




Newborn blood spot screening – knowledge of genetic testing among mothers

Angeline David ¹, Meredith O. Cruz ¹, Grzegorz Telega ¹, Pippa Simpson ¹,
Amy Pan ¹, Małgorzata Nagórska  ²

¹ Medical College of Wisconsin, Milwaukee, USA

² Medical College of Rzeszow University, Rzeszow, Poland

ABSTRACT

Introduction and aim. Newborn blood spot screening (NBS) uses genetic technology to screen for selected genetic, endocrine, and metabolic disorders. The purpose of the study was to assess the knowledge of newborn blood spot genetic screening among expectant mothers.

Material and methods. Between October 2015 and January 2016, a 20 question, multiple-choice questionnaire was administered to expectant mothers presenting for a pre-natal ultrasound at the Maternal Fetal Care Center in Milwaukee, Wisconsin Froedtert Hospital. Statistical analysis used Chi-Square or Fisher's exact test for categorical variables.

Results. 103 women completed the survey; 34% believed that education regarding screening is incomplete and 39% believed that it needs improvement. 27% knew the purpose of newborn screening.

Conclusion. Many mothers lack general and specific knowledge about NBS and the diseases screened for. Health education that provides accurate and complete information on the newborn blood spot screening should be provided to all parents prior to the administering of any genetic testing. Key areas that should be targeted include: purpose of NBS, screened diseases and how to interpret the results of the test.

Keywords. newborn, newborn blood spot screening, screening, perinatal care

Introduction

Newborn screening is the most widely used form of genetic technology. Newborn screenings uses a few drops of blood from the newborn's heel to screen for the presence of genetic, endocrine, and metabolic disorders. These screening tests identify infants at increased risk for selected genetic disorders, allowing for diagnoses and treatment to occur after follow-up tests. Currently in the United States, screening tests are mandated and are available for over 60 different disorders.^{1,2} The benefits of such testing include early intervention and appropriate management to prevent intellectual and physical disabilities. As NBS is mandatory, targets large popula-

tions, and offers preventative interventions. The widespread adoption of NBS, albeit beneficial, also poses a question of informed decision making.^{2,3}

The paternalistic nature of medicine often overshadows patient autonomy. When this happens, patients are denied the ability to make informed decisions for themselves. In this modern era of medicine, with advanced technologies developing each day, a discussion of informed consent is crucial. It is notable that a report by the Committee for the Study of Inborn Errors of Metabolism of the National Academy of Sciences recommended that "participation in a genetic screening program should not be made mandatory by law, but should be

Corresponding author: Małgorzata Nagórska, e-mail: mnagorska@ur.edu.pl

Received: 23.10.2022 / Accepted: 27.11.2022 / Published: 25.03.2023

David A, Cruz MO, Telega G, Simpson P, Pan A, Nagórska M. *Newborn blood spot screening – knowledge of genetic testing among mothers.* Eur J Clin Exp Med. 2023;21(1):52–57. doi: 10.15584/ejcem.2023.1.7.



left to the discretion of the person tested or, if a minor, of the parents or legal guardian”.⁴ Physicians must present information accurately and sensitively, including the diagnosis, the nature and purpose of the recommended interventions, and the burdens, risks and benefits of all options including no treatment. Informed consent for newborn screening should involve the discussion of guidelines established by the American College of Obstetricians and Gynecologists and the Royal College of Paediatric and Child Health⁵ that require information on the condition for which the test is being offered, purpose of the test, likelihood of positive and negative results, possibility of false positive and false negatives, and the risks and implications of test results.^{6,7}

Literature concerning NBS suggests that there is a discrepancy in the informed consent process between healthcare professionals and parents.⁸⁻¹⁰ Parents report that they feel that although screening is a routine test, they are not aware they even have a choice in the matter.¹¹

Despite efforts to decrease the knowledge gap between healthcare professionals and expectant and new mothers, a gap remains.¹² Healthcare professionals may not be providing consistent and reliable information to parents regarding newborn screening, which affects patient decision-making and informed consent. Informed consent creates an open dialogue between patient and provider. It allows patients to take an active role in their health care by providing autonomy and allows for trust in the health care relationship.

