Status of Screening for Recommended Disorders in the US

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Association of Public Health Laboratories

The development of this presentation was supported by Cooperative Agreement #U22MC24078 from the Health Resources and Services Administration (HRSA). Its contents are solely the responsibility of the authors and do not necessarily represent the official views of HRSA.

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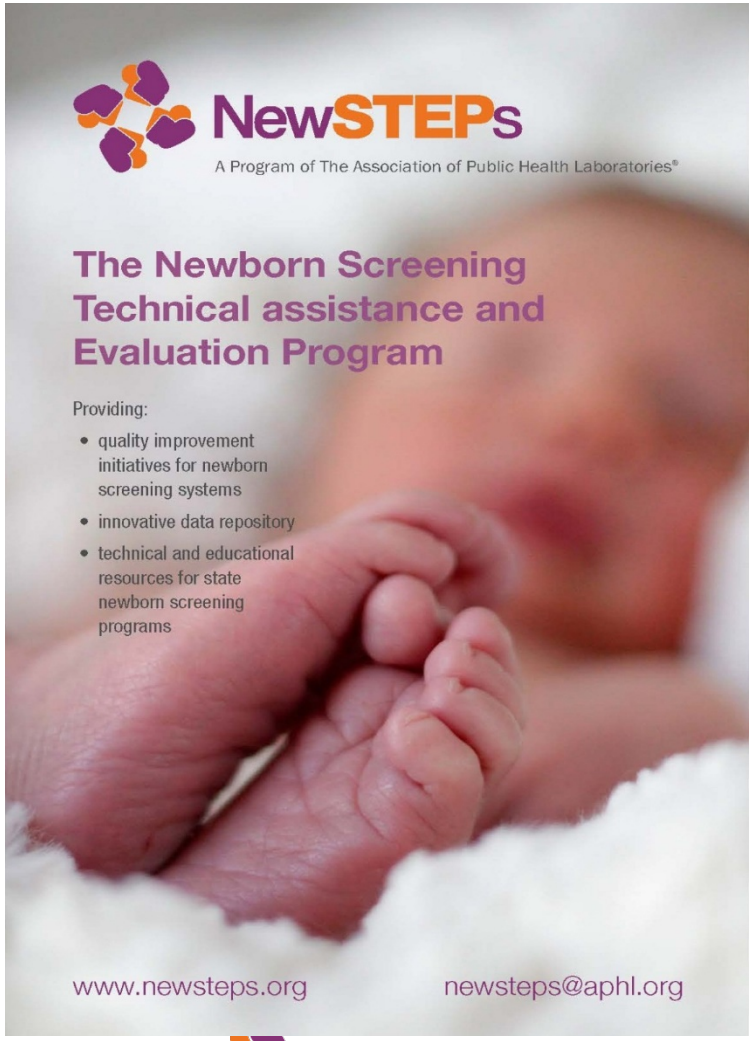
Vision

Dynamic newborn screening systems have access to and utilize accurate, relevant information to achieve and maintain excellence through continuous quality improvement.

Mission

To achieve the highest quality for newborn screening systems by providing relevant, accurate tools and resources and to facilitate collaboration between state programs and other newborn screening partners.



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Recommended Uniform Screening Panel

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ACMG Code	Core Condition	Metabolic Disorder			Endocrine Disorder	Hemoglobin Disorder	Other Disorder
		Organic acid condition	Fatty acid oxidation disorder	Amino acid disorder			
PROP	Propionic Acidemia	X					
MUT	Methylmalonic Acidemia (methylmalonyl-CoA mutase)	X					
Cbl A,B	Methylmalonic Acidemia (Cobalamin disorders)	X					
IVA	Isovaleric Acidemia	X					
3-MCC	3-Methylcrotonyl-CoA Carboxylase Deficiency	X					
HMG	3-Hydroxy-3-Methyglutaric Aciduria	X					
MCD	Holocarboxylase Synthase Deficiency	X					
ßKT	ß-Ketothiolase Deficiency	X					
GA1	Glutaric Acidemia Type I	X					
CUD	Carnitine Uptake Defect/Carnitine Transport Defect		X				
MCAD	Medium-chain Acyl-CoA Dehydrogenase Deficiency		X				
VLCAD	Very Long-chain Acyl-CoA Dehydrogenase Deficiency		X				
LCHAD	Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency		X				
TFP	Trifunctional Protein Deficiency		X				
ASA	Argininosuccinic Aciduria			X			
CIT	Citrullinemia, Type I			X			
MSUD	Maple Syrup Urine Disease			X			
HCY	Homocystinuria			X			
PKU	Classic Phenylketonuria			X			
TYR I	Tyrosinemia, Type I			X			
CH	Primary Congenital Hypothyroidism				X		
CAH	Congenital adrenal hyperplasia				X		
Hb SS	S,S Disease (Sickle Cell Anemia)					X	
Hb S/ßTh	S, βeta-Thalassemia					X	
Hb S/C	S,C Disease					X	
BIOT	Biotinidase Deficiency						X
CCHD	Critical Congenital Heart Disease						X
CF	Cystic Fibrosis						X
GALT	Classic Galactosemia						X
GSD II	Glycogen Storage Disease Type II (Pompe)						X
HEAR	Hearing Loss						X
SCID	Severe Combined Immunodeficiencies						X

RUSP Core Conditions (As of March 2015)

THE SECRETARY OF HEALTH AND HUMAN SERVICES
WASHINGTON, D.C. 20201

FEB 16 2016

Joseph A. Bocchini, Jr., M.D.
Committee Chairperson
Advisory Committee on Heritable
Disorders in Newborns and Children
5600 Fishers Lane
Room 18W68
Rockville, MD 20857

Dear Dr. Bocchini:

I want to take this opportunity to advise you of my decisions, taking into account the Interagency Coordinating Committee on Newborn and Child Screening's (ICC) review, regarding the Advisory Committee on Heritable Disorders in Newborns and Children's (ACHDNC) recommendations to add Mucopolysaccharidosis type I (MPS I) to the Recommended Uniform Screening Panel (RUSP) and to provide federal funding to state newborn screening programs to implement the screening of MPS I.

The ICC reviewed the ACH
new information from agenc
report to me, the ICC noted
screening for MPS I. Howe
help increase the number of
the disorder.

I would like to commend the
newborn screening for MPS I. The information from the ev
for Mucopolysaccharidosis Type I (MPS I), was taken
report.
reviewed the ICC's

Based on the information presented in these reports, I accept the ACHDNC's recommendation to add MPS I to the RUSP. The Affordable Care Act requires that most health plans cover the evidence-based preventive care and screenings provided for in the comprehensive guidelines supported by Health Resources and Service Administration (HRSA). Because the RUSP is a component of these guidelines, a condition added to the RUSP must be covered without cost-sharing. Plans and insurers will have until the first plan year that is one year after the date of adoption of the recommendation to implement coverage. However, it should be understood that addition of MPS I to the RUSP does not constitute a requirement for states to implement screening, only a recommendation.

I accept the ACHDNCS's recommendation to add MPS I to the RUSP.

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Joseph A. Bocchini, Jr., M.D.
Committee Chairperson
Advisory Committee on Heritable
Disorders in Newborns and Children
5600 Fishers Lane
Room 18W68
Rockville, MD 20857

Dear Dr. Bocchini:

I accept the ACHDNCS's recommendation to expand the RUSP to include the addition of X-ALD.

benefits and harms of newborn scree
X-ALD as well as the capability of state newborn screening programs to offer com
and follow-up services for infants identified with X-ALD. After reviewing the
report, *Newborn Screening for X-Linked Adrenoleukodystrophy (X-ALD): A Syst
ic Review of Evidence*, and taking into consideration the utility of current screening technologies, treatment for X-ALD, and the impact on public health systems, I accept the ACHDNC's recommendation to expand the RUSP to include the addition of X-ALD. As you may know the Affordable Care Act requires that most health plans cover without cost-sharing certain children's preventive services. Because the RUSP is a component of preventive services guidelines supported by the Health Resources and Services Administration, a condition added to the RUSP must be covered without cost sharing. I also want to clarify that the addition of X-ALD to the RUSP does not constitute a requirement for states to implement screening and is only a recommendation.

At this time, I am unable to identify new funding consistent with the ACHDNC's second recommendation to provide funding to state newborn screening programs to implement screening of X-ALD. However, I recognize the ongoing challenges that state newborn screening programs are experiencing in maintaining robust quality programs with the increasing demands of adding new conditions. This is why I have asked federal agencies to consider ways within their existing research and technical assistance resources to support state programs as they begin to implement comprehensive population-based screening for X-ALD.

ACMG Code	Secondary Condition	Metabolic Disorder			Hemoglobin Disorder	Other Disorder
		Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders		
Cbl C,D	Methylmalonic acidemia with homocystinuria	X				
MAL	Malonic acidemia	X				
IBG	Isobutyrylglycinuria	X				
2MBG	2-Methylbutyrylglycinuria	X				
3MGA	3-Methylglutaconic aciduria	X				
2M3HBA	2-Methyl-3-hydroxybutyric aciduria	X				
SCAD	Short-chain acyl-CoA dehydrogenase deficiency		X			
M/SCHAD	Medium/short-chain L-3-hydroxyacl-CoA dehydrogenase deficiency		X			
GA2	Glutaric acidemia type II		X			
MCAT	Medium-chain ketoacyl-CoA thiolase deficiency		X			
DE RED	2,4 Dienoyl-CoA reductase deficiency		X			
CPT IA	Carnitine palmitoyltransferase type I deficiency		X			
CPT II	Carnitine palmitoyltransferase type II deficiency		X			
CACT	Carnitine acylcarnitine translocase deficiency		X			
ARG	Argininemia			X		
CIT II	Citrullinemia, type II			X		
MET	Hypermethioninemia			X		
H-PHE	Benign hyperphenylalaninemia			X		
BIOPT (BS)	Biopterin defect in cofactor biosynthesis			X		
BIOPT (REG)	Biopterin defect in cofactor regeneration			X		
TYR II	Tyrosinemia, type II			X		
TYR III	Tyrosinemia, type III			X		
Var Hb	Various other hemoglobinopathies				X	
GALE	Galactoepimerase deficiency					X
GALK	Galactokinase deficiency					X
	T-cell related lymphocyte deficiencies					X

1. Selection of conditions based upon "Newborn Screening: Towards a Uniform Screening Panel and System." *Genetic Med.* 2006; 8(5) Suppl: S12-S252" as authored by the American College of Medical Genetics (ACMG) and commissioned by the Health Resources and Services Administration (HRSA).
2. Disorders that can be detected in the differential diagnosis of a core disorder.
3. Nomenclature for Conditions based upon "Naming and Counting Disorders (Conditions) Included in Newborn Screening Panels." *Pediatrics.* 2006; 117 (5) Suppl: S308-S314.

