

The World of Newborn Screening

Dallas, Texas - June 24, 2006



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Newborn Screening and Genetics Resource Center

Austin, Texas



NNSGRC Cooperative Agreement

Purpose

To establish and maintain a National Center to serve as a resource for:

- ◆ *refining state newborn screening programs,*
- ◆ *enhancing state capacity to incorporate new developments in genetics and disease prevention into public health, and*
- ◆ *serving as a contact center for consumers, public health professionals and government officials.*



NNSGRC INFRASTRUCTURE

MCHB
Project Officer

NBS Technical
Consultation Team

NNSGRC Office – Austin

Project Advice
Committees

Genetics Technical
Consultation Team

Executive Director
[Genetics Program Director]
Administrative Assistant
Program Coordinators (1.5)

Various
Subcontracts

Executive Advisory Committee
Includes 3 Center Administrators,
2 HRSA Advisors, 1 CDC Advisor
1 Consumer Liaison

On-Line newborn screening program information and assistance:

Genetics and Newborn Screening
Resource Center of the U.S.

NNSGRC Website:

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

National Newborn Screening Information System (NNSIS)



National Newborn Screening Information System

[Home](#)[NNSGRC Home](#)[Help on NNSIS](#)[NNSGRC Staff Only](#)[Contact NNSGRC](#)[Logoff](#)

Welcome to the National Newborn Screening Information System (NNSIS) database.

The database is hosted by the **National Newborn Screening and Genetics Resource Center (NNSGRC)** and is designed to provide a secure, Internet based, real-time, information collection and reporting system for capturing state and territorial newborn screening information.

The system uses existing reporting requirements specified in the former National Newborn Screening Annual Report.

-
- » **Obtain a Report**
 - » **Enter Individual or Annual Case Information**
 - » **Contact NNSGRC**
-



Welcome to our Newborn Screening Data System

Newborn Screening is
MORE than a blood test!



Newborn Screening Is:

A System that includes

- *Medical Practitioners (Doctors, Nurses, Mid-wives)*
- *Laboratory Personnel*
- *Administrative and Follow-up Personnel*
- *Specialty Care Providers*
- *Source(s) of Payment*
- *Family Members*
- *Other Interested People (e.g. Politicians)*

Newborn Screening Is:

A System that:

- *Usually begins with an interested individual (or group of individuals) who cares about improving health.*
- *Evolves in individual cultural, economic and political environments.*
- *Is successful because of the efforts of dedicated newborn screening staff members and concerned professionals.*

