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Assessing Public Attitudes on the Retention and Use of Residual Newborn Screening Blood Samples: A Focus Group Study

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Abstract

This paper discusses attitudes and opinions of a diverse group of participants toward the retention and use of residual newborn blood samples for research. Data were drawn from focus groups based in six states in the USA, and results provide support for the retention and use of residual newborn blood samples for research when parental permission is asked beforehand. However, there were a number of concerns that also warrant attention for the development of policy and maintaining trust with the public, such as timing of permission, use of samples already stored, level of personal control of sample use and education. The results demonstrate the complexity of the topic and the ethical ambiguities associated with the retention and use of residual newborn blood samples.

Keywords

newborn screening; focus groups; public attitudes; biobank; USA

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Background

Biorepositories that include biological samples from large segments of the population raise ethical challenges that are not addressed in current regulatory standards and public engagement is one important aspect to help address these ambiguities (O'Doherty & Hawkins, 2010). One set of programs that has grown dramatically in recent decades, and in some states, is the development of biorepositories from leftover blood samples is newborn screening (NBS). NBS is a core service for public health and is collectively the largest application of genetic testing in the United States, but the lack of effective public engagement is leading to controversy over current policies and practices within a number of NBS programs.

NBS is conducted by state health departments within the first few days of life from a heelprick on the infant and blood is dried on filter paper. A number of tests are conducted on the dried blood spots (DBS) that identify infants at high risk for particular conditions and would benefit from early treatment. In most states, NBS is mandatory and conducted without informed consent of the parents, but some states provide parents the option to opt out of NBS for religious or philosophical purposes (Lewis et al., 2011).

After screening is complete, there are leftover DBS for almost all of the infants born and many states retain these specimens for a variety of purposes. Residual DBS are a particularly valuable resource for genetic and population based research. Already, the residual DBS have been used for a number of biomedical research such as infectious disease epidemiology, genetic and environmental epidemiology, and population based studies (Olney et al., 2006). Such research projects are typically conducted without parental knowledge or choice, which raises several ethical and legal dilemmas (Kharaboyan et al., 2004).

The retention and use of residual DBS expands NBS program goals that are outside of traditional screening purposes and will require the development of policies that have the potential for changing the way the state-based NBS programs are currently conducted. An American Academy of Pediatrics (AAP) Task Force recommended that the process of developing policies for newborn screening programs should address the concerns and include opinions of the public (AAP, 2000). The Institute of Medicine, the Association of Public Health Laboratories, and the Secretary Advisory Committee on Heritable Diseases in Newborns and Children have called for the development of state policies to address the retention and use of residual DBS (ACMG, 2009; APHL, 2005; HRSA, 2010). Non-expert opinion is critical to the development of public policy because it provides unique perspectives due to different life experiences and training.

Methods

This research was conducted as one component of a larger study aimed to assess public attitudes on the retention and use of residual DBS (5R01HG004970-02). The quantitative results which incorporated responses from 3,855 participants from a national sample are published separately (Botkin et al., 2011). Qualitative data in this report were obtained from 128 participants from Arizona, Colorado, New Mexico, Oregon, Utah, and Texas from February through July 2010. Data were collected from 14 groups consisting of general population (4 groups, n = 39), African-American (3 groups, n = 20), Hispanic (3 groups, n= 31), and mothers of young children (4 groups, n = 38). (See Table 1A for more information about the participants.)

Participants were all 18 years of age or older and could not have attended more than one previous focus group within their lifetime. Prior to beginning the focus group discussion, participants watched a 22-minute video about newborn screening and issues related to the

use of residual newborn dried blood samples (http://learn.genetics.utah.edu/content/health/ngs/samples.html). The informational video was validated for the level of information presented using a separate sample of respondents. Prior to beginning the research, approval from the University of Utah Institutional Review Board was obtained. (See Table 1B for a list of questions asked in the focus groups.)

The data were analyzed using a qualitative content analysis. Consistent with this approach used in our previous work (Rothwell et al., 2011), the codes are generated from the data from careful readings of the transcripts rather than using search algorithms. One member of the research team read and re-read the transcripts for the initial development of the codes. The codes were then reviewed and refined with another member of the research team who observed all of the focus groups. Then the codes were systematically applied to the transcripts with the ability to add additional codes that may have been missed with the development of the codebook. The codes were linked together until all of the data fell into distinct categories.

