

Maternal and Child Health Bureau

**MCH/CSHCN Director June 2004 Webcast**

**June 10, 2004**

Health Resources and Services Administration  
Maternal and Child Health Bureau

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Maternal and Child Health Bureau

Welcome  
**Peter C. van Dyck, MD, MPH**



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Maternal and Child Health Bureau

Moderator  
**Chris DeGraw, MD, MPH**



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Maternal and Child Health Bureau

**Michele A. Lloyd-Puryear, MD, PhD**




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**MCHB's Genetic Services Program**

Health Resources and Services Administration

Mission:

- Health promotion through the early identification of individuals with or at risk for heritable disorders,
- Development of genetic services that are comprehensive, accessible, family-centered and culturally competent,
- Understanding of the genetic contribution to health and disease upon which services are developed.

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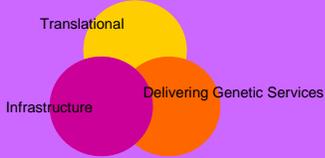
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**Genetic Service Program**



Agency and National Leadership

Translational

Infrastructure

Delivering Genetic Services

June 2004

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## Present Initiatives

Health Resources and Services Administration

- Education
- Training
- Capacity
  - Infrastructure
  - Workforce
- Research
- Public Policy

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## Program Aims

Health Resources and Services Administration

- Clarify resource and policy issues for a variety of stakeholders
- Develop education and training opportunities in genetics for consumers and health and public health professionals
- Integrate and translate genetics across state and community based genetic services and consumer and health provider initiatives

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## Program Goals

Health Resources and Services Administration

1. Facilitate the development of **public health and health care infrastructure** to enhance and expand newborn screening programs and to improve linkages among them and the state and community systems of care for CSHCN.
2. Examine emerging issues and evaluate emerging technologies in genetics with a special emphasis on the **financial, ethical, legal and social implications** of these issues/technologies for MCH populations.

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Health Resources and Services Administration

## Program Goals

3. Improve the **genetic literacy** of the MCH population by enhancing its understanding of the benefits, risks, limitations, and implications of genetic testing and the role of genetic information in improving health practices.
4. Provide leadership in defining the **educational needs** in genetics of health professionals working with the MCH population.

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Health Resources and Services Administration

## Program Goals

5. 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.
6. Build on the expertise gained from HRSA genetics activities to provide **national leadership** on expanding and enhancing genetics services for the entire population.

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Health Resources and Services Administration

## Current Newborn and Genetic Service Activities

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**HRSA's Vision for Newborn Screening**

Health Resources and Services Administration

- Systems approach with defined public health roles at state and national level
- Presence of Quality assurance
- Public-private partnerships for assurance of systems approach and comprehensive, efficient care and management
- Equity for families

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**HRSA Contract With ACMG National Newborn Screening Guidance**

Health Resources and Services Administration

Expert panel convened to review available information on newborn screening (NBS) based upon accumulation and analysis of best scientific evidence:

1. To address model policies and procedures and minimum standards for state NBS programs.

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**HRSA Contract With ACMG National Newborn Screening Guidance**

Health Resources and Services Administration

2. To create a model decision matrix for changing newborn screening panels.
3. To develop a uniform panel of conditions for screening.

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Understanding, Informing and Educating Parents About Newborn Screening

Health Resources and Services Administration

- Analysis of:
  - State Statutes/Regulations/policies regarding consent for newborn screening including recommendations for a state resource tool kit.
  - State Statutes/Regulations/policies regarding storage and use of residual blood spots following newborn screening including recommendations for a state resource tool kit.
- Develop sample newborn screening educational toolkit:
  - Analyze content and suitability of one set of prime educational materials from 50 states.
    - Prepare draft content for an educational program for parents on newborn screening

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Understanding, Informing and Educating Parents About Newborn Screening

Health Resources and Services Administration

- Convene listening groups to evaluate one or more options for an educational process for parents on newborn screening.
- Develop a preliminary toolkit
- Conduct Pilot Testing

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Understanding, Informing and Educating Parents About Newborn Screening

