide. The March of Dimes bestowed its inaugural Newborn Screening Quality Award upon Will Humble, acknowledging his spearheading efforts to expedite the transportation of DBS samples from hospitals to the state laboratory, a feat that earned Arizona national acclaim (2014). Furthermore, in 2016, Arizona's commitment to excellence was reaffirmed when the National Institute for Children's Health Quality reported the state was chosen as one of only 20 states to receive funding from HRSA under the NewSTEPS 360 program, aimed at optimizing the NBS process (NICHQ, 2016). Leveraging innovative approaches such as improving courier services for out-of-hospital births and implementing optical character recognition software for electronic data entry, Arizona has emerged as a trailblazer in newborn health initiatives. Moreover, the state's adoption of the RUSP and the implementation of the two-screen model underscore its dedication to comprehensive screening for genetic and hearing disorders. With an average of 120 newborns identified with genetic disorders and 200 with hearing disorders annually, Arizona's NBS program continues to set the standard for excellence, ensuring a healthier start for the next generation (Wangness, 2023).

2.6 Arizona's Current NBS Process

The Arizona Newborn Screening program encompasses comprehensive DBS screening for 35 rare and serious disorders (Table 1), along with measuring for hearing loss and surveillance of pulse oximetry screening, both vital POCT conducted at hospitals or birthing centers shortly after birth (AHCCCS, 2018). Utilizing just a few drops of blood from the newborn's heel, these screenings detect genetic, endocrine, and metabolic disorders. Hearing loss and congenital heart defects are also included on the NBS but utilize other screening technologies as described previously. While these early screenings do not diagnose disorders outright, they indicate an increased risk, prompting further specialized testing if necessary. Arizona mandates newborns to undergo screening twice: the initial screening occurs within 24 hours of birth or before discharge from the hospital, with midwives often collecting samples within 3 days for babies born outside hospital settings. A second screening is conducted around two weeks post-discharge. Upon identification of abnormal results, infants are promptly referred to specialists for evaluation and treatment. Samples are collected and processed swiftly, with results typically available within 24 hours. Specimens are stored at room temperature for 90 days before undergoing autoclaving to render them unidentifiable, after which they are safely discarded (ADHS Specimen Collection). Finalized results are disseminated via mail and electronically, accessible to hospitals and physicians for further action.

2.7 Arizona's Physician Responsibilities Guidelines

The *Arizona Newborn Screening Panel Guidelines* describes responsibilities for physicians who are involved with NBS: 1. Guarantee the timely and accurate collection of NBS specimens, 2. Facilitate rapid transfer to the State Lab, and 3. Follow-up on

abnormal results. First, physicians must order NBS tests for each newborn and educate parents about the importance of screening. They are encouraged to distribute informational brochures to parents and document any refusals for testing. Second, physicians must complete specimen cards accurately and collect acceptable DBS samples following specified guidelines. They should adhere to recommended timelines for specimen collection, ensuring samples are obtained within 24 to 72 hours of birth or before discharge from the hospital (2011).

As stated above, physicians are responsible for sending specimens promptly to the ADHS and documenting NBS test results in the baby's medical records. In the event of positive results, physicians must initiate appropriate follow-up measures, including additional testing and specialist referrals if necessary. Moreover, physicians are required to submit results from subsequent tests performed at other labs to the NBS program for comprehensive monitoring and evaluation (*Arizona Newborn Screening Panel Guidelines*, 2011). These conscientious steps reinforce Arizona's commitment to protecting the health of its youngest residents through an effective NBS program.

2.8 Arizona's Protocol for NBS Consent

The consent process for NBS revolves around the concept of autonomy, which encompasses the freedom to choose for oneself and issues of informed consent. While the American Academy of Pediatrics (AAP) has recommended since 2001 that parental informed consent should be standard practice for NBS, the reality is that most states employ a system of passive consent. This means parents can opt out of NBS by signing a waiver, but are not explicitly required to provide written consent before the testing.

Informed consent plays a crucial role in the NBS process, ensuring that parents are fully informed about the benefits and potential risks associated with testing their newborns for serious medical conditions. In Arizona, healthcare physicians are mandated by law to order NBS for all infants born in the state for potentially fatal conditions. Parents who choose to refuse the screening after being informed of its benefits and associated risks may be asked by their physician to sign a waiver, acknowledging their responsibility for any adverse consequences resulting from this decision. However, it's important to note that, currently, a waiver is not legally required. Any refusal of screening is documented on the specimen collection card and reported to the Newborn Screening Follow-Up Program, as per Baby's First Test guidelines from 2015. Currently, religious exemptions are the only valid reason for refusing NBS ("Baby's First Test" 2015).

CHAPTER 3

MATERIALS AND METHODS

3.1 Participants

Approval for this study was obtained from the Arizona State University (ASU) Institutional Review Board (IRB)(Appendix A). The population of interest for this study were licensed physicians that practice medicine in the state of Arizona who are 18 years and older. Participants were required to have been involved with the ordering, interpreting, and communicating of NBS. No other exclusionary criteria applied.

3.2 Research Methods

The identification and recruitment of participants for this study employed a multifaceted approach, utilizing several methods to reach the target population. First, recruitment efforts were facilitated through prominent healthcare institution websites identifying physician emails, including the Phoenix Children's Hospital (PCH) website, the Banner Health website, and the HonorHealth website. These platforms served as valuable resources for disseminating information about the study and encouraging participation among eligible individuals. Additionally, recruitment initiatives were extended to professional networks within the medical community, with targeted outreach through the Arizona Medical Association Newsletter. A pediatric resident, Rasa Tiano, was instrumental in getting responses amongst her peers at PCH.

Arizona physicians were contacted via email. The email (APPENDIX B) and the flier (APPENDIX C) were abbreviated versions of the summary and importance of the

study and explained the eligibility criteria for participation. The email then concluded with a link to the survey, or a scannable QR code was provided if the survey was to be accessed via handheld device. The contact information for the lead researcher and the principal investigator was provided in the email and the flier. Both email and flier were distributed via the internet; therefore, links provided were clickable and would open directly to the consent form. Formal consent was obtained directly at the beginning of the document prior to the start of the survey. The consent outlined in further detail the information in the email and flier. A thorough description of the study was also included. Participants could then select "I consent, begin the survey", and they were directed to start the survey. Non-responses were not included in the dataset.

3.3 Survey Development

The survey (APPENDIX D) consisted of 37 total questions. Qualtrics survey platform was used in development of the survey and consisted of 7 main sections. The first section consisted of multiple-choice questions pertaining to demographics. The second section included questions regarding their involvement in the NBS process, if they participate in an informed consent, and the time and method of communicating NBS results. The third section addressed their encounters with NBS results, materials and strategies used in result follow-up, and satisfaction of training and resources. The fourth section of the survey investigated the challenges encountered, preferred resources utilized, provision of patient support materials, and cultural awareness practices during discussions about NBS results with physicians. The last section of the survey contained

two open-ended questions for the participants to add any additional thoughts or experiences related to communicating NBS results.

