

Genetic Alliance Comments

Secretary's Advisory Committee on Heritable Disorders in Newborns and Children Considerations and Recommendations for National Guidance Regarding the Retention and Use of Residual Dried Blood Spot Specimens after Newborn Screening BRIEFING PAPER

Introduction

Genetic Alliance thanks the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (Advisory Committee) for its leadership in developing national guidance for the storage and use of residual dried blood spot specimens, and for the opportunity to comment on this issue of growing national importance. Genetic Alliance's primary goal regarding residual blood spot storage and use is to protect this unique and highly valuable resource. We agree that, as stated in your briefing paper, residual newborn blood repositories provide "critical information about risk for certain inherited conditions" and "present[s] an opportunity to generate population-based knowledge that can improve the health of children, support families, and provide information critical to understanding the antecedents of both child and adult diseases."

Our comments here emphasize our commitment to consumer perspectives as a critical component of policymaking. In addition, we describe new technological solutions available to facilitate informed decision-making in health.

Summary

The Advisory Committee correctly identifies the lack of existing guidelines for stewardship and operational requirements for successful biorepositories as a barrier to the development of appropriate guidelines for newborn blood repositories. We support the development of national guidelines and best practices and policies for biobanks in general and newborn sample biorepositories in particular. As Genetic Alliance developed its own BioBank, we struggled to define how best to protect subjects' interests and further our novel platform for accelerating translational research. The BioBank now empowers and protects participants, while helping dissolve boundaries between advocacy organizations, academia, industry, and government. As science rapidly advances, guidelines and best practices need to accelerate as well. We think the process the Advisory Committee follows for open, diverse stakeholder engagement will produce thoughtful and effective public policies.

The tremendous potential contribution of residual newborn bloodspots to future medical advances – and thus to helping individuals and families – is impossible to quantify today. No other biobank or database has the potential to advance science in the way that stable blood samples from such an extremely broad demographic cohort have. Genetic Alliance's comments addressing the Advisory Committee's recommendations are based on our core intention of protecting the appropriate storage and use of residual newborn blood spots.

Comments on Recommendations

- 1) *All state newborn screening programs should have a policy in place that has been reviewed by the state attorney general or other appropriate legal authority addressing the disposition of dried blood specimens remaining after newborn screening. Policymakers should consider the value of the specimens as a promising resource for research, the importance of protecting the privacy and confidentiality of families and the necessity of ensuring the public's trust.*

We support this recommendation. Public trust in government actions that lack transparency seems to be waning, especially when the government actions affect such a tender subject as newborn babies. Privacy fears are mounting steadily, and consumers sometimes feel devalued and frustrated when they have little knowledge about government practices affecting them and no opportunity to participate in the decisions. State governments, with the assistance of the federal government, could address these apprehensions related to newborn blood sample retention and research use by taking several actions, including ensuring full compliance with existing law, strengthening safeguards for stored blood and data, helping the public understand the tremendous potential benefits that may result from research involving retained specimens, explaining existing safeguards to the public, and considering new ways to involve parents in decisions regarding their children's specimens.

As the briefing paper explains, compliance with federal privacy regulations, the Health Insurance Portability and Accountability Act of 1996 (HIPAA) Privacy Rule, has been required of newborn screening programs since April 2003. Newborn screening programs currently comply with HIPAA by releasing samples, or information from samples, only on a de-identified basis, unless explicit parental consent has been obtained. The programs also comply with the federal Common Rule protecting human subjects in research by not permitting outside researchers to know the identity of the subjects, unless informed consent for participation has been obtained. Some states require additional measures, such as giving parents a right to opt out of having their children's blood saved and used in research even on a de-identified basis. Despite these existing legal protections, lingering fears about the privacy and security of the samples exist, and in some cases, those fears lead to opposition and litigation.

In response to these concerns and the threats posed to the viability of retention and research uses of newborn blood repositories, Genetic Alliance offers several recommendations regarding privacy and security:

First, we support your recommendation that state programs undergo a fresh legal review by state legal authorities to ensure compliance with all existing laws.

Second, transparency and communication practices about state screening practices could be improved in several respects:

- State policies regarding sample retention and use should be readily available. Specific previous disclosures should be listed publicly, and

perhaps states could be required to submit these policies, and keep them current, to a centralized repository.

