

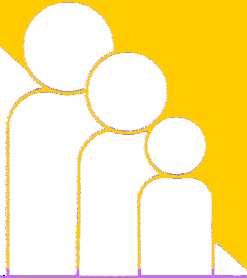
# **Regional Genetics and Newborn Screening Collaboratives**

## **January 2005**

Health Resources and Services Administration  
(HRSA)

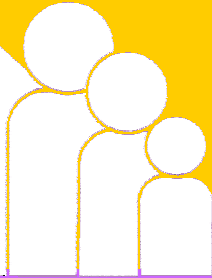
Maternal And Child Health Bureau (MCHB)

Michele A. Lloyd-Puryear, M.D., Ph.D.



## MCHB Strategic Plan Mission

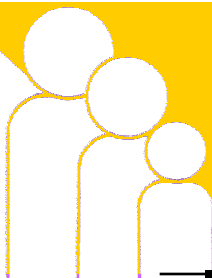
- “To provide national leadership and to work, in partnership with states, communities, public-private partners, and families to strengthen the MCH infrastructure, assure the availability and use of medical homes, and build the knowledge and human resources, in order to assure continued improvement in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



# The MCH Block Grant (Title V) “SPRANS” 501(a)(2)

- “To provide for SPRANS, research, and training for MCH and CSHCN, for **genetic disease testing, counseling, and information development and dissemination programs**, for grants (including funding for **comprehensive hemophilia diagnostic treatment centers**) relating to **hemophilia without regard to age**, and for the screening of newborns for sickle cell anemia, and other genetic disorders and follow-up services”

# CORE PUBLIC HEALTH SERVICES DELIVERED BY MCH AGENCIES



**MCH**



**DIRECT  
HEALTH CARE  
SERVICES  
(GAP FILLING)**

Examples:  
Basic Health Services and  
Health Services for CSHCN

**ENABLING SERVICES**

Examples:  
Transportation, Translation, Outreach, Respite Care,  
Health Education, Family Support Services, Purchase of  
Health Insurance, Case Management, Coordination with  
Medicaid, WIC and Education

**POPULATION--BASED SERVICES**

Examples:  
Newborn Screening, Lead Screening, Immunization, Sudden Infant Death  
Counseling, Oral Health, Injury Prevention, Nutrition and Outreach/Public Education

**INFRASTRUCTURE BUILDING SERVICES**

Examples:  
Needs Assessment, Evaluation, Planning, Policy Development, Coordination,  
Quality Assurance, Standards Development, Monitoring, Training, Applied Research,  
Systems of Care and Information Systems



# Current Environment

- Changing technology
  - Multiplex
- Rare Diseases
  - Few providers with required expertise
- New technology on the horizon



# Current Environment, continued

- Legislation for Heritable Disorders Program
  - 2000 enacted
  - 2003 Committee created
- Recommendations from the ACMG-Newborn Screening Expert Group



# Heritable Disorders Program

- In fiscal year 2004 appropriations language, Congress designated funds be applied to the implementation of the Heritable Disorders Program (HDP)



# Heritable Disorders Program

## Legislation

- **IN GENERAL.**—The Secretary shall award grants to eligible entities to 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.





# Heritable Disorders Program

## (b) USE OF FUNDS.—

- (1) **establish, expand, or improve systems or programs to provide screening, counseling, testing or specialty services** for newborns and children at risk for heritable disorders;
- (2) **establish, expand, or improve programs or services to reduce mortality or morbidity** from heritable disorders;
- (3) **establish, expand, or improve systems or programs to provide information and counseling on available therapies** for newborns and children with heritable disorders;



# Heritable Disorders Program

- (4) **improve the access of medically underserved populations** to screening, counseling, testing and specialty services for newborns and children having or at risk for heritable disorders; Or
- (5) **conduct such other activities as may be necessary to enable newborns and children having or at risk for heritable disorders to receive** screening, counseling, testing or specialty services, regardless of income, race, color, religion, sex, national origin, age, or disability.

# Regional Newborn Screening and Genetics Collaborative Groups

- 7 Regional Collaborative Groups; 1 national coordinating center
- To enhance and support the genetics and newborn screening capacity of States
  - Addresses maldistribution of genetics resources
  - Promotes translation of genetic medicine into public health and health care services
  - Shifts services into local communities



# Regional Collaborative Group

Enhance activities:

- Screening and follow-up services;
- Augment capacity needs: training, education;
- Strengthen Linkages to medical homes and tertiary care;
  - expansion of long term follow-up activities;
- Strengthen genetic counseling services; and
- Enhance communication/education to families and health practitioners and other forms of information sharing



# Regional Collaborative Group

- Serve as Regional Center for genetic and newborn screening services
  - counseling, testing, information dissemination, education and training
- Activities based on capacity needs of region
- Organizationally represent the collaborative partnerships between public health and clinical providers
  - CDC's Centers of Excellence for Birth Defects Prevention Research and NIH's Rare Diseases Clinical Research Network



# Regional Collaborative Group

- Year 1: Regional coordinating plan to address the maldistribution of genetic and newborn screening services and expertise
  - present practice models and materials needed for optimal diagnosis, follow-up and management of children identified with genetic conditions in the defined regions
- Year 2: Strategy to implement the practice model developed in Year 1 to provide genetic service/expertise and newborn screening expertise to the majority of rural and urban areas within the region



# Regional Collaborative Group

- Year 3: Demonstrate public-private regional and collaborative relationships that represent a variety of health systems organizations within all States in the defined region
  - community health centers,
  - health care insurers and health maintenance organizations,
  - state-based primary care organizations, and academic institutions.
  - State genetics and/or newborn screening Advisory Committees within the defined region

