



SMA

Current Research Funding at NICHD

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Sample of SMA Projects Recently Funded by NICHD



- **Optimization of drug-like compounds for treatment of spinal muscular atrophy**
 - Elliot J. Androphy – University of Massachusetts Medical School
- **Spinal muscular atrophy: SMN's role in MRNA localization and local translation**
 - Gary J. Bassell – Emory University
- **Creation of hypomorphic panel of Smn mice that mimic human SMN2 splicing**
 - Christine Didonato – Children's Memorial Hospital (Chicago)
- **Stimulating SMN2 exon 7 inclusion with short RNAs**
 - Christian Lorson – University of Missouri-Columbia
- **Spinal muscular atrophy: A novel role of SMN in axonal ribonucleoprotein complexes**
 - Wilfried Rossoll – Emory University
- **Therapeutic Opportunities in Spinal Muscular Atrophy**
 - Kathryn Swoboda – University of Utah
- **Newborn Screening for Spinal Muscular Atrophy**
 - Thomas W. Prior- Ohio State University

PUBLIC LAW 110-204-NEWBORN SCREENING SAVES LIVES ACT OF 2008



- ***This Bill amends the Public Health Service Act to establish grant programs to provide for education and outreach on newborn screening and coordinated follow-up care once newborn screening has been conducted...***

- This new Bill requires the Secretary of the department of Health and Human Services to:
 - Ensure the quality of laboratories involved in NBS activities and
 - To develop a national contingency plan for newborn screening.
 - The Bill gives NIH the authority to carry out research in newborn screening, including identifying new screening technologies and researching disease management strategies for the conditions that can be detected through the Hunter Kelly Newborn Screening Research Program. This NBS program is within NICHD

Newborn Screening Translational Research Coordinating Center



In September 2008, the NICHD awarded a 5 year contract to the American College of Medical Genetics (ACMG) to create a Coordinating Center that will establish a research infrastructure for Newborn Screening studies.

The NBSTRN Coordinating Center (NBSTRN-CC) will facilitate research to develop new screening methods and support the conduct of clinical trials for new therapeutic interventions.

Newborn Screening Translational Research Network Coordinating Center - Objectives



- Establish an organized network of State newborn screening programs and clinical centers
- Develop, implement and refine a national research informatics system for investigators and policy makers
- Establish and administer an efficient and reliable repository of residual dried blood spots
- Provide expertise and support to researchers related to regulatory requirements associated with informed consent, IRBs and state and local research policy associated with NBS.

Newborn Screening Translational Research Network Coordinating Center - Objectives



- Facilitate research on the development of new methods and technologies
- Facilitate research on screened and treated patients to define effectiveness of treatments and long-term outcomes
- Provide statistical leadership and clinical trial design expertise for the individualized needs of researchers through the NBSTRN Coordinating Center
- Facilitate the timely dissemination of research findings

Program Announcements



■ Innovative Therapies and Clinical Studies for Screenable Disorders

(PAR 06-342 – R21; PAR 06-341 – R03; PAR 07-184 – R01) (Standard Due Dates)

- National Institute of Child Health and Human Development (NICHD)
- National Institute on Deafness and Other Communication Disorders (NIDCD)
- National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK)

- The purpose of this ongoing Funding Opportunity Announcement is to stimulate translational research on potential therapeutic interventions for currently screened conditions and “high priority” genetic conditions for which screening could be possible in the near future.

- Targeted disorders include Heritable Gamma-Hydroxybutyric Aciduria, Hyperammonemia, Gaucher Disease, **Spinal Muscular Atrophy**, Galactosemia, Globoid-Cell Leukodystrophy, Phenylketonuria.