



National Quality Improvement Projects



Heartland Genetics and Newborn Screening Collaborative Annual Conference

August 25, 2011


Amy Brower, PhD



Presentation Overview


- ◆ **Setting the Stage**
- ◆ **Federal Agency Initiatives**
 - CDC
 - APHL
 - HRSA
 - NICHD



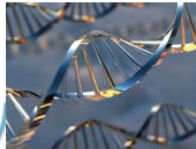


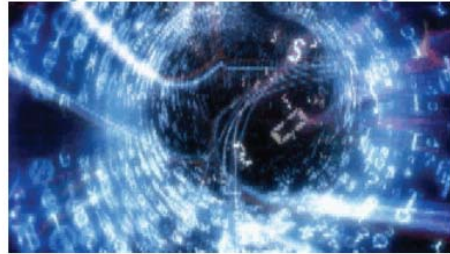
Setting the Stage – Technology Advancements


Information Technology



Genetics & Genomics





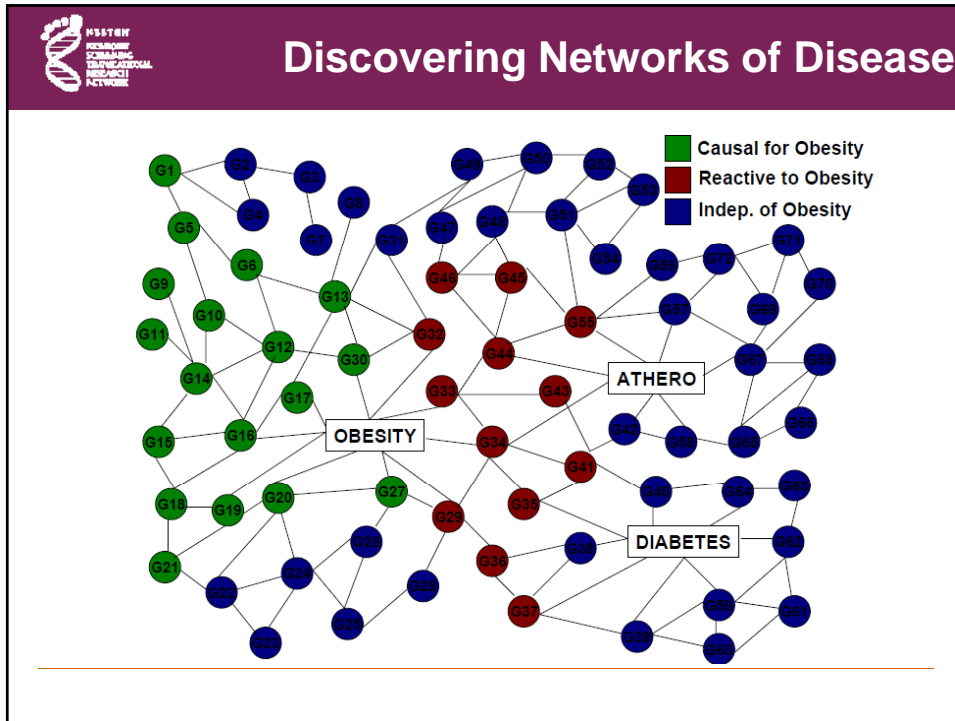


Setting the Stage – Costs Versus Outcome

The U.S. pays more per person for health care than most countries

But fails on health indicators such as life expectancy

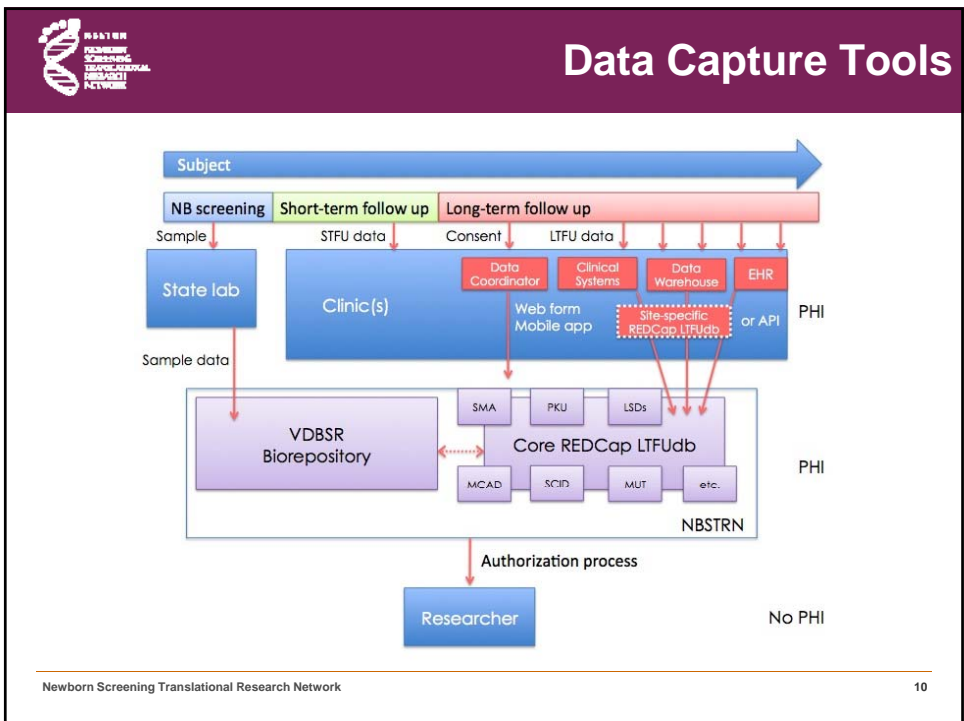
Legend: — U.S., — UK, — Other OECD countries




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- Setting the Stage – Key Areas**
- ◆ Information Standards
 - ◆ Data Capture Tools
 - ◆ User Friendly Data Analysis Tools
 - Health Care Provider
 - Public Health
 - Consumer
 - Researcher
 - ◆ Data Aggregation
 - ◆ Federal, State, Local Policy Development
- Newborn Screening Translational Research Network 8

Information Standards

- ◆ **Federal initiatives defining key aspects of information exchange**
 - **Meaningful use**
 - **Performance measurement**
- ◆ **Content – standard codes for test names, diagnosis, treatments**