- Explaining the technology and medical benefits anticipated from saving and using the samples could be extremely helpful in allaying concerns and fostering trust and public support.
- Communications to the public and state legislators should include more information about the legal mandates and operational safeguards currently applied to the repositories. The fact that unconsented uses or disclosures are made only where the samples or data have been de-identified and Institutional Review Board(s) have evaluated and approved the particular study should be made clear. Other security measures should also be explained. Neither parents nor policymakers can make wise decisions in the absence of such vital information.
- Where parents have legal rights to deny permission for retention and research uses, those rights often need to be better publicized and enforced. Because anecdotal reports exist that hospital staff have occasionally impeded parents from exercising their legally available choices. Educational outreach to health care staff about newborn screening in general should include information about the choices available to parents, and parental rights should be enforced.

Third, while we are aware of no evidence of security breaches involving newborn sample data or repositories, it is likely that the protective controls used need to be modernized and improved to meet contemporary security standards. States, as well as any agents they employ to manage these repositories, should be required to comply with the full range of administrative, technical, and physical safeguards required by the HIPAA Security Rule. In an era where a rogue employee or a determined hacker can cause vast damage by misappropriating and misusing confidential information, these repositories should employ comprehensive and up-to-date security controls, including role-based access management, audit logs and flags, intrusion detection, and periodic reassessment. These repositories are a vital public trust, and they should be protected as such by their custodians.

Fourth, in response to privacy fears and concerns, the issue of parental consent needs careful examination by policymakers. All constituents and interests need to be heard and considered, including those fearful of privacy or autonomy violations but also those families affected by diseases or conditions that make them highly motivated to see medical advances to benefit their loved ones. Litigation or opposition generated by a handful of parents anxious about privacy should not automatically trump the wishes of parents anxious to see bloodspots used to accelerate medical progress. For example, in Texas, where more than five million specimens were incinerated this year as part of a settlement of a case brought by five parents, other parents have since come forward to express their unhappiness that their children's samples were destroyed. These consent issues thus require careful and thorough

analysis by policymakers, considering input from diverse constituents, including not only consumers but also researchers able to explain future potential benefits from their studies.

In analyzing the complex subject of consent, policymakers should also be mindful of new technology that can facilitate specific and dynamic consent opportunities. These technology capabilities, which are considerably easier and more efficient than choice options would be in a paper-based setting, are explained in more detail in our response to Recommendation (5).

- 2) ***All state newborn screening programs should have a policy in place that has been reviewed by the state attorney general or other appropriate legal authority that specifies who may access and use dried blood specimens once they arrive at the state- designated newborn screening laboratory, including further access after newborn screening tests are completed.***

We support this recommendation. Recognizing the complementary nature of developing appropriate research use, specimen access, and informed consent policies, Genetic Alliance recommends that the Advisory Committee specify that these policies be developed in an inclusive and parallel process. Specifically, Genetic Alliance suggests that the Advisory Committee include in its recommendation a requirement that program specimen access policies be easily accessible (for example, via the program's website) to the public. Such access policies would promote accountability and build trust without constraining or restricting the capacity of biorepositories to facilitate discovery research. The Advisory Committee should recommend that states implement audit trails and flags and other internal controls, as discussed above. Technology platforms for clinical research participation management now enable individuals to 1) set their preferences for what is deemed acceptable research to the participant (as opposed to a general policy applied to all) and 2) track who accesses an individual's biological sample or clinical data, and for what reasons.

Tightly related to appropriate research use and specimen access policies, Genetic Alliance recommends that the Advisory Committee solicit comments from the newborn screening community on the establishment of a voluntary virtual national repository and oversight board, similar to an Institutional Review Board (IRB) but with more consumer/community involvement, as referenced in the briefing paper. The Advisory Committee should convene a forum to discuss such a solution to expedite the translation of discoveries to benefit the newborn screening system and research community as a whole, to complement and leverage state-by-state initiatives. The voluntary national repository could serve as a virtual biobank, allowing samples to remain with state programs, while utilizing information technology to facilitate resource and data sharing within and across programs. A national oversight board, with majority representation from consumers, would be appropriate to expedite studies, encourage collaboration, and serve as a foundation for the development of consistent policies about appropriate research use and specimen access. Careful collaboration with primarily state-based or institution-based research models should mitigate the problems associated with a national IRB.

