Newborn Screening: Current Status of State Newborn Screening Programs

Newborn Screen Positive Infant ACTion Project
Learning Session 2
February 12, 2011

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University of Texas Health Science Center at San Antonio
Newborn Screening and Genetics Resource Center
Austin, Texas

I have no relevant financial relationships with the manufacturers of any commercial products and/or provider of commercial services discussed in this CME activity. I do not intend to discuss an unapproved/investigative use of a commercial product/device in this presentation

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Newborn Screening is MORE than a hearing test!

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Newborn Screening is a SYSTEM!

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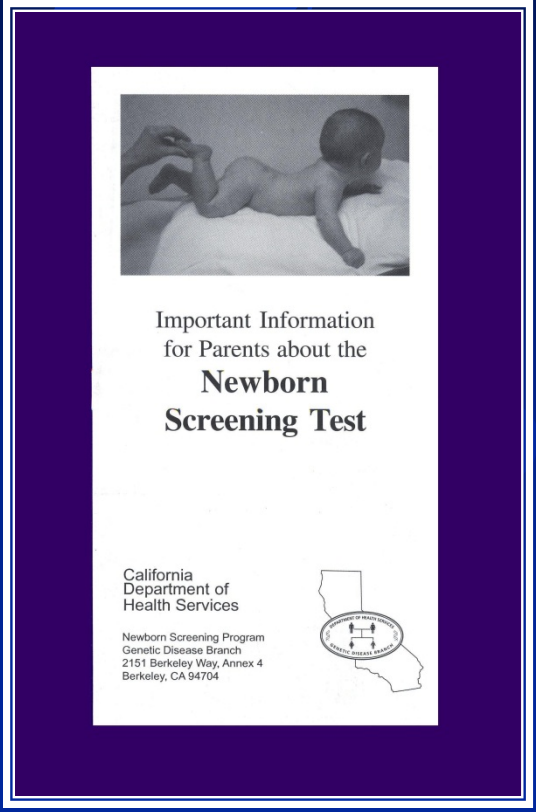
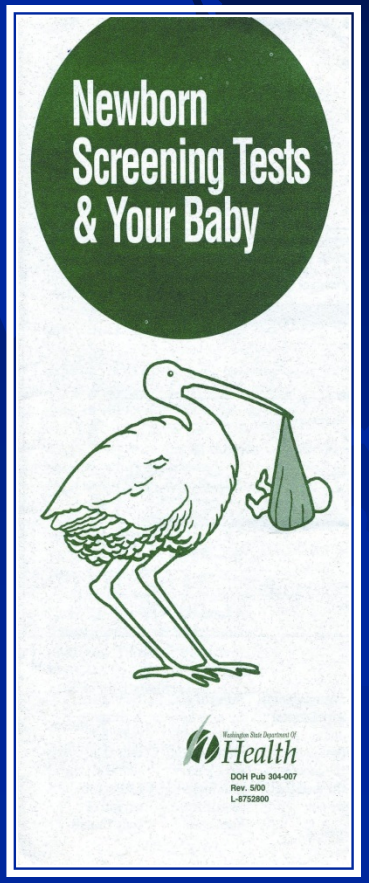
Newborn Screening Education for Parents





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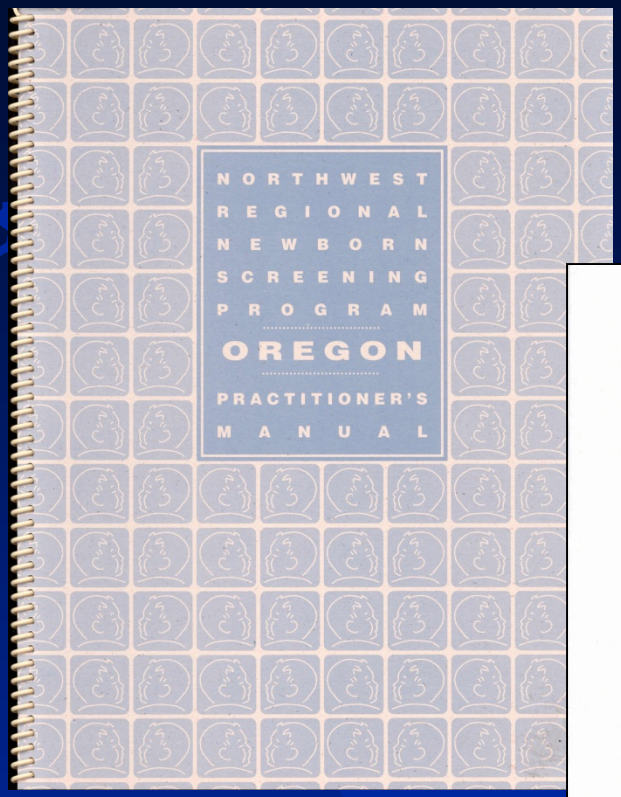
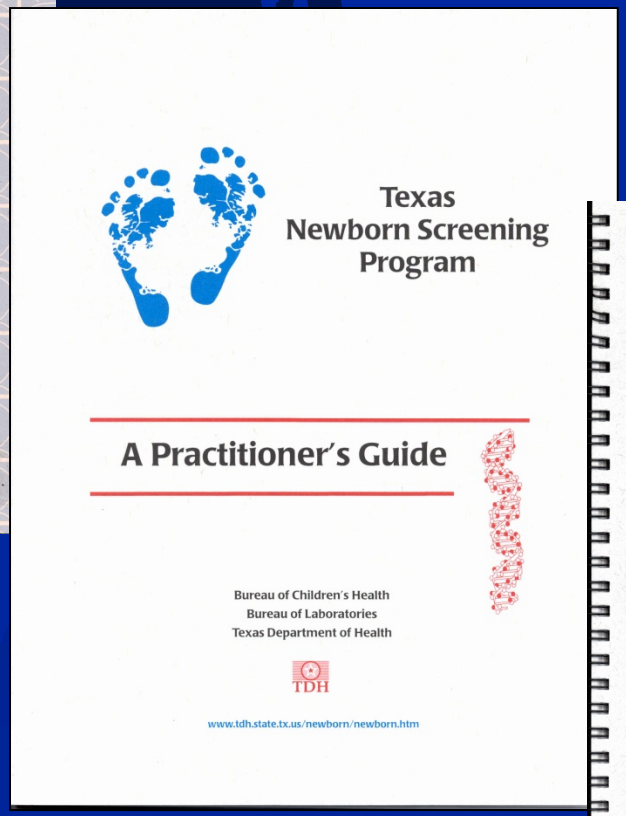
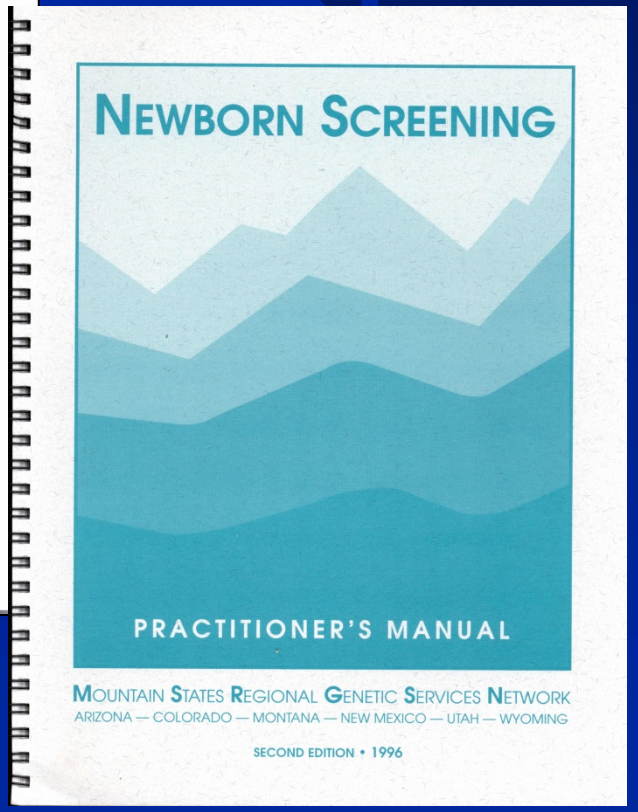
Practitioner Manuals







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CLSI Newborn Screening Collection