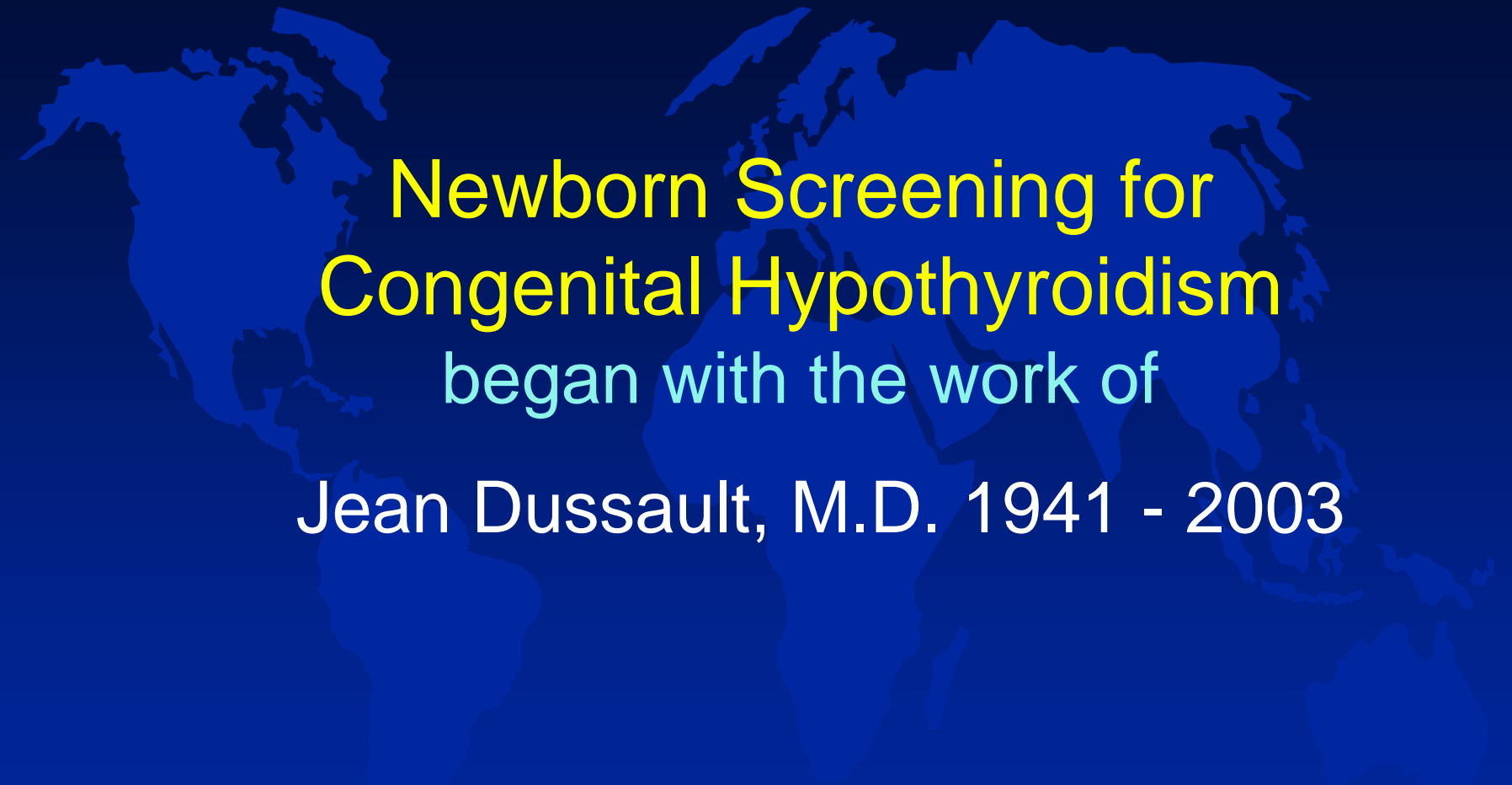
Newborn Screening

*Began in the 1960s with the work of
Bob Guthrie (1916-1995)*

Brief Review: Newborn Screening History

Early 1960s

1. Guthrie developed filter paper test for PKU (Identified newborns with PKU whose diet could be modified thus preventing mental retardation.)
2. Parents pressed for testing through organized lobbying.
3. Legislatures began to pass mandatory newborn screening laws to reduce institutionalization.

A dark blue silhouette of a world map is centered in the background of the slide.

Newborn Screening for
Congenital Hypothyroidism
began with the work of
Jean Dussault, M.D. 1941 - 2003

Brief Newborn Screening History In the United States

1970s

- Other filter paper tests became available – CH, CAH, SS, etc.
- Technology improved allowing program expansion.
- Programs expanded to higher incidence disorders – congenital hypothyroidism – and disorders that result in death – CAH, GAL.
- Legislatures began asking programs to become self-supporting.

Brief Newborn Screening History In the U.S.

1980s

- Programs became computerized.
- Expansion continued, including DNA studies.

1990s

- DNA tests used as second tier – Sickle Cell Disease screening, Cystic Fibrosis screening.
- Tandem mass spectrometry developed allowing simultaneous detection of multiple disorders.

Brief Newborn Screening History In the U.S.

