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What Parents Want to Know about the Storage and Use of Residual Newborn Bloodspots

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Abstract

Many state newborn screening programs retain residual newborn screening bloodspots for a variety of purposes including quality assurance, biomedical research, and forensic applications. This project was designed to determine the information that prospective parents want to know about this practice.

Eleven focus groups were conducted in four states. Pregnant women and their partners and parents of young children (N=128) were recruited from the general public. Focus group participants viewed two educational movies on newborn screening and DBS retention and use. Transcripts

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Contributor's Statement

Jeffrey R. Botkin, M.D., M.P.H.: Dr. Botkin conceptualized and designed the study, participated in development of the focus group guided, production of the movies for the focus groups, analysis of the data, drafted the initial manuscript and approved the final manuscript as submitted.

Erin Rothwell, Ph.D.: Dr. Rothwell participated in the conceptualization and design of the project, designed and finalized the focus group guide, participated in the development of the movies, analyzed the focus group data, review and revised the manuscript and approved the final manuscript as submitted.

Rebecca A. Anderson, R.N., Ph.D.: Ms. Anderson participated in the conceptualization and design of the project, coordinated and supervised the focus groups, participated in the data analysis, critically reviewed the manuscript and approved the final manuscript as submitted.

Aaron Goldenberg, Ph.D.: Dr. Goldenberg participated in the conceptualization and design of the project, development of the focus group guide, development of the movies, reviewed the data analysis, the manuscript and approved the final manuscript as submitted.

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were analyzed with qualitative methods and the results were synthesized to identify key information items.

We identified 14 categories of information from the focus groups that were synthesized into seven items prospective parents want to know about residual DBS. The items included details about storage, potential uses, risks and burdens, safeguards, anonymity, return of results, and parental choice.

For those state programs that retain residual dried bloodspots, inclusion of the seven things parents want to know about residual dried bloodspots in educational materials may improve parental understanding, trust, and acceptance of the retention and use of stored bloodspots.

Keywords

newborn screening; dried bloodspots; education; research; public health

INTRODUCTION

Newborn bloodspot screening is conducted by state public health departments to enable the early identification and treatment of asymptomatic infants with certain genetic, metabolic, and endocrine disorders. The education of both clinicians and parents about newborn screening (NBS) is an important component of the newborn screening system that may enhance the efficacy of programs [American Academy of Pediatrics, 2000]. NBS programs offer information to parents primarily through printed material provided in the newborn nursery and in some prenatal clinics.

Davis and colleagues identified “seven things parents want to know about newborn screening” through focus group research [Davis et al., 2006], concluding that most parents want to know a few, relatively simple facts regarding these programs. However, their research did not address information about the potential uses of NBS residual dried bloodspots (DBS). Many states’ programs store residual DBS after the completion of screening [Lewis et al., 2011]. Currently more than half of the states retain specimens for more the 6 months and at least 20 for greater than one year, longer than what is necessary for clinical use [Olney et al., 2006]. These can be used for quality improvement, biomedical research, and forensic applications. Controversy has emerged over this practice: two state health departments (TX, MN) were sued by parents who objected to the retention of these samples without parental knowledge and consent, leading to fundamental changes in those programs [Lewis et al., 2011].

Public opinion on the research use of DBS has demonstrated that the public, and particularly parents of young children, are supportive of NBS programs and the majority are willing to have DBS used for research [Bombard et al., 2012; Botkin et al., 2012; Rothwell et al., 2012]. There is also a clear preference for receiving information prenatally about NBS and specific information about the retention and use of DBS [Botkin et al., 2012; Rothwell et al., 2012]. The Division of Health and Human Services Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children made recommendations regarding the management of residual DBS including that public and primary health care providers be

better educated about newborn screening and the retention and use of residual DBS [Therrell et al., 2011]. We conducted focus groups in four states to determine what parents want to know regarding the retention and use of residual DBS to help them make informed choices regarding this practice. This was synthesized to develop recommendations on the key information items. This research is part of a larger NIH funded project to develop and formally evaluate prenatal education tools that address newborn screening and the retention and use of residual DBS.

