



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

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October 19, 2006



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

- ◆ **Who are we?**
 - **Staff and ACMG resources**
- ◆ **NCC Project Goals**
- ◆ **National Activities and Resources**
 - **Communication**
 - **Technical Assistance**
 - **Workgroups and Projects**
- ◆ **Opportunities for Collaboration**



The Players



Structure of NCC

- ◆ **Cooperative agreement between GSB/MCHB/HRSA and American College of Medical Genetics (ACMG)**
- ◆ **Staff:**
 - **Michael Watson, PhD - Project Director**
 - **Judith Benkendorf, MS, CGC - Project Manager**
 - **Lori Oxendine, BFA, AIGA - Meetings/Publications**
 - **Gloria Weissman, PhD - MCH Consultant**
- ◆ **Advisory Committee**
 - **Jon Zonana, MD - Chair**
 - **RCs and MCHB/HRSA**
 - **18 Partner Organizations**



NCC Advisory Committee: Partner Organizations and Representatives

- ◆ Jonathan Zonana, MD (Chair)
- ◆ Cheryl Aldridge, MN, RN, CPNP (NAPNAP)
- ◆ Steven Downs, MD
- ◆ Debra Doyle, MS, CGC (UW-GSPP)
- ◆ Nancy Green, MD (MOD)
- ◆ Stephen Groft, PharmD (NIH-ORD)
- ◆ James Hanson, MD (NICHD)
- ◆ Alissa Johnson (NCSL)
- ◆ Norman Kahn, MD (AAFP)
- ◆ Christopher Kus, MD, MPH (AMCHP)
- ◆ Harvey Levy, MD (ACMG)
- ◆ Jelili Ojodu, MPH (APHL)
- ◆ Lauren Ratner, MPH, MSW (ASTHO)
- ◆ Sonya Ross, BA (SCDAA)
- ◆ April Studinski, MS, CGC (NSGC)
- ◆ Fan Tait, MD (AAP)
- ◆ Brad Therrell, PhD (NNSGRC)
- ◆ Tracy Trotter, MD
- ◆ Mendel Tuchman, MD, PhD (SIMD)
- ◆ Ajay Vatave, MD PhD (CDC)
- ◆ Lisa Wise, MA (Genetic Alliance)



American College of Medical Genetics

- ◆ Provides education, resources and a voice for ~1500 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other health care professionals committed to the practice of medical genetics.
- ◆ **ACMG initiatives include activities to:**
 - **Advance the practice of medical genetics**
 - From promulgating laboratory and practice guidelines to advocating for fair health policies;
 - **Increase access to genetic services and improve the public's health;**
 - **Promote development and implementation of methods to diagnose, treat and prevent genetic disease.**



Part Two

The Process



Goals of the NCC

- ◆ **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**
- ◆ **Develops management guidelines and tools for RCs, providers, policymakers and other stakeholders**
- ◆ **Interfaces with ACMG when approval needed for NCC work products**



Part Three

The Projects



Communication

- ◆ **NCC website: www.nccrcg.org**
- ◆ **Quarterly e-newsletter**
 - **Featuring activities of NCC, RC and Partner Organizations**
 - **First issue in Fall 2006**
- ◆ **Conference calls**
 - **With each RC, Spring 2006**
 - **Monthly calls for all PIs, NCC and GSB/MCHB**
- ◆ **Calendar circulated monthly**



Technical Assistance (1)

- ◆ **NCC Teleconferences (2005 - 2007)**
 - **Telemedicine (July 2005)**
 - **Financing Genetics and NBS Services (Sept. 2005)**
 - **Ideas needed for future web casts**
 - **Carrier screening**
 - **Transition to adult care**
- ◆ **Teleconferences for state legislators**
 - **NCSL/NCC, May 2006**
 - **Telegenetics**
 - **Emergency Preparedness**



Technical Assistance (2)

