

Genetic Services Branch

Who we are and what we do

December 10, 2008

Genetic Services
Agency and National Leadership
Advisory Committee on Heritable Disorders

Translational Genetics

Resources and Services-ongoing

- Regional Collaboratives

Education-ongoing)

- CGEN
- Genetic Resources
- Family History Tool

Applied Research-ongoing

Parental Attitudes

Training-completed

- GPC
- GENE Tools
- NBS education tools

NBS Infrastructure and Services

National Newborn Screening
and Genetic Resource Center

Implementation NBS Task Force
Report

- NBS Information Infrastructure
Development
 - Practice Network

Training/Education

- MS-MS training

Evaluation and Applied Research

- System Monitoring

Screening Initiatives

Vision, etc.

Genetic Services and Health
Care Delivery Systems

Congressional Programs

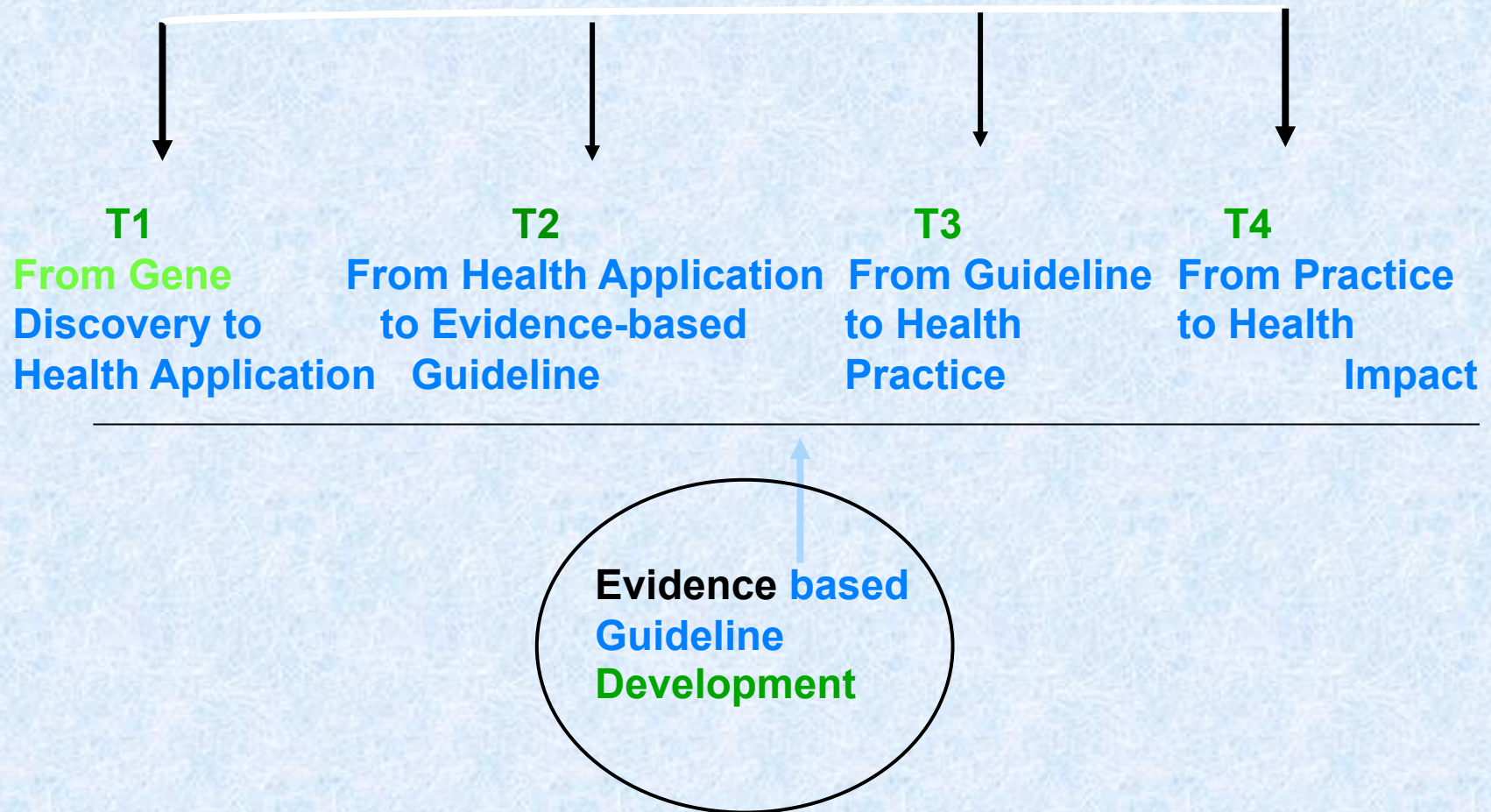
- National Hemophilia Program
- Sickle cell Disease NBS Program
- Thalassemia Comprehensive Care Program

