U.S. Newborn Screening System: NewSTEPs Summary

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colorado school of public health

On behalf of the Newborn Screening Community

August 2015

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NewSTEPs and related activities are funded through a cooperative agreement (#U22MC24078) to APHL by the Genetic Services Branch of the Health Resources and Services Administration (HRSA).

NewSTEPs Vision

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

NewSTEPs 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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NewSTEPs: Data to Support NBS

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National Data Repository for NBS

Purpose: Provide tools to state newborn screening systems to adequately evaluate, analyze, and benchmark the performance of their tests and the quality of their newborn screening programs

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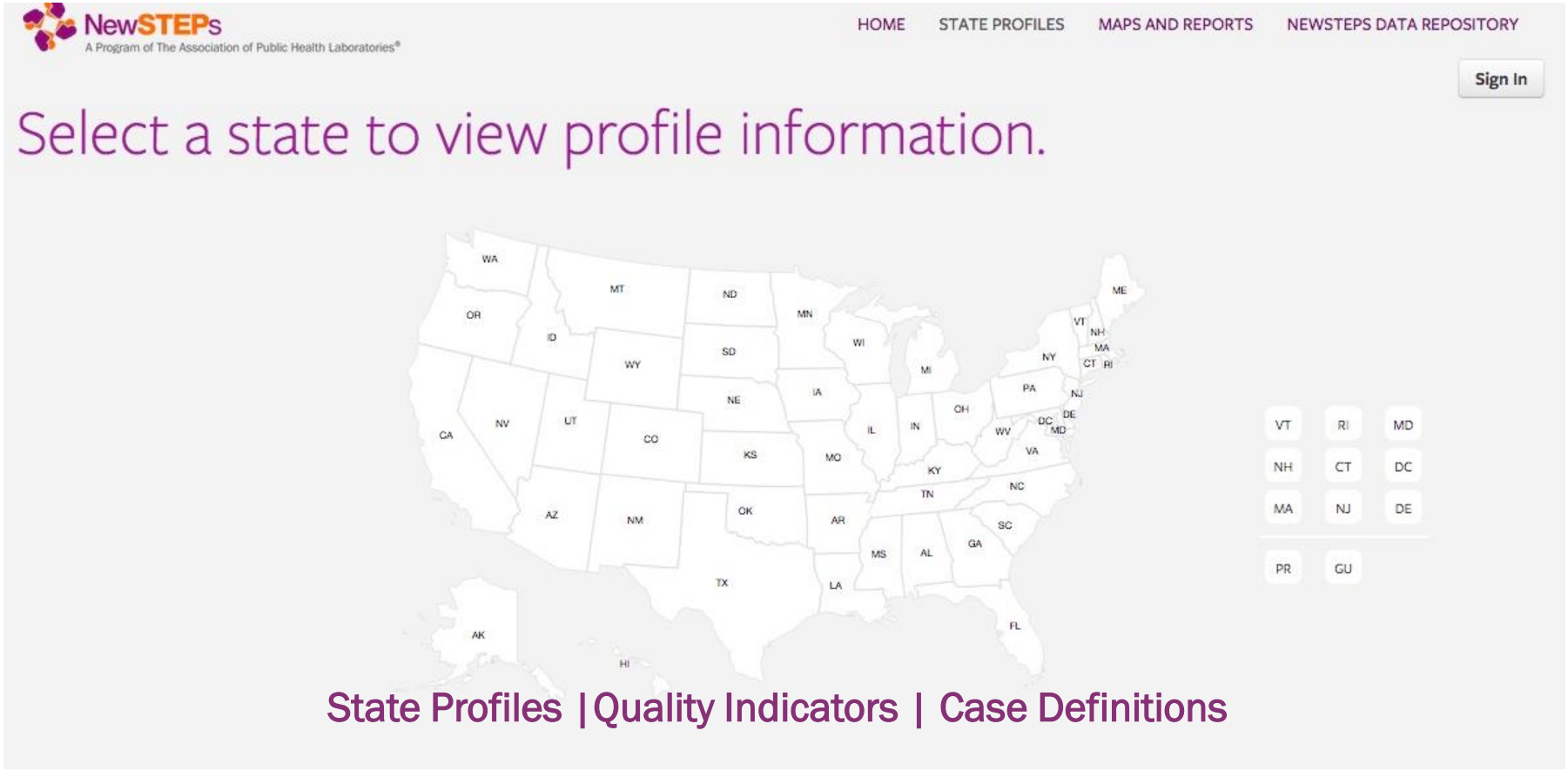
Components of Data Repository

www.newsteps.org



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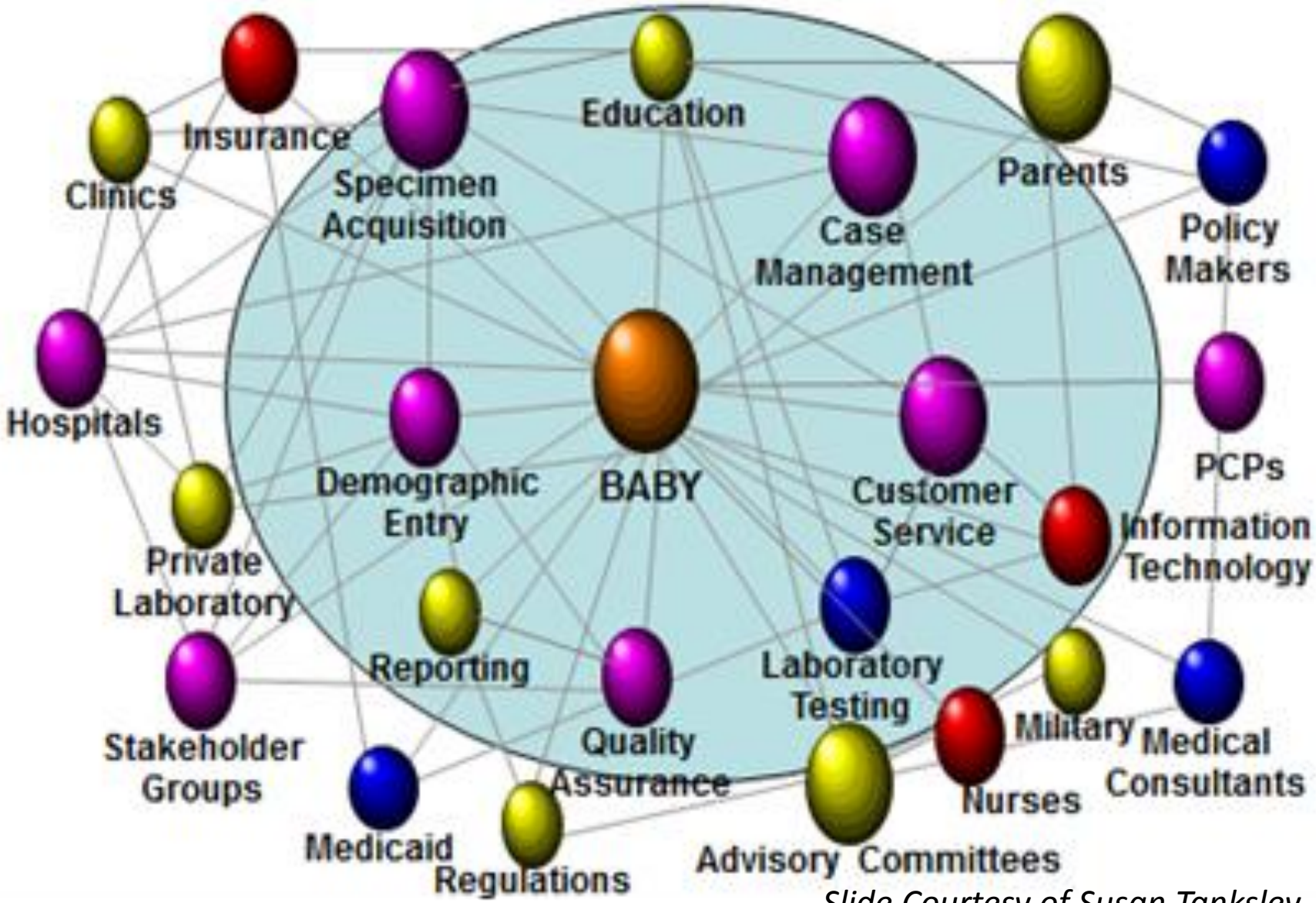
Data Collection and Confirmation

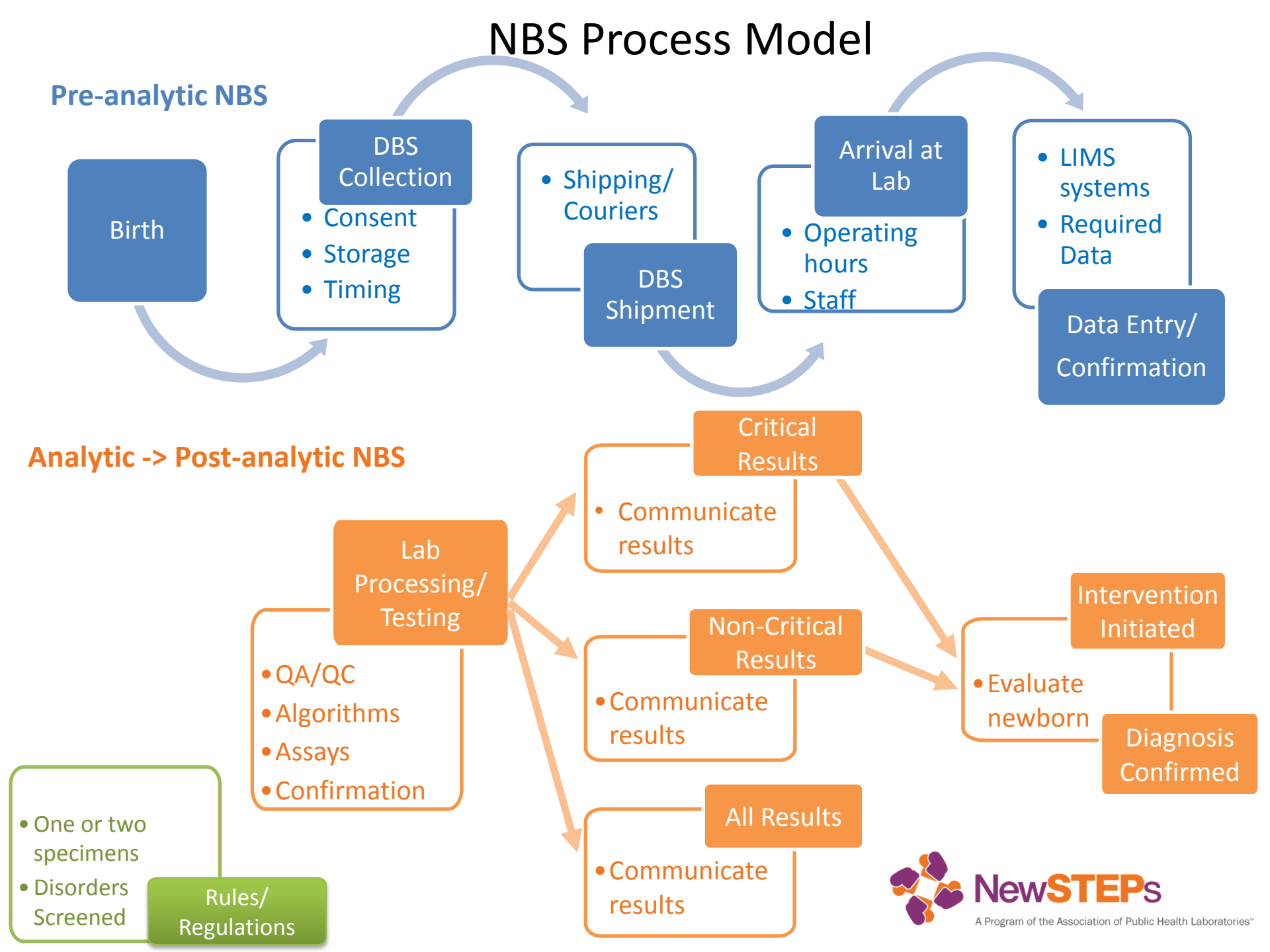
- Data Repository
- Interviews with and training of each NBS program
- Data entry from NBS programs
- Confirmation of data via printed summary reports
- Iterative process

Newborn Screening System Partners

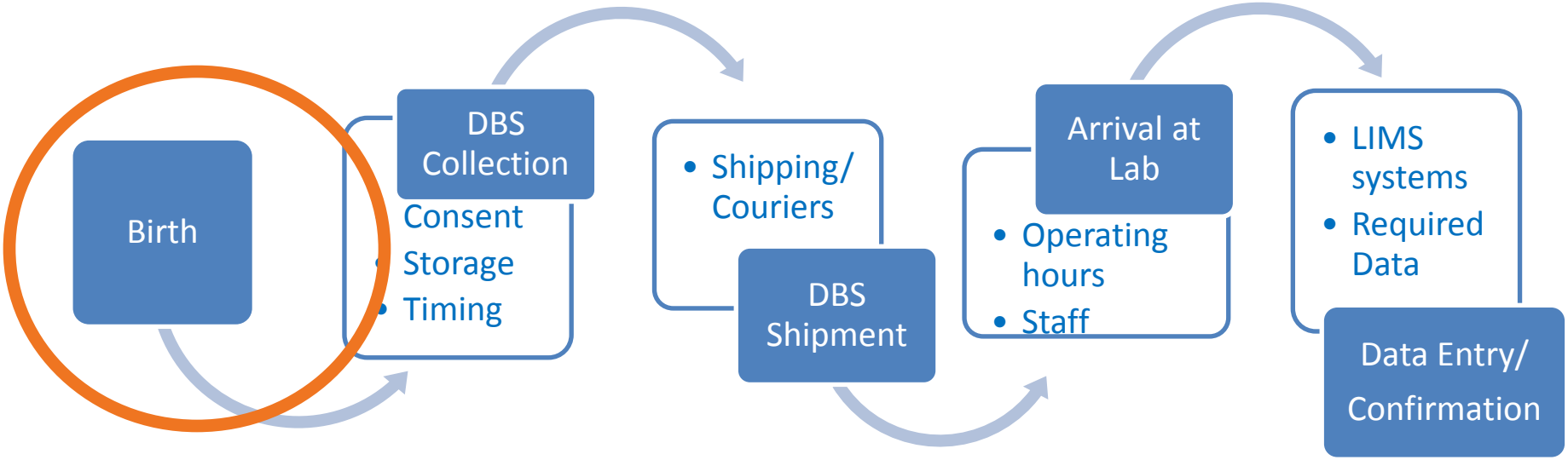


Slide Courtesy of Susan Tanksley, PhD

NBS Process Model



Pre-Analytic



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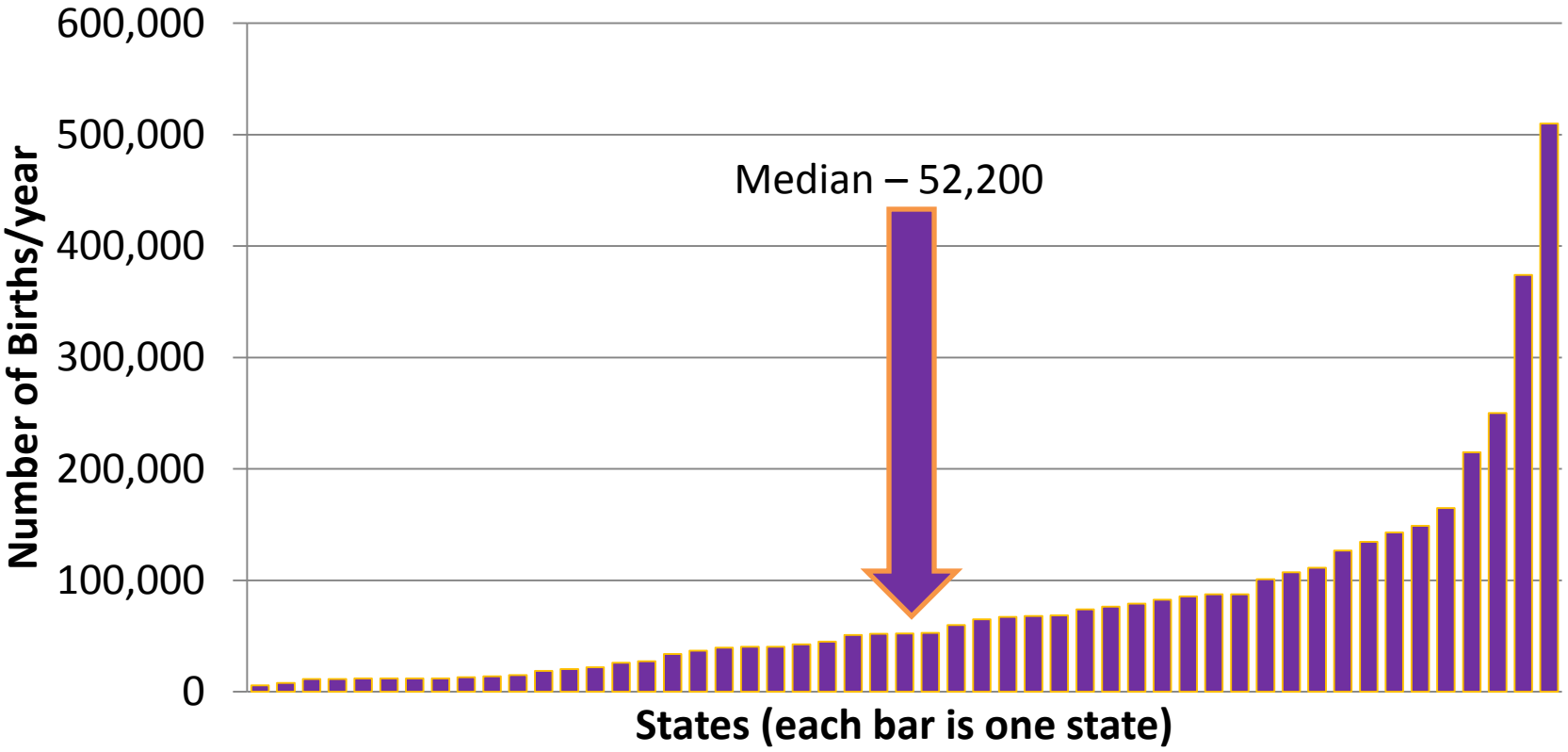
Number of Annual Births in the U.S.