RUSP Secondary Conditions (As of March 2015)

Status of Screening of Core RUSP Conditions

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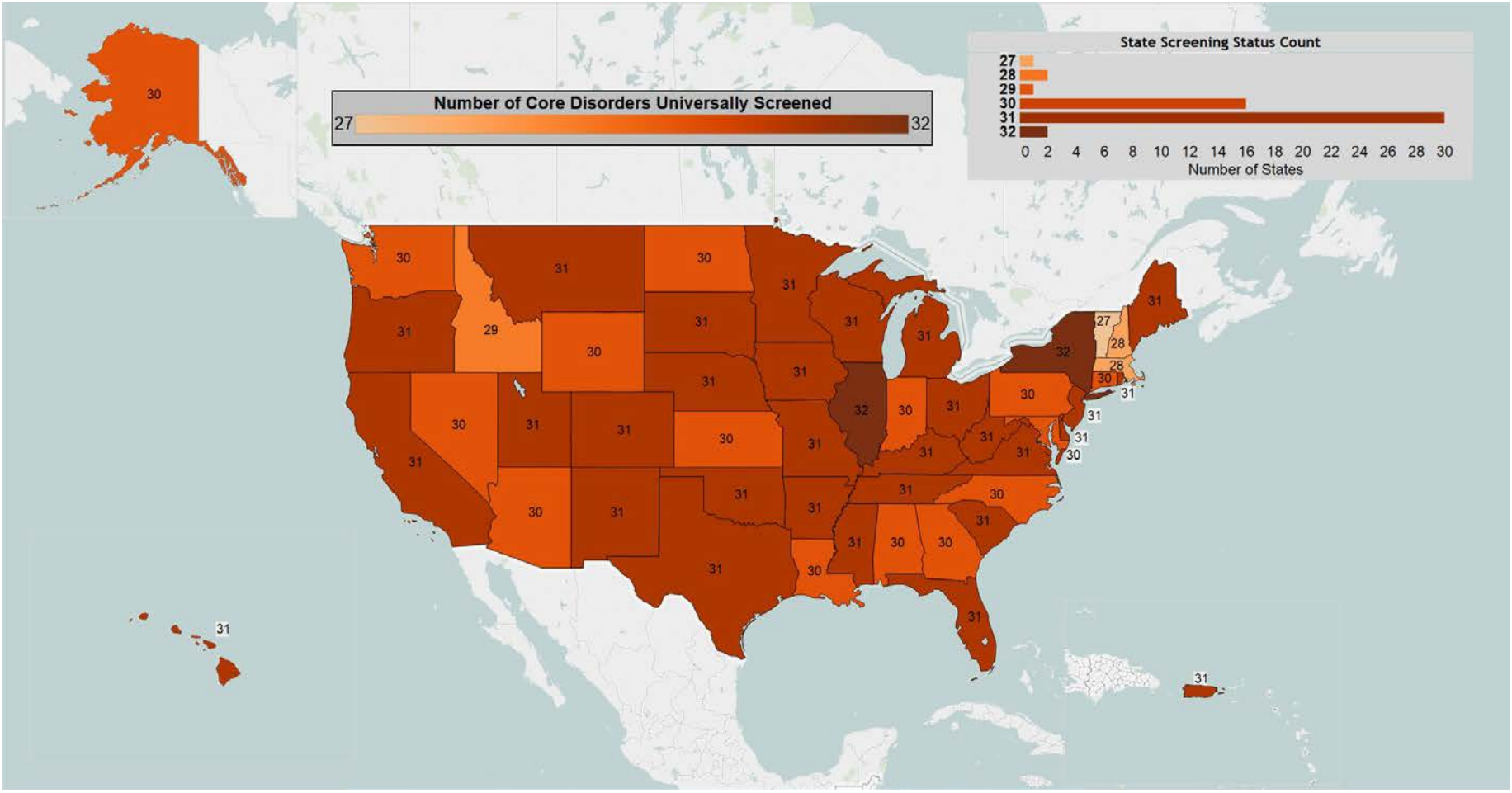
Universal Screening Status of the 32 Core Disorders (January 2016- does not include MPS 1 and x-ALD)



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Status of Screening of Newly Added RUSP Conditions

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Progress In SCID NBS Implementation

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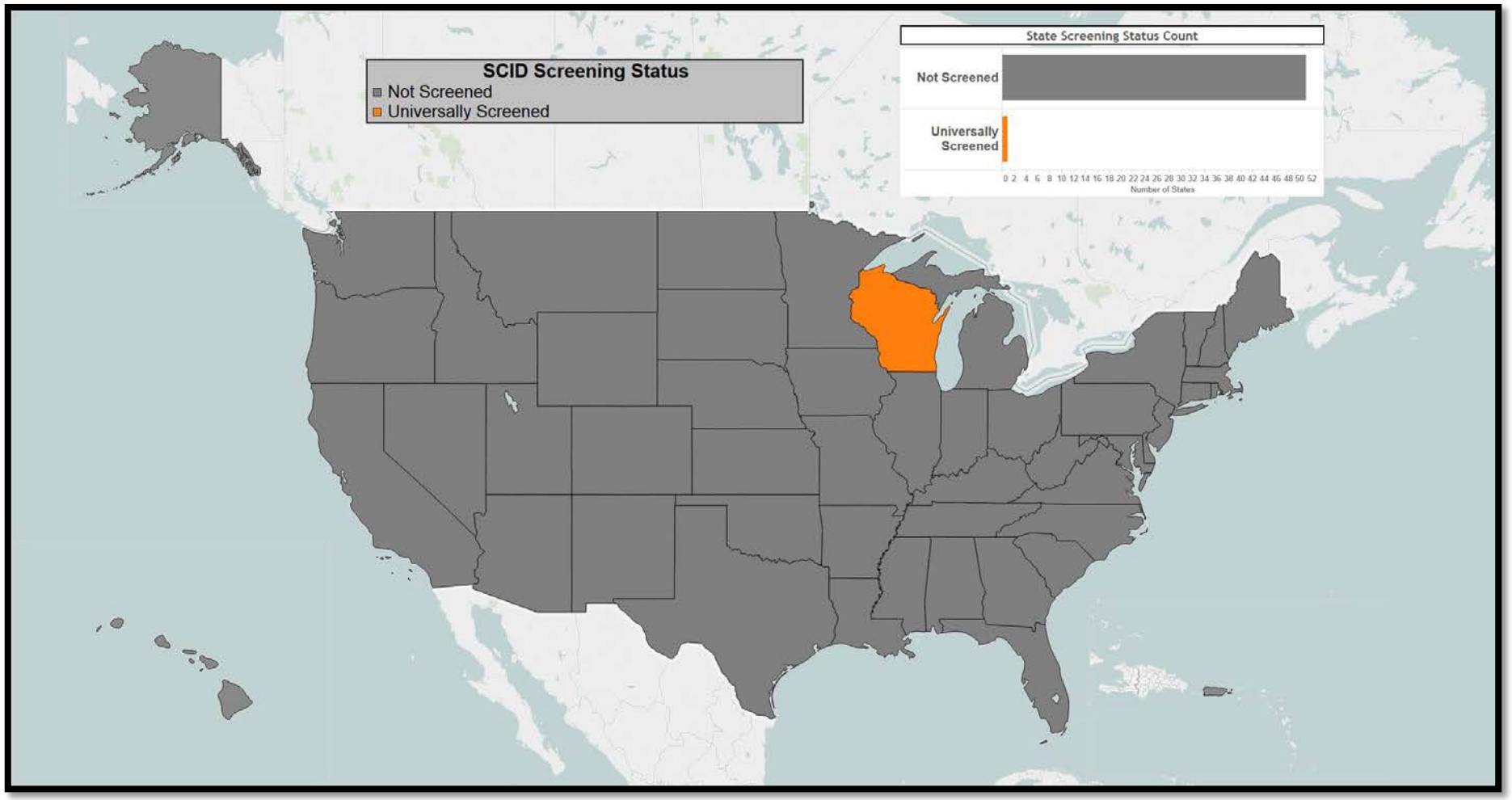
2008



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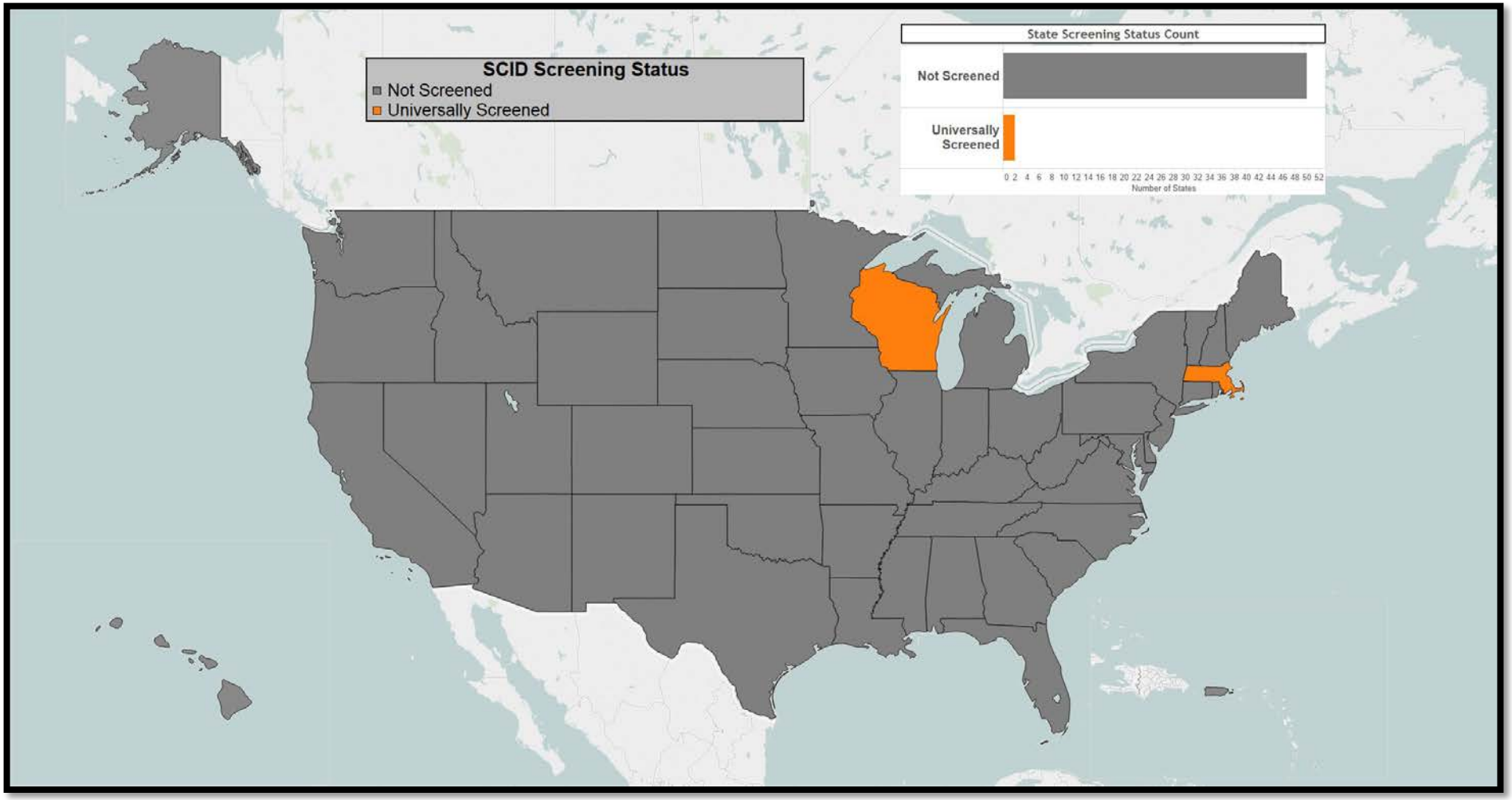
2009



