Providing Newborn Screening Specimens for Research

Legal Issues Faced by State Health Departments

Session 3
June 28, 2012
Agenda

- Sessions 1 & 2 Recap
- Speaker Introductions

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Sessions 1 and 2

- Overview of the Newborn Screening Translational Research Network (NBSTRN)
- Newborn screening and secondary use issues
- Challenges in storing and using residual dried blood spots
  - Role of the law, state variations, public concerns
- Case example – how Michigan has handled many of these issues
- State regulations, legislations and genetic privacy laws
- Minnesota case and impact on public health
- Clinical Utility of Residual Specimens
Speaker Introductions

- Edward Goldman, J.D.
  - Associate Professor at the University of Michigan
  - U of M Health System Legal Office, Deputy General Counsel
  - University of Michigan Law School Graduate
  - State and National Commissions
  - Michigan BioTrust for Health
Speaker Introductions

- Aaron Goldenberg, Ph.D., M.P.H.
  - Assistant Professor, Case Western Reserve University, Department of Bioethics
  - Assistant Director, Center for Genetic Research Ethics and Law
  - Co-Director, Bioethics Masters Degree Program, CWRU
Speaker Introductions

- Ann Willey, Ph.D., J.D.
  - Wadsworth Center, NY State Department of Health
  - Director of Cytogenetics
  - Director of the Laboratory of Human Genetics
  - Director, Division of Laboratory Quality Certification
  - Director, Office of Laboratory Policy and Planning
  - Albany Law School
Session 3 Overview

- Privacy, privacy laws, HIPAA, consent, de-identification, honest broker
- Application to genetics and newborn screening research
- New York state newborn screening program
- Questions and Answers
Privacy, Privacy Laws, HIPAA, Consent, De-identification, Honest Broker

Ed Goldman, J.D.
University of Michigan
What is Privacy?

- Privacy: I have a secret and I tell no one so my information is private.
- Confidentiality: I have a secret and I tell my doctor, lawyer or priest since they are required both by ethics and law to not tell anyone else my secret.
- These are often confused and we refer to privacy as information personal to the patient/subject.
Ethical Rules for Privacy

- It is wrong to disclose information given to you, no matter what your professional responsibility, when it is given to you in confidence.
- Here, there is no legal requirement but rather a moral imperative to honor a request that information not be shared.
Legal Rules for Privacy

- Law can provide a “shield” protecting someone from having to reveal information provided under an expectation of privacy.
- Ex: priest-penitent, lawyer-client, doctor-patient all have state laws that prohibit disclosure of information provided in the context of the professional relationship with specific exceptions (child abuse, etc.) built into the laws.
Laws Specific to Research with Human Subjects

- Federal:


HIPAA Privacy

- Requires researchers preserve privacy of subjects, even after death.
- Requires permission (or waiver from an IRB or Privacy Board) before a researchers can review data in a subject’s medical record.
IRB Rules

- IRB (Institutional Review Board) is required for federal sponsored or FDA research. Its job is to review and approve, modify or reject research protocols.
- The IRB is guided by federal regulations found in the “Common Rule” (common to 17 federal agencies with some modifications for FDA research). See: http://www.hhs.gov/ohrp/index.html
IRB Rules

- Subject information must be protected from disclosure.
- How to think about HIPAA and IRB rules? In general, specific informed consent, including details about privacy, is required prior to human subject research (IRB). If the researcher wants to review medical records then a HIPAA specific consent is also necessary.
Is Consent Always Required?

- An IRB can waive consent if it is not possible to conduct the research without a waiver.
- Ex: Researcher wants to study 100,000 medical records from the 1950’s to see how polio was treated. Records exist but patients cannot practically be located. IRB can waive need for consent if satisfied that information will be confidential.
How Can Medical Information Be Protected?

- Informed consent from the subject.
- Separation of the data from names or other identifying information but researcher keeps a link.
- Complete removal of all identifying information with no link.
- Honest Broker: Where a third party has the link.
Honest Broker Example 1

- The Michigan BioBank (MBB) ([http://www.michigan.gov/mdch/0,1607,7-132-2942_4911_4916-209738--,00.html](http://www.michigan.gov/mdch/0,1607,7-132-2942_4911_4916-209738--,00.html)) obtains de-identified dried blood spots from the Michigan Dept. of Community Health (MDCH). The spots are bar coded so that MDCH can identify the newborn. A researcher is given spots with a new bar code.
Honest Broker Example 2