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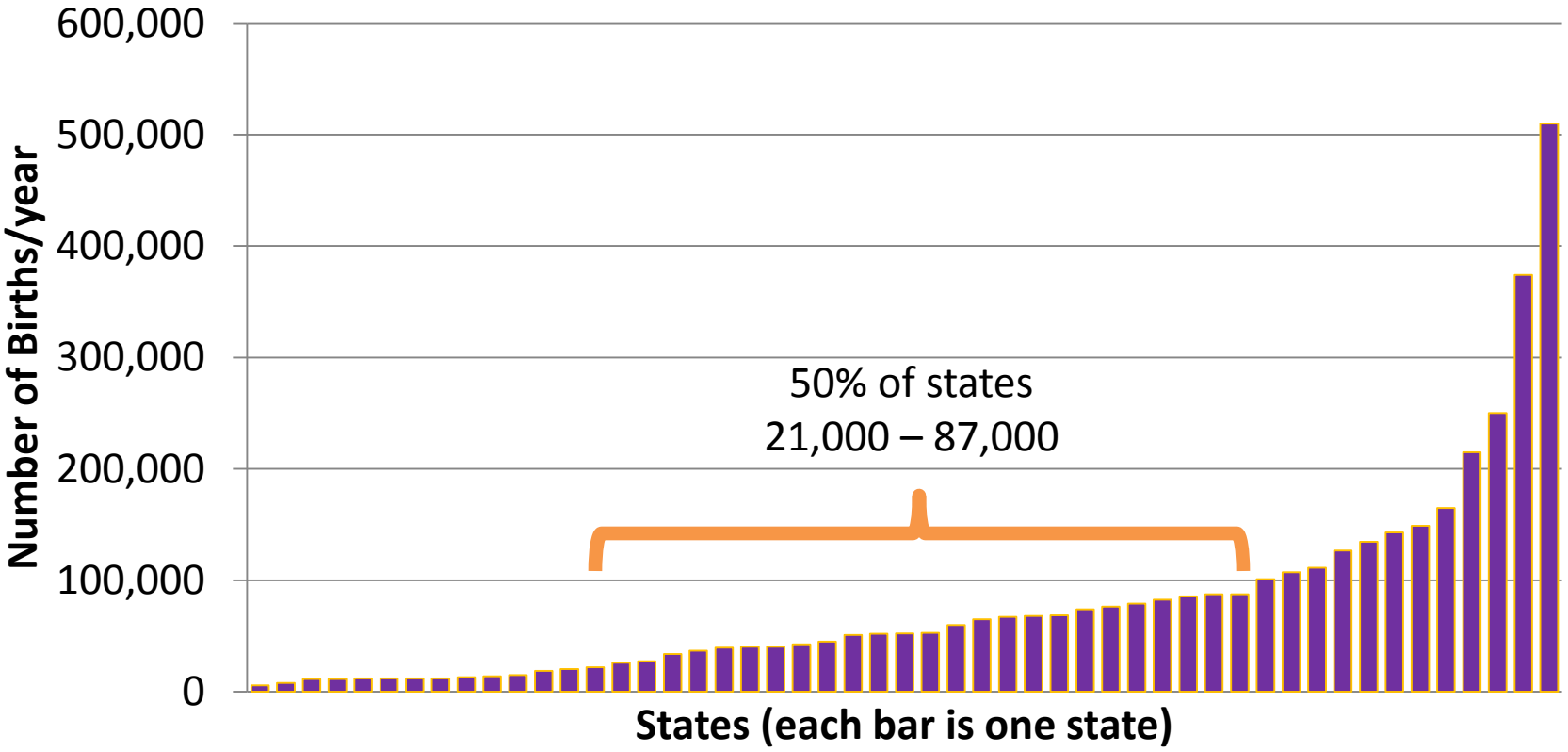

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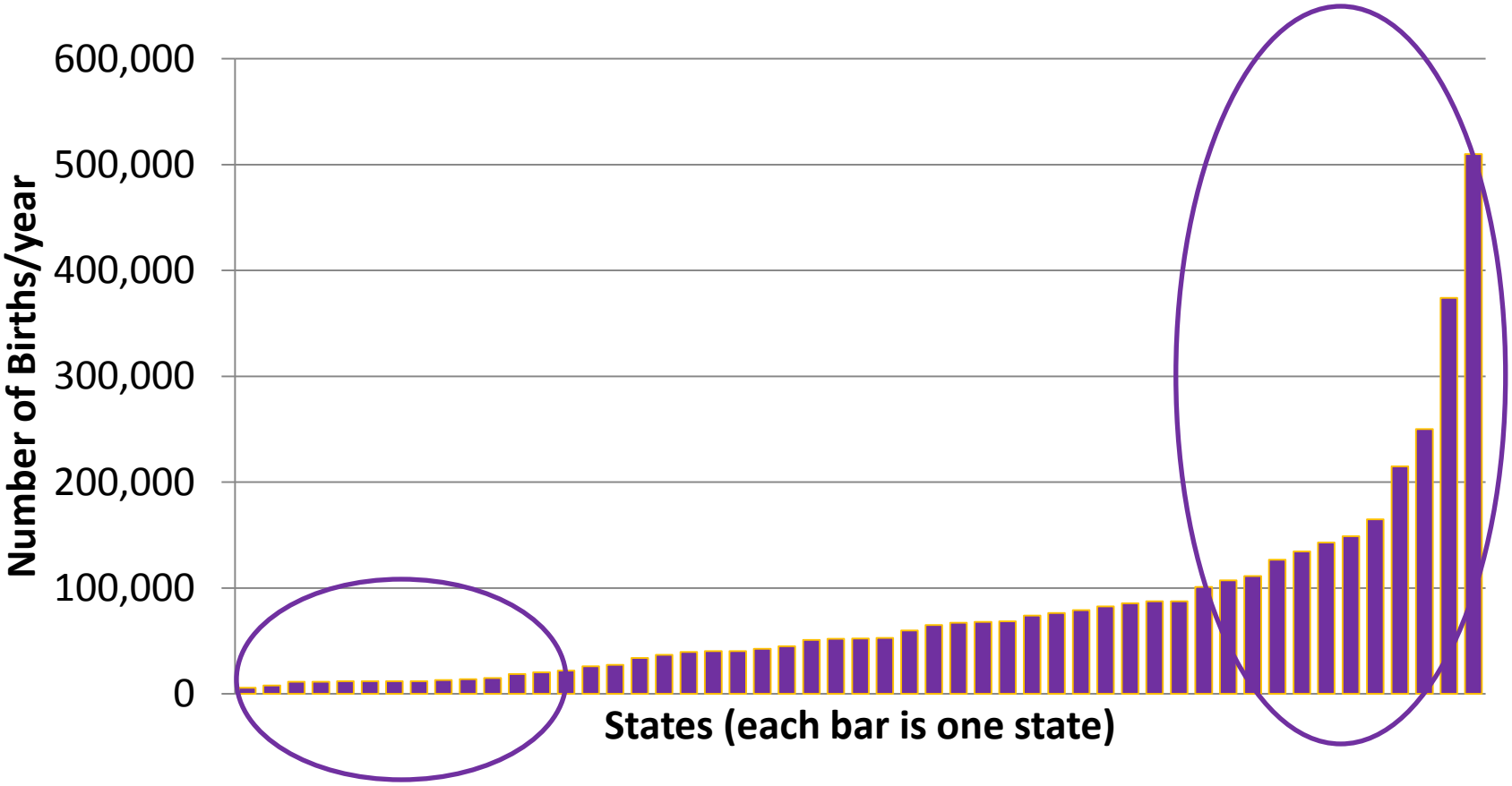
Number of Annual Births in the U.S.



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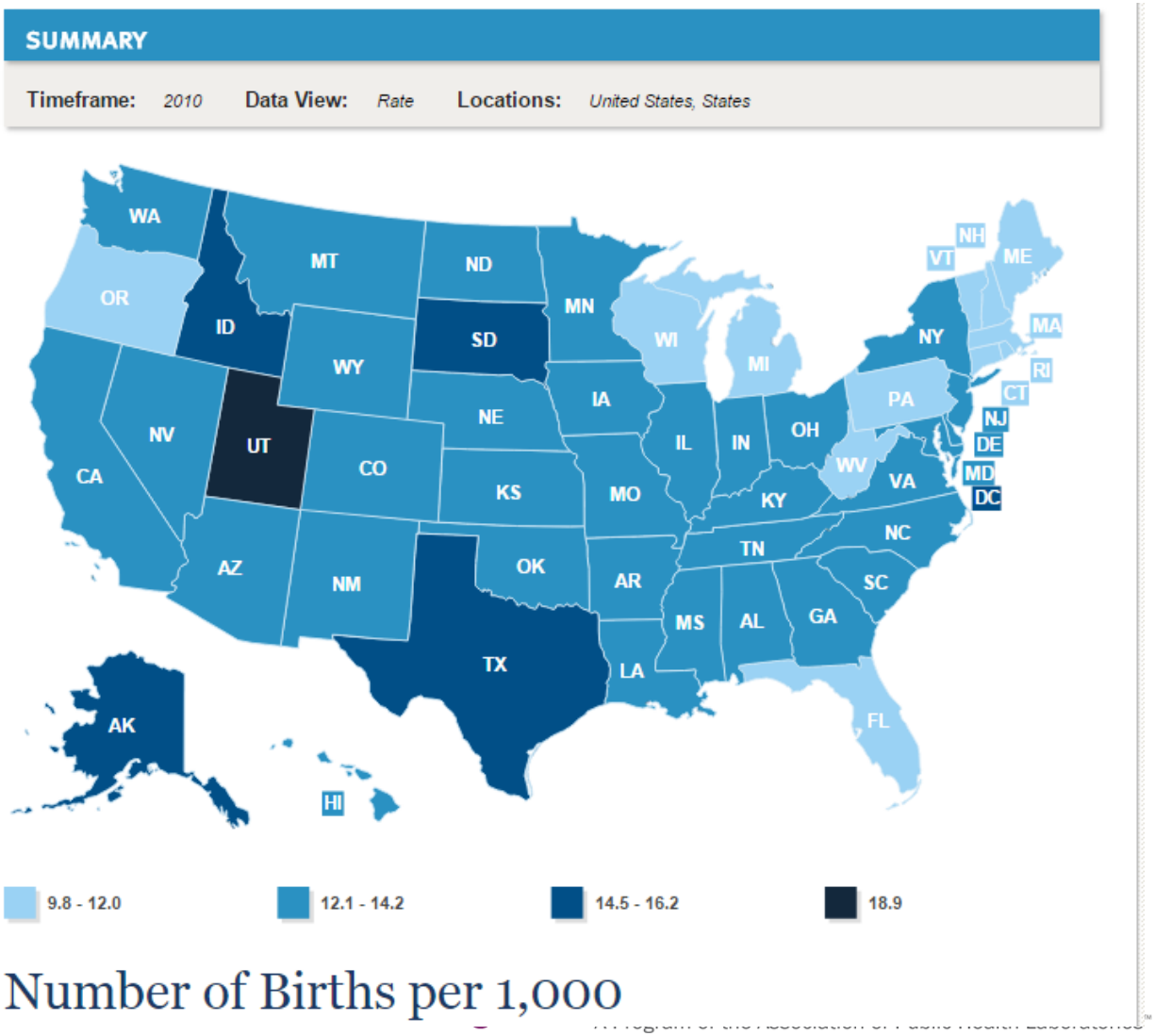
Birth rates vary between states

- The number of live births per 1,000 population varies
- May point to different needs



http://kff.org/other/state-indicator/birth-rate-per-1000/#map

What do we know about our newborn screening systems in the U.S.?

- 52 newborn screening programs
- 36 newborn screening labs
- Geographically diverse states
  - 663,000 Square Miles to 1,212 square miles

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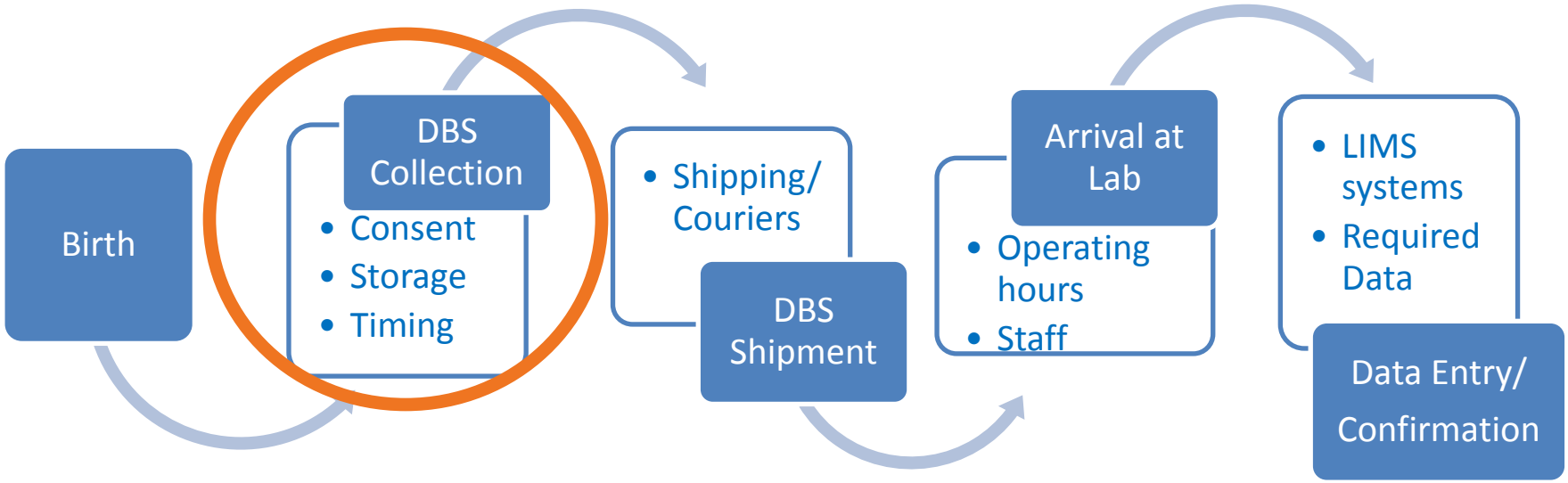
Pre-Analytic



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States practices on parental refusal of newborn screening

- Consent is implied in most states; Most states allow parents to opt-out of newborn screening (religious, other)

Table 1. State refusal provision by documentation form type.

