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Challenges and Opportunities in Using Newborn Screening Samples for Translational Research

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Newborn Screening: A Public Health Program

Newborn screening is a public health program that identifies rare genetic, metabolic, hormonal, and functional disorders in infants right after birth and assures early management and comprehensive follow-up care for those affected.

Newborn Screening: Sample Collection

Newborn Screening Programs

- Sample collection and submission to the laboratory
- Laboratory testing
- Reporting of results
- Diagnostic confirmation
- Referral for treatment
- Long term support of patients and families
- Program evaluation

Newborn Screening -- Principles

- Serious disease or disorder that significantly impairs health beginning in infancy;

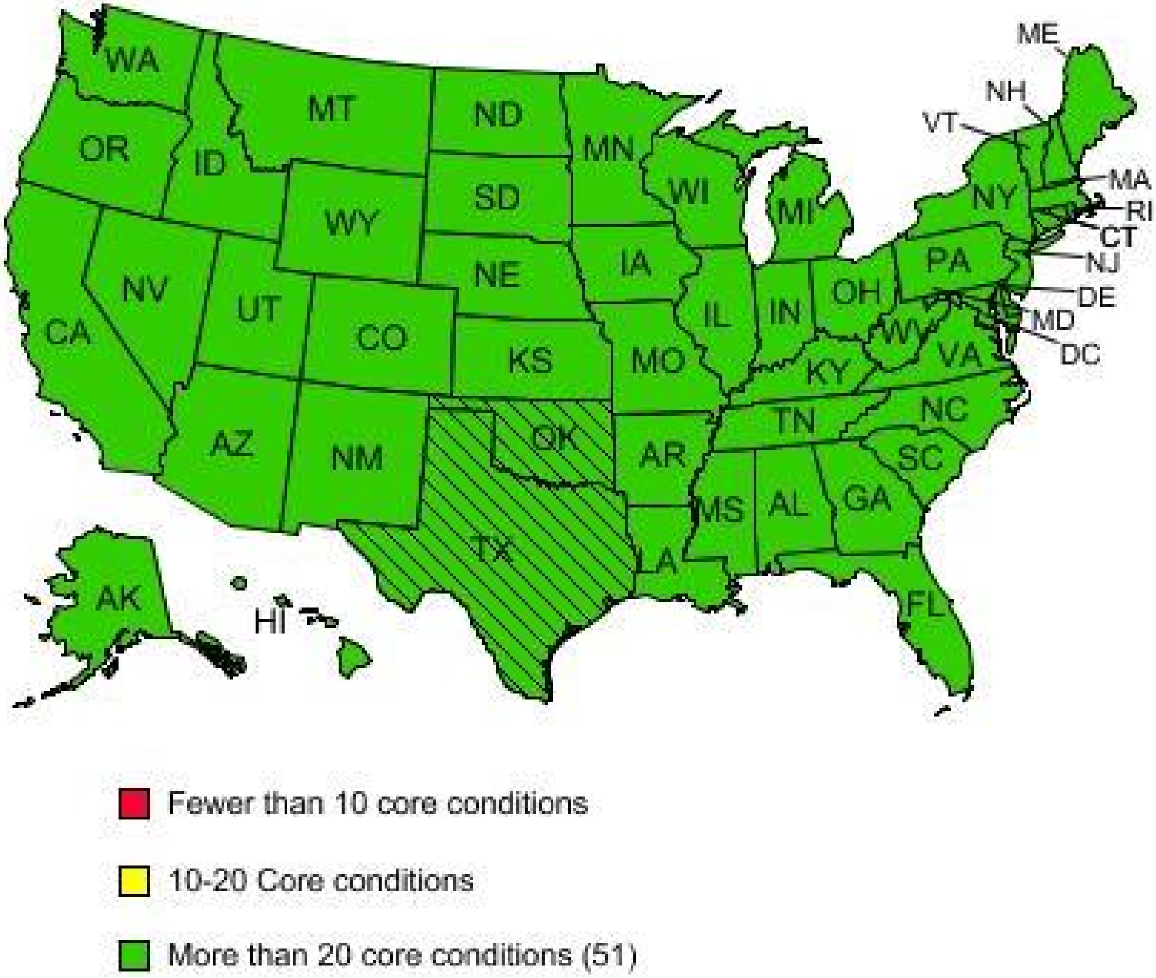
- Able to identify disorder before symptomatic;

