Massachusetts Newborn Screening – Public Health Service, Research and Public Trust

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2016 APHL Annual Meeting

Albequerque, New Mexico

June 8, 2016

University of Massachusetts Medical School

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Proposed changes to the Common Rule

- General Background
- High level summary of proposed changes
- Background related to Newborn Screening
- Massachusetts experience and current plans

Good Intentions…

Proposed Changes to the Common Rule: Impact on Public Health Laboratories

Proposed changes: Background

- Good Intentions, thoughtful consideration
- Covers a wide spectrum of projects
- Protect research participants
  - Advancing technologies
  - Emerging scope and breadth of clinical trials
  - Enhanced call for informed decision-making

https://s3.amazonaws.com/public-inspection.federalregister.gov/2015-21756.pdf

Proposed changes: 8 Point Summary

1. Biospecimens will be considered to be human subjects.
   - *The term "de-identified" or "non-identifiable" is moot*
   - *Covers the obtaining, use, and study of biospecimens*
   - *QA excluded, specific exclusion if characteristic already known*
   - ***Informed consent will be generally required before research use of any biospecimen that is not covered by an exclusion.***
   - ***Introduction of the term "Broad Consent" see later points too***

Proposed changes: 8 Point Summary

2. Proposed Explicit Exclusions
   - *Program Improvement Activities (internal monitoring)*
   - *Quality Assurance and Improvement Activities*
   - *Public Health Surveillance Activities*
3. Proposed Exclusions of Activities that are low risk and already subject to controls
   - *Does not include secondary use of biospecimens*
   - *Surveys, information gathering, analyses*

Proposed changes: 8 Point Summary

4. Exemption for secondary use of private identifiable information with caveats:
   - *Notice of potential research use given*
   - *Privacy safeguards are in place*
   - *Limited to specified request*
5. Exemptions – Documentation, Limited IRB Review and Broad Consent
   - *Written consent for storage or maintenance*
   - *Use of HHS-developed form*
   - *Anticipated applications for newborn blood spots*

Proposed changes: 8 Point Summary

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Proposed changes: 8 Point Summary

6. Obtaining, Waiving, Documenting Informed Consent
   - *3 new elements (commercial, disclosure, and option to refuse re-contact)*
7. Details on Broad Consent
   - *Good for collection of specimens and data for 10 years*
   - *Notes on withdrawing consent*
8. *Waiver of consent or documentation*
   - *Considering that NO waivers be allowed.*

Background: Newborn Screening Saves Lives Reauthhorization Act Section 12 (2014)

SEC. 12. INFORMED CONSENT FOR NEWBORN SCREENING RESEARCH.

(a) IN GENERAL.-Research on newborn dried blood spots shall be considered research carried out on human subjects meeting the definition of section 46.102(D(2) of title 45, Code of Federal Regulations, for purposes of Federally funded research conducted pursuant to the Public Health Service Act until such time as updates to the Federal Policy for the Protection of Human Subjects (the Common Rule) are promulgated pursuant to subsection (c). For purposes of this subsection, sections 46.116(c) and 46.116(d) of title 45, Code of Federal Regulations, shall not apply.

(b) EFFECTIVE DATE.-Subsection (a) shall apply only to newborn dried blood spots used for purposes of Federally funded research that were collected not earlier than 90 days after the date of enactment of this Act.

(c) REGULATIONS.-Not later than 6 months after the date of enactment of this Act, the Secretary of Health and Human Services shall promulgate proposed regulations related to the updating of the Federal Policy for the Protection of Human Subjects (the Common Rule), particularly with respect to informed consent. Not later than 2 years after such date of enactment, the Secretary shall promulgate final regulations based on such proposed regulations.

Newborn Screening is

a public health program that

provides an opportunity for early identification and early treatment

of infants with conditions that otherwise would go unrecognized

prior to irreversible clinical damage.

Newborn Screening is

highly successful.