2000s

- Newborn hearing screening
- Public pressure to expand testing with MS/MS
- Emphasis on program integration (especially data)
- Privacy concerns – residual blood spot, federal HIPAA rules (data sharing)

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

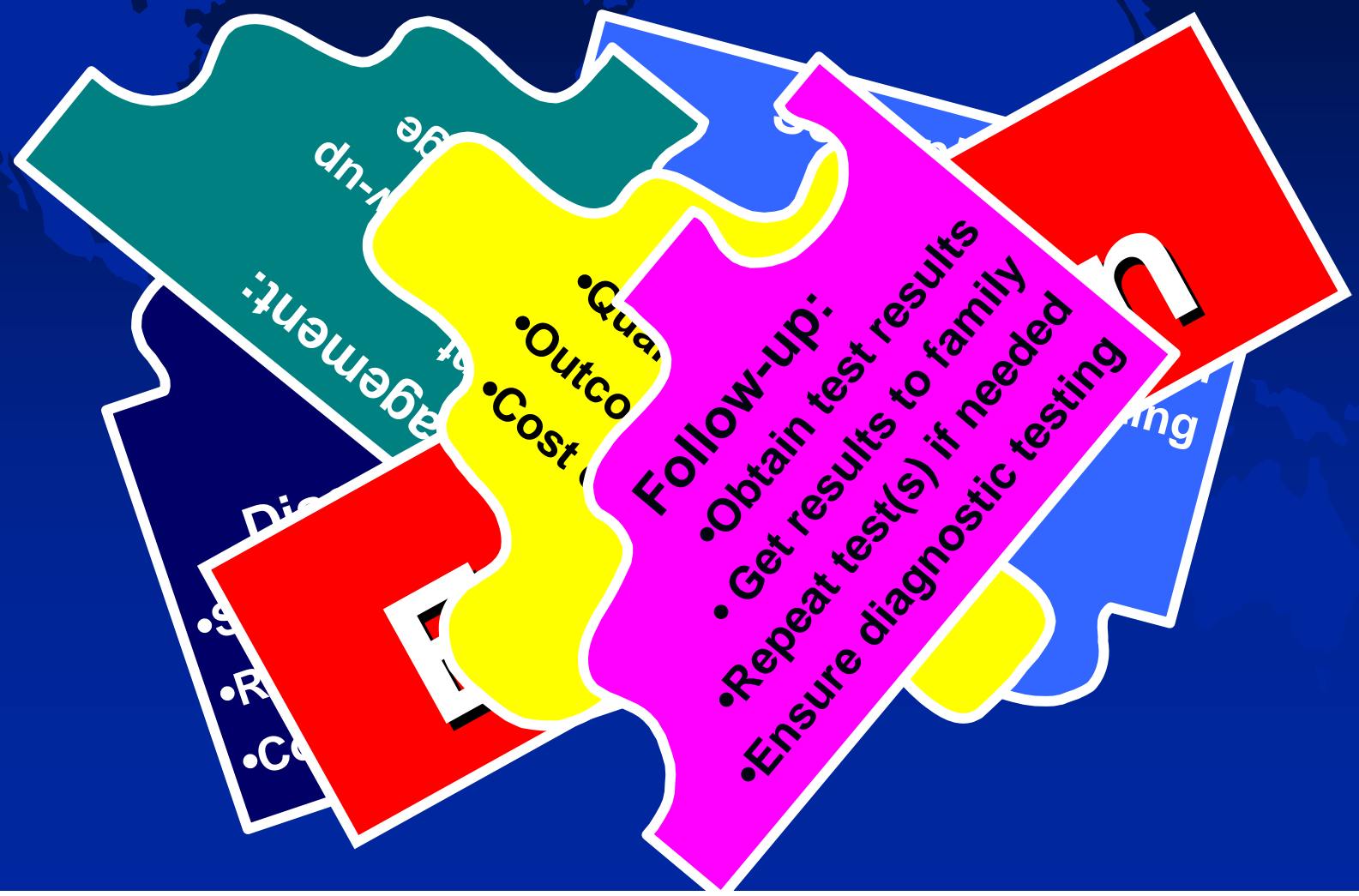
- ◆ Advisory Committees (All but 2 have standing advisory committees)

