

Achieving Community Service Systems For Genetic And Newborn Screening Services

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MCHB Strategic Plan: Its Mission

To provide national leadership and to work, in partnership with states, communities, public-private partners, and families to strengthen the MCH infrastructure, assure the availability and use of medical homes, and build the knowledge and human resources, in order to assure continued improvement in the health, safety and well-being of the MCH population.

MCHB Genetic Services Branch

- Help State MCH officials, public health professionals, and families respond to scientific findings and technologies in the fields of genetic services and newborn screening.
- Clarify genetic services, resources and policy issues.
- Establish capacity infrastructure for delivering services.
- Improve access to services for medically underserved populations.
- Integrate genetics research, technologies and medicine into State programs and their local communities.
- Develop education and training opportunities in genetic medicine for consumers and health professionals.

Regional Genetic and Newborn Screening Service Collaboratives

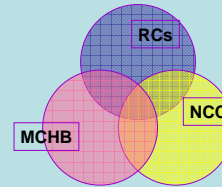
The Primary Goal

- Ensure that children with heritable disorders and their families have access to quality care and appropriate genetic expertise and information in the context of a medical home that provides accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally effective care, with the following objectives:
 - To strengthen communication and collaboration among public health, families, primary care providers, and genetic medicine and other subspecialty providers
 - To quantitatively and qualitatively evaluate outcomes of projects undertaken to accomplish their goals.

Moving from Networks to Collaboratives

The Continuum of Collaboration

- **Networking**-Exchanging information for mutual benefit
- **Coordination**-Exchanging and modifying activities for mutual benefit
- **Cooperation**- Exchanging information, modifying activities, sharing resources for mutual benefit to achieve a common purpose
- **Collaboration**- Exchanging information, modifying activities, sharing resources and enhancing the capacity of another for mutual benefit and to achieve a common purpose, by sharing risks, resources, responsibilities, and rewards



Our Resources

- The **Genetic Services Policy Project** - describes models of genetic services delivery, including economic and policy issues; in partnership with Washington State to develop an agenda to translate genetic research into practice. Website, <http://depts.washington.edu/genpol>.
- A **Child Health Data Integration Project** - supports a newborn screening best practice model; developed with the Public Health Informatics Institute. A long standing effort to combine and coalesce newborn screening technology and computerized systems at the state level.
- **Newborn screening ACTION (ACT) sheets (ACMG)** and confirmatory algorithms - describe the short-term actions that a health professional should take for follow-up of a newborn who screens positive for a condition in the uniform newborn screening panel. Websites <http://www.acmg.net>, and <http://mchb.hrsa.gov/>.
- The May 2006 **Supplement to Pediatrics** entitled "A Look at Newborn Screening: Today and Tomorrow". It highlights key developments and issues in the past 6 years.

- A parent brochure, "**These Tests Could Save Your Baby's Life**"; and 2 Guides for health professionals- "**7 Things Parents Want to Know About Newborn Screening**" and "**7 Things Parents Want To Know From their Child's Health Professional**". These materials have been distributed to providers (obstetricians, family practice physicians, and nurse midwives) in partnership with the AAP, ACOG, and the AAFP. Website, <http://mchb.hrsa.gov/programs/default.htm>.
- A consumer-based **Family History Tool** that utilizes family traditions and oral history for health promotion. MCHB partners are: the Library of Congress, the Genetic Alliance, and the American Society of Human Genetics. Website, <http://www.geneticalliance.org>.
- **Gene Tests-GeneClinics**, a medical genetics information resource developed for health care providers and researchers. Website, www.geneclinics.org.

National Newborn Screening Genetics Resource Center

- **Performance Evaluation and Assessment Scheme (PEAS)**. An interactive self-assessment checklist to support improvement of newborn screening (NBS) State systems, and ultimately patient services, with emphasis on follow-up.
- **Tandem Mass Spectrometry (MS/MS) Training**. A week long, hands on course hosted by Duke Medical School (NC) & Baylor Institute of Metabolic Disease (TX). Offered to newborn screening laboratory and follow-up staff anticipating or using MS/MS technology.
- **Emory University Newborn Screening Data Evaluation Project**. A project utilizing statistical methods to examine ten years of newborn screened confirmed cases information to determine if any spatio-temporal associations exist on a national scale.

National Newborn Screening Genetics Resource Center

- **Clinical Genetic Services Data Collection Project.** To assess whether individuals identified with conditions through newborn screening receive genetic services, and to define gaps in data collection and potential barriers to services.
- **National Newborn Screening Data System Web site:** <http://www2.uthscsa.edu/nnsis/>. This site, is a data base for collecting and providing data on newborn screening in the states and U.S. territories.
- **National Newborn Screening and Genetics Resource Center (NNSGRC) Web site:** <http://genes-r-us.uthscsa.edu>.