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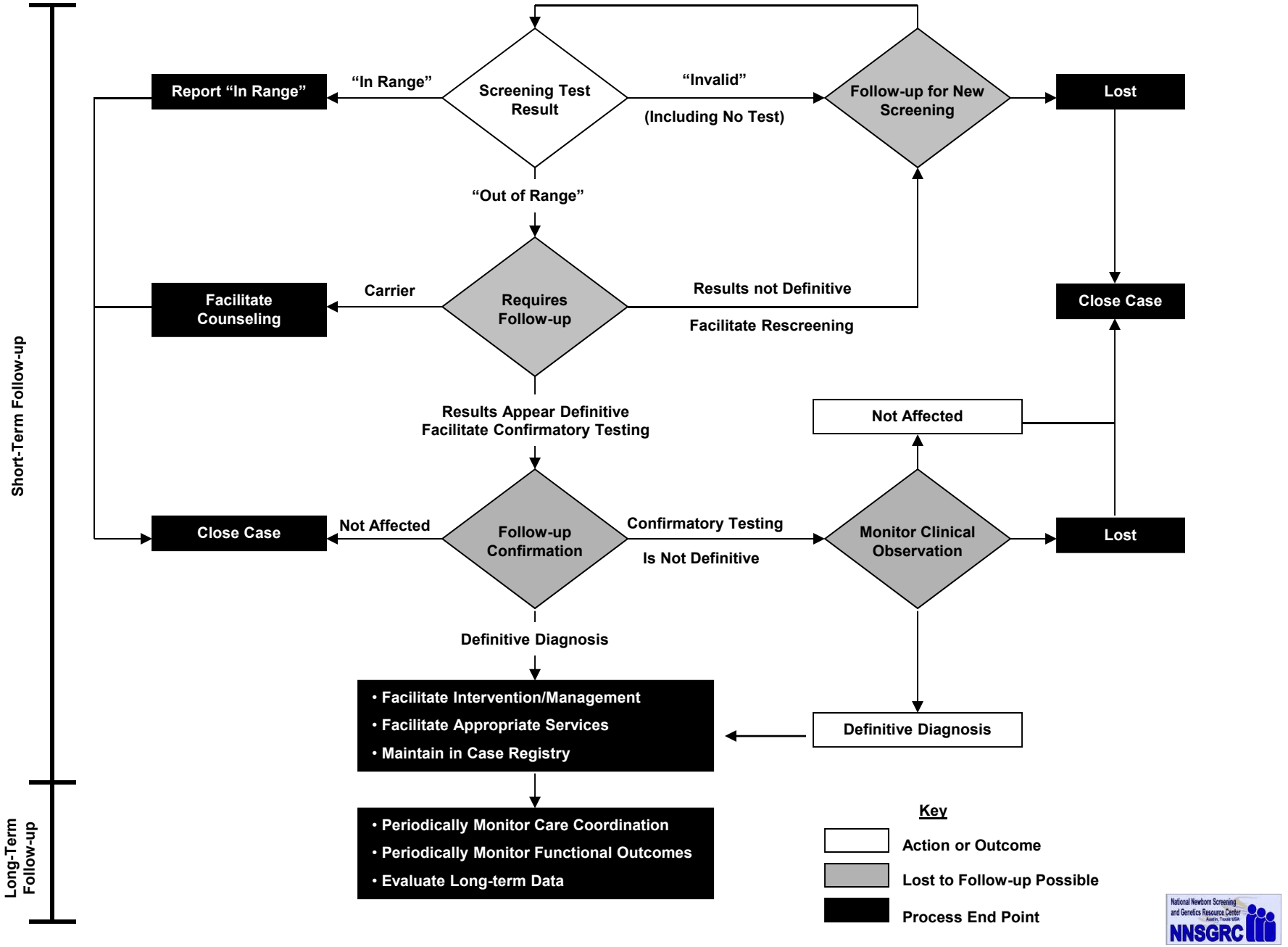
The Newborn Screening Follow-up Process (CLSI I/LA 27-A)



Role of the Obstetrician

Clinical Obstetrics and Gynecology
45:697-710 (2002).

CLINICAL OBSTETRICS AND GYNECOLOGY
Volume 45, Number 3, 697–710
© 2002, Lippincott Williams & Wilkins, Inc.
DOI: 10.1097/01.GRF.0000016420.79751.55

Newborn Screening: The Role of the Obstetrician

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The history of newborn screening is relatively short, spanning only 40 years since the pioneering work by Guthrie.[1] It was he who first realized that a blood sample could be taken from a newborn, absorbed and dried onto standardized filter paper, transported to a central testing laboratory, and then analyzed for biochemical indicators of inborn disorders of metabolism such as phenylketonuria (PKU). Because inborn metabolic errors are relatively rare (PKU in U.S. populations was shown to be about 1:15,000 at the time), it took the efforts of parents lobbying in behalf of the health of their newborns to convince health policy makers of the value of this type of population screening.[2] In 1965, the American Academy of Pediatrics Committee on the Fetus and Newborn finally recommended a newborn screening blood test for PKU for all newborns "no sooner than 24 hours after onset of milk feeding and previous to discharge."[3] Within a few years, most states in the United States and many other countries in the industrialized world were performing newborn screening for these "rare" disorders. As screening developed, it was inevitable that automated testing and data handling systems would evolve, and that new procedures would improve disease detectibility in newborns. Indeed, by the end of the 1970s, automated sample preparation and improved testing sensitivity and specificity had led to expansion of many screening programs to include dried blood spot screening for congenital hypothyroidism[4,5] (worldwide incidence of about 1:3,500, except in iodine-deficient areas, where it is much more prevalent). Today newborn screening is perhaps the best example of a successful and ongoing preventive public health population-screening program.

Definitions of newborn screening have traditionally been limited to biochemical

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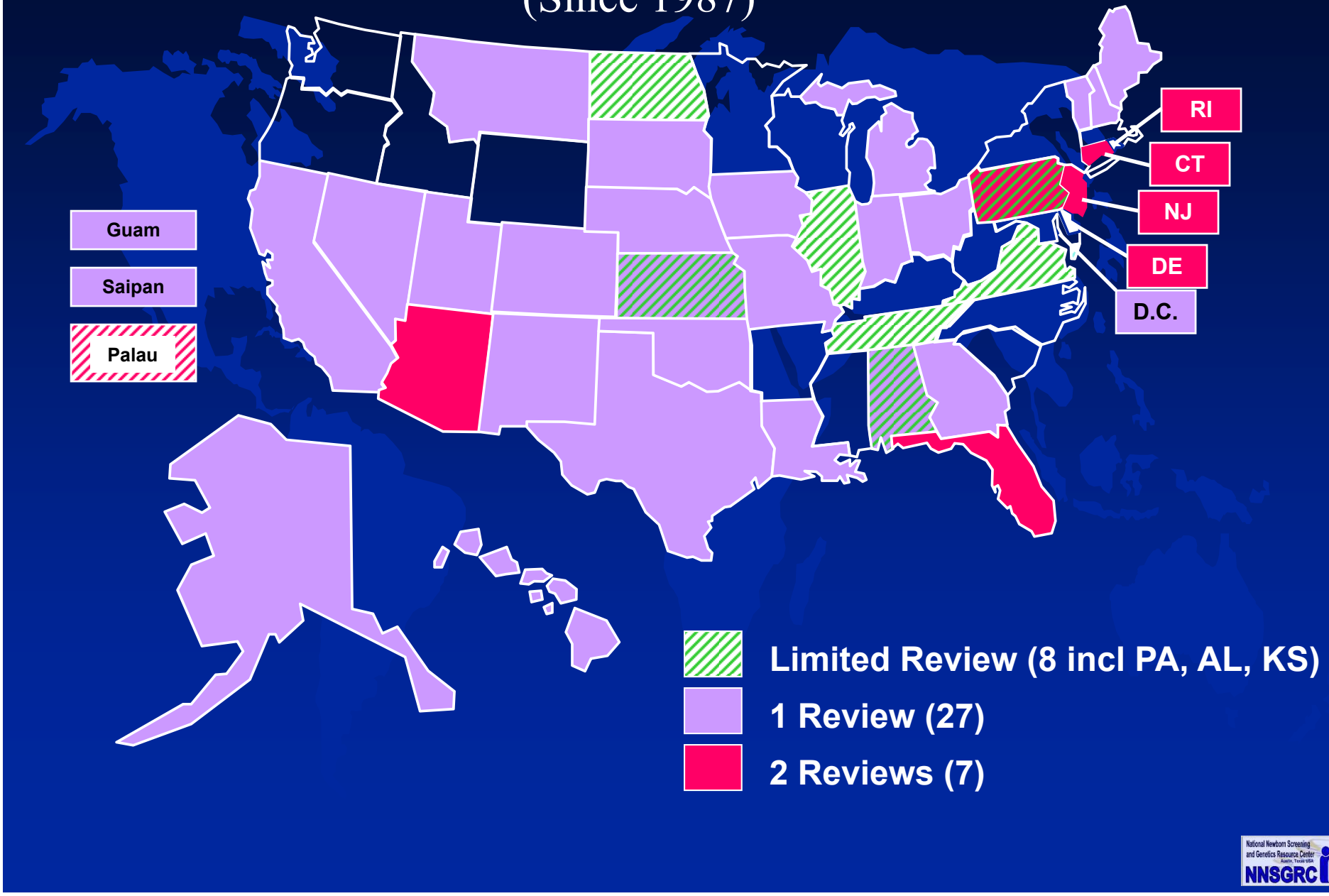
Quality Improvement Reviews

(Since 1987)