~14,000 infants treated annually who otherwise would succumb to an illness that could have been treated

Newborn Screening Advances are

An example of continuous learning

through research

for quality improvements

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1998 - present
MA DPH Newborn Screening Committee Recommends Processes for NBS Expansions

1. Keep the focus on conditions proposed to expand NBS
2. Evaluate known benefits and harms
3. **Offer** expanded screening for conditions that do not meet criteria for state-authorized mandate

    Run Statewide pilot program(s)

    Collect data for further evaluation

    Report back to committee

Massachusetts Human Subject Review

2 independent HSRBs note:

- When state-authorized Mandate is not justified by Committee

  And

- Data collection for the evaluation of benefit is planned

This is a study, requiring informed consent

2 HSRBs also note:

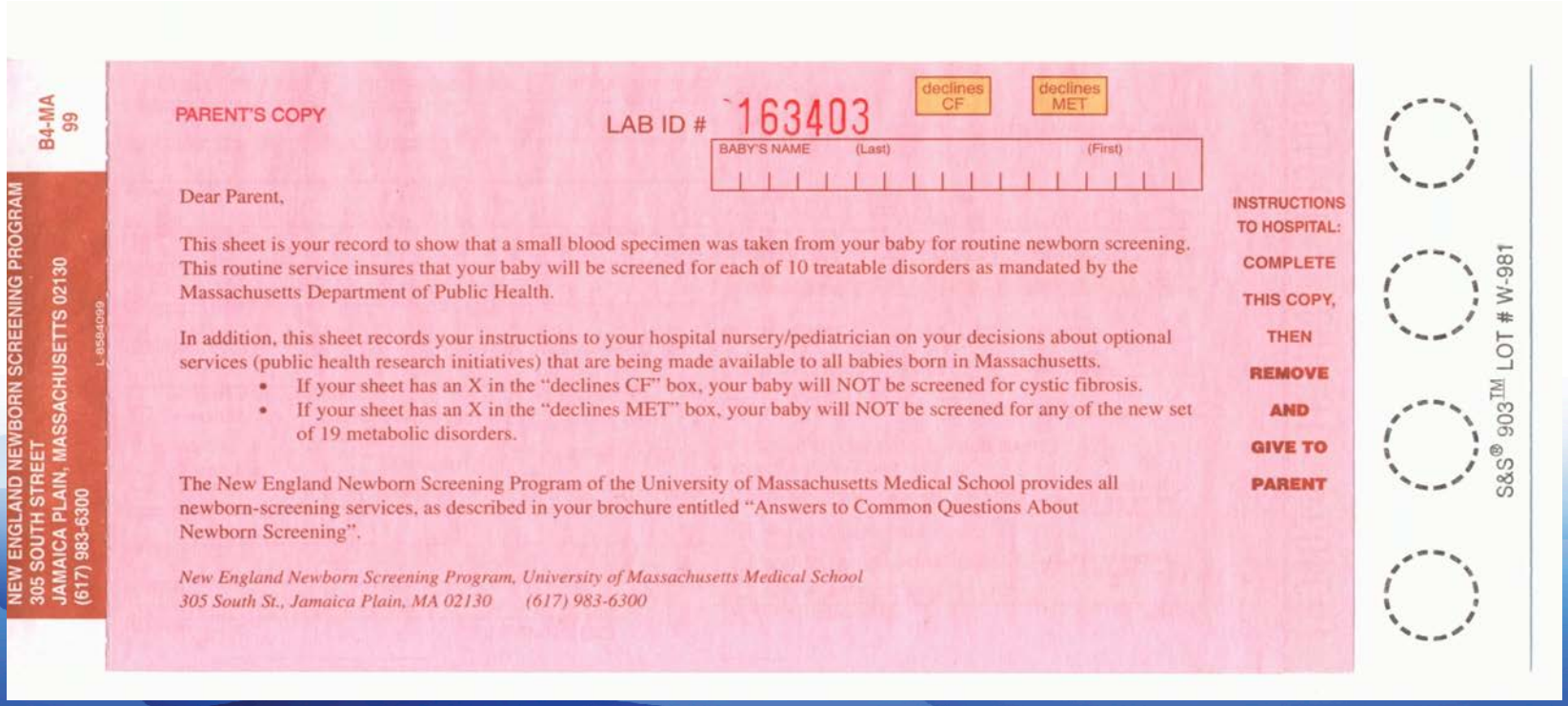
- Presumed benefit to infant
- Low risk- no additional blood and clinical follow up available
- Operational practicalities- informed consent for birth cohort

  Verbal Informed Consent

  OHRP buy-in

Statewide NBS Pilots : Informed consent protocol (current)

- Education via brochure
- Ask each infant's parent
- Provide parent with record of their decision



B4-MA 99

NEW ENGLAND NEWBORN SCREENING PROGRAM
305 SOUTH STREET
JAMAICA PLAIN, MASSACHUSETTS 02130
(617) 983-6300

L-0584099

PARENT'S COPY

LAB ID # 163403

declines CF

declines MET

BABY'S NAME (Last) (First)

Dear Parent,

This sheet is your record to show that a small blood specimen was taken from your baby for routine newborn screening. This routine service insures that your baby will be screened for each of 10 treatable disorders as mandated by the Massachusetts Department of Public Health.

