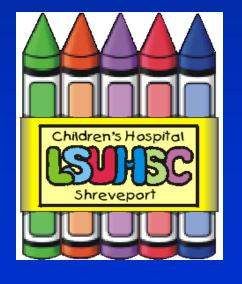
Updating requirements for Nomination Packet: Pilot Study



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Chairperson, SACHDNC

May 30, 2014

DACHDNC Form for Nomination of a Condition: Pilot Study

SECTION II, PART C

POPULATION- BASED PILOT STUDY	STATEMENT
Location of	
Prospective Pilot	
Number of	
Newborns	
Screened	
Number of	Positive by primary test vs. 2 nd tier test if applicable.
Screen Positive	
Results	
False Positive	False positive by primary test vs. 2 nd tier test if applicable.
Rate; False	
Negative Rate (if	
known)	
Number of	How is diagnosis confirmed [clinical, biochemical, molecular]?
Infants	
Confirmed with	
Diagnosis	

Pilot study data utilized in past reviews

- Anonymous testing of residual dried blood spots
- States which pilot a test
 - Legislative requirement
 - Grants/Contracts
 - NBSTRN, Modell Foundation
- Other countries

Pilot study = Pilot program

- Evaluate how testing can be incorporated into the state screening program
 - Laboratory
 - Short-term follow-up
 - Long-term follow-up

Data Elements needed from Pilot Program

1. Clinical Evidence

- Test (analytic, cutoffs, etc.)
- Treatment

2. Laboratory

- Equipment
- Staff and/or expertise to run test
- High throughput

Data Elements needed from Pilot Program

- 3. Follow-up Activities
 - Clinical experts in place for handoff
 - Treatment available, feasible

Recommendation for DACHDNC consideration

- Standardize pilot program data required for nomination of condition
- Work with partners to create opportunities for states to participate in pilot programs to generate needed evidence to support nomination







Planning Your Research

Research Tools

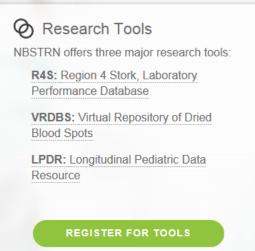
Research Projects

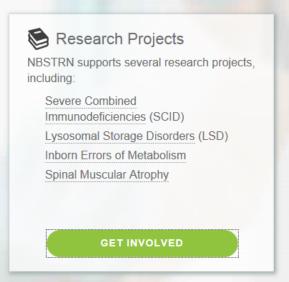
About NBSTRN

Q Planning Your Research

NBSTRN has developed a centralized point of access to information valuable to conducting and facilitating research related to newborn screening.

VIEW RESOURCES







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Planning Your Research

Research Tools

Research Projects

About NBSTRN

Research Projects

Severe Combined Immunodeficiencies

Lysosomal Storage Disorders

Inborn Errors of Metabolism

Spinal Muscular Atrophy



Lysosomal Storage Disorders Collaborative

Principal Investigator: Melissa Wasserstein, MD

Institution: Mount Sinai School of Medicine
Project Start Date: September 4, 2012

Project End Date: May 31, 2017

Project Uses: R4S, VRDBS, LDPR







"LSDs: A Pilot NBS and Examination of the Associated Ethical Legal and Social Issues"

Newborn screening for several LSDs (Fabry, Gaucher, Niemann Pick Types A and B, and Pompe) has been proposed in several states. Each of these diseases has a broad phenotypic spectrum ranging from severe infantile-onset disease to adult-onset, milder phenotypes. Thus, newborn screening for these disorders presents a unique set of complex issues that require investigation prior to the initiation of mass newborn screening. These issues include determining the clinical and diagnostic accuracy of the screening assay, investigating how to correctly predict phenotype in asymptomatic newborns, and developing algorithms to assist with clinical decision-making about if and when to initiate therapy. In addition, there are novel ethical, legal, and social issues associated with testing infants for potentially later-onset disorders. This proposal will explore these issues by implementing a pilot newborn screen in approximately 80,000 infants born in high birth rate, ethnically diverse New York City hospitals in order to evaluate the analytic and clinical validity of the screening test and to



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Planning Your Research

Research Tools

Research Projects

About NBSTRN



NICHD Request for Proposal: Newborn Screening Pompe Pilot Study

On May 6th, 2014 the Eunice Kennedy Shriver National Institutes of Child Health and Human Development (NICHD) released a request for proposal for a Newborn Screening Pompe Pilot Study.

The purpose of this solicitation is to support the development, implementation and proof of concept in newborn screening for Pompe disease. See the full request for proposal for greater detail.

NICHD Request for Proposal ₪

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PLANNING YOUR RESEARCH RESEARCH TOOLS RESEARCH PROJECTS ABOUT NBSTRN

Proposal and Discussion

DACHDNC: Process for evaluation of a condition for inclusion on RUSP

- Nomination package submitted
- Nomination and Prioritization Workgroup
 - Recommendation to DACHDNC
 - DACHDNC votes whether to move to CRW
- Condition Review Workgroup
 - Completes a systematic review and PHI assessment, provides updates, and presents a final report
- DACHDNC votes to recommend, or not recommend