

Heritable Disorders Program UPDATE AND UPBEAT

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Genetic Services Branch
HRSA Maternal and Child Health Bureau
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HRSA Administrator

■ **Mary Wakefield, Ph.D., R.N.**

- University of North Dakota (UND), associate dean for rural health at the School of Medicine and Health Sciences, and director of the university's Center for Rural Health.
 1. Rural health care
 2. Quality improvement
 3. Expand and improve services for the uninsured or underserved
 4. Expand and improve care provided at the community health centers
 5. Population-based screening and preventive health
- Capitol Hill - In the 1990s, she served as chief of staff for two North Dakota senators: Kent Conrad (D) and Quentin Burdick (D).
- Served on the IOM committees that produced the [landmark reports](#)
 - *To Err is Human and Crossing the Quality Chasm*
 - *Health Professions Education*
 - *Quality through Collaboration: Health Care in Rural America.*
- Served on National Advisory Councils/Commissions for :
 - HRSA / Health Resources and Services Administration/
 - CMS Medicare / Centers for Medicare and Medicaid
 - AHRQ / Agency for Healthcare Research and

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HRSA MCHB Genetic Services Branch

■ **Sara Copeland, MD**

- Medical Director and Deputy Chief, 9/28/2009.
- Division of Medical Genetics
University of Iowa Children's Hospital
Medical Director of Newborn Screening for Iowa

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ACMG NCC LTFU

■ **Amy Brower, PhD**

- Project Manager, American College of Medical Genetics, NCC Long-Term Follow-up
- Past ACHDNC Committee Member
- Past Chair and current Member, Subcommittee on Laboratory Standards and Procedures
- Third Wave Molecular Diagnostics
- South Sioux City, NE

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Legislation – Heritable Disorders Program

- Title XXVI of the Children’s Health Care Act of 2000, **Screening for Heritable Disorders**
 - To assist and improve the ability of States to provide newborn and child screening for heritable disorders.
 - Established a grant program (Cooperative Agreements)
 - 7 Regional Genetic and Newborn Screening Service Collaboratives /RC
 - 1 National Coordinating Center/ NCC
 - Established the Advisory Committee on Hereditary Disorders in Newborns and Children Committee)
 - Committee is advisory to the HHS Secretary
 - Committee is advisory to the **Regional Collaborative Program**

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Public Law 110-204
Newborn Screening Saves Lives Act of 2008 (NBSSLA)
Established for Expanded Screening

A BILL

To amend the Public Health Service Act to establish grant programs to provide for education and outreach on newborn screening and coordinated followup care once newborn screening has been conducted, to reauthorize programs under part A of title XI of such Act, and for other purposes.

1 *Be it enacted by the Senate and House of Representa-*
2 *tives of the United States of America in Congress assembled,*

3 **SECTION 1. SHORT TITLE.**

4 *This Act may be cited as the “Newborn Screening*
5 *Saves Lives Act of 2008”.*

6 **SEC. 2. IMPROVED NEWBORN AND CHILD SCREENING FOR**
7 **HERITABLE DISORDER.**

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NBSSLA: Summary of Provisions

- **Establish Grant programs for improved Newborn and Child Screening for Heritable Disorders** to provide:
 - Education and outreach on newborn screening and coordinated follow-up care once newborn screening has been conducted
- Facilitate the creation of Federal guidelines on newborn screening.
- Assist State newborn screening programs in meeting Federal guidelines
- Reauthorize programs under Part A of Title XI of the Act

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NBSSLA: Authorization of Grant Program

1. Enhance, improve or expand the ability of **State and local public health agencies** to provide screening, counseling, or health care services to newborns and children having or at risk for heritable disorders.
2. Assist in providing **health care professionals and newborn screening laboratory personnel** with education in newborn screening and training in relevant and new technologies in newborn screening and congenital, genetic, and metabolic disorders.
3. Develop and deliver educational programs (**at appropriate literacy levels**) about newborn screening counseling, testing, follow-up, treatment, and specialty services to **parents, families, and patient advocacy and support groups**.
4. Establish, maintain, and operate a **system to assess and coordinate treatment** relating to congenital, genetic, and metabolic disorders.