The survey used skip logic, as well as branching, to personalize the survey based on how participants answered a question. Three genetic counselors and a NBS coordinator with expertise in NBS and survey development reviewed the survey.

3.4 Data Collection

The survey was open to participants from July 2023 to January 2024. Access to the survey was through a web link and/or a QR code given to participants via an email or flier. A reminder was sent to unresponsive participants after three weeks of no progress. Data was collected anonymously through the Qualtrics system.

3.5 Data Analysis

Raw data were input into Microsoft Office Excel from Qualtrics for descriptive statistical analysis. Daniel Coven, a biostatician employed by ASU was consulted for direction and statistical recommendations on the data collected. Free-text questions were analyzed for themes. Themes were coded and analyzed for frequency using a grounded theory approach. Tables and figures were constructed with Microsoft Office Excel.

CHAPTER 4

RESULTS

4.1 Demographics

The survey was distributed to 462 physicians who practice in the state of Arizona. Because the survey was distributed randomly, the specialty breakdown of those 462 is not known. Of the estimated 462 physicians contacted, 70 responded resulting in a response rate of 15%. 57 participants utilized the QR code, 9 accessed the survey via email, and 4 participants used the anonymous link to access the survey. Two of those physicians that consented were not licensed in the state of Arizona, three participants that are licensed in Arizona are not involved in the NBS process, and five participants did not complete the survey. Due to their responses, the survey was instructed to close. The total number of surveys analyzed was 60.

Forty-three participants were female (71.67%) and seventeen participants were males (28.33%). The majority of respondents were between the ages of 25 and 34 years old, five were reported to be between the ages of 35 and 44 years old, five were reported to be between the ages of 45-54 years old, two were reported to be between the ages of 55 and 64 years old, and one reported to be 65 years or older. Ages reflect the participants' age at the time of survey completion. Predominantly, the surveyed physicians were based in Maricopa county (57/60, 95.0%) while the remaining three licensed physicians practice in Pima county (3/60, 5.0%). The majority of participants had received medical training in pediatrics (56/60, 93.33%), one licensed physician was trained in family medicine (1/60, 1.67%), and four physicians reported to have received

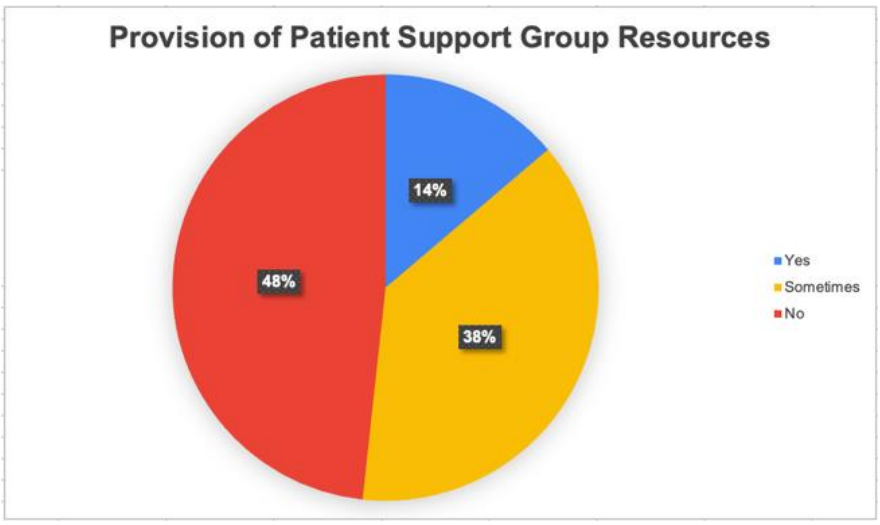
training in both pediatrics and internal medicine (3/60, 5.0%). Among the physicians surveyed, 48 respondents reported having less than five years of experience, while three indicated having 5-10 years of experience, and another four reported having 10-20 years of experience. Five participants stated they have 20 or more years of experience.

4.2 Current Communication Practices

More than half of the participants often discuss NBS with families prior to conducting the initial screening. On a scale of 1 to 5 with 5 being very confident, a majority of respondents, comprising 43% of the sample, rated their confidence level as a 4, indicating a substantial level of confidence in explaining NBS. About 40% of respondents rated their confidence level as a 3 or lower in explaining NBS to families reflecting a moderate to minimal level of confidence in explaining NBS to families.

Most physicians (91%) report normal NBS results to parents. Of the physicians who disclose the normal NBS results, the majority discuss the results during in-person appointments, and all of the participants allocated less than 30 minutes to the discussion. Most physicians (93%) report abnormal NBS results to parents. Of the physicians who disclose the abnormal NBS results, the majority discuss the results via phone calls with reiteration during in-person appointments, and nearly all of the participants allocated less than 30 minutes to the discussion.

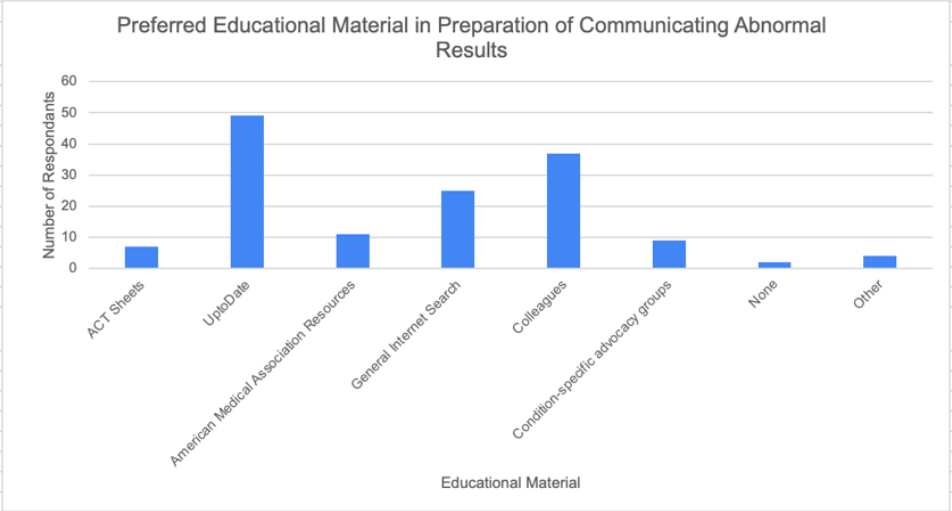
Nearly half of all respondents do not provide resources to support groups for those families that have abnormal screening results.



4.3 Effectiveness and Timeliness of NBS Results

When reporting normal NBS results, physicians who contacted patients within a week versus more than one week was about equal (52% versus 48%). While the time for reporting normal NBS results was equally split between respondents, the majority of physicians reported the abnormal results within three days (51 out of 58).