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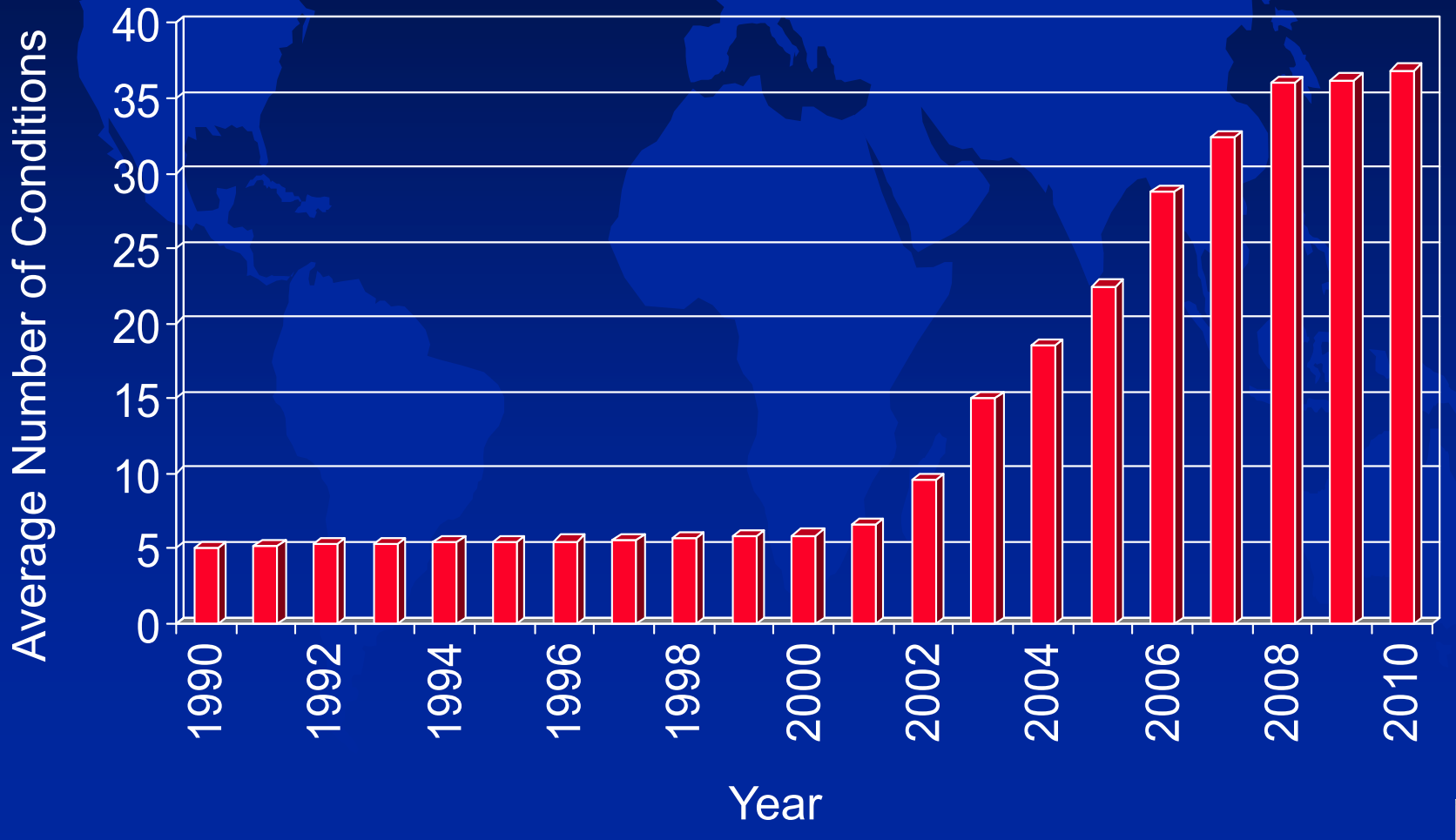
Average Number of Newborn Screening Conditions Required in US Programs 1990-2010



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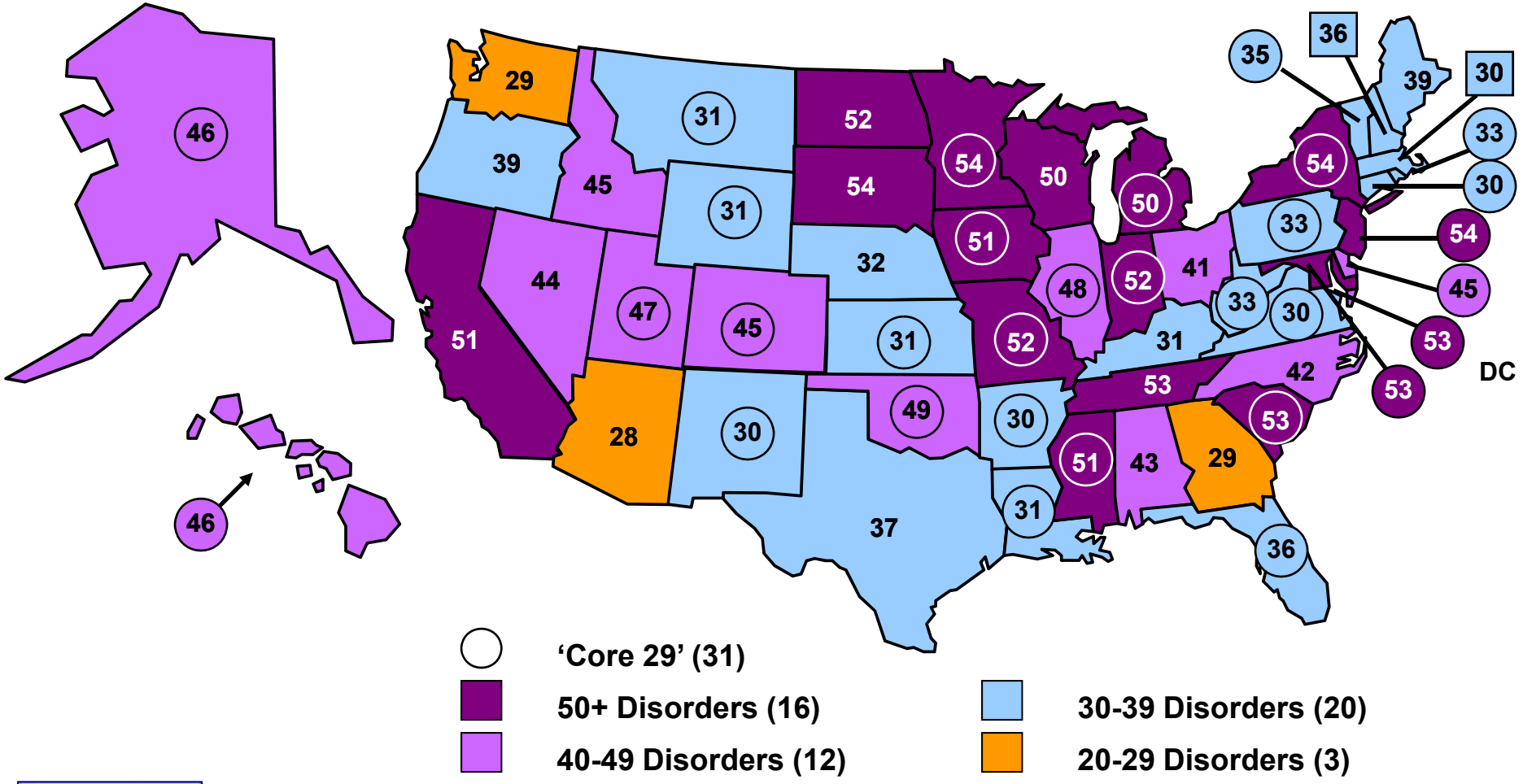
U.S. Newborn Screening

Conditions Required – Feb 1, 2011

(Conditions available as an option to a selected population are not counted – Must be universally required)



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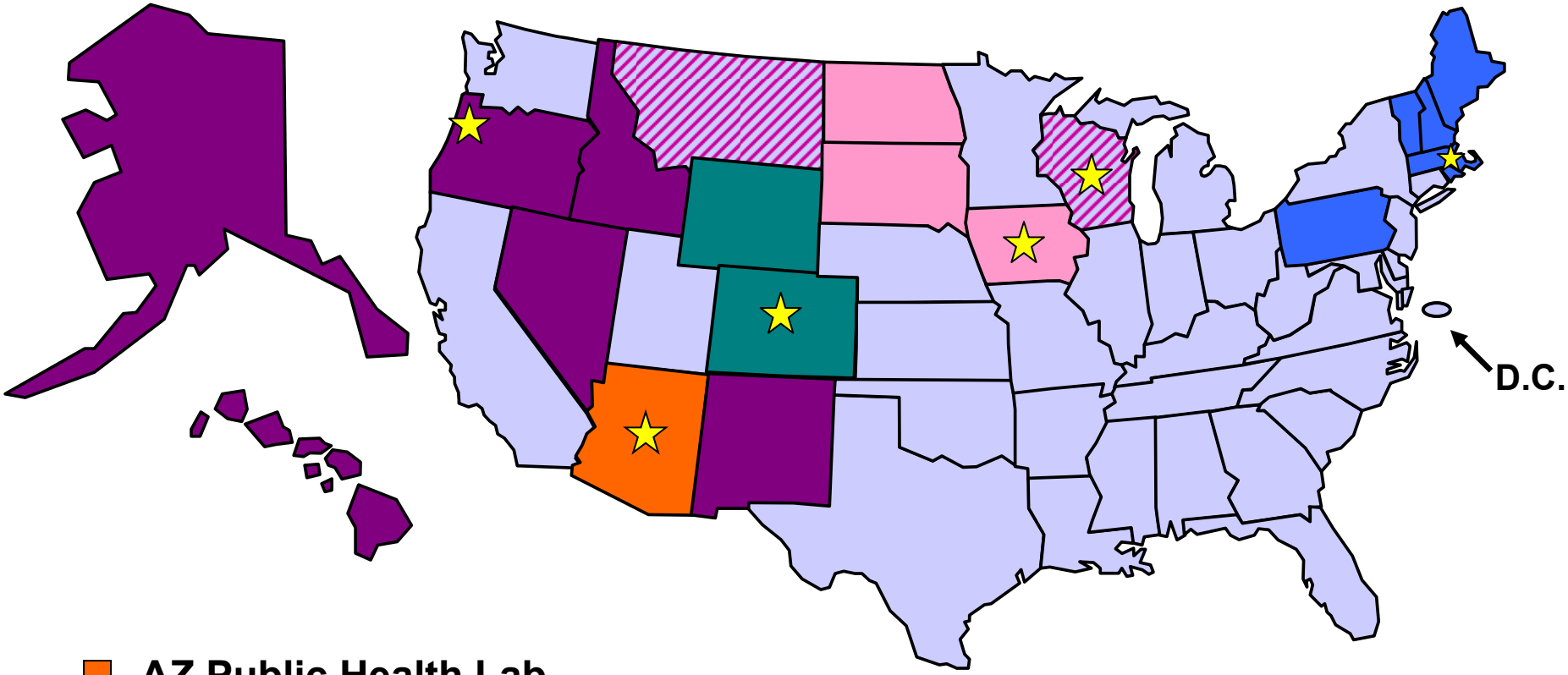