	No State Form		Optional State Form		Required State Form		Total	
Refusal provision	n	%	n	%	n	%	n	%
Refuse for any reason	5	10%	6	12%	5	10%	16	31%
Refuse for religious reasons	12	24%	6	12%	14	27%	32	63%
No provision for refusals	3	6%	0	0%	0	0%	3	6%
Total:	20	39%	12	24%	19	37%	51	100%

Review of Best Practices in Documenting Newborn Screening Refusals for States
Jeremy Penn, Ph.D., and Eric Sondreal, M.P.H., NDSU
Supported in part by a grant from HRSA H46MC24089
Report for the Heartland Genetic Services Collaborative, April, 2015
https://drive.google.com/file/d/0BwP8F0nwpufqMFpib0o5VWhIYm8/view

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Storing Samples and Storing Data

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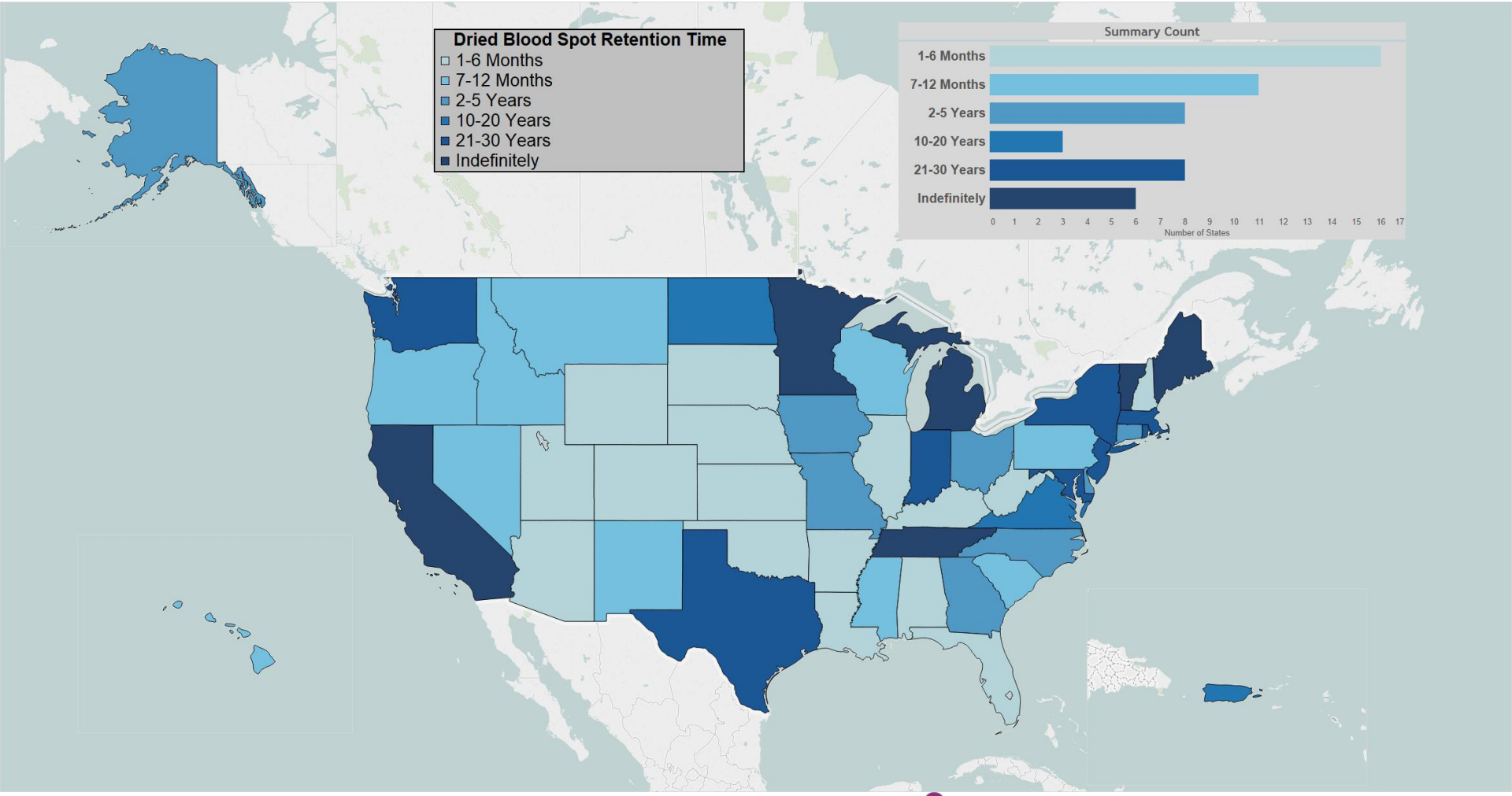
Dried Blood Spot Retention Time



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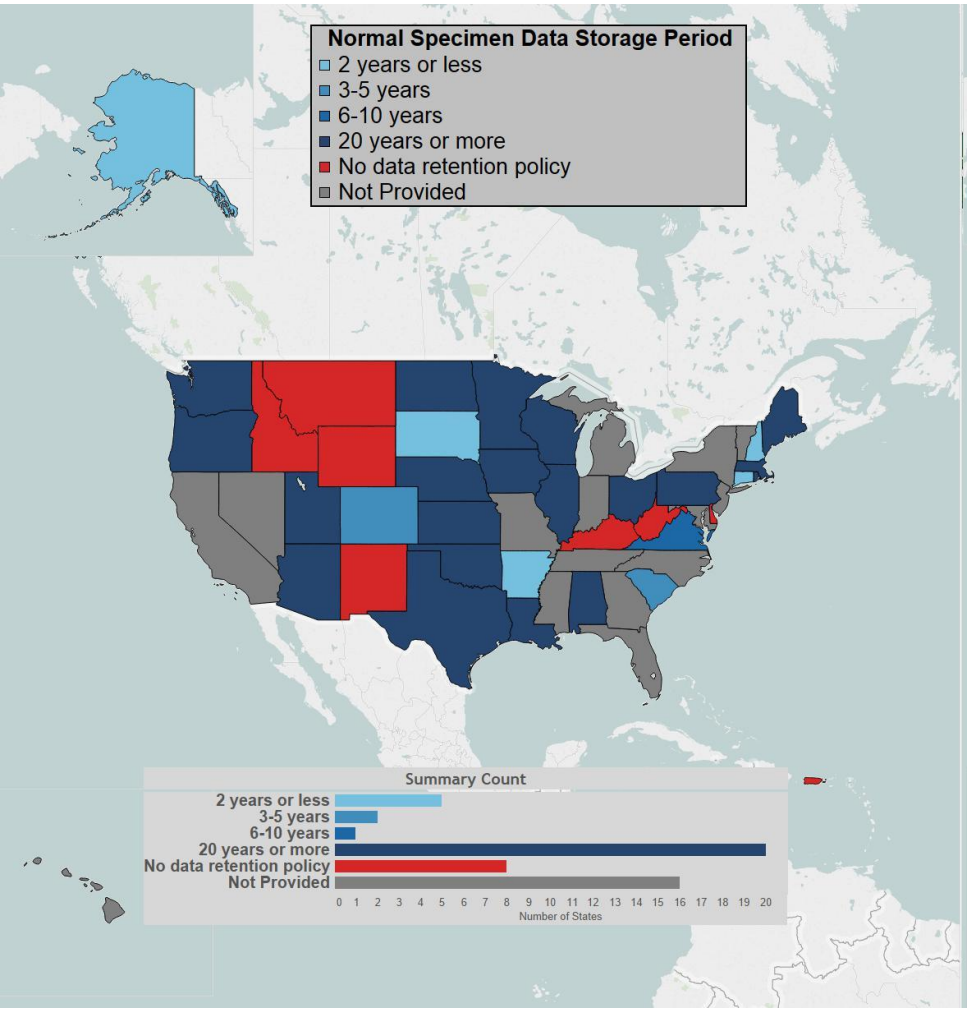
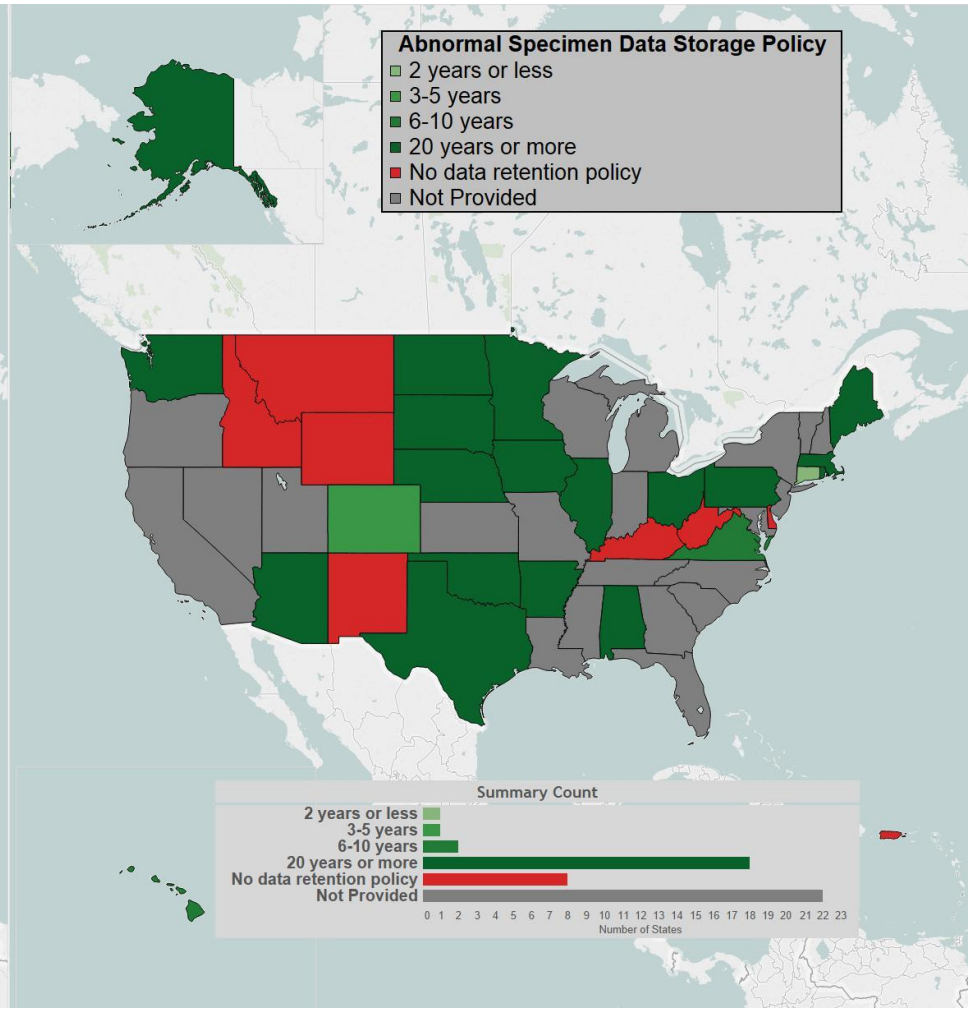
Data Storage Periods





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Informed Consent for NBS Research and Storage of Dried Blood Spots

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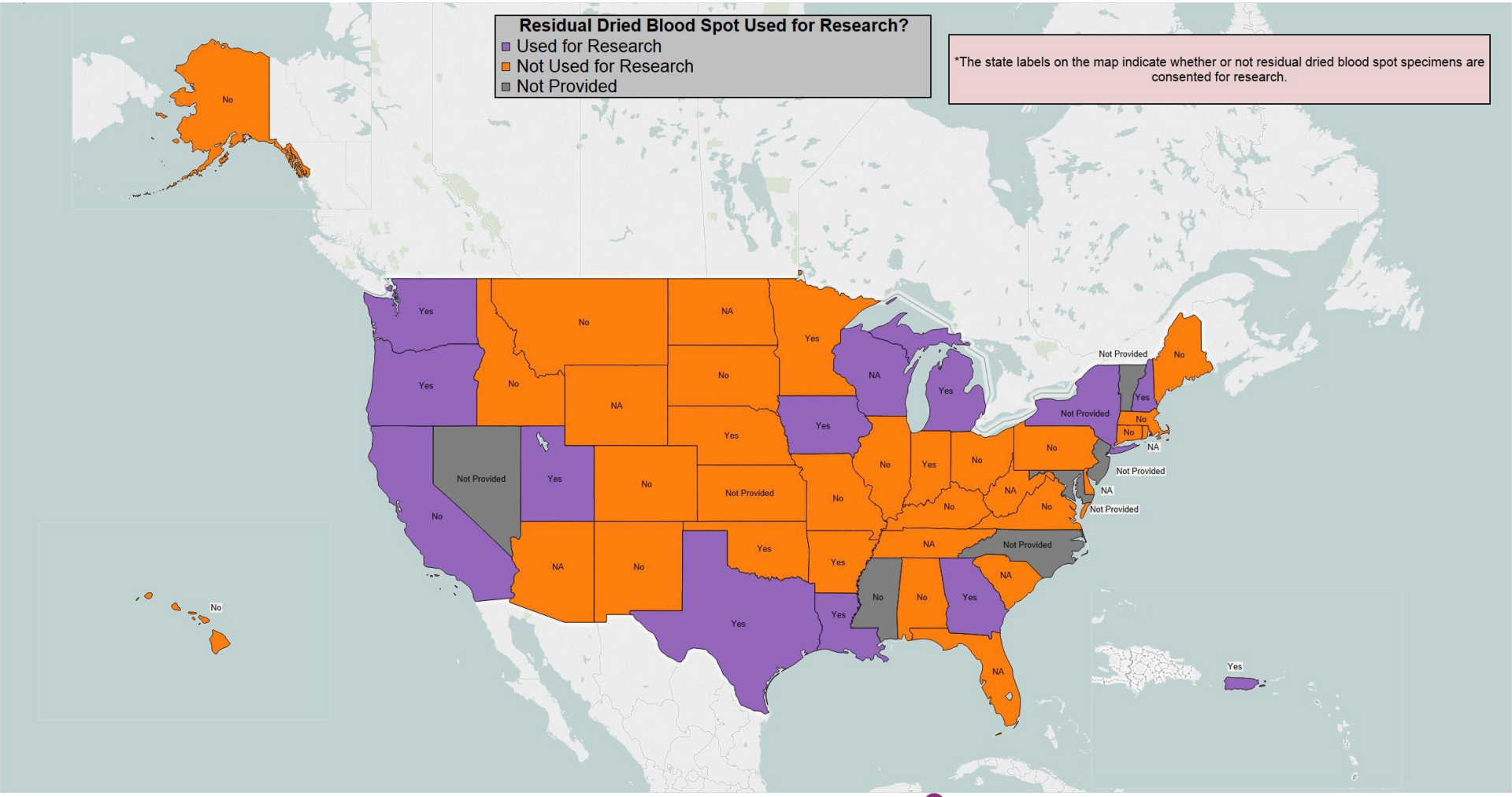
NBSSLA Research Amendment

- Federally funded research on NBS blood spots is considered research on human subjects, regardless of whether the specimens are identifiable
- Eliminates the ability of an IRB to approve alterations or waivers of informed consent
- Applies to samples collected 90 days after enacted date
- Secretary must promulgate proposed revisions to Federal Policy for the Protection of Human Subjects within six months and final regulations within two years

Which States Use Dried Blood Spots for Research?



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Timeliness

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ACHDNC Timeliness Recommendations

In order to achieve the best outcomes for babies:

A. Presumptive positive results for time-critical conditions should be communicated immediately to the child's healthcare provider but no later than 5 days of life.

B. Presumptive positive results for all other conditions should be communicated to the child's healthcare provider as soon as possible but no later than 7 days of life.