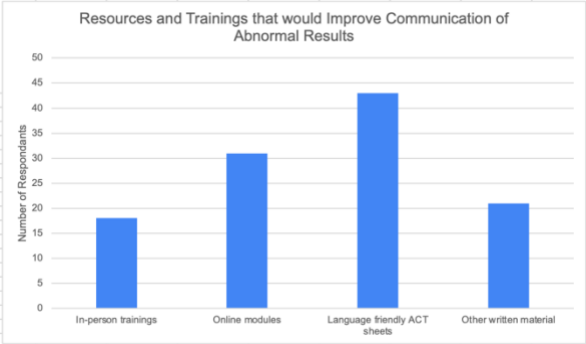
The main support material used by surveyed physicians to assist with how to communicate abnormal NBS results is a subscription-based resource designed to provide physicians access to current clinical information called UpToDate (Garrison, 2003). Asking colleagues was the second most utilized resource used by surveyed physicians. In a free-text option, one physician responded utilizing some handouts available from the ADHS.



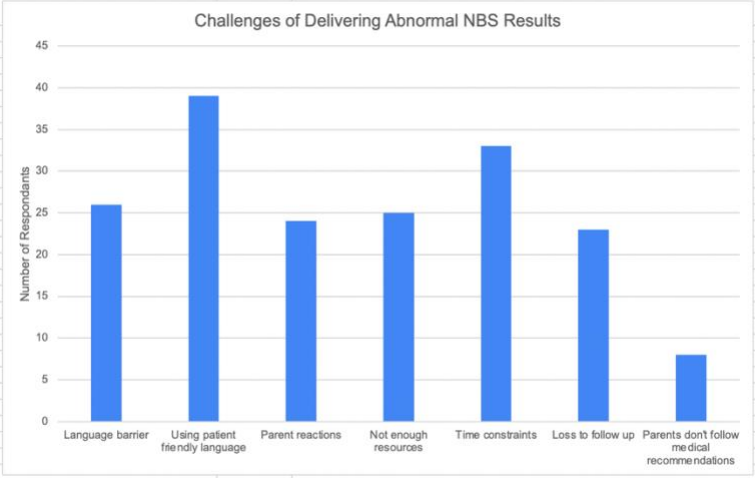
The most commonly employed strategies by physicians to assist patients in understanding the implications of abnormal genetic screening results were the use of an interpreter, scheduling an in-person follow-up appointment, and referring patients to a geneticist. 51% of respondents involve a geneticist, other specialist(s), or other experts in the management of an abnormal result. Of the roughly half that do not involve a geneticist or other specialist when an abnormal result requires follow-up, 50% of those physicians order further genetic testing in office.

4.4 Potential Barriers and Challenges

In the study, a significant majority of the surveyed physicians (69%) indicated that they receive fewer than 5 abnormal NBS results annually. Regarding the satisfaction with training and available resources, 60% of the respondents expressed a neutral or dissatisfied stance. Additionally, there was a consensus among participants that the introduction of language-friendly ACT sheets could enhance their communication skills when conveying abnormal results.



The primary challenges faced by physicians when communicating abnormal NBS results were identified as using patient-friendly language and managing time constraints.



Despite the reported lack of formal training on culturally sensitive communication in the context of NBS by 86% of the respondents, 85% acknowledged considering cultural factors when discussing NBS results with parents.

4.5 Open-Ended Responses

The open-ended responses from the survey offer valuable insights into the challenges and considerations faced by physicians in Arizona when communicating NBS results. Some physicians highlighted the additional burden that postpartum families face, emphasizing the importance of providing support in setting up transportation and

ensuring necessary follow-ups to alleviate the added stress of additional appointments and blood draws. Others expressed concerns about the lack of comprehensive information and clear recommendations for abnormal screening results, indicating a need for more accessible and easy-to-understand information for both physicians and parents.

Suggestions for improving communication included the development of video training with acted-out examples, next steps flowsheets for abnormal results, and providing educational visits with specialists for specific diagnoses like sickle cell trait. Some physicians also stressed the importance of tailoring the communication approach based on the family's needs and the type of abnormality detected.

A final theme noted is, a few respondents mentioned the need for efficient handling and coordination of additional testing and referrals, highlighting the importance of streamlining processes and leveraging available resources within their clinics. The complex nature of communicating NBS results is obvious by these responses and emphasize the need for tailored, accessible, and supportive approaches to effectively communicate with families during this critical time.

CHAPTER 5

DISCUSSION

5.1 Current Practices and Processes of NBS Result Communication

In the realm of NBS in the United States, there exists a notable discrepancy in reporting requirements between physicians and laboratories. While the Office of Newborn Screening has established specific time frames for NBS systems to communicate results to healthcare physicians, there is a lack of federal and state mandates compelling physicians to relay these critical results to parents in a timely manner. Currently, HRSA states that "All NBS tests should be completed within seven days of life with results reported to the healthcare provider as soon as possible" (2017). However, there are no established guidelines for physicians regarding the timing of notifying caregivers or parents about the NBS results. This disparity reveals a gap in the responsibilities physicians should abide by and showcases the need for enhanced regulatory measures to ensure consistent and timely communication of NBS results to parents, thereby facilitating timely diagnosis, treatment, and improved healthcare outcomes for newborns.

5.2 Informed Consent in NBS Process

A significant majority of participants, over half, engage in discussions about NBS with families before conducting the initial screen. This proactive approach likely helps build trust and prepares families for potential outcomes.

Informed consent for NBS is an ethical consideration that often catches many parents off guard when they receive screening results requiring further diagnostic testing. The procedure involves a heel prick to obtain a DBS sample from the newborn, which carries inherent risks such as infection and bruising. Many parents argue that they have the right to be fully informed about these potential risks, akin to any other medical procedure (Beal et al., 2014). While the importance of public education in NBS has been well documented, there are currently no specific guidelines on the best way to ensure that parents understand NBS' role in disease detection. With the expansion of NBS and advancements in genomic technologies enabling the screening for a broader range of conditions, the need to educate parents about the process and actively involve them in the informed consent process becomes increasingly apparent. As emphasized, "Meaningful parental understanding about NBS is a prerequisite for informed consent, but the importance of education is relevant even where screening is mandated (Etchegary et al., 2016). Interestingly, studies have shown varying perspectives among healthcare providers regarding the consent process. While some providers, such as midwives, view obtaining consent as relatively straightforward and advocate for an informed consent approach, others, like pediatricians, perceive the process as time-consuming and impractical (Beal et al., 2014). Nevertheless, there is a growing consensus supporting the preference for full disclosure of all pertinent information, with some parents valuing this transparency even more than having a choice in the matter. The National Society of Genetic Counselors (NSGC) supports opt-out NBS programs, or conversely, opposes policies requiring parents to opt in to NBS. NSGC defends their stance by arguing that opt-in screening policies could result in considerable medical harm to babies who are not

screened because their parents are confused or misinformed about the consent process due to insufficient provider education. Opt-in programs also undermine the public health function of NBS and have the potential to introduce unnecessary health disparities (Blout et al., 2014).