- 3) ***All state newborn screening programs should develop a well-defined strategy to educate health care professionals who provide patients with pre-and post-natal care about newborn screening and the potential use of residual newborn screening specimens for research.***

Genetic Alliance suggests that the Advisory Committee strengthen Recommendation 3 to explicitly state that the primary target of a provider education initiative should be prenatal care providers. The importance of the prenatal care provider as newborn screening educator has been defined in multiple studies and forums. This is in part because of the benefits to the parent of obtaining newborn screening information early in pregnancy: emphasis on educating expectant parents ensures the process and benefits of newborn screening are clearly communicated when families are prepared to learn about and accept them in the context of pregnancy and their baby's birth, as opposed to closely following the birth of a child, which is typically an emotional and overwhelming time. With a strong foundation of education and engagement initiated in the prenatal period, pre- or postnatal providers can more easily engage their patients in a dialogue on residual blood spot storage and use.

The Advisory Committee should include in its recommendation, in addition to clarification of responsibility for educating families on residual blood spot storage and use, that policies surrounding strategies to educate providers include incentives for providers to educate their patients. The articulation of responsibility and available incentives should be extended to prenatal education sources outside of the obstetrician: nurse-midwives, doulas, and childbirth educators all participate as part of an expectant mother's prenatal care team, and their ability to serve in newborn screening education should not be overlooked.

Genetic Alliance also requests that Recommendation 3 be strengthened by specifying that newborn screening programs must share best practices and strategies to educate healthcare professionals through collaborative entities, such as the HRSA genetics and newborn screening regional collaboratives, in a unified effort to improve provider education. These strategies must utilize novel education methods beyond paper-based resources and include a range of health and community professionals involved in pre- and postnatal care. These entities will facilitate making educational information for providers readily accessible at the point-of-care, recognizing that many providers will work in multiple states across their careers and will need to re-learn and re-train themselves on specific state policies for residual blood spot retention and use.

- 4) ***All state newborn screening programs should work proactively to ensure that all families of newborns are educated about newborn screening as a part of prenatal and postnatal care.***

Genetic Alliance supports the Advisory Committee's recommendation, especially the details provided in the recommendation's supporting paragraph proposing that "processes should be in place to evaluate the extent, timing and understanding of parental education with an eye towards educational program improvement." Genetic Alliance requests the Advisory Committee

not limit its recommendation to education of families of newborns, but simultaneously support the accessibility of information on storage and use policies for the general public.

At various national meetings, newborn screening personnel have expressed concern surrounding integrating education for parents and families on residual blood spot storage and use too tightly with education about newborn screening in general. The concern is grounded in fears that confusion surrounding residual blood spot storage and use could cause parents to unwittingly opt-out of screening for their child all together because they do not understand storage and use policies. Conducting public education in phases, focusing first on the core purpose of newborn screening and then discussing retention and use of residual blood spots, will both protect the newborn screening system and serve to build and facilitate dialogue with the public. Genetic Alliance requests the Advisory Committee support this phased education of parents and the public alike. Further, we ask the Advisory Committee to acknowledge the varied information needs and seeking behaviors of parents and consumers, therefore programs should emphasize providing information that is most actionable and applicable at the point of education, but also provide a place for more layered and deeper learning, perhaps on program websites. Genetic Alliance proposes that Recommendation 4 also include additional information about the format of educational materials. Especially to reach parents that do not have ready access to prenatal or postnatal care services, materials on residual blood spot storage and use, and newborn screening in general, must be readily available online and in various formats where individuals and families have easy access to them.

The recommendation should specify that materials developed for public consumption should adhere to institutional standards and reflect community consensus-building in appropriate education strategies and processes. State programs have recently placed one-pagers on Frequently Asked Questions (FAQs) on their websites that feature the details of their policies, and others have created informational videos that are community and state-specific, customized to the unique characteristics of a population. States must develop materials with an eye toward sustainable, paperless solutions, especially when considering the ongoing nature of an education campaign. The recommendation should specify that newborn screening programs share best practices and strategies to educate consumers through collaborative entities, such as the HRSA genetics and newborn screening regional collaboratives, in a unified effort to improve consumer/parent education. Genetic Alliance recommends the Advisory Committee convene program leaders in newborn screening education, or those programs that have implemented novel education strategies, to craft best practices that can be applied to education on residual blood spot storage and use.