In addition, this sheet records your instructions to your hospital nursery/pediatrician on your decisions about optional services (public health research initiatives) that are being made available to all babies born in Massachusetts.

- If your sheet has an X in the "declines CF" box, your baby will NOT be screened for cystic fibrosis.
- If your sheet has an X in the "declines MET" box, your baby will NOT be screened for any of the new set of 19 metabolic disorders.

The New England Newborn Screening Program of the University of Massachusetts Medical School provides all newborn-screening services, as described in your brochure entitled "Answers to Common Questions About Newborn Screening".

New England Newborn Screening Program, University of Massachusetts Medical School
305 South St., Jamaica Plain, MA 02130 (617) 983-6300

INSTRUCTIONS TO HOSPITAL: COMPLETE THIS COPY, THEN REMOVE AND GIVE TO PARENT

S&S® 903™ LOT # W-981

Education

- Pilot program is different from routine newborn screening services
  - Pilot programs are research
  - We are studying whether newborn screening for the list of conditions included in pilot is helpful
  - To do this, we will report our findings to your baby's doctor and we will study the findings from many babies who participate.
  - If your baby has a pilot condition, newborn screening for that pilot conditions may be lifesaving for your baby - or – it may present significant challenges to you or your baby without benefit for your baby; we do not have enough information to tell you what will happen.
  - Participation is optional
  - If you want to participate, you must consent

MA NBS Committee Interim Conclusions

- DATA
  - Move some, not all conditions to mandatory panel

- CONSENT FORMAT
  - DO-ABLE
  - Disseminates knowledge about the service in general
  - PROVIDES FRAMEWORK FOR FUTURE QI / RESEARCH

Continuation of the Pilot Consent Protocol Facilitated Expansion in Massachusetts.

CF and metabolics 1999 -2009

Total Number of Babies screened	>	**784,000**	
Total number declined		**8,000**	**1%**

SCID 2009-present

Total Number of Babies screened	>	**461,000**	
Total number declined		**3,600**	**0.78%**
Year one declined			**1.6%**

Experience

- CONSENT FORMAT
  - Simpler may be safer
  - Protocols included in competencies
  - Highest rates of declining in affluent communities
  - Disseminates knowledge about the service in PARTICULAR
    - Awareness of genetic testing among providers
    - Awareness of genetic testing among population
  - PROVIDES FRAMEWORK FOR FUTURE QI / RESEARCH

We were able to answer Massachusetts and National Questions 1999-2015 in strict compliance with ethical standards

- How would we define positive screen?
- Who would we recommend have immediate diagnostic evaluation *vs* sending a repeat specimen?
- Who would we find?
- Would population-based clinical outcomes be as promising as originating treatment data?

New Challenges

Technologies may greatly expand list of conditions at the same time as expanding questionable outcomes–

We were able to answer Massachusetts and National Questions…

Could we do it again now?

Example:
Massachusetts SCID NBS Statewide Pilot

ASSAY DEVELOPMENT

SCREENING IMPLEMENTATION

ALGORITHM REFINEMENT

TECHNOLOGY TRANSFER

Grant # IV01-EH000362-03

Implementing SCID NBS with Multiplexed Assays in an Integrated Program Approach
CDC National Center for Environmental Health

University of Massachusetts Medical School
UMASS
umassmed.edu

Good intentions dot dot dot…

MA will now have to seek 2 permissions:

- One specific for pilots
- One broad for use of dried blood spots

How does anyone develop an assay that is universally applicable when available specimens represent only a portion of population?

Dot dot dot….

Does broad consent increase protections for participants in research ?

Or maybe just practitioners of research?

Mamma Mia!

Thank you