- Valid, reliable, sensitive and specific screening methods able to be performed shortly after birth;

- Mandatory that all babies are screened without parental informed consent; some states allow opting out for various reasons;

- Documented benefit of early detection, timely intervention and efficacious treatment;

Newborn Screening Tests



* Hatch marks indicate testing for some conditions required but not yet implemented.

In 2010:

42 States +DC screen for all 29 disorders;

and All States screen for 26 or more disorders

Newborn Screening
Estimated Number of Cases in U.S. (2006)

Disorder	Estimated Number of cases
Amino Acid Disorders	**283**
Organic Acid Metabolism Disorders	**260**
Fatty acid Oxidation Disorders	**408**
Hemoglobinopathies	**1775**
Primary Hypothyroidism	**2156**
Cystic Fibrosis	**1248**
Other (Cong Adrenal Hyper, Biotin, Galactosemia)	**488**

Total 6618

MMWR Morb Mortal Wkly Rep.
2008 Sep 19;57(37):1912-5.

Potential Uses of Residual Blood Spots from Newborn Screening

- Program quality assurance and test validation
- Develop new screening methods
- Parental requests for additional testing (particularly in cases of an infant death)
- Forensic tests—police, FBI, Homeland Security
- Public health research--populations
  - Prevalence of disease
  - Identify new infectious agents, toxins, bioterrorism, etc.
- Health related research--individuals
  - Identify affected children
  - Assess long term health status

Residual Blood Spots from Newborn Screening

Stakeholders

Child

Residual Blood Spots from Newborn Screening

Stakeholders

Child

Family

Residual Blood Spots from Newborn Screening

Stakeholders

Scientists and Clinicians

Child

Family

Residual Blood Spots from Newborn Screening

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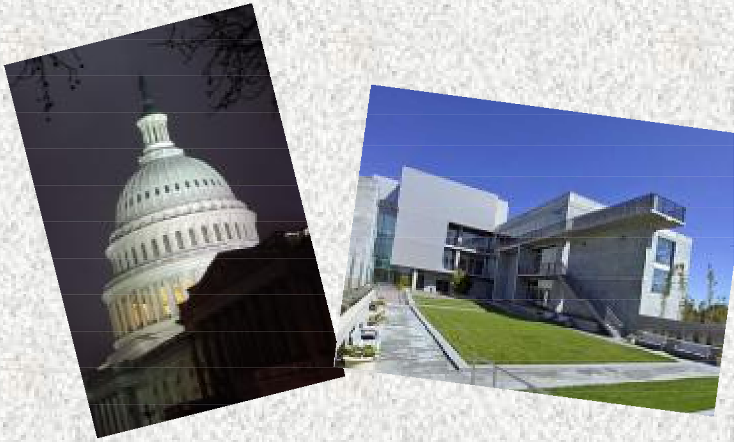


Government and Public
Health Departments

Family

Research with Residual Blood Spots from Newborn Screening

In the interests of all children there are two critical questions:

1-Can residual blood spots be stored and used for future research without jeopardizing the important public health goals of newborn screening programs?

2-Can we balance respect for parental involvement in decisions about storage and use of residual blood spots with the importance of research?

Research with Residual Blood Spots from Newborn Screening

- Anonymous
  - Samples separated from all possible identifiers
  - Cannot be re-linked to individual

- De-Identified (sample coded and separated from all identifiers)
  - Cannot be linked to specific individuals by investigator
  - Data access agreements—investigator will make no attempts to re-identify samples to subject
  - Possible to re-link to individual if finding critical to health (honest broker)

Research with Residual Blood Spots from Newborn Screening

- Health related research on Individuals
  - Requires Identifiers
  - Requires Institutional Review
  - Generally requires full informed consent

- Families may be approached for willingness to participate after Newborn Screening testing has been completed