METHODS

Sample

Institutional review board approval was obtained and all participants provided informed consent for their participation. Eleven focus groups were conducted between February and May 2012 in the states of Utah, Washington, California, and Minnesota. Participants included women who were pregnant, partners of a pregnant woman, or parents of children who were 5 years of age or younger. Professional survey research companies recruited English-speaking participants from the local communities. Focus group participants were recruited from the research companies' panel members. The panels include individuals who have expressed willingness to participate in future research during previous surveys and polls, were referred by other panel members, or responded to an opportunity on a company's website. Panel members were contacted about participation in the groups, screened for eligibility and those who met inclusion criteria wre invited to participate. Eligibility criteria included 18 years or older, pregnant women and/or their partners (attended separate focus groups), parents with children less than 5 years of age, and they could not attended more than one focus group in their lifetime. A total of 128 individuals participated. The demographics of the participants are shown in Table I.

Procedures

Focus groups lasted between 1.5 and 2 hours. All focus groups were audio recorded and transcribed by a professional transcriptionist. A member of the research team verified all transcription work (ER). The focus groups followed recommendations by Krueger and Casey regarding preparation, engaging participants and moderating the discussion [Krueger and Casey, 2009]. The questions for the focus groups were created from a review of the literature and input from experts in newborn screening and were designed to evoke conversation, be open ended, and follow a logical questioning route [Stewart et al., 2007].

Prior to the group discussion, each group watched a short movie about NBS based on the "Seven Things Parents Want to Know about Newborn Screening" [Davis et al., 2006] and a second movie about the management of residual dried bloodspots. Both movies can be viewed at http://gslcutah.org/2013/05/newborn-bloodspot-videos/. The movies were created by the University of Utah Genetic Science Learning Center for this project. A qualitative researcher (ER) with expertise in focus groups and a pediatrician (JB) with expertise in NBS moderated the focus groups. An observer (RA) also was present.

Analysis

Qualitative content analysis was conducted to code data with similar content and relevance [Miles and Huberman, 1994]. The coding framework was created from reading the transcripts and the semi-structured interview guide, and was systematically applied to the transcripts with the ability to add additional codes (open coding) that may have been missed with the development of the codebook. A researcher with experience in qualitative research analysis (AG) who was not present at the focus groups conducted an independent analysis of the data to further establish trustworthiness of the analyses. Data from both analyses were compared and no discrepancies emerged.

All coded data was queried and reviewed for content using a qualitative software program (Atlas.ti). This allowed the researchers to assess the extent to which there may have been differences between states in regard to the density and type of codes. No differences between states were identified. The codes were then linked together based on similarity of the content to form the categories [Miles and Huberman, 1994]. The codes also provided a measure of the density of statements made and were used to help identify prominent categories among the groups. Less frequently occurring codes were collapsed into the more prominent similar codes within each of the main categories.

The emerging categories from these analyses were presented and discussed by the research team and by an advisory committee comprised of national experts in newborn screening (see Acknowledgments). Our final results regarding key information items for parents represent a synthesis of the focus group data by the investigators and the external advisory group to promote simplicity of the content for educational purposes [Baddeley, 1994].

RESULTS

The focus group participants identified 22 topics of potential information for parents regarding the retention and use of DBS. Of the 22 topics, 14 had numerous comments by participants in multiple groups. As previously described the 14 topics were synthesized in to the seven things parents wanted to know The results presented below provide a general description of the categories that emerged within the focus group discussions.

General Knowledge about NBS and DBS

Consistent with other research, we found that parents were generally aware that NBS had been conducted on their children but were not familiar with the details of the programs [Botkin et al., 2012]. Most participants were strongly supportive of newborn screening and reported that they were not familiar with the practice of retention and use of residual DBS. Frequently, participants said they thought the leftover bloodspots were immediately discarded and DBS were not stored (“I just kind of assumed they tested it, and threw it away”). A number of participants indicated the fact that DBS are stored and other information about storage such as length and location of storage should be communicated to parents. Representative quotes indicated what participants would like to know included: “I think how long the blood would be stored, especially since it’s going to vary from state to state.”

Parental Preference for Choice about DBS

Participants consistently identified issues regarding parental choice for DBS storage and use. Many of the participants stated that they would allow the DBS to be stored and used for research but wanted to be adequately informed and to have a choice. One representative quote that captured this perspective by the participants included:

> I have the expectation that I should be asked. Now, I think this gives a great case for me to say, "Yes," but I still think that individuals should be given that choice of whether the residuals are provided for research because, fundamentally, I think of it this way: You could make a case that all should be tested because it's important for the greater good of society. And that information should go to the parents because they have a vested interest. But the residuals are used for general research, which doesn't directly benefit the individual, so that they ought to give permission.