5.3 NBS Results Disclosure

When it comes to disclosing the screening results, the majority of physicians surveyed in this study, 91%, report normal NBS results to parents, indicating a consistent practice of transparency that may alleviate parental anxiety and foster confidence in the healthcare system. Interestingly, both normal and abnormal results are predominantly discussed during in-person appointments, emphasizing a preference for direct and personal communication. The Office of Disease Prevention and Health Promotion (OASH) states that babies need to go to the doctor or nurse for a "well-baby visit" 6 times before their first birthday (2023). This directive could also explain why results are typically disclosed during in-person appointments. However, the data raises a potential concern as nearly all participants allocate less than 30 minutes for these vital discussions, raising questions about the adequacy of time to convey comprehensive information and address parental concerns effectively.

There is a pressing need to allocate more time, beyond the current 30-minute average, for discussions about NBS results. Given that physicians already cite time constraints as their primary challenge, increasing the time allocated for discussing NBS appears to be unrealistic. Primary care providers interact with the family for a median of

16.3 minutes (LeBaron, 1999) and see approximately twenty patients a day, expecting more than 30 minute allotments to discuss NBS results may not be feasible.

The discovery that nearly half of the respondents do not offer resources to support groups for families with abnormal NBS results reveals a concerning gap in the support and resources available during this critical and potentially distressing period. The lack of awareness or access to local support groups and services among physicians handling NBS results should be further explored. It is imperative to recognize that families facing abnormal screening outcomes may require specialized guidance, emotional support, and information to navigate this challenging situation effectively. Local support groups and resources can play a powerful role in providing families with the necessary support and connections to professionals who can offer expert advice and assistance.

5.4 Effectiveness and Timeliness of NBS Communication Methods

More than half of the physicians contact parents within a week of receiving normal NBS results. However, it is concerning that 48% of respondents wait more than one week to communicate these normal results to parents, which might cause unnecessary anxiety and uncertainty for families during this waiting period.

Conversely, when it comes to abnormal NBS results, there is a more concerted effort to contact parents within three days. This timely communication is crucial given the potential implications of abnormal results.

The main educational material utilized by surveyed physicians for communicating abnormal NBS results is a subscription-based resource called UpToDate, designed to

provide access to current clinical information (Garrison, 2003). This resource is followed by colleagues as the second most utilized support material. Only one physician mentioned utilizing handouts from the ADHS, indicating potential gaps in the availability or awareness of state-provided resources. While UpToDate serves as a valuable resource, there is a need to diversify and expand the range of support materials available to physicians, including state-provided resources and handouts, to ensure comprehensive and accessible information for families.

Furthermore, the strategies employed by physicians to assist patients in understanding the implications of abnormal genetic screening results predominantly include the use of an interpreter for non-English speakers, scheduling in-person follow-up appointments, and referring patients to a geneticist. These strategies reflect a commitment to personalized care and the recognition of the complex nature of genetic screening results, which often require specialized knowledge and expertise to interpret and explain effectively to patients.

Despite these strategies, it is concerning **that only 51% o**f respondents involve a geneticist or other specialists in the management of abnormal results, while the remaining half either order further genetic testing in-office or do not involve specialized professionals at all. This suggests that a substantial portion of physicians either rely solely on in-office genetic testing, which may not provide comprehensive information or insights into the implications of abnormal results, or do not involve specialized professionals at all, potentially leaving patients without the necessary expertise and support to understand and navigate the complexities of their results.

To improve understanding and support for families receiving abnormal results, there should be an emphasis on involving geneticists or other specialists in the management and follow-up of abnormal results. This will be discussed more in the recommendations section. The significance of these findings lies in the potential implications for patient care and outcomes. Inadequate or inconsistent management of abnormal genetic screening results can lead to misunderstandings, increased anxiety, and delayed or inappropriate interventions.

5.5 Confidence of Explaining Significance of NBS

A barrier in the process of explaining NBS results, is the level of confidence physicians have in their ability to explain the results. The results shed light on the varying levels of confidence among physicians when discussing and explaining NBS to families. A combined analysis of the confidence levels reveals that a significant majority of respondents, rating themselves at either 4 or 5 on a scale ranging from 1 to 5, demonstrate a high level of capability and comfort in communicating NBS information effectively to families. This indicates that most physicians feel well-prepared and confident in explaining the importance and procedures of NBS, demonstrating their readiness to educate families about early detection and intervention in potential health conditions.

When combining the confidence levels of 3 and 1 collected from the responses, we find that a combined 40% of the respondents rated their confidence in explaining NBS as either moderate (33% at level 3) or minimal (7% at level 1). This indicates that a significant number of physicians may have reservations or lack confidence in communicating this critical information to families.

The mixed confidence levels among respondents raises questions about the adequacy, accessibility, and effectiveness of current training and resources available to physicians to enhance their confidence and proficiency in communicating NBS information to families. While a majority of physicians feel well-equipped to handle these conversations, addressing the concerns of those with lower confidence levels is essential to ensure consistent and effective communication across the board. By providing additional resources and training opportunities, healthcare organizations can help improve confidence levels and ensure that all families receive clear and accurate information about NBS.

5.6 Additional Barriers and Challenges with the Communication of NBS Results

A notable majority of physicians (69%) reported receiving a relatively low number of abnormal NBS results annually, which could potentially limit their experience and confidence in handling such cases effectively. Limited exposure to abnormal results can impact physicians' confidence, expertise, and preparedness in managing and communicating these complex and sensitive findings to patients and their families. Without adequate exposure and experience, physicians may struggle to navigate the intricacies of abnormal screening results, leading to potential misunderstandings, increased anxiety among families, and suboptimal patient care. This limited exposure might be further exacerbated by the reported dissatisfaction with available training and resources, with 60% of respondents expressing a neutral or dissatisfied stance. The results

suggest that current educational and support mechanisms may be inadequate, outdated, or not effectively meeting the needs of NBS physicians.

Despite these challenges, there was a consensus among participants regarding the potential benefits of incorporating language-friendly ACT sheets to improve their communication skills when delivering abnormal results and supplemental algorithms with the proper protocol for result follow-up and treatment.

Despite the reported lack of formal training on culturally sensitive communication in their medical programs, it is encouraging to note that a significant majority (85%) of physicians consider cultural factors when discussing NBS results with parents. This encouraging statistic demonstrates physicians' awareness, efforts, and willingness to accommodate and respect diverse cultural backgrounds . This proactive approach to considering cultural factors can help build trust, enhance patient-physician relationships, and improve communication effectiveness, ultimately leading to better patient understanding, satisfaction, and healthcare outcomes.

The significance of these findings lies in their potential impact on improving cultural competence, communication effectiveness, and patient-centered care in healthcare settings. By recognizing and accommodating cultural differences, physicians can create a more inclusive and respectful healthcare environment, where patients and their families feel valued, understood, and supported.

