

Newborn Screening: Update on National Priorities and Perspectives

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Presentation Outline

- ◆ **ACMG Overview**
- ◆ **NCC Overview**
- ◆ **LTFU Efforts**



Alphabet Soup

- ◆ **ACMG = American College of Medical Genetics**
- ◆ **NCC = National Coordinating Center**
- ◆ **NICHD = *Eunice Kennedy Shriver* National Institute of Child Health and Human Development**
- ◆ **NBSTRN = Newborn Screening Translational Research Network**
- ◆ **NBSTRN CC = Newborn Screening Translational Research Network Coordinating Center**

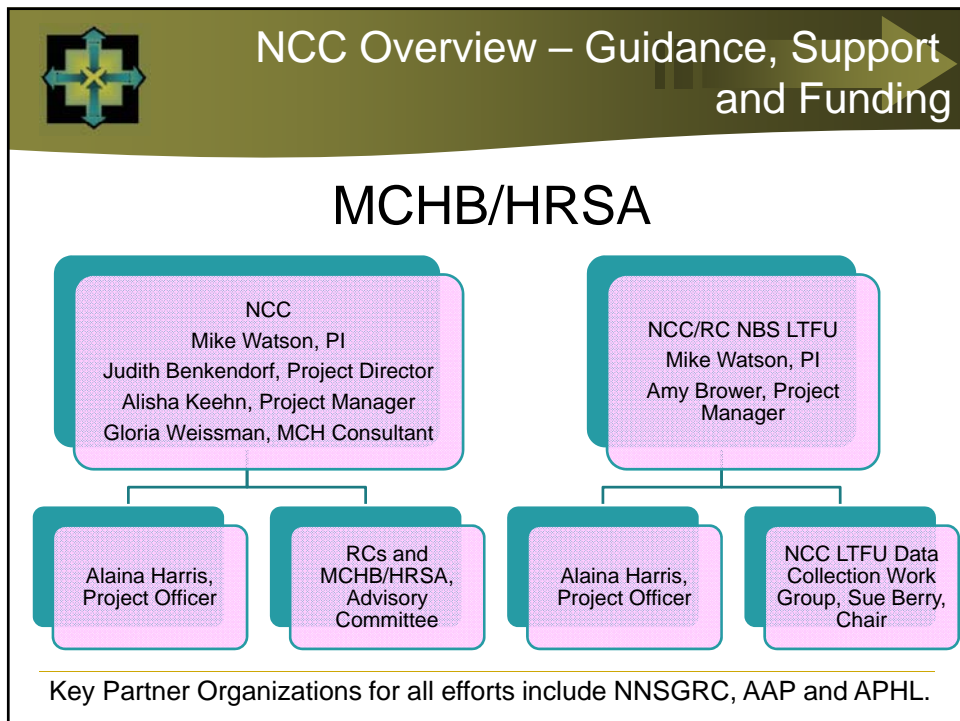
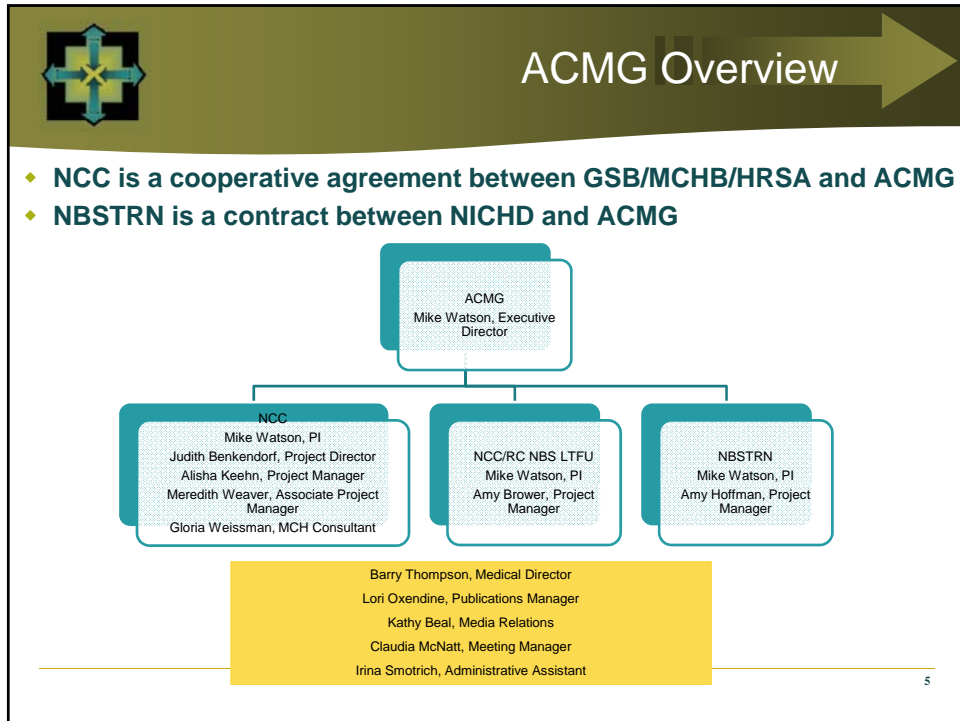
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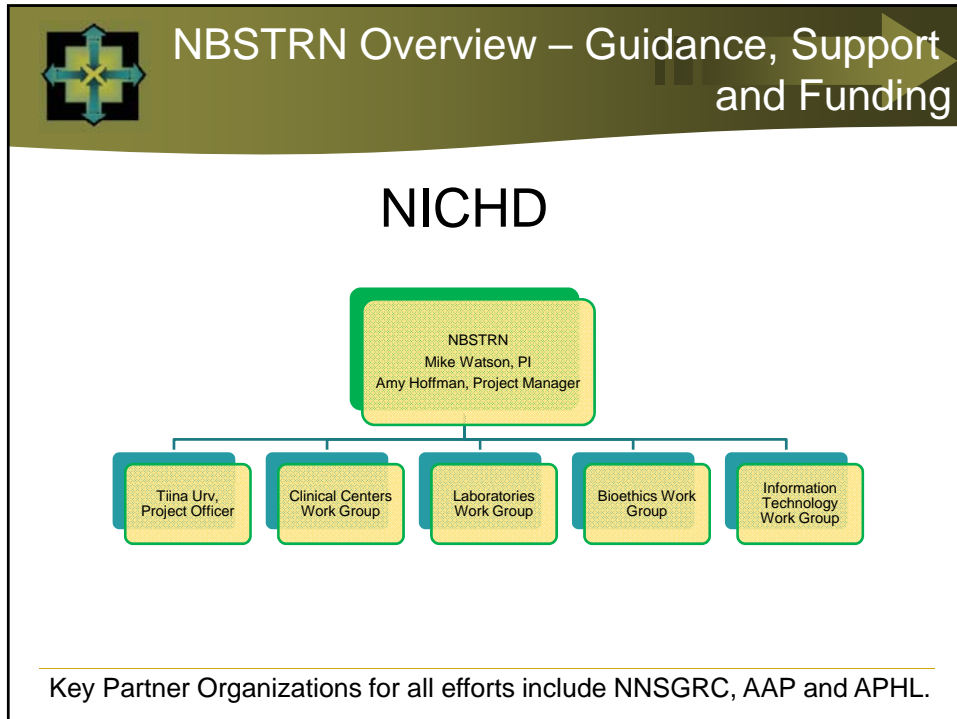


