

Newborn Screening: Current Status of State Newborn Screening Programs

June 5, 2006

Brad Therrell, Ph.D.

University of Texas Health Science Center at San Antonio

Newborn Screening and Genetics Resource Center

Austin, Texas





Alaska



- Hearing screening now universally mandated



Arizona



- April 2006 – Expansion rules promulgated; Fee up from \$20/screen to \$30 (1st) and \$40 (2nd)
- April 2006 – Added citrullinemia, tyrosinemia I, and arginosuccinic acidemia – Hearing screens required to be reported; ADHS doing follow-up
- May 2006 – Pilot testing for 5 FAOs and 9 OAs with reporting to begin in September. [CF in '07]



Colorado



- July 1, 2006 – Expand to 'core 29'
- Possibility of limiting MS/MS to 1st specimen
- Fee increase of ~ \$5 to \$60/baby (2 specimens)
- CAH protocol change – any 2nd specimen result higher than 1st is treated as presumptive positive



District of Columbia



- February 1, 2006 – Expanded to 53 conditions
- Expansion has been successful to date



Florida



- Clarification – Not screening for variant hemoglobins.
- Advisory Committee to consider question of 24 hr. vs. 48 hr. for satisfactory specimen. Currently 48 hr.



Georgia



- May 2006 – Law changes:
 - January 2007 – Fee allowed restricted to \$40/baby
 - January 2007 – Expand to the ‘core 29’ condition panel
 - Department of Audits instructed to evaluate effectiveness and efficiency of GDHL. Negative findings – RFP outside



Idaho



- Planning expansion to CF for 2007



Kansas



- May 2006 – Legislature authorized money for advisory committee to develop newborn screening guidelines and any rules or statutory changes necessary.
- Committee must report to appropriate Senate and House committees by January 1, 2007.
- Amended law requiring payment for PKU treatment formula and required instead a sliding fee scale.



Kentucky



- January 1, 2006 – Expanded to the ‘core 29’
- January 1, 2006 – Fee increased from \$14.50 to \$53.50
- CF detected by IRT/IRT protocol.



Louisiana



- August 1, 2006 – Will expand to 28 of ‘core 29’ – not CF
- August 1, 2006 – Fee increased from \$18.00 to \$30.00
- CF scheduled for July 1, 2007



Maryland



- June 1, 2006 – Added CF to screening panel
- Screening procedure for CF is IRT/IRT – no fee increase.



Nebraska



- January 1, 2006 – Added CF and CAH to required screening panel
- Screening procedure for CF is IRT/DNA



New Hampshire



- May 1, 2006 – After 2 ½ years of work, added 5 conditions to required panel: BIO, CAH, MCAD, CF and SCD
- Held 3 meetings of Advisory Committee (March, April, May) finally voting to expand the current panel of 11.
- Expansion will likely reflect MA panel minus SCAD and TYR – internal process has begun!!



New York



- Clarified mandated panel of tests – removed indication for S-beta-thalassemia and TYR
- Testing for Krabbe still in pilot.



New Mexico



- 2005 – Legislature mandated expansion
- 2005 - 2006 – Evaluation process to determine best approach
- 2006 – Western States Buying Cooperative process to obtain bids – CA, CO, OR, UT, MN ??
- October 1, 2006 – Will expand to Core 29
- October 1, 2006 – NM will contract all newborn screening services to Oregon
- October 1, 2006 – Cost expected to increase from \$32/2 screens to ~ \$89/2 screens



Ohio



- August 30, 2006 – Adding CF and CUD



Oklahoma



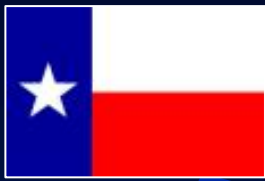
- June 5, 2006 – Adding MCAD
- Others in 'Core 29' by end of the year



Rhode Island



- Expansion to 29 core disorders scheduled for July 1, 2006.



Texas



- 2006 – RFP reissued
- May 2006 – No bid awarded; Texas will maintain screening lab
- Anticipated start of expanded testing – November 2006
- Fee increase expected – \$19.50 to 29.50/screen (2 req'd)



Utah



- January 1, 2006 – Expanded to MS/MS panel (contract with ARUP)



Washington



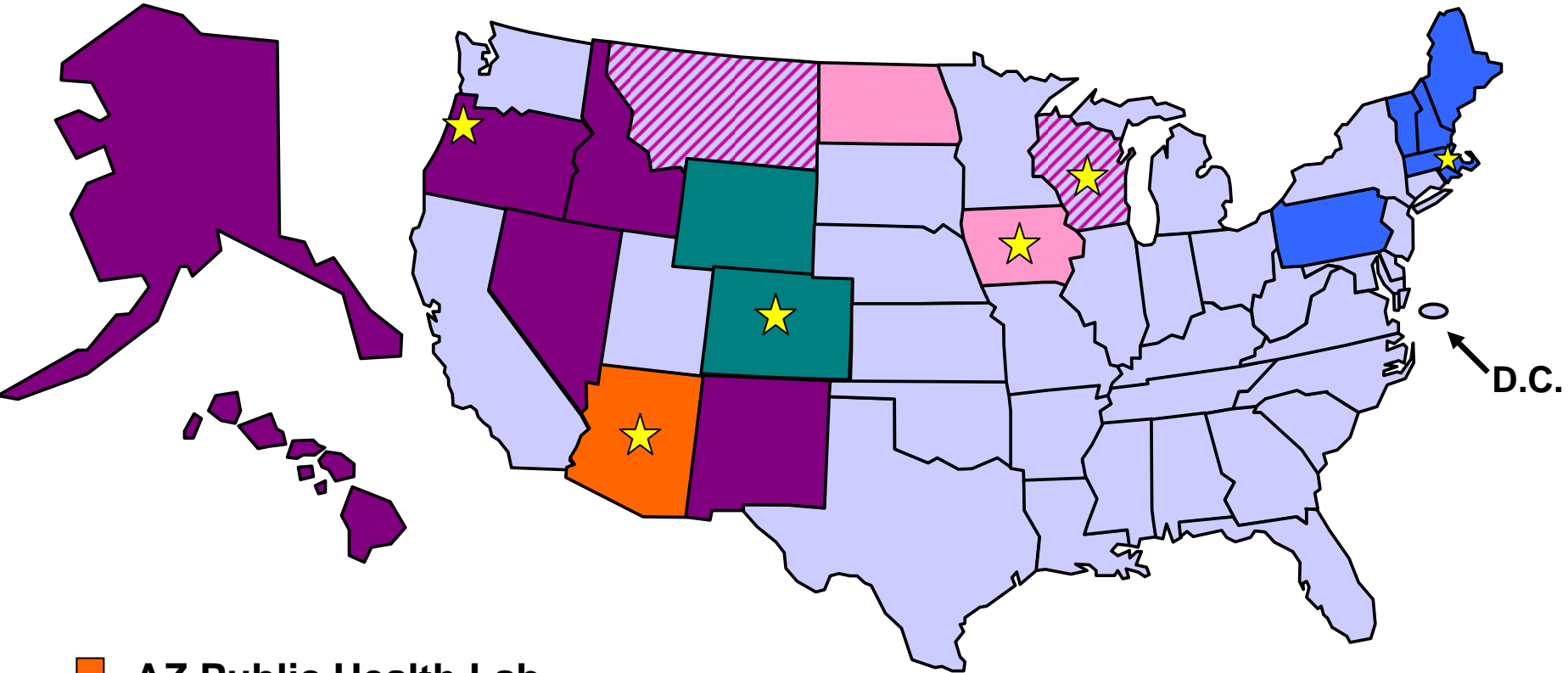
- November 1, 2005 – Raised clinical surcharge by \$3.10 to \$6.60. Overall fee is \$60.90 + \$6.60 = \$67.50
- March 15, 2006 – Added CF (IRT/IRT Protocol).
- Coding revision process underway for expansion to other 16.