5.7 Recommendations

These recommendations outline key strategies aimed to enhance the communication and management of NBS results, focusing on critical areas such as time

allocation, training and understanding among physicians, resources for physicians, resources for patients and families, and cultural competency. By implementing these targeted recommendations, healthcare organizations can improve the quality of care, enhance communication effectiveness, and ultimately optimize outcomes for families undergoing NBS.

As mentioned above, there is a need to establish comprehensive federal guidelines that provide clear protocols for physicians on when and how to communicate screening results, the information to be conveyed, and strategies to support parents psychosocially during this process. Implementing standardized follow-up procedures for abnormal results, ensuring timely and appropriate support and guidance for parents throughout the screening process and any subsequent diagnostic evaluations or treatments, is essential. Regularly monitoring and evaluating physician communication practices regarding NBS results can help identify areas for improvement and ensure adherence to guidelines. While it may be unrealistic to extend the current 30-minute average consultation time due to physicians' busy schedules, encouraging pre-screen discussions by perinatal providers can prepare families for potential outcomes and foster trust.

The ADHS can bolster physician training by offering targeted programs aimed at enhancing their knowledge and communication skills regarding NBS. This can also be balanced by physician interest and attendance at educational opportunities. Such training could be delivered through online modules or videos that simulate the NBS process from beginning to end. This presents a unique opportunity for the state laboratory to distinguish itself by supporting physicians with essential resources and information, equipping them to effectively communicate and provide care to their patients.

Personalizing communication approaches to individual parent characteristics, such as cultural background, educational level, and emotional readiness, can help minimize distress and enhance understanding. This training should equip physicians with the necessary knowledge and skills to communicate effectively with families from diverse cultural backgrounds, ensuring clear, respectful, and culturally appropriate information dissemination. Utilizing evidence-based communication methods that have proven effective in reducing parental distress and improving outcomes when conveying positive NBS results is crucial. Providing physicians with comprehensive and up-to-date educational materials, brochures, and resources can further support their communication efforts and enhance parental understanding.

The American College of Medical Genetics (ACMG) has compiled informational guides on genetic conditions that healthcare physicians can consult for quick insights into the condition, diagnosis, and subsequent steps. For each marker(s), there is 1) an ACTion (ACT) sheet that describes the short term actions a health professional should follow in communicating with the family and determining the appropriate steps in the follow-up of the infant that has screened positive, and 2) an algorithm that presents an overview of the basic steps involved in determining the final diagnosis in the infant (ACMG ACT Sheets and Algorithms, 2001). The ACMG ACT sheets are provided to the physician by the NBS program with the abnormal results (Fran Altmaier, NBS Follow-up Coordinator, personal communication, 2024). ACT sheets tailored to Arizona's specific NBS panel can serve as invaluable one-page clinical decision support tools for physicians. By incorporating patient-friendly language, these sheets can facilitate clearer communication and enhance understanding for both physicians and families. These ACT sheets, designed

to address genetic conditions identified through NBS and beyond, can be readily accessible on the ADHS website. The supplemental inclusion of treatment algorithms can further assist physicians in determining appropriate testing to confirm a diagnosis, follow-up care, and interventions. Disseminating these sheets to local physicians involved in NBS ensures widespread availability and utilization. It is imperative for physicians to be aware of and utilize this resource for quick reference when encountering abnormal NBS results, thereby supporting timely and informed clinical decision-making.

Given the apparent lack of awareness or access to crucial support systems among respondents, there is a pressing need to explore and compile a comprehensive list of available services, support groups, and resources. Creating a centralized database or resource guide that physicians can easily access and refer families to can help bridge this gap and ensure that families receive timely and appropriate support. By raising awareness and facilitating access to local support groups and resources, physicians can better support families during this challenging period, ultimately enhancing the overall experience and outcomes for families undergoing NBS. Addressing this gap and promoting collaboration between physicians and local support groups can substantially improve the support available to families, emphasizing the importance of enhancing awareness and accessibility to vital resources in healthcare settings.

Moreover, employing genetic counselors (GC) at state labs to act as liaisons for physicians can facilitate smoother communication and enhance the quality of information provided to physicians. In general, a GC's role involves providing information on genetics, inheritance, and specific genetic conditions. They discuss the benefits, risks, and limitations of genetic testing, interpret test results, and assist patients in making informed

decisions. Additionally, they offer support to patients, their families, other healthcare providers, and the community (Resta et al., 2006).

State laboratories can effectively utilize GCs in NBS to enhance follow-up and support. GCs play a central role in developing educational materials for parents and physicians, either independently or in collaboration with other NBS professionals. They source specialized parent support and provide educational materials for complex disorders to accompany abnormal screening results. Additionally, they assist physicians in navigating newly added conditions to the screening panel and provide guidance on patient-specific results. GCs could act as consultants for physicians seeking advice on specific conditions and address parent questions when primary doctors are not available or comfortable discussing certain results. In cases where a physician has not been established, or at the request of a physician, GCs directly report results to parents. With their training in empathy and cultural sensitivity, GCs help alleviate anxiety by providing context to results, supporting both physicians unfamiliar with the screening process and families during the diagnostic testing waiting period. A GC could initiate the process for additional testing and place referrals as needed, when appropriate.

The themes identified in the results of physicians' perceptions, such as time constraints, dissatisfaction with educational materials, awareness of local support groups, and the provision of patient support material, encompass the multifaceted role of a GC. GCs are equipped to address these challenges by providing comprehensive support and guidance. They can assist physicians by offering tailored, time-efficient educational materials and resources. Furthermore, GCs are well-positioned to connect patients with local support groups and provide personalized patient support materials. By addressing

these themes, GCs play a crucial role in enhancing the communication and understanding of genetic information, thereby facilitating more effective patient care and support.

By implementing these recommendations, it is anticipated that physician communication of NBS results will be significantly improved, leading to better parental understanding, reduced distress, and improved overall outcomes for children and families.

5.8 Limitations

My research stemmed from an initial group discussion with families whose children were identified with PKU through NBS. This conversation inspired me to explore further and engage with more families whose children were diagnosed with various conditions via NBS. The objective was to understand the communication process: who contacted them, how they were notified, and what information they were given. Essentially, I aimed to evaluate parents' perceptions of how NBS results are communicated in Arizona, seeking comprehension into what parents would consider most helpful when informed of abnormal results by their physician.

However, identifying and reaching out to families proved challenging. Despite my efforts to contact every organization that had an Arizona-specific condition listed on the NBS panel, I either received no response or was informed that third-party surveys could not be disseminated. Fortunately, the Arizona Network for PKU and Allied Disorders (ANPAD) agreed to distribute the survey. I am grateful to the 12 participants who responded, as their feedback helped shape specific questions for my current physician survey. My subsequent attempt to obtain IRB approval through ADHS and ASU’s

Honest Program encountered delays that did not align with my graduation timeline. Despite these setbacks, the challenges I encountered have deepened my understanding and appreciation for the complexities involved in research.

