



Public Consultation Regarding State Newborn Screening Policies

Michelle Huckaby Lewis, M.D., J.D.

Genetics and Public Policy Center, Berman Institute of Bioethics

Johns Hopkins University, Washington, DC

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Acknowledgements

- **Collaborators:**
 - **Aaron Goldenberg**
 - **Rebecca Anderson**
 - **Erin Rothwell**
 - **Jeff Botkin**
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Background

- **Operation of state nbs programs has become increasingly controversial**
- **Key element in lawsuits was perceived lack of transparency**
- **Since most infants born in the U.S. undergo mandatory nbs, parents of healthy children, parents of children affected by nbs d/o's, & public at large have an interest in nbs policies**
- **Policies related to expansion of nbs (use of public resources), retention/use of dbs**
- **NBS Advisory Committees are a possible mechanism by which public consultation could be achieved**

Need for Public Participation in Medical Policy-Making

- Individuals should have a voice in policy decisions that have implications for the public**
- Interests of state and individual may differ**
- “Exclusion of relevant communities from policy-making can alienate presumed beneficiaries of genetic services.”**

E Hiller, G Landenburger, M Natowicz, “Public Participation in Medical Policy-Making and the Status of Consumer Autonomy: The Example of Newborn Screening Programs in the United States,” American Journal of Public Health, Aug. 1997, Vol. 87, No. 8. pp. 1280-1288.

Role of State NBS Advisory Committees

- **“State advisory committees constitute an important mechanism for the involvement of the public in newborn-screening programs.”**
- **1997 data indicate “there is ample room for increased public participation in the construction of the policies of newborn-screening programs.”**

Hiller, 1997.

Recommendations re Advisory Committees

- **Newborn Screening Committee of the Council of Regional Networks for Genetics Services (CORN)**
- **Basic program guidelines:**
- **“The use of at least one advisory committee, including outside professional and consumer representatives, is encouraged. . . .”**
- **“This committee. . . . Should act as a group of consultants that helps in developing approaches, planning future directions, and problem solving.”**

BL Therrell, SR Panny, A Davidson, et al, “US Newborn Screening System Guidelines: Statement of the Council of Regional Networks for Genetics Services,” Screening, 1 (1992) 135-47.

AAP NBS Task Force Report

- “States. . . Should include public participation in medical policy-making.”
- “Each state should establish and fund a newborn screening advisory body with public participation to advise on newborn screening policy developments.”
- “Such an entity should include a broad range of public advisors representing parents, health professionals, third-party payers, appropriate government agencies, and other concerned citizens.”
- “Such an entity should be involved in the ongoing evaluation of all aspects of the state’s process for newborn screening.”

AAP Newborn Screening Task Force, “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.

Recent Calls for Public Engagement

- **“Public engagement in developing policies for biobanking initiatives takes time and resources. But the payoff-trust in the research enterprise and willingness to provide biospecimens-is worth the effort.”**

K Maschke, “Disputes over Research with Residual Newborn Screening Blood Specimens,” Bioethics Forum, 9/8/09, <http://thehastingscenter.org/Bioethicsforum/post.aspx?id=3826>, accessed 11/1/11.

Focus Groups

- **Focus groups were held with state Newborn Screening Advisory Committee members**
- **Purpose: To “ascertain attitudes and opinion regarding the storage and secondary research use of DBSs.”**
- **NBS advisory committees from Mountain States Region**

EW Rothwell, RA Anderson, MJ Burbank, AJ Goldenberg, MH Lewis, LA Stark, B Wong, JR Botkin, “Concerns of Newborn Blood Screening Advisory Committee Members Regarding Storage and Use of Residual Newborn Screening Blood Spots,” American Journal of Public Health, November 2011, Vol. 101, No. 11, pp. 2111-2116.

Focus Group Results

- **“Participants were asked about the role the Newborn Screening advisory committee should play in developing policies for storage and secondary research use of DBSs.”**
- **Noted “differing views regarding whom the advisory committee represented. Some participants felt they represented the community or at least understood the concerns of the community, and others disagreed.”**
- **The most commonly held view was that the committees represented medical experts in the community.**
- **“(I would say we represent the medical community, we haven’t always had great luck with getting consumers involved in our advisory committee.)”**

Focus Group Results

- **“Participants supported involvement by parents with and without affected children as a necessary component of NBS advisory committees.”**
- **“([What we need are] parents on the advisory committee. If you’re going to have an advisory committee that releases samples for research, you should have parents.)”**
- **“Participants were unsure, however, about public representation-how the public should be defined and whether an advisory committee would be representative of the public.”**

State Law Analysis: Methods

- **State statutes and regulations related to nbs of all 50 states +DC were accessed online between 11/08 and 12/09.**
- **Reviewed by 2 independent reviewers to determine extent to which the retention and use of DBS were addressed.**
- **What states required to do (may be doing more)**

MH Lewis, A Goldenberg, R Anderson, E Rothwell, J Botkin, "State Laws Regarding the Retention and Use of Residual Newborn Screening Blood Samples," Pediatrics, published online 3/28/11; DOI: 10.1542/peds.2010-1468.