NCC Objectives

- ◆ **Coordinate shared activities between the seven RCs and other HRSA supported resource centers and projects**
- ◆ **Develop educational materials and management guidelines at the national level**
- ◆ **Identify policy issues or other barriers to improving access, utilization and quality of services**

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Focus on Long-Term Follow-Up (LTFU)

- ◆ **The goal of LTFU is to “assure the best possible outcome for individuals with disorders identified through newborn screening”.***
- ◆ **Four components identified**
 - **Care coordination through a medical home**
 - **Evidence-based treatment**
 - **Continuous quality improvement**
 - **New knowledge discovery**

*Kemper et al. Genet Med 2008;10(4):259-261.

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Health Information Exchange

- ◆ **LTFU requires health information exchanges throughout the lifetime of NBS identified patients.***
- ◆ **LTFU information systems (IS) should employ best practice approaches to information technology development.**
- ◆ **LTFU IS should connect stakeholders, processes and outcomes through the collection, integration, evaluation and sharing of key data and metrics.**

*Hinman et al. Genet Med 2009;11(6):418-424.

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LTFU Data Collection Project

- ◆ **Goals**
 - Determine the LTFU IS needs of state newborn screening programs to conduct LTFU of NBS identified patients.
 - Incorporate findings into a public health evaluation tool that can be used to monitor and improve LTFU.
- ◆ **Structure**
 - Supplement to NCC LTFU activities.
 - Cooperative agreement with HRSA.
 - Work Group – Sue Berry, Chair
- ◆ **Staff**
 - Project Manager – Amy Brower
- ◆ **Project Officer**
 - Alaina Harris

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LTFU – Data Collection Project

- ◆ **Common Elements for LTFU**
 - Demographics
 - Basic diagnostic information
 - Genotype
 - Morbidity/Mortality
 - Clinical Measurements
 - Types of providers
 - Types of services
 - Treatments
 - Hospitalizations
 - Developmental Benchmarks

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LTFU - Current Activities

- ◆ **Collaboration with NBSTRN**
 - Establish initial clinical histories of disorders identified in NBS for use in public health and research initiatives.
 - Joint meeting of NCC LTFU Data Collection Work Group with NBSTRN Clinical Centers Workgroup.
 - Initial focus on metabolic diseases utilizing several LTFU data collection efforts as a baseline.
- ◆ **Assessment of ongoing LTFU data collection projects within the seven Regional Collaboratives (RC).**

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LTFU - Future Activities

- ◆ Establish clinical histories of all NBS identified disorders
- ◆ Survey and summarize the types of information required for LTFU.
- ◆ Evaluate and consider issues related to LTFU information systems.
- ◆ Communicate findings to groups developing HIT standards for consideration.
- ◆ Conduct quarterly conference calls and meetings of the NCC Data Collection Work Group.
- ◆ Report efforts to HRSA.

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