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)

Components of the Newborn Screening System



Components of the Newborn Screening System

Management:

- Treatment
- Long-term follow-up
- Specimen storage

Screening:

- Sample collection
- Sample submission
- Laboratory testing

Evaluation:

- Quality assurance
- Outcome evaluation
- Cost effectiveness

Diagnosis:

- Subspecialist Assessment
- Results shared with family
- Counseling if necessary

Follow-up:

- Obtain test results
- Get results to family
- Repeat test(s) if needed
- Ensure diagnostic testing

Components of the Newborn Screening System

Management:

- Treatment
- Long-term follow-up
- S

Screening:

- Sample collection
- Sample submission
- ng

Education

- Subspecialist Assessment
- Results shared with family
- Counseling if necessary

- Obtain test results
- Get results to family
- Repeat test(s) if needed
- Ensure diagnostic testing

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

Scoring Criteria

HRSA Contract

National Policy Development for NBS Test Selection

American College of
Medical Genetics



- Incidence
- Difficulty of diagnosis (birth)
- Disease impact
- Test sensitivity/specificity
- Test characteristics
- Treatment availability & cost
- Treatment efficacy
- Benefits to individual
- Benefits to family & society
- Mortality prevention
- Diagnosis availability
- Management availability
- Simplicity of therapy

HRSA Contract National Policy Development for NBS Test Selection

American College of
Medical Genetics

Expected Completion
June 2004

CRITERIA INCLUDED IN THIS SURVEY	CATEGORIES	SCORE
Incidence of condition	>1:5,000	100
	>1:25,000	75
	>1:50,000	50
	>1:75,000	25
	<1:100,000	0
Phenotype clinically identifiable at birth	Never	100
	<25% of cases	75
	<50% of cases	50
	<75% of cases	25
	Always	0
Burden of disease if untreated	Profound	100
	Severe	75
	Moderate	50
	Mild	25
	Minimal	0
Does a sensitive <u>AND</u> specific screening test currently exist?	YES	200
	NO	0
Test characteristics (Yes = apply score; No = zero)	Doable in neonatal blood spots OR by a simple, in-nursery physical method	50
	High throughput (>200/day/FTE)	20
	Cost (supplies + equipment) per test <1\$	20
	Multiple markers in same analysis	20
	Detection of secondary targets	20
	Utilizes a multiplex platform	20
Availability of treatment (Expensive IF >\$50,000/patient/year)	Inexpensive and widely available	100
	Expensive OR limited availability	50
	Expensive AND limited availability	0
Potential efficacy of existing treatment	To prevent ALL negative consequences	200
	To prevent MOST negative consequences	100
	To prevent SOME negative consequences	50
	Treatment efficacy not proven	0
Benefits of early intervention (INDIVIDUAL OUTCOME)	Clear scientific evidence that intervention IN THE FIRST WEEKS OF LIFE maximizes outcome	100
	Early intervention improves outcome	50
	No evidence of improved outcome	0
Benefits of early intervention (FAMILY & SOCIETY)	Early intervention maximizes benefits (education, understanding prevalence and natural history, cost effectiveness)	100
	Early intervention improves benefits	50
	No evidence of benefits	0
Early diagnosis and treatment prevent mortality	YES	100
	NO	0
Diagnostic confirmation	Widely available	100
	Reduced availability	50
	Available only in a few laboratories	0
Clinical management	Widely available	100
	Reduced availability	50
	Available only in a few centers	0
Simplicity of therapy	Very high	200
	High	100
	Average	50
	Low	0

