Newborn Blood Spot Screening:

Assessing the level of knowledge of genetic testing among mothers in Rzeszow, Poland

**ABSTRACT**

Newborn blood spot screening has quickly become the most widespread use of genetic technology. It uses blood from the newborn’s heel to screen certain genetic, endocrine, and metabolic disorders. Currently in Poland, screening tests are available for over 40 different disorders, compared to the United States’ 60. Literature suggests that there is a discrepancy between parents and healthcare professionals regarding informed choice (Green, JM. 1993). This study assesses the knowledge gap regarding newborn screenings in an urban community in Poland with a questionnaire. During June 2015 to August 2015, pregnant and postpartum women were identified and approached for enrollment while admitted to Szpital Wojewódzki Nr 2 Hospital. An anonymous 20 question multiple-choice questionnaire was collected from 127 women. Despite efforts to improve the knowledge barrier, the data shows that there is still a gap that remains to be filled. Most women believed that screening is necessary but received incomplete information from the Internet and medical staff such that they answered incorrectly for many questions. These findings indicate that newborn screening in Poland is a fait accompli. Future efforts can attempt to address the knowledge gap by teaching health professionals how to better educate patients as well as creating accurate and accessible material on the Internet.

Newborn blood spot screening has quickly become the most widespread use of genetic technology. Newborn screenings are defined by using a few drops of blood from the newborn’s heel to screen certain genetic, endocrine, and metabolic disorders. One of the earliest screening tests was for phenylketonuria (PKU), which was developed by Dr. Robert Guthrie in the late 1950s. As a result of its effectiveness and efficiency, PKU screening was required in every state. By the late 1960s, newborn screening became a permanent part of health care for infants. Currently in the United States, screening tests are available for over 60 different disorders but differ by state as the state’s public health department determines each screening panel, (Baby's First Test 2015). In Poland, however, screening tests are only available for around 40 disorders.

Newborn blood spot screening is not a diagnostic test. It identifies individuals with increased risk and allows diagnosis and treatment to occur after follow up tests. Literature suggests that there is currently a discrepancy between parents and healthcare professionals regarding informed choice in newborn screening. (Green, JM. 1993).  Informed choice requires health care professionals to convey information, such that the patient has sufficient information to make a choice based on advantages and disadvantages of all possible outcomes in accordance with one’s own beliefs and values.

           Informed consent for newborn screening should involve the discussion of guidelines that have been established by numerous organizations, including the American College of Obstetricians and Gynecologists and the Royal College of Paediatric and Child Health (Michie, S. 2003). These guidelines include 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, as well as the risks and implications of test results (Marteau et al., 2001, Baumiller et al., 1996).

Despite efforts to improve the knowledge barrier between healthcare professionals and pregnant women, there is still a gap that remains to be filled (Gourounti K., 2011). Informed choice is a vital part of health care. If patients are not making informed choices, it is the responsibility of the healthcare profession to advocate for services that allow primary caregivers access to accurate information in order to create informed decisions (Gourounti, K., 2011).

In order to gauge the level of knowledge that mothers possess regarding newborn screenings, a questionnaire can be used. This questionnaire provides a categorization of informed choice to be used with both pregnant and postpartum women who have or will have newborn screening tests done for their infant.

The aims of this study are to assess the knowledge gap regarding newborn blood spot screenings in an urban community in Poland with a questionnaire. In addition to general knowledge about newborn screening (NBS), we have included questions on general experience and opinions with NBS and the current state of knowledge among mothers regarding NBS. This study will help to identify what measures can be put into place to meet the needs of patients regarding knowledge about newborn screenings.