- AZ Public Health Lab
- OR Public Health Lab
- WI Public Health Lab
- IA Public Health Lab
- CO Public Health Lab
- U Mass Lab

Laboratory Service Delivery Models
States Using Contract Screening Laboratories
(Public)

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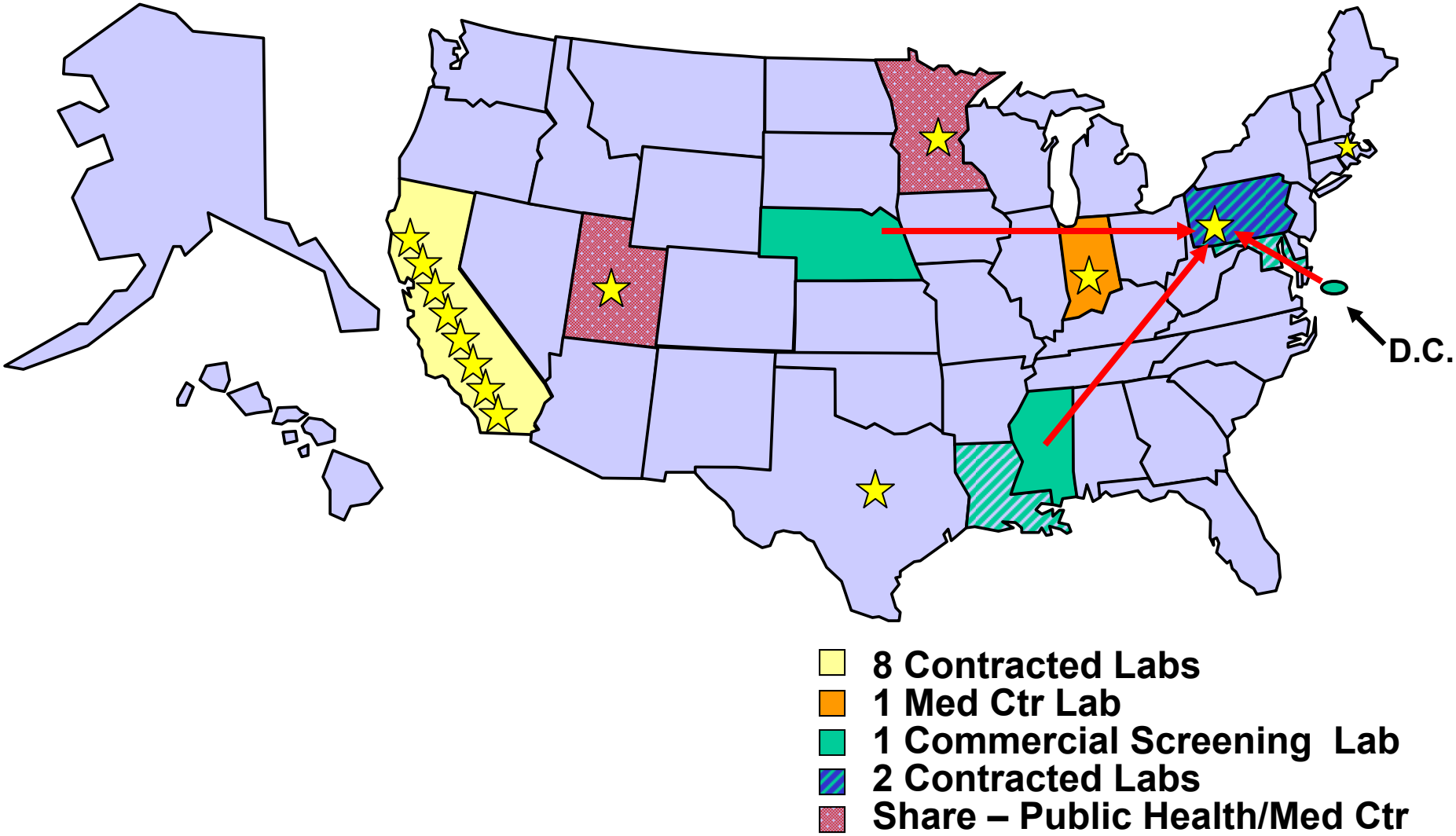

Laboratory Service Delivery Models
States Using Contract Screening Laboratories
(Commercial/Non-profit)

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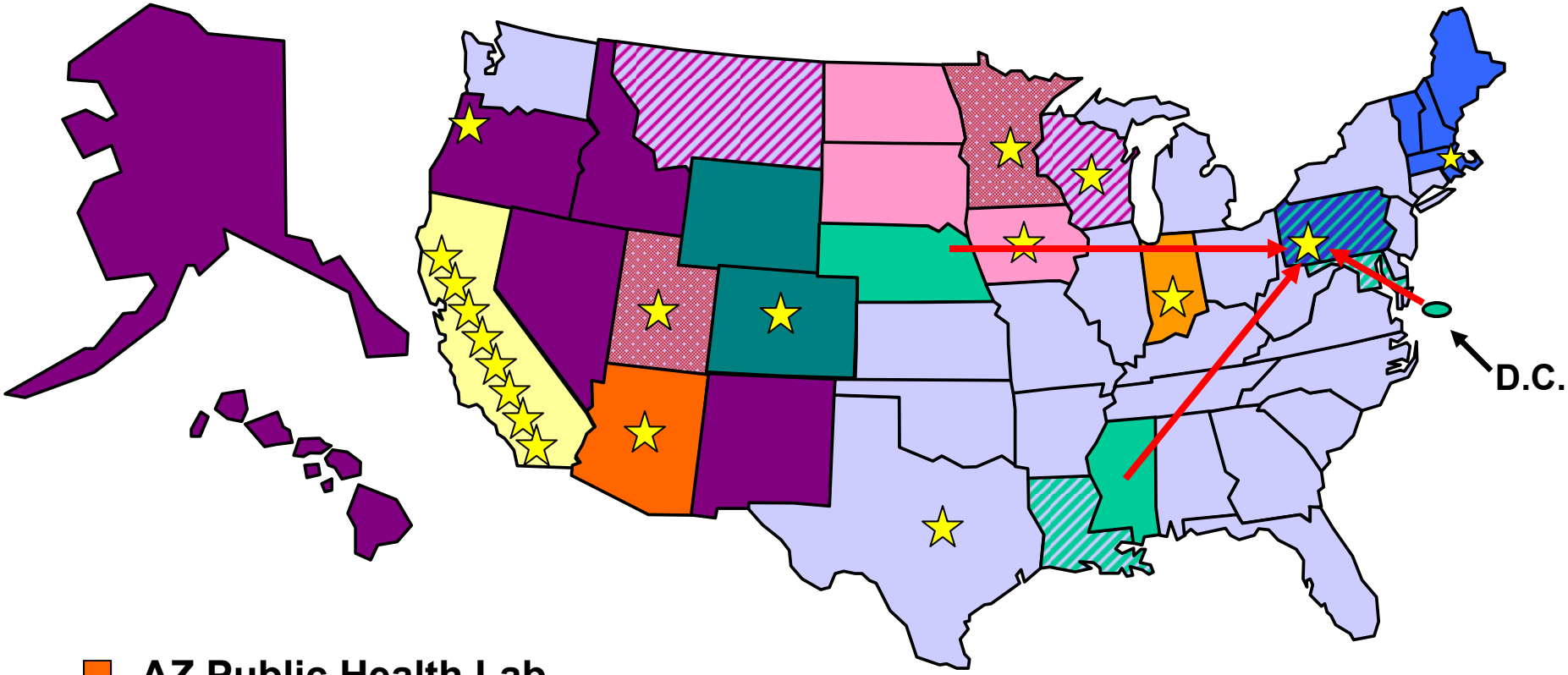

- AZ Public Health Lab
- OR Public Health Lab
- WI Public Health Lab
- IA Public Health Lab
- CO Public Health Lab
- U Mass Lab
- Allows Commercial Lab Competition
- 8 Contracted Labs
- 1 Med Ctr Lab
- 1 Commercial Screening Lab
- 2 Contracted Labs
- Share – Public Health/Med Ctr

Laboratory Service Delivery Models
States Using Contract Screening Laboratories
(Public and/or Commercial/Non-profit)