- ◆ **Messaging format – standard messaging format to convey the content electronically**






User Friendly Data Analysis Tools

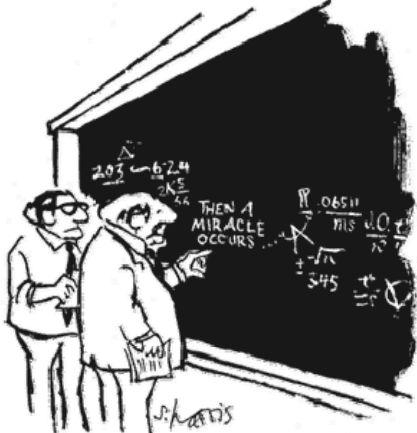
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User Friendly Data Analysis Tools

- Health Care Provider
- Public Health
- Consumer
- Researcher



"I THINK YOU SHOULD BE MORE EXPLICIT HERE IN STEP TWO."

NBSTRN
Newborn Screening Translational Research Network

Data Aggregation

HOME | WORKGROUP INFORMATION | LINKS | STEERING COMMITTEE

Welcome

ABOUT | CALENDAR | RESOURCES | PARENTS | RESEARCHERS | CONTACT

The NBSTRN is a research resource for investigators engaged in newborn screening-related research. The network includes new technological developments, the clinical history of newborn disorders, and new biopsy developments.

Resources
Education Services

The NBSTRN is funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development, National Institutes of Health

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Newborn Screening Research Tools

Virtual Repository of Dried Blood Spots
Laboratory Proficiency Program (LPP)
Data Tools Follow Up Data Collection

Building a research infrastructure for a healthier world.

REQUEST DRIED BLOOD SPOTS FOR RESEARCH
Search and request de-identified residual dried blood spots (DBS) to use in newborn screening related research projects.