ACMG
Report on
Newborn
Screening
May 2006

Genetics
IN
Medicine®

Official Journal of the American College of Medical Genetics



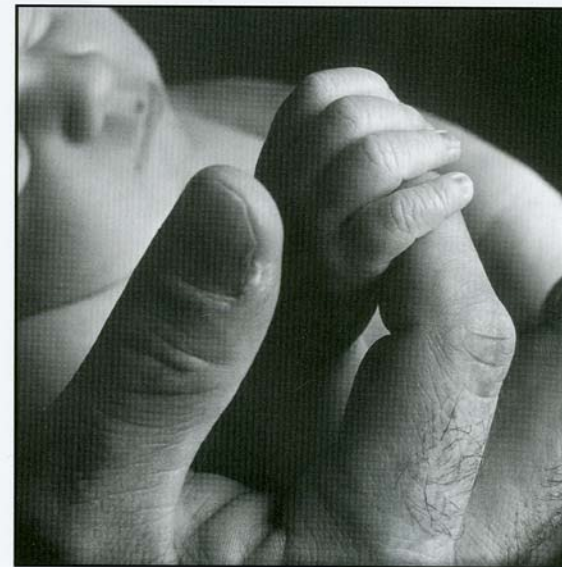
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www.geneticsinmedicine.org

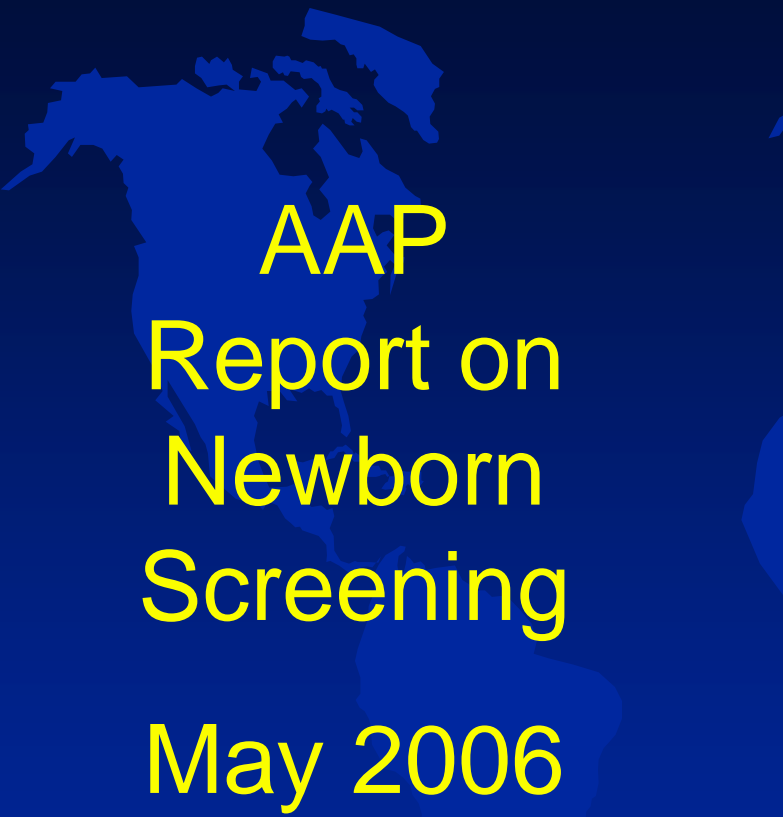
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Newborn Screening: Toward a Uniform
Screening Panel and System