Wyoming

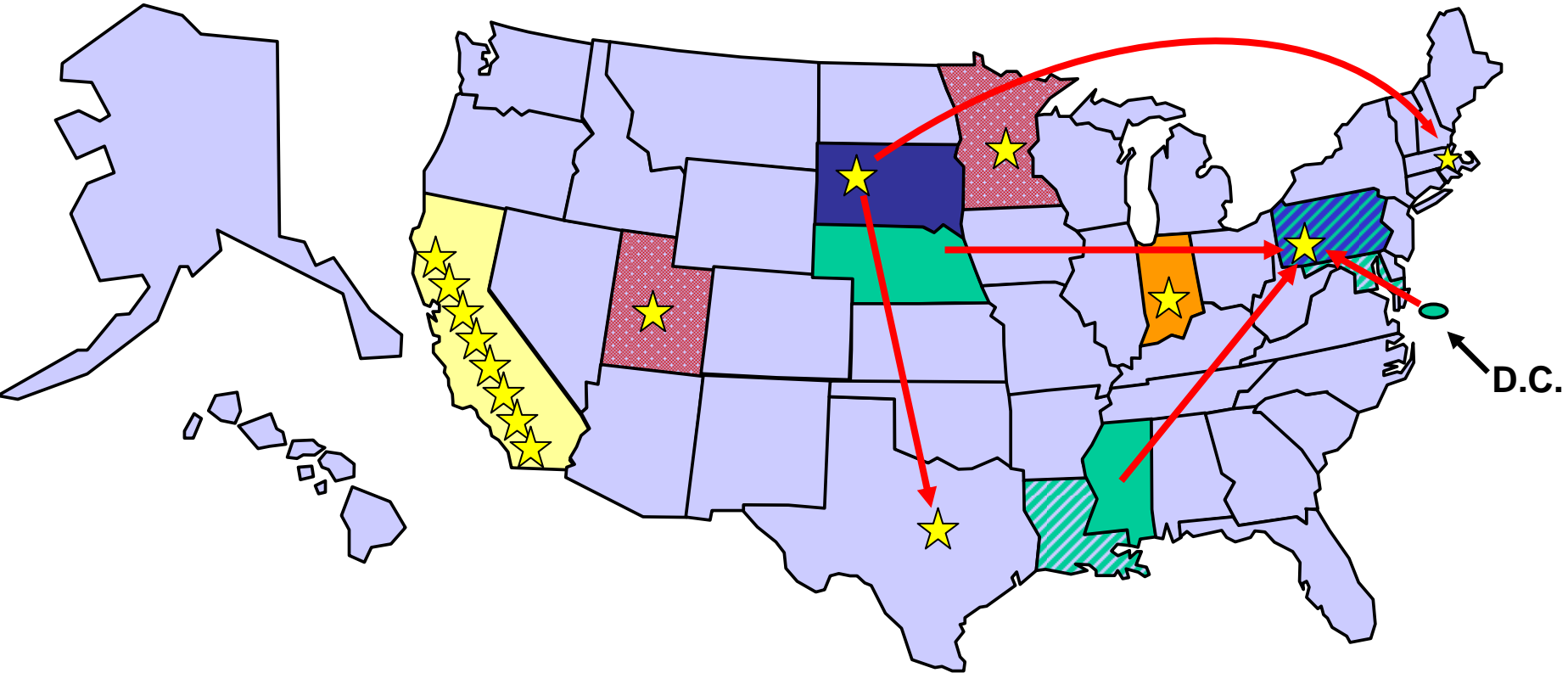


- July 1, 2006 – Scheduled to expand to 'Core 29' along with Colorado (their contract laboratory)



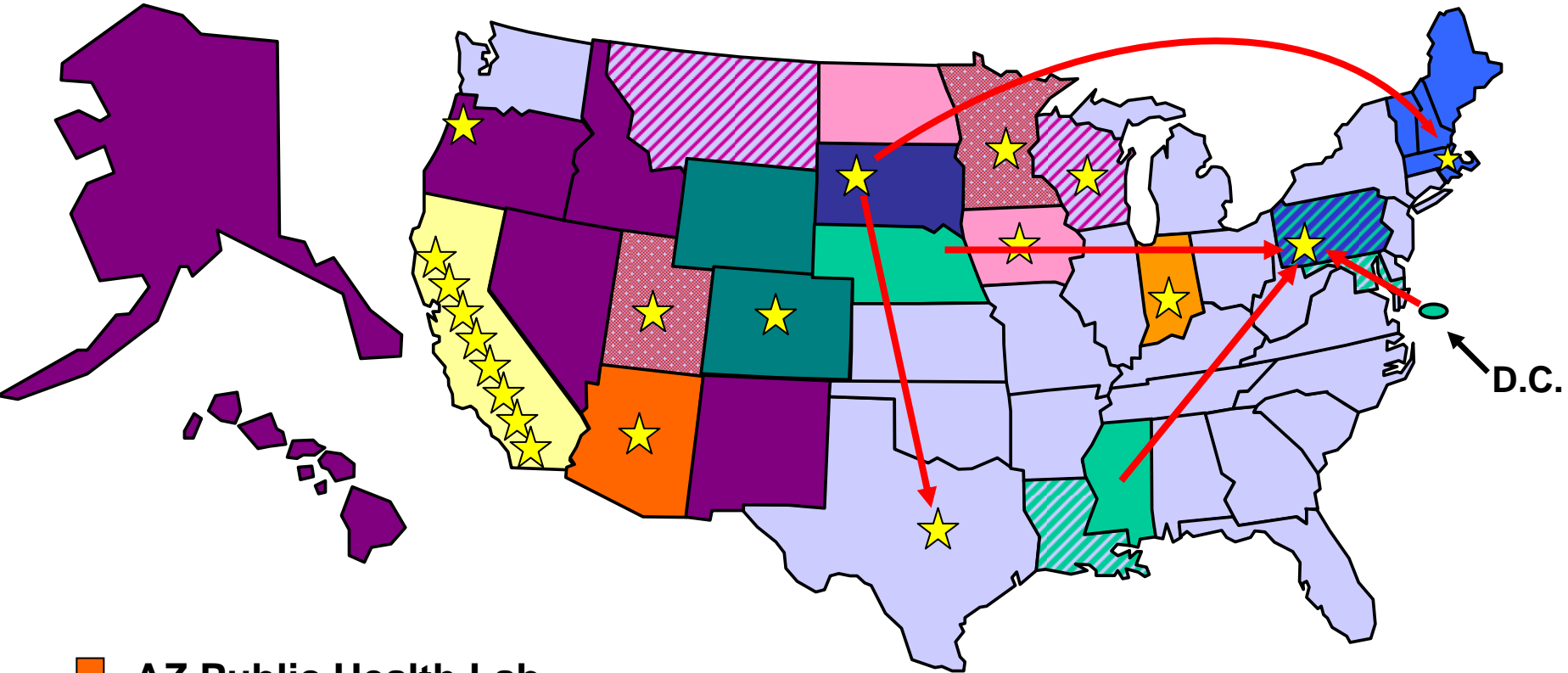
- AZ Public Health Lab
- OR Public Health Lab
- WI Public Health Lab
- IA Public Health Lab
- CO Public Health Lab
- U Mass Lab

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



- 8 Contracted Labs
- 1 Pathology Lab
- 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
(Commercial/Non-profit)



- 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 Pathology Lab
- 1 Med Ctr Lab
- 1 Commercial Screening Lab
- ▨ 2 Contracted Labs
- ▨ Share – Public Health/Med Ctr