The codes also provide a measure of the frequency of statements made and were used to help identify prominent themes among the groups in this study as opposed to only the researchers' own interpretation of the data. Less frequently occurring codes were collapsed into the more prominent codes within each of the main categories. The second component and most time-intensive part of the analyses was returning to the transcripts and assessing the context of the codes for detailed description of why the statements were made. In addition, each of the subgroups' transcripts were analyzed separately (e.g. general population, mothers of young children, Hispanic, African American) and these data were compared across these subgroups. The results presented below are the themes that emerged within and across all of the 14 groups in this study.

Results

Due to the complexity of the topic, categories were used to help organize and present the data. The categories were created from codes that were grouped together based on meaningful relatedness. Within each of these categories several themes emerged across all of the groups in this study. The focus of this analysis and presentation of the results is on the breadth of the data as opposed to in-depth examination of specific categories. The data are presented in this manner because the purpose of the research was to inform policy development within NBS and an overview of all of the categories provides guidance on these issues. Table 1 contains representative quotes for each of the six categories.

Research with Residual Samples

Permission for Current and Future Samples—The main premise expressed by most of the participants was giving them a choice to participate or not. Many participants stated preference for an opt-in approach. Reasons for why participants preferred this opt-in approach included respect for parents and that parents should be responsible for making the decision about a child's participation in research. However, some participants were not sure how the logistics of an opt-in approach would work and therefore, an opt-out approach was discussed by some participants.

Timing of Permission—Most participants stated permission for retention and use of residual DBS should be obtained before newborn screening is conducted. The most commonly mentioned time period was during prenatal care. There were some participants who stated permission should also be asked before conducting NBS itself.

Mixed Opinions on Permission for Past Samples—There were mixed opinions on how samples that have already been collected and stored by states should be used for research. Some participants suggested that parents should be contacted and permission obtained. If the family could not be contacted, then the sample should not be used. Some participants stated that it was acceptable to use the residual samples without permission for research if they were anonymous. Overall, no consensus emerged from the data on this topic.

Conditions for Allowing Research

Type of Research—Many of the participants expressed support for research if it was for the benefit of improving health/medical and environmental research, and/or conducted by university or medical departments. Research with the samples by private pharmaceutical companies was not supported because it was expressed by some participants that these companies would use the samples for their own gain by targeting the most profitable diseases and not the greater good.

Identifiable Samples for Research—Most participants indicated that, if they gave permission, they would want to keep identifiable information with the sample in order to be notified if something was wrong with the child. However, a few participants stated that anonymous research, when communicated back to the community, could also be beneficial. Participants questioned the value of research if the individual results could not be returned to the families.

Personal Control—Many of the participants stated it was their right to individually choose whether the child's sample would be anonymous or identifiable for research, to decide on the type of research, and to be informed when the sample was going to be used and results from the research. Overall, participants stated mechanisms should be in place to provide on-going communication. They also indicated that they should have the option to change their minds and have the sample removed.

Length of Storage and Ownership of the Sample for Research

Length of Storage—Numerous comments were expressed that there should be a time limit on how long the samples are stored. There were also questions by the participants if they did give permission for use of the sample for research, what happened when the child turn 18 years? Concerns also included if when the child became of legal age, would the child's values differ from the parents and how the "now consenting adults" would react when they find out that their parents allowed their samples to be used for research.

Ownership of the Samples—Most of the participants stated that the residual samples were the property of the parent and not the state. As with the length of storage, most of the participants stated that when the child became an adult, ownership transferred to that individual. Participants stated the reason why the parent/child owned the sample was because the sample was a part of them.

Perceived Risks and Benefits of Research

Risk of Research on NBS Participation—Many participants stated that asking for permission for research with residual DBS would not interfere with them participating in the NBS program. The biggest concern reported by participants that would interfere with participation in the NBS program was the poor communication about the program and the residual samples. It is also important to note, that there were a few participants in each of the

groups who indicated that they were unsure about whether they would participate with research on residual DBS, but stated they were still highly supportive of the NBS program.

More Risks Identified than Benefits—Participants more readily identify risks that affected them and most of these risks identified were focused on the individual. The most common risks reported were denial of insurance or employment if research found they were genetically predisposed to a disease. There were substantially fewer comments by the participants and these comments focused on the community level benefits rather than individual level. Examples of benefits included how research could improve public health, especially with respect to children, and notification of environment toxins within a community.