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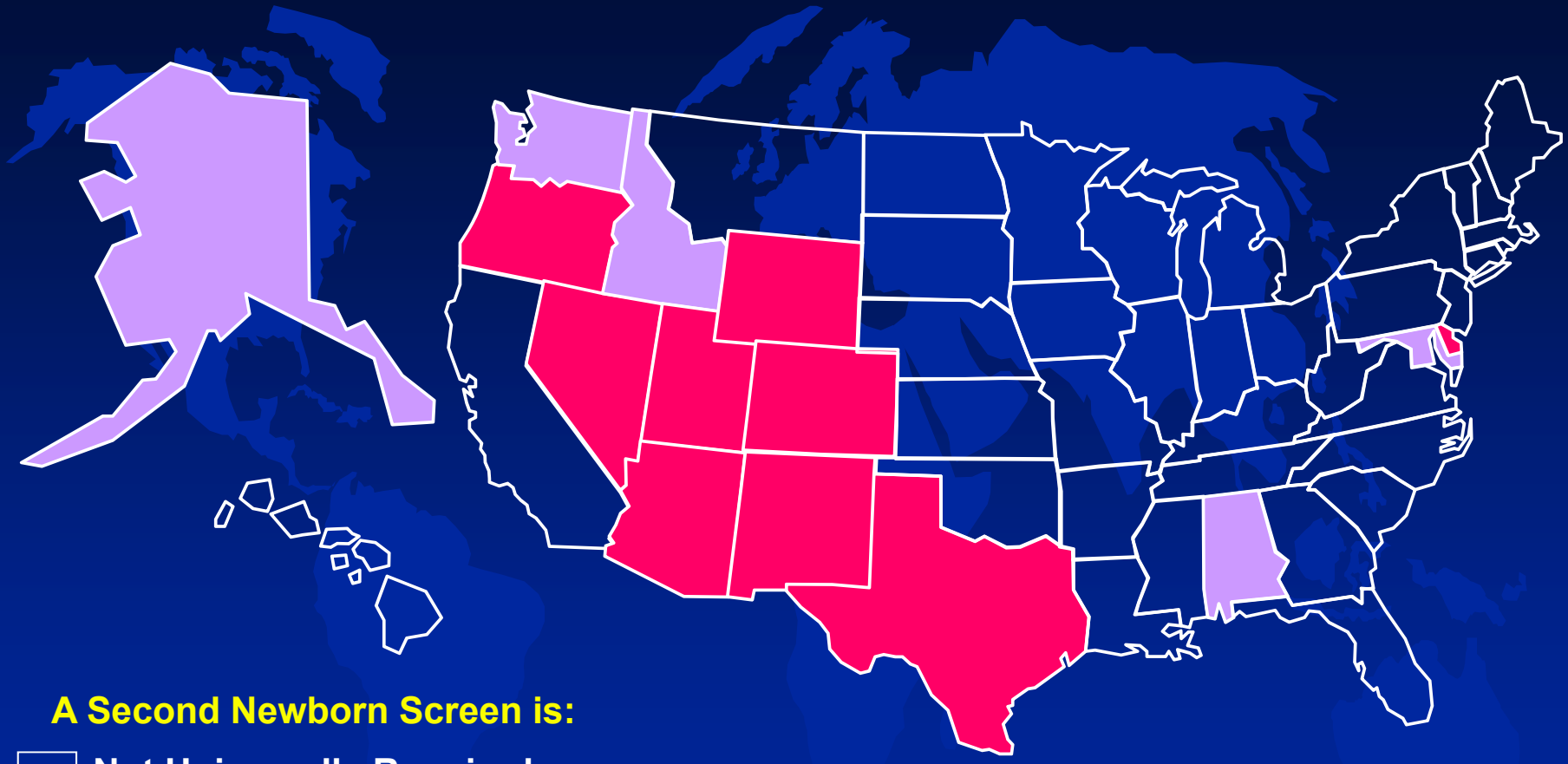
States With Two 'Required' Screens



A Second Newborn Screen is:

- Not Universally Required
- Universally Required (CO Limits Analytes on 2nd)
- Strongly Recommended (>85%)

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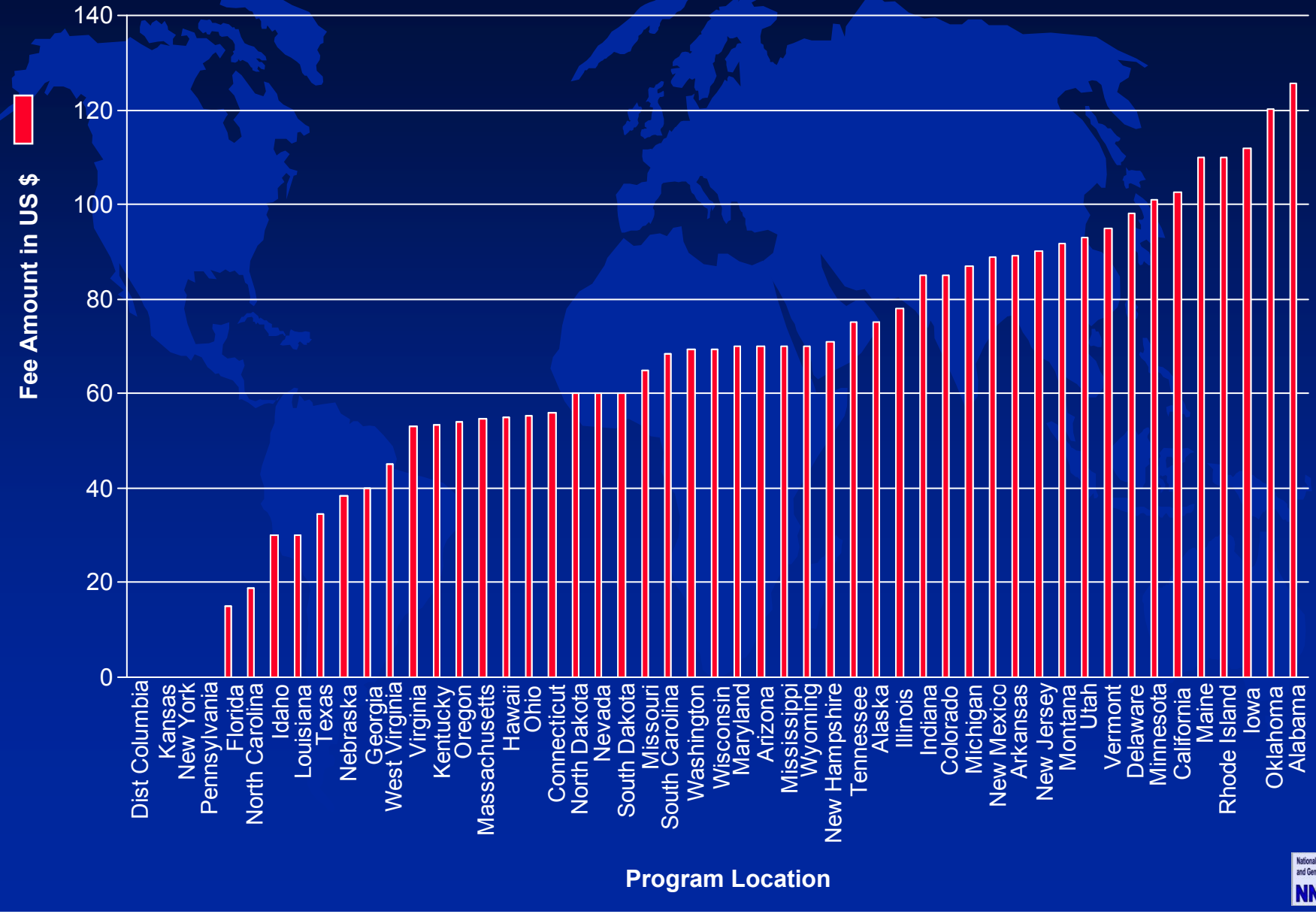
U.S. Newborn Screening Fees – 2011

(Ascending Amount with Number of Mandated Disorders Overlayed and Normalized)