Laboratory Service Delivery Models

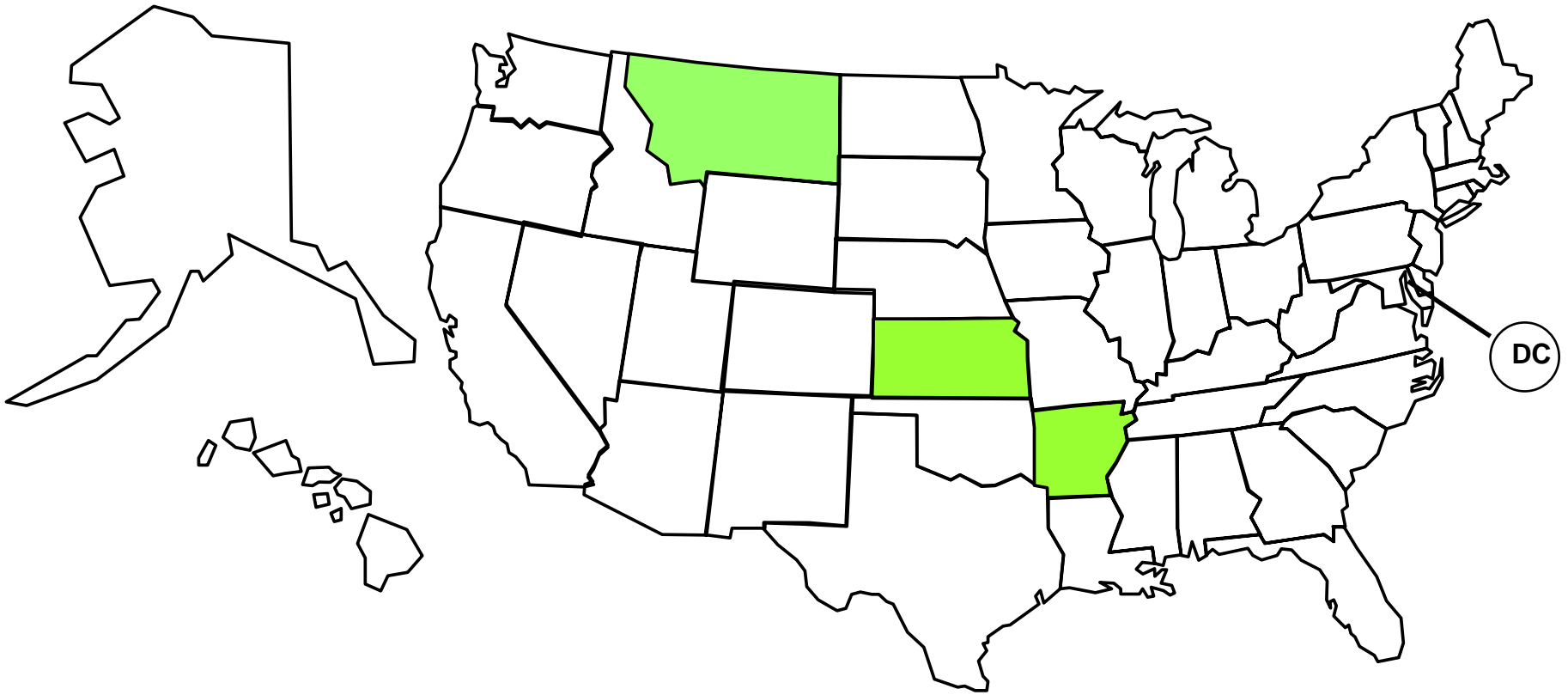
States Using Contract Screening Laboratories (Public and/or Commercial/Non-profit)



U.S. Newborn Screening

Conditions Required – June 1, 2006

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



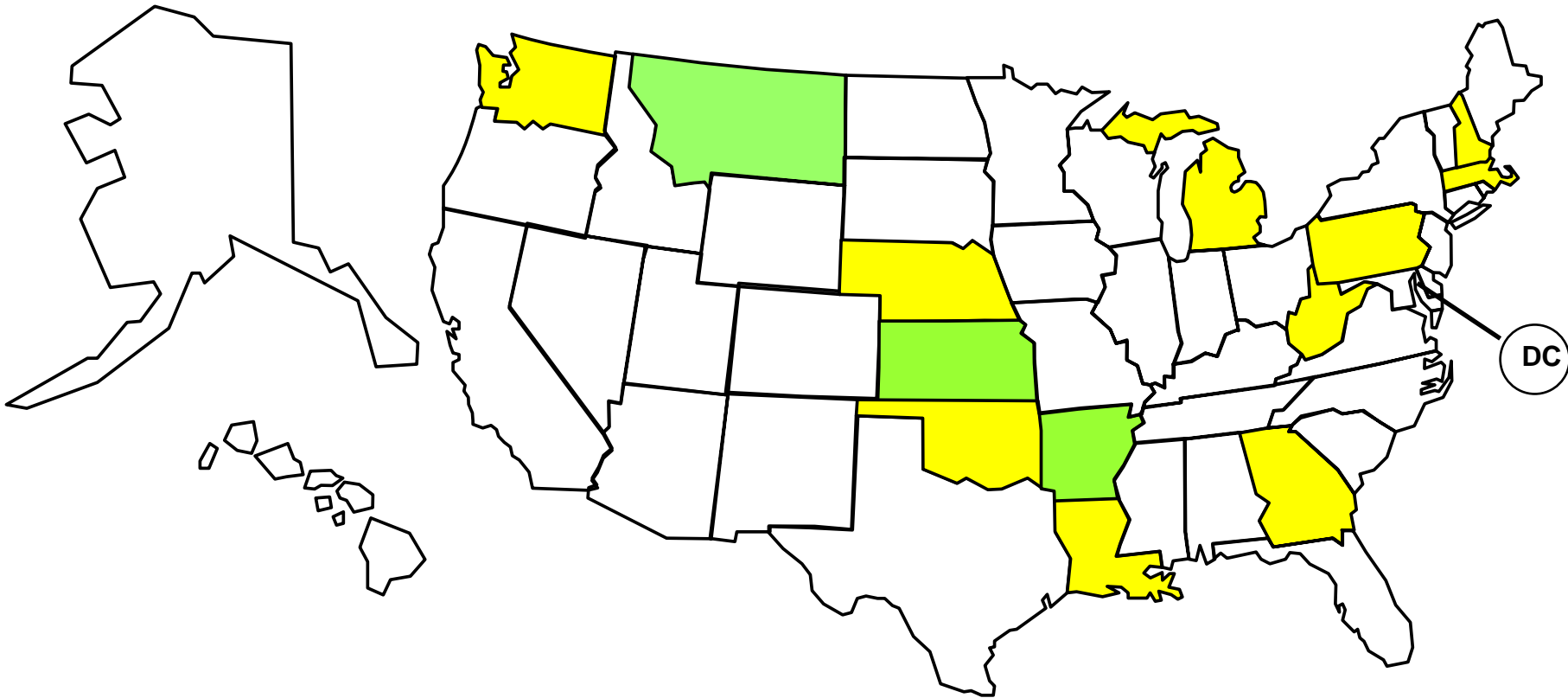
U.S. Newborn Screening

Conditions Required – June 1, 2006

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



<10 Disorders (3)



