NIH Newborn Screening Research Activities

Gilian H. Engelson, MPH
National Institute of Child Health and Human Development, NIH
### NIH Institutes/Offices/Centers Relevant to NBS

<table>
<thead>
<tr>
<th>Institute/Center</th>
<th>Responsibilities</th>
</tr>
</thead>
<tbody>
<tr>
<td>NICHD</td>
<td>Lead institute on newborn screening</td>
</tr>
<tr>
<td>NINDS</td>
<td>Developmental neurological disorders</td>
</tr>
<tr>
<td>NIDDK</td>
<td>Metabolic conditions</td>
</tr>
<tr>
<td>NIBIB</td>
<td>Point of care technologies</td>
</tr>
<tr>
<td>NHGRI</td>
<td>ELSI, genomics, linkages to conditions</td>
</tr>
<tr>
<td>NHLBI</td>
<td>Hemoglobinopathies and cardiomyopathies</td>
</tr>
<tr>
<td>NIAMS</td>
<td>Neuromuscular conditions</td>
</tr>
<tr>
<td>NIDCD</td>
<td>Hearing impairment</td>
</tr>
<tr>
<td>NIEHHS</td>
<td>Environmental factors associated with congenital defects</td>
</tr>
<tr>
<td>NLM</td>
<td>Newborn screening resource pages including the Genetics Home Reference</td>
</tr>
<tr>
<td>FIC</td>
<td>Genetics and informatics training, international research efforts</td>
</tr>
<tr>
<td>ORD</td>
<td>Rare diseases</td>
</tr>
</tbody>
</table>
NICHD Newborn Screening Research Priorities

- Development of Translational Research Infrastructure Programs
- Development of Screening Technology
- Improved Therapies
- Studies of Natural History and Long Term Outcomes of Treatment
- Behavioral and Social Sciences Research
- Creation of Appropriate Public Policies
PA: Innovative Therapies and Clinical Studies for Screenable Disorders

- Program Announcement to support the development of therapeutic interventions (new, improved or supplemental) for screenable conditions
  - Funding Mechanisms: R01, R21, R03
- Applications deadlines 3 times each year until 2009
- Co-sponsored by NICHD, NIDDK, NIDCD
- [www.grants.gov](http://www.grants.gov)
Active Grants PA: Innovative Therapies and Clinical Studies for Screenable Disorders

- Awarded grants starting Spring 2007
- Current successful grants researching therapeutic interventions for:
  - galactosemia
  - spinal muscular atrophy
  - hearing loss due to cytomegalovirus
  - globoid-cell leukodystrophy
Contracts on Novel Technologies in Newborn Screening

Support the development of comprehensive multiplex technologies

Sept 2006: Awarded 2 three-year contracts to:

- University of Washington
  
  Principal Investigator: C. Ronald Scott, MD
  
  Expansion of MS/MS to measure enzyme activity for lysosomal storage disorders

- New York State Department of Health/Wadsworth Center
  
  Principal Investigator: Kenneth Pass, Ph.D.
  
  Utilization of Luminex florescent bead array technology
Potential NIH Initiative: Newborn Screening Translational Research Network

- Validate screening technologies and therapeutic interventions for rare diseases
- Increase access to dried bloodspots and other samples for research use
- Perform longitudinal health outcomes research on individuals identified through newborn screening
- Establish an informatics system to link researchers with potential human subjects for clinical trials
- Develop informed consent and recommended research policies
Potential NIH Initiative: Newborn Screening Translational Research Network

Development of a network that improves communication and collaboration among:

- Databases
- Biospecimen repositories
- Clinical Researchers
- Public Health Programs
InfoRx

- InfoRx pads to assist HCPs refer patients to up-to-date, authoritative, consumer-friendly website - the Genetic Home Reference - for info on genetic conditions detected through NBS
- Developed by NLM, supported by NICHD
- Direct outreach by AAP, ACMG, ACOG, AAFP
- Order free pads at: http://www.informationrx.org
How can ACHDGDNC help?

- Guidance/Advice on:
  - Newborn screening research needs
    - Development of a translational research network
      - Infrastructure
      - Components
      - Linkage to other public health programs
      - Policy and legislative issues
    - ELSI Issues: IRB, human subject and social issues
  - Other