Health Resources and Services Administration

- Develop educational materials for Prenatal providers for educating parents
  - Partnership with ACOG, AAFP
  - Target: health professionals with the primary responsibility for prenatal health care, labor and delivery services (obstetricians, family practice physicians and nurse midwives)

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## Heritable Disorders

- Title XXVI of the Children's Health Care Act of 2000 (Title XI of PHS act)
  - 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**

U.S. Department of Health and Human Services  
  
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Health Resources and Services Administration

## Heritable Disorders

**Section 1109** provides funds for grants to establish or expand or improve:

- Systems or Programs:
  - for genetic services
  - for services to reduce mortality and morbidity
  - 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.

U.S. Department of Health and Human Services  
  
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## Advisory Committee on Heritable Disorders

- **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.

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## Advisory Committee on Heritable Disorders

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- Medical, technical, or scientific professionals with special expertise in heritable disorders, or in providing screening, counseling, testing or specialty services for newborns and children at risk for heritable disorders;
- Members of the public having special expertise about or concern with heritable disorders; and
- Representatives from such Federal agencies, public health constituencies, and medical professional societies as determined to be necessary by the Secretary, to fulfill the duties of the Advisory Committee

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## Committee Members

Health Resources and Services Administration

**William J. Becker, D.O., M.P.H.**  
Bureau of Public Health Laboratories  
Ohio Department of Health

**Amy Brower, Ph.D.**  
Third Wave Technologies

**Peter B. Coggins, Ph.D.**  
PerkinElmer Life and Analytical Sciences

**Gregory A. Hawkins, Ph.D.**  
Department of Internal Medicine  
Wake Forest University School of Medicine

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## Committee Members

Health Resources and Services Administration

**R. Rodney Howell, M.D.**  
**(Chairperson)**  
The University of Miami School of Medicine  
Department of Pediatrics (D820)

**Piero Rinaldo, M.D., Ph.D.**  
Mayo Clinic

**Derek Robertson, M.B.A., J.D.**

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**Committee Members**

**Liaison Members**  
**James W. Collins, Jr., M.D., M.P.H.**  
 Advisory Committee on Infant Mortality

**E. Stephen Edwards, M.D., F.A.A.P.**  
 American Academy of Pediatrics  
 President

**Jennifer L. Howse, Ph.D.**  
 March of Dimes Birth Defects Foundation

**Reed Vaughn Tuckson, M.D.**  
 Advisory Committee on Genetics, Health and Society

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**Committee Members**

**Ex-officio Members**  
**Duane Alexander, M.D.**  
 National Institutes of Health

**Coleen Boyle, Ph.D.**  
 Centers for Disease Control and Prevention

**Denise Dougherty, Ph.D.**  
 Agency for Healthcare Research and Quality

**Peter C. van Dyck, M.D., M.P.H.**  
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**Heritable Disorders Program**

**Regional Genetic Service and Newborn Screening Collaboratives**

- Enhance and support the genetics and newborn screening capacity of States within defined regions
- These projects will undertake a regional approach toward addressing the maldistribution of genetic resources
- 7 regions

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## Heritable Disorders Program

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- Willingness to serve as a regional center
- Collaborative and regional approach toward facilitating access to the genetics expertise, services and technology that providers and families need to diagnose and manage children identified with genetic disorders
- Infrastructure of public-private regional and collaborative partnerships to provide the **genetic, newborn screening** and other relevant subspecialty expertise and services

U.S. Department of Health and Human Services  


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**NNSGRC Website:**  
<http://genes-t-us.uthscsa.edu>

*(Genetics and Newborn Screening Resource Center of the U.S.)*

U.S. Department of Health and Human Services  


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## Newborn Screening: Current Challenges and Opportunities

Brad Therrell, Ph.D.  
 University of Texas Health Science Center at San Antonio  
 and  
 Newborn Screening and Genetics Resource Center  
 Austin, Texas



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## Newborn Screening

The term is used to refer to two programs that may or may not have linkages:

1. Traditional biochemical screening for inherited conditions (metabolic, endocrine, hematological, etc.)
2. Screening for congenital hearing loss

In this presentation, 'newborn screening' will usually refer to the traditional heelstick biochemical testing program.