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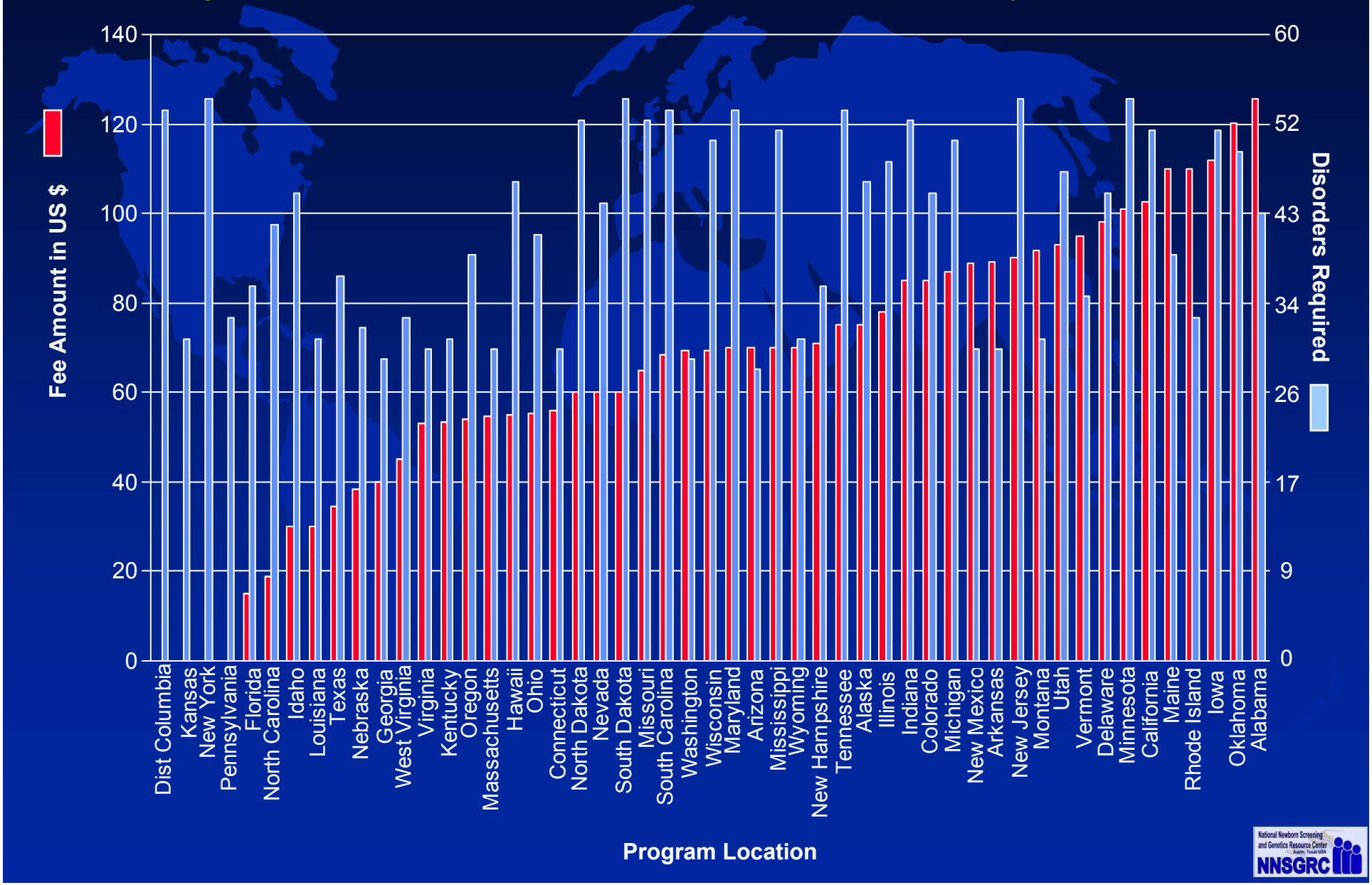
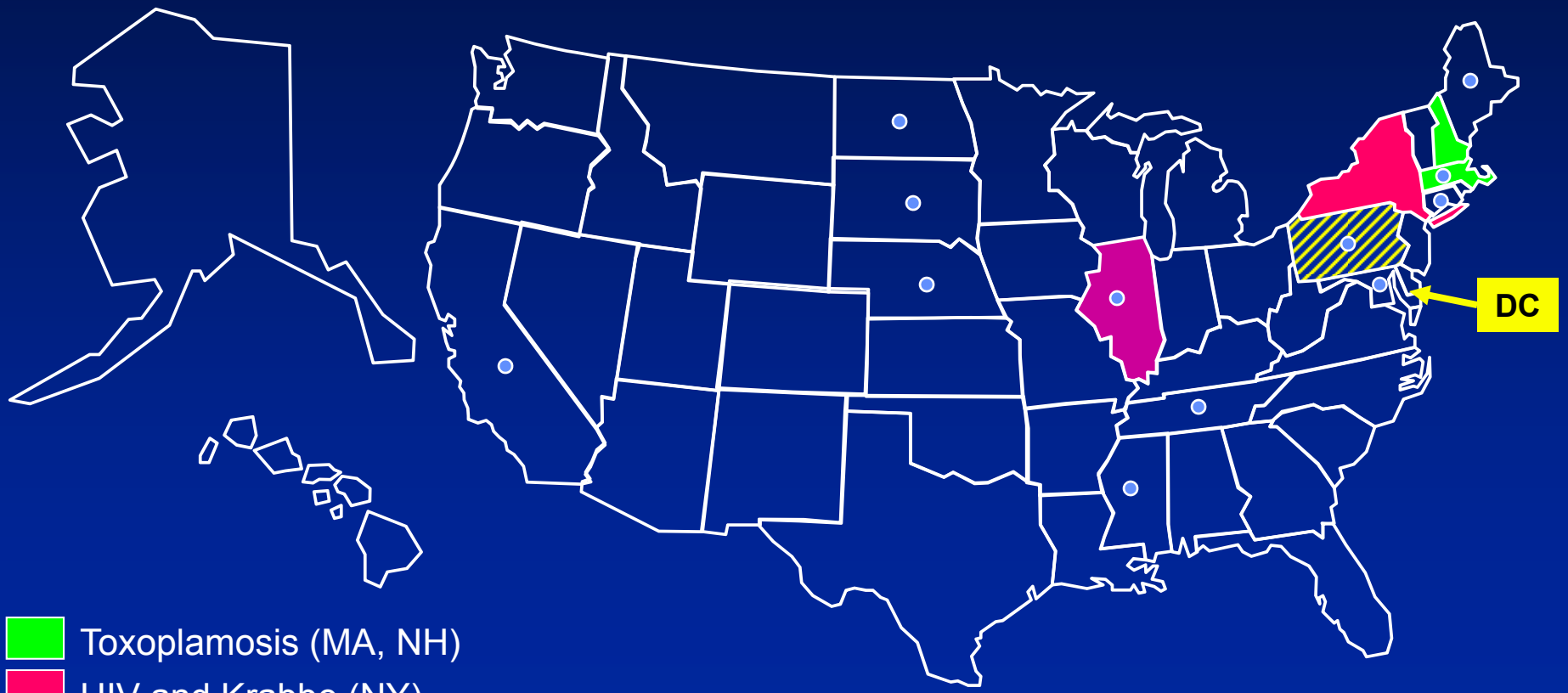
U.S. Newborn Screening Fees – 2011

(Ascending Amount with Number of Mandated Disorders Overlayed and Normalized)



U.S. Newborn Screening

Conditions Not On SACHDNC List



- Toxoplamosis (MA, NH)
- HIV and Krabbe (NY)
- G6PD [DC; PA (part)]
- LSDs – Pompe, Gaucher, Fabry (pilot)

- Note: Additional MS/MS disorders also included in CA, CT, IL, ME, MD, MA, MS, NB, ND, PA, SD, TN

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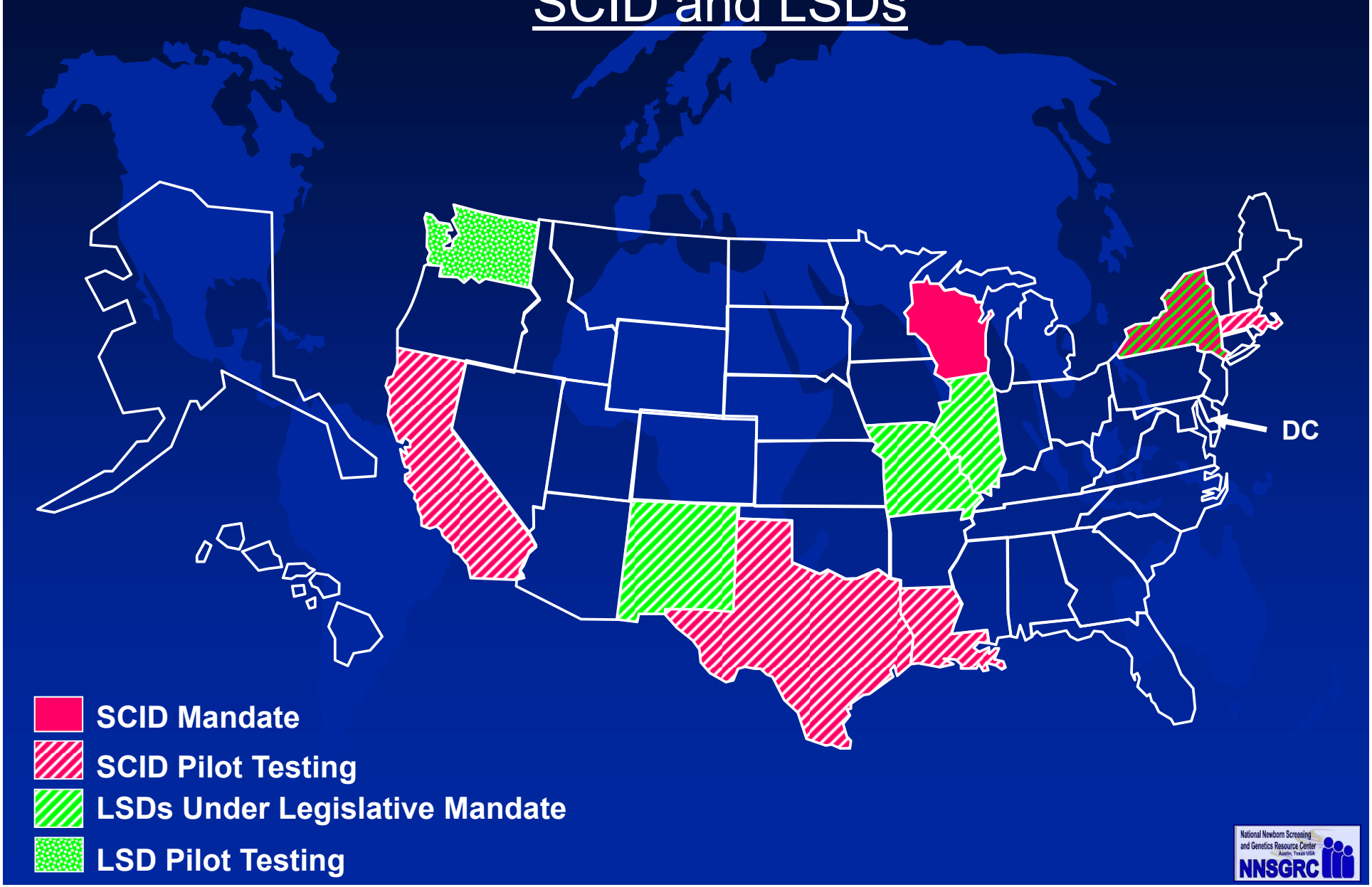
U.S. Newborn Screening