U.S. Newborn Screening

Conditions Required – June 1, 2006

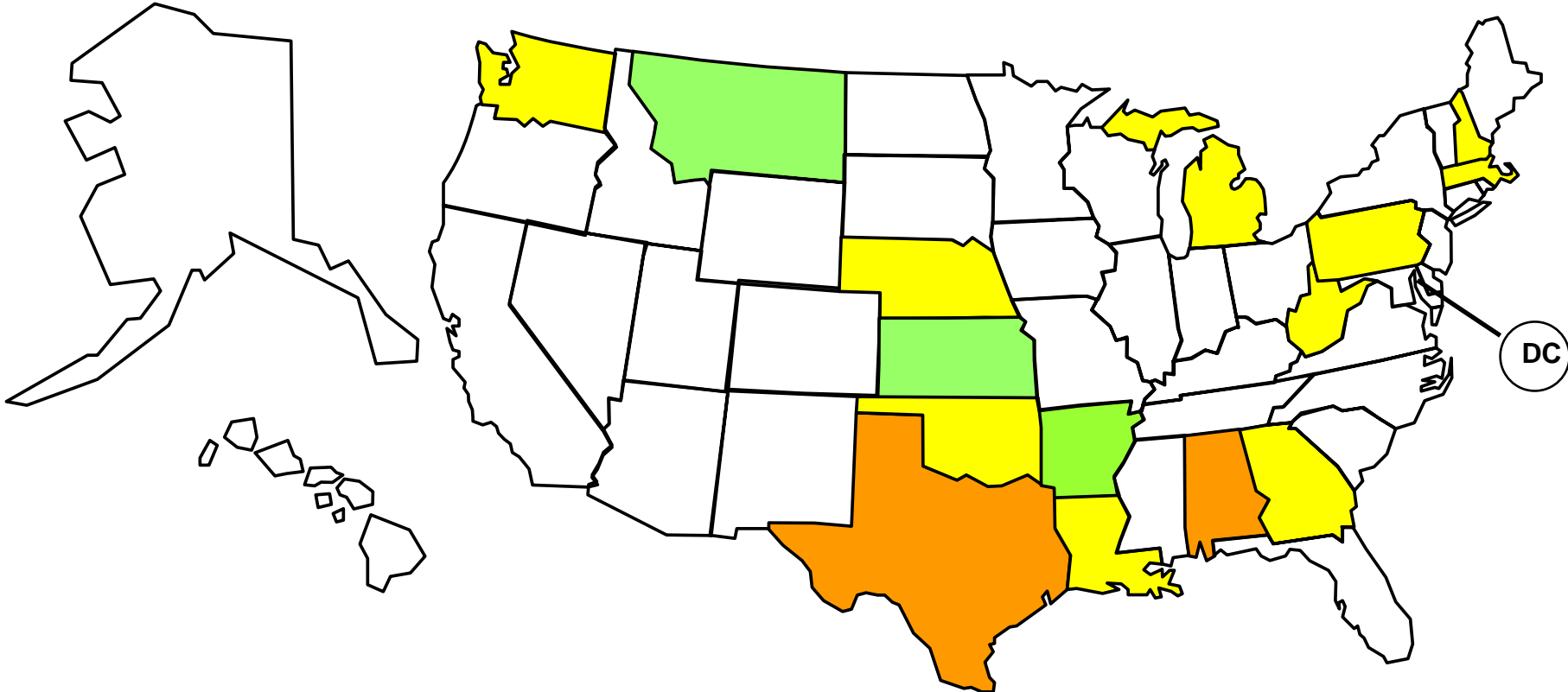
(Conditions available as an option to selected population are not counted)



10-19 Disorders (10)



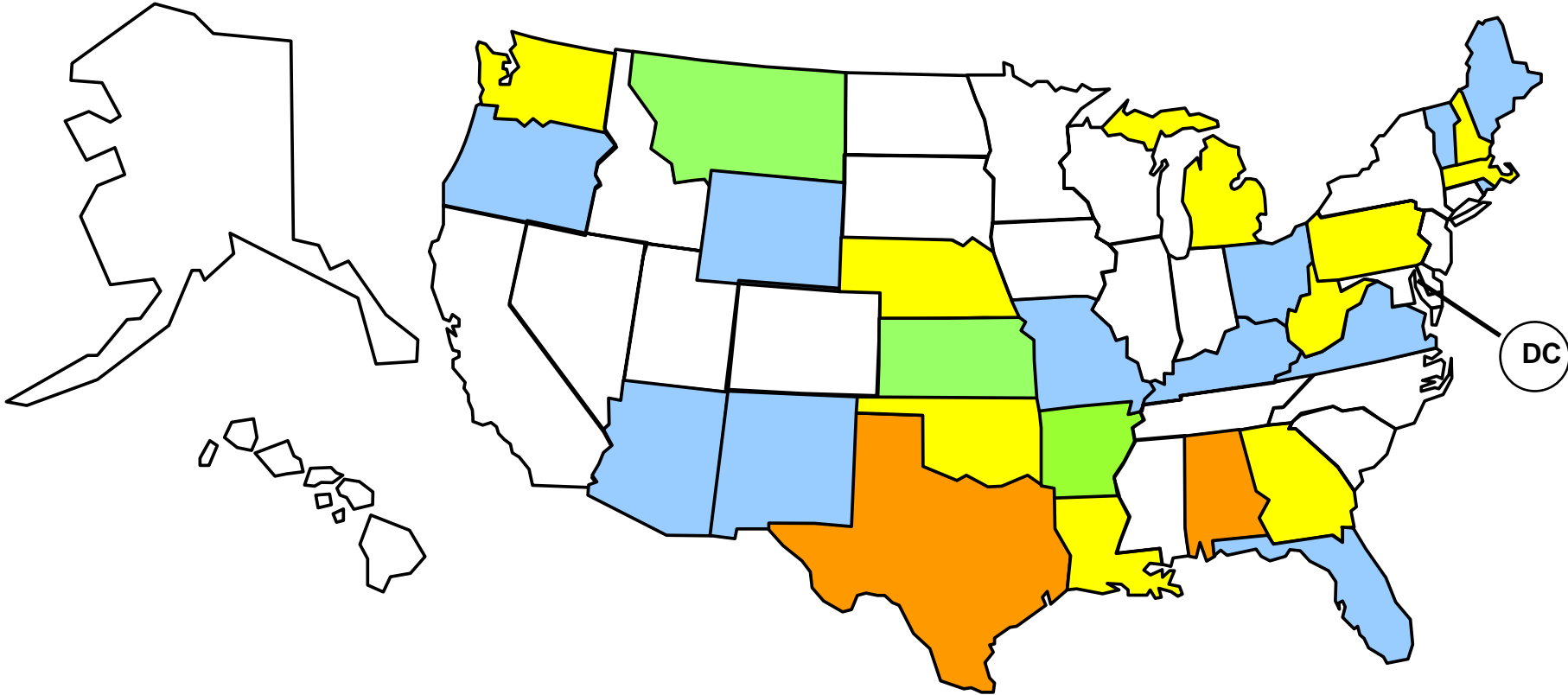
<10 Disorders (3)



U.S. Newborn Screening

Conditions Required – June 1, 2006

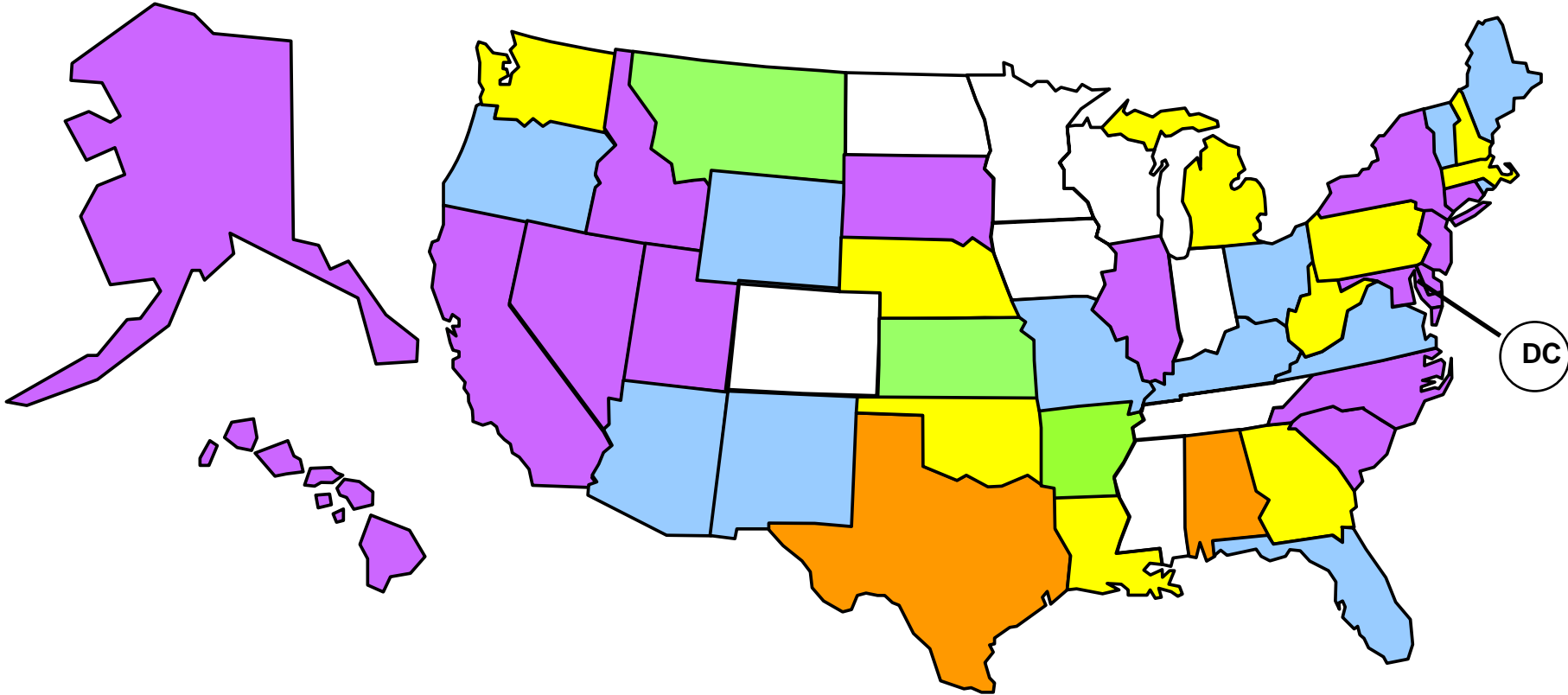
- 20-29 Disorders (2)** (Conditions available as an option to selected population are not counted)
- 10-19 Disorders (10)** (Conditions available as an option to selected population are not counted)
- <10 Disorders (2)**



