Newborn Screening at NICHD

TIINA K. URV, PH.D.
An Act
To amend 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, to reauthorize programs under part A of title XI of such Act, and for other purposes.

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

SECTION 1. SHORT TITLE.
This Act may be cited as the “Newborn Screening Saves Lives Act of 2007”.

SEC. 2. IMPROVED NEWBORN AND CHILD SCREENING FOR HERITABLE DISORDER.
Section 1109 of the Public Health Service Act (42 U.S.C. 300b-8) is amended—
(1) by striking subsections (a), (b), and (c) and inserting the following:
“(a) AUTHORIZATION OF GRANT PROGRAM.—From amounts appropriated under subsection (f), the Secretary, acting through the Administrator of the Health Resources and Services Administration (referred to in this section as the ‘Administrator’) and in consultation with the Advisory Committee on Heritable Disorders in Newborns and Children (referred to in this section as the ‘Advisory Committee’), shall award grants to eligible entities to enable such entities—
“(1) to enhance, improve or expand the ability of State and local public health agencies to provide screening, counseling, or health care services to newborns and children having or at risk for heritable disorders;
“(2) to assist in providing health care professionals and newborn screening laboratory personnel with education in newborn screening and training in relevant and new technologies in newborn screening and congenital, genetic, and metabolic disorders;
“(3) to develop and deliver educational programs (at appropriate literacy levels) about newborn screening counseling, testing, follow-up, treatment, and specialty services to parents, families, and patient advocacy and support groups; and
“(4) to establish, maintain, and operate a system to assess and coordinate treatment relating to congenital, genetic, and metabolic disorders.
“(b) ELIGIBLE ENTITY.—In this section, the term ‘eligible entity’ means—
“(1) a State or a political subdivision of a State;
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.
Newborn Screening Condition Nomination Criteria

**Condition**
- Well-defined
- Natural history
- Clinically important phenotypic or genotypic variations
- Prevalence/Incidence

**Screening Test**
- Availability
- Method and accuracy
- Adequate distinction between early and late onset
- Pilot testing and experience (population or other)
- Is there a diagnostic test

**Treatment**
- Is there a treatment
- Treatment strategies
- Are treatments: Standardized, available, FDA approved
- Potential harms and risks of treatment

**Ethical, Legal and Societal Implications**
- Potential benefits of screening
- Potential harms and risks of screening
- Treatment early vs. late
- Cost of Screening

**Feasibility of Implementation**
- Cost of screening and Follow-up
- Impact on Public Health Systems
- Other indirect of induced costs

**References**
Communication, Collaboration and Coordination
Collaboration Across HHS Agencies

- **CDC**
  - Quality Control and Improvement
  - Quality Assurance Materials

- **HRSA**
  - Clinical Decision Support for Health Care Providers
  - Patient Care Support

- **NIH**
  - Identify, develop and test new screening technologies
  - Increase the specificity of NBS and expand the number of conditions for NBS
  - Develop experimental treatments and disease management

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**H.H.S.**
Approach
In the beginning...
...and in the more recent past!
What if it were possible to provide universal sequencing of infants???

While for some this is an exciting possibility...
...for others it is a daunting challenge!
The Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), the National Human Genome Research Institute (NHGRI), and the NIH Office of Rare Diseases Research (ORDR) sponsored a workshop, *Newborn Screening in the Genomic Era: Setting a Research Agenda*.

The purpose of the meeting was to identify elements of a trans-NIH research agenda that would lead to the application of new genomics concepts and technologies to newborn screening and child health.

The meeting was attended by experts from academia, industry, and federal agencies in the fields of newborn screening (NBS) and genomics.

Chaired by Drs. David Valle (Johns Hopkins, University) and Piero Rinaldo (Mayo Clinic).