One significant limitation was the low response rate of 15%, which could have been influenced by budgetary restrictions that prevented the provision of incentives or reminder letters to participants, potentially affecting the representativeness of the sample and making data comparisons challenging. Accessing physicians is difficult at best, as much of the external electronic communication is through patient portals. Emails are often not advertised or kept confidential. Third-party websites that claimed to have physician contact information had an unfeasible cost, ranging in the thousands of dollars for a one-time email.

Another potential limitation relates to the age and experience of the physicians participating in the study, as newer trainees might have different communication practices compared to those with more experience. Additionally, the omission of exploring participants' beliefs regarding the responsibility of informing parents about NBS by obstetricians could shed light on the existing dynamics, responsibilities, and potential areas for improvement in the communication of NBS information. Obstetricians play a crucial role in prenatal care and may be the first point of contact for expectant parents, making their views on this topic particularly relevant.

Specifically, the study did not assess the timing of initial discussions about NBS, particularly in relation to when parents meet with a pediatrician either before delivery or prior to the screening test being conducted. This is particularly relevant considering that while over three-quarters of pediatricians offer prenatal visits, surveys indicate that only

5% to 39% of first-time parents attend one, highlighting potential disparities in access to prenatal care and education about NBS (Stamford, 2021).

5.9 Future Directions

Assessing the current communication practices of physicians in relaying NBS results not only provides an understanding into the existing practices and challenges, but also offers direction for future research and improvements in healthcare delivery. Further research could explore how NBS is taught to residents in training, including the communication of results, how to effectively convey NBS results to patients, and assessing potential psychosocial implications in order to identify specific areas for targeted training and support. Additionally, conducting surveys of follow-up NBS labs who do or do not employ GCs could help evaluate the effectiveness and efficiency of current communication practices and identify opportunities for improvement. There is also a need to explore the awareness and satisfaction levels of pediatricians and primary care providers regarding genetic services, including specific feedback on what could be improved and how to optimize the communication process in Arizona. Further advocacy efforts are essential to emphasize the importance of optimizing communication services, ensuring that families receive clear, timely, and supportive information about NBS results to make informed decisions and navigate the healthcare system effectively. By addressing these areas and investing in continuous research, training, and advocacy, healthcare systems can enhance communication practices, improve patient satisfaction, and ultimately contribute to better health outcomes for newborns and their families.

CHAPTER 6

CONCLUSION

The study emphasizes the critical need for comprehensive education among physicians and parents regarding the NBS process to enhance early detection and treatment of infants with metabolic or genetic disorders. Recognizing the emotional distress and financial implications linked to delayed diagnosis, it is imperative for physicians to be well-versed in NBS and to approach each screening result with the utmost urgency.

While NBS programs are state-specific and poised for expansion with advancements in genetic testing and gene therapy, the absence of federal standardization presents challenges. This lack of uniformity extends to the criteria for adding conditions to screening panels. Despite improvements in courier services and laboratory turnaround times, there's a clear need for enhanced physician communication regarding NBS results. The current absence of federal guidelines leaves physicians without clear directives on result communication, information provision, or psychosocial support for parents navigating abnormal results.

Navigating the communication of positive NBS results is a complex and sensitive task, demanding precise planning, preparation, and evidence-based approaches. Initial communication of positive NBS results should be approached with sensitivity, accounting for individual parent characteristics and reflecting the severity of the identified condition and required response urgency to mitigate distress effectively.

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APPENDIX A

ARIZONA STATE UNIVERSITY IRB APPROVAL

ASU Knowledge Enterprise Development

EXEMPTION GRANTED

Katherine Hunt Brendish
CHS: Health Solutions, College of
480/314-7437
KHuntBrendish@asu.edu

Dear Katherine Hunt Brendish:

On 8/25/2023 the ASU IRB reviewed the following protocol:

Type of Review:	Initial Study
Title:	Assessing the Current Communication Practices and Physicians' Perceptions of Newborn Screening in Arizona
Investigator:	Katherine Hunt Brendish
IRB ID:	STUDY00018511
Funding:	None
Grant Title:	None
Grant ID:	None
Documents Reviewed:	• Physician Communication of NBS results Protocol, Category: IRB Protocol; • Physician NBS Survey Consent, Category: Consent Form; • Physician NBS Survey Questions, Category: Measures (Survey questions/Interview questions /interview guides/focus group questions); • Physician Recruitment Material, Category: Recruitment Materials;

The IRB determined that the protocol is considered exempt pursuant to Federal Regulations 45CFR46 (2)(ii) Tests, surveys, interviews, or observation (low risk) on 8/25/2023.

In conducting this protocol you are required to follow the requirements listed in the INVESTIGATOR MANUAL (HRP-103).

If any changes are made to the study, the IRB must be notified at research.integrity@asu.edu to determine if additional reviews/approvals are required. Changes may include but not limited to revisions to data collection, survey and/or interview questions, and vulnerable populations, etc.

Sincerely,

IRB Administrator

cc: Tessa Nelson
Tessa Nelson
Katherine Hunt Brendish

APPENDIX B

RECRUITMENT EMAIL

Good morning Doctor,

On average, over 200 infants are born per day in the state of Arizona. Newborn screening is performed on most of these infants to help identify conditions that may affect the child's long-term health or survival. It is widely understood that the timeliness of testing and communication is essential to the child's quality of life. You are receiving this message because you are eligible to participate in a research study about the Current Communication Practices and Physicians' Perceptions of Newborn Screening in Arizona. We are trying to learn more about what barriers and challenges there may be with communication of newborn screening results. Participation is voluntary and you have the right to opt out at any time. No information will be gathered that could personally identify you. The survey takes about 10-15 minutes to complete. If this sounds like an exciting opportunity please click here , or follow the link **https://asu.co1.qualtrics.com/jfe/form/SV_bsjsKnyUavdcY9o**, for more information or scan the QR code attached at the bottom. Thank you for your time and consideration in helping us answer this important question.

Sincerely,

Tessa Nelson
Arizona State University
Genetic Counseling Master's Program
Tknelso5@asu.edu

Katherine Hunt Brendish
Arizona State University
Genetic Counseling Master's Program
Khuntbrendish@asu.edu

APPENDIX C

RECRUITMENT FLIER

Physician Responses Needed for Research Study on Newborn Screening in Arizona

Thank you for your interest in participating in this research study.

This survey is a part of a Masters thesis research project to assess the current communication practices and physicians' perceptions of newborn screening results in Arizona.

Participants: Physicians licensed in Arizona that facilitate, interpret, and/or communicate newborn screening. Participation is voluntary and response are anonymous.

When: At your earliest convenience. The survey will take 10-15 minutes to complete.

Questions?
You can contact me, Tessa Nelson, via e-mail at tknelso5@asu.edu. Katherine Hunt Brendish is the Chair of my thesis committee available at khuntbrendish@asu.edu.