U.S. Newborn Screening

Conditions Required – June 1, 2006

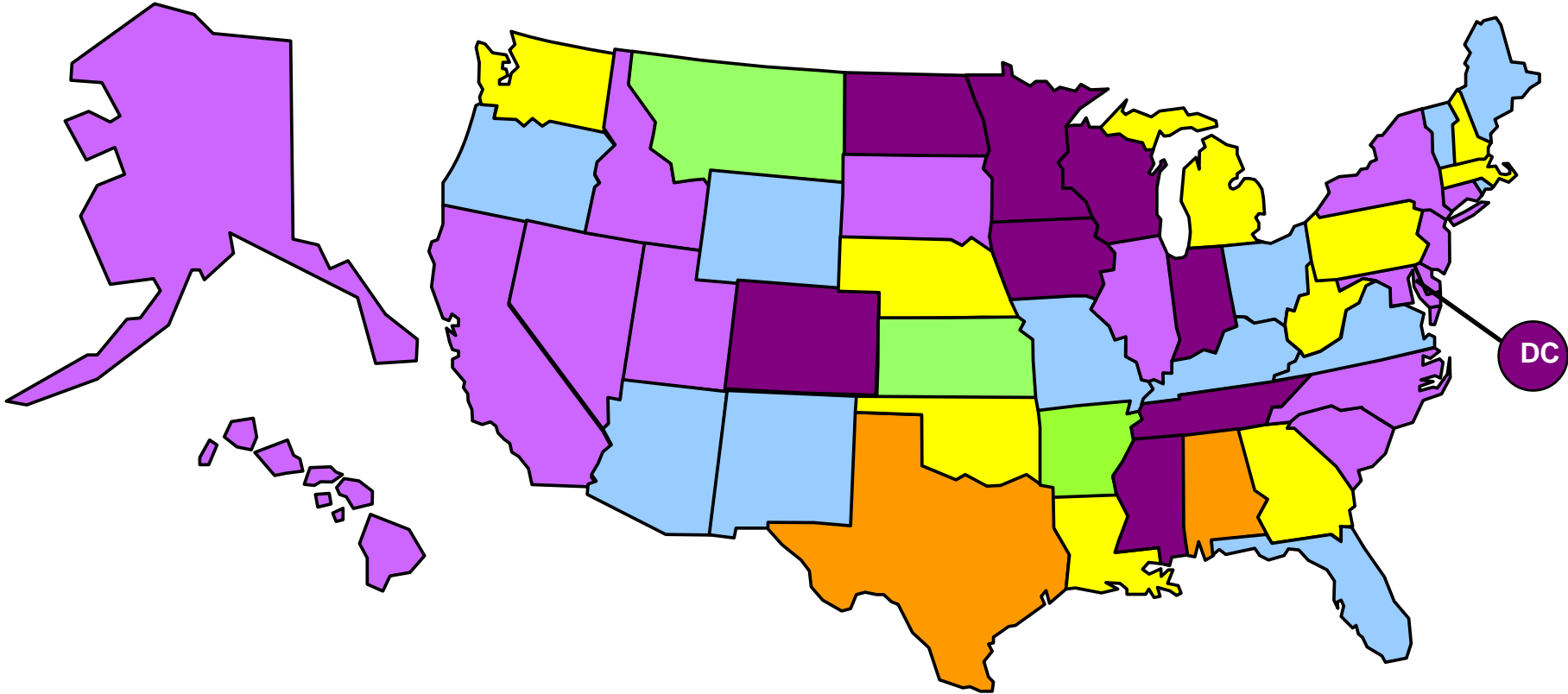
- 30-39 Disorders (12)
- 20-29 Disorders (2) (Conditions available as an option to selected population are not counted)
- 10-19 Disorders (10)
- <10 Disorders (3)



U.S. Newborn Screening

Conditions Required – June 1, 2006

- 40-49 Disorders (15)
- 30-39 Disorders (12)
- 20-29 Disorders (2) (Conditions available as an option to selected population are not counted)
- 10-19 Disorders (10)
- <10 Disorders (3)

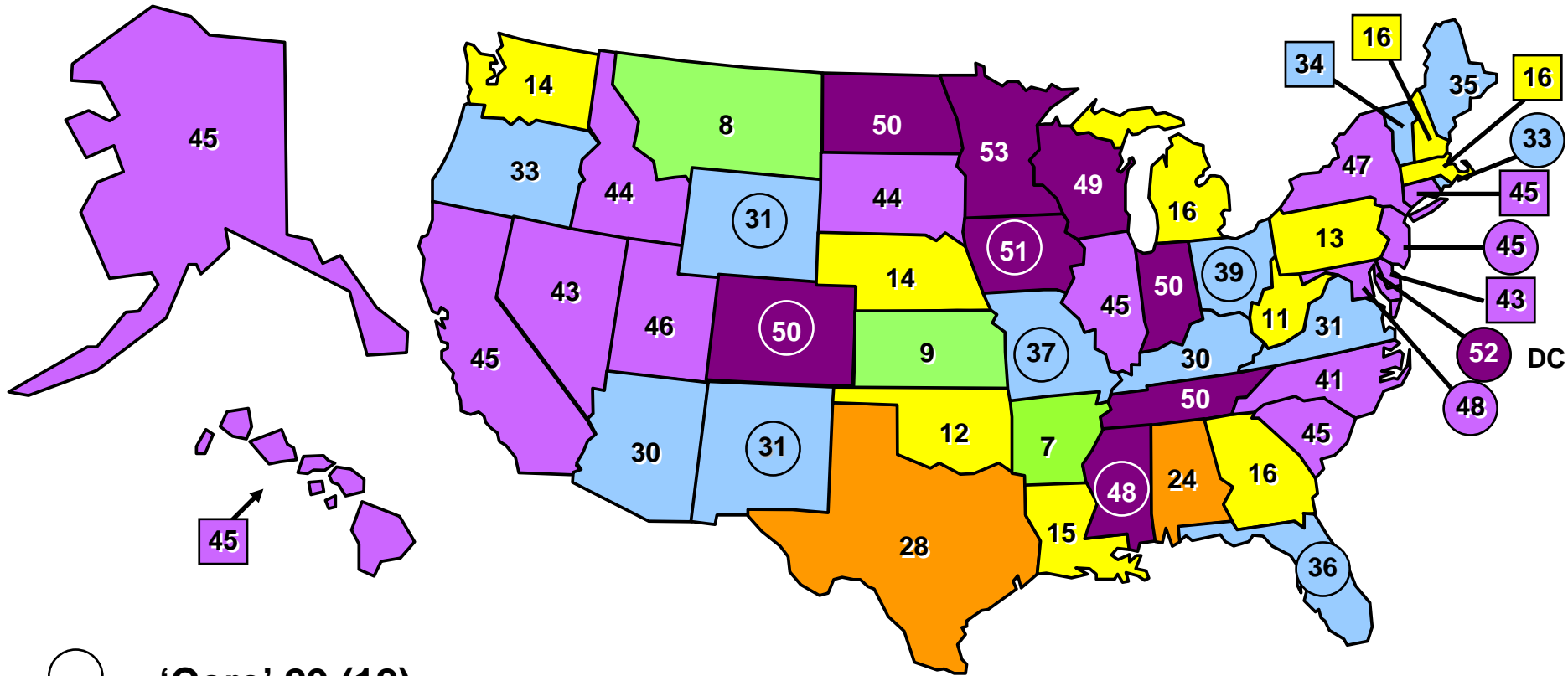


- 50+ Disorders (9)
- 40-49 Disorders (15)
- 30-39 Disorders (12)
- 20-29 Disorders (2)
- 10-19 Disorders (10)
- <10 Disorders (3)