- Executive summary
- Main report



AAP
Report on
Newborn
Screening
May 2006

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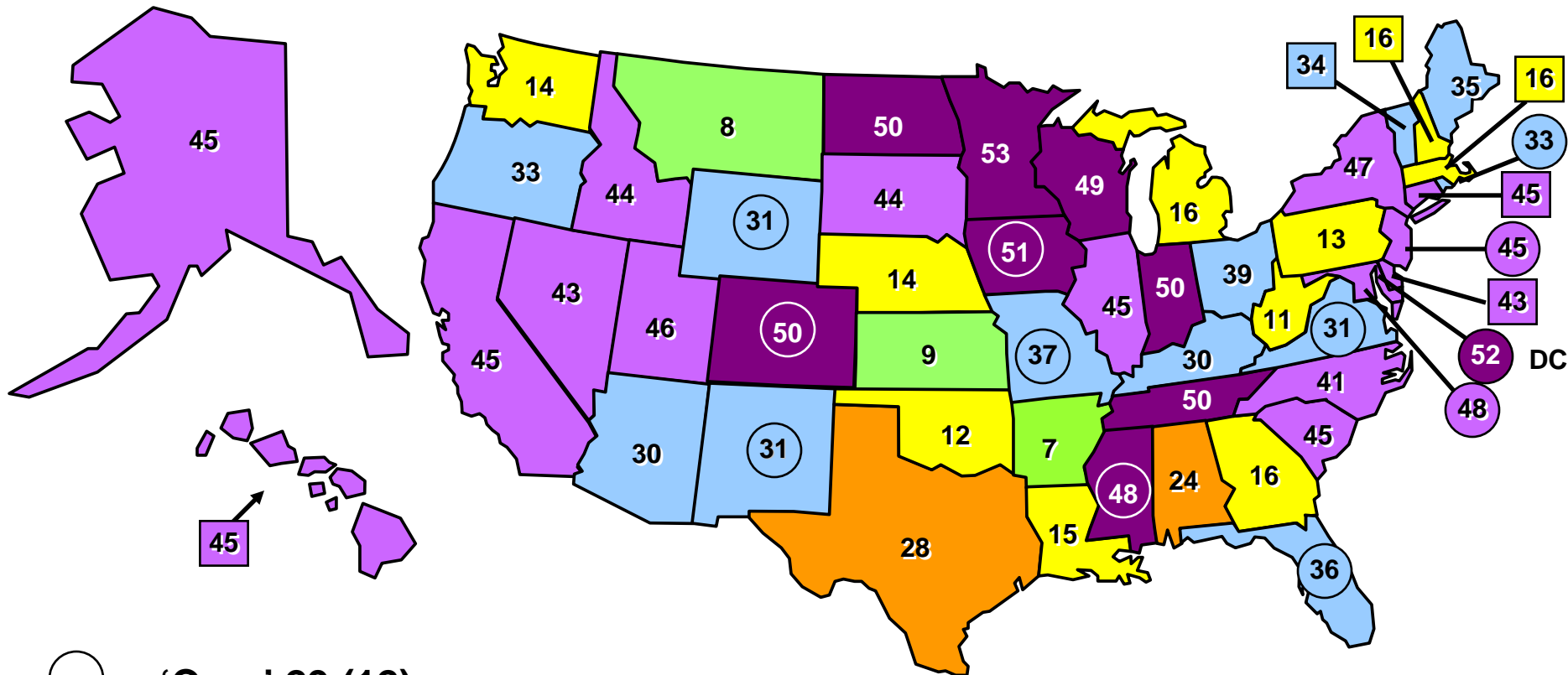
www.pediatrics.org

A SUPPLEMENT TO PEDIATRICS

A Look at Newborn Screening: Today
and Tomorrow

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Administration*

American Academy of Pediatrics 
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- 'Core' 29 (12)
- 50+ Disorders (9)
- 40-49 Disorders (15)
- 30-39 Disorders (12)
- 20-29 Disorders (2)
- 10-19 Disorders (10)
- <10 Disorders (3)

U.S. Newborn Screening

Conditions Required – June 1, 2006

(Conditions available as an option to selected population are not counted)



Thank You!

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

<http://www2.uthscsa.edu/nnsis/>

<http://www.marchofdimes.com/peristats/>