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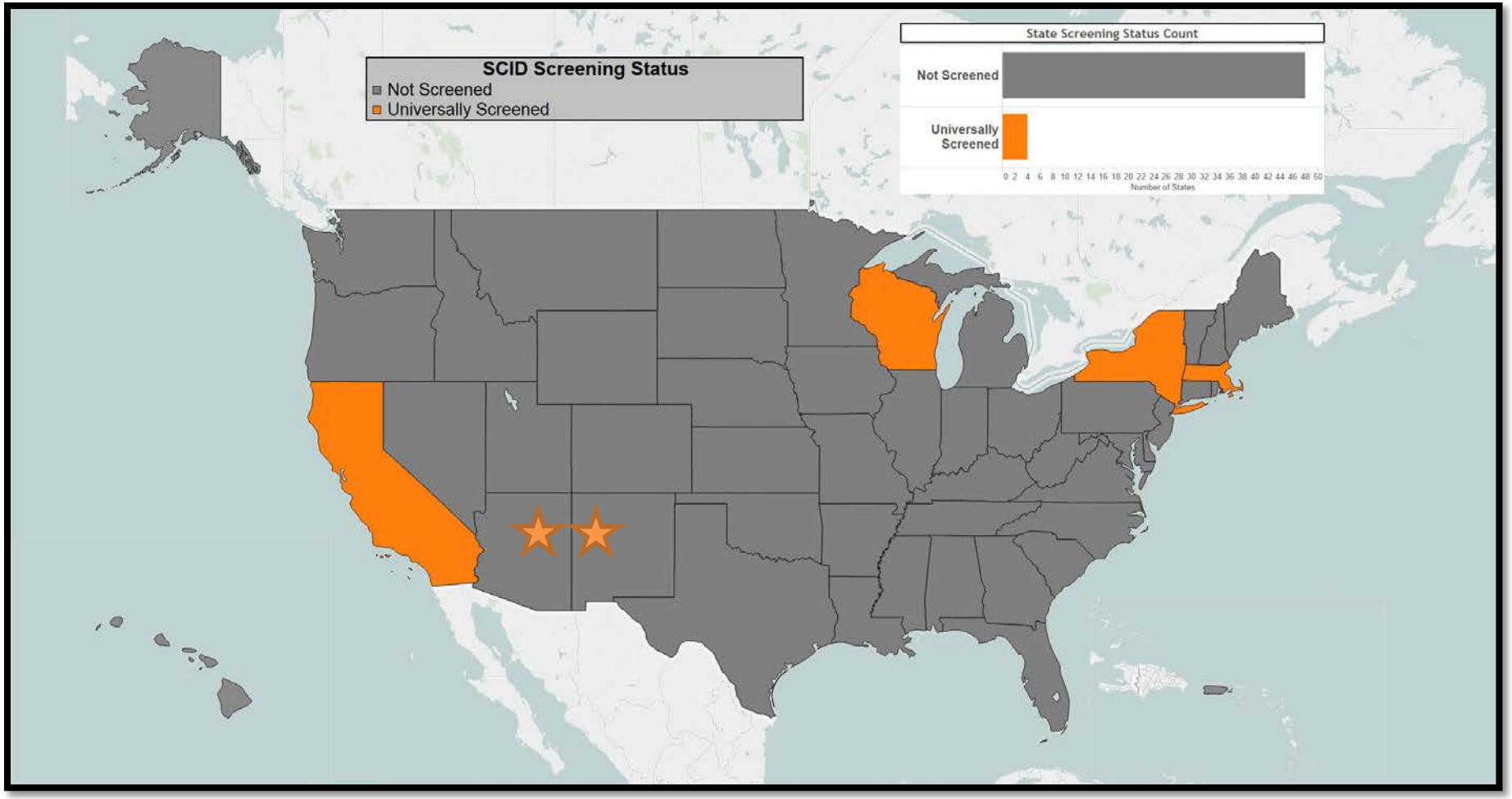
2010



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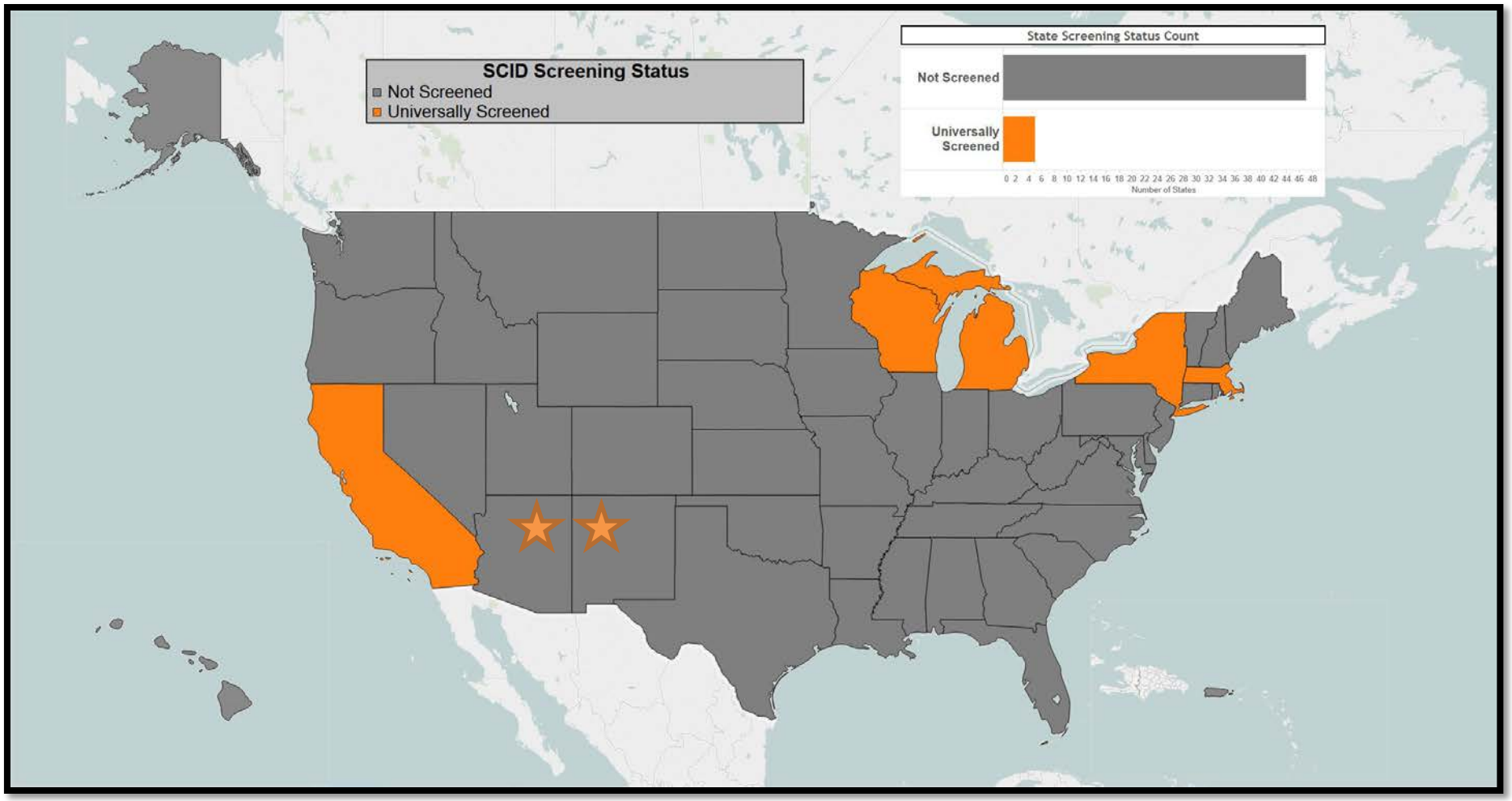
2011



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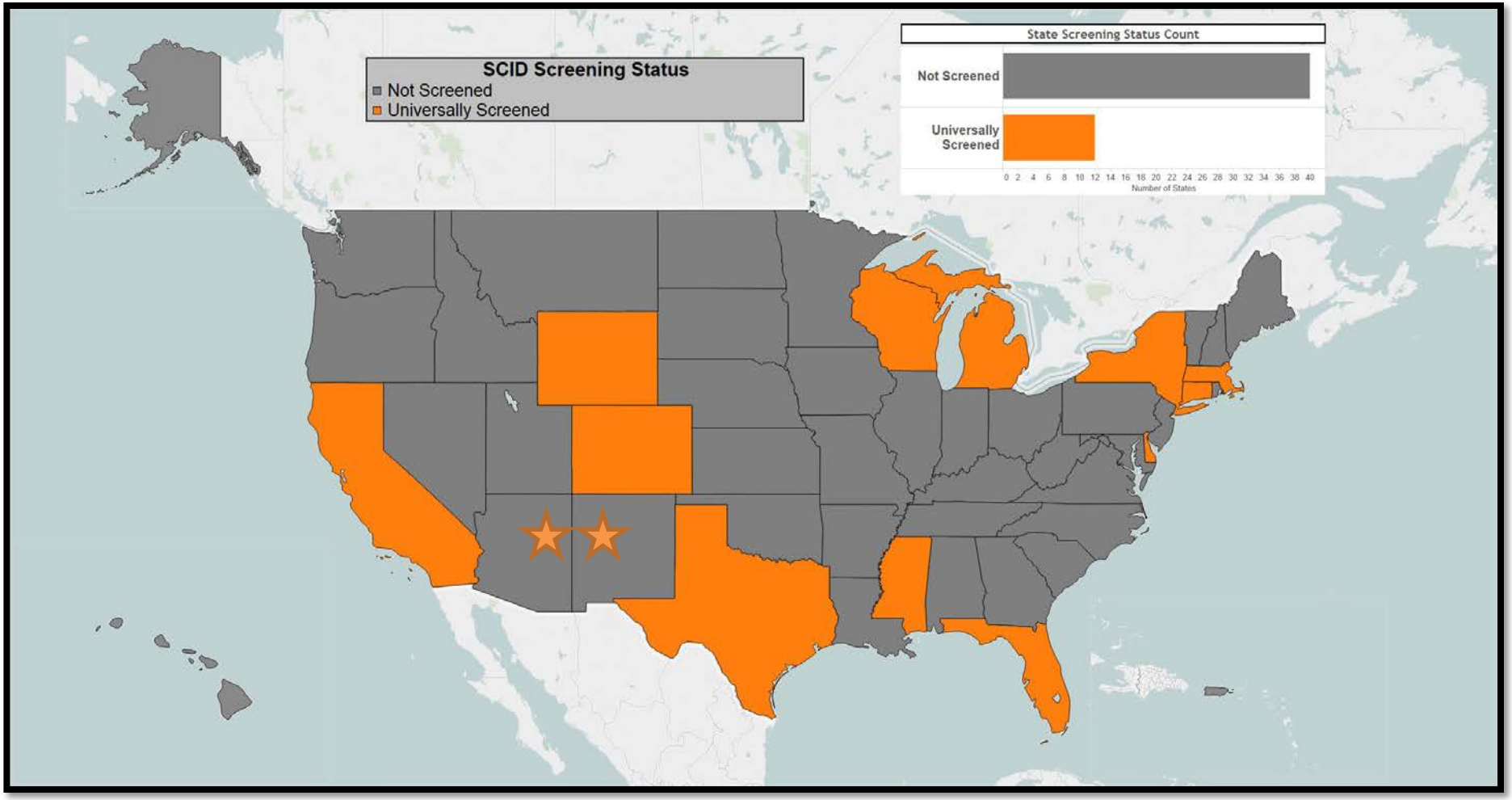
2012