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## Newborn (Heelstick) Screening Multiple Ways of Defining:

An essential public health program that prevents catastrophic health consequences through early detection, diagnosis and treatment.

A complex system of testing, evaluation and treatment that is dependent upon the dedication of persons working within the system.

A public health prevention strategy that, when designed properly, reaches all newborns rapidly and effectively.



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## Newborn Screening Is:

A Public Health Prevention System that includes

- Primary Health Care Professionals
- Laboratory Personnel
- Administrative and Follow-up Personnel
- Specialty Care Centers
- Source(s) of Payment
- Family Members
- Policy Makers
- Manufacturers
- Other Interested Persons or groups (e.g. Legislators)



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## Newborn Screening Refresher

- Began in the 1960s
- Newborn screening was initially available in public health and hospital laboratories, and funding was usually included in state budgets
- State legislators gradually encouraged funding from other sources, and testing became centralized
- All states now have a law that can be cited as a basis for newborn screening
- There is wide variation in the way newborn screening occurs from state to state.



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## Newborn Screening Refresher

- Newborn screening is a public health "SYSTEM"
  1. Education
  2. Screening
  3. Follow-up
  4. Diagnosis
  5. Treatment/Management
  6. Evaluation
- The "SYSTEM" must function seamlessly and be responsive to politics, culture, and economics
- Functioning of the "SYSTEM" must be central to all considerations about changes
- Multiple models exist regarding service provision



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## Newborn Screening

Given the history of newborn screening since 1960, what does the typical system look like today and how do the components mesh?



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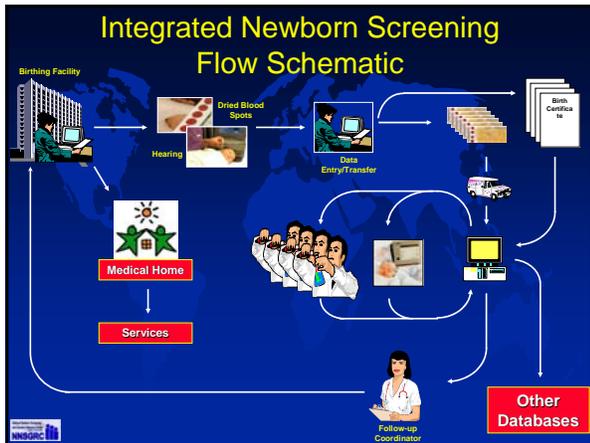
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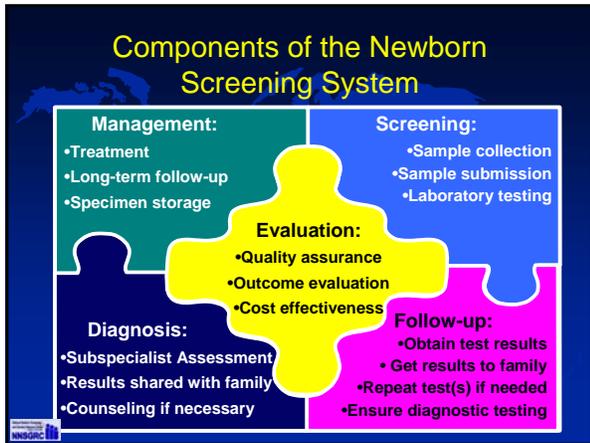
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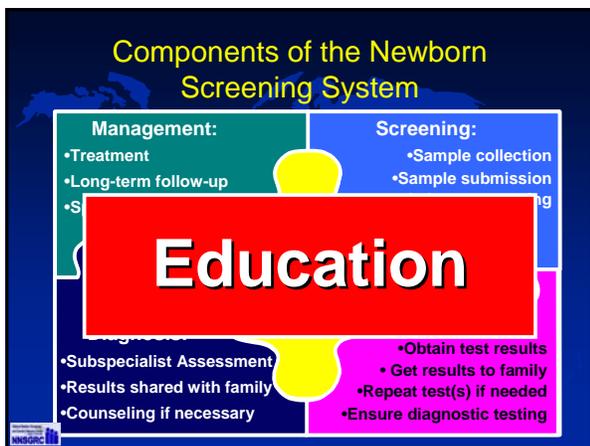
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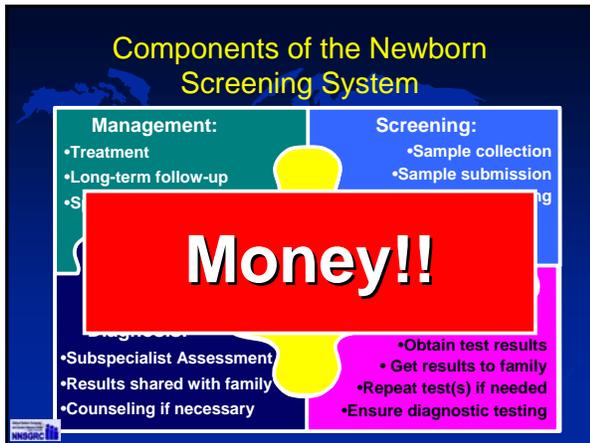
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### Newborn Screening