- 5) ***If residual newborn screening specimens are to be available for any purpose other than the legally required newborn screening process for which they were obtained, an indication of the parents' awareness and willingness to participate should exist - in compliance with federal research requirements, if applicable.***

We support this Recommendation, subject to the perspectives regarding development of new public policies regarding consent that we expressed in response to Recommendation (1), and subject to our reservations about blanket consent expressed here. Genetic Alliance

acknowledges that a blanket consent process for future retention and across-the-board research uses may seem easier to implement and thus more attractive to states. We have two responses to this observation, however.

First, a process involving only blanket consent for a wide array of uses, such as all future research uses, is frequently criticized for being meaningless and intrinsically contrary to the principle that consent should be informed by relevant knowledge. As parents realize this and as the entire newborn screening landscape becomes increasingly politicized and controversial, we can expect ever-larger numbers of parents to refuse to grant blanket consent if that is the only choice offered. Large-scale refusals would seriously limit the eventual pool of samples available to researchers and thus would impede research advances.

Second, it is important to be aware that new consent management technology can be a game-changer with respect to difficult consent issues. Technology is available that could readily and efficiently permit parents to grant or withhold consent on a granular, specific basis. For example, parents could allow their child's blood specimen to be used for research improving the newborn screening program itself, any research involving a particular disease, any research done by a particular institution, or on a project-by-project basis. They could specify that they are allowing such research involving fully identifiable or only de-identified data. Moreover, individuals' preferences and perspectives change over time. A parent who declines all future research uses at the emotion-laden time of birth might feel differently later, particularly if someone close to them were stricken with a particular disease. Modern consent management technology can allow for dynamic changes of consent directives. Provided the bloodspots have been retained, parents can change their minds over time, allowing or disallowing uses they may have felt differently about earlier. Each family's tolerance for research participation is unique and variable, and consent management technology that allows for both granular and dynamic consent respects those individual variations. Genetic Alliance encourages the Advisory Committee and states to evaluate and consider adoption of such technology[†], for we think that that granular and flexible parental control is likely to lead to broader public support for vital research uses of newborn blood specimens.

Genetic Alliance requests the Advisory Committee revise Recommendation 5 to offer critical characteristics of: "an indication of the parents' awareness and willingness to participate." Furthermore, the Advisory Committee should provide guidance on the appropriateness of blanket consent versus separate consent, acknowledging the necessity of preserving the primary purpose of newborn screening programs and clarifying research to benefit newborn screening programs from research that falls outside of that scope.]

6) ***Provide administrative support and funding to the SACHDNC to:***

- Facilitate a national dialogue among federal and state stakeholders about policies for the retention and use of residual newborn screening specimens, including model consent and dissent processes;
- Develop national guidance for consent or dissent for the secondary use of specimens and mechanisms to ensure privacy and confidentiality, including methods for opting in or out of repositories; and

- Collect and analyze national data on the utility of any additional consent or dissent processes implemented relative to potential research uses of residual newborn screening specimens;

Genetic Alliance requests that the Advisory Committee add to its activities for which it recommends administrative support and funding that it provide a forum for states to collaborate and share best practices for crafting policies for residual blood spot retention and use.

7) ***Provide administrative support and funding to the Health Resources and Services Administration - Maternal and Child Health Bureau to award grants to states to:***

- Develop model educational programs for the general public on the importance of newborn screening and the potential uses of residual newborn screening specimens to generate population-based knowledge about health and disease; and
- Create educational materials directed to health care professionals and consumers with facts about potential uses of residual newborn screening specimens and other related issues, including those outlined in Recommendation 4.

Genetic Alliance requests the Advisory Committee revise Recommendation 7 to include the revision or customization of existing educational programs for the general public, as opposed to only development. This would include an assessment of existing resources and tools.

Conclusion

Genetic Alliance acknowledges the financial constraints of the states, the reality of the financial burden extended storage and use places on programs, and the support needed to execute the technical aspects of these recommendations. We further acknowledge the support states need to properly fund and execute educational programs on newborn screening generally, not limited to residual blood spot retention and use. We are committed to working with the Advisory Committee and newborn screening programs in identifying and implementing methodologies built upon existing evidence, best practices, and novel education models that efficiently and cost-effectively create meaningful impact across communities and states.