Sign In
First Name
Last Name
Email
Phone
Program/Department
Register


Newborn Screening Translational Research Network

NBSTRN
Newborn Screening Translational Research Network

Policy Development

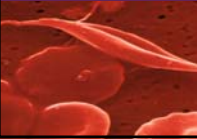

- ◆ Screening Mandates
- ◆ Program Policies
- ◆ Federal Oversight
- ◆ Ethical, Social & Legal

Newborn Screening Translational Research Network



Law - Screening Student Athletes

- ◆ Sickle Cell Trait (SCT)
- ◆ Prevalence
 - Worldwide > 300 million
 - US > 4 million
- ◆ Newborn screening – 1.31%
- ◆ NCAA approved mandatory testing for all Division I student athletes
- ◆ Estimate ruling affects 166,900



Chromosome 11

HBB Sequence in Normal Adult Hemoglobin (Hb A):

Nucleotide	CTG	ACT	CCT	GAG	GAG	AAG	TCT
Amino Acid	Leu	Thr	Pro	Glu	Glu	Lys	Ser

HBB Sequence in Mutant Adult Hemoglobin (Hb S):

Nucleotide	CTG	ACT	CCT	GAG	GAG	AAG	TCT
Amino Acid	Leu	Thr	Pro	Glu	Glu	Lys	Ser







Regulation - Oversight



 <h2 style="text-align: right;">Ethics – Ethical and Policy Issues</h2>	
Area	Question
Fairness	Who should have access to personal genetic information and how will it be used?
Privacy and confidentiality	Who owns and controls genetic information?
Psychological impact and stigmatization	How does personal genetic information affect an individual and society's perceptions of that individual?
Reproductive issues	Do health care personnel properly counsel parents about the risks and limitations of genetic testing?
Clinical issues	How will genetic tests be evaluated and regulated for accuracy, reliability, and utility?
Uncertainties	Should testing be performed when no treatment is available?
Commercialization of products	Who owns genes and other pieces of DNA?
Health and environmental issues	Are GM foods and other products safe to humans and the environment?
Philosophical implications	Do people's genes make them behave in a particular way?


 <h2 style="text-align: right;">Newborn Screening Saves Lives Act of 2008</h2>	
<p>▶ “The Secretary of the National Institutes of Health recommends that we continue expanding newborn screening - to be known as the Hunter Kelly Newborn Screening Research Program”</p>	
<p>“Hunter Kelly Newborn Screening Research Program”</p>	
<p>Newborn Screening Translational Research Network 18</p>	



National Quality Improvement


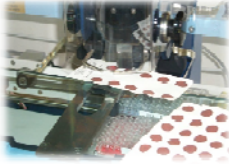

- ◆ **CDC**
- ◆ **APHL**
- ◆ **HRSA**
- ◆ **NICHD**

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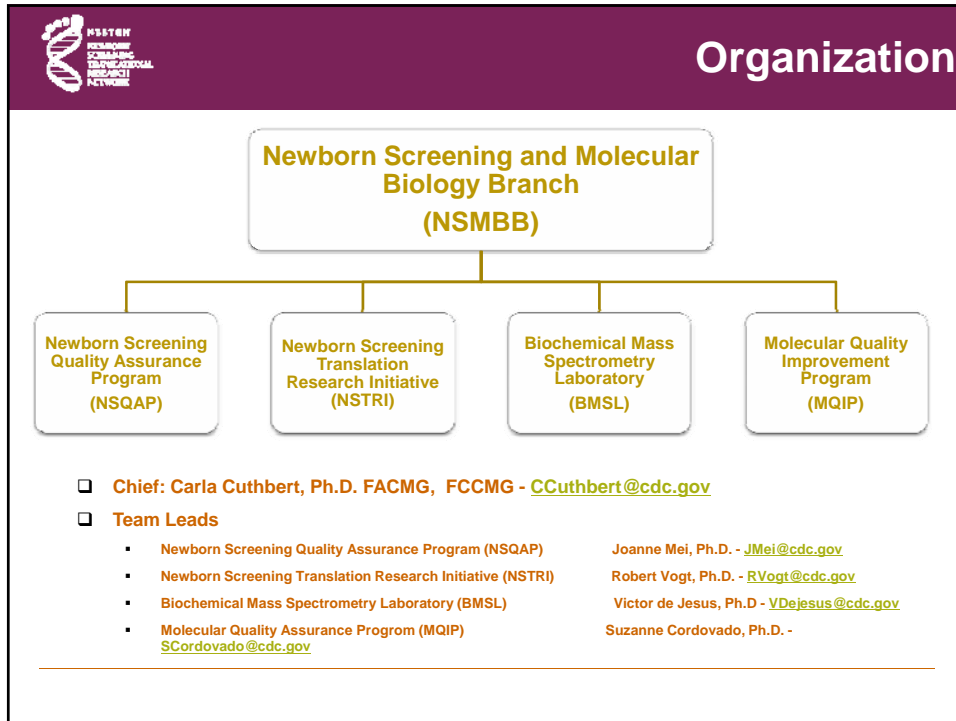