What The Collaboratives Are Doing

Building The Capacity of State Genetics And Newborn Screening Programs

- **Western States**
 - Needs Assessments for State Genetics and NBS plans completed for **Guam, Alaska** and **Nevada**.
 - **Solutions: Nevada** - re-establish the Genetics Advisory Comm. and develop a State Genetics Plan; **Guam**- initiated genetic consults via telemedicine and in-person genetics clinics, assisted in providing a user fee-based plan for NBS, promoting NBS legislation, and will increase medical education with in person lectures, teleconferencing, CD-ROMS, and web-based conference.

Communication Within the Region

- **SERGG**
 - Telecommunications system to link States.
 - Webcast and face-to-face technology.
 - Academic geneticist and public health representative from each State.
 - Purchased equipment and provided training.
 - Education and Committee meetings.

Maldistribution of Genetic Services: Telemedicine

- **Western States**
 - Washington State Dept of Health is continuing to pilot telehealth genetic services in rural **Washington** and **Alaska**.
 - Oregon Health & Science University to plan, pilot and evaluate project activities in Oregon communities, including the Southern and Southwest coasts of **Oregon** for hemoglobinopathies, and **Idaho**.
 - State of **Idaho** to plan, pilot and evaluate the practice model for delivering genetic services to its rural areas.
 - **Hawaii** will provide telemedicine to **neighbor islands**.
 - **Hawaii** is initiating telemedicine genetic evaluation and counseling visits for **Guam**.

Maldistribution of Genetic Services: Mapping

NYMAC

- Limited to **PKU, Hemoglobinopathies, and Trisomy 21**
- Cases diagnosed by State NBS programs in the last 5 years.
- Calculated the average distance from patients to treatment centers- **there are pockets of patients far removed from services.**
- To be determined- availability and distribution of services.

NERGG

- Epidemiology mapping of genetic services for **CF, hemoglobinopathies & metabolic disorders.**
- Patients are in **close proximity to service centers.**
- Addressing "border babies"; interstate access to services, tracking, privacy, medical licensure; and immigrant populations.

Laboratory Test Performance And Uniformity of Testing

Region 4

- To improve **analytical performance, laboratory quality improvement, and uniformity of the testing panel using MS/MS.**
 - Completed -Implementation of **ACMG/HRSA uniform panel is highest in the nation.** 100% screening by MS/MS. 99% implementation of 20 primary targets. 80% implementation of the 22 secondary targets.
 - Completed - Established a regional **true positive database** for the region. Achieved False Positive Rate <0.3% in the whole region.
 - Introduced **2nd Tier Testing for Congenital Adrenal Hyperplasia (CAH).** Prospective collection of anonymized blood spots of CAH false and true positive cases

Laboratory Test Performance And Uniformity of Testing

Region 4 con't:

- A "**Sample Exchange Program**" protocol was developed - View protocol on Region 4 website.
- A **data collection site** is set-up on the website.
- **To expand current regional activities to include other state programs** that have expressed interest to participate in data collection and sharing for the purpose of defining disease specific disease ranges and clinically defined cut-off ranges. ([Supplement Project](#))
- Evaluation of NBS for detection of **Endocrine Disorders** to conduct an epidemiological evaluation of **CH and CAH** to improve detection rates and reduce false positive and negative rates in the region. ([Supplement Project](#))

Laboratory Test Performance And Uniformity of Testing

SERGG

- To establish a regional **MS/MS database in concert with the Mayo Clinic Lab,** and to document the **impact of nutrition intervention and low birth weight infants on positive MCADD screens with 5 states in the Region.** ([Supplement](#))

NYMAC - Children's Hospital of Philadelphia

- **Duarte Galactosemia (DG): A Pilot Study of Biochemical and Neurodevelopment in Children Detected by Newborn Screening** ([Supplement](#))
 - Compare clinical outcome of patients who receive a galactose-free diet with those on an unrestricted diet, and assess the relationship between developmental and clinical outcome and biochemical parameters.