- ◆ **Program Evaluation**
 - **MCH consultant reviewed all RC programs for relationship with:**
 - MCHB and Heritable Disorders Program goals
 - GSB projects and partners
 - **Updated based on progress reports**
 - **Used by NCC to assist RCs**
- ◆ **Small grant guidance (forthcoming)**
- ◆ **Workgroup products**



Workgroups

- ◆ **Telegenetics: Capacity Development**
 - RC representatives and national experts
 - Fall 2006: Survey to assess existing technology and capacity
 - Resources for NCC website to be identified and developed
 - Addressing legal issues with interstate licensing
- ◆ **Emergency Preparedness**
 - RC and national representatives
 - Integrating NYMAC NBS lab planning (NERGG, NYMAC, Region 4, SERGG and Western States) with SERGG patient management planning
 - Funding from NIH-ORD to sponsor formal workgroup meeting, to include national stakeholders (e.g., FEMA EMAC, HHS agencies, informatics and e-communication experts)



Management Guidelines (1)

- ◆ **NBS ACT(ion) Sheets and Diagnostic Algorithms**
 - **For all conditions in uniform panel**
 - **Approved by ACMG Board of Directors**
 - **Posted on ACMG website**
 - **Genetics Home Reference, NNSGRC, and many others link to these**
 - **Distributed to NBS labs and programs**
 - **To accompany all “screen positive” lab reports**
 - **Distributed to RCs to coordinate use with local and regional plans**
 - **Survey of utility presently being conducted**



Management Guidelines (2)

- ◆ **NBS ACT Sheets currently being written for non-S hemoglobin alleles**
 - **Beyond SCD and the thalassemias**
- ◆ **Support for meeting in Morocco (11/05) to strengthen NBS in N. Africa and Middle East**
- ◆ **NB Hearing Screening 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**



Management Guidelines (3)

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**
- ◆ **Model management guidelines for primary care providers around transition to adult care**
 - **Pilot project using hemoglobinopathies**
 - **Information to be collected on dissemination methods (pediatricians versus adult medicine)**



Educational Program Development (1) (One-year supplement)

- ◆ **Assessment of genetics content on US Medical Licensing Exams (through NBME)**
 - **Step One showed improvement after prior review**
 - **Review of Steps Two and Three**
 - Tests examine applying genetics knowledge to practice
 - Recognized area of deficiency among graduating medical students
- ◆ **Medical school genetics curriculum evaluation**
 - **Jefferson Medical College as case study**
- ◆ **Data useful for making recommendations for improvement and observing changes over time**



Educational Program Development (2) (One-year supplement)

- ◆ **Meetings to be convened with major developers of EMR and HIS:**
 - **To include NCC partners, Public Health Informatics Institute and AAP**
 - **Seeking to integrate ACT sheets as high quality educational pop-ups within electronic systems**
 - **Direct input from genetics community to be sought for developing management guidances in EMRs to assist providers in fulfilling their role(s) in genetic service delivery.**



Infrastructure Development: Genetic Service Delivery (1)

- ◆ **Developing and mapping (using GIS) a national network of genetic service providers**
- ◆ **Services to be categorized (lab, clinical, special populations, etc.)**
 - **ACMG and RCs to assist with developing standards for genetic services and providers**
 - **SIMD to assist with developing standards for metabolic service providers**
 - **Subspecialty/organ/disease clinics included**



Infrastructure Development: Genetic Service Delivery (2)

- ◆ **Locations of identified NBS patients to be added**
- ◆ **Data will be useful to:**
 - **Identify provider gaps**
 - **Justify support for services to fill gaps**



Data Collection and Related Projects

- ◆ **PI Meeting (Sept. 7th) to discuss prospective collaborations around data collection and uses**
- ◆ **Building the Business Case will involve collection of outcomes data**
 - **Integration with Quality of Genetic Services Project**
- ◆ **NBS data collection projects**
 - **Tracking NBS pilot studies**
 - **Genetics patient registry**
 - **Organizing metabolic centers around follow-up**



The Future

- ◆ **Is busy**
- ◆ **Is exciting**
- ◆ **Is dependent on successful communication, collaboration and shared goals.**