U.S. Newborn Screening

Conditions Required – June 1, 2006

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

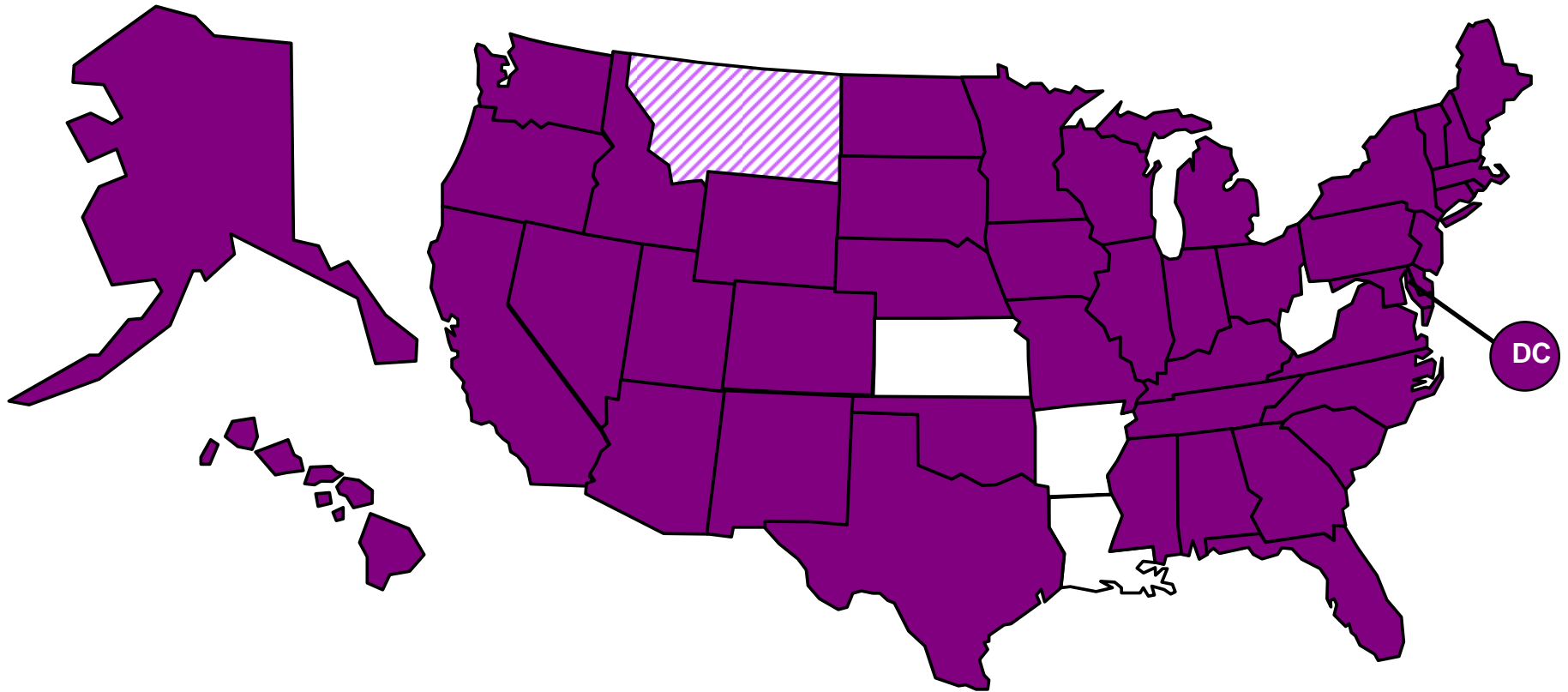


- 'Core' 29 (12)
- 50+ Disorders (9)
- 40-49 Disorders (15)
- 30-39 Disorders (12)
- 20-29 Disorders (2)
- 10-19 Disorders (10)
- <10 Disorders (3)

U.S. Newborn Screening

Conditions Required – June 1, 2006





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

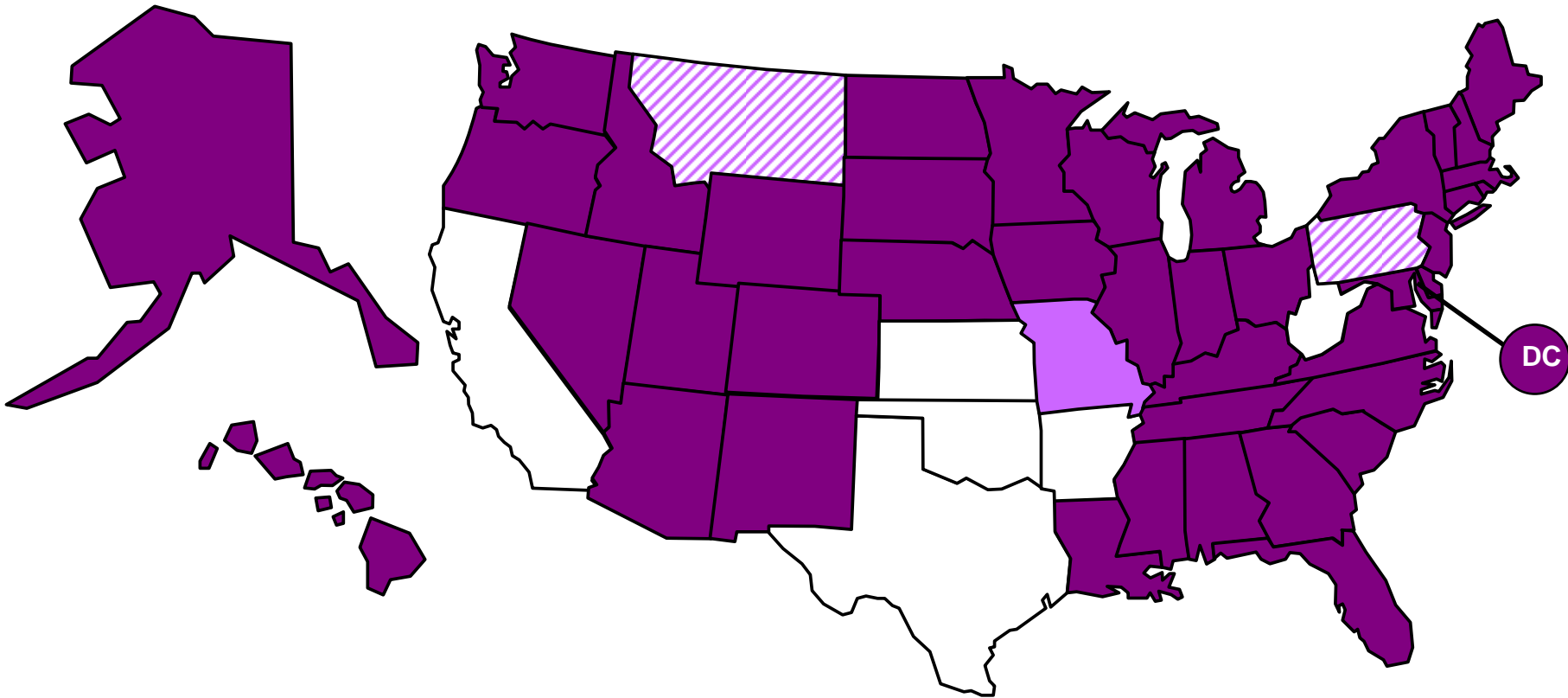


U.S. Newborn Screening

Congenital Adrenal Hyperplasia – June 1, 2006

Available to ~ 97% of US Newborns





-  Universally Required (46)
-  Required but not yet implemented (0)
-  Screening not required but available as option or pilot (1)
-  Screening not required or piloted (4)

