Committee Report on the Retention and Use of Residual Dried Blood Spot Specimens after Newborn Screening

Advisory Committee on Heritable Disorders in Newborns and Children

Presented by Alissa Johnson, January 21, 2010

Preparation for requesting comment

- Added statement at the beginning of paper regarding potential to advance science and clinical care
- Added language "***a policy in place that has been reviewed by the state attorney general or other appropriate legal authority***" to recommendations 1 and 2

Preparation for requesting comment

- Removed validation from recommendation 1: Now reads "The policy should specify appropriate use and storage after the completion of newborn screen testing and verification of results according to laboratory QA procedures."
- Combined recommendations 3 and 4 concerning the educational process of the newborn screening system and educating parents

Preparation for requesting comment

- Kept optional recommendation in the paper to obtain additional feedback

Responses received

- The Association of State and Territorial Health Officials (ASTHO)
- Centers for Disease Control and Prevention (CDC)
- Centers for Medicare and Medicaid Services (CMS) - no comments
- National Institutes of Health (NIH)
- Office of Civil Rights (OCR)
- Office of Human Research Protections (OHRP)

Other comments requested

- American Hospital Association
- Council of State Governments
- International Society of Nurses in Genetics
- Midwives Alliance of North America
- National Association of Attorneys General
- National Conference of State Legislatures
- National Governors Association

NIH comments

NIH urges the Committee to become an advocate for research use by setting forth actual recommendations for States to consider. When might consent for secondary use be necessary and what mechanisms might be used to ensure privacy and confidentiality? (pg. 1)

NIH comments

The committee could propose voluntary national standards, including provisions for broad research use that each state could consider for adoption. (pg. 1)

NIH comments

Recommend that the Secretary provide resources to facilitate a national dialogue with the relevant stakeholders across the States, perhaps through the National Conference of State Legislatures. (pg. 1)

NIH comments

The issue of education around newborn screening is critically important and merits a fuller treatment. Recommendation 3 should lay out the two currently uninformed "audiences," i.e., parents of newborns and health care professionals who provide them with pre- and post-natal care. (bottom of pg. 1)

NIH comments

Also with regard to recommendation 3, states may need federal funding support to implement educational programs. Why not recommend that the Secretary provide funds for these activities? (pg. 2)

NIH comments

Although reference is made to the use of opt-in or opt-out approaches in recommendation 4, the paper does not discuss these approaches or when they would be appropriate. Support to States might also be needed to help them address this recommendation. (pg. 2)

NIH comments

Recommendation 5 calls for the development of model consent/dissent processes for the use of residual specimens. A concerted, nationwide effort is needed to develop a national policy and best practices that could be adopted by individual states. (pg. 2)

NIH comments

The Committee should remove the Optional Recommendation in the paper. (pg. 2)

NIH comments

The Committee should consider the potential benefit of suggesting the creation of a voluntary national research repository for blood spots into which parents could voluntarily “opt” their children. (pg. 2)

NIH comments

Additional general comments:

- Add information about current state practices with regard to research use of residual specimens (pg. 2)
- Add information about examples of scientific and medical discoveries made possible using residual dried blood specimens (pg. 3)

NIH comments

Topics that may need further discussion:

- Potential benefits and risks that screening programs should anticipate as they approach the use of residual specimens (pg. 3)
- Anticipated scope of future uses of these resources (e.g., genetic vs. genomic; public health vs. clinical medicine-oriented)

NIH comments

Topics that may need further discussion:

- Possible impact of increased data-generation (e.g., sequencing) and data-sharing on privacy
- Ongoing governance and oversight of future research using these specimens (oversight of distribution, including to whom, for what, and how the specimens will be distributed)

NIH comments

Topics that may need further discussion:

- Policies for the return of various kinds of results
- More robust discussion of (re)consent once subjects reach adulthood, which is an issue that relates back to the question of ongoing oversight and the intent to give results

NIH comments

Topics that may need further discussion:

- Given that residual blood spots are finite resources, what is the optimal approach for allocating the resource among competing uses and needs?
- Do policies for stored blood spots apply to other types of archived newborn specimens (i.e., peripheral blood, buccal swabs, urine specimens)?

NIH comments

Pre-meeting discussion of section-by-section comments:

- **Executive summary – define consumers (pg. 3)**
- **Policy, ethical and legal issues – add international guidelines for specimen repositories (pg. 4)**
- **Ownership – add case law (pg. 4)**

NIH comments

Pre-meeting discussion of section-by-section comments:

- **Stewardship – define stewardship, shorten discussion of examples in Michigan and Denmark (see appendix) and remove discussion of a global consortium (pg. 4)**
- **Privacy protections – accept OCR comments (pg. 5)**

NIH comments

Pre-meeting discussion of section-by-section comments:

- **Awareness and education – add discussion of the role of prenatal care providers in educating parents and themselves and cite more published references on the subject (pg. 5)**

NIH comments

Pre-meeting discussion of section-by-section comments:

- **Consent/dissent – work OHRP comments into the paper and add text box explaining anonymized, unidentified, linked with identifiers, identifiable, completely de-identified, private unless decoded and double coded samples (pgs. 5 and 6)**

NIH comments

Pre-meeting discussion of section-by-section comments:

- **Financial considerations – shorten significantly but include examples of the cost of storage and retrieval (pg. 6)**

OHRP Comments

Points to consider regarding how HHS human subjects regulations may apply in the context of newborn screening activities:

1. The collection of newborn blood spots would not involve research under HHS regulations for the protection of human subjects if the specimen collection for the newborn screening is not modified in any way for a research purpose. This is the case even if it is known the specimens will subsequently be used for research purposes. (pg. 1)

OHRP Comments

2. If the specimens were collected for solely clinical purposes, the retention of specimens for future research studies may involve research, depending on whether the retention of the specimens is being altered due to the plan to carry out research using the specimens. If the retention of the specimens is not altered by the future research plans, then the retention of the specimens is not a research activity. (pg. 2)

OHRP Comments

3. If the creation or maintenance of a specimen repository is a research activity and associated individually identifiable information will be retained with the specimen, then the existence of the repository would involve non-exempt human subjects research. In this case, the repository would require review by an institutional review board, and the informed consent of the subjects or the subjects' legally authorized representative, unless the IRB determines that informed consent may be waived. Another consideration for such studies involving newborns is that the additional regulatory protections for children involved in research will be applicable (45 CFR 46, subpart D) if the research is conducted before the subject reaches the age of majority. (pg. 2)

OHRP Comments

4. The research use of individually identifiable specimens from a repository would involve human subjects research that would require IRB review and consideration of the informed consent requirements under the HHS human subject protection regulations, unless the research meets the criteria for exemption under 45 CFR 46.101 (b) (4). If the research involves non-exempt human subjects research and the subjects will not have reached the age of majority when the research is to be conducted, then the additional regulatory protections for children involved in research will be applicable (45 CFR part 46, subpart D). (pg. 2)