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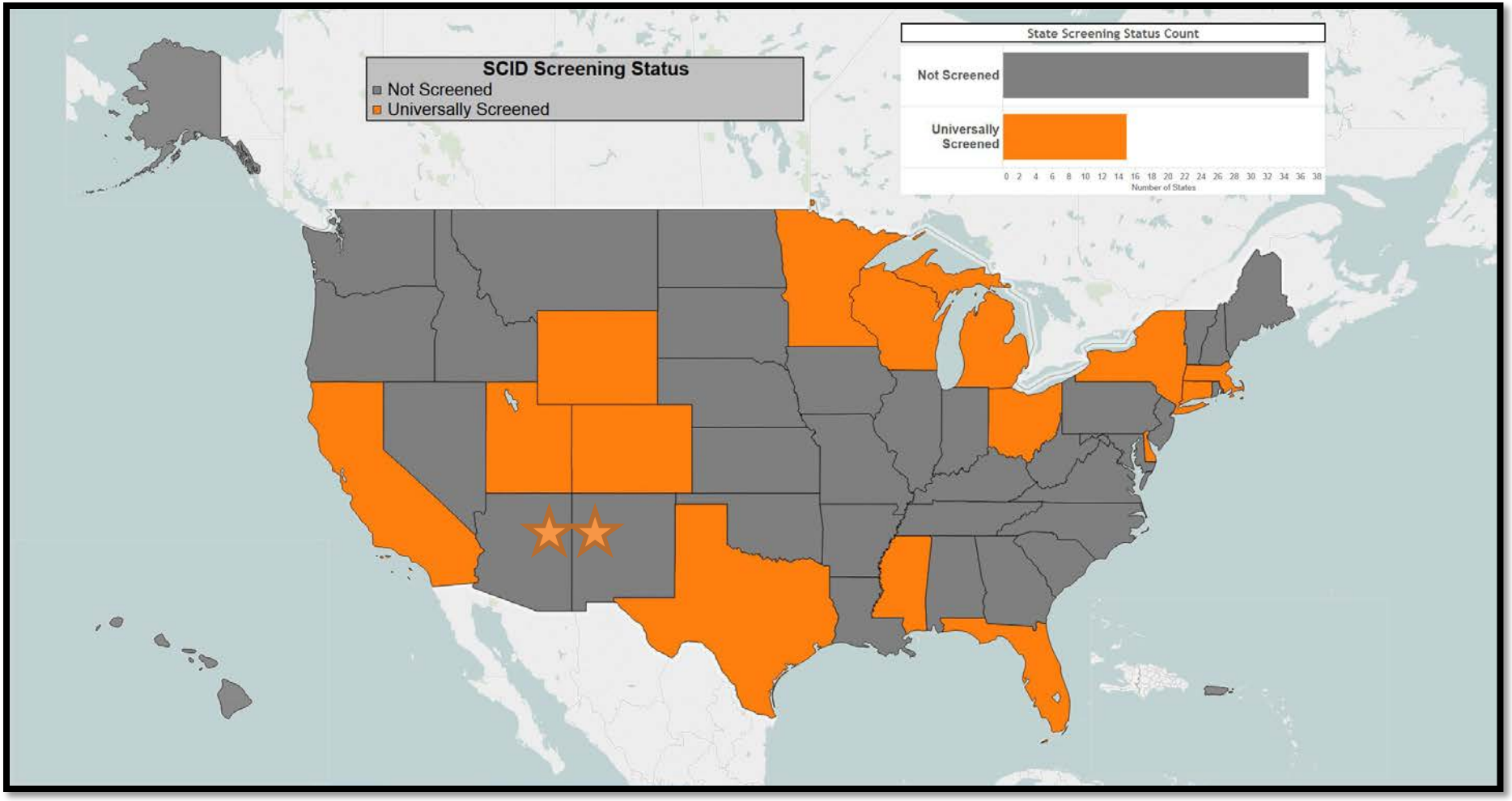
2013



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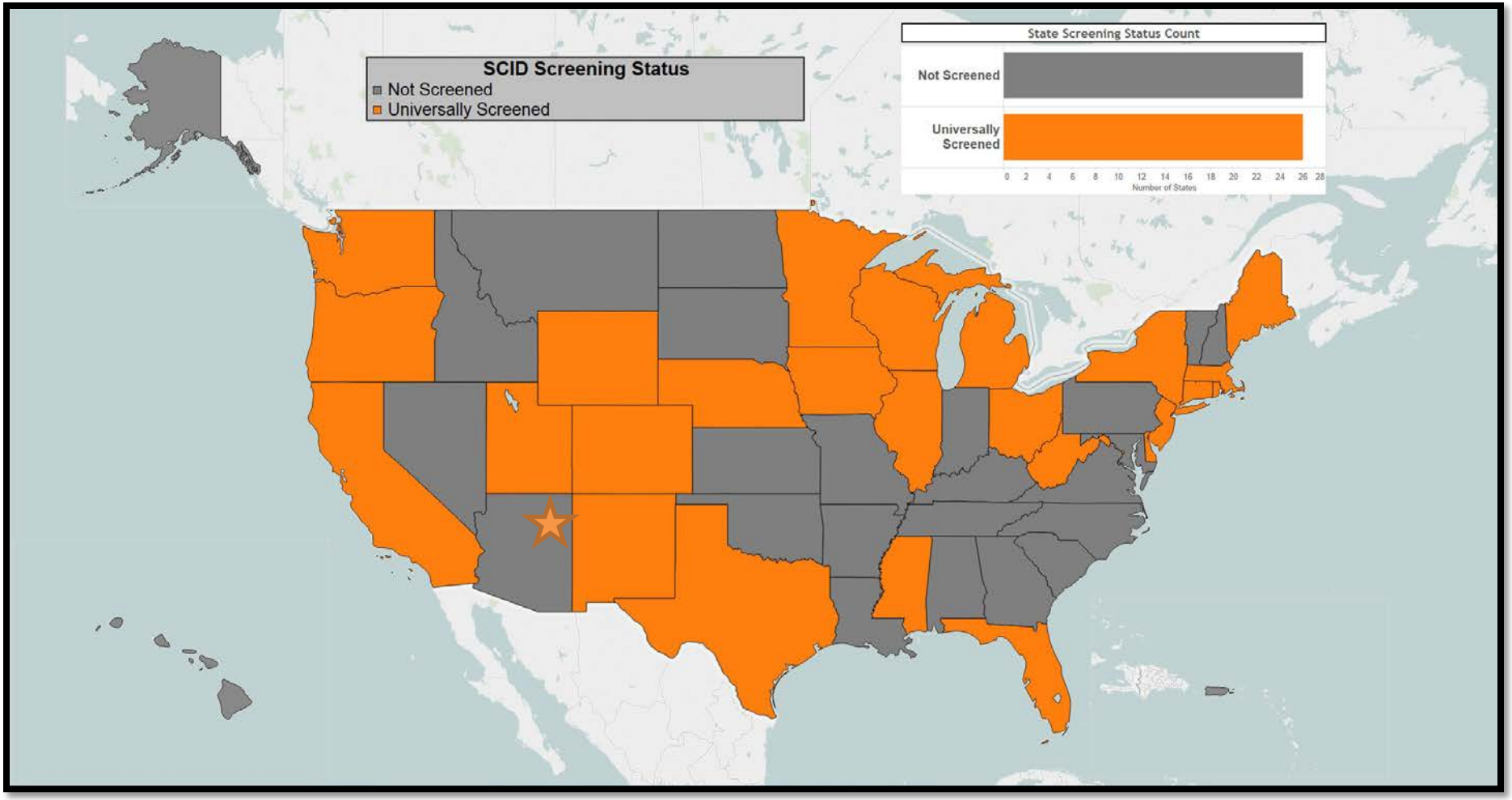
2014



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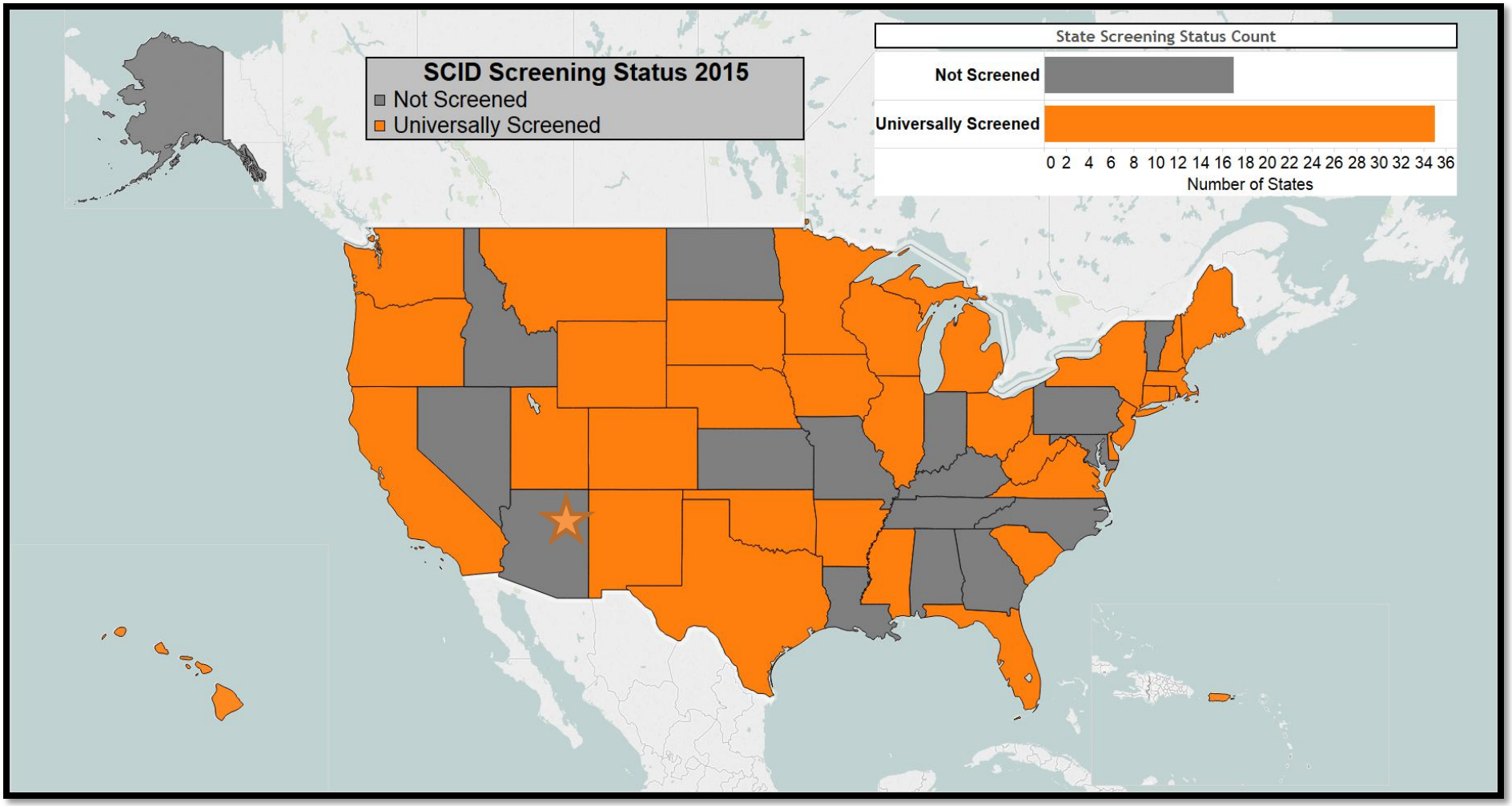
2015