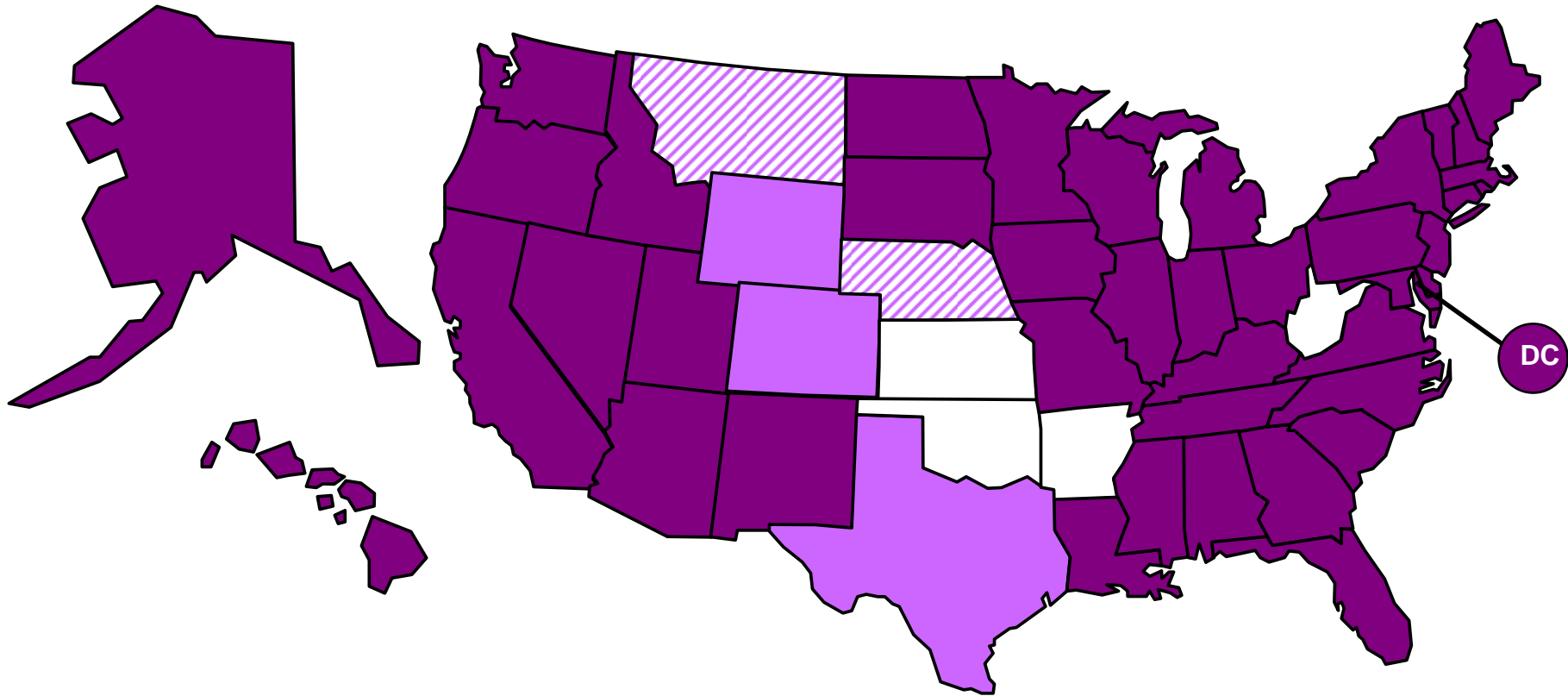


U.S. Newborn Screening

Biotinidase Deficiency – June 1, 2006

Available to 83% of US Newborns

-  Universally Required (42)
-  Required but not yet implemented (1)
-  Screening not required but available as option or pilot (2)
-  Screening not required or piloted (6)