*Current Congressional interests are focused on differences in programs within the states.*

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### U.S. Senate Committee on Health, Education, Labor and Pensions Subcommittee on Children and Families







14/06/2002

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**GAO Report on Newborn Screening Programs prepared at the request of Sen. Dodd (CT) and Sen. DeWine (OH)**

U.S. Government Accounting Office  
March 2003

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**Some of the Newborn Screening Program Differences**

- 51 States (incl. D.C.) have laws allowing or mandating NBS.
- 3 Programs require consent for NBS (MD, WY, DC). Some do not allow dissent for any reason.
- 8 Programs mandate 2 screens (>90% comply) and several others strongly suggest (>80% comply) 2<sup>nd</sup> screen.
- 8 Programs do not charge a fee but for others fees exist up to \$70 (excluding hospital administrative costs).
- The amount of Medicaid reimbursement varies widely and there are about 1/3 of all births are Medicaid.
- The storage time and protocols for accessing and using residual blood that remains after testing varies widely.

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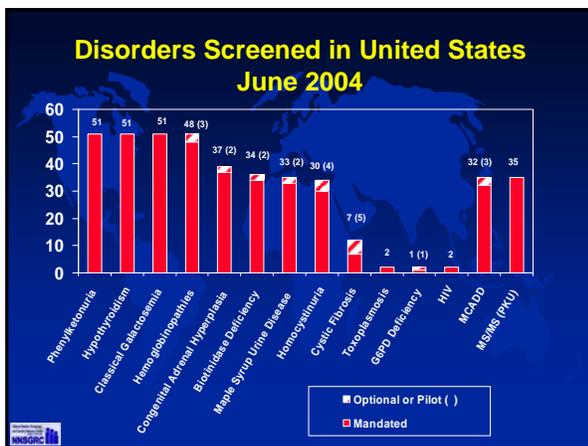
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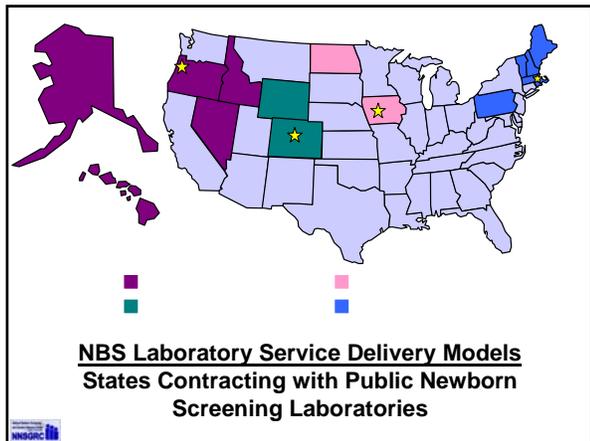
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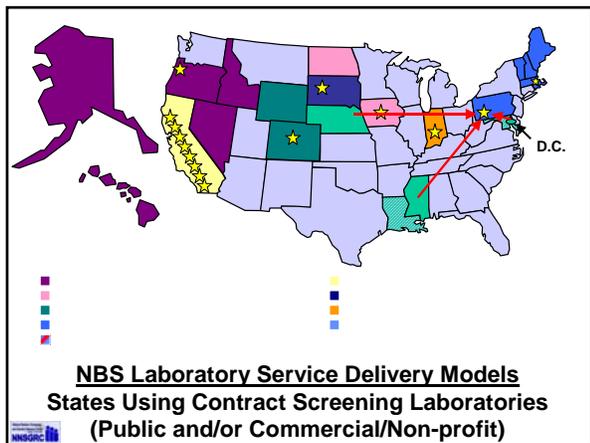
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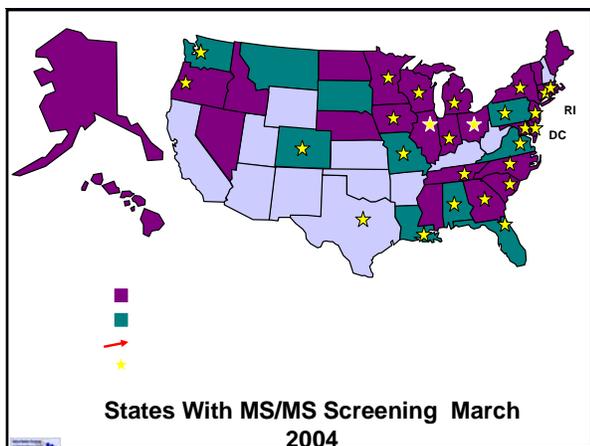
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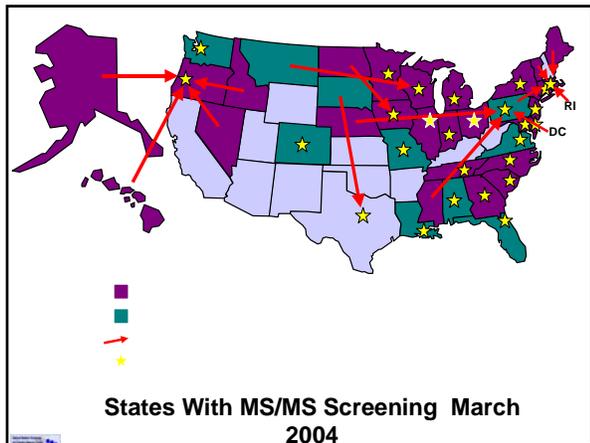
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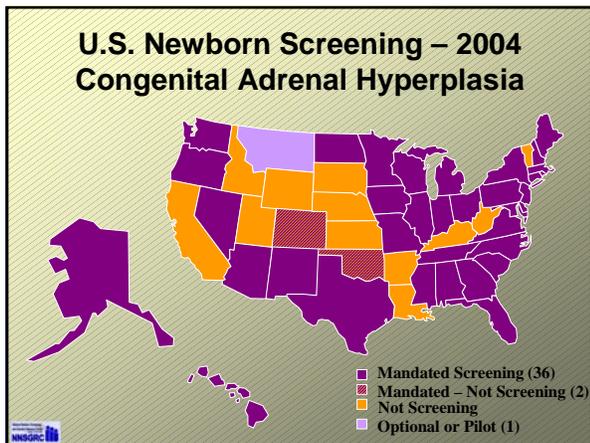
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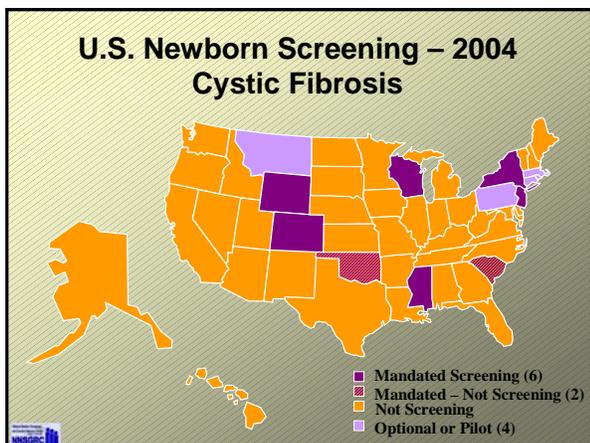
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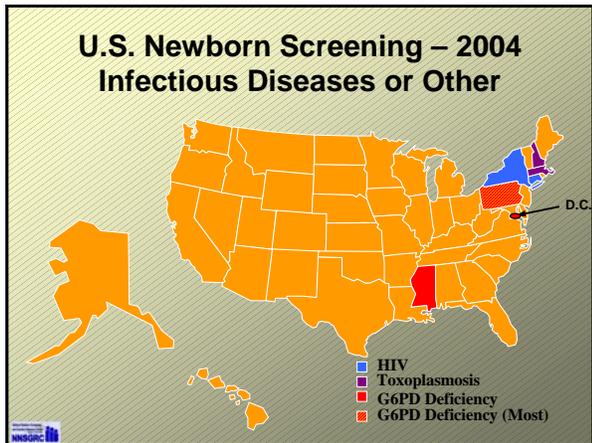
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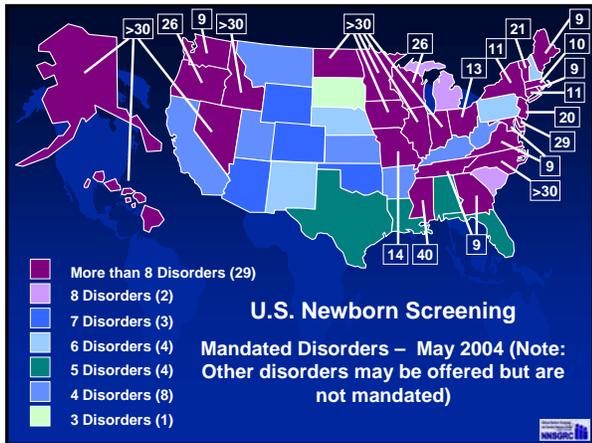
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### U.S. Newborn Screening Data 1990-1999\*