Advisory Committees

- **22 states require the appointment of a Newborn Screening Advisory Committee or a Genetics Advisory Committee (AK, AZ, DE, DC, FL, GA, IL, IO, KS, ME, MD, MA, MS, MO, NE, NH, OH, OK, TN, VA & WY)**
- **The composition of the advisory committee is at least partially specified by either statute or regulation in 16 states. (AZ, DE, DC, FL, GA, IO, MD, MA, MN, MS, MO, NE, NH, OH, TN & WY)**

Committee Composition Reqts

- **Ten states require the committees to have lay members, defined as parents, “consumers,” or “representatives of a consumer group.” (DC, FL, GA, IO, MD, MA, MN, MO, NE & OH)**
 - GA requires prof’l & consumer representatives w/ knowledge & expertise in nbs
 - NE requires membership to include consumers w/ technical, prof’l &/or personal experience w/ nbs
 - In OH, members should include individuals w/ interest & expertise in nbs, incl members of the public
- **Four states require that the membership include at least one parent of a child affected with a d/o included in the state’s nbs panel or a parent with experience w/ an abnml screen (AZ, DE, MS & NH)**

Committee Membership Reqts

- **14 states specify the minimum # of consumer representatives**
 - **Some states specify # (AZ, DE, MS, MO, NH-at least one; DC-4; FL-2; MD-5)**
 - **Other states mandate that there should be consumer representatives but don't set a specific number (must be at least 1)**

Role of Members

- In genl, the role of the committee is to provide advice and guidance re the administration of state nbs programs.
- In 3 states, committee has role to play in deciding whether DBS may be used for research purposes.
 - In IO, b/f research proposals using dbs can commence, proposal must be approved by the advisory committee
 - In ME, dbs may be released for anonymous research w/o consent if approved by DOH w/ input from the advisory committee. Q 5 years, the program advisory committee shall review the policy to store residual specimens indefinitely.
 - In MN, 1 of the roles of the advisory committee is to assess the ethical considerations surrounding the testing, tx, and handling of data and specimens generated by the testing equipment.

Public Consultation Reqts

- **Public consultation is req'd w/ respect to the activities of only 3 nbs advisory committees.**
 - **In DC, one of the roles of the committee is to consult w/ the public, esp groups affected by metabolic d/o programs.**
 - **In NH, any proposal for recommending new tests or fees shall req a public hearing to be held on the proposal by the nbs advisory committee. The committee shall give public notice of the proposed agenda 30 days in advance.**
 - **In MD, before DOH adopts rules, regs, or stds re nbs, the DOH is req'd to consult the public, especially communities & groups particularly affected by hereditary d/o's programs.**

Conclusions

- **Who the “public” is is not clearly defined**
- **“Currently existing mechanisms for inclusion of the public in determining nbs program polices are underutilized.” Hiller, 1997.**
- **For effective public participation, NBS Advisory Committees should include:**
 - **Parents of health children**
 - **Parents of children affected by nbs d/o**
 - **Public at large b/c of duration of specimen retention**
- **Decision-making re operation of nbs programs & retention/use of dbs**
- **Goal is to enhance public participation and foster public trust**

**Bioethics and Legal Issues
Predominant in Discussions
Regarding Newborn Screening
Translational Research Network**

**M.H. Lewis, E. Goldman, J. Brosco, M. Watson, B.
Thompson, A. Brower, A. Hoffman**

**Johns Hopkins University, University of Michigan,
University of Miami, American College of Medical
Genetics**

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Background

- ◆ American College of Medical Genetics awarded contract by NICHD to develop a National Coordinating Center for NBS research (2008)
- ◆ Newborn Screening Translational Research Network (NBSTRN) created to develop a comprehensive infrastructure that allows investigators access to robust resources for NBS research
- ◆ NBSTRN comprised of Standing Committee and 4 work groups: Clinical Centers, Laboratories, Bioethics and Legal Issues