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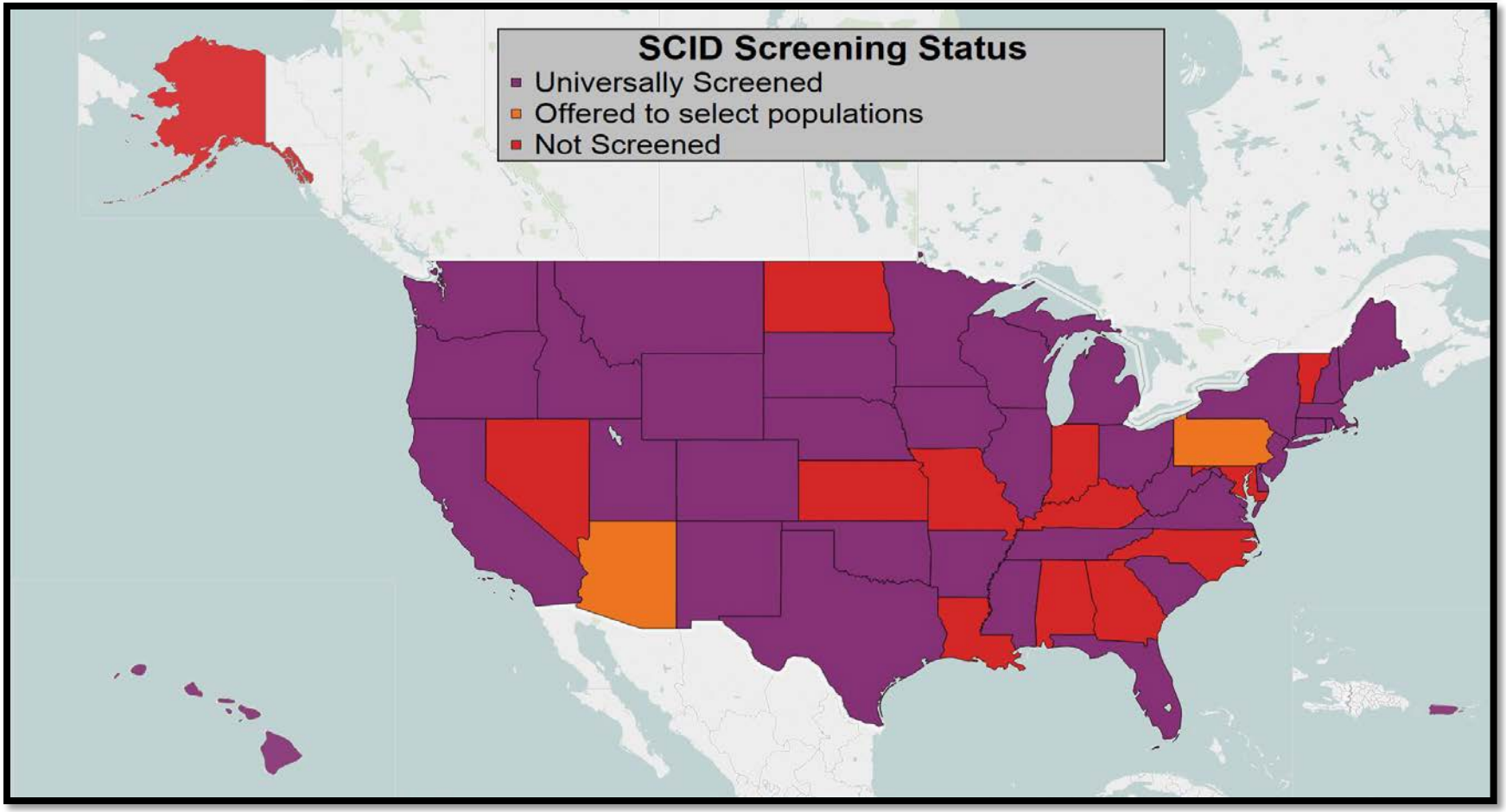
2016

SCID Newborn Screening Status Chart

Screening Status	Regulatory Details	Implementation Details	Summary Count
Universally Screened	Required by law or rule	N/A	34
	Not required by law or rule	N/A	3
Offered to select populations	N/A	N/A	2
Not Screened	Pursuing implementation	Performing validation/pilot studies	1
		Required but not yet fully implemented	3
		Seeking authorization/funding	8
	Data Not Provided	Data Not Provided	1

0 2 4 6 8 10 12 14 16 18 20 22 24 26 28 30 32 34 36
Number of States



Progress in CCHD NBS Implementation

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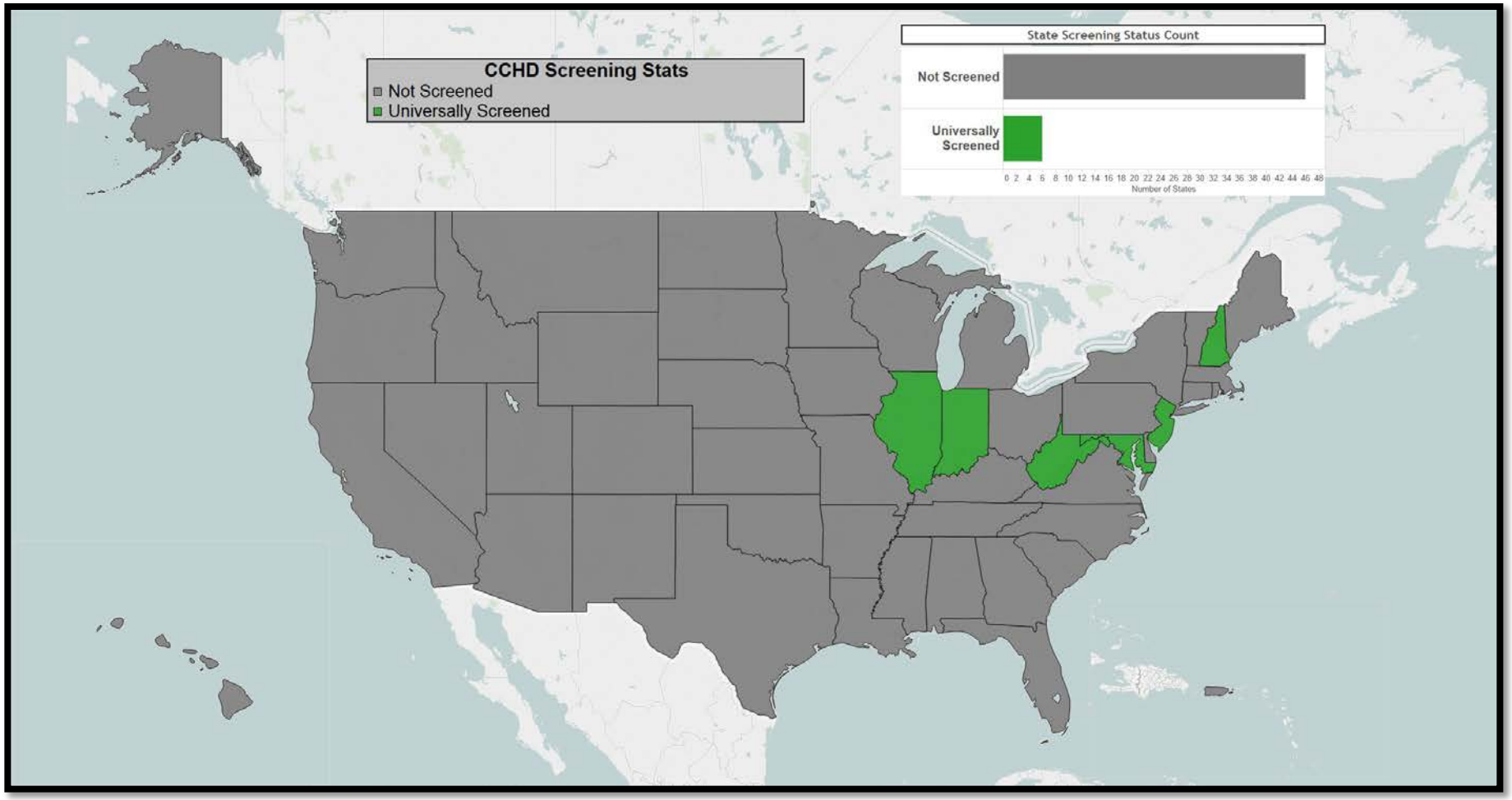
2012