DISORDER	SCREENED	CASES	INCIDENCE
Sickle Cell Diseases**	28,149,621	12,976	1:2,170
Primary Hypothyroidism	40,214,946	13,213	1:3,044
S,S Disease	28,149,621	7,565	1:3,721
Cystic Fibrosis	1,459,834	372	1:3,924
S,C Disease	28,149,621	3,811	1:7,386
Toxoplasmosis	989,402	95	1:10,415
Classical PKU	40,028,546	2,098	1:19,079

\* Still in process of validation    \*\*Includes SS-Disease, SC-Disease, Thalassemias

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**U.S. Newborn Screening Data 1990-1999\***

DISORDER	SCREENED	CASES	INCIDENCE
Classical PKU <small>(from previous page)</small>	40,028,546	2,098	1:19,079
PKU Variant Clin. Sig.	40,028,546	772	1:51,850
Clin. Sig. Hyperphe	40,028,546	2,870	1:13,947
Classical CAH	13,347,888	703	1:18,987
Classical Galactosemia	35,897,634	674	1:53,261
Biotinidase	12,754,403	208	1:61,319
MSUD	13,801,657	60	1:230,028
Homocystinuria	12,027,751	35	1:343,650

\* Still in process of validation

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## Newborn Screening

*One of the big questions today –  
How are decisions made and is the  
process appropriate (modern)?*

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## Who Decides About Newborn Screening in the U.S. and How?

Who?

- ◆ State Legislatures  
*Every State has a law mandating screening –  
sometimes specifying disorder(s) and  
laboratories*
- ◆ State Health Officers
- ◆ State Boards of Health
- ◆ Advisory Committees (All but 2 have  
standing advisory committees)

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## Who Decides About Newborn Screening in the U.S. and How?