Delivering Genetic Services

- Clinical Paradigm
(subspecialist/specialist/families/patient)
- Financing Treatment and Management

Applied Research

The Four Phases of Translation For Genomic Applications in Population Health



Shifting Focus:

From Single Gene Conditions
To Common, Complex Conditions



Human Genome Project



FUNCTIONAL GENOMICS



Genetic Information and Technology

FUNCTIONAL GENOMICS

Population-based research to understand gene-environment interactions

– But, how will we --

- Engage large numbers in this effort?
- Involvement a diverse population?
- Assure equal access to this effort?

– Thus far --

- Little involvement of broad population
- No fiscal commitment

What does it have to do with us?

What role will public health agencies play in translating the Human Genome Project into resources and services to improve health?



Communication Arts, Jackson MS

Agency and National Leadership

- **Title XXVI of the Children's Health Care Act of 2000**
 - Advisory Committee on Heritable Disorders
- **Partnerships with National Organizations**
 - March of Dimes, ACMG, AAP, NCSL, Genetic Alliance ACOG, SIMD etc

Agency and National Leadership

- Title XXVI of the Children's Health Care Act of 2000
 - Establishes a program to improve the ability of States to provide newborn and child screening for heritable disorders.
 - PHS Act: Section 1109
 - PHS Act: Section 1110
 - **PHS Act: Section 1111- Advisory Committee on Heritable Disorders**

Advisory Committee on Heritable Disorders-Section 1111

Provide advice and recommendations to the Secretary **concerning Section 1109** which provides funds for grants to establish or expand or improve:

-Systems or Programs:

- a. for genetic services
- b. for services to reduce mortality and morbidity
- c. to provide information and counseling on available therapies

-Access of medically underserved populations to genetic services

-Other activities as may be necessary to enable infants and children to receive genetic services.

Advisory Committee on Heritable Disorders-Section 1111

Provide technical information to the Secretary for the development of policies and priorities for the administration of grants under Section 1109 of the PHS Act; and

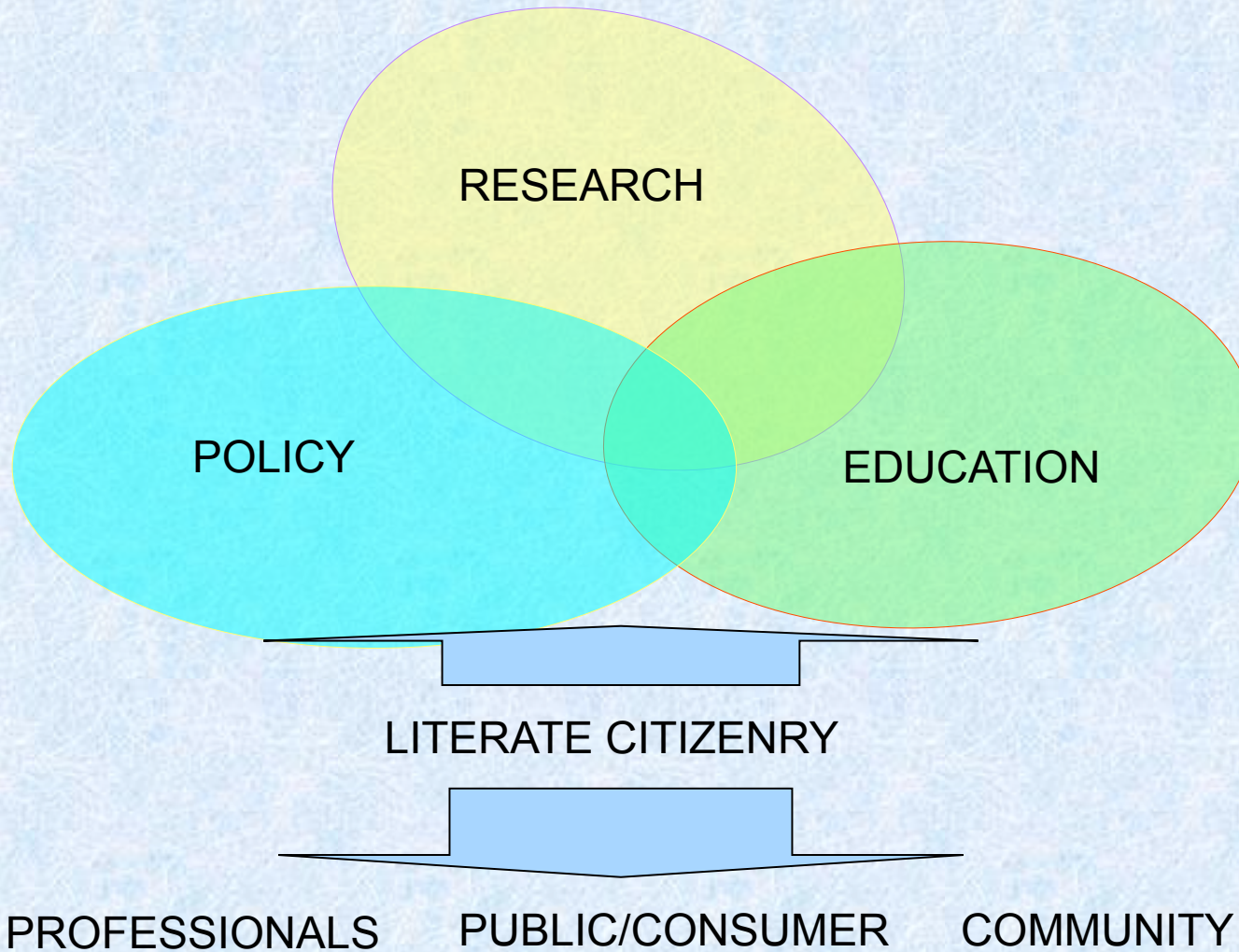
Provide such recommendations, advice or information as may be necessary to enhance, expand or improve the ability of the Secretary to reduce the mortality or morbidity in newborns and children from heritable disorders.