SCID and LSDs



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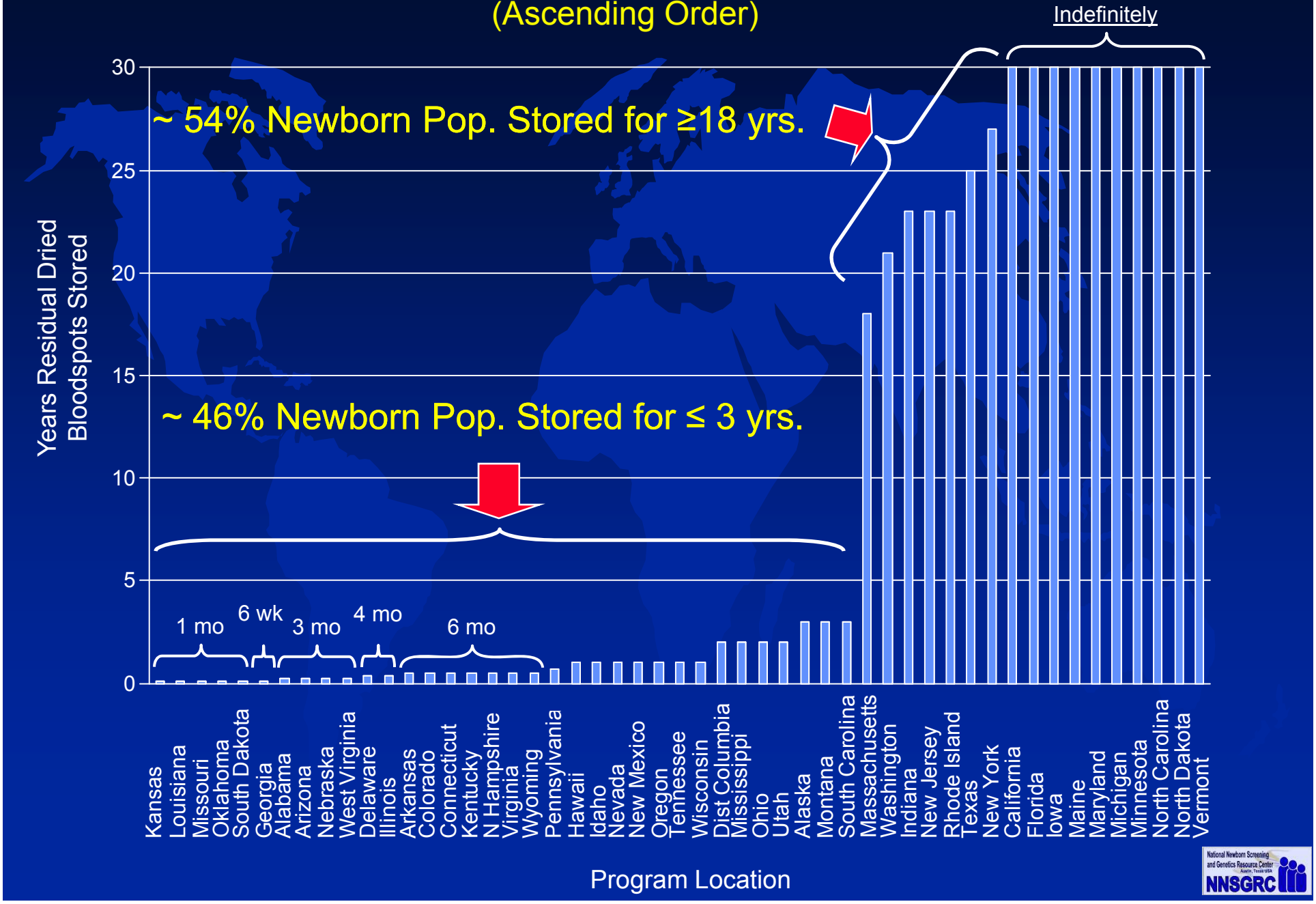
Reported Residual Bloodspot Storage – 9/1/2009

(Ascending Order)



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Arkansas

• Births (2008)	39,502
• Medicaid Births (2003)	51.7%
• Screens (2008)	39,826
• Screens (2009)	41,069
• No. Screens Required	1
• No. Disorders Required	30
• Fee	$89.25
• Residual Storage	3 - 6 mo. (freezer space) -20 °C

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Colorado

- Births (2008) 70,527
- Medicaid Births (2003) 37.3%
- Screens (2008) 134,702
- Screens (2009) 131,921
- No. Screens Required 2 (for 4 only)
- No. Disorders Required 45
- Fee $85.00
- Residual Storage 6 mo.
  Room temp.

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Kentucky

• Births (2008)	56,621
• Medicaid Births (2003)	43.7%
• Screens (2008)	60,408
• Screens (2009)	60,136
• No. Screens Required	1
• No. Disorders Required	31
• Fee	$53.50
• Residual Storage	6 mo. 2 - 8 °C

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Missouri

• Births (2008)	81,992
• Medicaid Births (2003)	45.4%
• Screens (2008)	90,713
• Screens (2009)	89,230
• No. Screens Required	1
• No. Disorders Required	52
• Fee	$65.00
• Residual Storage	1 mo (zip lock bags) -30°C (5 yr. July 1, 2011)

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New York

• Births (2008)	252,360
• Medicaid Births (2003)	40.5%
• Screens (2008)	277,449
• Screens (2009)	273,915
• No. Screens Required	1
• No. Disorders Required	54
• Fee	no fee
• Residual Storage	27 yr. 4 °C

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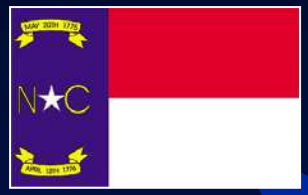
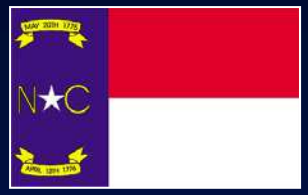


North Carolina



- Births (2008) 132,106
- Medicaid Births (2003) 47.9%
- Screens (2008) 130,703
- Screens (2009) no report
- No. Screens Required 1
- No. Disorders Required 42 (SCID Rec.)
- Fee $19.00

Issues: testing unsat specimens, fee for repeat to physician

- Residual Storage 5 yr.
  Room temperature

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Ohio

- Births (2008) 149,346
- Medicaid Births (2003) 32.1%
- Screens (2008) 151,583
- Screens (2009) no report
- No. Screens Required 1
- No. Disorders Required 41 (SCID under review – will take about a year if adv. approved this month)
- Fee $55.16
- Residual Storage 2 yr.
  Room Temperature

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Pennsylvania

- Births (2008) 148,460
- Medicaid Births (2003) 31.0%
- Screens (2008) 148,460
- Screens (2009) 148,474
- No. Screens Required 1
- No. Disorders Required 33 (6) (DBS issue)
- Fee no fee (discussions concerning possible legislative change; OZ system)
- Residual Storage 8 mo. (w/ desiccant) -20°C

Texas

- Births (2008) 412,127
- Medicaid Births (2001*) 47.6%
- Screens (2008) 795,974
- Screens (2009) 789,467
- No. Screens Required 2
- No. Disorders Required 51 (7 not impl; 14 likely to be detected)
- Fee $34.50
- Residual Storage 25 yr. (under review) No information

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Utah

• Births (2008)	56,787
• Medicaid Births (2003)	30.2%
• Screens (2008)	111,915
• Screens (2009)	108,870
• No. Screens Required	2
• No. Disorders Required	47
• Fee	$93.00
• Residual Storage	2 yr. (7 day room temp) -20 °C

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Virginia

• Births (2008)	104,990
• Medicaid Births (2003)	27.6%
• Screens (2008)	113,922
• Screens (2009)	108,656
• No. Screens Required	1
• No. Disorders Required	30
• Fee	$53.00
• Residual Storage	6 mo (positives 10 yr.) room temperature

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Summary – Basic Program Information

State	Births	Medicaid Births	Screens 2008	Screens 2009	Screens	Tests	Fee	Storage
AR	39,503	51.7%	39,826	41,069	1	30	$89.25	3-6 mo
CO	70,527	37.3%	134,702	131,921	2	45	(2) $85.00	6 mo
KY	56,621	43.7%	60,408	60,136	1	31	$53.50	6 mo
MO	81,992	45.4%	90,713	89,230	1	52	$65.00	1 mo
NY	252,360	40.5%	277,449	273,915	1	54	None	27 yr
NC	132,106	47.9%	130,703	No report	1	42	$19.00	5 yr
OH	149,346	32.1%	151,583	No report	1	41	$55.16	2 yr
PA	148,460	31.0%	148,474	145,367	1	33	None	8 mo
TX	412,127	†47.6%	795,974	789,467	2	‡51	(1) $34.50	25 yr
UT	56,787	32.2%	111,915	108,870	2	47	(2) $75.00	2 yr
VA	104,990	27.6%	113,922	108,656	1	30	$53.00	6 mo