CDC

NSMBS shall provide for:

- Quality assurance for laboratories involved in screening newborns and children for heritable disorders
 - Quality assurance for NBS tests
 - Performance evaluation services
 - Technical assistance and technology transfer to NBS laboratories
 - Assistance to ensure analytic validity and utility of screening tests
- Appropriate quality control and other performance test materials to evaluate the performance of new screening tools


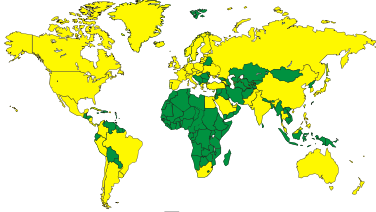





Newborn Screening Saves Lives Act of 2008



Newborn Screening Quality Assurance Program Statistics in 2010

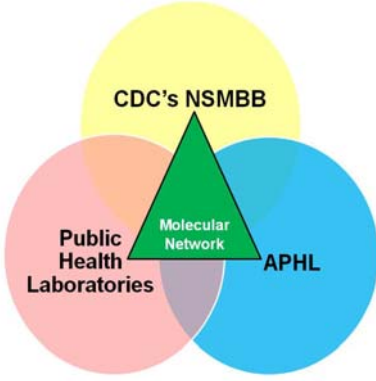
- 100% participation of US states
- 456 labs reported data
- 67 countries participated
- 391 labs participated in PT
- 717,255 DBS produced
- 337 labs participated in QC
- 28 employees
- 17 reports to participants
- 36 new enrollments
- 4 filter paper lots evaluated
- 463 labs enrolled at year end







Molecular Quality Improvement Activities

- ❑ Establishment of the NBS Molecular Network
- ❑ Implementation of NBS Molecular Assessment Program (MAP)
- ❑ QA research to identify and develop quality molecular methods for the DBS matrix
- ❑ Molecular characterization of quality assurance (QA) materials (e.g. cystic fibrosis and hemoglobinopathies)
- ❑ Translational research to address NBS community identified needs and QA protocols





Newborn Screening Molecular Biology Branch FY2011 Priorities

- ❑ Implement quality assurance programs for recent additions to the newborn screening panel (e.g. SCID)
 - NSMBB, as early advocates of SCID NBS, provided greater than \$3 million over 3 years to support SCID screening in Wisconsin and Massachusetts NBS laboratories and in the Navajo Population
 - In September 2011, new funds will support two new states implement SCID screening for 2 years
 - Ongoing SCID proficiency testing program - TREC Model Performance Evaluation Survey (MPES) - currently 11 participants
 - CDC developed *in situ* qPCR assay to detect TREC in dried blood spots



Newborn Screening Molecular Biology Branch FY2011 Priorities

- ❑ Identify gaps and address laboratory needs as NBS laboratories introduce routine molecular testing
 - Creation of the Molecular Quality Improvement Program (MQIP)
 - **NBS Molecular Network**
 - Molecular Assessment Program (MAP)
 - **Collaborative laboratory research to assure quality molecular testing**
-



APHL

- ◆ Strengthens the role of NBS PHL's
 - ◆ Facilitates implementation
 - ◆ Policy statements
 - ◆ Provides input to CDC's Newborn Screening Quality Assurance Program (NSQAP) on quality control and proficiency testing issues
 - ◆ Training and technical assistance
-






2011 Newborn Screening and Genetic Testing Symposium

- ◆ **November 7-10, 2011**
 - ◆ **Theme: “Fair Winds for the Future”**
 - ◆ **Sheraton San Diego Hotel & Marina**
 - ◆ **Call for Abstracts: June 24, 2011**
 - ◆ **Pre conference on QA/QC and Short and Long term Follow Up**
 - ◆ **Harry Hannon and George Cunningham Awards**
 - ◆ **For more information, visit www.aphl.org**
-




HRSA

- ◆ **Case Definition**
 - Surveillance: making sure we are counting cases the same
 - Determine the final disposition of a patient with regard to whether they were a detected case or not.
 - This is not a clinical algorithm to help in determining who should be treated or not.
 - The purpose is to help standardize the determination of outcomes of newborn screening conditions.
 - ◆ **NBS Quality Indicators**
 - Common measures that can be defined, compared and used for quality improvement
 - Need agreed upon definitions, where difference in practice, will try to stratify in order to compare similar programs
 - Measures that should be measured and compared at a national level that reflect the functioning of the state NBS programs
-





What are we comparing?