Translational Genetics

Translational Genetics

Applying fundamental science and technology discoveries to advances in medical genetics resources, services and systems

Translational Genetics



Translational Genetics

- **Education**
 - National Consumer Center for Genetic Information
 - Family History Tool
 - CGEN
 - Genetics
- **Training (completed)**
 - GPC
 - GENE Tools
 - NBS education tools
- **Capacity**
 - Regional Collaboratives
 - **Applied Research**
 - Parental Attitudes regarding false positive screening

Goal 1

- **Clarify genetic resources, services, technology and policy issues for a variety of stakeholders**

Objectives

– Fiscal years 2008-2012:

- Build upon current grant works to focus on areas of identified needs for families, health and public health professionals and communities including family health history and access to genetic services

Current Fiscal Year

- Expansion of Regional Collaboratives initiatives: emergency preparedness, telegenetics outreach, genetics education for primary care providers and linkage to medical home
- Study the efficacy of community based family health history tool in 10 communities
- Dissemination of genetics education products in 4 communities

Goal 2

- **Prepare a literate genetics citizen**

Objectives

- **Increase involvement of families, communities, health and public health providers access to and use of genetic resources and services**
- **Provide education and training opportunities for consumers and health and public health profession.**

Goal 3

- **Integrate genetics research with a broader mandate.**

Objectives

- **Infuse translational genetics expertise into MCHB programs**
- **Provide context of genetic for other maternal and child health populations.**
 - Fiscal year 2008-2012
 - Regional Genetics and Newborn Screening Collaboratives
 - LEND program addition of genetics into program
 - Perinatal Programs use of family health history

FY 2008-2009 Initiatives

- Family History as an Educational Tool
 - utilize a consumer-based family history tool to increase awareness about genetics
- Regional Genetics and Newborn Screening Collaboratives
 - enhance and support the genetics and newborn screening capacity of states within defined regions
- Advisory Committee
 - Subcommittee activities: Long term follow-up, food s and nutrition survey, collaboration with professional organizations for educational foci

FY 2008-2009 Initiatives

- Efficacy projects regarding parental attitudes
 - Iowa
 - Hawaii
 - Maryland and Georgia
- National Consumer Information Center

Future Initiatives

- Through Cooperative Agreements, we will **continue to explore the issues of the maldistribution of all aspects of genetic service capacity, including many of the policy and education and training issues** that will surround us. These issues include such areas as: family health history, emergency preparedness, education of providers and consumers, utilization of telegenetics/telehealth options.