Research Tools and Resources

◆ Tools:

- Virtual Repository of Dried Blood Spots
- Laboratory Performance Database (R4S)
- Long-term Follow-up Data Collection

◆ Resources:

- Investigator FAQs to help researchers navigate bioethical and legal issues related to NBS research
- In process of developing model consent form

Methods

- ◆ June 16-17, 2011: NBSTRN convened Standing Committee and all 4 Work Groups to review the scope of their work and identify goals of the NBSTRN for the next 2 years
- ◆ Work Group members attended Work Group sessions that were different from their primary Work Group membership in order to learn more about the other Work Groups
- ◆ Then reconvened with primary Work Group upon completion of the sessions

Results

- ◆ All 4 Work Groups spent a significant amount of time discussing unresolved Bioethics and Legal issues.
- ◆ Several themes emerged:
 - Consent issues remain unresolved, opt-in vs. opt-out
 - DBS are scarce resource, social justice issues
 - Who may have access?
 - For what purposes?
 - Who decides?
 - Data protection/sharing
 - Need to have NBSTRN serve as an “honest broker” for long-term follow-up data

Parental Permission for Pilot NBS Research-SMA Project

- ◆ Principles and underlying assumptions used to develop recommendations
 - Need for QI/QA activities to improve research
 - Must not jeopardize the public health mission of nbs
 - Need for evidence base to support decision-making
 - Recognize that sometimes decisions are made based upon incomplete evidence-politics!

What is a Pilot Study?

- ◆ For these purposes, a “pilot study” is:
 - Something that looks like regular screening
 - Mimics nbs program, population-based evaluation
 - Can pilot nbs system relevant to new test/condition
 - Intent is to gather data for decisions re expansion population-wide
 - Can be at hospital level or statewide
 - May or may not utilize the public health system
 - Did not distinguish btw “pilot” and “research”

Education and Choice in Pilot Studies

- ◆ There was strong support for parental education and choice re pilot nbs protocols
- ◆ Parents should receive high-quality linguistically appropriate education materials
- ◆ Multiple avenues and levels of information should be included: brochures, websites
- ◆ Educational programs should occur in prenatal period when possible
- ◆ Prenatal and postnatal care providers should be aware of the research protocol and be able

Form of Choice

- ◆ When traditional informed consent is not req'd, an opt-out approach may be appropriate in some circumstances (not defined)
- ◆ Reqts for opt-out:
 - Ability to opt-out should be clearly communicated in educational materials
 - Mechanism for opting out should be readily available

◆ When traditional IC is waived, an opt-in approach may be important

Incidental Findings

- ◆ “Finding concerning an individual research participant that has potential health or reproductive importance and is discovered in the course of conducting research but is beyond the aims of the study.” (Wolf, “Managing Incidental Findings in Human Subjects Research: Analysis and Recommendations, J Law, Med & Ethics, 2008)
- ◆ Distinguish from results that are not the target of the activity but are predictable, i.e. SS trait
 - Should researchers have duty to consider these findings and whether to report them? Should they have a plan?
- ◆ Does it matter how information was discovered, i.e., in diagnostic vs. research vs. Next Gen sequencing?
- ◆ Is there a duty to look for information beyond target of investigation? (Dist fr clinical scenario-CXR)

Incidental Findings

- ◆ Presumption in favor of providing information b/c the information may be important
- ◆ But what if we don't know whether info is important?
- ◆ Are there limitations to providing all knowable information? Negative results?
- ◆ What is the obligation to do something just because we can?
- ◆ Conclusions:

Why is NBS Different?

- ◆ NBS is a population-based public health program. Not equivalent to other types of biomedical research or clinical settings.
- ◆ Can be performed w/o parental knowledge or permission.
- ◆ Duty to return results in other contexts hinges upon the relationship of the investigator with the subject.
- ◆ Participation is mandated by state law.

Community as Stakeholder

- ◆ For whose benefit are DBS stored?
- ◆ At whose expense are DBS stored?
 - May implement fee structures to recoup these costs
 - Initial burden borne by the state
- ◆ Who should benefit from research use of DBS?
- ◆ What obligations are imposed upon researchers?
- ◆ Researcher may not have independent obligation to community to share results.
- ◆ Obligations must be spelled out in terms of use agreements with individual states

Conclusions

- ◆ Many bioethical issues related to nbs, nbs research, and use of residual dbs remain unresolved
- ◆ Focus on bioethics by all of the Work Groups demonstrates the need to pay careful attention to these issues in the development of future policies related to nbs research

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