We need to be sure that the diagnoses are comparable in reliability before we can compare numbers for surveillance and outcomes for long term follow-up


We want to compare outcomes, but are we comparing outcomes of the same certainty of diagnosis?







NICHD - Goals of the Hunter Kelly Newborn Screening Research Program

- 


Identify, develop and test the most promising new screening technologies
- 

Increase the specificity of newborn screening and expand the number of conditions for which screening tests are available
- 

Develop experimental treatments and disease management strategies for additional newborn conditions, and other genetic, metabolic, hormonal and or functional conditions that can be detected through newborn screening for which treatment is not yet available.




NICHD - Novel Technologies




Development of efficient and effective screening strategies for Lysosomal Storage Disorders, Friedreich ataxia, Wilson disease, and X-linked Adrenoleukodystrophy

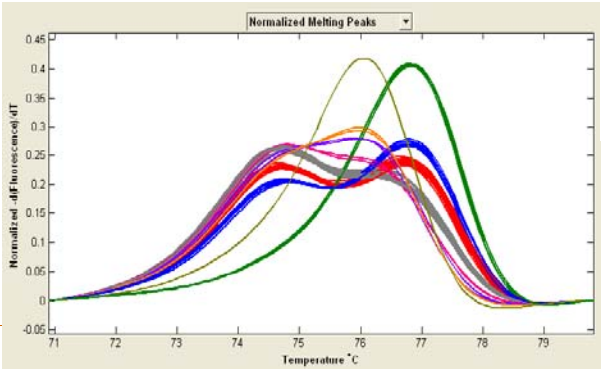
Method	Multiplex	Platform	Complexity	Performance
Enzyme Assay	No	Fluorometry	Low	?
Multiplex Enzyme	Yes	MS/MS	High	?
Multiplex Immune-Quantification Assay	Yes	Luminex	Low	??
Digital Microfluidics	Yes	“Lab-on-a-chip”	Low	??




NICHD - Current Technologies




Increase the specificity of newborn screening and expand the number of conditions for which screening is available





NICHD – Novel Treatments



- **Innovative Therapies and Tools for Screenable Disorders PAR**
- **Basic understanding and development of therapeutic interventions for currently screened conditions and “high priority” genetic conditions for which screening could be possible in the near future.**



NICHD - Natural History Studies

- ◆ **Pilot Newborn Screening Project for Identification and Prospective Follow-up of Infants with Spinal Muscular Atrophy**
 - **Kathy Swoboda – University of Utah**
- ◆ **Inborn Errors of Metabolism Collaborative: Defining the Natural History of Inborn Errors of Metabolism**
 - **Cynthia Cameron – Michigan Public Health Institute**

NICHD - Health Outcomes

Intake

- Demographics
- SES
- Family History
- Prenatal History
- Neonatal History
- Birth Measurements
- Newborn Screening
- Hearing Screening
- Diagnostic Testing

Childhood

- Monitoring Labs
- Diet
- Therapies
- Emergency Management
- Developmental Screening
- Imaging Studies
- Intercurrent Complications

Adulthood

- Monitoring Labs
- Diet
- Therapies
- Emergency Management
- Imaging Studies
- Intercurrent Complications

- Create a uniform dataset for all conditions "80%"
- Create condition-specific datasets

NBSTRN/NCC-RC Joint Workgroup

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NBSTRN

Newborn Screening Translational Research Network

RESEARCHERS
PARENTS
WORKGROUPS

[Home](#) [About](#) [Resources](#) [Research Tools](#) [Services](#) [Education](#) [Supported Projects](#) [Calendar](#)

The **Newborn Screening Translational Research Network (NBSTRN)** seeks to improve the health outcomes of newborns with genetic or congenital disorders through an infrastructure that provides the research community access to robust newborn screening resources.