Residual Blood Spots from Newborn Screening

Privacy Protections

Scientists and Clinicians

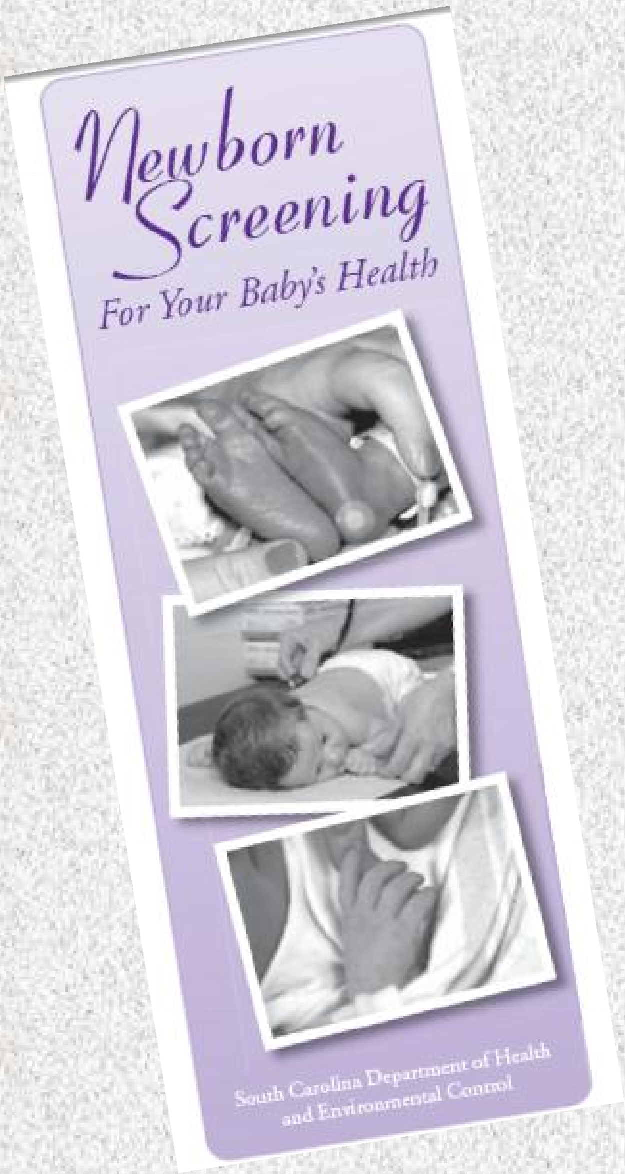
Child and
Family

Government and
Public Health
Departments

Universities and

Institutional Review Boards

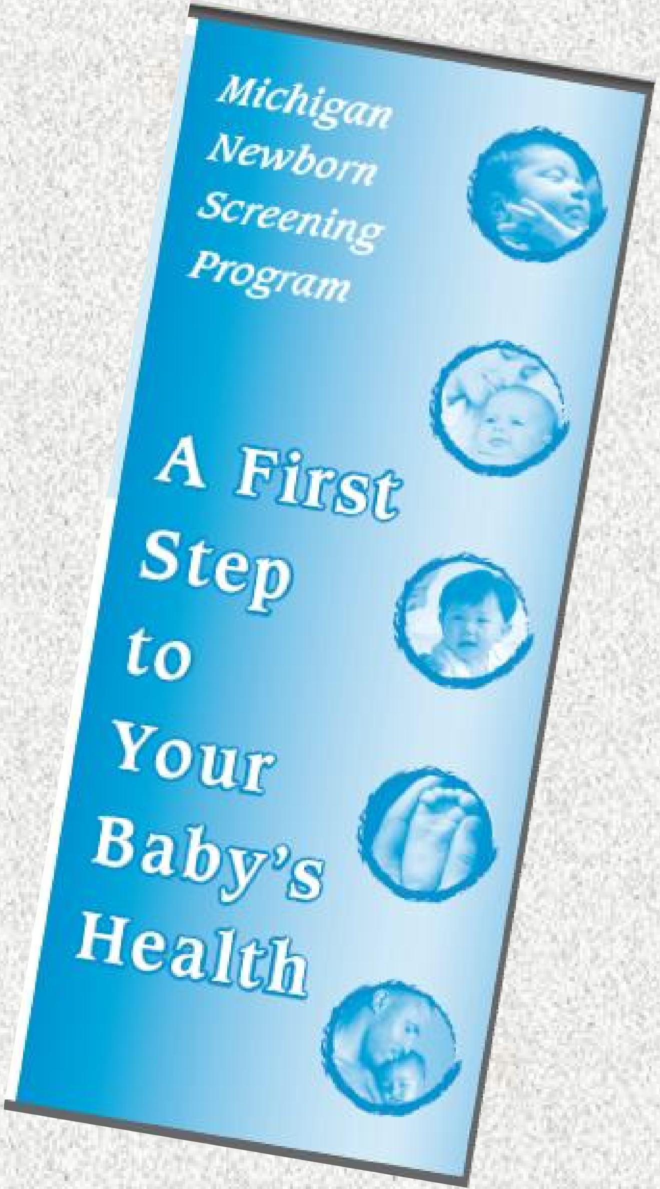
State Approaches to Research with Residual Blood Spots



