


Report of the National Coordinating Center

Providing Resources for Bringing Genetics and NBS Services to Local Communities


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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


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Part One

The Players

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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, ALGA - Meetings/Publications
 - Gloria Weissman, PhD - MCH Consultant
- ◆ Advisory Committee
 - Jon Zonana, MD - Chair
 - RCs and MCHB/HRSA
 - 18 Partner Organizations


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NCC Advisory Committee: Partner Organizations and Representatives

<ul style="list-style-type: none"> • 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) 	<ul style="list-style-type: none"> • Jellii 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)
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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.

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Part Two

The Process


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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


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Part Three

The Projects


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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


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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

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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


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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)


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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


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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

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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)


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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**

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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.

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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

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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

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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

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The Future

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



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