NBSSLA: Provisions and Activities

(Responsible Agencies)

- Authorize grants/Cooperative Agreements (HRSA):
 - Services, infrastructure, and care coordination
 - **Regional Genetic and Newborn Screening Service Collaboratives**; current funding 2007-2012
 - Education Programs
 - Will expand with additional funding in 2010
- Authorize demonstration programs to evaluate the effectiveness of screening, counseling, or health care services in reducing the morbidity and mortality caused by heritable disorders in newborns and children (HRSA):
 - **Effective Follow-Up in Newborn Screening**: Electronic health information exchange
 - 3 Awards: start 9/1/09 (3 YRS)
 - New York
 - Utah
 - Indiana

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NBSSLA: Provisions and Activities

- Reauthorizes and expands the role of the Advisory Committee on Heritable Disorders in Newborns and Children (Committee) (HRSA)
- Creates a **Clearinghouse of Newborn and Child Screening Information for Heritable Disorders** (HRSA)
 - 1 Award - Genetic Alliance; start 10/1/09 (5 YRS)
 - Partnership:
 - Regional Collaboratives
 - National Newborn Screening and Genetics Resource Center / NNSGRC
 - APHL
 - Internet based and interactive forum
 - Information - Non-duplication
 - Supplement - building on existing infrastructure
 - Audience - expectant individuals/parents/consumers and providers

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NBSSLA: Provisions and Activities

- Ensures laboratory quality and surveillance involved in NBS activities (CDC); **CDC Proficiency Testing Program**
- **Creates Interagency Coordinating Committee (ICC)** on Newborn and Child Screening: Co-Directors **HRSA & CDC; FDA, NICHD**
- Creates National Newborn Screening Contingency Plan
 - The Contingency Plan is done by CDC in consultation with **HRSA, *Newborn Screening Concept of Operations Plan (CONPLAN)***
- Research in newborn screening, identify new screening technologies, and research disease management strategies for conditions detected through screening - **Hunter Kelly Newborn Screening Research Program (NIH)**.

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Prenatally and Postnatally Diagnosed Conditions Awareness Act, 2009

- **Provide information and support services to families receiving a positive test diagnosis for down syndrome, spina bifida, dwarfism, or other prenatally and postnatally diagnosed conditions.**
- To collect and disseminate current evidence-based information and to coordinate the provision of supportive services to parents who receive a positive diagnosis prenatally, at birth, or up to 1 year after the affected child's birth.
- To expand and further develop **national and local peer-support pro**
- Create a telephone hotline which would provide parents with information on the physical, developmental, educational, and psychosocial aspects of the condition.
- Provide awareness and education programs for the healthcare providers who provide, interpret, and inform parents of the results of positive test diagnoses for congenital disabilities.

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Congenital Conditions Program

- ***Congenital Conditions Program Translating Medical Genetics into Services***

- 1 Award – Genetic Alliance; start 9/1/09 (3 YRS)
 - Partnership
 - National Coalition for Health Professional Education in Genetics / NCHPEG
 - Regional Collaboratives
 - Poised to demonstrate the most partnership between families and providers of all grants

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RC Supplemental Funding

- Heartland RC

- Support a regional **Newborn Screening Data Integration Evaluation Project** by the participating states to improve health outcomes for newborns and children receiving long-term follow-up services in the Heartland region.

- Mountain States RC

- Support the expansion of the project, **Community Conversation on Genetics with Native American Peoples**, and include; a meeting to follow-up the First Community Conversation on Genetics with the **Navajo people**; and a Community Conversation on Genetics with a second Native American group.

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RC Supplemental Funding

■ New England RC

- Improve the quality of clinical genetic health care services for children with global developmental delays and intellectual disabilities seeking services in rural areas, by developing a “Quality Improvement Registry”, mapping data into the registry from clinics in four New England states, and creating a prototype model for other genetic conditions.

■ Region 4 RC

- Support the expansion of two projects: 1) Inborn Errors of Metabolism Information System (IEMI-IS); and 2) Midwest Emergency Medical System for Children Information System (MEMSCIS).

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RC Supplemental Funding

■ Western States RC

- Increase the knowledge about the screening, diagnosis, and clinical outcomes of children diagnosed with VLCADD (very long chain acyl-CoA dehydrogenase deficiency) by newborn screening.
- Complete the project to compile, analyze, and submit for peer-reviewed publication screening, diagnosis, and management data from:
 - California
 - Northwest NBS Program (Alaska, Hawai'i, Idaho, Oregon, Nevada, and New Mexico)
 - Washington
- Advance collaboration among the WSGSC newborn screening programs and clinicians providing treatment/management along the continuum of collaboration from the current state of “Networking” (Exchanging information for mutual benefit), to “Cooperation” (Exchanging information, modifying activities, sharing resources for mutual benefit to achieve a common purpose).

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Thank You

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