How?



- ◆ Public Interest
- ◆ Professional Interest
- ◆ Political Interest
- ◆ Cost Savings (Benefits Outweigh Costs)
- ◆ Scientific Evidence (Incidence/Outcome)

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## Traditional Criteria for Newborn Screening

Wilson and Jungner – WHO, 1968

10 Criteria for Population Screening

1. Important health problem
2. Accepted treatment
3. Diagnosis and treatment facilities available
4. Recognizable latent or early symptomatic state
5. Suitable test or examination
6. Test is acceptable to the population
7. Natural disease history adequately understood
8. Agreed policy on whom to treat as patients
9. The cost balanced relative to possible expense for medical care
10. Case finding is a continuous process

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## Latest Major Attempt to Consider National Issues

### Newborn Screening Task Force (1999)

- ◆ Convened in by: American Academy of Pediatrics (AAP)  
 Funding Support – Maternal and Child Health Bureau (MCHB)  
 [Health Resources and Services Administration (HRSA)]
- ◆ Chairs: Edward McCabe and Thomas Tonniges
- ◆ Co-sponsors:
  - National Institutes of Health (NIH)
  - Centers for Disease Control and Prevention (CDC)
  - Agency for Health Care Policy & Research (AHCPR)
  - Genetic Alliance (Alliance)
  - Association of State & Territorial Health Officials (ASTHO)
  - Association of Maternal & Child Health Programs (AMCHP)
  - Association of Public Health Laboratories (APHL)

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## Newborn Screening Task Force Report

- ◆ Vol. 106, Aug. 2000, Suppl.
- ◆ Approved by:
  - AAP Board of Directors
  - AAP Committee on Genetics
  - AAP Committee on Fetus and Newborn
  - Medical Home Initiatives for Children with Special Needs-Project Advisory Committee
  - AAP Task Force on Newborn and Infant Hearing




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## AAP Task Force Report Based on 5 Major Principles Concerning Newborn Screening

1. Infants should benefit from and be protected by NBS programs
2. Public health agencies should assume responsibility for oversight of NBS systems
3. Standards and guidelines for NBS should be more consistently applied
4. Greater uniformity would benefit families, professionals, and public health agencies
5. NBS systems should link to a medical home




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## AAP Task Force Report Key Recommendation Areas

1. Public Health Infrastructure
2. Public and Professional Involvement
3. Surveillance and Research
4. Financing




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## AAP Task Force - Agenda for Action

"Public health, partnering with health professionals & consumers, should continue a process that:"

1. Defines responsibilities – federal and state
2. Models regulations for NBS systems
3. Defines minimum standards for NBS
4. Models guidelines and protocols for professionals
5. Models systems of care from infancy to adulthood
6. Designs strategies to inform and involve families and the general public
7. Funds demonstration projects to evaluate technology, quality assurance, and health outcomes



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## MOD Current Recommendation 9 Biochemical Tests + Hearing Screening

"We believe that a test (even for a rare disease) as long its early discovery makes a difference to the child must be conducted for every newborn."

From Howse J, Katz M. Pediatrics 2000;106:595

1. Phenylketonuria
2. Congenital Hypothyroidism
3. Congenital Adrenal Hyperplasia
4. Sickle Cell Diseases
5. Galactosemia
6. Biotinidase Deficiency
7. Homocystinuria
8. Maple Syrup Urine Disease
9. Congenital Hearing Loss
10. Medium Chain Acyl-CoA Dehydrogenase Deficiency  
(Added after the initial press release)



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## Congressional Interest in NBS

[Has encouraged activities by HRSA, NIH, CDC AHRQ.]

- ◆ "...the Committee urges the availability and accessibility of newborn screening services to apply public health recommendations for expansion of effective strategies."
- ◆ "HRSA, in collaboration with the CDC and the NIH, is encouraged to implement a strategy for evaluating and expanding newborn screening programs, pilot demonstration projects, and the use of contemporary public health recommendations on specific conditions, such as cystic fibrosis and the fragile X syndrome."
- ◆ "... the Committee directs that tangible steps be taken to protect patient privacy and to avert discrimination based upon information derived from screenings."