To access the survey please click following the link: https://asu.co1.qualtrics.com/jfe/form/SV_bsjsKnyUavdcY9o

Scan the QR code below to complete survey my mobile device

Genetic Counseling Master's Program
ASU Health Futures Center
6161 E Mayo Blvd.
Phoenix, AZ 85054

ASU Arizona State University

APPENDIX D

SURVEY

Informed Consent

Welcome to the research study!

Thank you for your interest in this research study! I am a genetic counseling graduate student at Arizona State University. I am conducting a research study to assess Arizona's current communication of newborn screening results and evaluating physicians' perceptions. Please read below to find out more information about this study:

Newborn screening (NBS) is a critical public health program aimed at identifying infants who may have certain genetic or metabolic disorders early in their lives. The timely and effective communication of NBS results from healthcare providers to the parents is crucial for ensuring appropriate follow-up, early intervention, and improved health outcomes for affected infants. This research protocol outlines a study designed to assess the communication of newborn screening results and investigate the potential areas of improvement in this process.

The primary objectives of this research are as follows: a.) To evaluate the current practices and processes of communicating newborn screening results to parents. b.) To assess the effectiveness and timeliness of the communication methods used for conveying NBS results. c.) To identify potential barriers and challenges associated with the communication of NBS results.

I am inviting you to participate in my research study. Participants must be 18 years or older. Participation is voluntary. If you choose to participate in the study, it will take approximately 10-15 minutes of your time. There is no penalty for not participating in the survey. Responses are anonymous and will be kept confidential. De-identified data collected as a part of current study will be shared with only those directly involved with this project. No aspect of the survey asks for personal identifiers and the survey will not record any information about your email, phone number, or IP address. There are no anticipated risks of this study. The results of this study may be used in reports, presentations, or publications. Any shared results will be in aggregate form. There is currently no plans to use results in future studies.

If you have any questions about the study, please contact the me at tknelso5@asu.edu. Study PI is Katherine Hunt

Brendish, reachable at khuntbrendish@asu.edu. If you have any questions about your rights as a subject/participant in this research, or if you feel you have been placed at risk, you can contact the Chair of the Human Subjects Institutional Review Board through the ASU Office of Research Integrity and Assurance at (480) 965-6788. Thank you for taking the time to assist me with this research.

By clicking the button below, you acknowledge:

- Your participation in the study is voluntary.
- You are 18 years of age.
- You are aware that you may choose to terminate your participation at any time for any reason.

○ I consent, begin the study

Demographics

Are you a licensed physician in the state of Arizona?

- ○ Yes
- ○ No

In what county of Arizona do you practice in? (Select all that apply)

- ☐ Apache
- ☐ Cochise
- ☐ Coconino
- ☐ Gila
- ☐ Graham
- ☐ Greenlee
- ☐ La Paz
- ☐ Maricopa
- ☐ Mohave
- ☐ Navajo
- ☐ Pima
- ☐ Pinal
- ☐ Santa Cruz
- ☐ Yavapai
- ☐ Yuma

What is your medical training in?

- ☐ Family Medicine

- [ ] Internal Medicine
- [ ] Pediatrics
- [ ] Other (please specify) _____

How old are you

- ○ 18 - 24 years old
- ○ 25 - 34 years old
- ○ 35 - 44 years old
- ○ 45 - 54 years old
- ○ 55 - 64 years old
- ○ +65 years old

How do you describe yourself?

- ○ Male
- ○ Female
- ○ Non-binary / third gender
- ○ Prefer to self-describe _____
- ○ Prefer not to say

Choose one or more races that you consider yourself to be

- [ ] White or Caucasian
- [ ] Black or African American
- [ ] American Indian/Native American or Alaska Native
- [ ] Asian
- [ ] Native Hawaiian or Other Pacific Islander
- [ ] Other
- [ ] Prefer not to say

Are you of Spanish, Hispanic, or Latino origin?

- ○ Yes
- ○ No

Newborn Screening Process

Are you involved in the process of ordering and interpreting newborn screening tests?

- ○ Yes
- ○ Maybe
- ○ No

How many years of experience do you have with communicating newborn screening results?

- ○ Less than 5 years
- ○ 5-10 years
- ○ 10-20 years
- ○ More than 20 years

How often do you discuss newborn screening with parents before conducting the tests?

- ○ Often
- ○ Sometimes
- ○ Rarely
- ○ Never

Normal Results

Do you inform your patients of normal newborn screening results?

- ○ Yes

- ○ No
- ○ Sometimes

If normal, do you inform the parents of the first newborn screening result before receiving the second newborn screening result?

- ○ Yes
- ○ No
- ○ Sometimes

Do you inform the parents of the second normal newborn screening results?

- ○ Yes
- ○ No
- ○ Sometimes

How do you typically communicate normal newborn screening results to parents? (Select all that apply)

- ☐ In-person appointments
- ☐ Phone Call
- ☐ Email

- [ ] Printed Material
- [ ] Other (please specify) _____

After receiving normal newborn screening results, how soon do you contact the parents?

- ○ Same day
- ○ Less than 3 days
- ○ Less than 1 week
- ○ More than 1 week

How much time do you usually allocate to discussing normal newborn screening results with parents?

- ○ Less than 30 minutes
- ○ 30 minutes - 1 hour
- ○ More than 1 hour

On a scale of 1 to 5, how confident do you feel in explaining the purpose and significance of each screening test to parents? (1 = Not confident, 5 = Very confident)

Select rating

On a scale of 1 to 5, how satisfied are you with the training and resources available to support you in this area? (1 = Not satisfied, 5 = Very satisfied)

Select rating

Abnormal Results

Do you inform your patients of abnormal newborn screening results?

- ○ Yes
- ○ No
- ○ Sometimes

If abnormal, do you inform the parents of the first newborn screening result before receiving the second newborn screening result?

- ○ Yes
- ○ No
- ○ Sometimes

Do you inform the parents of the second abnormal newborn screening results?

- ○ Yes
- ○ No
- ○ Sometimes

After receiving abnormal newborn screening results, how soon do you contact the parents?

- ○ Same day
- ○ Less than 3 days
- ○ Less than 1 week
- ○ More than 1 week

How do you inform parents about abnormal screening results? (Select all that apply)

- ☐ In-person appointments
- ☐ Phone calls

- [ ] Email
- [ ] Printed materials
- [ ] Other (please specify)

How much time do you usually allocate to discussing abnormal newborn screening results with parents?

- ○ Less than 30 minutes
- ○ 30 minutes - 1 hour
- ○ More than 1 hour

How often do you encounter abnormal newborn screening results in your practice per year?