[More about NBSTRN](#)

Building a research infrastructure for a healthier world.

For Researchers >

The NBSTRN was developed as a resource for researchers [more](#)

For Parents >

This section is organized into collection of useful resources [more](#)

For NBSTRN Workgroups >

The NBSTRN is comprised of an advisory Standing [more](#)

Newborn Screening Research Tools

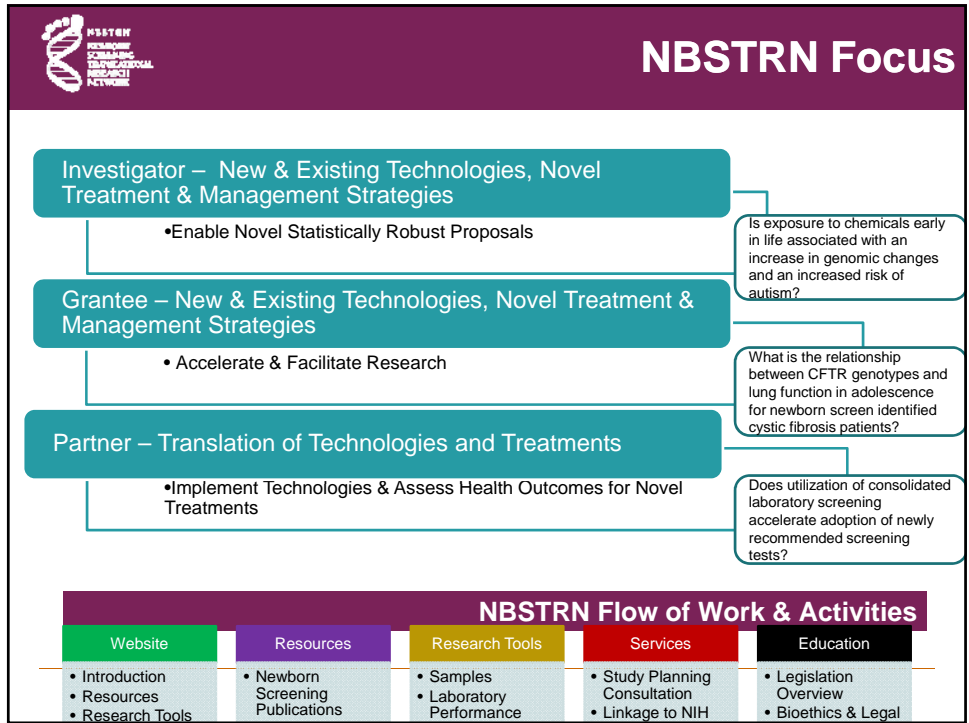
Virtual Repository of Dried Blood Spots

Laboratory Performance Database (R4S)

Long Term Follow-Up Data Collection

What's New

- NBSTRN Newsletter
- SACHDNC Presents New Recommendations for Storing DBS
- NBSTRN Next Generation Meeting June 16-17, 2011
- Save the Dates for Upcoming NBSTRN Meetings
- Funding Opportunity Announcements and Notices



NBSTRN
NEWBORN SCREENING TRANSLATIONAL RESEARCH NETWORK

Biorepository

NBSTRN Virtual Repository of Dried Blood Spots

Home About Research Support Search nbstrn.org

My Selections My Requests Help Sign In

REQUEST DRIED BLOOD SPOTS FOR RESEARCH
Search and request de-identified residual dried blood spots (DBS) to use in newborn screening related research projects.

Register Now Learn More Privacy Policy

Sign In
Email Address
Password
Sign In
Forgot password? Register

Welcome About NBSTRN How To

The NBSTRN Virtual Repository for Dried Blood Spots is a centralized, web-based tool to access specimens for newborn screening related research and program development. This virtual repository presents information from participating states and provides a centralized, de-identified view of dried blood spots, allowing researchers to browse and query for specimens. For public health departments, the virtual repository provides the ability to review and manage specimen requests, generate orders, track shipments, and view researcher feedback regarding the specimens they have received.

Learn More Register Now

Diagnosed Conditions in Repository

- No Conditions Diagnosed
- 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
- 3-Hydroxy-3-methylglutaric aciduria (HMG)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
- Argininosuccinic aciduria (ASA)
- beta-Ketothiolase deficiency (BKT)
- Biotinidase Deficiency (BIO)

Participating States

Select a U.S. State

Why is a state not listed?