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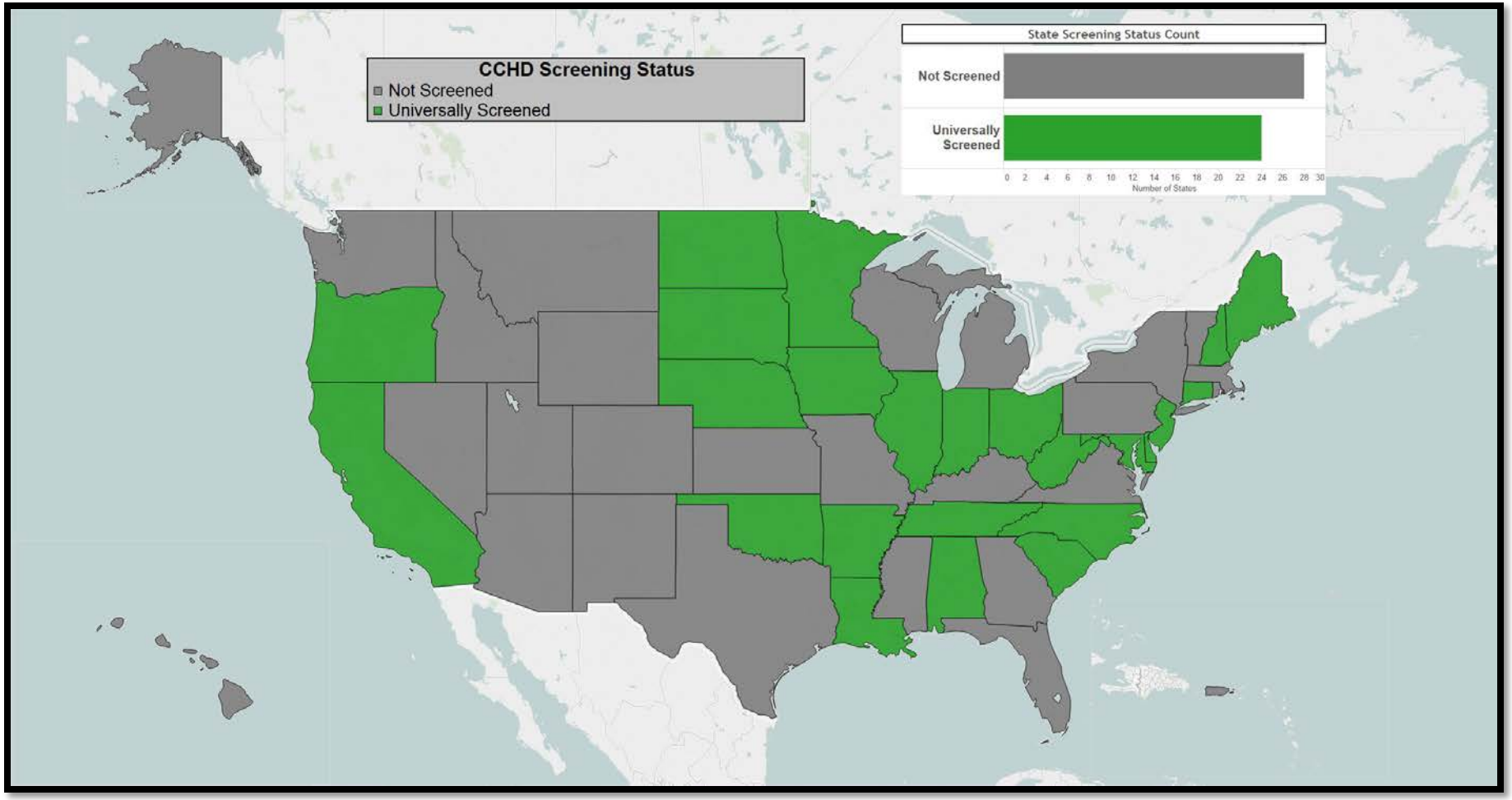
2013



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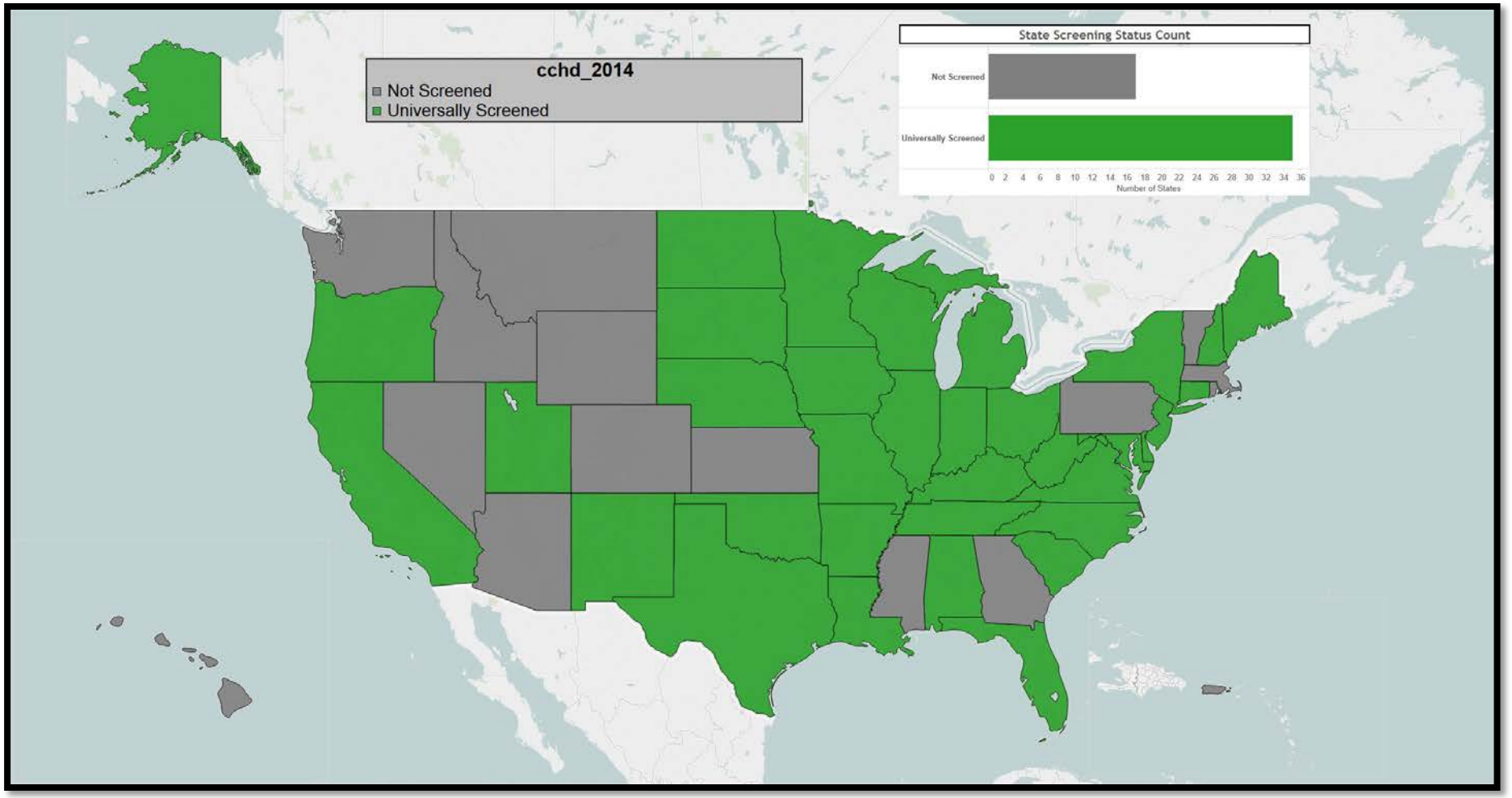
2014



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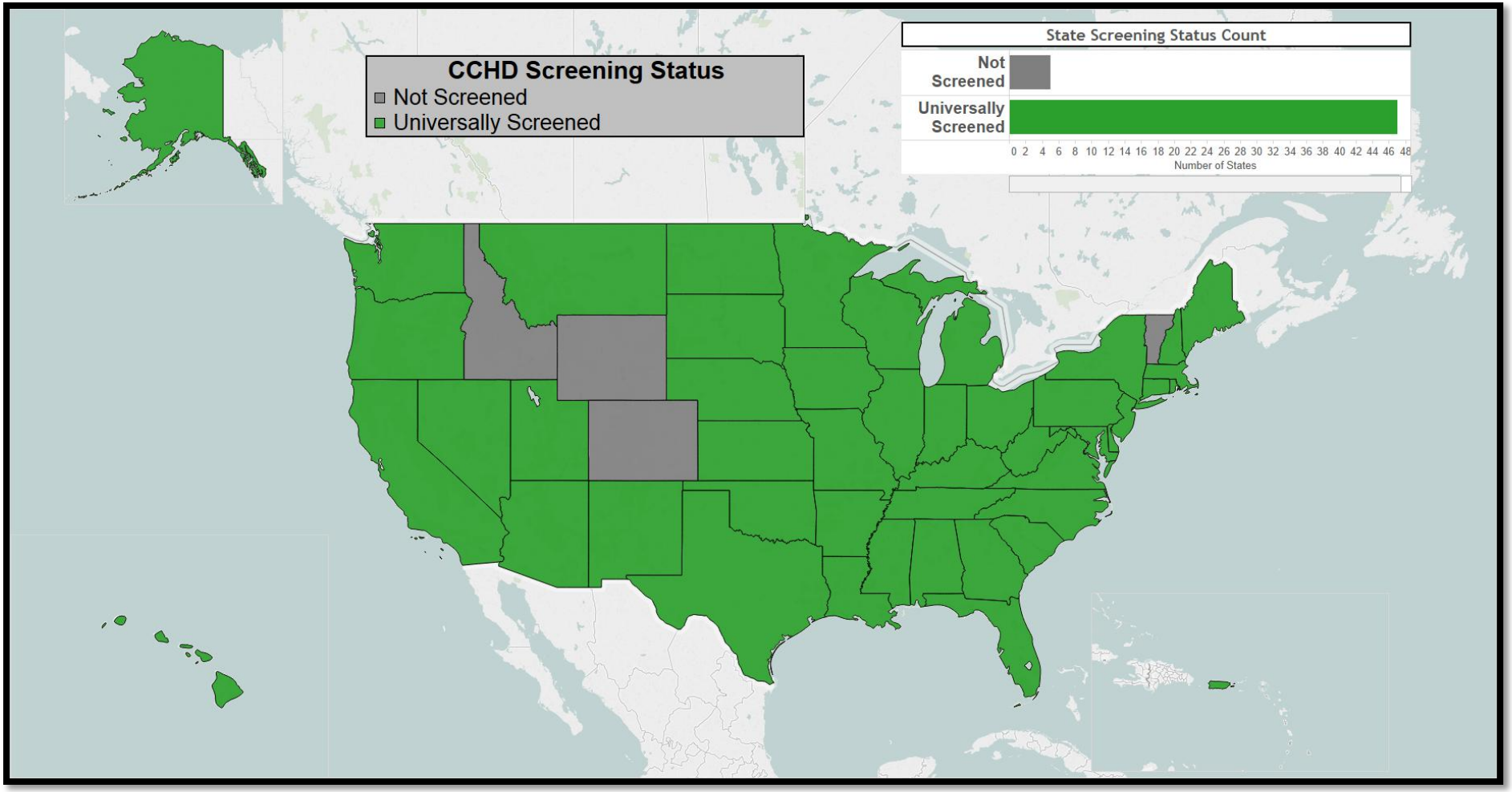
2015