- ○ Never
- ○ Less than 5
- ○ More than 5

What support material do you use to help you prepare in communicating abnormal results? (select all that apply)

- [ ] ACT Sheets
- [ ] UpToDate

- [ ] American Medical Association Resources
- [ ] General Internet Search
- [ ] Colleagues
- [ ] Condition-specific advocacy groups
- [ ] None
- [ ] Other (please specify)

What strategies do you use to ensure parents understand the implications of abnormal results? (select all that apply)

- [ ] Interpreter
- [ ] Written support material
- [ ] Nurse educator
- [ ] Referral to geneticist
- [ ] Referral to other specialist
- [ ] Referral to a genetic counselor
- [ ] Follow-up in-person appointment
- [ ] Follow-up phone call
- [ ] Other (please specify)

On a scale of 1 to 5, how satisfied are you with the training and resources available to support you in this area? (1 =

Not satisfied, 5 = Very satisfied)

Select rating

Do you involve a counselor or a support system when communicating abnormal results to parents?

- Yes
- Sometimes
- No

How often do you involve geneticists, specialists, or other experts in managing abnormal results?

- Always
- Most of the time
- About half the time
- Sometimes
- Never

How do you handle cases where a newborn screening result requires further testing or follow-up? (please select

all that apply)

- [ ] In-office order further genetic testing
- [ ] Refer to geneticist w/ or w/out genetic counselor
- [ ] Refer to genetic counselor
- [ ] Refer to other specialist
- [ ] Other (please specify) _____

What are the main challenges you encounter when delivering abnormal newborn screening results to parents?

- [ ] Langauge barrier
- [ ] Using patient friendly language
- [ ] Parent reactions
- [ ] Time constraints
- [ ] Not enough resources
- [ ] Loss to follow-up
- [ ] Parents don't follow medical recommendations
- [ ] Other (please specify) _____

What kind of training or resources do you believe would be most helpful in improving your communication skills for abnormal results? (select all that apply)

- [ ] In-person trainings
- [ ] Online modules
- [ ] Language friendly ACT sheets
- [ ] Other written material
- [ ] [ ] Other resources (please specify)

Do you provide patients with resources to support groups?

- ○ Yes
- ○ Sometimes
- ○ No

Cultural Sensitivty

Do you consider cultural factors when discussing newborn screening results with parents?

- ○ Yes
- ○ Sometimes
- ○ No

Have you received training on culturally sensitive communication in the context of newborn screening?

- ○ Yes
- ○ Maybe
- ○ No

Concluding Statements

Please share any additional thoughts or experiences related to communicating newborn screening results.

APPENDIX E

CORN FRAMEWORK

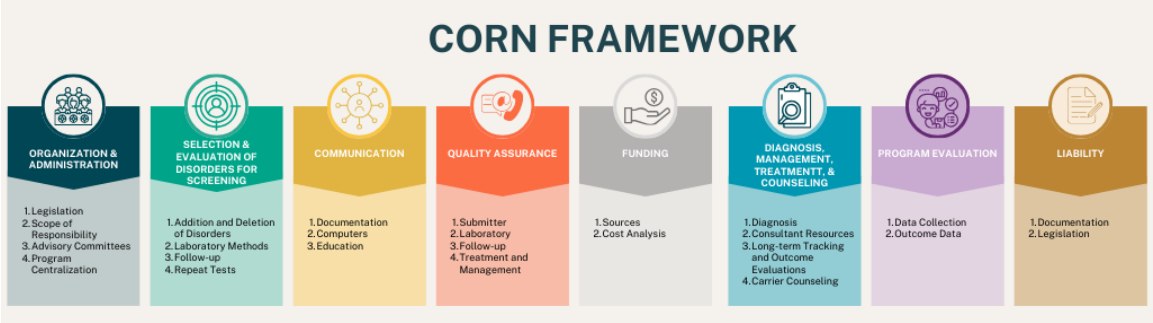
CORN FRAMEWORK
ORGANIZATION & ADMINISTRATION
1. Legislation
2. Scope of Responsibility
3. Advisory Committees
4. Program Centralization
SELECTION & EVALUATION OF DISORDERS FOR SCREENING
1. Addition and Deletion of Disorders
2. Laboratory Methods
3. Follow-up
4. Repeat Tests
COMMUNICATION
1. Documentation
2. Computers
3. Education
QUALITY ASSURANCE
1. Submitter
2. Laboratory
3. Follow-up
4. Treatment and Management
FUNDING
1. Sources
2. Cost Analysis
DIAGNOSIS, MANAGEMENT, TREATMENTT, & COUNSELING
1. Diagnosis
2. Consultant Resources
3. Long-term Tracking and Outcome Evaluations
4. Carrier Counseling
PROGRAM EVALUATION
1. Data Collection
2. Outcome Data
LIABILITY
1. Documentation
2. Legislation

TABLE 1: ARIZONA NEWBORN SCREENING PANEL

Table 1. Arizona Newborn Screening Panel: 35 Conditions Screened			
Category	**Core Conditions**	**First Screen Panel**	**Second Screen Panel**
Organic Acid Disorders (9):		✓	✓
	Isovaleric Acidemia (IVA)		
	Glutaric Acidemia type 1 (GA-1)		
	3-Hydroxy-3-Methylglutaric Aciduria (HMG)		
	Multiple Carboxylase Deficiency (MCD)		
	Methylmalonic Acidemia-Cobalamin Defect (Cbl A,B)		
	Methylmalonic Acidemia-Mutase Deficiency (MUT)		
	3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)		
	Propionic Acidemia (PROP)		
	Beta-Ketothiolase Deficiency (BKT)		
Fatty Acid Oxidation Disorders (5):		✓	✓
	Carnitine Uptake Defect (CUD)		
	Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)		
	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD		
	Long-Chain L-3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)		
	Trifunctional Protein Deficiency (TF)		
Amino Acid Disorders (6):		✓	✓
	Argininosuccinic Aciduria (ASA)		
	Citrullinemia Type 1 (CIT-1)		
	Classic Phenylketonuria (PKU)		
	Homocystinuria (HCY)		
	Maple Syrup Urine Disease (MSUD)		
	Tyrosinemia Type 1		
Endocrine Disorders (2):		✓	✓
	Primary Congenital Hypothyroidism (CH)		
	Congenital Adrenal Hyperplasia (CAH)		
Hemoglobin Disorders (3):		✓	✓
	Sickle Cell Anemia (Hb SS)		
	S, Beta-Thalassemia (Hb S/β Th)		
	S, C Disease (Hb S/C)		
Other			
	Biotinidase Deficiency (BIOT)	✓	✓
	Galactosemia (GALT)	✓	✓
	Cystic Fibrosis (CF)	✓	*
	Severe Combined Immunodeficiency (SCID)	✓	*
	Spinal Muscular Atrophy (SMA)	✓	*
	X-Linked Adrenoleukodystrophy (X-ALD)	✓	*
	Pompe Disease (by 1/1/2024)	✓	*
	Mucopolysaccharidosis Type I (MPSI) (by 1/1/2024)	✓	*
	Hearing Differences (HEAR)	"Point of care" tests completed by hospital or midwife	
	Critical Congenital Heart Defects (CCHD)	"Point of care" tests completed by hospital or midwife	

*These disorders are not normally performed on the second screen specimen after a successful first screen. However, the laboratory may add them to the second screen panel as needed.