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NBSTRN's Role in Pilot Projects

- Identify collaborations that would benefit from NBSTRN infrastructure, resources and expertise
- Focus on needs of researchers and overarching goal of effort
- Current efforts
 - Natural history of newborn screened disorders
 - Novel technologies
 - Newborn screening pilots
- Future efforts
 - Consider potential needs




Current Pilots

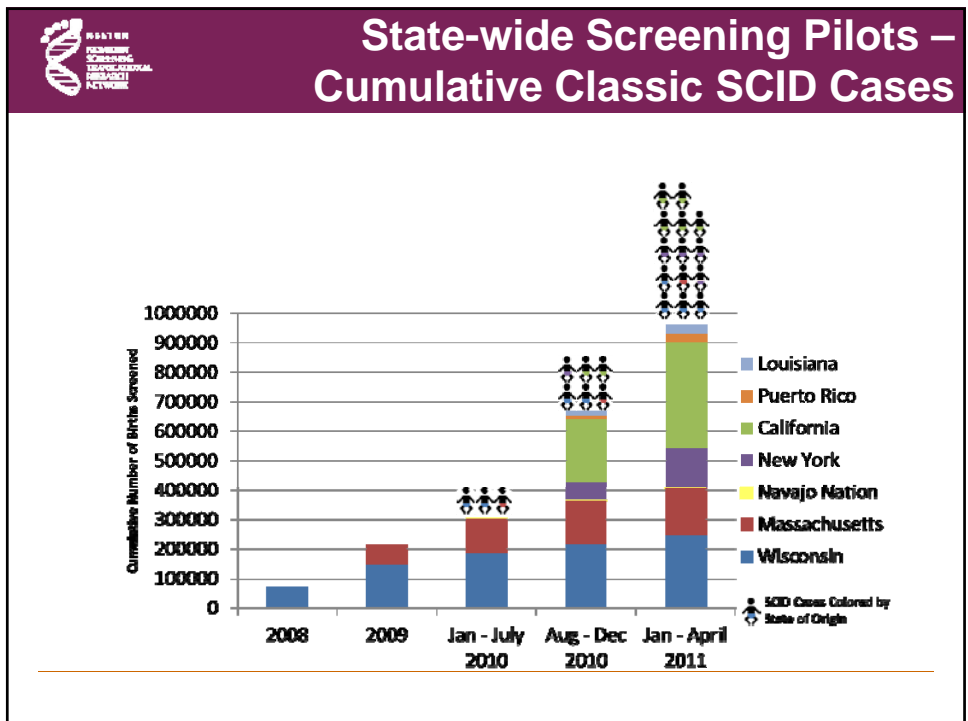
- ◆ LSD
 - Compare screening technologies for lysosomal storage disorders (LSDs)
- ◆ SCID
 - Expand pilot of newborn screening for SCID and related T cell lymphopenias
 - Establish novel technologies for screening
 - Draft and disseminate laboratory protocols for screening
 - Support expansion of screening to non-pilot states