- Neither MBB nor the researcher can identify the subject but MDCH could, if necessary for clinical or other reasons, identify the subject. Thus, MDCH and MBB act as the honest broker to insure confidentiality.
Tricky Terms

- Confidential: Researcher will not disclose known subject information to any third party.
- De-identified: Data is stripped of identifiers as required by the HIPAA privacy rule. See next slide for details. But a link can be retained.
- Anonymous: All identifying information has been removed and there is no link.
- Protected Health Information (PHI): A HIPAA term covering patient medical, financial info.
De-Identification Elements 1

- Names
- All geographic subdivisions smaller than a state
- Zip Code (can retain initial 3 digits if 20,000 plus people)
- All dates except year (unless over age 89)
- Phone numbers
- Fax number
De-Identification Elements 2

- Electronic mail address
- Social security number
- Medical record number
- Health plan beneficiary number
- Account number
- License/certificate number
- Vehicle numbers
De-Identification Elements 3

- Device identifiers
- Web Universal Resource Locators (URLs)
- Internet Protocol (IP) address number
- Biometric identifiers (finger/voice prints)
- Full face photographic images
- Any other unique identifier
- Note: Can assign a code to re-identify if code is kept secure.
Who Does the De-Identification?

- Investigator must de-identify. May assign a code to re-identify IF:
  1. Code is not derived from information about the individual;
  2. Investigator does not use/disclose the code for any other purpose and does not disclose the mechanism for re-identification. 164.514 (c)
Summary

- Confidentiality is a critical requirement in human subjects research.
- Confidentially is governed by ethics, law, subject permission (consent) and common sense.
- The federal laws are detailed and specific.
- State laws are more general.
- Rare disease research presents special problems.
Residual Dried Bloodspot Use: Parental Attitudes and Privacy

Aaron Goldenberg, Ph.D. M.P.H.
Case Western Reserve University
Center for Genetic Research Ethics and Law
Parental Attitudes in the Media

- “I know the government says my baby's data will be kept private, but I'm not so sure. I feel like my trust has been taken.” *(CNN.com Feb 2010)*

- "Why do they need to store my baby's DNA indefinitely? Something on there could affect her ability to get a job later on, or get health insurance.” *(CNN.com Feb 2010)*

The decision protects individual rights, privacy rights, patient’s rights”…“When parents or individuals find themselves used for research without their knowledge or consent, they begin to distrust not only the research enterprise but also the clinics, the hospitals and the doctors who take that information.” (Brase in Nature Feb 2012)  

“What we wanted to do was to make sure that our children's privacy was being protected and that the state is respecting our rights, because if we don't stand up and make the government do that, nobody's going to do it for us.” (Andrea Beleno, Chron.com Dec 2009)
"People put more information obtainable about their own personal lives out on Facebook and MySpace than from their little blood spots," she says. She urges better public information "to really calm this issue.” (Jana Monaco, AP Feb 2010)  

Moreover, since the bloodspots contain deeply private medical genetic information, Plaintiffs are concerned about the potential misuse of that information and fear the possibility of discrimination against their children and perhaps even relatives though the use of such blood samples and research activity thereon. (Beleno v. Texas-Claim Filed Mar 2009)
Methods for Promoting Public Dialogue on the use of Residual Newborn Screening Samples for Research

- Funded by NIH National Human Genome Research Institute (1R01HG004970), from 9/08 to 7/11
- Research Team:
  - Jeffrey Botkin, M.D.\(^1\), M.P.H, PI
  - Rebecca Anderson R.N.\(^1\), Project Director
  - Erin Rothwell Ph.D.\(^2\), Co-Investigator/Qualitative Research
  - Louis Stark Ph.D.\(^3\), Genetic Science Learning Expert
  - Aaron Goldenberg Ph.D.\(^4\), Bio-ethicist
  - Michelle Lewis, M.D.\(^5\), J.D., Law
  - Matthew Burbank Ph.D.\(^6\), Politic Science
  - Bob Wong Ph.D.\(^2\), Statistician

1 U of U Department of Pediatrics; 2 U of U College of Nursing; 3 U of U Department of Human Genetics; 4 Case Western Department of Bioethics; 5 Berman Institute of Bioethics, Genetics and Public Policy Center, Johns Hopkins University, Baltimore, Maryland; 6 U of U Department of Political Science
Methods

- Focus groups;
  - In-person in Mountain states region (N=14).
  - Online with Knowledge Networks, General public (N=3)
  - Movie, discussion and paper survey.