C. All NBS tests should be completed within 7 days of life.

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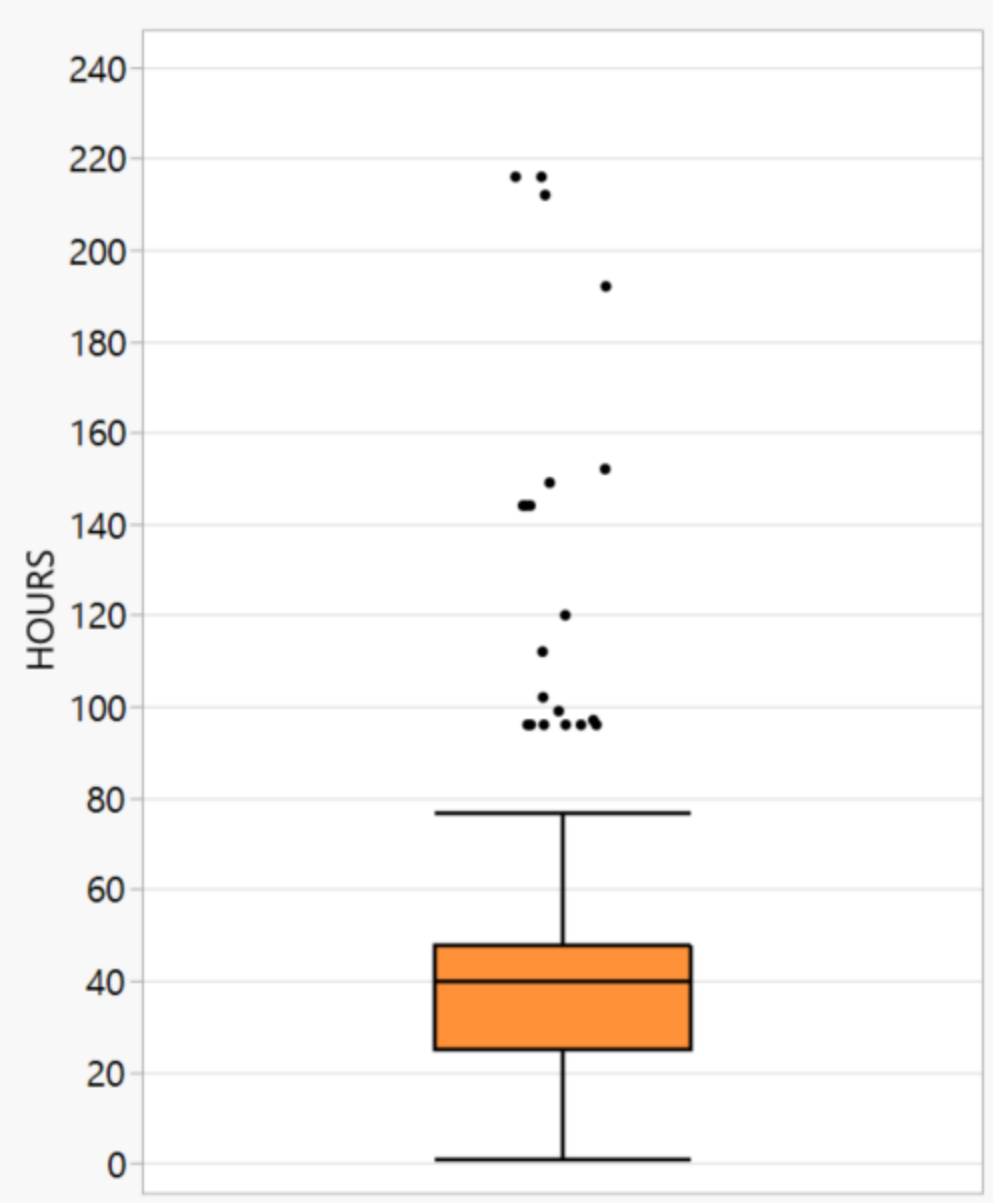
ACHDNC Timeliness Recommendations

In order to achieve these goals and reduce delays in newborn screening:

D. Initial NBS specimens should be collected in the appropriate time frame for the baby's condition but no later than 48 hours after birth.

E. NBS specimens should be received at the Laboratory as soon as possible; ideally within 24 hours of collection.

Specimen collection



- NewSTEPs Data
  - Cases with disorders diagnosed by NBS
  - States with signed MOUs
  - Not all disorders

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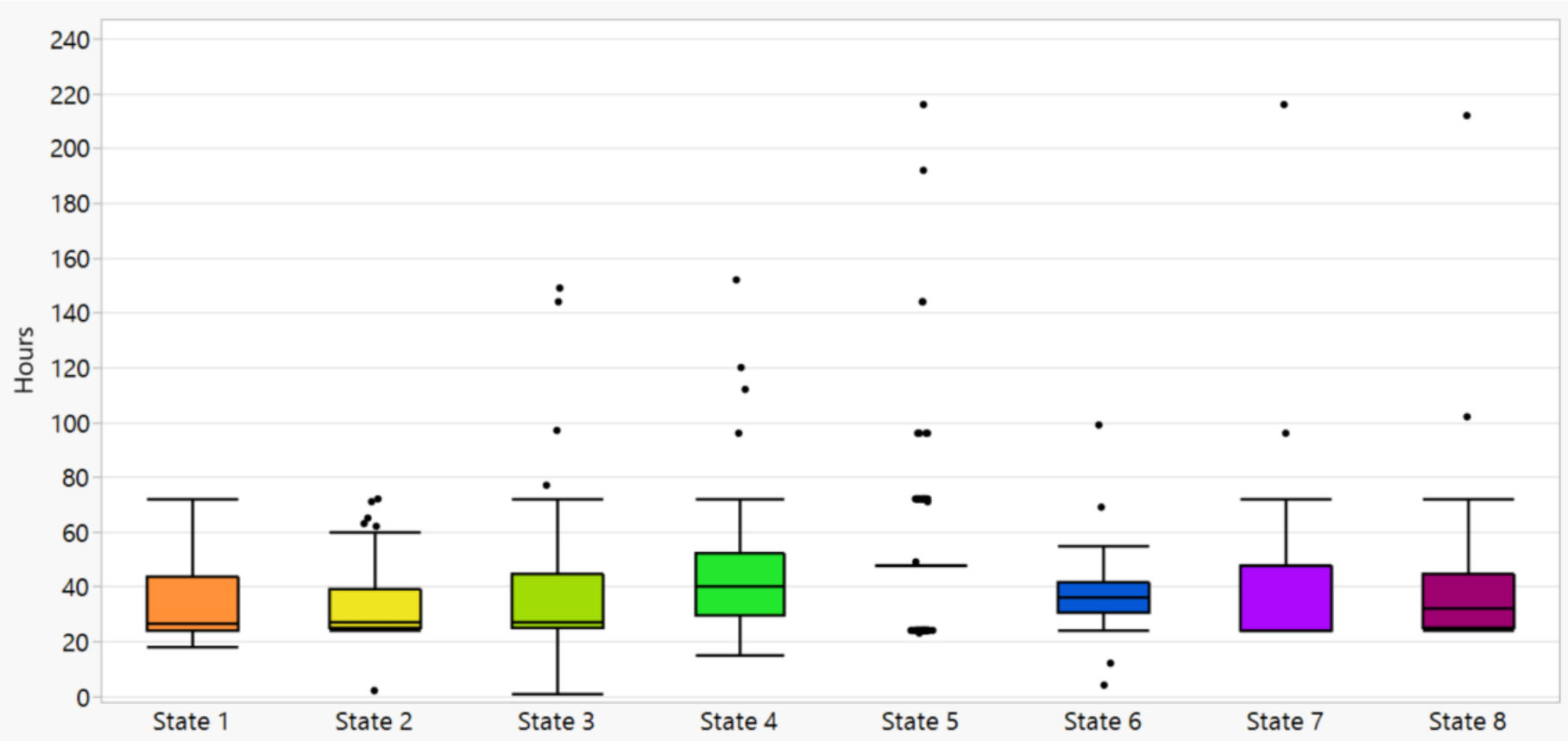
State variation in Specimen Collection



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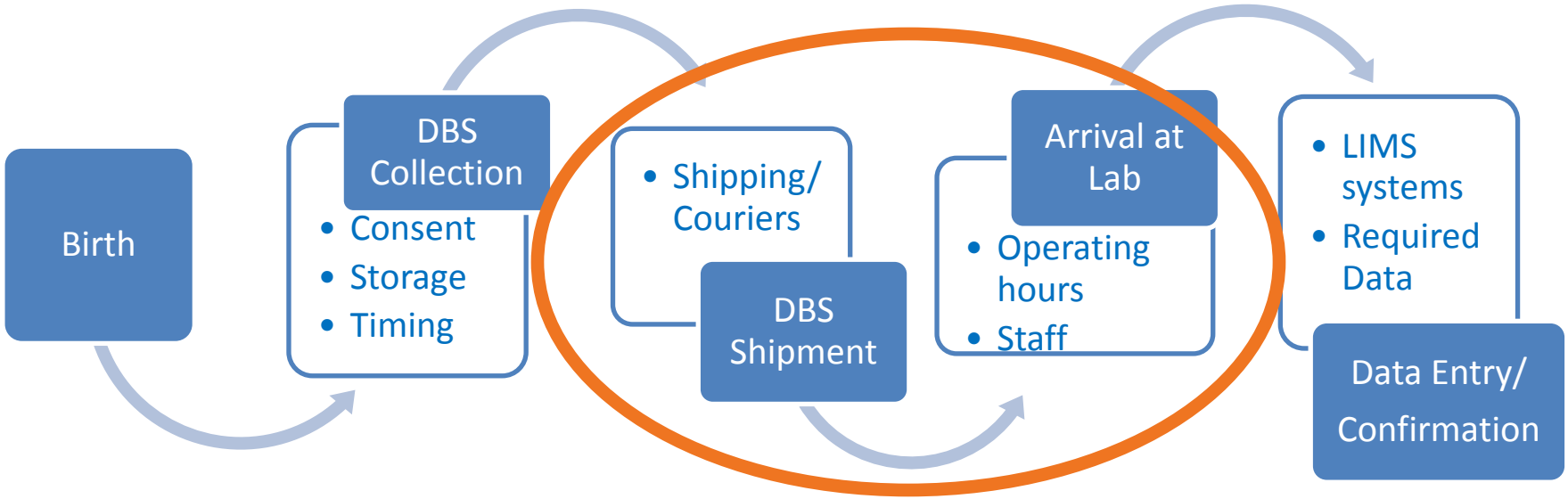
Pre-Analytic



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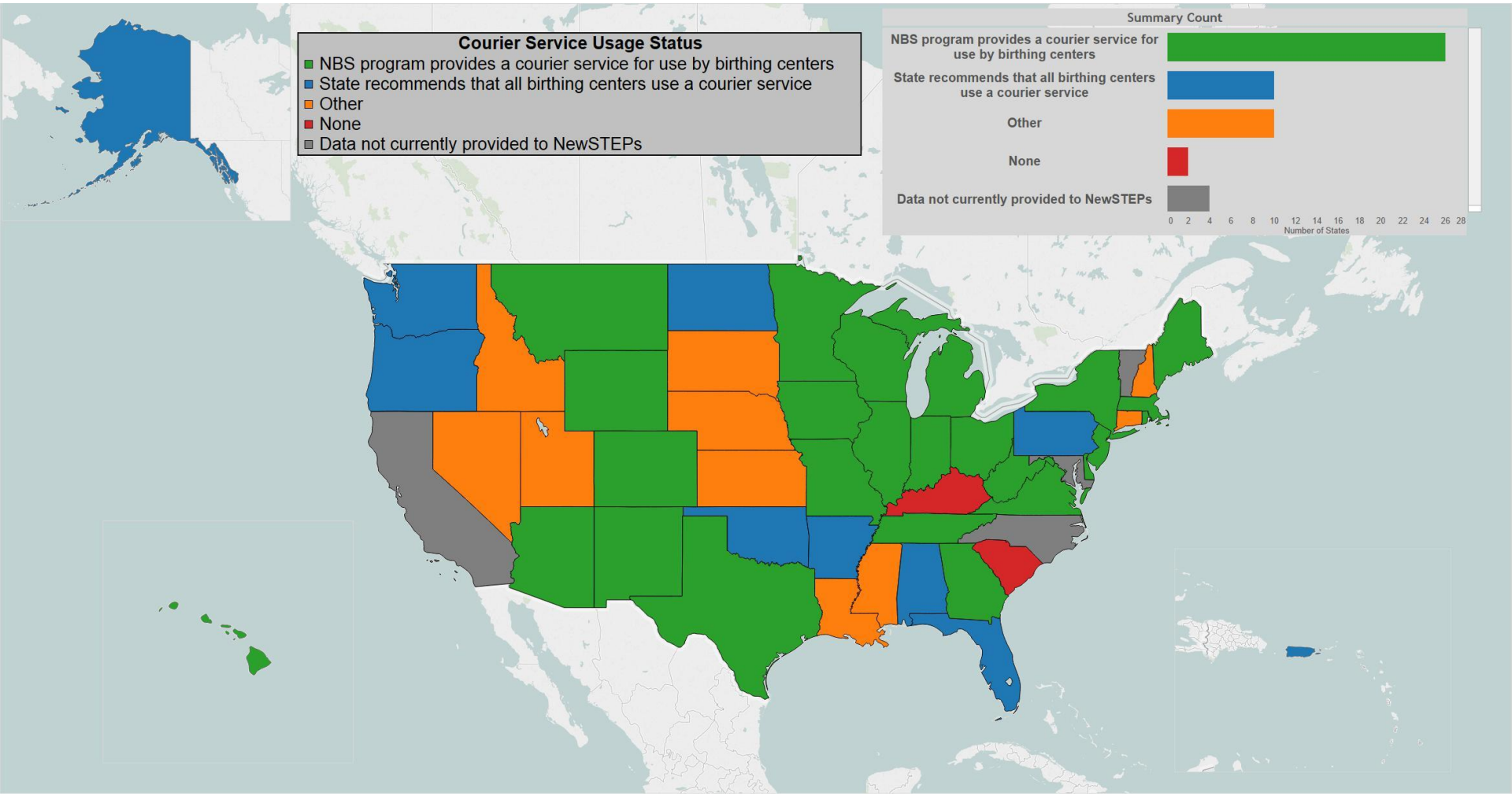
Courier Service Usage Status



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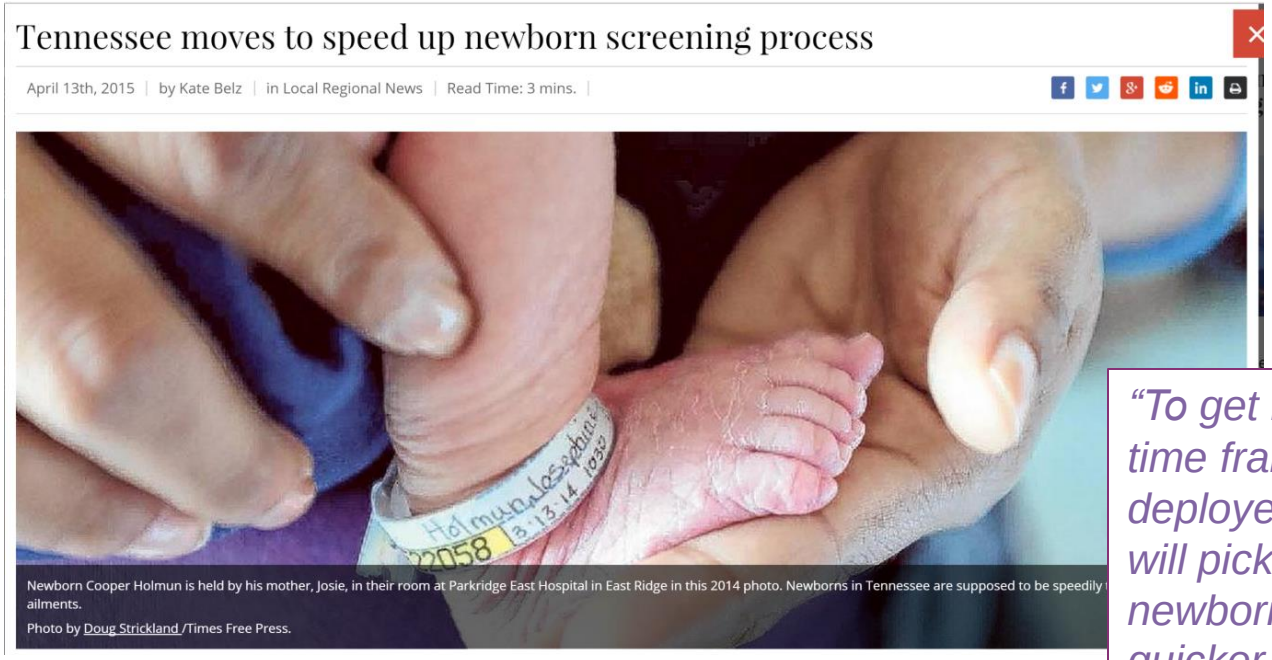
States are changing policies for delivery of samples



Tennessee moves to speed up newborn screening process

April 13th, 2015 | by Kate Belz | in Local Regional News | Read Time: 3 mins. |

Newborn Cooper Holmun is held by his mother, Josie, in their room at Parkridge East Hospital in East Ridge in this 2014 photo. Newborns in Tennessee are supposed to be speedily ailments.

Photo by Doug Strickland /Times Free Press.

"To get nearer the [recommended] time frame, the state last week deployed a new courier service that will pick up and shuttle hospitals' newborn screening specimens on a quicker, more consistent schedule."

