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# Challenges and Opportunities in Using Newborn Screening Samples for Translational Research: A Workshop of the Institute of Medicine

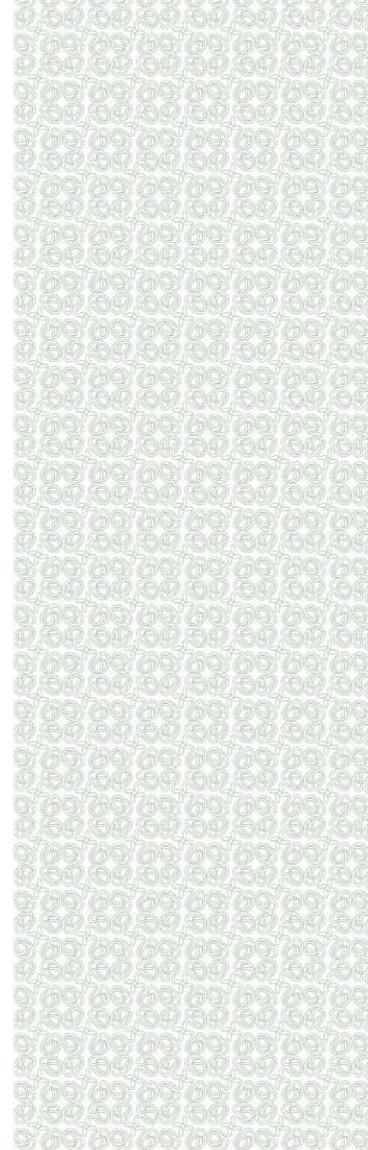
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Roundtable on Translating Genomic-  
Based Research for Health

June 16, 2010



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# Workshop Goals

1. Ensure that the various stakeholders were identified and their views and interests were heard
2. Promote a discussion where potentially opposing viewpoints could be heard and considered
3. Potentially find common ground

Disclaimer: All presented information comes from the statements and opinions of the individual presenters and participants and should not be construed as reflecting any group consensus from the members of the Roundtable or the Institute of Medicine.



# Focal Questions

1. What are the benefits of making newborn screening samples available for research?
2. How do we protect the privacy and rights of individuals if we allow samples to be used for research?
3. How can we make these samples available without compromising the main function of the newborn screening program?



# Session Topics

1. CURRENT STATUS REGARDING STORAGE OF NBS SAMPLES
2. RESEARCH OPPORTUNITIES ON NBS SAMPLES
3. SURMOUNTING CHALLENGES
4. WEIGHING THE VALUE



# CURRENT STATUS REGARDING STORAGE OF NBS SAMPLES

- Current state practices and policies
- Rationale for storing newborn screening samples



# CURRENT STATUS REGARDING STORAGE OF NBS SAMPLES

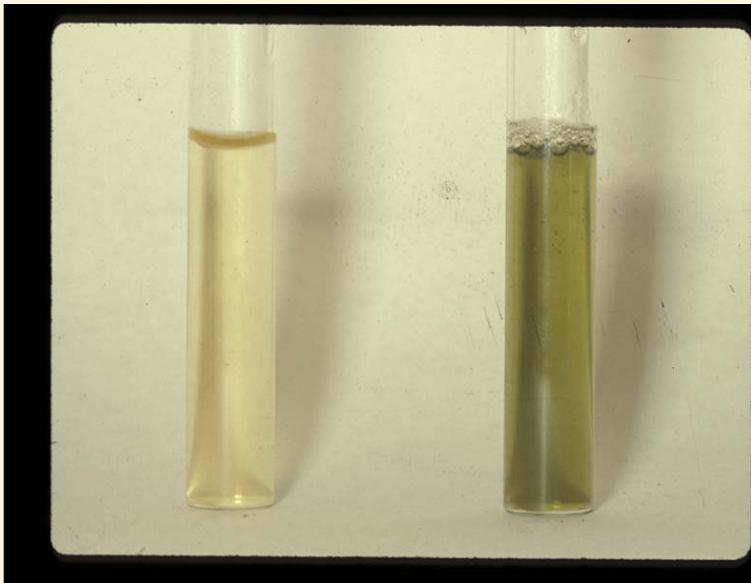
- As of May 2010, 18 states have legislation regarding storage and use
  - Many states are currently re-examining their policies
- Individual states store these samples not only for use in newborn screening, but also for secondary uses
  - Analysis of 162 analytes, development of new tests, public health surveillance, reflection of fetal exposures, sources of DNA, retrospective studies, unexpected uses, and discovery



# CURRENT STATUS REGARDING STORAGE OF NBS SAMPLES

Why store residual dried blood spots?