[https://www.genome.gov/Pages/PolicyEthics/StaffArticles/Newborn_Screening_Meeting_Summary.pdf](https://www.genome.gov/Pages/PolicyEthics/StaffArticles/Newborn_Screening_Meeting_Summary.pdf)
To explore, in a limited but deliberate manner, opportunities to use genomic information for broadening our understanding of diseases identified in the newborn period.
Required 3 Components

- Clinical Research
- Genomic Sequencing
- Ethical, Legal, and Social Implications
Part I Overview Information

Department of Health and Human Services

Participating Organizations
National Institutes of Health (NIH). (http://www.nih.gov)

Components of Participating Organizations
Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), (http://www.nichd.nih.gov)
National Institute on Deafness and Other Communication Disorders (NIDCD) (http://www.nidcd.nih.gov)
National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) (http://www2.niddk.nih.gov)
National Institute of Neurological Disorders and Stroke (http://www.ninds.gov)

Title: Innovative Therapies and Tools for Screenable Disorders in Newborns (R01)

Announcement Type
This is a reissue of PAR-07-164

Update: The following updates relating to this announcement have been issued:

- **July 2, 2014** - This PAR has been reissued as PAR-14-270.
- **September 28, 2010** (NOT-OD-11-007) - NIH to Require Use of Updated Electronic Application Forms in 2011. Adobe B1 forms are required for due dates on or after May 8, 2011.
- **August 16, 2010** - IMPORTANT NOTE! NIH has eliminated the error correction window for due dates of January 25, 2011 and beyond. As of January 25, all corrections must be complete by the due date for an application to be considered on-time. See NOT-OD-10-123.

Program Announcement (PA) Number: PAR-10-230

NOTICE: Applications submitted in response to this Funding Opportunity Announcement (FOA) for Federal assistance must be submitted electronically through Grants.gov (http://www.grants.gov) using the SF424 Research and Related (R&R) forms and the SF424 (R&R) Application Guide.

APPLICATIONS MAY NOT BE SUBMITTED IN PAPER FORMAT.

This FOA must be read in conjunction with the application guidelines included with this announcement in Grants.gov/Apply for Grants (hereafter called Grants.gov/Apply).
# Department of Health and Human Services

## Initiatives

### Part 1. Overview Information

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<th>National Institutes of Health (NIH)</th>
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<tr>
<td>Components of Participating Organizations</td>
<td>National Human Genome Research Institute (NHGRI)</td>
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<tr>
<td>Funding Opportunity Title</td>
<td>Methods Development for Obtaining Comprehensive Genomic Information from Human Specimens that are Easy to Collect and Store (R43/R44)</td>
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<tr>
<td>Activity Code</td>
<td>R43/R44 Small Business Innovation Research (SBIR) Grant - Phase I, Phase II, and Fast Track</td>
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<td>Announcement Type</td>
<td>New</td>
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<td>Related Notices</td>
<td>- June 4, 2014: Notice NOT-14-074 supersedes instructions in Section III.3 regarding applications that are essentially the same.</td>
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<td>- May 12, 2014 (NOT-OD-14-039) - Updated Grant Application Forms (FORMS-C) Now Available for SBIR/STTR Funding Opportunities. Forms-C applications are required for due dates on or after August 5, 2014.</td>
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<td>- August 21, 2013: Removed reference to ASSIST in section IV.3, since ASSIST is currently only available for multi-project applications.</td>
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<td>Funding Opportunity Announcement (FOA) Number</td>
<td>PAR-13-203</td>
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<td>Companion Funding Opportunity</td>
<td>RFA-HD-13-010, U19 Research Program – Cooperative Agreements</td>
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<td>Number of Applications</td>
<td>See Section III, 3. Additional Information on Eligibility.</td>
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<td>Catalog of Federal Domestic Funding (CFDA) Numbers</td>
<td>93.172, 93.865</td>
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Awarded Soon (Probably Friday)

- Pompe Pilot
  - That is all I can say
A Resource to provide a research infrastructure to support investigators with projects related to newborn screening

Currently managed by the American College of Medical Genetics
Feel free to contact me anytime

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