Newborn Screening and Health Systems Infrastructure Program

Newborn Screening and Health Systems Infrastructure Program

Facilitate the development of **health care and public health infrastructure** to enhance and expand newborn screening programs and to improve linkages among them and the state and community systems of care for CSHCN.

Principle

Effective newborn screening (NBS) systems are available for all children in the United States

Reference: Serving the Family From Birth to the Medical Home – A Report From the Newborn Screening Task Force Convened in Washington DC, May 10-11, 1999

Resource Center

- National Newborn Screening and Genetics Resource Center -

<http://genes-r-us@uthscsa.edu>

Serves as a focal point for national newborn screening and related genetics activities, collects and shares information and resources to benefit consumers, health professionals, researchers, the public health community, and government officials.

Goal 1

Support a framework for effective partnerships between parents and professionals and among professions, agencies, and officials at the Federal, State, and community levels and between the public and private sector.

Objective

- Promote the linkage of NBS programs to medical homes and family support networks.
 - *AAP medical home initiative*
 - *Sickle Cell Disease and Newborn Screening Program (ref: Congressional appropriation language)*
 - *Regional Newborn Screening and Genetic Services Collaboratives*

Objective

- Strengthen network of specialty-subspecialty health professionals to provide an adequate system of follow-up, diagnosis, referral, and management.
 - *Regional Newborn Screening and Genetic Services Collaboratives*

Goal 2

Strengthen existing public health infrastructure and allow it to be integrated with the health care delivery system.

Objective

- Support State and Territorial efforts to coordinate activities among different programs and integrate child related public health agency information to allow improved coordination, tracking, assessment, and evaluation.
 - Support an integrated child health information system Community of Practice (Connections)
 - NNSGRC national data system on State and Territorial NBS activities.

Objective

- Ensure information among the various groups, including medical homes, is expeditiously and appropriately shared.
 - Public Health Informatics Institute project to analyze newborn dried blood spot screening business processes, including long term follow-up
 - Public Health Informatics Institute project to develop HL-7 implementation guide for newborn screening (allow electronic transmission of NBS results)
 - Regional Newborn Screening and Genetic Services Collaboratives

Objective

- Assist States in their efforts to monitor and evaluate system performance.
 - *Implement program evaluation and assessment scheme for quality assessment of State NBS programs*
 - *NNSGRC conducts expert review of State NBS programs.*

Objective

- Support pre-screening and screening education and training initiatives.
 - *Distribution of NBS educational materials for obstetrical providers.*
 - *Distribution of NBS educational materials for parents, taking into consideration ethnicity, culture, and literacy concerns.*

Objective

- Support State implementation of technological innovations.
 - *NNSGRC, in collaboration with the Association of Public Health Laboratories, convenes training workshops for State laboratory and follow-up staff.*

Goal 3

Provide ongoing leadership and support for the development of NBS standards, guidelines, and policies.

Objective

- Support the development of models, strategies, and materials for implementation of effective NBS systems.
 - With Translational Genetics Program, facilitate through *Regional Genetics and Newborn Screening Collaboratives*
 - *NNSGRC activities*

Genetic Services and Health Care Delivery Programs

Genetic Services and Health Care Delivery

Support the Hemophilia Diagnostic and Treatment Centers and Thalassemia and Sickle Cell Disease programs as **models of comprehensive care** for the delivery of genetic services: testing, counseling, education and coordinated system of services.

Congressional Mandates

- **Hemophilia**-Provide access to hemophilia diagnostic and treatment centers to provide comprehensive care through adulthood
 - **340B Drug Pricing Program**-Provide access to factor at a discounted price and pass the savings on to the consumers or back into services
- **Sickle Cell Disease and Newborn Screening**-Enhance the sickle cell disease and carrier follow-up services of State newborn screening programs with community-based efforts that provide hemoglobinopathy counseling, education, referrals to primary care providers and comprehensive SCD treatment centers. Funding for a National Coordinating & Evaluation Center

Congressional Mandates

- **Sickle Cell Disease Treatment Demonstration Program**- the development and establishment of infrastructure and partnerships to enhance the prevention and treatment of sickle cell disease. Fund a National Coordinating Center
- **Thalassemia**- Support implementation of demonstration grants regarding a model system of comprehensive care and medical management for individuals and families at risk for or affected by Cooley's Anemia/Thalassemia-tertiary care

Goals

- Manage GSB's genetic disease-specific programs according to their legislative mandates

In Process

- Paper on emerging practices regarding SCD & NBS
- Hemoglobinopathy counselor certification program
- Tool to educate parents of children with SCD about disease symptom and management and how to navigate the health care system