There was a clear expectation that adequate information about choice be delivered effectively to parents, although no clear preference on the type of permission process emerged (i.e., opt-in vs opt-out). One participant stated, "I think nine out of 10 parents don't know whether they verbally say it or on paper saying it. We just need a better understanding and information about what's going on." Another participant noted, "All the more information you can give about this, the better cooperation you're going to get from people." Participants consistently supported the notion that parents should be informed of whether there is a choice about DBS retention and use and how choice might be offered.

Benefits and Risks

Another consistent theme that emerged was the desire for information about the risks and benefits of research that might be conducted with DBS. Specifically, participants wanted to know the types of research, typical examples of projects, benefits and risks from research, and why DBS are useful in research. Representative quotes included: "Benefits for the population as a whole, plus benefits to you as an individual they did cite that there could be cures that are developed and diseases we identify in the future that we don't know now. " Some participants also indicated that information regarding how the bloodspots would not be used (e.g., cloning) would be helpful. Further, many participants said that parents should be informed of the risks and burdens of the retention and use of DBS for research ("*I'd like to know what the risks are.*"; and "The cons I think, because I think a lot of people are skeptical about stuff like that, 'Oh, they're keeping my baby's blood'). When asked what they thought were the risks, many of the comments focused on potential employment or insurance discrimination. Some participants commented on the pain of the heel prick, noting that it should be clearly communicated that no additional heel pricks are needed for the storage and use of residual DBS.

Safeguards

Participants were generally unaware of the types of safeguards used in the conduct of quality assurance activities and research, including the existence of institutional review board review, data security practices, and the de-identification of specimens. Accordingly, participants stated that it would be important to inform parents that safeguards that are in place to secure the physical samples and to protect the privacy of information generated in

research or QA activities (*"What are the safeguards?"*; "What it takes in order to be able to access those samples."; "My biggest concern is that discrimination in the future with healthcare, providing us care or jobs."; and "And it seems like the whole issue is kind of over privacy and people are struggling for privacy today"). There also were several questions concerning how access to the residual DBS is regulated and who governs access ("Who decides who is in charge, who really owns that and who is responsible for it?").

Information how DBS will be used in Research

With respect to the de-identification of residual DBS used in research, many of the participants expressed a preference for knowing whether samples would be used with or without identifiers. There were many comments by participants about the pros and cons of research with anonymous samples versus identifiable samples (*"Does anonymization limit the type of research?"*; and "What are the benefits or the pros and cons of anonymizing or not anonymizing ?"). There were a few comments by participants about the parental choice for having the DBS used anonymously in research ("I think the anonymization should be parent's choice from birth.").

More importantly, many of the participants asked questions about the level of parental control for how the DBS would be used in research. For example, some participants stated preference for the type of studies the DBS could be used, notification of when the DBS would be used, and the ability to not allow research to be conducted on the DBS. In general, many participants stated it was important that there was some level of control by the parents on how DBS would be used in research. Representative quotes include:

> Is there anyway that you can have as a parent a little bit of control over that? I would rather be notified before what test is being done to give me the choice of do I want my residual blood to be tested for that? I want to have that option.

Return of Research Results

We found that many participants expected that research results would be made available to parents if useful information was identified about their child ("I'd also like to know any results of any testing that you might have used my sample for, whether our blood sample was part of a positive or even part of the negative, it was still part of an overall research into whatever disorders or studies that they were look at."). Participants also discussed that it would be important to know general information about the research on DBS and how research with DBS improved the health of a group or community. When the participants were asked whether they would prefer greater privacy through de-identification or the return of results, the overwhelming response was return of results because many participants stated they would want to know if something was found to be wrong with their child ("I would want to know if they found out something about my kid like ten years down the road").

The Seven Things Parents Want to Know about Dried Bloodspots

From the items consistently identified by participants in all of the focus groups and the analysis of experts, we developed the key items parents want to know about DBS. These are included in Table III. These basic points provide a description of the retention practice and

note that the bloodspots can be used for promoting public health. The risks and burdens are addressed by stating that extra heelpricks are not performed and by noting that bloodspots are usually used in a de-identified fashion. The common misperception that research results would be returned to parents is included. Noting the existence of safeguards to protect privacy of babies and families is another important element. The last point emphasizes that parents often have a choice regarding the retention of their child's residual bloodspot although this point should be revised to accurately reflect the policy and practices of a state program. In general, we anticipate that these core items would be customized for individual state practices and supplemented with additional information or discussion by a care provider.