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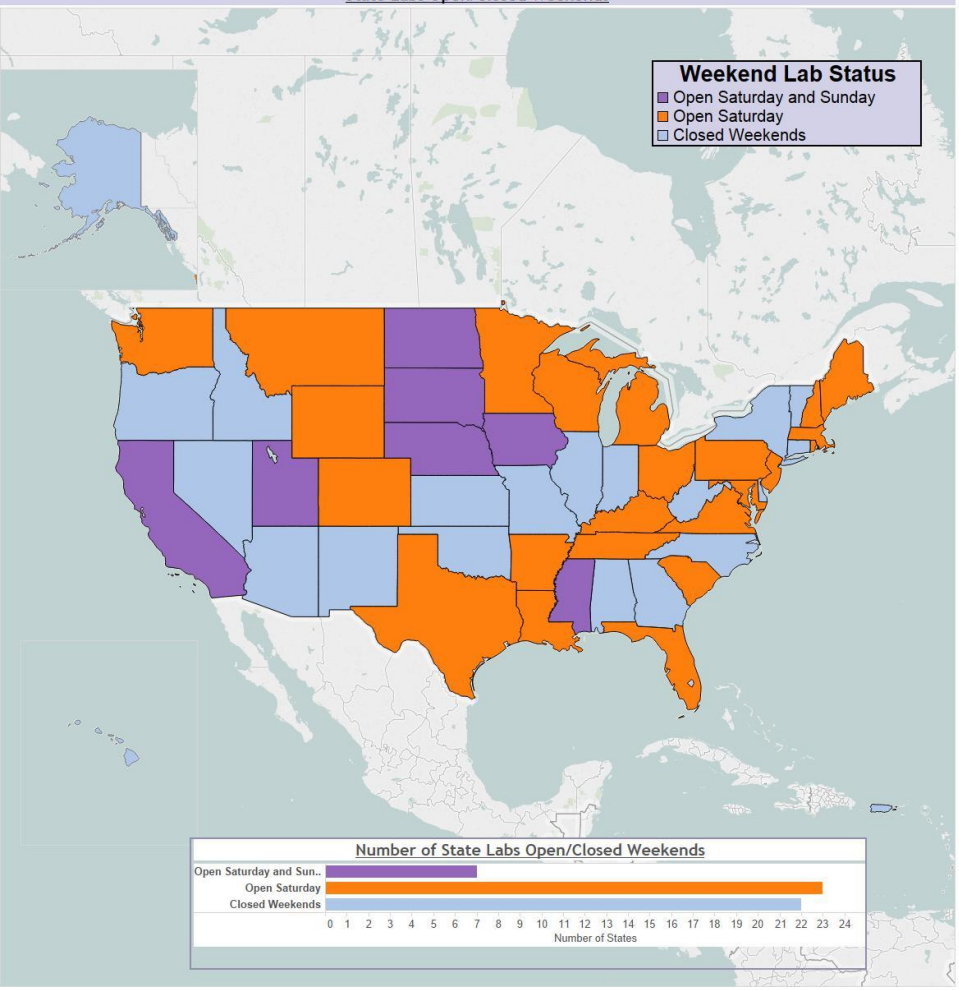
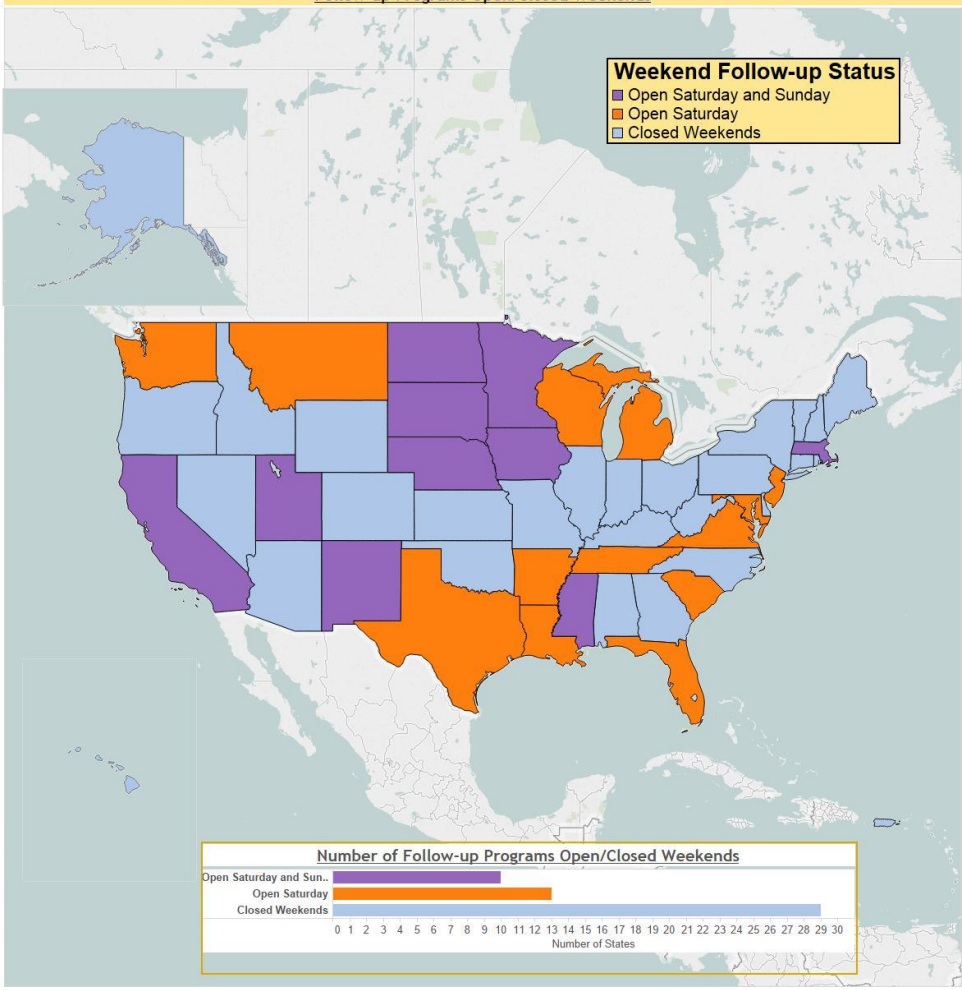
Weekend Operating Status

State Labs Open/Closed Weekends



Follow-up Programs Open/Closed Weekends



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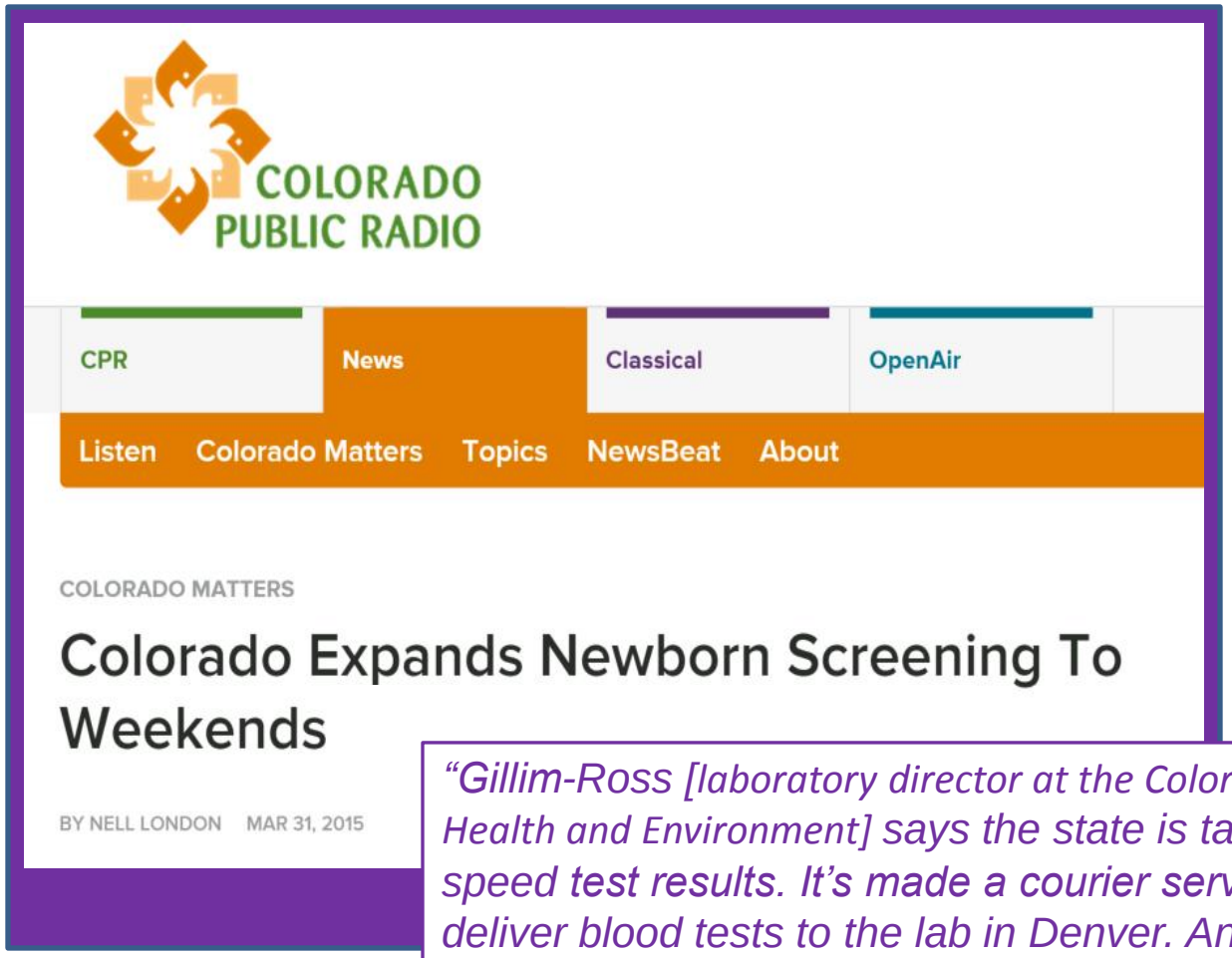

"Gillim-Ross [laboratory director at the Colorado Department of Public Health and Environment] says the state is taking other steps to speed test results. It's made a courier service available to hospitals to deliver blood tests to the lab in Denver. And its working with the Colorado Hospital Association to educate medical professionals on taking the blood samples promptly and correctly."

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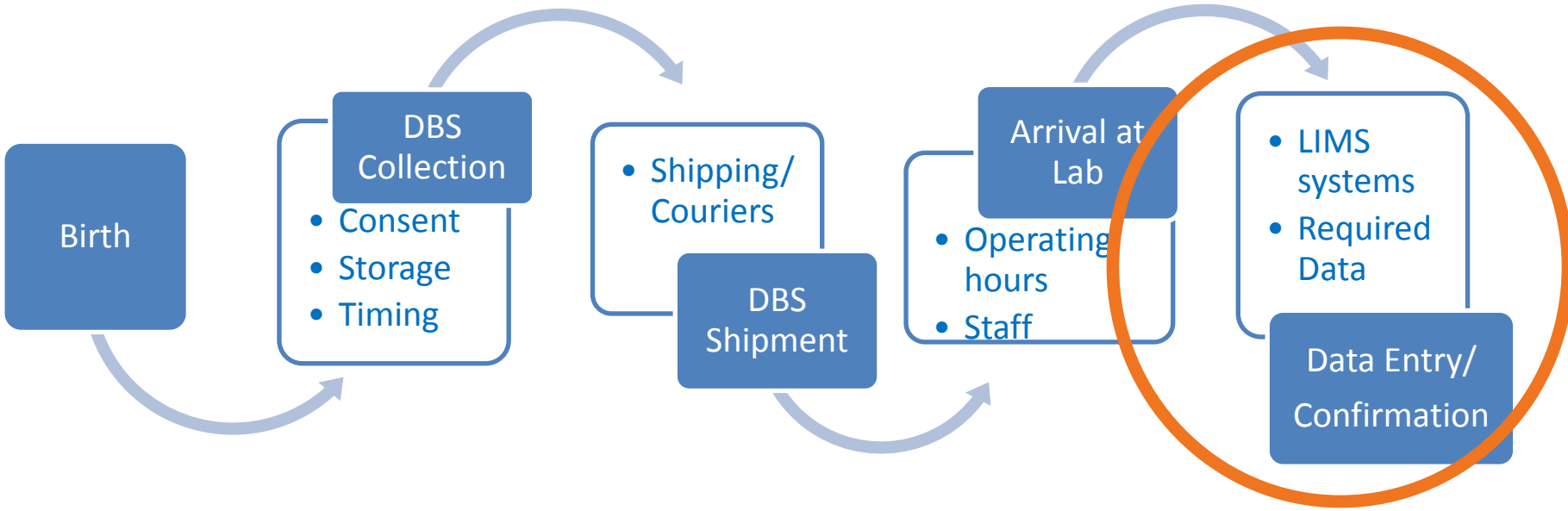
Pre-Analytic



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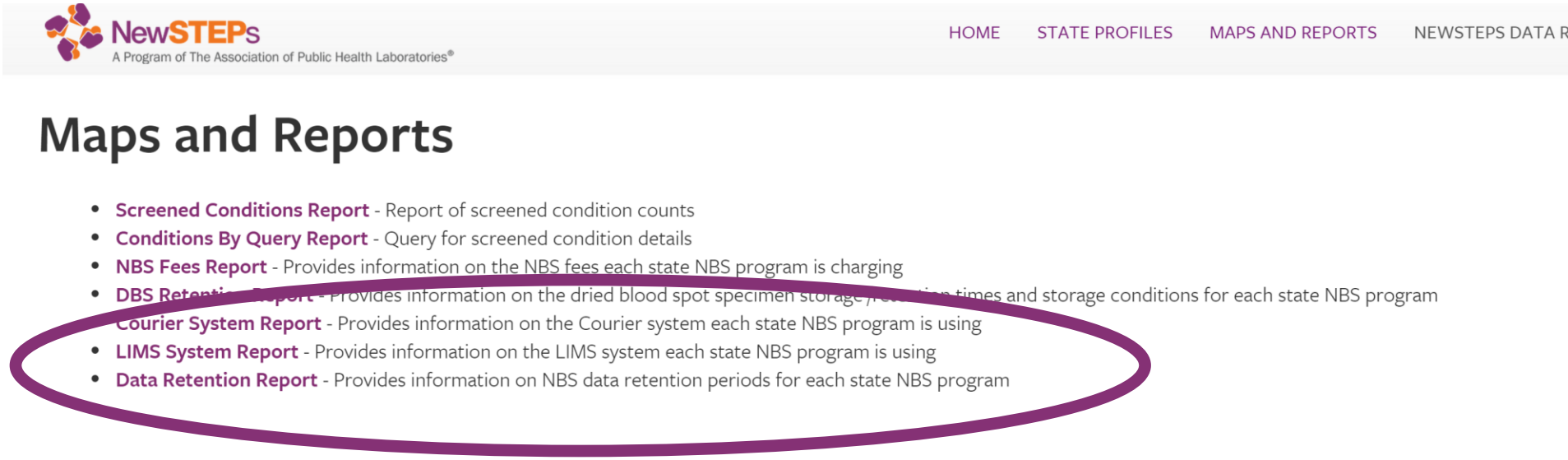
LIMS SYSTEMS



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LIMS System Summary

1-6 of **6** Results

LIMS System	Total
Internally Developed	4
Neometrics/ Natus	14
Other	8
PerkinElmer	15
StarLims	1
Unanswered	11

LIMS System by State

1-53 of **53** Results

State NBS Program	LIMS System	Other LIMS Specified	Follow-up System	Other Follow-up System Specified
Alabama	Neometrics/ Natus		Neometrics/ Natus	
Alaska	Neometrics/ Natus		Internally Developed	
Arizona	PerkinElmer		Neometrics/ Natus	
Arkansas	Unanswered		Unanswered	
California	PerkinElmer		Internally Developed	
Colorado	PerkinElmer		Internally Developed	
Connecticut	StarLims		StarLims	
Delaware	Neometrics/ Natus		Neometrics/ Natus	

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Screening Practices in States