“They are irreplaceable! ....and they sure beat daily wet diapers by the hundreds !!” (K. Pass)



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Adapted from K. Pass

# RESEARCH OPPORTUNITIES ON NBS SAMPLES

- Continuum from service to research
- Opportunities for broader research
- Implications of dataset linkage



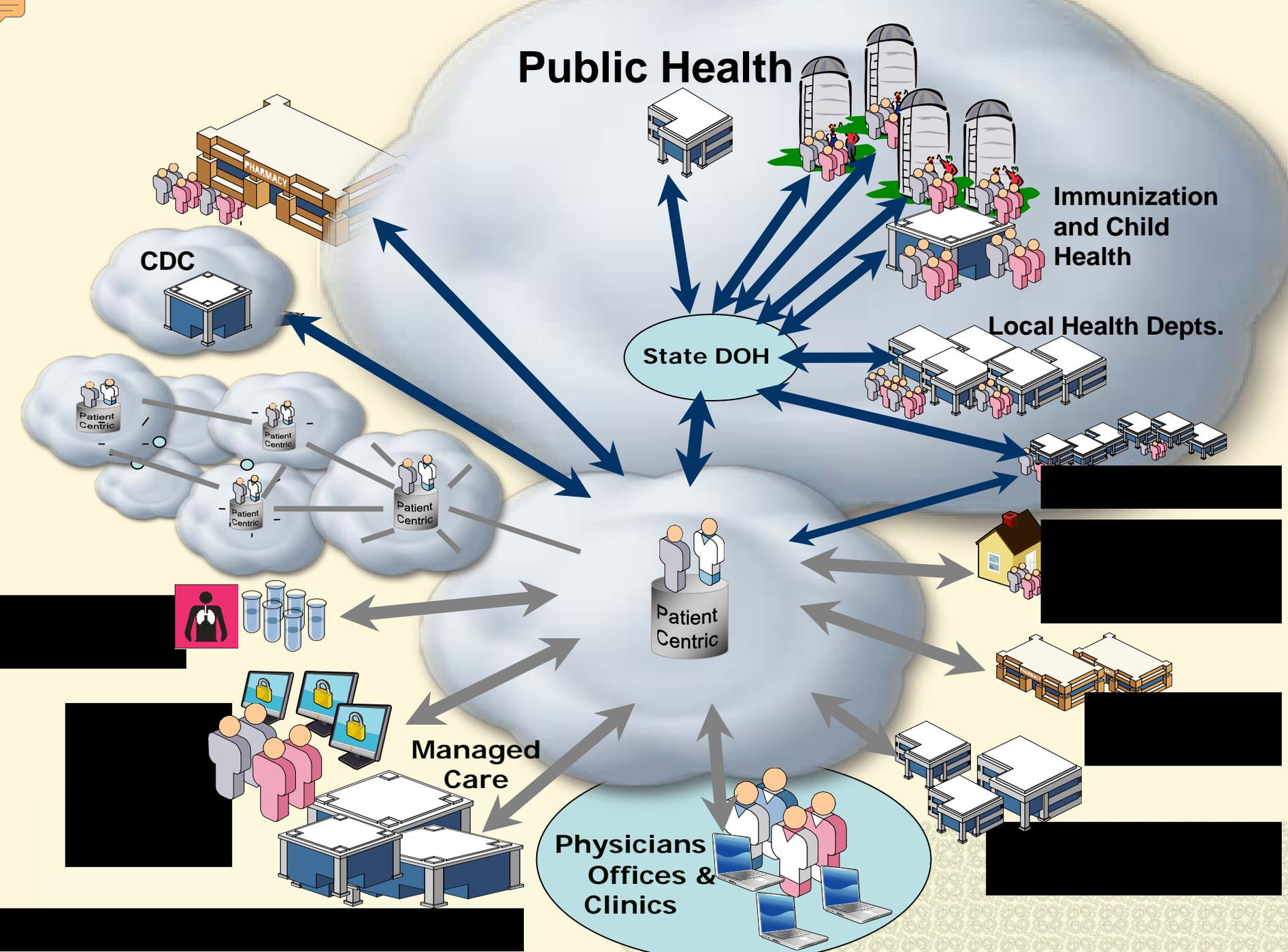
# RESEARCH OPPORTUNITIES ON NBS SAMPLES

- Epidemiological/Public Health Benefit
  - Disease prevalence or susceptibility, global health, research in developing countries, case control studies, birth cohort studies, and longitudinal studies
- Expanded Screening
- Data Linkage
  - Creation of a virtual health profile which integrates much of the siloed health databases of the state
    - Done with protections for privacy and taking into account legal, regulatory, and technology issues





# Public Health



CDC

State DOH

Immunization and Child Health

Local Health Depts.

Patient Centric

Patient Centric

Patient Centric

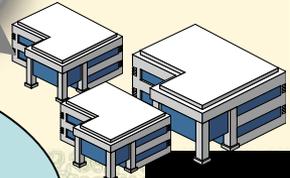
Patient Centric

Patient Centric

Managed Care

Physicians Offices & Clinics

PHARMACY



# RESEARCH OPPORTUNITIES ON NBS SAMPLES

- Data Linkage
  - Benefits
    - Allows better coordination of care and treatment
    - Have meaningful health information exchange between clinicians and the public health department
    - Creates a standardization of data and diagnostic criteria



# SURMOUNTING CHALLENGES AND WEIGHING THE VALUE

- Importance of retaining the core mission of newborn screening programs
- Balancing issues from the patient/parent perspective
- Informed consent and stewardship
- Legal issues related to the usage of newborn screening samples