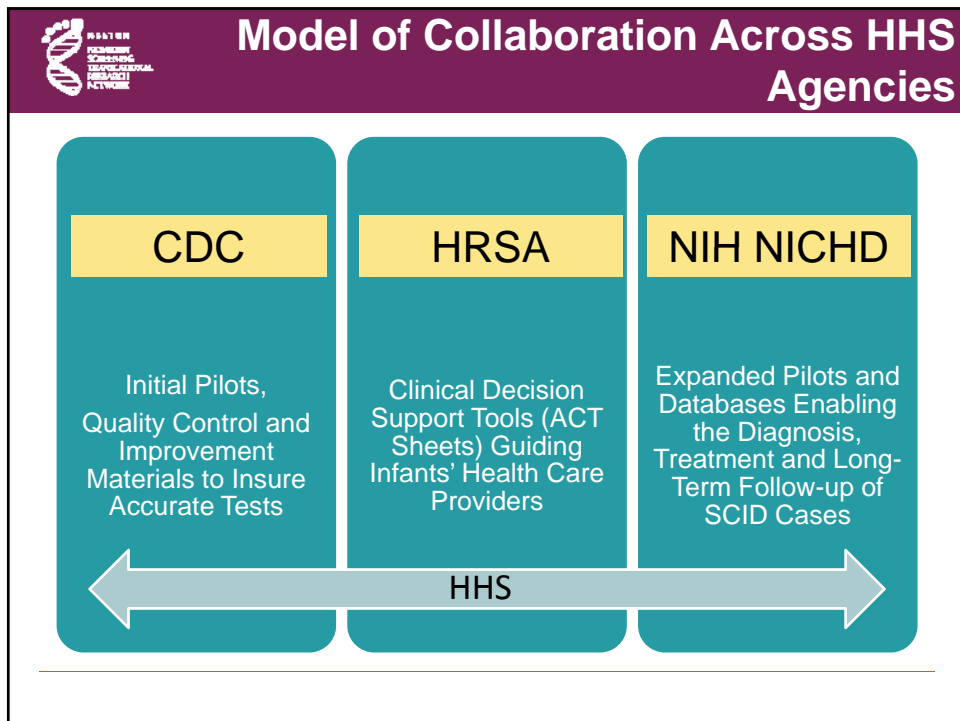
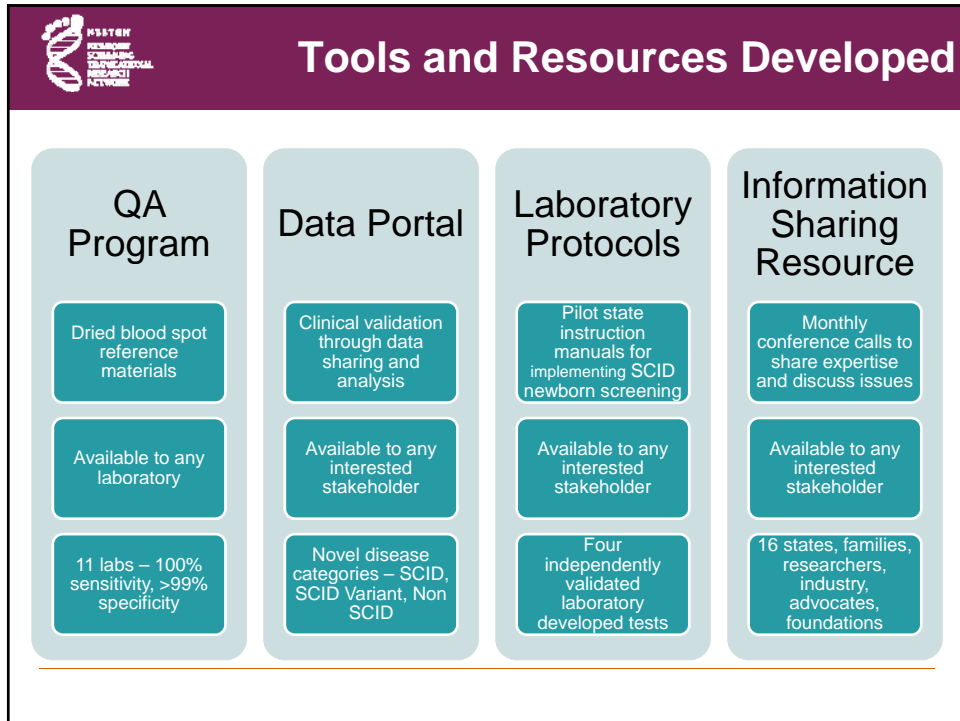



SCID Data Portal

- ◆ Goal was to collect, aggregate and analyze de-identified screening data generated during the pilot
- ◆ Enables real-time laboratory performance quality improvement
- ◆ Stores laboratory protocols
- ◆ Facilitates tracking of emerging findings
- ◆ Provides disease definitions
- ◆ Available to any newborn screening program and or researcher













Federal Institutions

- ◆ **HHS Personalized Health Care (PHC) Initiative**
 - Focus on infrastructure and creation of “network of networks”
 - <http://www.hhs.gov/myhealthcare>
- ◆ **Office of the National Coordinator on Health Information Technology**
 - Focus on development and adoption of health care information technology
 - <http://www.hhs.gov/healthit>
- ◆ **Agency for Healthcare Research**
 - Evidence-based Practice Centers (EPCs)
 - Centers for Education and Research on Therapeutics (CERTs)



Acknowledgements

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SCID Trial -HHSN267200603430**

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