This study sought to investigate the current understanding of newborn screening among expectant mothers. We hypothesize that there is a lack of knowledge among mothers regarding newborn screening. The results of the study will help develop targeted health education for expectant mothers whose children are undergoing NBS so that they can be fully informed of the process.

Aim

The purpose of the study was to assess the knowledge of newborn blood spot genetic screening among expectant mothers.

Material and methods

Design

Cross-sectional descriptive study was used.

Ethical approval

The authors declare that the research was carried out in accordance with the Declaration of Helsinki of 1964 and its last revision of 2013. This study was approved by the Medical College of Wisconsin IRB as well as the Office of Clinical Research and Innovative Care Compliance (1/06/2015). All patients enrolled in the study were in-

formed of the aims and intentions of the study, that their participation was anonymous and voluntary.

Data collection

Data were collected between October 2015 and January 2016, 103 expectant mothers were approached for enrollment during their outpatient visits in the Maternal and Fetal Care Center in Milwaukee, Wisconsin Froedtert Hospital. Inclusion criteria included: pregnant women of legal age who speak and read English. Members of the study team as well as ultrasound technologists approached women who met inclusion criteria to participate in the study.

Expectant mothers were approached during a waiting period while in the ultrasound suites of the Maternal and Fetal Care Center. Participants received the questionnaire, which included an informational cover letter stating the purpose of the study, the voluntary nature of their participation, a description of the procedures, and a statement assuring their anonymity.

Twenty questions multiple choice survey collected quantitative information regarding the patients' experiences with newborn screening and the informed consent process in an urban hospital in Milwaukee, Wisconsin. It was developed by the last author. Participants were encouraged to answer all questions and were also allowed to leave answers blank if they did not know the answer. The survey took approximately 10 minutes to complete and was collected soon after completion. Participants were allowed to leave answers blank if they did not know the answer. Blank answers and multiple answers for certain questions about facts of NBS were counted as incorrect.

The questionnaire asked about basic facts about newborn screening, perceptions of informed consent, experiences of obtaining medical knowledge, and mothers' general opinions regarding newborn screening. Demographic information including age, parity, education level, and residence were also included on the questionnaire.

Statistical analysis

Analysis included Chi-Square or Fisher's exact test for categorical variables. Univariate analyses were used for continuous variables, and only complete data were used. All statistical analyses were performed using SAS version 9.4 (SAS Institute, Cary, North Carolina) software. And $p \leq 0.05$ was considered significant.

Results

A total of 103 expectant mothers completed the questionnaire. The response rate for this study was 100%. The average age was 29 years, with a range of 18 to 33 years. The majority of women (68%) had a college education or higher (Table 1).

Table 1. Survey sample characteristics (n=103)

Item	n	%
Age		
18-25	29	39%
26-30	17	23%
31-35	32	27%
>35	8	11%
Level of education		
Elementary School	0	0%
Less than High School	1	1%
Completed High School	28	28.3%
College or Higher	70	70.7%
Place of Residence		
Urban	87	87.9%
Rural	12	12.1%
Number of Children		
0	18	18%
1	33	33%
2	29	29%
3	13	13%
4+	7	7%

Table 2. Knowledge of mothers on newborn blood spot screening

Purpose of Newborn Screening	n	%
Diagnosis of disease	41	40.59
Confirmation of the possibility of disease in children in the future	27	26.73
Confirmation of carriers of certain diseases in children	29	28.71
Combination of answers	4	3.96
Total	101	100
Necessity of Screening		
Necessary	83	80.58
Not Necessary	4	3.88
No Opinion	16	15.53
Total	103	100
Knowledge of which diseases are screened		
Yes-Detailed	18	17.65
Yes-Incomplete	45	44.12
No	39	38.24
Total	102	100
Medical knowledge of diseases that are screened		
Yes-Detailed	18	17.82
Yes-Incomplete	41	40.59
No	42	41.58
Total	101	100
Source of Knowledge		
Internet	8	12.12
Medical Staff	28	42.42
Books, Magazines	5	7.58
Family, Friends	3	4.55
Other	9	13.64
Combination of answers	13	19.72
Total	66	100
When does NBS occur?		
24 hours after birth	76	75.25
Day 2	14	13.86
Between 4-6 days	9	8.91