METHODS

During June 2015 to August 2015, 127 pregnant and postpartum women were identified and approached for enrollment while admitted to the Obstetrics and Gynecology department of Szpital Wojewódzki Nr 2 Hospital in Rzeszow, Poland. Mothers were eligible for this study if they spoke and were literate in Polish. A midwife from the study team approached inpatients and outpatients who qualified to take the questionnaire. Patients received the questionnaire which includes an informational cover letter stating that the questionnaire is for research, a description of the procedures, participation is voluntary with no penalties or loss of benefits for not participating, and that the subjects’ privacy will be protected as personal identifiers were not collected. The Bioethics Review Board of the University of Rzeszow approved this study. Using the experiences of mothers regarding newborn screening knowledge and important facts about newborn screening, a 20 question multiple-choice survey was developed to assess the knowledge gap between healthcare providers and expectant and new mothers regarding newborn blood spot screening. The questions tested facts about newborn screening, knowledge about the diseases screened for, opinions and perceived availability on informed consent regarding testing, as well as sources of knowledge regarding newborn screening.  
 Data was analyzed using simple descriptive statistics to see frequencies and percentages of various answers among women. Variables such as age, parity, education level, and residence will be analyzed in order to answer the aims of the study: assess the knowledge gap regarding newborn blood spot screenings among pregnant women and mothers of newborns in Rzeszow, Poland.

RESULTS

A total of 127 mothers completed and returned the questionnaire. These mothers differed in terms of age, place of residence, number of children, educational level, and parity status. The majority of the sample was aged between 23 to 33 years old (71%) with the average age of mothers being 29 years old. The majority of women were educated with 72 out of 127 (57%) having an education of college or higher, (Table 1).

As shown in Table 2, when given the survey, many women incorrectly believed that newborn screening was a diagnostic test (61%) with only 20% of women correctly answering that it was a confirmation of the possibility of a disease. Over 50% of women also had incomplete knowledge of the diseases that are screened for with 40% of information being obtained from the Internet. In concordance with incorrect knowledge, many women received no information on the process of newborn screening (54%) as well has when and how the results will be interpreted (59%).

Only 58% of women ultimately gave consent for newborn screening. However, 36% of women report not having enough time to make a decision. Presented with the question on whether education for women on newborn screening is sufficient, only 14% say it is sufficient, with 46% reporting that education is incomplete and the other 40% reporting that the current education needs improvement. The majority of mothers (54%) also report their preferred method of receiving medical knowledge would be through medical staff with 33% preferring the internet for their source.

Statistical analyses show that the effect of age on knowledge is not significant. However, the amount of correct fact-based answers for newborn screening is significantly higher (p=0.02) for women with college or higher education. There is no difference in number of correct answers between rural and urban mothers.

DISCUSSION

This study is one of the first created to identify the knowledge gap between patients and healthcare professionals, while exploring informed consent regarding newborn blood spot screening in Poland. The findings indicate that the majority of women believe that screening is necessary yet education regarding newborn screening is insufficient. Although the majority of women do have knowledge about the diseases that are screened for, 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.

In light of these results, the limitations of this study should also be taken into consideration. As these results came from pregnant women and mothers of newborns, perhaps the knowledge of the father can be an additional insight or contributor to maternal knowledge. While this information was learned from patients, the responses and opinions of the health care professionals responsible for communicating knowledge of newborn screening to patients would also be beneficial. Despite these limitations, it is evident through these findings that there is still a large knowledge gap between health care professionals and their patient population regarding informed choice in the context of newborn blood spot 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.

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, (Nicholls, SG., 2012). The same holds true in this study. General knowledge about testing is lacking, as well as knowledge about which diseases are screened for and knowledge about those specific diseases. In regards to the demographics of women, there was no significant difference in knowledge across ages or place of residence. This shows that healthcare professionals should be weary 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, (Campbell, ED., 2004). 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.

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 (Hargreaves, K., 2005). Rather, information given by health care professionals was more beneficial, (Davis TC., et. al., 2006).

In order to address the knowledge gap between patients and health care providers regarding 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 of benefit to patients. These next steps will promote informed choice and consent among mothers in regards to newborn blood spot screening.