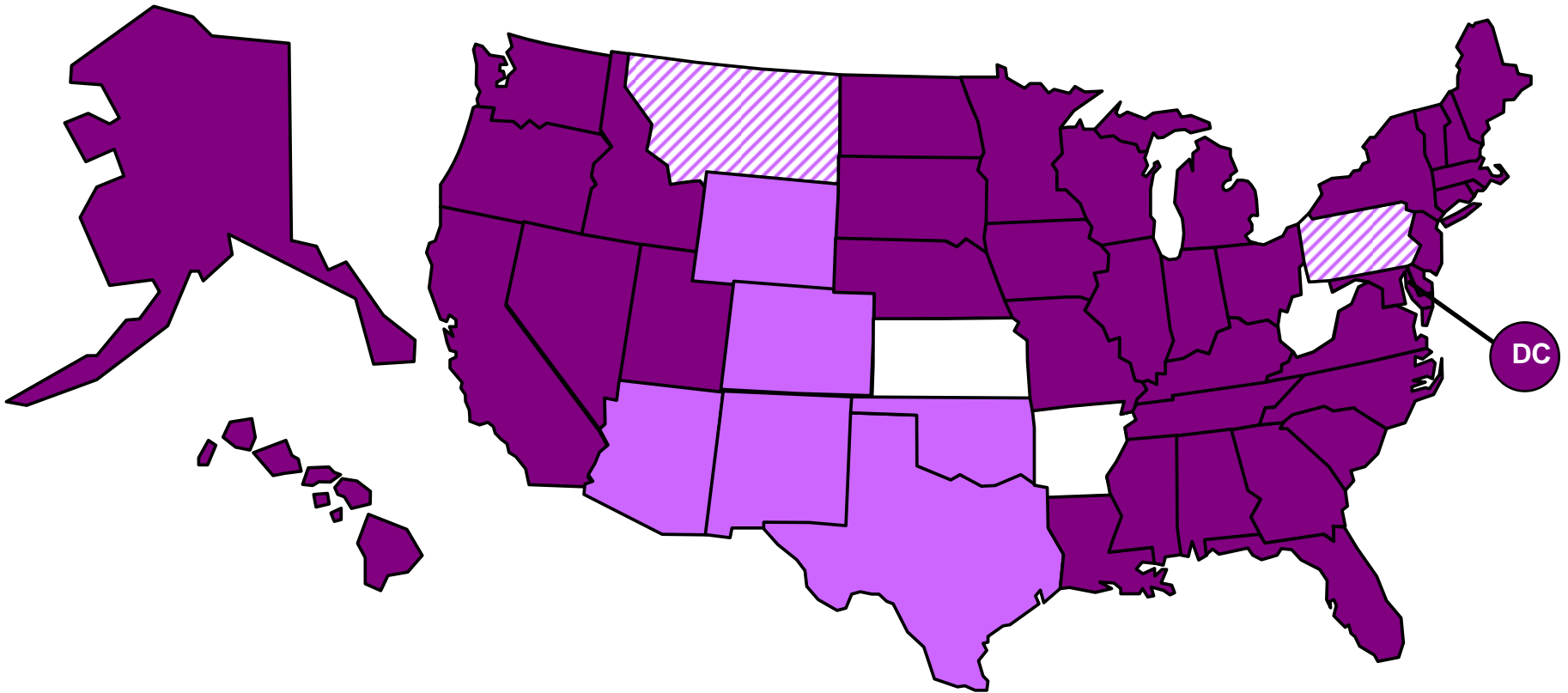


U.S. Newborn Screening

Maple Syrup Urine Disease – June 1, 2006





Available to ~ 96% of US Newborns

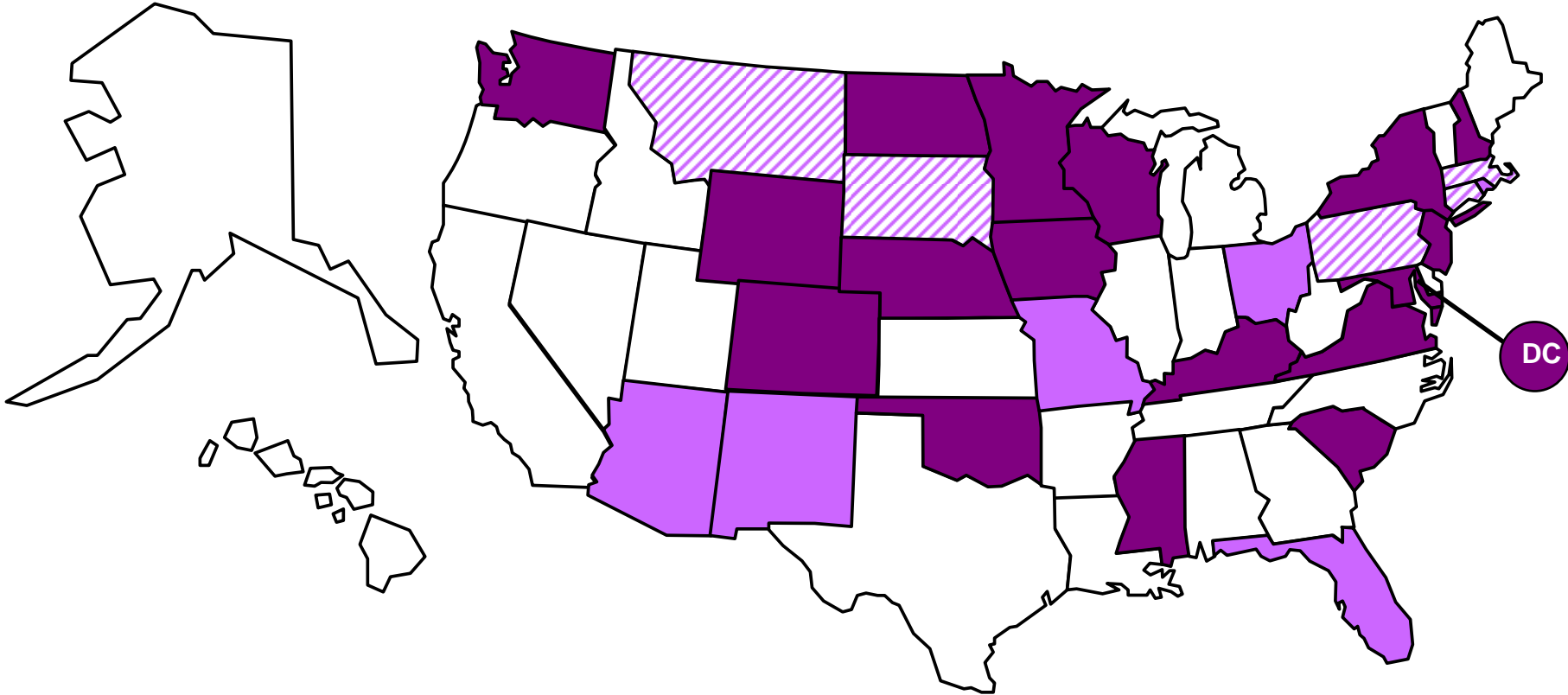
- Universally Required (43)
- Required but not yet implemented (3)
- Screening not required but available as option or pilot (2)
- Screening not required or piloted (3)



U.S. Newborn Screening

Tandem Mass Spectrometry – June 1, 2006
 (MCAD) Available to ~ 97% of US Newborns





-  Universally Required (40)
-  Required but not yet implemented (6)
-  Screening not required but available as option or pilot (2)
-  Screening not required or piloted (3)

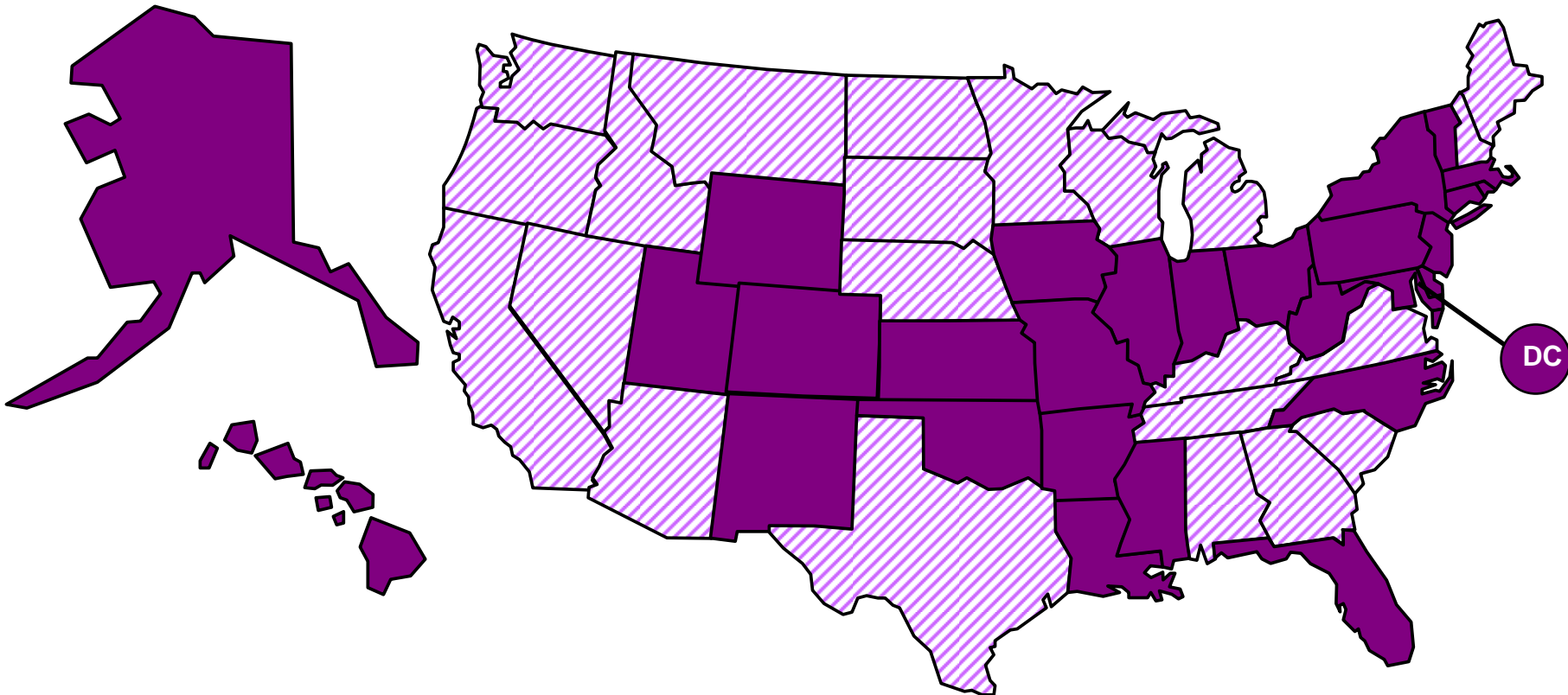


U.S. Newborn Screening

Cystic Fibrosis – June 1, 2006

Available to ~ 53% of US Newborns

-  Universally Required (18)
-  Required but not yet implemented (6)
-  Screening not required but available as option or pilot (5)
-  Screening not required or piloted (22)



U.S. Newborn Screening
Hearing Screening – June 1, 2006
Available to ~ 100% of US Newborns

- Universally Required (29)**
- Required but not yet implemented (0)**
- Screening not required but available as option or pilot (22)**
- Screening not required or piloted (0)**

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)
- Free long-term software available from OHSU
- Recall data in hearing screening
- Data sharing – patent issues



Thank You!

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

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

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