Genetic Alliance's activities related to newborn screening are rooted in the principles that families' needs must remain central to this important public health program and that consumer perspectives must be integrated into public policy development. Our newborn screening projects, namely the Consumer Focused Newborn Screening Quantitative and Qualitative Projects (U33MC07951 and U33MC07952) and the Newborn Screening Clearinghouse (U36MC16509) from the Genetic Services Branch, MCHB/HRSA/HHS, among others, demonstrate our commitment to consumers as proactive collaborators.

Genetic Alliance recognizes the significance of the task before the Advisory Committee. We acknowledge the challenge of bringing together diverse stakeholders to implement policy action on a complex, and at times, divisive issue that is imperative to the advancement of improved clinical care and discovery research. In the briefing paper's discussion of the policy, ethical, and legal issues associated with retention and use of residual dried blood spot specimens

after newborn screening, the Advisory Committee identifies the Genetic Information Nondiscrimination Act of 2008 (GINA) as a crucial stepping-stone toward addressing public concerns of misuse of genetic information. When GINA was enacted, Genetic Alliance chaired the Coalition for Genetic Fairness, a multi-stakeholder coalition of over 500 organizations committed to passing federal genetic nondiscrimination legislation. The legislation's thirteen-year history in Congress reflected the perseverance of the health community to achieve crucial legal protections against employment and insurance discrimination based on genetic information. These protections can encourage individuals to learn about relevant genetic risks without fearing that the information will be used against them. Better public knowledge about the protections mandated by GINA could further help to allay such fears, as well as parents' fears about discrimination risks their children might possibly face if their bloodspots are retained and someday subject to misuse.

Genetic Alliance thanks the Advisory Committee for the opportunity to comment on this historically significant briefing paper on considerations and recommendations for national guidance on the retention and use of residual dried blood spot specimens after newborn screening. The topic will continue to constrain the advancement of newborn screening programs until the implementation of a transparent, systematic, and widespread initiative to engage diverse stakeholders in policy and program development. The newborn screening community looks to the expertise of the Advisory Committee to lead this process, initiating the sharing of successful strategies across states and convening forums for exchanging ideas, but above all, to provide clear, national guidance. We look forward to partnering with you in this endeavor.

Respectfully submitted:



Sharon F. Terry, President and CEO

† Granular and Dynamic consent enables parents to assess research participation with their own family's values associated with privacy and confidentiality and advancement of scientific understanding. For example, some parents may wish to participate in research where the use of identifiers may be extremely valuable to them, especially if the research findings could indicate an increased health risk for a child whose sample is being used for research. For this reason, some parents may wish identifiers to be remain linked to their child's specimen in order to be contacted in the future if information becomes available that may inform the child's care. Other parents may wish their child's residual sample only be made available for research where all samples are anonymized, minimizing the ability to link data from the residual sample back to their child. Some parents may wish their child's sample to only be made available for studies for newborn screening research, such as research that would support the addition of conditions to state newborn screening panels; while others may allow their child's sample to be made available for more broad public health research initiatives, such as measuring the general population's exposure to a particular environmental toxin by measuring the toxin's level in newborn dried blood spots. Each family's tolerance for research participation in the context of

their values is unique, and dynamic consent accounts for this variability. Further, dynamic consent changes the context of ethical and policy issues identified by the Advisory Committee, including control and stewardship, as families are not only able to select research initiatives consistent with their values but also track compliance with those selections, such as through an audit trail.

There are other distinct benefits to granular and dynamic consent that streamline expansion of newborn screening research opportunities while respecting a family's autonomy. If a national voluntary biorepository of residual dried blood samples from newborn screening were to be established, dynamic consent could allow for parents to learn about the national biorepository and its policies regarding appropriate research use and researcher access as determined by its IRB, and determine if they would like to participate. Further, a dynamic consent process would take into account the differing consent rights of children as compared to adults, and that the decision to retain the residual blood sample is a decision the parent makes on the behalf of their child. Dynamic consenting procedures may take into account the rights of the child upon reaching the age of consent. A dynamic consent process facilitated by a technological interface would also allow parents to change their preferences for the use of their child's residual samples on an ongoing basis, as opposed to making the decision once at the point-of-care. By providing layered consenting options for parents, coupled by in-depth education on the benefits and risks associated with participating with certain research initiatives, states can enhance the value of their biorepositories while communicating the value of the parent in the research process.