DISCUSSION

While work by Davis et al [2006] regarding what parents want to know about NBS has been influential in guiding the development of informational brochures for many state programs, there is broad recognition that current educational approaches for parents about NBS are inadequate [Araia and Potter, 2011; Arnold et al., 2006]. The issues about the retention and use of DBS are related to NBS but public sensitivities are heightened because the uses of DBS are not intended to benefit individual newborns and many members of the public perceive a threat to parental rights as well as privacy risks due to this practice. The lawsuits in Minnesota and Texas have motivated extensive efforts across the country to better balance the advantages of DBS retention with respect to quality assurance and biomedical research with the rights and expectations of parents. We believe that informing prospective parents of these key issues could help improve patient understanding of the desirability of DBS retention and mitigate the adverse consequences associated with lack of information.

Previous research demonstrates that if DBS are retained after clinical testing is complete, parents want information regarding this practice [Bombard et al., 2012; Botkin et al., 2012; Tarini et al., 2010]. Our project is the first to ascertain what parents specifically want to know about DBS retention and use. However, research of this type is hampered by the fact that the general public has little or no prior knowledge of this topic, including what newborn screening programs entail. This means that individuals must be informed about the topic first before they can be asked what they think parents should be told about this practice.

A limitation of our project is that responses from the focus group participants were no doubt influenced by what we told them about the topic and the movie presentations. However, our protocol was designed to provide a broad background on the issues and extensive group discussion in the focus groups. Participants were then asked to prioritize and identify key information items that could be relayed efficiently in the prenatal or postnatal environment. Priorities emerged from the groups that we had not highlighted or predicted in our presentation of background information. For example, the researchers did not anticipate that some of the participants assumed additional heel pricks were conducted for the storage of DBS. Further, it was the assessment of the investigators and expert advisory group that several expectations expressed by our participants could not be accommodated by the system. For example, the expectation that research results would be returned to parents is not feasible given the strong preference for the use of de-identified specimens. This

expectation by parents is contrary to the majority of research using DBS that is conducted with de-identified or anonymized specimens in order to protect the privacy of infants and families and because many research results have uncertain clinical utility. Further, it is not feasible for care providers in the newborn nursery to describe the types of research being conducted with DBS, although health department websites could maintain a list of approved uses. Therefore, our recommendations reflect the constraints that exist in these public health programs and research protocols as they currently function.

In summary, we found that parents are open to the retention and use of DBS but expect to be told about storage of DBS and to have a choice regarding whether or not their infant's DBS will be retained for future research. The key informational requirements identified by the participants are relatively simple and could be conveyed through a brochure or audiovisual presentation. Routinely providing this information in those states that choose to retain residual dried bloodspots may help maintain public trust in newborn screening and public health agencies.

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Abbreviations

DBS	Dried Bloodspots
NBS	Newborn Screening
IRB	Institutional Review Board

REFERENCES

American Academy of Pediatrics. Serving the family from birth to the medical home. Newborn screening: A blueprint for the future - A call for a national agenda on state newborn screening programs. Pediatrics. 2000; 106:389–422. [PubMed: 10947682]

Araia M, Potter B. Newborn screening education on the internet: a content analysis of North American newborn screening program websites. J Community Genet. 2011; 2:127–134. [PubMed: 22109819]

Arnold CL, Davis TC, Frempong JO, Humiston SG, Bocchini A, Kennen EM, Lloyd-Puryear M. Assessment of newborn screening parent education materials. Pediatrics. 2006; 117:S320–325. [PubMed: 16735259]

Baddeley A. The magical number seven: still magic after all these years? Psychol Rev. 1994; 101:353–356. [PubMed: 8022967]

Bombard Y, Miller FA, Hayeems RZ, Carroll JC, Avard D, Wilson BJ, Little J, Bytautas JP, Allanson J, Axler R, Giguere Y, Chakraborty P. Citizens' values regarding research with stored samples from newborn screening in Canada. Pediatrics. 2012; 129:239–247. [PubMed: 22250019]

Botkin JR, Rothwell E, Anderson R, Stark L, Goldenberg A, Lewis M, Burbank M, Wong B. Public attitudes regarding the use of residual newborn screening specimens for research. Pediatrics. 2012; 129:231–238. [PubMed: 22250018]

Davis TC, Humiston SG, Arnold CL, Bocchini JA Jr. Bass PF 3rd, Kennen EM, Bocchini A, Kyler P, Lloyd-Puryear M. Recommendations for effective newborn screening communication: results of focus groups with parents, providers, and experts. Pediatrics. 2006; 117:S326–340. [PubMed: 16735260]

Krueger, R.; Casey, MA. Focus Groups: A pratical guide for applied research. Sage; Thousand Oaks CA: 2009.