Personal sample versus Child's sample—Many participants expressed concern and fear for their children's future more so than they did for themselves. When asked if permission for use of their "personal" sample for research, most of the participants were more willing to allow research than on their child's sample. Reasons for this included protecting their child from discrimination in the future (e.g. employment, insurance).

The Importance of Education

Prenatal Education—Many of the participants stated more should be done to educate participants about NBS program and for obtaining permission for the residual samples to be used for research. Most of the participants stated that education about NBS program and residual samples should occur during the prenatal period. Without education, many participants felt parental initial reactions to seeking permission for research on the samples would be to refuse or conversely, parents may not understand what they are signing.

Community Outreach—The role of the community was seen as a medium for education on a larger level. Many participants stated there should be awareness about NBS program and residual samples on the community level, and information needs to be accessible by the public.

Ethical Considerations with Respect to Residual Samples

Invasion of Privacy—There were numerous comments by participants that their privacy rights were invaded with the use of samples without their permission. Some participants stated they felt betrayed or invaded with the retention and use of the residual DBS without their permission and/or knowledge. In general, many participants stated there was an ethical obligation to know whether their child's blood was retained and used for any type of research.

Greater Good—Many of the participants also mentioned that there was an altruistic obligation for allowing these samples to be used to improve public and children's health. There would have to be some level of trust for medical advancements to occur, but it also needs to be weighed against individual rights. Individuals also stated they would like to contribute to the greater good but would also like to know about safeguards for protection.

Highlight the Samples Contain DNA—Many of the participants made a point to highlight that the residual DBS contained their DNA, specifically their children's DNA. Several participants also mentioned that additional protections should be in place for research with DNA. DNA not only communicated information about the child but also information about them and other family members.

Discussion

One of the implications of this research is the strong preference expressed by the participants to provide a choice for parents to allow or not allow secondary research use of the residual samples. This finding is consistent with other research that most parents would be willing to permit use of their children's DBS for future research studies if their permission was obtained (Tarini et al., 2010) The importance of obtaining parental permission for use of the DBS for research was reinforced when many participants were unsure how to manage past samples that have already been collected through the NBS program.

Parental permission for retention and use of the samples is also associated with the participants' desire for more personal control over how the samples should be used for research. Consistent with other research on public perspectives on biobanking (Murphy et al., 2009), participants in this study wanted ongoing choices and control over access to their child's sample. The desire for more personal control could stem from the perceived ownership and emotional connection over one's DNA and/or lack of trust in the state to provide safeguards on health information on these samples (Neidich, Joseph, Ober, & Ross, 2008). The increased desire for more personal control could also be associated with the higher number of perceived personal risks as compared to perceived community benefits with research on the residual DBS. The participants in this study stated they should own the sample because it contains DNA, and appeared to view research and genetic research differently with genetic research posing more risk.

Participants' attitudes toward the relationship between residual DBS and the NBS program, and more control over access to the residual DBS mentioned in this study could have a direct impact on the NBS program. Most of the participants stated it was an invasion of privacy to retain and use the residual DBS without parental permission. Some participants also stated they were misled due to the lack of information and communication not only about the residual samples but also about the NBS program. Many of the participants were unaware that NBS was even conducted on their children and/or that NBS was more than just a PKU test. However, it is important to note that most of the participants were supportive of research on the samples when their permission was asked before obtaining the sample and participants expressed a desire to help the greater good.

The results of this study shed light on the complexity of the topic, the ethical ambiguities associated with residual DBS, and the need for increased transparency to maintain public support for the NBS program. The support of the public is essential for the successful conduct of a population screening program. Understanding public attitudes and opinions about residual DBS is a key step to ensure public accountability of newborn screening.

There are limitations to this study that should be noted. Although there were no differences between the categories within each of the subgroups in this study, such group differences could still be present. The focus groups in this study covered a wide breadth of topics and a more focused discussion on one area (i.e. risks and benefits) may reveal more concerns than the ones expressed in this study. Finally, there were a few participants who were not supportive of research even if parental permission was asked and, conversely, there were a few participants who were supportive of research without any parental permission. The results presented here do not represent these minority opinions but the ones most frequently discussed in the groups.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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Research Highlights

- Residual newborn blood samples could serve as a valuable source for biomedical research but public support is essential.
- Results indicate that the US public is supportive of research with residual newborn blood samples when permission is asked.
- A number of concerns were identified that should be addressed for the development of policies for the retention and use of residual newborn samples.