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## Newborn Screening

*In addition to the contracts and grants currently being implemented, HRSA continues to work through the NNSGRC to provide NBS program support.*



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## NNSGRC Training Support for NBS

Cooperation between NNSGRC (HRSA) and APHL (CDC) for Laboratory and Follow-up Training Courses in Tandem Mass Spectrometry Methodologies.

1-week intensive course on the basics of tandem mass spectrometry methods and interpretations. Courses were initiated at Duke and the Institute for Metabolic Disease (Dallas) to provide an interim solution to training gaps resulting from rapid implementation of expanded screening in many states.



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## Training Courses in Tandem Mass Spectrometry Methodologies



Duke Medical School  
Durham



Institute for Metabolic Disease  
Baylor (Dallas)



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## NNSGRC Consultative Reviews

Cooperation between NNSGRC (HRSA) State Health Departments to evaluate and improve the newborn screening program at the state level.

A valuable external review system using experts in laboratory, follow-up, administration, quality assurance and medicine to address specific program needs at the request and invitation of a public health screening program.



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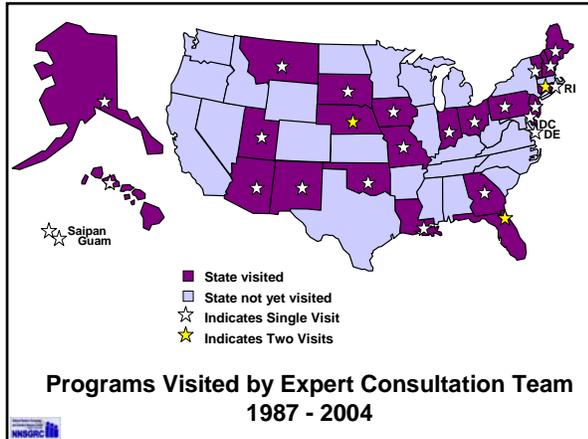
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## NNSGRC Consultative Newborn Screening Reviews



Missouri Review



Louisiana Review



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## Other Federal Interest and Support

Continued Support of a National Proficiency Testing Program for Newborn Screening Laboratories by the CDC.

An essential service providing external proficiency testing specimens for newborn screening laboratories and kit manufacturers around the world.



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## Newborn Screening Quality Assurance Program

- Services provided:
  - Filter paper QC
  - Reference materials
  - QC materials
  - Proficiency testing
  - Consultation and network resource support
- Partners
  - Association of Public Health Laboratories
  - ~ 64 domestic screening laboratories
  - > 350 laboratories in 53 countries



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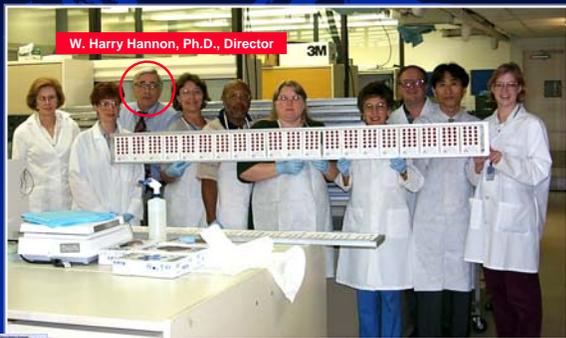
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## CDC NSQAP Production Team



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### Summary

- Newborn Screening Works – Approximately 4,000 are detected annually with one of the conditions being screened.
- There are many program differences across the nation.
- More than 1,000 newborns with detectable conditions go undetected because they are not screened for all conditions currently available.
- There is Federal and State interest and support for improving programs to provide equity between programs.
- A national screening mandate does not currently exist and would require likely require funding support if enacted.
- There is continuing national interest in expanding newborn screening programs.



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### On line genetics and newborn screening program information and assistance:

NNSGRC Website:

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

*(Genetics and Newborn Screening Resource Center of the U.S.)*

Various NBS Listserv memberships available upon request



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# Thank You



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