Summary – Case Finding Information (2009)

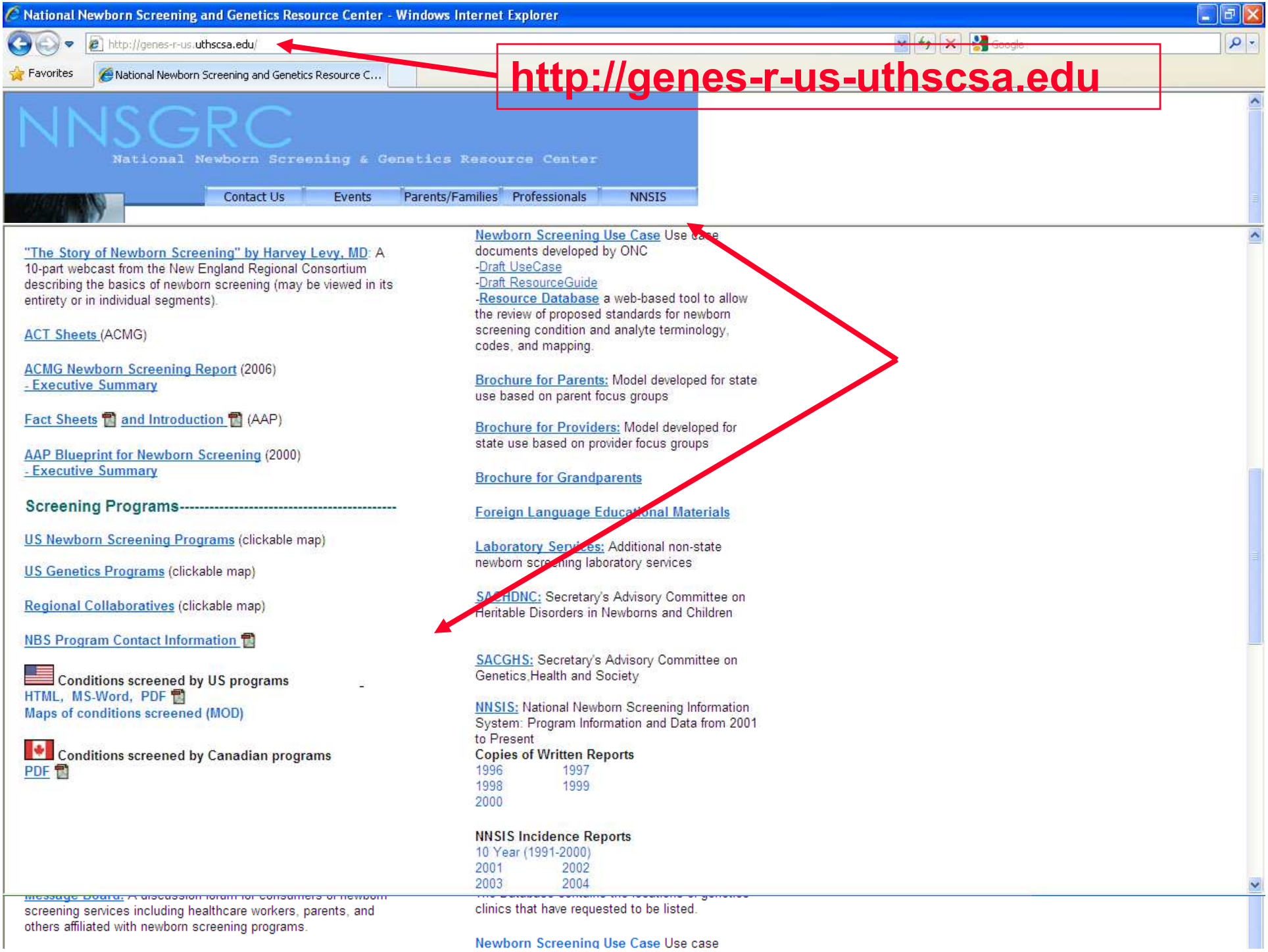
State	CAH			GAL			CH	BIO	
	SW	SV	Other	GALT	GALE	Variant		Profound	Partial
AR	3	0	0	0	0	13	23	2	
CO	3	0	0	1	0	0	29	1	3
KY	2	0	0	0	0	16	38	1	6
MO	1	0	0	2	0	1	31	1	10
NY	11*			6*			141	13*	
NC	6	0	0	1	0	14	68	0	0
OH	0	1	0	2	0	21	70	1	1
PA	8	0	1	4	6	14	54	2	14
TX	23	3	11	4	0	104	183	6	31
UT	3	1	0	1	0	59	18	1	2
VA	5	2	0	2	0	23	33	2	14

* Total, Not divided by type

Summary – Case Finding Information (2009)

State	Hgb		PKU		CF		MS/MS Groupings		
	All	S,S	Classical	Variant	Classical	Variant	AA	FAO	OA
AR	22	11	1	0	8		1	3	1
CO	14	9	2	0	17		1	5	4
KY	0		4	0	14		2	12	2
MO	0		1	0	21	1	0	14	4
NY	214	112	19 ?		71 ?		1	20	42
NC	No data		5	3	22	8	2	8	6
OH	69	35	8	2	34	6	3	16	7
PA	83	38	6	8	15	5	3	6	8
TX	210	106	17	3	* 9		4	28	17
UT	4	2	2	3	15	1	0	18	6
VA	80	46	5	I2	15		0	10	5

* Incomplete year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http://genes-r-us.uthscsa.edu/
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http://genes-r-us-uthscsa.edu
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Contact Us
Events
Parents/Families
Professionals
NNSIS
"The Story of Newborn Screening" by Harvey Levy, MD: A 10-part webcast from the New England Regional Consortium describing the basics of newborn screening (may be viewed in its entirety or in individual segments).
ACT Sheets (ACMG)
ACMG Newborn Screening Report (2006)
- Executive Summary
Fact Sheets and Introduction (AAP)
AAP Blueprint for Newborn Screening (2000)
- Executive Summary
Screening Programs----------
US Newborn Screening Programs (clickable map)
US Genetics Programs (clickable map)
Regional Collaboratives (clickable map)
NBS Program Contact Information
Conditions screened by US programs
HTML, MS-Word, PDF
Maps of conditions screened (MOD)
Conditions screened by Canadian programs
PDF
Newborn Screening Use Case Use case documents developed by ONC
-Draft UseCase
-Draft ResourceGuide
-Resource Database a web-based tool to allow the review of proposed standards for newborn screening condition and analyte terminology, codes, and mapping.
Brochure for Parents: Model developed for state use based on parent focus groups
Brochure for Providers: Model developed for state use based on provider focus groups
Brochure for Grandparents
Foreign Language Educational Materials
Laboratory Services: Additional non-state newborn screening laboratory services
SACHDNC: Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
SACGHS: Secretary's Advisory Committee on Genetics,Health and Society
NNSIS: National Newborn Screening Information System: Program Information and Data from 2001 to Present
Copies of Written Reports
1996 1997
1998 1999
2000
NNSIS Incidence Reports
10 Year (1991-2000)
2001 2002
2003 2004
screening services including healthcare workers, parents, and others affiliated with newborn screening programs.
clinics that have requested to be listed.
Newborn Screening Use Case Use case

National Newborn Screening Information System - Windows Internet Explorer provided by MSN & Bing

http://nnsis.uthscsa.edu/xreports.aspx?XREPORTID=5

Favorites | National Newborn Screening Information System

Welcome to the
National Newborn Screening Information System
database

The database is hosted by the
National Newborn Screening and Genetics Resource Center
(NNSGRC)

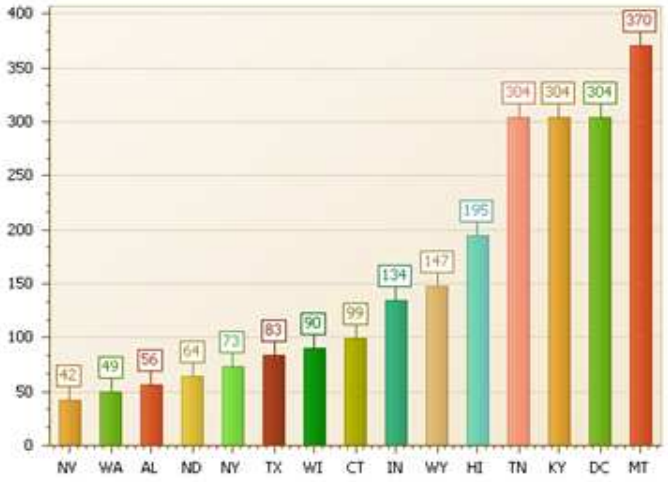
and is designed to provide a secure, Internet based, real-time, information collection and reporting system for capturing state and territorial newborn screening information

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

States Where Reported % Unsat >1 for 2010

Days Since Last Update

(have we heard from you lately?)



Current Issues

- 12 hr. vs. 24 hr. vs. 48 hr. for unsatisfactory specimens
- Required single screen vs. required two screens
- Financing – fees, Medicaid
- Best protocol for CF screening – IRT/DNA vs. IRT/IRT (carrier detection issues)
- Whether to mandate all conditions on the ACMG panel (detection and liability issues)
- Long-term follow-up responsibility
- Whether to universally mandate hearing screening
- National data reporting

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Thank You for Your Attention!


Thank You!!
http://genes-r-us.uthscsa.edu
http://www2.uthscsa.edu/nnsis/
http://www.marchofdimes.com/peristats/