Combination of answers	2	1.98
Total	101	100
Do you think there is a need for consent?		
Yes	66	64.71
No	17	16.67
I don't know	19	18.63
Total	102	100
When did you learn about NBS?		
Before Current Pregnancy	37	38.14
During Current Pregnancy	55	56.70
After birth	5	5.15
Total	97	100
Has your child already had screening?		
Yes	31	30.10
No	53	51.46
I don't know	19	18.45
Total	103	100
If YES, did you give consent?		
Yes	29	61.70
No	7	14.89
I don't remember	11	23.40
Total	47	100
Did you have enough time to make a decision?		
Yes	50	86.21
No	8	13.79
Total	58	100
Did you receive information on the collection procedure?		
Yes - Detailed	15	15.15
Yes - Incomplete	16	16.16
No information	68	68.69
Total	99	100
Did you receive information on when and how the results are interpreted?		
Yes	23	23.23
No	76	76.77
Total	99	100
Is education for newborn screening sufficient?		
Sufficient	24	25.53
Incomplete	32	34.04
Needs improvement	37	39.36
Combination of answers	1	1.06
Total	123	100
Preferred Method of Receiving Information		
Internet	6	6
Medical Staff	80	80
Books, Magazines	3	3
Family, Friends	1	1
Other	14	14
Total	100	100

As shown in Table 2, many women did not know the purpose of newborn screening, with only 26.73% of women correctly answering that it was a confirmation of the possibility of a disease. 38.24% of women also reported having no knowledge of what diseases were screened with only 17.65% of women having detailed knowledge. Many women received no information on

the process of newborn screening (68.69%) as well as when and how the results will be interpreted (76.77%).

Presented with the question on whether education for women on newborn screening is sufficient, only 25.53% say it is sufficient, with 34% reporting that education is incomplete and the other 39.36% reporting that the current education needs improvement. The majority of mothers (80%) also report their preferred method of receiving medical knowledge would be through medical staff with only 6% preferring the Internet for their source.

Analyses demonstrated that the effect of demographics or age does not influence correct answers. There is also no statistically significant difference in number of correct answers between rural and urban mothers.

Discussion

Our findings indicate that the majority of women believe that newborn screening is mandatory, yet education regarding newborn screening to be insufficient. Although the majority of women do have knowledge about the diseases that are screened for, their perception of their knowledge is often incomplete and lacking. The majority of women also incorrectly answered fact, based questions on newborn screening such as purpose of the test and methods of collection. The questionnaires also addressed sources of knowledge and preferred sources on knowledge in order to gauge potential points of intervention. The two most highly desired sources were medical staff and the Internet.

It is evident through these findings that there is still a knowledge gap regarding what patients know about newborn screening and how they understand the informed consent process. This study shows that education on newborn screening is lacking and as such, the informed consent process is incomplete. Patients are not receiving adequate medical knowledge for an important genetic test such as newborn screening. This study also presents us with an insight into the mothers' perceived understanding of newborn screening as well as a point of intervention in terms of current education.

Perhaps two of the most important discoveries in this study were the current method of receiving information among mothers, and their preferred method of receiving information. These data relate to informed choice stemming from education that should be given by the healthcare provider during the current pregnancy. The education can be in a written form but should also be communicated directly to the patient by the healthcare provider. Many of the women were divided between medical staff and the Internet, in terms of receiving knowledge about newborn screening. Ideally, the percentage of mothers receiving information from medical staff should be higher. Studies have shown that written information regarding screenings do not provide substantial informed choices.¹¹ Rather, infor-

mation given by health care professionals was more beneficial.⁹

It has been shown that parental knowledge regarding newborn screening as well as other genetic testing such as prenatal testing is often low and incomplete.¹⁵ The results of this study are consistent with earlier studies. General knowledge about testing is lacking, as well as knowledge about which diseases are screened for and knowledge about those specific diseases.¹⁶⁻²¹ Regarding the demographics of the women surveyed, there was no significant difference in knowledge across ages or place of residence. This shows that healthcare professionals should be wary of assuming that older women, or women who live in cities as opposed to rural areas, know more about newborn screening than their counterparts. The significantly lower understanding of newborn screening among women with lower education levels shows that healthcare providers should also take into consideration and address health literacy gaps. At the same time healthcare providers should not assume that women with higher education necessarily have more knowledge on newborn screening. The findings from the study also show a roughly equal distribution of when knowledge about newborn screening was obtained: before the current pregnancy, during the current pregnancy, and after delivery. Previous studies have shown that parents prefer information in the prenatal period.^{22,23} This fact, taken with the results of the study, could provide an opportunity to intervene and present relevant information regarding newborn blood spot screening early on in order to give mothers the most informed choice. Providing information during the prenatal period as opposed to postnatal allows time for mothers to develop and ask questions and gain a better understanding of a quick yet important diagnostic tool.