Table 1

Focus Group Results

Research with Residual Samples	
Permission for Current and Future Samples	• What's important is just giving people a choice. I don't like not having a choice and that's what bothers me. I want to have the choice. • I need to sign something to let me know that they're doing some sort of research. Because that my baby's blood. • It's respect. It should be an opt-in. I give my permission to do this. • I've worked for different state agencies and there's not a lot resources [for opt-in].
Timing of Permission	• I think when we go to the prenatal exams. • From the beginning of the pregnancy. It's the best way to start talking about that. • I think you should have consent to allow them to take the blood spot [NBS screening].
Mixed Opinions on Permission for Past Samples	• They should try to find me to use it [past samples]. • I would want to be contacted to give consent [for past samples already collected]. • I mean it's inconceivable that the fact that they could actually track down how many millions of babies are born each year. First of all, it's going to cost you way too much money.
Conditions for Allowing Research	
Type of Research	• Health-related research. • As long as it's for medical research. • Are drug companies going to start targeting what they found within me? • I trust the medical community.
Identifiable Samples for Research	• Keep names on them because if they find something in my kid's sample that can help have a healthier life, I'd want to know about it immediately. • It's got to be all or nothing. If you don't have that person name, that research is useless.
Personal Control	• I think they should ask them would you like us to keep with the baby's name or would you like us to keep it anonymous. • I think that in the consent there should be specific questions about what you want to use the blood to research for. • Inform them [parents] when they're going to use their blood, informing them of the progress if anything is found. • The opportunity to change your mind in the future or the child.
Length of Storage and Ownership of the Sample for Research	
Length of Storage	• Some type of time limit is needed. • As long as it's possible you could do it with the understanding that once the child does reach adult age they have the opportunity to change the decision that their parents made if they so choose.
Ownership of the Samples	• Because that's your child's blood. It's not [the] state's blood. It 's your child, part of your child. • At some point like at 18 years, there's some process within the system where you can reassert your permission for the sample. • Parent owns them and then later on the child, and let the child know, "We have a sample of your blood.
Perceived Risks and Benefits of Research	

Risk of Research on NBS Participation	• I'm somewhat embarrassed to say that I did not understand that they use it for more than one test [PKU]. • When they do the test on the child - I never knew it was for that [NBS]. I don't have the knowledge about it. • For the first initial tests that they do, yes [NBS], I think that is a necessity. But as far as keeping my blood samples, I don't want that to be done.
More Risks Identified than Benefits	• But even though GINA is in existence, the anonymity, I don't believe in that. It's not dependable. And I wouldn't want my child in 21 years or 18 years of age going for health insurance for there to be a back door to see what might be coming up, especially even if it's private or federal insurance.
Personal sample versus Child's sample	• I don't care about me, but my daughter's. I don't want my daughter's DNA being used for anything. • I think with strict policies and rules in place, research could be beneficial but it can also be very dangerous for our kids. • I think research is necessary and there's a lot of benefit, but I want my child's name off.
The Importance of Education	
Prenatal Education	• Informing the parents in the hospital before it's done, what these are going to be used for, what they could be used for in the future. • As women, while we're getting prenatal care. Because once we have the children, all of focus is taking our ourselves and the child. We wouldn't have time to be reading brochures and letters.
Community Outreach	• So that they [public] can be aware of it, don't just limit it to the people having babies. • Like they advertise immunization, the public needs to be informed. • Advertisements on the television.; and Put a billboard up about it.
Ethical Considerations with Respect to Residual Samples	
Invasion of Privacy	• There's an ethical thing with this. Whether people have the right to collect samples and use it for research without people's knowledge. • It's a little bit of a violation of your privacy. • Well, my attitude is who are you to invade my child and take his blood or her blood and use it for your purposes? You don't know better than me. I am the parent. That's my child, not yours.
Greater Good	• I think the very least we could do is take a little more of a gamble and give permission for the residual samples to be used in research that could possibly benefit someone just besides ourselves. • At some point you have to trust that these people are doing the right thing, and that there's not going to be an abuse.
Highlight the Samples Contain DNA	• It's your DNA. • They [children] need to know where their DNA is. • You guys invade my privacy, you're holding my blood, my DNA somewhat hostage. • When you're taking something like that, blood or samples of DNA, that there should be some very, very strict, I mean very strict policies.