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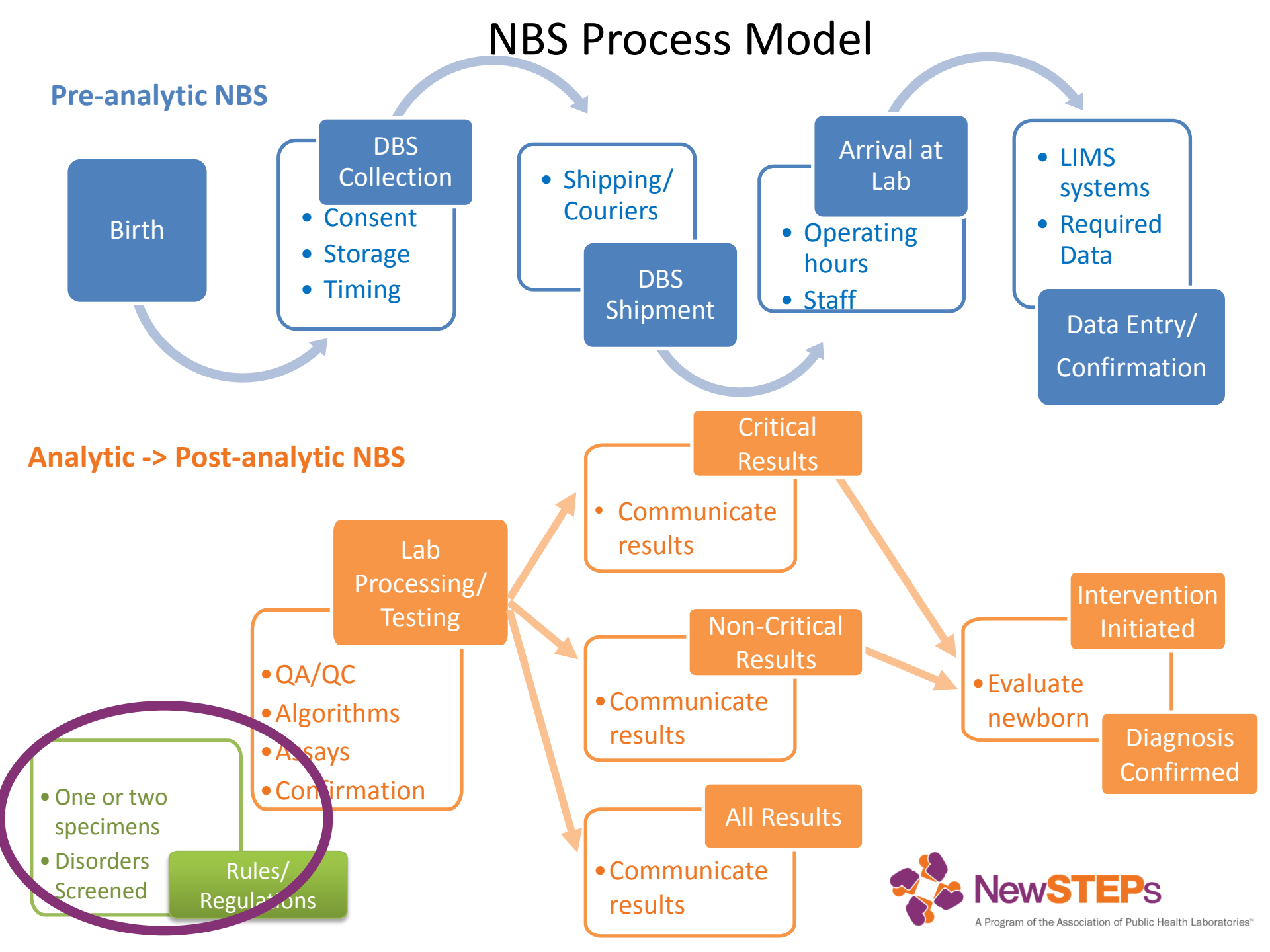
NBS Process Model



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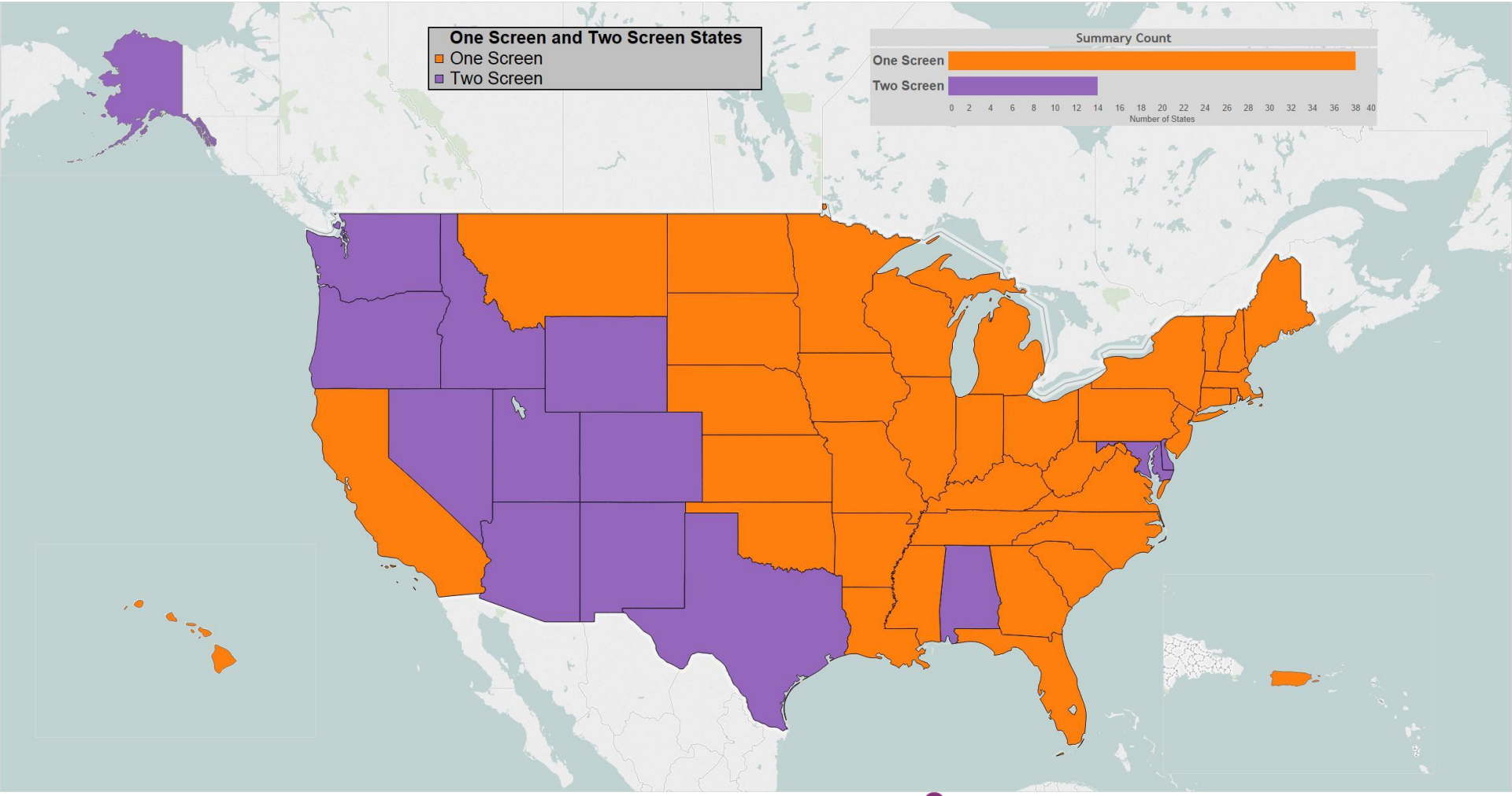
Number of screens (1 vs. 2 Screen States)



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Single newborn screen or routine second screening for primary congenital hypothyroidism☆

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ABSTRACT

Routine second screening of most newborns at 8–14 days of life for a panel of newborn conditions occurs in 12 U.S. states, while newborns in the other states typically undergo only a single routine newborn screen. The study objective was to evaluate screening consequences for primary congenital hypothyroidism (CH) in one- and two-screen states according to laboratory practices and medical or biochemical characteristics of screen-positive cases. Individual-level medical and biochemical data were retrospectively collected and analyzed for 2251 primary CH cases in one-screen (CA, WI) and two-screen (AL, DE, MD, OR, TX) states. Aggregate data were collected and analyzed for medical and biochemical characteristics of all screened newborns in the states. Among the states evaluated in this study, the detection rate of primary CH was higher in the one-screen states. In the two-screen states, 11.5% of cases were detected on the second screen. In multivariate analyses, only race/ethnicity was a significant predictor of cases identified on the first versus second screen, which likely reflects a physiologic difference in primary CH presentation. Newborn screening programs must heed the potential for newborns with CH not being detected by a single screen, particularly newborns of certain races/ethnicities. If the two-screen states converted to a single screen using their current algorithms, newborns currently identified on the routine second screen would presumably not be detected, resulting in probable delayed diagnosis and treatment. However, based on the one-screen state experiences, with appropriate modifications in screening method and algorithm, the two-screen states might convert to single screen operation for CH without loss in performance.

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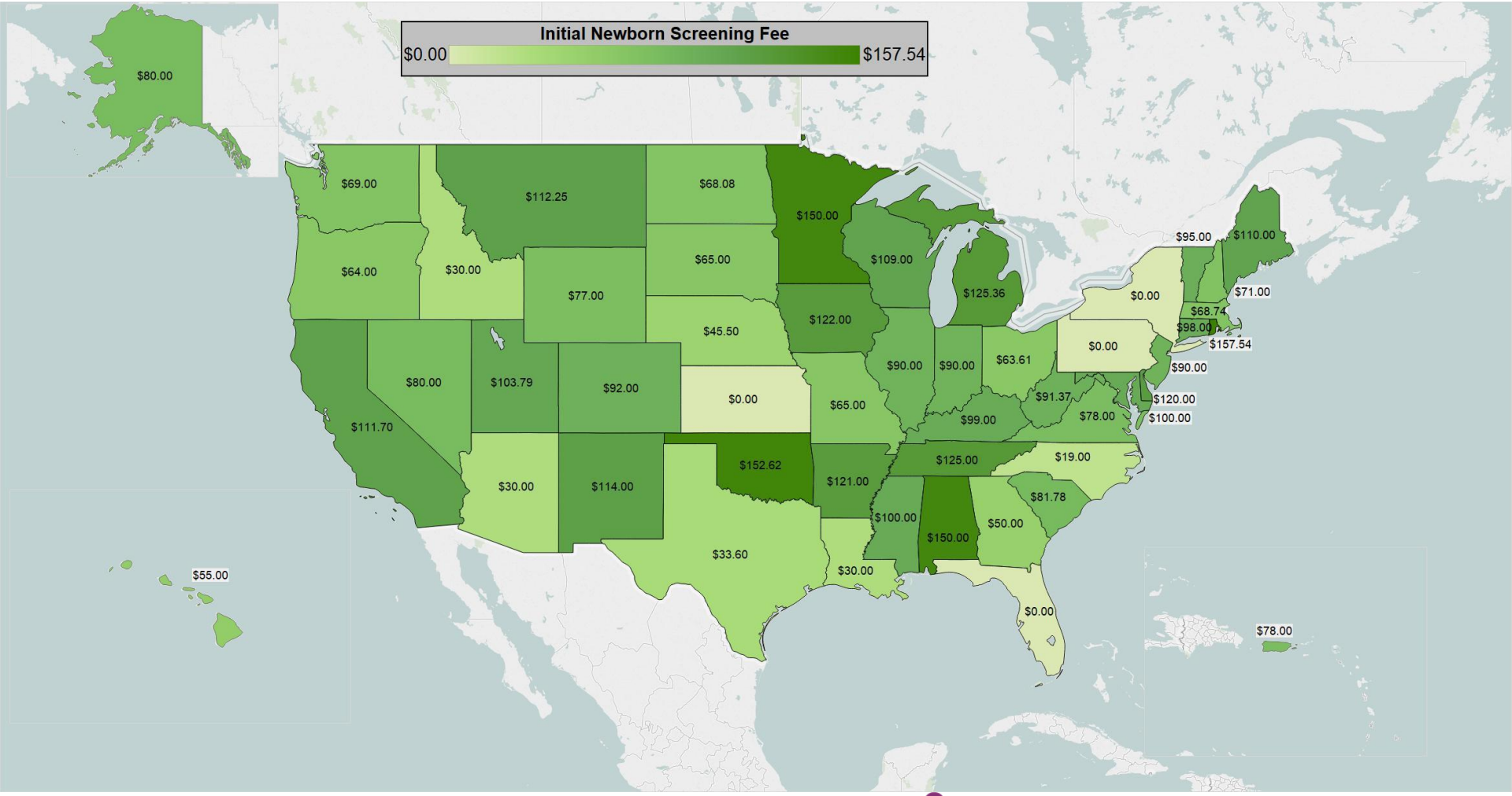
Newborn Screening Fees



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What is covered by the newborn screening fees?

- Program administration
- Laboratory tests (includes salaries of laboratory personnel, supplies, instruments and equipment maintenance)
- Information technology support (lab and general)
- Short-term follow-up services (includes salaries and educational materials)
- Courier services
- Long-term follow-up services
- Bio-bank program
- Metabolic foods and formula

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State Practices: Screening for Disorders

- Recommended Uniform Screening Panel
- Counting the Disorders
  - Core
  - Secondary
  - Other
- Screening for disorders on the Recommended Uniform Screening Panel

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Recommended Uniform Screening Panel
Core Conditions
(As of March 2015)

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 Immunodeficiences						X

Recommended Uniform Screening Panel[1]
SECONDARY[2] CONDITIONS[3]
(As of March 2015)

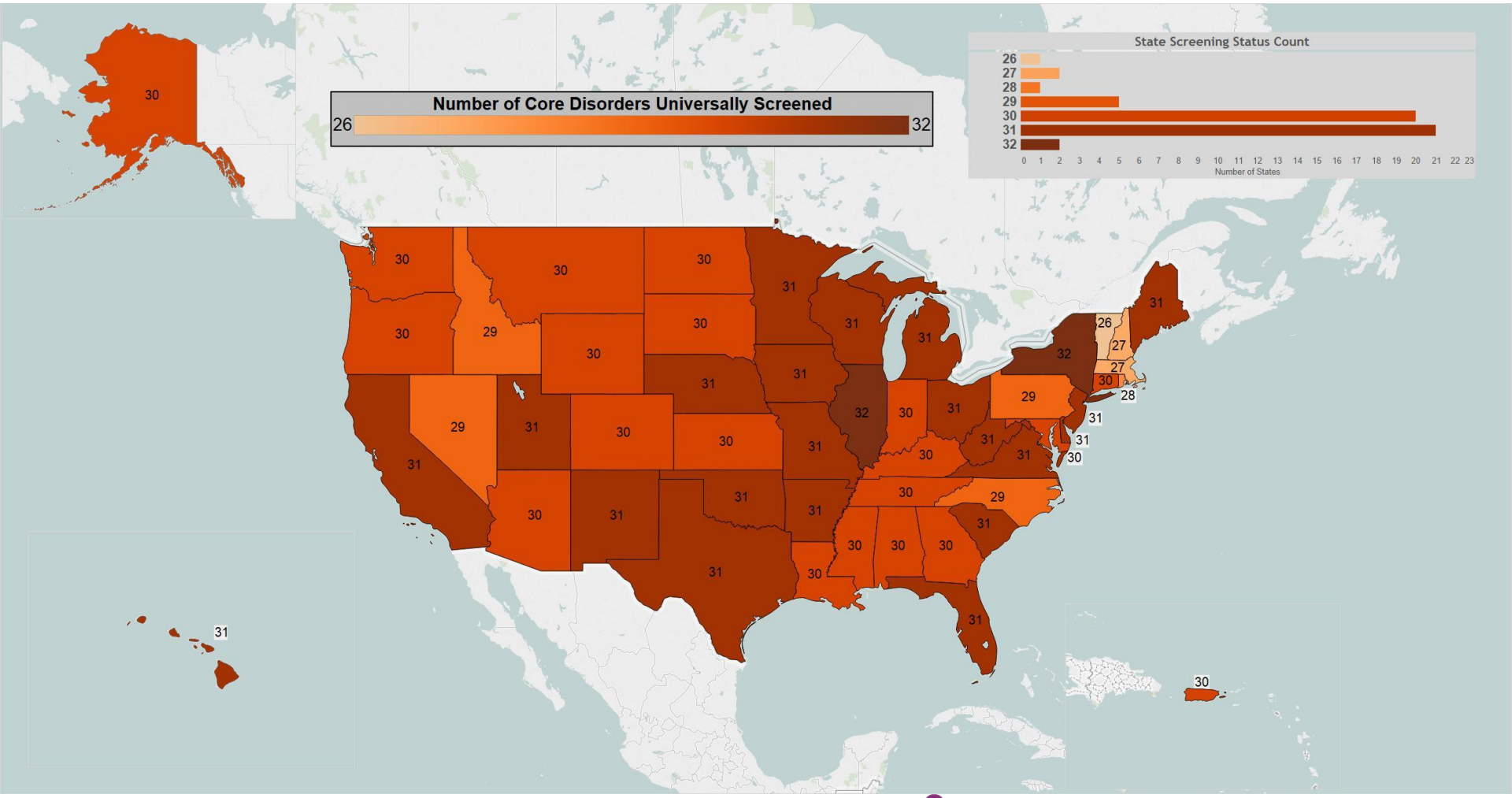
ACMG Code	Secondary Condition	Metabolic Disorder			Hemoglobin Disorder	Other Disorder
		Organic acid condition	Fatty acid oxidation disorders	Amino acid disorders		
Cbl C,D	Methylmalonic acidemia with homocystinuria	X				
MAL	Malonic acidemia	X				
IBG	Isobutyrylglycinuria	X				
2MBG	2-Methylbutyrylglycinuria	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.

Secondary Disorders

"Disorders that can be detected in the differential diagnosis of a core disorder."

Screening of the 32 Core Disorders



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** Screening is on the state panel and fully implemented in the state*

Other disorders screened in the U.S.