In light of these results, the limitations of this study should also be taken into consideration. Participants may not feel comfortable providing honest answers if they perceived their knowledge to be inadequate or their answers to be incorrect, which would affect the reliability of the survey data. Participants might also have an unclear understanding of the questions, which would lead to inaccurate answers.

With an understanding of the lack of knowledge that mothers have regarding newborn screening, there can be an educational intervention during prenatal care. In order to provide better knowledge to mothers about newborn screening and in a grander sense other tests or procedures, health care providers must be diligent in providing accurate and timely information to all patients, regardless of education level, age, or other demographic factors. In addition to advising healthcare providers about conveying newborn screening information to patients, perhaps more accurate and easily accessible material on the Internet would be beneficial to patients. Key areas that should be targeted include:

purpose of NBS, screened diseases and how to interpret the results of the test. These next steps will promote informed consent among mothers in regards to newborn blood spot screening.

Conclusion

Many mothers lack general and specific knowledge about NBS and the diseases screened for. Health education that provides accurate and complete information on the newborn blood spot screening should be provided to all parents prior to the administering of any genetic testing. Key areas that should be targeted include: purpose of NBS, screened diseases and how to interpret the results of the test.

Acknowledgements

Dr. Elaine Kohler Summer Academy of Global Health Research, the Medical College of Wisconsin Office of Global Health, Froedtert Memorial Lutheran Hospital, and those involved in distribution of questionnaires.

Declarations

Funding

The authors received no financial support for the research, authorship, and/or publication of this article.

Author contributions

Conceptualization, A.D., M.N. and G. T.; Methodology, A.D., M.N. and G. T.; Software, A.D., M.C., G. T.; P.S. A.P. and M.N. Validation, A.D., M.C. PS, AP, and M.N.; Formal Analysis, A.D., M.N. and G. T.; Investigation, A.D., M.N. and G. T.; Resources, A.D., M.N. and G. T.; Data Curation, A.D., M.C. PS, AP, and M.N.; Writing – Original Draft Preparation, A.D., G. T. and M.N. Writing – Review & Editing, A.D., M.N. and G. T.; Visualization, A.D., M.C., G. T.; P.S. A.P. and M.N. Supervision, A.D., M.N. and G. T.; Project Administration, A.D., M.C., G. T.; P.S. A.P. and M.N. Funding Acquisition, A.D., M.C., G. T. and M.N.

Conflicts of interest

The authors declare that they have not conflict of interests.

Data availability

Data available on request from the authors.

Ethics approval

The study was approved by the Medical College of Wisconsin/Froedtert Health IRB as well as the Office of Clinical Research and Innovative Care Compliance, USA (1/06/2015).