- Mailed paper surveys, PRAMS like;
  - New mothers N=200
  - Not feasible to add items to PRAMS survey: Mimic methodology
Methods Cont.

- Telephone survey, BRFSS like; Survey only.
  - TX, CO, NM, AZ, MT, NV,
  - N= 1218; General pop, AA, Hispanic, IA and Asian

- Online panel, Knowledge Networks;
  - N= 2280
  - Some viewed movie, some only the survey

*All groups received same survey, differed by delivery format only.*
### Survey Demographics (N=3385)

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<th>Characteristic</th>
<th>n or Mean</th>
<th>% or SD</th>
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<tbody>
<tr>
<td>Age, y</td>
<td>48.73</td>
<td>17.21</td>
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<td>Gender</td>
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<tr>
<td>Male</td>
<td>1404</td>
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<tr>
<td>Female</td>
<td>2451</td>
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<td>Ethnicity</td>
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<td>Non-Hispanic</td>
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<td>Hispanic</td>
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<tr>
<td>Race</td>
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<td>Black or African American</td>
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<tr>
<td>White</td>
<td>2268</td>
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<td>Mother with young children (&lt;1 y)</td>
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<td>No</td>
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<td>Yes</td>
<td>464</td>
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<td>Parent</td>
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<td>No</td>
<td>840</td>
<td>22.4</td>
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<tr>
<td>Yes</td>
<td>2911</td>
<td>77.6</td>
</tr>
</tbody>
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Focus Group Results - Privacy Issues

- Some participants felt that their privacy was invaded by the retention and use of the residual DBS without their permission or knowledge:
  - “It’s a little bit of a violation of your privacy.”
  - “You guys invade my privacy, you’re holding my blood, my DNA somewhat hostage.”
  - “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.”
Public Survey Results


- Bloodspot Use Into: Leftover blood samples from babies can be used for different purposes. One use is to maintain the quality of the screening tests for babies. The samples also can be very valuable for research on diseases that affect mothers and babies, or for general health research. National regulations require that all research be approved by a committee that oversees the protection of the rights, welfare, and privacy of people involved in research.
Risk and Identifiability

- If the leftover blood samples are used for research, how much risk would this be for babies...if the baby’s name is NOT attached?
  - No Risk at all (51%)
  - A little Risk (23.5%)
  - Some Risk (14%)
  - High Risk (11.5%)

- If the leftover blood samples are used for research, how much risk would this be for babies...if the baby’s name is attached?
  - No Risk at all (17.9%)
  - A little Risk (17.6%)
  - Some Risk (31%)
  - High Risk (33.5%)
Sample Use and Identifiers

- Would it be alright to use your sample for research if information connecting you to the sample was REMOVED?
  - Definitely/Probably Alright (70.2%)
  - Definitely/Probably NOT Alright (29.8%)

- Would it be alright to use your sample for research if information connecting you to the sample is STILL ATTACHED?
  - Definitely/Probably Alright (47.5%)
  - Definitely/Probably NOT Alright (52.5%)
"It may be very difficult and costly to find many parents after several years. If parents cannot be contacted, what would be the best thing to do with their baby’s leftover samples?

- With Privacy Safeguards in Place, Allow the Samples To Be Used for Important Health Research (71%)
- Do Not Allow the Samples To Be Used for Important Health Research (29%)
Privacy vs. Return of Research Results

- Do you think it's more important to: Allow Notification of Parents If Something Important Is Learned vs. Provide Greater Privacy Protection
  - Allowing Notification More Important (64%)
  - Greater Privacy Protection (36%)
Privacy and Confidentiality Protection Policies

- 26 states have regulatory language that information related to newborn screening is considered confidential\(^1\)
  - 14 state regs. state that confidential information may be released with parental consent
- State genetic privacy laws
  - ~8 address NBS (few exempt NBS info)
- Genetic Information Non-Discrimination Act (GINA)

Genetic Information
Non-Discrimination Act (GINA)

- GINA protects individuals from discrimination based on genetic information:
  - Health insurers cannot use a person’s genetic information to set eligibility requirements or establish premiums
  - Employers cannot use a person’s genetic information in decisions about hiring, firing, job assignments, or promotions.

- GINA Does NOT Apply To
  - life, disability, or long-term-care insurers
  - employers with fewer than 15 employees.
  - members of the United States military, veterans obtaining health care through the VA
  - individuals using the Indian Health Service, or
  - federal employees in the Federal Employees Health Benefits program.