# Regional Newborn Screening and Genetics Collaborative Groups

- Region 1: Connecticut, Massachusetts, Maine, Rhode Island, Vermont and New Hampshire
- Region 2: District of Columbia, Maryland, Virginia, West Virginia, Pennsylvania, New York, New Jersey
- Region 3: Alabama, Mississippi, Georgia, Louisiana, North Carolina, Tennessee, Florida, South Carolina,, Puerto Rico, Virgin Islands
- Region 4: Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio, Wisconsin



# Regional Newborn Screening and Genetics Collaborative Groups

- Region 5: Arkansas, Iowa, Kansas, Missouri, North Dakota, Nebraska, Oklahoma, South Dakota
- Region 6: Arizona, Colorado, Montana, New Mexico, Texas, Utah, Wyoming
- Region 7: Alaska, California, Hawaii, Idaho, Nevada, Oregon, Washington, Pacific Basin

# Region 1

CT, MA, ME, NH, RI, VT

- New England Genetics and Newborn Screening Collaborative
  - Project Director: Thomas Brewster, MD
- Enhance collaboration
  - Web site
- Enhance and improve current practice models
  - Best practices models using an “equality of access” to NBS concept
  - Develop geographic and epidemiological approach to needs assessment
- Improve educational opportunities
  - Management guidance for Rx
  - Education Committee to develop programs to improve genetic literacy

# Region 2

DC, DE, MD, NJ, NY, PA, VA

- New York State DOH
  - Project Director: Kenneth Pass, PhD
- Regional Coordinating Plan
- Local solutions to barriers to access to specialty care for congenital abnormalities
  - (Teratogen hotline; Special regional projects)
- Emergency back-up system for NBS
- Standardize NBS throughout region
- Educate providers, payers, patients, and families

# Region 3

AL, FL, GA, LA, MS, NC, PR, SC, TN, USVI

- Southeastern Regional Genetics Group
  - Project Directors: David Ledbetter, PhD  
Jess Thoene, MD
- Identify existing gaps in genetics services
- Improve regional communication infrastructure
  - Telecommunications to link States' academic and public health representatives

# Region 4

IL, IN, KY, MI, MN, OH, WI

- Michigan Public Health Institute
  - Project Director: Cynthia Cameron, PhD

## Three Cluster Areas:

- MS/MS Project
  - Achieve uniformity of testing panel in region
- Reduce inequities in access to services
  - Telemedicine, long distance consultation, satellite clinics
- Public health infrastructure
  - Practice model for optimal diagnosis, follow up and management of children with heritable disorders and birth defects.

# Region 5

IA, KS, MO, NB, ND, OK, SD, AR

- Oklahoma Health Sciences Center
  - Project Director: John Mulvihill, MD
- Develop regional infrastructure
  - Communications
  - Education
  - Resource sharing
- Develop *Heartland Regional Genetics Strategic Plan*
- Special regional projects
- Identify gaps in service and education

# Region 6

AZ, CO, MT, NM, TX, UT, WY

- Mountain States Genetics Foundation
  - Project Director: Joyce Hooker
- Establish MoStGeNe Regional Center
- Update and regionalize existing needs assessment
- Develop regional plan for collaborative genetics activities

# Region 7

AK, CA, ID, HA, NV, OR, Guam, WA

- State of Hawai'i DOH
  - Project Directors: Sylvia Au, MS, CGC  
Kerry Sylvey, MA, CGC
- Plan, pilot, and evaluate a regional practice model
  - Improve access to specialty metabolic genetic services, primary care
- Needs assessment process to identify activities to increase the capacity genetics and newborn screening programs to perform their assessment, policy development, and assurance functions





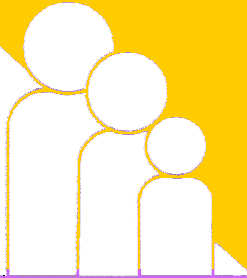
# National Coordinating Center (NCC): Genetics and NBS Regional Collaboratives

- American College of Medical Genetics
  - Project Director: Michael Watson, PhD
- Support the Regional Collaborative efforts to identify issues specific to utilization of genetic and newborn screening services at all levels



# National Coordinating Center (NCC): Genetics and NBS Regional Collaboratives

- Minimize duplication of efforts
- Identify “best practices” developed by regions
- Further information exchange and professional collaboration
- Maximize interregional collaboration
  - Language and terminology compatibility



# Plans for NCC

- Develop networks of centers of genetic services (COGs) and primary care provider
- Facilitate data collection collaborating with:
  - NIH rare disease centers
  - CDC genomics centers
- Involve national programs to which American College of Medical Genetics, AAP, and AAFP link
  - CPT code development
  - JCAHO role in hospital activities related to newborn screening
- Information sharing between projects with overlapping interests



# Areas of Shared Need

- Videoconferencing
- Telemedicine
- Genetics networks
- Interstate satellites
- Management information
- Web-based clinical management system suitable to telemedicine
- Reimbursement (e.g. for telephone consultation)



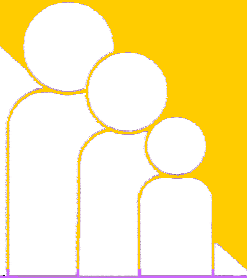
# Areas of Shared Need

- Evaluation
- Expansion of newborn screening
- Legislation and regulation
  - Interstate licensing
  - liability
- Financing
  - Matching funding mechanisms to regional needs
- Expanding access to genetic services
  - Training of geneticists and primary care providers
  - Increase diversity of trainees
  - Geographical Maldistribution



# Coordinating Center Partners

- National Conference of State Legislators (NCSL)
- American Academy of Pediatrics (AAP)
- Association for State and Territorial Health Officials (ASTHO)
- Genetic Alliance



# Contact Information

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