Lewis MH, Goldenberg A, Anderson R, Rothwell E, Botkin J. State laws regarding the retention and use of residual newborn screening blood samples. Pediatrics. 2011; 127:703–712. [PubMed: 21444595]

Miles, M.; Huberman, MM. Qualitative Data Analysis: An Expanded Sourcebook. Sage Publications, Inc.; Thousand Oaks CA: 1994.

Olney RS, Moore CA, Ojodu JA, Lindegren ML, Hannon WH. Storage and use of residual dried blood spots from state newborn screening programs. J Pediatr. 2006; 148:618–622. [PubMed: 16737872]

Rothwell E, Anderson R, Goldenberg A, Lewis MH, Stark L, Burbank M, Wong B, Botkin JR. Assessing public attitudes on the retention and use of residual newborn screening blood samples: A focus group study. Soc Sci Med. 2012; 74:1305–1309. [PubMed: 22398141]

Stewart, DW.; Shamdasani, PN.; Rook, DW. Focus groups : theory and practice. Vol. xii. SAGE Publications; Thousand Oaks: 2007. p. 188

Tarini BA, Goldenberg A, Singer D, Clark SJ, Butchart A, Davis MM. Not without my permission: parents' willingness to permit use of newborn screening samples for research. Public Health Genomics. 2010; 13:125–130. [PubMed: 19602864]

Therrell BL Jr. Hannon WH, Bailey DB Jr. Goldman EB, Monaco J, Norgaard-Pedersen B, Terry SF, Johnson A, Howell RR. Committee report: Considerations and recommendations for national guidance regarding the retention and use of residual dried blood spot specimens after newborn screening. Genet Med. 2011; 13:621–624. [PubMed: 21602691]

Table I

Demographics of Focus Group Participants

	N	%
Gender		
Female	70	54.7
Male	58	45.3
Ethnicity/Race		
White	41	32.0
Latino	43	33.6
African American/Black	36	28.1
Asian/Pacific Islander	7	0.8
Multi-Racial	1	5.5
Income (missing n = 12)		
< $25,000	23	19.8
$25,000-$45,000	43	37.1
$46,000-$65,000	25	21.6
>$65,000	25	21.5
Age		
18-29 years	46	35.9
30-39 years	71	55.5
40-49 years	11	8.6
Education		
< High School	4	3.1
High School	18	14.1
Some College/Bachelors Degree	72	55.3
Technical/Associate Degrees	22	12.2
Graduate Degree	12	9.4

Table II

Semi-Structured Interview Guide

1.	Before we begin, do you have any questions about newborn screening or residual samples?
2.	What would you want to know about the storage and/or use of leftover newborn screening samples?
3.	If you were to identify the top 5 things to be told to parents about residual samples, what would they be?
4.	What additional information should be included?
5.	The health department is concerned that people may opt out of newborn screening because of the storage and use of the leftover residual samples. What are your thoughts about this risk?
6.	How do you think the educational information about leftover samples should be provided to you?
7.	What aspects of this conversation or video might affect your choice to allow or not allow your child's sample to be stored and used?

Table III

The Seven Things Parents Should Know about Residual Dried Bloodspots

1.	Some states save leftover bloodspots after newborn screening is complete.
2.	Leftover bloodspots can be used to improve the public's health in many ways.
3.	No extra heel pricks are done to collect blood for other potential uses of the spots.
4.	Safeguards are in place to protect the privacy of babies and families and to ensure the ethical conduct of research.
5.	The baby's name or other identifiable information is not attached to the leftover bloodspots used in most research.
6.	Because most research with leftover bloodspots is done anonymously, parents will usually not get results back from the research.
7.	A parent may request that their baby's bloodspot not be used in research after newborn screening.