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Figures

Table 1:

|  |  |  |
| --- | --- | --- |
| **Survey Sample Characteristics (n=127)** | | |
| **Item** | **n** | **%** |
| **Age Group (y)** |  |  |
| 15-25 | 22 | 17% |
| 26-30 | 59 | 46% |
| 31-35 | 30 | 24% |
| 36-40 | 16 | 13% |
|  |  |  |
| **Highest Education Level** |  |  |
| Primary School | 1 | 1% |
| Less than High School | 8 | 6% |
| Completed High School | 46 | 36% |
| College or Higher | 72 | 57% |
|  |  |  |
| **Place of Residence** |  |  |
| Village | 74 | 58% |
| City | 53 | 42% |
|  |  |  |
| **Number of Children** |  |  |
| 0 | 30 | 24% |
| 1 | 47 | 37% |
| 2 | 33 | 26% |
| 3 | 11 | 9% |
| 4+ | 6 | 5% |
|  |  |  |
| **Parity** |  |  |
| Pregnant | 48 | 38% |
| After Delivery | 79 | 62% |

Table 2:

|  |  |  |
| --- | --- | --- |
| **Q1: Purpose of Newborn Screening** | | |
| **Response Item** | **n** | **%** |
| Diagnosis of disease | 75 | 61% |
| **Confirmation of the possibility of disease in children in the future** | **25** | **20%** |
| Confirmation of carriers of certain diseases in children | 23 | 19% |
| **Total** | **123** | **100%** |
|  | | |
| **Q2: Necessity of Screening** | | |
| Response Item | n | % |
| **Necessary**  Not Necessary  No Opinion | **116**  0  10 | **92%**  0%  8% |
| Total | 126 | 100% |
|  | | |
| **Q3: Knowledge of which diseases are screened** | | |
| Response Item | n | % |
| **Yes-Detailed**  Yes-Incomplete  No | **9**  64  52 | **7%**  51%  42% |
| Total | 125 | 100% |
|  | | |
| **Q4: Knowledge of diseases that are screened** | | |
| Response Item | n | % |
| **Yes-Detailed**  Yes-Incomplete  No | **3**  66  57 | **2%**  52%  45% |
| Total | 126 | 100% |
|  |  |  |
| **Q5: If Yes - Source of Knowledge** | | |
| Response Item | n | % |
| Internet  **Medical Staff**  Book, Magazines  Family, Friends  Other | 38  **39**  17  9  2 | 40%  **41%**  18%  10%  2% |
| Total | 94 | 100% |
|  |  |  |
| **Q6: When does NBS occur?** | | |
| Response Item | n | % |
| **24 hours after birth**  Day 2  Between 4-6 days | **54**  55  8 | **46%**  47%  7% |
| Total | 117 | 100% |
|  | | |
| **Q7: Do you think there is a need for consent?** | | |
| Response Item | n | % |
| **Yes**  No  I don't know | **83**  9  33 | **66%**  7%  26% |
| Total | 125 | 100% |
|  | | |
| **Q8: When did you learn about NBS?** | | |
| Response Item | n | % |
| **Before Current Pregnancy**  During Current  After birth | **39**  40  40 | **33%**  34%  34% |
| Total | 119 | 100% |
|  | | |
| **Q9: Has your child already had screening?** | | |
| Response Item | n | % |
| Yes  No  I don't know | 40  58  24 | 33%  48%  20% |
| Total | 122 | 100% |
|  | | |
| **Q10: If YES, did you give consent?** | | |
| Response Item | n | % |
| **Yes**  No  I don't remember | **49**  19  16 | **58%**  23%  19% |
| Total | 84 | 100% |
|  | | |
| **Q11: Did you have enough time to make decision?** | | |
| Response Item | n | % |
| **Yes**  No | **54**  30 | **64%**  36% |
| Total | 84 | 100% |
|  | | |
| **Q12: Did you receive information on the collection procedure?** | | |
| Response Item | n | % |
| **Yes - Detailed**  Yes - Incomplete  No information | **15**  35  58 | **14%**  32%  54% |
| Total | 108 | 100% |
|  | |  |
| **Q13: Did you receive information on when and how the results are interpreted?** | | |
| Response Item | n | % |
| **Yes**  No | **47**  68 | **41%**  59% |
| Total | 115 | 100% |
|  |  |  |
| **Q14: Is education for newborn screening sufficient?** | | |
| Response Item | n | % |
| Sufficient  Incomplete  Needs improvement | 17  57  49 | 14%  46%  40% |
| Total | 123 | 100% |
|  |  |  |
| **Q15: Preferred Method of Receiving Information** | | |
| Response Item | n | % |
| Internet  **Medical Staff**  Book, Magazines  Family, Friends  Other | 49  **81**  19  8  1 | 33%  **54%**  13%  5%  1% |
| Total | 149 | 100% |