Newborn Screening

For Your Baby's Health

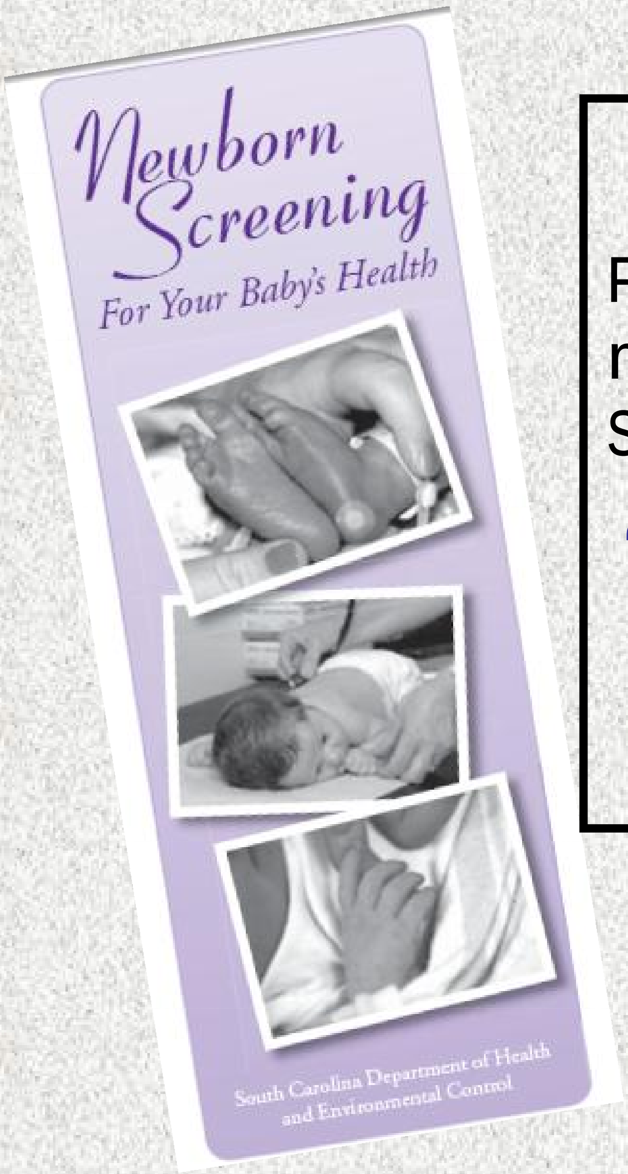
South Carolina Department of Health and Environmental Control



Michigan Newborn Screening Program

A First Step to Your Baby's Health

State Approaches to Research with Residual Blood Spots



Newborn Screening

For Your Baby's Health

South Carolina Department of Health
and Environmental Control

South Carolina

Parents receive educational materials at time of Newborn Screening:

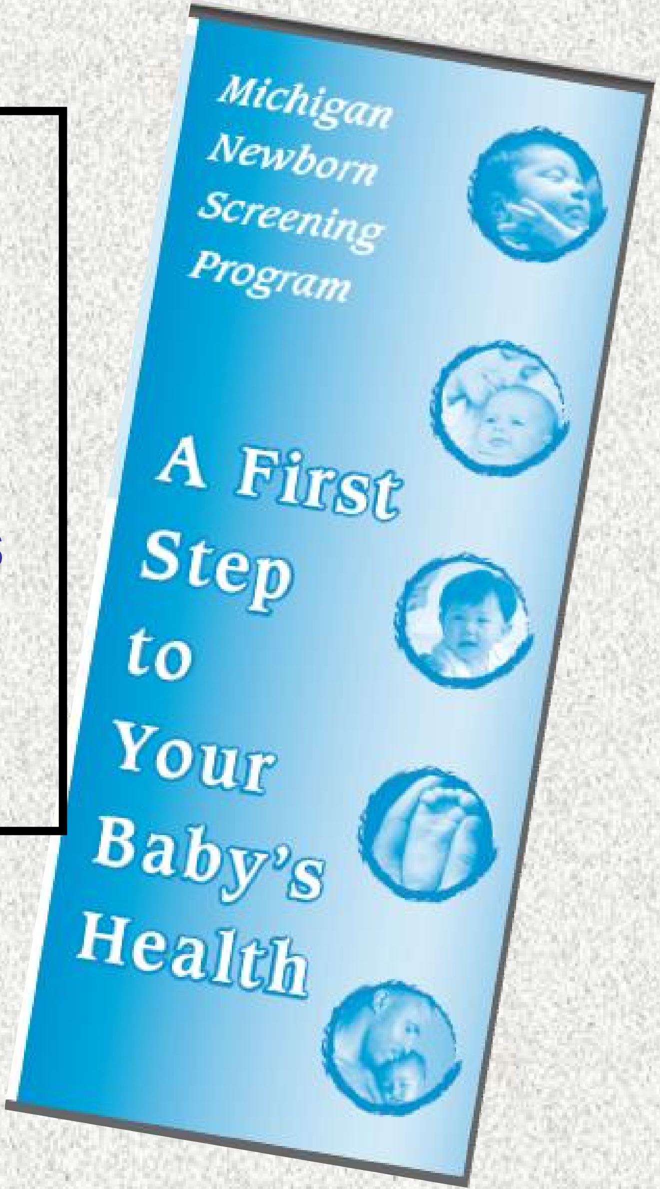
"What happens to my baby's blood sample after the lab tests? -- You decide…."

State Approaches to Research with Residual Blood Spots

Michigan

Parents receive educational pamphlet at time of Newborn Screening…

"What happens to my baby's blood specimen after testing?"



Michigan
Newborn
Screening
Program

A First
Step
to
Your
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Health

Residual Blood Spots from Newborn Screening

- Newborn Screening is a highly successful public health program
- Residual blood spots are a unique resource
- Residual blood spots can be stored while assuring confidentiality and privacy of the individual child and family
- The are many important uses of anonymized or de-identified residual blood spots that do not require prior informed consent
- Institutional Review Boards for research with human subjects should assure compliance with all federal regulations for informed consent

Residual Blood Spots from Newborn Screening

- All families receiving prenatal care, and all new parents, should be educated about newborn screening programs, including the potential to store and use residual blood spots for research;
- Health professionals who serve families receiving prenatal care, and new parents, should be knowledgeable about newborn screening programs, including the potential uses of residual blood spots for research;
- The public should be educated about newborn screening programs and reassured that research utilizing residual blood spots can enhance health and be performed with respect for and protection of privacy and confidentiality

Residual Blood Spots from Newborn Screening

- Parents should be informed and involved in decisions regarding storage and use of post-screening residual samples;
- If a state chooses to use a consent process for storage of samples, offering parents the opportunity to “opt out” of storage is preferable and less likely to jeopardize the public health program;
- All research using residual samples should be reviewed and approved by an Institutional Review Board;
- If any form of parental consent is required (including opting out) it should be addressed only after the blood sample for screening has been obtained.

Comments??? Questions???