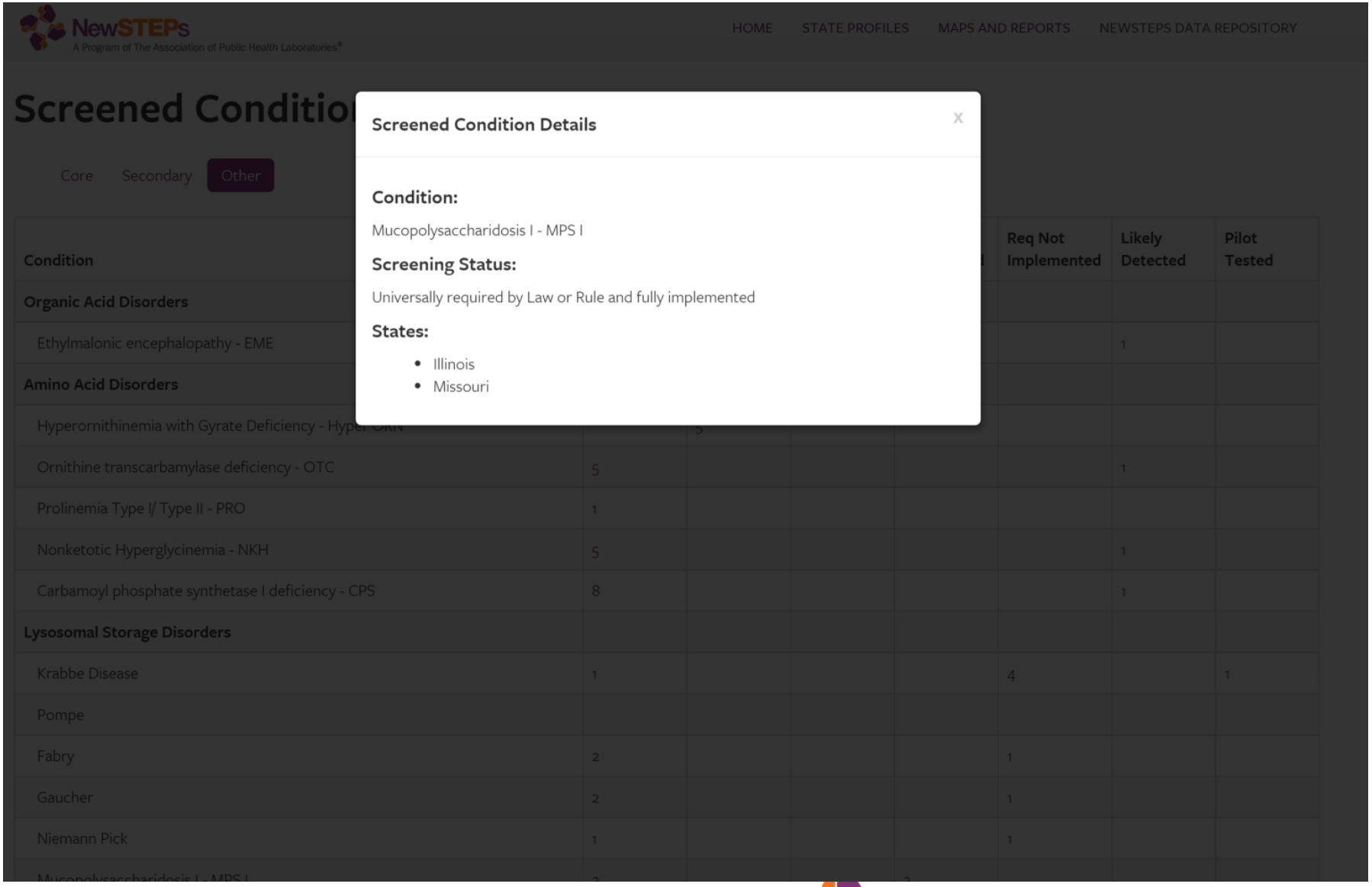
Universally Screened:

- Ethylmalonic encephalopathy – EME (4)
- Hyperornithinemia with Gyrate Deficiency - Hyper ORN (5)
- Ornithine transcarbamylase deficiency – OTC (5)
- Prolinemia Type I/ Type II – PRO (1)
- Nonketotic Hyperglycinemia – NKH (5)
- Carbamoyl phosphate synthetase I deficiency – CPS (8)
- Krabbe (1)
- Fabry (2)
- Gaucher (2)
- Niemann Pick (1)
- Mucopolysaccharidosis I - MPS I (2)
- Glucose-6-phosphate dehydrogenase deficiency - G6PDD/G6PD (2)
- Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome – HHH (10)
- Pyroglutamic acidemia - 5-OXO (3)
- Congenital Toxoplasmosis – TOXO (5)
- Human Immunodeficiency Virus - HIV Exposure (1)
- X-linked Adrenoleukodystrophy (1)

Screened Conditions Report

Core Secondary Other

Condition	Universally Required	Universally Offered	Offered Select	Considered	Req Not Implemented	Likely Detected	Pilot Tested
Organic Acid Disorders							
Ethylmalonic encephalopathy - EME	4					1	
Amino Acid Disorders							
Hyperornithinemia with Gyrate Deficiency - Hyper ORN		5					
Ornithine transcarbamylase deficiency - OTC	5					1	
Prolinemia Type I/ Type II - PRO	1						
Nonketotic Hyperglycinemia - NKH	5					1	
Carbamoyl phosphate synthetase I deficiency - CPS	8					1	
Lysosomal Storage Disorders							
Krabbe Disease	1				4		1
Pompe							
Fabry	2				1		
Gaucher	2				1		
Niemann Pick	1				1		
Mucopolysaccharidosis I - MPS I	2			2			



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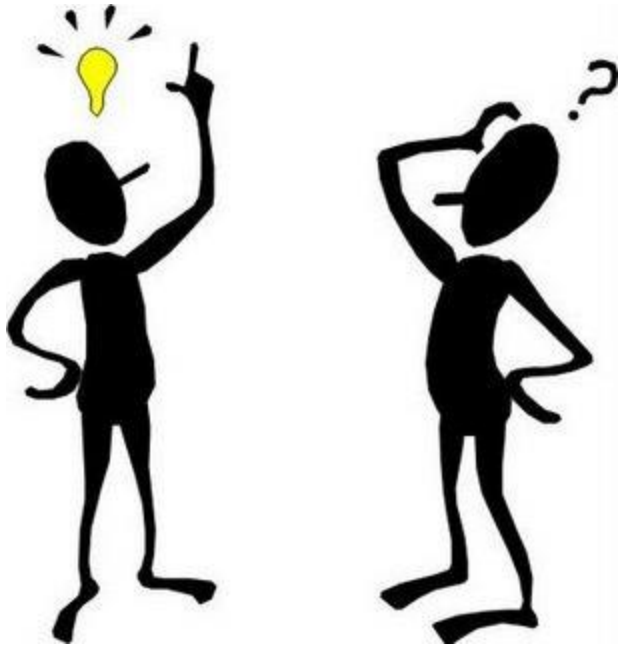
Decision making, policies

- Advisory committees, board of health, commissioner of health
- Legislators



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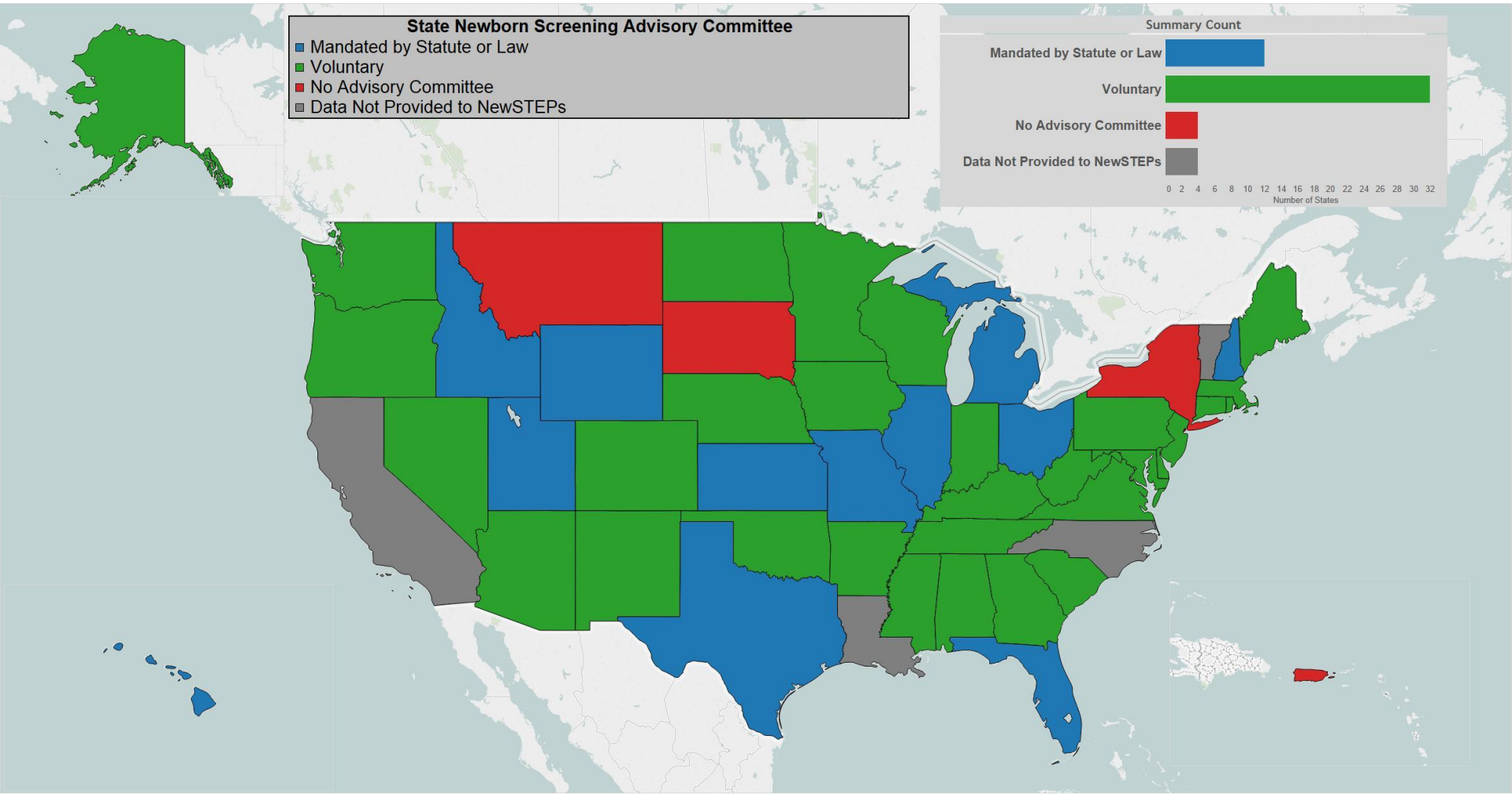
Advisory committees across the U.S.



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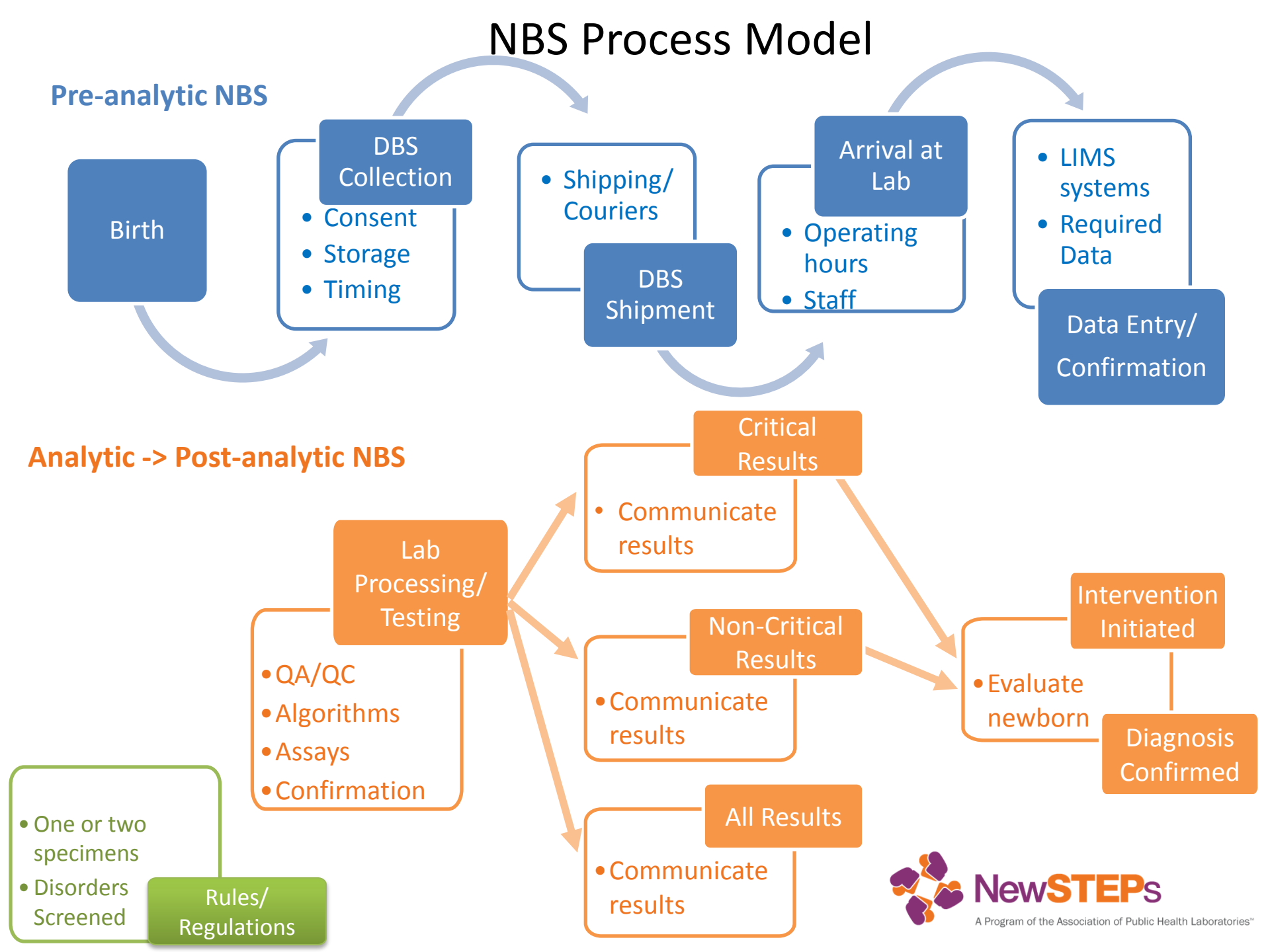
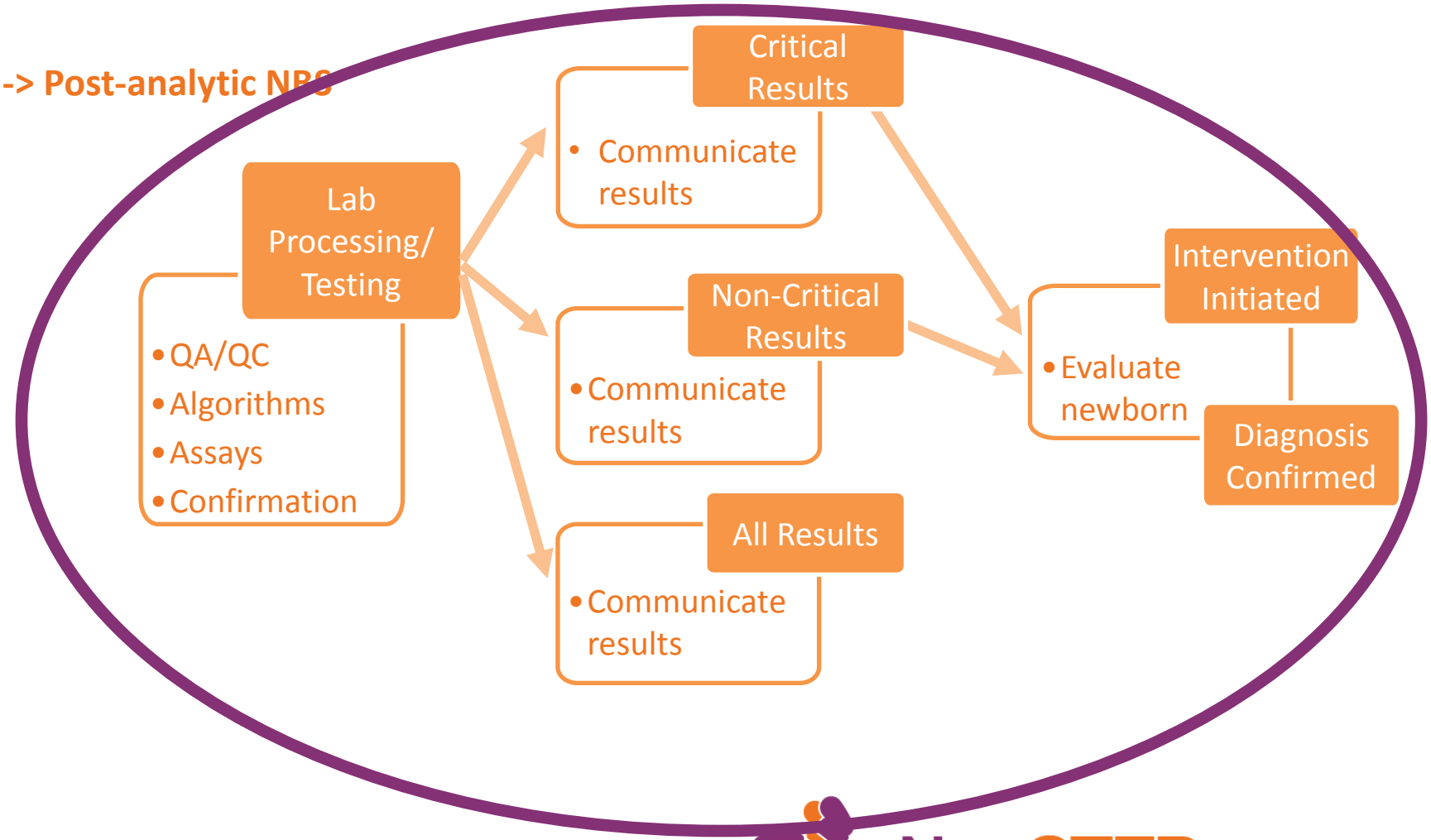
Composition of advisory committees