Laboratory Emergency Preparedness

NYMAC

- **A multi-level back-up plan.**
- **Development of an Emergency Shut-down Back-up Template for State agreements:** authorities, contacts, implementation, communication, specimen transport, reporting test results, retention etc. Add a follow-up component
- **Concept of a model of a centralized back-up NBS Lab facility (CDC)** when multiple labs lose function: staffing; maintenance; instrumentation, funding, vendor participation, platforms, scope of testing, and secure data exchange.
- Participants are- **SERGG, Region 4, NERGG, NNSGRC, APHL, CDC.**
- **APHL NBS Committee** – Lab ER Preparedness a priority.

SERGG

- Follow-up **treatment of metabolic disorders:** command control, leaders, communication, national ER telephone number, list of care centers, pharmaceutical companies and pharmacies for specialized medicines, & collaboration with specialty centers.

Short Term and Long Term Follow-up

Region 4

- Pilot study of a **MCADD Disease Registry** that may become a national activity.
- Initiated a process to collect diagnostic and medical management data from metabolic specialists on patients with MCADD.
- Identified data elements, >30 metabolic geneticists, and a software vendor.

Education and Training

NERGG

- Power Point presentation to educate **health care providers** about the importance of the **Family History in clinical care.** Training scheduled for medical practices and community health centers

SERGG

- Regional **metabolic nutrition** assessment and services training; an international meeting, "**First Annual Genetic Metabolic Nutrition Conference**", April 2005; a new professional organization, Genetic Metabolic Dietitians, International (GMDI); and a website component of presentations from the conference.

Education and Training

Western States

- Development of the “**Genetic Conditions and Congenital Malformations**” section of the “**Nursing Care of Children with Special Health Needs in the Community**” resource manual.

Region 4

- Online NBS Education module for **prenatal care providers** including the availability of CME credits, “**Newborn Screening: What Prenatal Caregivers Need to Know**”.

Education and Training

NERGG

- Regional modification of the **Genetic Alliance** manual for health care providers and consumers.

Region 4

- Implementation of “**Medical Homes in Action**” website to provide **primary care physicians and families** with information and resources related to disorders diagnosed by newborn screening: **GIS map** of location of metabolic geneticists, hematologists and endocrinologists; NBS disorder specific info; State NBS program info; national, state & local resources.

SERGG

- **Ask the Geneticist**” – an online education website component that answers questions from primary care providers.

Newborn Screening Information Systems

Region 4

- **Indiana University – Pilot Testing Adaptive Turnaround Documents to Link Newborn Screening Programs, Sub-specialists, and the Medical Home**
 - communication between the NBS program and the medical home using tailored, computer-readable paper document technology called *adaptive turnaround documents*. The technology will support: effective clinical data collection at the point of care; provide a follow-up system for tracking time from birth to diagnosis, monitoring disposition of all cases, collecting confirmatory biochemical data, and identifying reasons for late diagnosis of cases. ([Supplement](#))

New Funding New Cycle

- Program: **U22: Heritable Disorders (93.110)**
- Announce Number: **HRSA-07-016**
- Cooperative Agreement
- Type: Competitive
- Eligibility: any public or private entity, including an Indian tribe or tribal organization, and a faith based and community based organization.
- No. of Funding Years: **5 years**
- Due dates, Preview & Guidance – N/A and in process

Next Steps

- Serve as the regional center
- Take a regional, collaborative approach to facilitating access to the genetics expertise, services, and technology that providers and families need
 - This approach should be based on the regional and/or State genetics plans developed during the first three years
- Design, develop, and implement practice models and systems for delivering recommended genetic screening and testing, diagnosis, treatment, and short- and long-term follow-up services
 - Special attention should be paid to the development of effective practice models for adolescents and young adults with heritable diseases who are transitioning from pediatric to adult health care. Any data on the effectiveness of practice models and systems that are currently in place should be included in the narrative application.

Next Steps, cont

- Have public-private partnerships in place to provide the genetic, newborn screening and other relevant subspecialty expertise and services in the region and to link those services with the medical home.
 - Priority partners are families, primary care providers and public health entities
- Serve as a focal point for effective dissemination of relevant, up-to-date information on genetics aimed at both consumers and service providers,
 - Information developed by the NCC (central role of development)
- Work, through the NCC and GSB to share those projects that have potential interregional and national significance with the other RCs and their partners.

Next Steps, cont

- Demonstrate active participation in the National Newborn Screening Information System (NNSIS) by the States in the Region for newborn screening data submission.
- If the applicant establishes a small project program to accomplish particular ongoing activities relevant to the needs of the region, this program and its activities should be **clearly linked** to the Regional and/or State genetics plans developed during the first three years (FY 04-07) of funding for RCs.
- Two Priority Projects:
 - **Laboratory Quality Assurance Activity.**
 - **Follow-up Activity.**

GSB Contact Information

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