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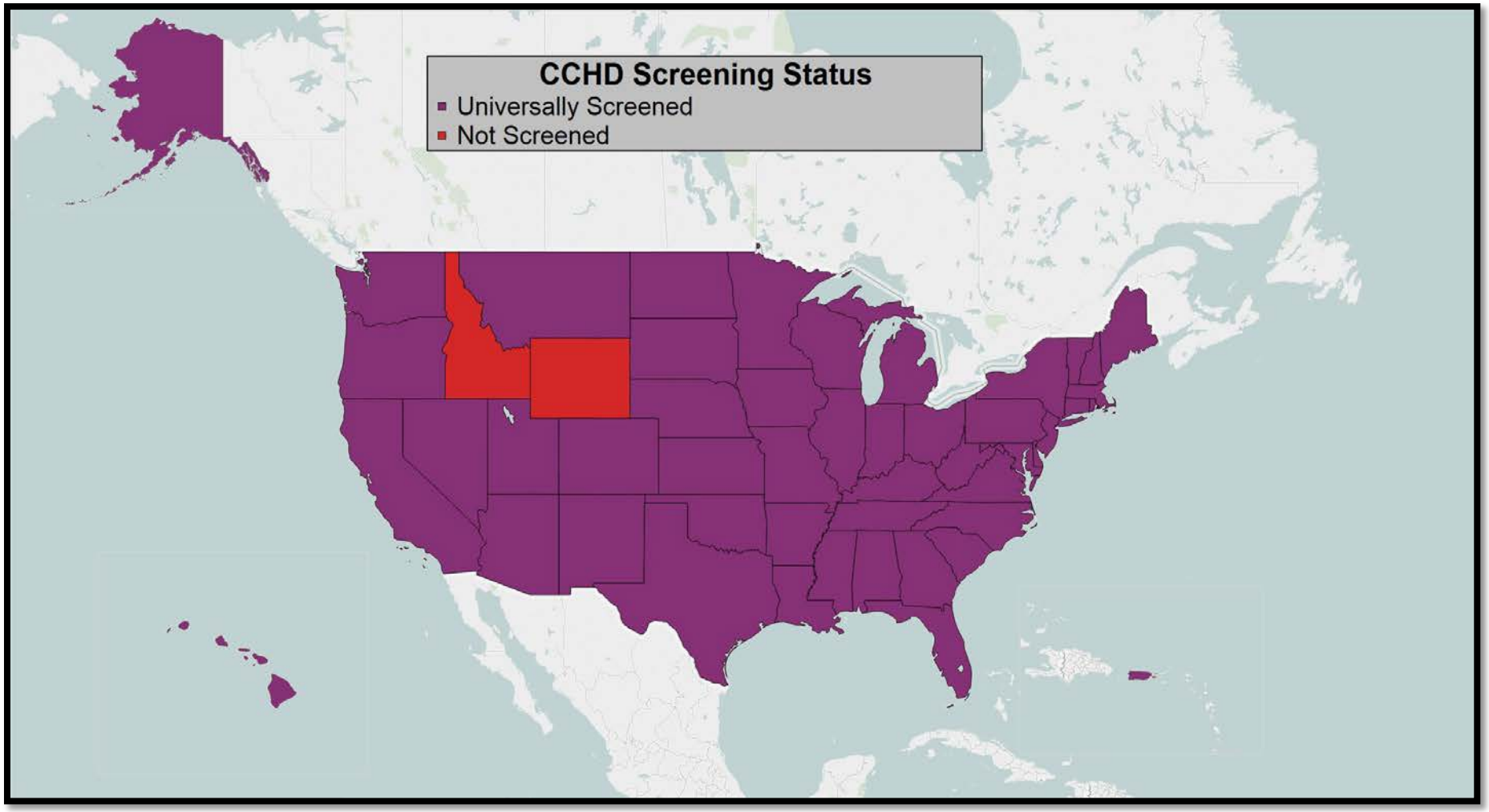
2016

CCHD Newborn Screening Status Chart

Screening Status	Regulatory Details	Implementation Details	Summary Count
Universally Screened	Not required by law or rule	N/A	2
	Data Not Provided	N/A	
	Required by law or rule	N/A	44
Not Screened	Pursuing implementation	Seeking authorization/funding	1
	Data Not Provided	Data Not Provided	1

0 2 4 6 8 10 12 14 16 18 20 22 24 26 28 30 32 34 36 38 40 42 44 46 48
Number of States



Progress in Pompe NBS Implementation

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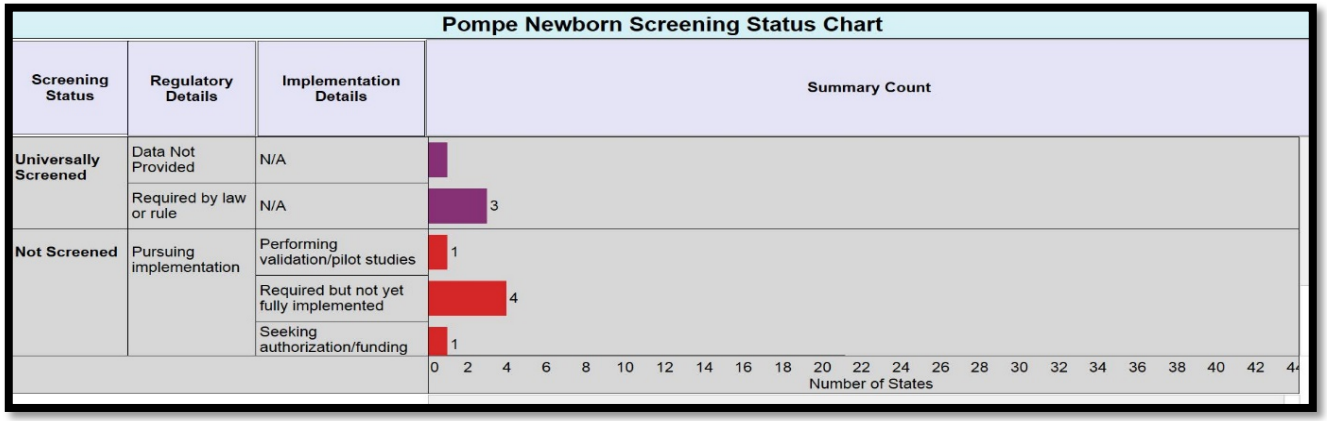
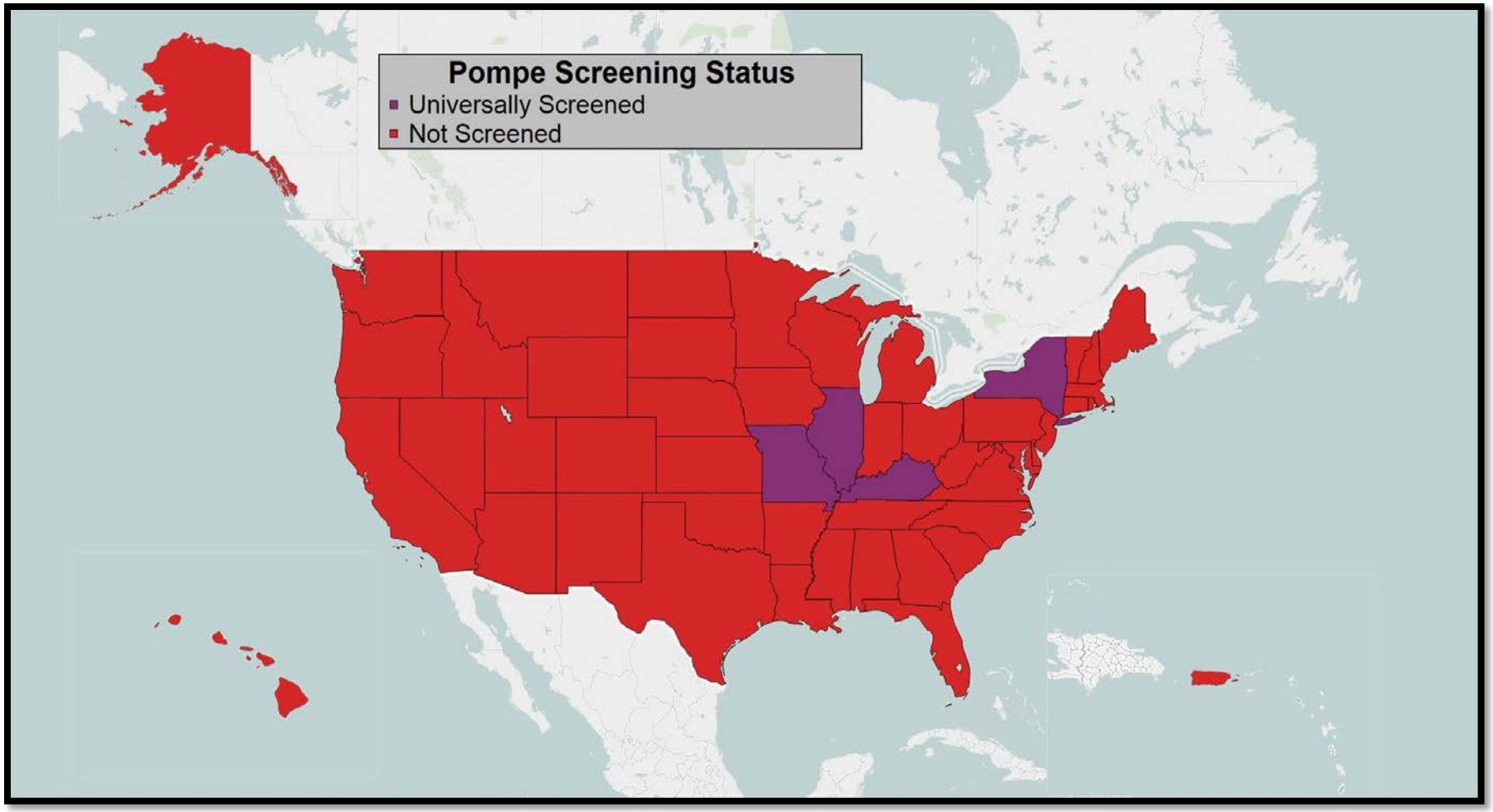
2016


Pompe Newborn Screening Status Chart
Screening Status
Regulatory Details
Implementation Details
Summary Count
Universally Screened
Data Not Provided
N/A
Required by law or rule
N/A
3
Not Screened
Pursuing implementation
Performing validation/pilot studies
1
Required but not yet fully implemented
4
Seeking authorization/funding
1
0 2 4 6 8 10 12 14 16 18 20 22 24 26 28 30 32 34 36 38 40 42
Number of States