- Examining the potential benefits that could be derived from the use of dried blood spots for translational research versus the issues that need to be overcome

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# Tempering the Opportunities

- Clear issues which participants indicated needed to be resolved
  - Education
  - Funding
  - Consent
  - Transparency
  - Stewardship and Accountability
  - Policies



# Education

- Public outreach to inform about the newborn screening program and use of dried blood spots
- Education about the public health infrastructure
- Provider education
- Risk of destroying samples
- Financial resources to provide education are not available



# Funding

- Newborn screening programs have minimal funding to complete their main mission of providing screening
- Many states are considering or are destroying samples due to financial constraints of storage



# Consent

- The mandated language of consent may hamper program function
- New innovative ways of obtaining consent (adaptive and manageable by consentee)
- Project-specific approval, consent, and review



# Transparency

- Allowing the public to access their own data
- Integrating participants into the process
- Building relationships proactively



# Stewardship and Accountability

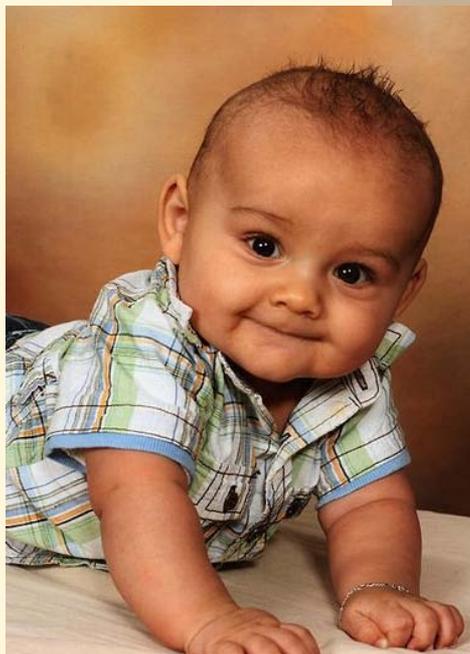
- Composition of oversight committees
- Definition of ownership and what constitutes fair use
- Project-specific approval, consent, and review
- Repercussions for failing to uphold agreements
- On-going follow-up
- Prioritization of different uses of samples



# Policies

- HIPAA
- Law Enforcement
- Return of results





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