

# Updating requirements for Nomination Packet: Pilot Study

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# 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 Screen Positive Results                   | Positive by primary test vs. 2 <sup>nd</sup> tier test if applicable.       |
| False Positive Rate; False Negative Rate (if known) | False positive by primary test vs. 2 <sup>nd</sup> tier test if applicable. |
| Number of Infants Confirmed with Diagnosis          | How is diagnosis confirmed [clinical, biochemical, molecular]?              |

# 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**



**NBSTRN**

Newborn Screening  
Translational Research  
Network

 For Parents

[Planning Your Research](#)

[Research Tools](#)

[Research Projects](#)

[About NBSTRN](#)



## 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](#)



## Research Tools

NBSTRN offers three major research tools:

[R4S: Region 4 Stork, Laboratory Performance Database](#)

[VRDBS: Virtual Repository of Dried Blood Spots](#)

[LPDR: Longitudinal Pediatric Data Resource](#)

[REGISTER FOR TOOLS](#)



## Research Projects

NBSTRN supports several research projects, including:

[Severe Combined Immunodeficiencies \(SCID\)](#)

[Lysosomal Storage Disorders \(LSD\)](#)

[Inborn Errors of Metabolism](#)

[Spinal Muscular Atrophy](#)

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CONNECTING RESEARCHERS  RESOURCES



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



## 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.

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[NICHD Request for Proposal](#)

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# 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**