Pompe Screening Status
Universally Screened
Not Screened

Progress in MPS-1 NBS Implementation

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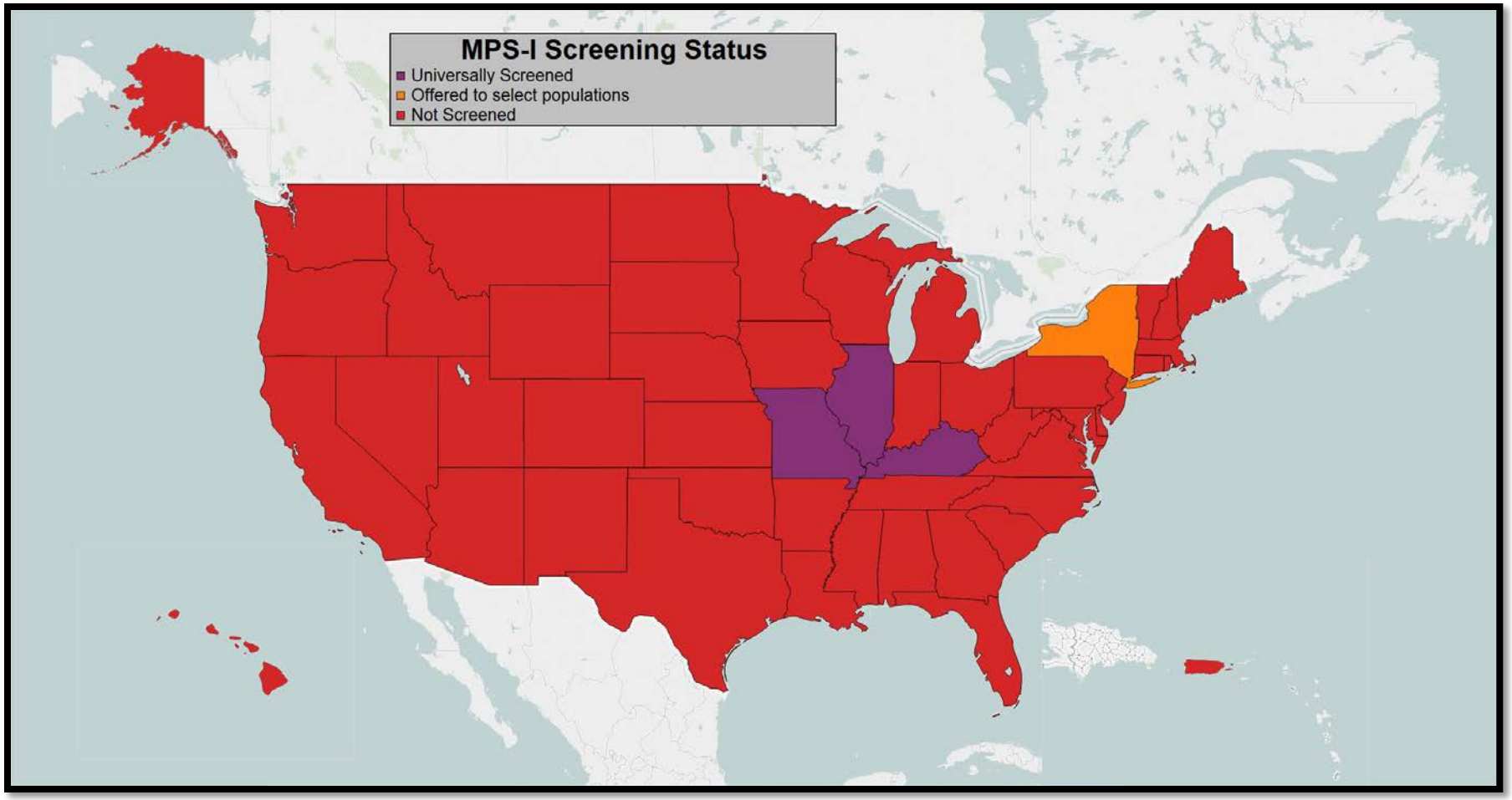
2016

MPS-I Newborn Screening Status Chart

Screening Status	Regulatory Details	Implementation Details	Summary Count
Universally Screened	Data Not Provided	N/A	
	Required by law or rule	N/A	2
Offered to select populations	N/A	N/A	1
Not Screened	Data Not Provided	Data Not Provided	46

0 2 4 6 8 10 12 14 16 18 20 22 24 26 28 30 32 34 36 38 40 42 44 46 48
Number of States



States Currently Screening for MPS 1

Illinois

- Statewide screening for MPS 1 (and Pompe, Fabry, Gaucher, and Neimann-Pick) began in June 2015
- 113,711 births to date
- 43 referrals (presumptive positives), 1 confirmed case

Missouri

- Statewide screening for MPS 1 (and Pompe) began in November 2013
- 237,000 births to date
- 98 referrals, 2 confirmed cases

New York

- Pilot testing began in May 2015
- ~13,500 infants, no cases to date

Kentucky

- Statewide screening for MPS 1 began February 2016

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Progress in x-ALD NBS Implementation

NewSTEPs

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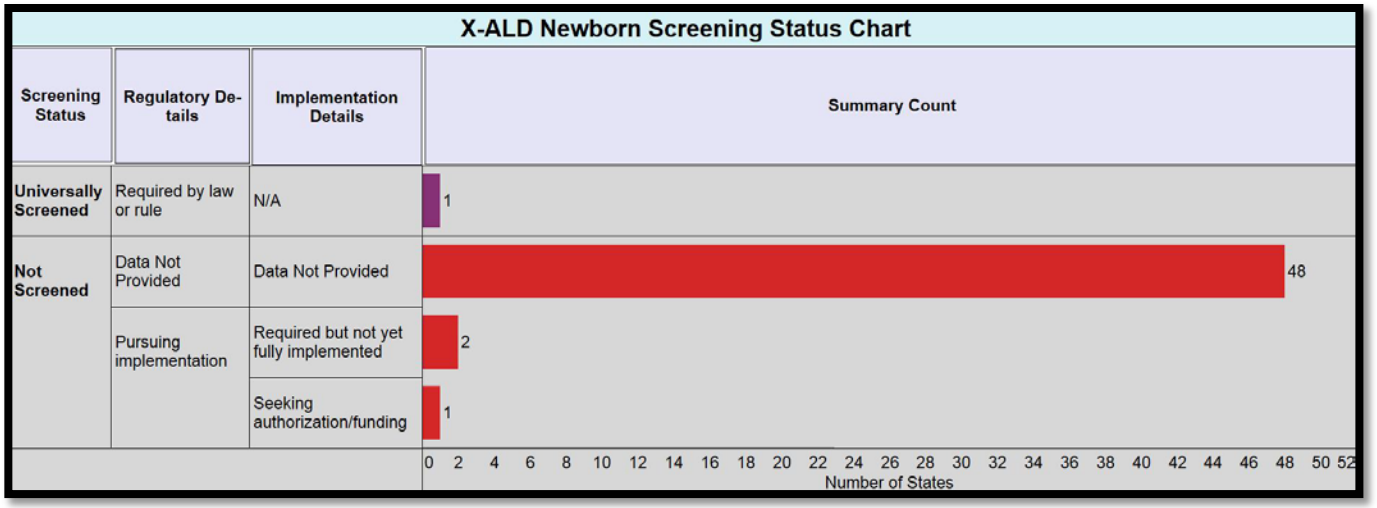
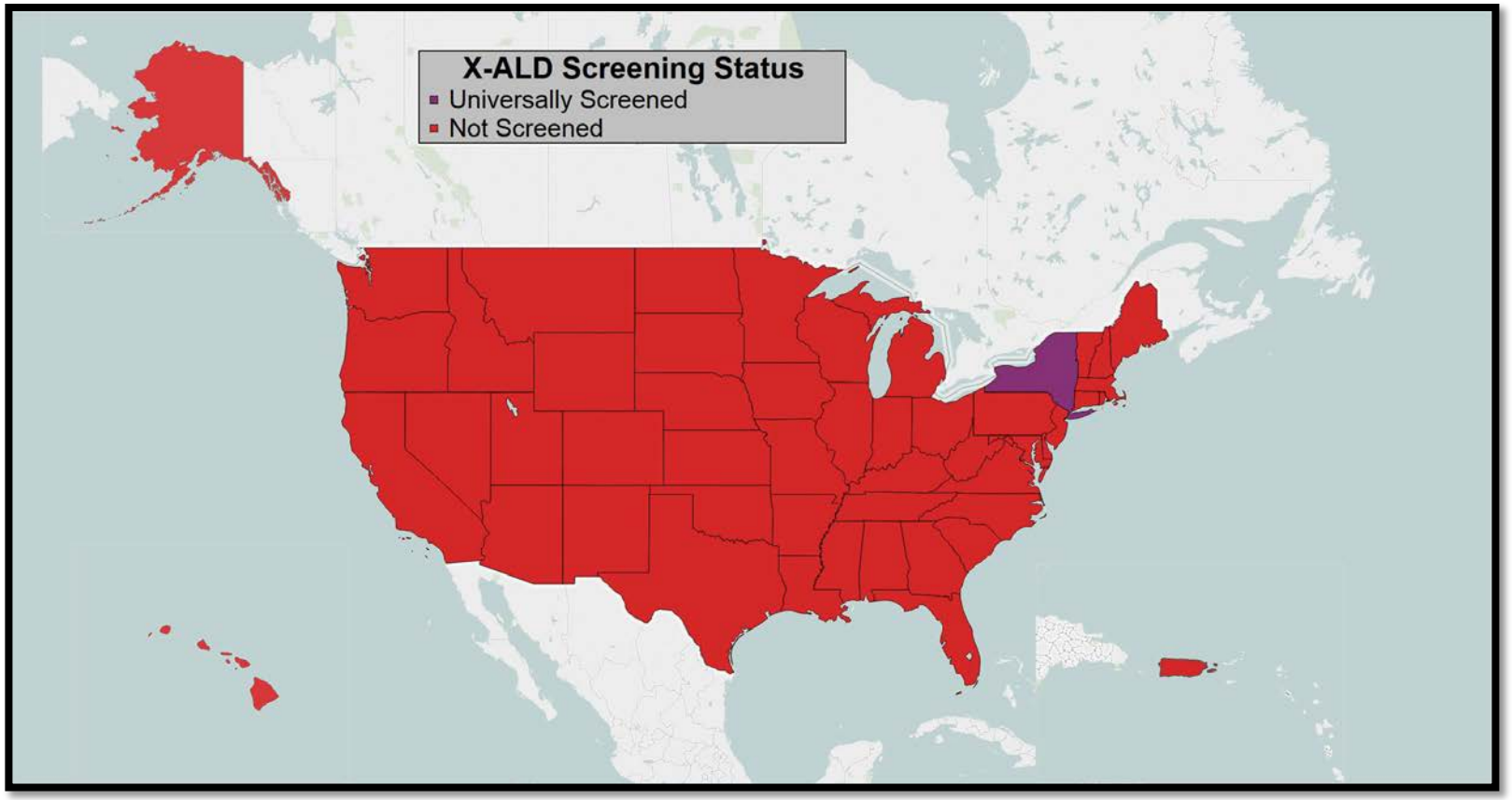
2016


X-ALD Newborn Screening Status Chart
Screening Status
Regulatory Details
Implementation Details
Summary Count
Universally Screened
Required by law or rule
N/A
1
Not Screened
Data Not Provided
Data Not Provided
48
Pursuing implementation
Required but not yet fully implemented
2
Seeking authorization/funding
1
0 2 4 6 8 10 12 14 16 18 20 22 24 26 28 30 32 34 36 38 40 42 44 46 48 50 52
Number of States


X-ALD Screening Status
Universally Screened
Not Screened

States Currently Screening for X-ALD

New York

- Statewide screening for X-ALD began December 30, 2013
- ~500,000 births to date
  - 17 males with elevated marker and a mutation in ABCD1 gene
  - 20 female carriers

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Acknowledgements
Thank you NBS Programs!