



Report of the National Coordinating Center □ **Providing Resources for Bringing Genetics and NBS Services to Local Communities □**

Michael S. Watson, PhD, FACMG

American College of Medical Genetics

**National Coordinating Center for the Genetics and Newborn
Screening Regional Collaborative Groups**

July 25, 2007



The National Coordinating Center (NCC) for the Genetics and Newborn Screening Regional Collaborative (RC) Groups

- ◆ **A quick review of who we are**
 - **NCC Staff and ACMG resources**
- ◆ **NCC Project Goals: *Includes next 5 years***
- ◆ **National Activities and Resources**
 - **Communication**
 - **Technical Assistance**
 - **Content expertise**
 - **Networking and facilitation**
- ◆ **Workgroups and Projects**
 - **Opportunities for collaboration**



Goals of the NCC (1)

- ◆ **Facilitates communication and collaborations between RCs and nationally**
- ◆ **Responds to issues best addressed nationally**
 - **Minimize duplication of efforts**
 - **Take advantage of shared resources**
- ◆ **Organizes trans-RC workgroups**
- ◆ **Provides guidance and technical resources**
- ◆ **Translates best practices and RC activities into national initiatives**
- ◆ **Focus on non-genetics providers**



What is the NCC doing?



What is the NCC Doing?

- ◆ **Communication**
- ◆ **Technical Assistance**
 - **Teleconferences**
 - **Program Evaluation**
 - **Monitoring Legislation Impacting Genetics**
- ◆ **Work Groups**
 - **Telegenetics**
 - **Emergency Preparedness**
 - **Provider Networks**



Technical Assistance: Program Evaluation

- ◆ **Prior to non-competitive renewal MCH consultant reviewed all RC programs for relationship with:**
 - **MCHB and Heritable Disorders Program goals**
 - **GSB projects and partners**
- ◆ **Matrix to be updated for new grant cycle**
- ◆ **Workgroup currently convening with consultant to develop shared evaluation measures for new grant cycle**
 - **To be used by RCs and NCC, as applicable**



Management Guidelines: NBS ACT(ion) Sheets and Diagnostic Algorithms

- ◆ **Designed to aid in evaluation and treatment of newborns with abnormal NBS test results**
 - **Developed for all conditions in uniform panel**
 - **Approved by ACMG Board and endorsed by AAP Board**
 - **Posted on ACMG website, with links from**
 - **Genetics Home Reference, NNSGRC, and many others**
 - **Distributed to NBS labs and programs**
 - **To accompany all “screen positive” lab reports**
 - **Distributed to RCs to coordinate local and regional use**
- ◆ **Facilitate communication between labs and PCPs**



Management Guidelines: Next Steps (1)

- ◆ **NBS ACT Sheets currently being completed for non-S hemoglobin alleles**
 - Includes α -globin conditions and the thalassemias
- ◆ **NB Hearing Screening Practice Guideline and ACT Sheet being adapted into a brochure for parents of “screen positive” infants**
 - Tailored to low-literate populations
 - Emphasis on genetic workup as part of etiologic diagnosis
- ◆ **ACT sheets to be developed to support assessment of family history**



Management Guidelines: Next Steps (2)

- ◆ **Supplemental grant to NCC to develop similar primary care guidance for genetic testing**
 - **To be disseminated by genetics laboratories in the US with “positive” test results**
 - **Fragile X, cystic fibrosis and hemoglobinopathy testing to serve as models for genetic tests**



Educational Program Development (One-year supplement: June 2006-7)

- ◆ **Meeting to be convened Fall 2007 with major developers of EMR and HIS:**
 - **To include key RC experts and NCC partners (e.g., Public Health Informatics Institute, AAP)**
 - **Seeking to integrate ACT sheets as high quality educational pop-ups within electronic systems**
 - Point-of-care education and decision support
 - Personalized Healthcare Workgroup (HHS-OS) using NBS as a model for integrating genetic testing into EHR
 - **Integration of genetics management guidelines into EMR will be one way to address challenge of communicating updates to users/adopters**



Data Collection and Related Projects: The Future is Now

And in with the new:

- ◆ **National Collaborative Research in Rare Genetic Diseases - NICHD award to ACMG**
- ◆ **First meeting February 2007**
 - **Broad consensus recommendations**
 - **Overlap with NCC/RC activities and goals**
- ◆ **Collaborators include:**
 - **NCC and RCs**
 - **Newly forming Metabolic Disease Consortium (SIMD, ACMG and others)**
 - **HRSA, NNSGRC, ASHG and more**



National and International Data Collection Activities

- ◆ **Regional Genetics and Newborn Screening Collaboratives (MCHB/HRSA)**
 - **Region IV**
 - **Mountain States**
 - **SERGG**
 - **New England**
- ◆ **NBSRN (NICHD)**
- ◆ **Other countries**
 - **Denmark and EU activities**
- ◆ **International Human Variome Project**



Data Collection Needs Related to Newborn Screening

	Newborn Screening	NBS Candidates	Other Genetic Diseases
Data Needed	LTFU	Natural history	Natural history
Why Data is Needed	NBS Program Evaluation; 2 nd generation tests and treatments	Inform decision - making on use; New test and treatment trials	Research, clinical investigation, and diagnostic uses
Conditions	29 core conditions >25 secondary targets 51 markers 175 loci	SCID SMA LSDs Hyperbilirubinemia Connexin 26 Connexin 30 CMV Pendred	Huntington Disease Alzheimer BRCA Parkinson Disease
Funding Sources	States CDC, HRSA RCs	NIH/NICHD (NBSRN), HRSA RCs	NIH Industry Foundations



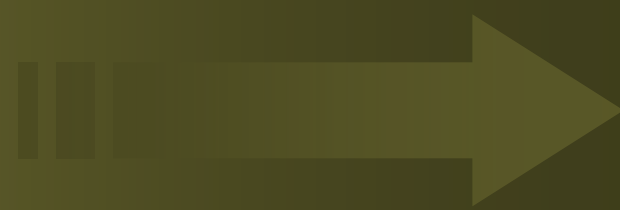
Issues to Address to Realize Goals

- ◆ **State sovereignty vs. realistic scientific needs for as many cases as possible to be gathered**
- ◆ **Databases**
 - **Standardizing laboratory and clinical languages for EMR compatibility**
 - HHS to use NBS as a model
 - **EHRs to support patient needs**
- ◆ **Role of NCC/RC system will be critical**
 - **Development of system (s)**
 - **Data collection**
- ◆ **International integration**
 - **Other countries**
 - **International Human Variome Project**



Moving into an Electronic Health System World

- ◆ **Organizing meetings with Electronic Medical Record (EMR) and Clinical Decision Support tool developers**
- ◆ **Working with AHIC's Personalized Medicine Work Group**
 - **NBS as demonstration project**
 - **Would utilize the RCs to implement**



Thank you