NIH Newborn Screening Research Activities

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National Institute of Child Health and Human Development, NIH

National Institute of Child Health and Human Development (NICHD)

NIH Institutes/Offices/Centers Relevant to NBS

NICHD	lead institute on newborn screening
NINDS	developmental neurological disorders
NIDDK	metabolic conditions
NIBIB	point of care technologies
NHGRI	ELSI, genomics, linkages to conditions
NHLBI	hemoglobinopathies and cardiomyopathies
NIAMS	neuromuscular conditions
NIDCD	hearing impairment
NIEHS	environmental factors associated with congenital defects
NLM	newborn screening resource pages including the Genetics Home Reference
FIC	genetics and informatics training, international research efforts
ORD	rare diseases

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

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 flourescent bead array technology

Potential NIH Initiative: Newborn Screening Translational Research Network

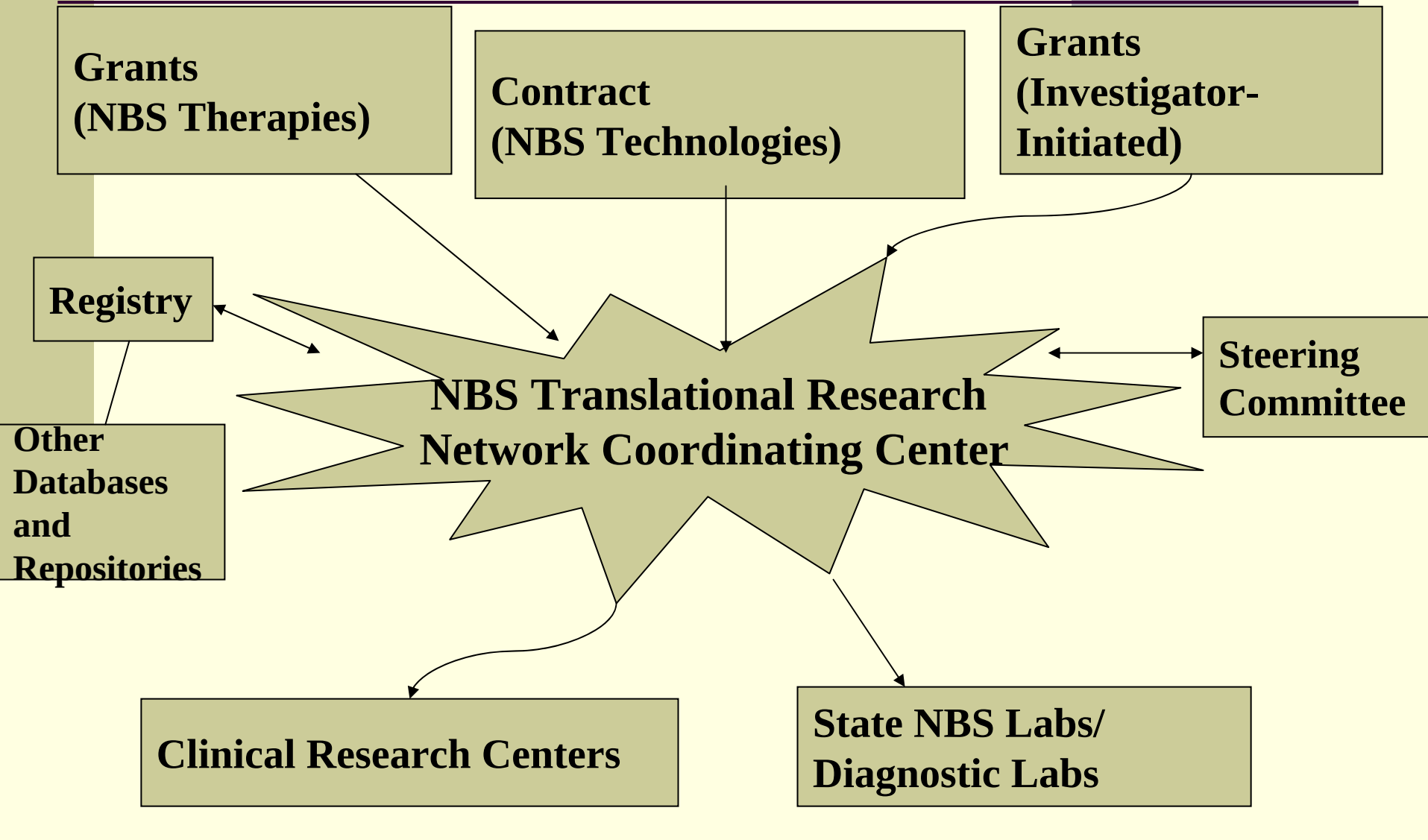
- Validate screening technologies and therapeutic interventions for rare diseases
- Increase access to dried bloodspots and other samples for research use
- Perform longitudinal health outcomes research on individuals identified through newborn screening
- Establish an informatics system to link researchers 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:

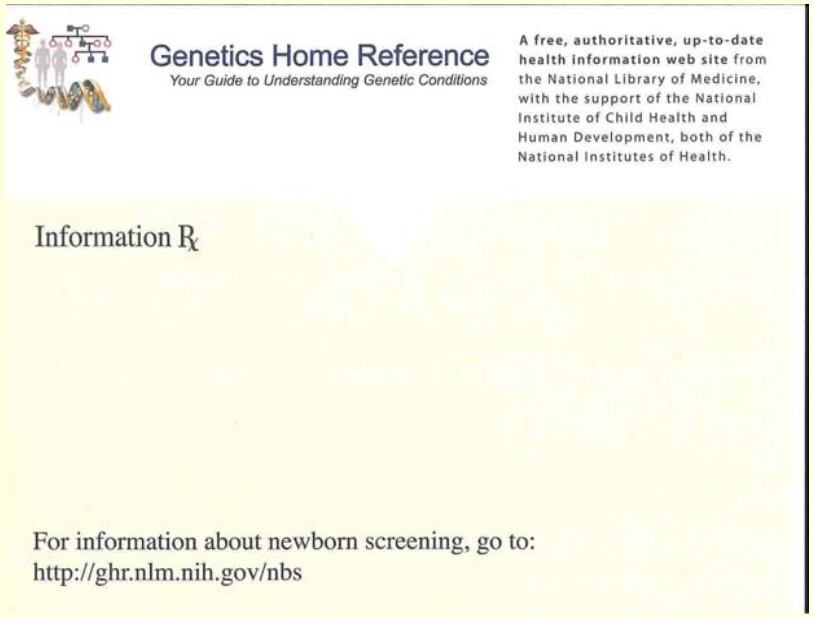
- Databases
- Biospecimen repositories
- Clinical Researchers
- Public Health Programs

Newborn Screening Translational Research Network



InfoRx

- InfoRx pads to assist HCPs refer patients to up-to-date, authoriative, 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
- GHR: http://ghr.nlm.nih.gov



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