From NCHPEG Website:
http://www.nchpeg.org/index.php?option=com_content&view=article&id=97&Itemid=120&limitstart=1
Limits of Privacy Assurances

- Hard to characterize actual harms---makes privacy protections more difficult
- The Identifiability of DNA? Changes to fed HS policies- ANPRM
- Desire for Control:
  - “I think the screening program is good. They test for things that can make babies really sick initially,”...“if they’d asked me if I would consent for this blood to be used for specific medical research, ... I would have probably said yes.”
Thank You!

- Jeff Botkin and the Residual Newborn Screening Samples Team
- The Center for Genetic Research Ethics and Law NHGRI-ELSI Grant #P50-HG003390
New York State Newborn Screening Program

Ann Willey, Ph.D., J.D.
State Specific Newborn Screening Specimen Access

- Enabling Statute
  - NYS PHL 2500a
- Implementing Regulations
  - 10 NYCRR 69-1
- Program Policy
  - <www.wadsworth.org/newborn/index.html>
- Program Procedures
Protections for Specimen Use and Access

- Parental Notice
- Institutional Review Board review of overall project and individual requests for specimen or data access
- De-identification of specimens
  - Limited data set
  - Aggregate Data groups
  - Minimum group size
- Parental Consent prior to use of individually identifiable specimens
Parental Notice

- Opportunities
  - Preconceptual?
  - Prenatal
  - Hospital discharge
  - First pediatric visit
Message

“Your child’s specimen(s) will be stored by the Newborn Screening Program for up to 27 years under secure conditions where access is strictly controlled. Should the need arise, the specimen(s) may be used for diagnostic purposes for your child with appropriate consent. A portion of the specimen will also be stripped of all information that might identify your child and may be used in public health research that has been reviewed and approved by a Board charged with overseeing compliance with all applicable laws and ethical guidelines. You may have your child’s specimen(s) destroyed or prevented from being used in public health research by calling xxx-xxx-xxxx.”
“IRB” Review and Program Policy

- A document describing the state program specific guidelines should be posted and reviewed regarding:
  - requirements or limitations for use, publication and data retention
  - State program IRB information and expectations for IRB review at the participating investigator’s institution
  - Execution of MTAs (Material Transfer Agreements) if required by the state program
    - May require return of unused residual specimen
    - May prohibit sharing or sale of specimens to other investigators
  - Assurance of compliance with all applicable originating state program terms and conditions
De-Identification of Specimens

- Limited data associated with the specimens
  - Year of collection
  - State of collection
  - Age at collection
  - Birth weight
  - Feeding status
  - Storage Conditions
  - Gender
  - Race/ethnicity (as assigned by person collecting the specimen)
  - Screen Negative or Positive diagnosis of specified condition in the state screening profile

- Data provided as aggregate summary rather than by individual specimen
- Minimum group size
Parental Consent for Use of Identified Specimens

- Logistics
  - When
  - By whom

- Content
  - Specific to the intended project
  - Who will have access to results
  - Possible Certificate of Confidentiality
Other Possible State Specific Protections

- Applicability of State Genetic Confidentiality Statutes
  - NYS Civil Rights Law 79-L
- State Anti-genetic-discrimination laws
  - NYS Executive Law 292, 296
- Laboratory certification (release of any test results obtained from the specimens)
  - NYS PHL Article 5 Title V, 10 NYCRR 58, 19
Question and Answer
Please use the dialog box on the webinar to submit questions
Continuing Education Credits

For information about how to pursue continuing education credits for your attendance at this webinar, contact:

Irina Smotrich
American College of Medical Genetics & Genomics
Project Coordinator
ismotrich@acmg.net
Upcoming in the Webinar Series

<table>
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<tr>
<th>Session 4: Protection of human research subjects</th>
<th>July 26th</th>
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<tr>
<td>Speakers: Harry McGee, M.P.H., Rachel Nosowsky, J.D.</td>
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<td>Session 5: Ownership and control of research results</td>
<td>Aug 30th</td>
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<td>Speakers: Natasha Bonhomme, Anne Comeau, Ph.D., TBD</td>
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<td>Session 6: Summary of core issues</td>
<td>Sept 27th</td>
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<td>Speakers: Brad Therrell, Ph.D., Michelle Lewis, M.D., J.D., Denise Chrysler, J.D.</td>
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All sessions at 1:00 pm EST
Thank You

Contact Info:

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- Amy Hoffman, MPH, Project Manager
- Email: nbstrn@nbstrn.org
- Phone: 301-718-9603
- Website: www.nbstrn.org