References

1. Newborn Screening Frequently Asked Questions (FAQ). 2016. Retrieved August 20, 2016, from Wisconsin State Laboratory of Hygiene, <http://www.slh.wisc.edu/clinical/newborn/parents-guide-to-newborn-screening/frequently-asked-questions-faqs/> Accessed August 19, 2022.
2. Detmar S, Hosli E, Dijkstra N, et al. Information and informed consent for neonatal screening: Opinions and preferences of parents. *Birth*. 2007;34(3):238-244. doi: 10.1111/j.1523-536x.2007.00176.x.
3. Nicholls SG. Proceduralisation, choice and parental reflections on decisions to accept newborn bloodspot screening. *Journal of Medical Ethics*. 2011;38(5):299-303. doi: 10.1136/medethics-2011-100040.
4. The President's Council on Bioethics. The changing moral focus of newborn screening: An ethical analysis by the president's council on bioethics. https://bioethicsarchive.georgetown.edu/pcbe/reports/newborn_screening/chapter4.html Accessed August 19, 2022.
5. Michie S, Dormandy E, Marteau TM. Informed choice: understanding knowledge in the context of screening uptake. *Patient Educ Couns*. 2003;50(3):247-253. doi: 10.1016/s0738-3991(03)00044-2.
6. Marteau TM, Dormandy E. Facilitating informed choice in prenatal testing: how well are we doing? *Am J Med Genet*. 2001;106(3):185-190. doi: 10.1002/ajmg.10006.
7. Baumiller RC, Cunningham G, Fisher N, et al. Code of ethical principles for genetics professionals: an explication. *Am J Med Genet*. 1996;65(3):179-183. doi: 10.1002/(SICI)1096-8628(19961028)65:3<179::AID-AJMG2>3.0.CO;2-L.
8. Statham H, Green J, Snowdon C. Mothers' consent to screening newborn babies for disease. *BMJ*. 1993;306(6881):858-859. doi: 10.1136/bmj.306.6881.858-c.
9. Davis TC, Humiston SG, Arnold CL, et al. Recommendations for effective newborn screening communication: results of focus groups with parents, providers, and experts. *Pediatrics*. 2006;117(5/2):326-340. doi: 10.1542/peds.2005-2633M.
10. Holtzman NA, Faden R, Chwalow AJ, et al. Effect of informed parental consent on mothers' knowledge of newborn screening. *Pediatrics*. 1983;72(6):807-812.
11. Hargreaves KM, Stewart RJ, Oliver SR. Informed choice and public health screening for children: the case of blood spot screening. *Health Expect*. 2005;8(2):161-171. doi: 10.1111/j.1369-7625.2005.00324.x.
12. Gourounti K, Sandall J. The validation and translation of multidimensional measure of informed choice in Greek. *Midwifery*. 2011;27:170-173. doi: 10.1016/j.midw.2009.04.006.
13. Gourounti K, Sandall J. Do pregnant women in Greece make informed choices about antenatal screening for down's syndrome? A questionnaire survey. *Midwifery*. 2008;24(2):153-162. doi: 10.1016/j.midw.2006.09.001.
14. Alio AP, Lewis, CA, Scarborough K, et al. A community perspective on the role of fathers during pregnancy: A qualitative study. *BMC Pregnancy and Childbirth*. 2013;13(1):60. doi: 10.1186/1471-2393-13-60.

15. Nicholls, S., Southern, K. Informed choice for newborn blood spot screening in the United Kingdom: A survey of parental perceptions. *Pediatrics*. 2012;130:1527-1533. doi: 10.1542/peds.2012-1479.
16. Kemper AR, Fant KE, Clark SJ. Informing Parents about Newborn Screening. *Public Health Nurs*. 2005;22:332-338. doi: 10.1111/j.0737-1209.2005.220408.x.
17. Tluczek A, Orland KM, Nick SW, Brown RL. Newborn screening: an appeal for improved parent education. *J Perinat Neonatal Nurs*. 2009;23(4):326-334. doi: 10.1097/JPN.0b013e3181a1bc1f.
18. Jansen ME, Van Den Bosch LJ, Hendriks MJ, et al. Parental perspectives on retention and secondary use of neonatal dried bloodspots: A Dutch mixed methods study. *BMC Pediatrics*. 2019;19:1-11. doi: 10.1186/s12887-019-1590-8.
19. Clayton EW. Talking with parents before newborn screening. *J Pediatr*. 2005;147(3):26-29. doi: 10.1016/j.jpeds.2005.08.001.
20. IJzebrink A, van Dijk T, Franková V, et al. Informing Parents about Newborn Screening: A European Comparison Study. *Int J Neonatal Screen*. 2021;202;7(1):13. doi: 10.3390/ijns7010013.
21. Fitzgerald C., Heery E, Conneally, et al. An evaluation of pregnant women's knowledge and attitudes about newborn bloodspot screening. *Midwifery*. 2017;45:21-27. doi: 10.1016/j.midw.2016.11.007.
22. Campbell E, Ross L. Incorporating newborn screening into prenatal care. *Am J Obstet Gynecol*. 2004;190(4):876-877.
23. Franková V, Dohnalová A, Pešková K, et al. Factors Influencing Parental Awareness about Newborn Screening. *Int J Neonatal Screen*. 2019;5(3):35. doi: 10.3390/ijns5030035.