REFERENCES

Baby's First Test. (2015). About newborn screening: Screening facts. Retrieved from [http://www.babysfirsttest.org/newborn-screening/screening-facts](http://www.babysfirsttest.org/newborn-screening/screening-facts%22%20%5Ct%20%22_blank)

Bailey, D., & Gehtland, L. (2015).  Newborn screening: Evolving challenges in an era of rapid discovery  *Journal of the American Medical Association, 313*(15), 1511-1512.

Baumiller, R. C., Cunningham, G., Fisher, N., Fox, L., Henderson, M., Lebel, R., . . . Roper Willson, N. (1996). Code of ethical principles for genetics professionals: An explication. *American Journal of Medical Genetics, 65*(3), 179-183. doi:10.1002/(SICI)1096-8628(19961028)65:3<179::AID-AJMG2>3.0.CO;2-L [pii]

Campbell, E., & Ross, L. (2004).  Incorporating newborn screening into prenatal care  *American Journal of Obstetrics and Gynecology, 190*(4), 876-877.

Davis, T., Humiston, S., Arnold, C., Bocchini, J. J., Bass, P. 3., Kennen, E., . . . Lloyd-Puryear, M. (2006). Recommendations for effective newborn screening communication: results of focus groups with parents, providers, and experts. *Pediatrics, 117*(5), 326-340.

Gourounti, K., & Sandall, J. (2011).  The validation and translation of multidimensional measure of informed choice in greek  *Midwifery, 27*, 170-173.

Gourounti, K., & Sandall, J. (2008). Do pregnant women in greece make informed choices about antenatal screening for down's syndrome? A questionnaire survey. *Midwifery, 24*(2), 153-162. doi:S0266-6138(06)00102-1 [pii]

Hargreaves, K., Stewart, R., & Oliver, S. (2005).  Newborn screening information supports public health more than informed choice.  *Health Education Journal, 63*(2), 110-119.

Marteau, T. M., & Dormandy, E. (2001). Facilitating informed choice in prenatal testing: How well are we doing? *American Journal of Medical Genetics, 106*(3), 185-190. doi:10.1002/ajmg.10006 [doi]

Michie, S., Dormandy, E., & Marteau, T. (2003).  Informed choice: Understanding knowledge in the context of screening uptake  *Patient Education and Counseling, 50*, 247-253.

Nicholls, S., & Southern, K. (2012).  Informed choice for newborn blood spot screening in the united kingdom: A survey of parental perceptions  *Pediatrics, 130*, 1527-1533.

Padilla, C., Krotoski, D., & Therrell, B. (2010). Newborn screening progress in developing countries - overcoming internal barriers. *Seminars in Perinatology, 34*(2), 145-155.

Quinlivan, J., & Suriadi, C. (2006). Attitudes of new mothers towards genetics and newborn screening.. *Journal of Psychosomatic Obstetrics and Gynaecology, 27*(1), 67-72.

Statham, H., Green, J., & Snowdon, C. (1993). Mothers' consent to screening newborn babies for disease.. *Bmj, 306*(6881), 858-859.

Therrell, B., Padilla, C., Loeber, J., Kneisser, I., Saadallah, A., Borrajo G, & Adams j. (2015). Current

status of newborn screening worldwide; 2015. *Seminars in Perinatology, 39*, 171-187.