- Consumers or parents of patients affected by screened conditions
- Laboratory representatives of pathology and chemistry
- Pediatric, neonatology and family practitioners
- Pediatric subspecialists (e.g. Endocrine, Hematology, Metabolic etc.)
- Metabolic nutritionists
- Hospital association representative
- March of Dimes representative
- Medical ethicist
- NBS program (management, follow-up and lab) representatives

NBS Process Model



Analytic -> Post-analytic NBS



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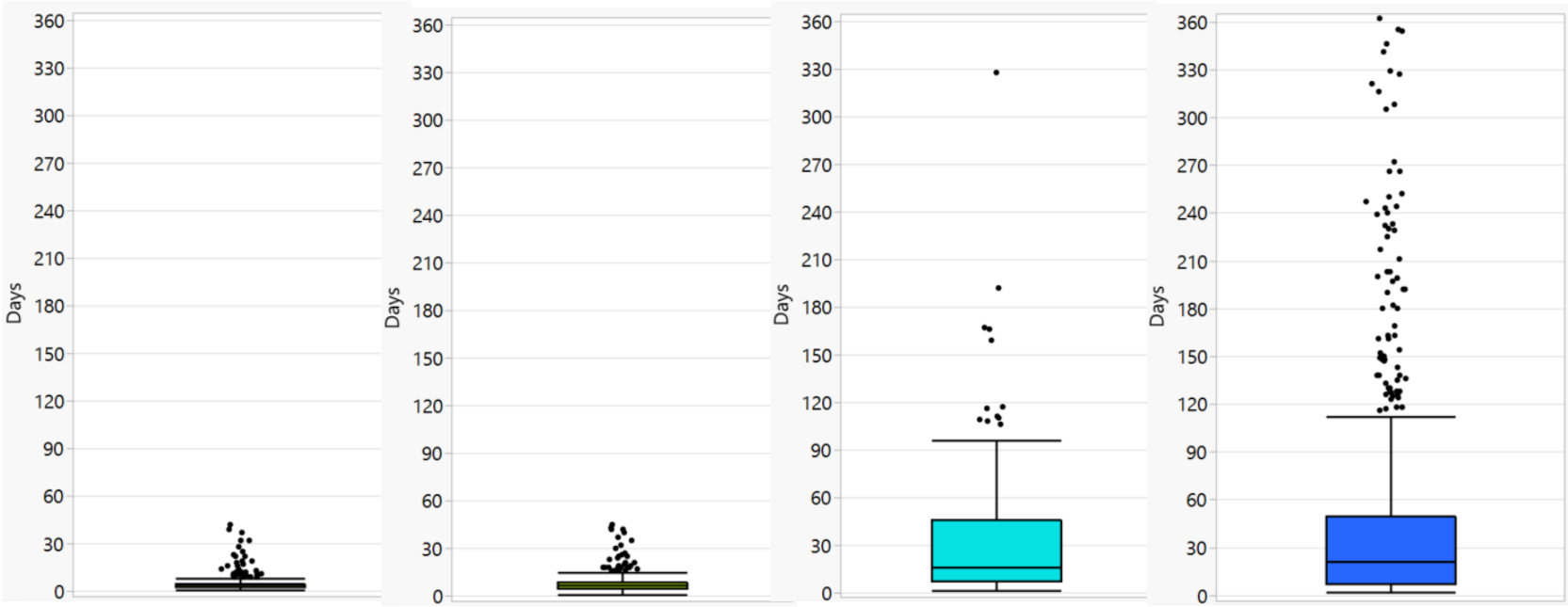
Timeliness outcomes in infants diagnosed with disorders



Birth to Receipt by lab

Birth to Report of Results

Birth to Intervention

Birth to Diagnosis

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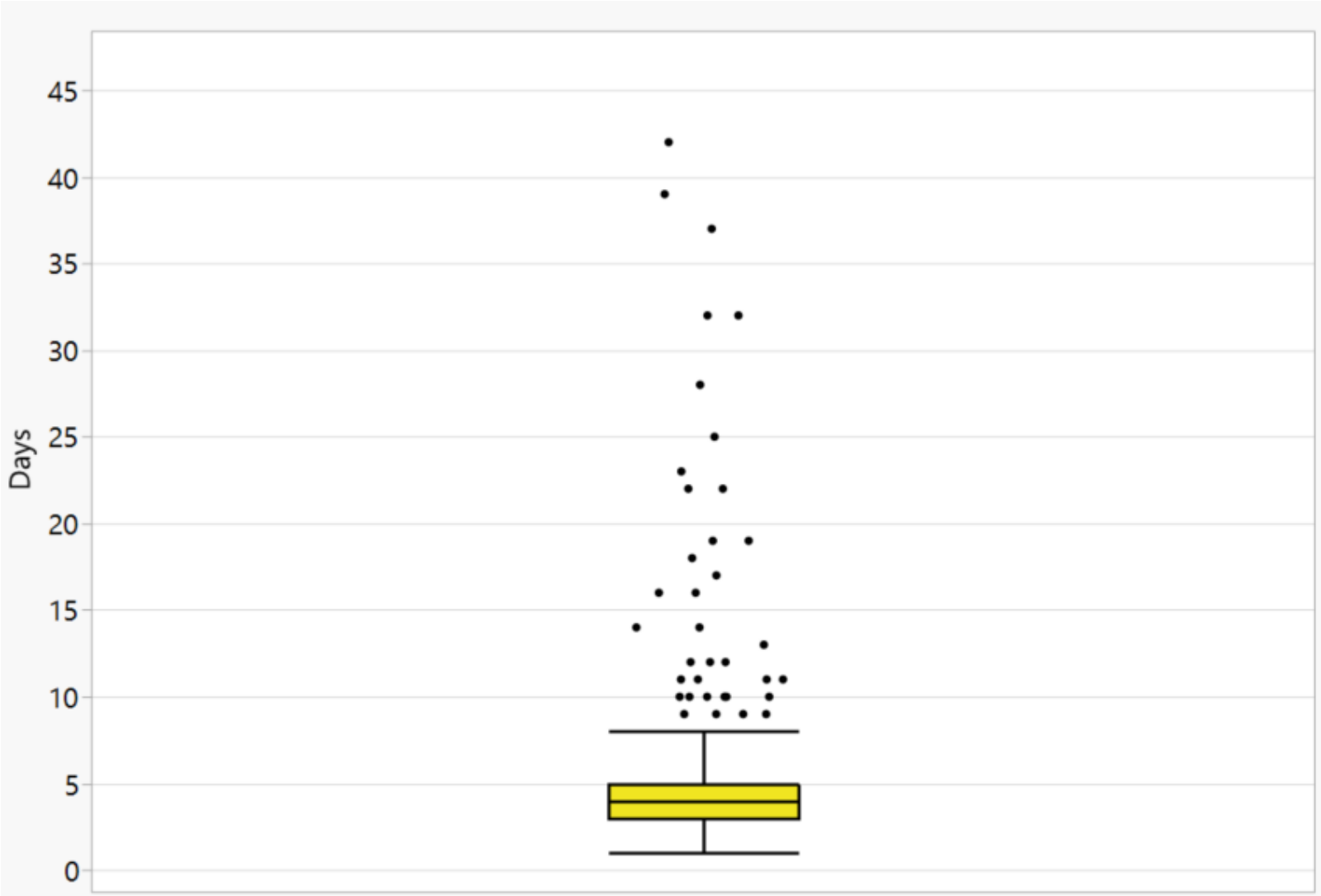
Time to Receipt by Lab



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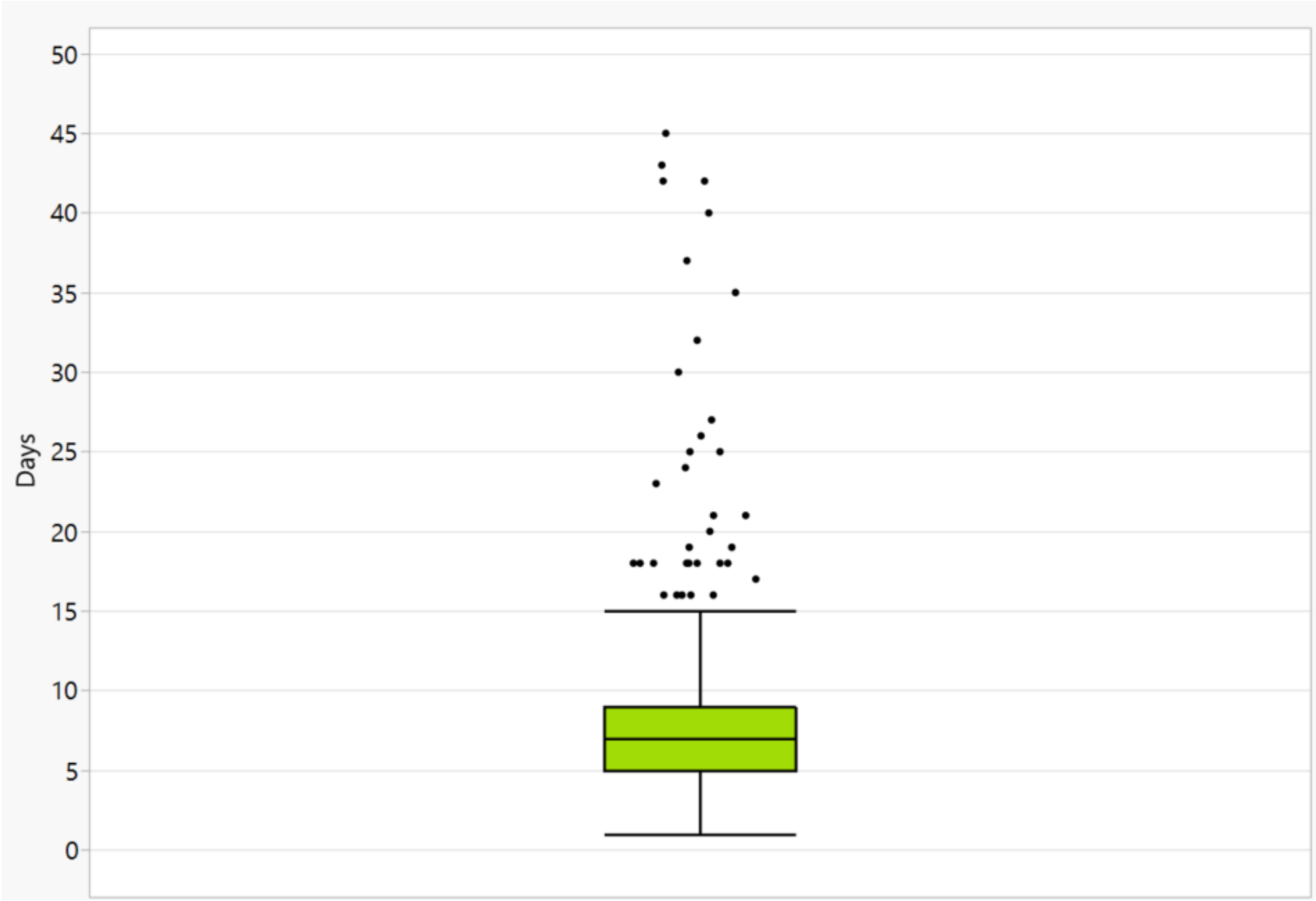
Time to Release of Out of Range Results



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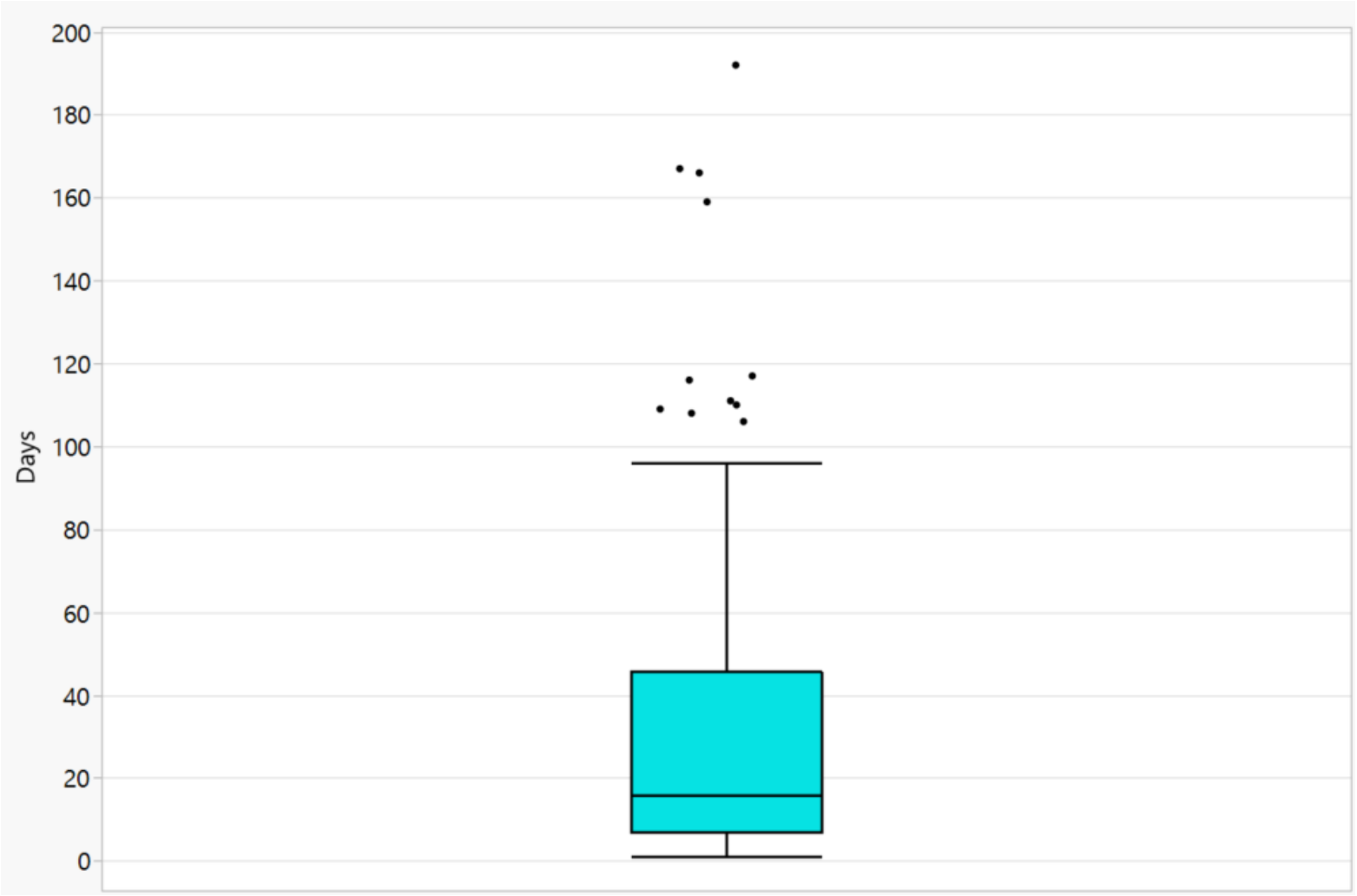
Time to Intervention



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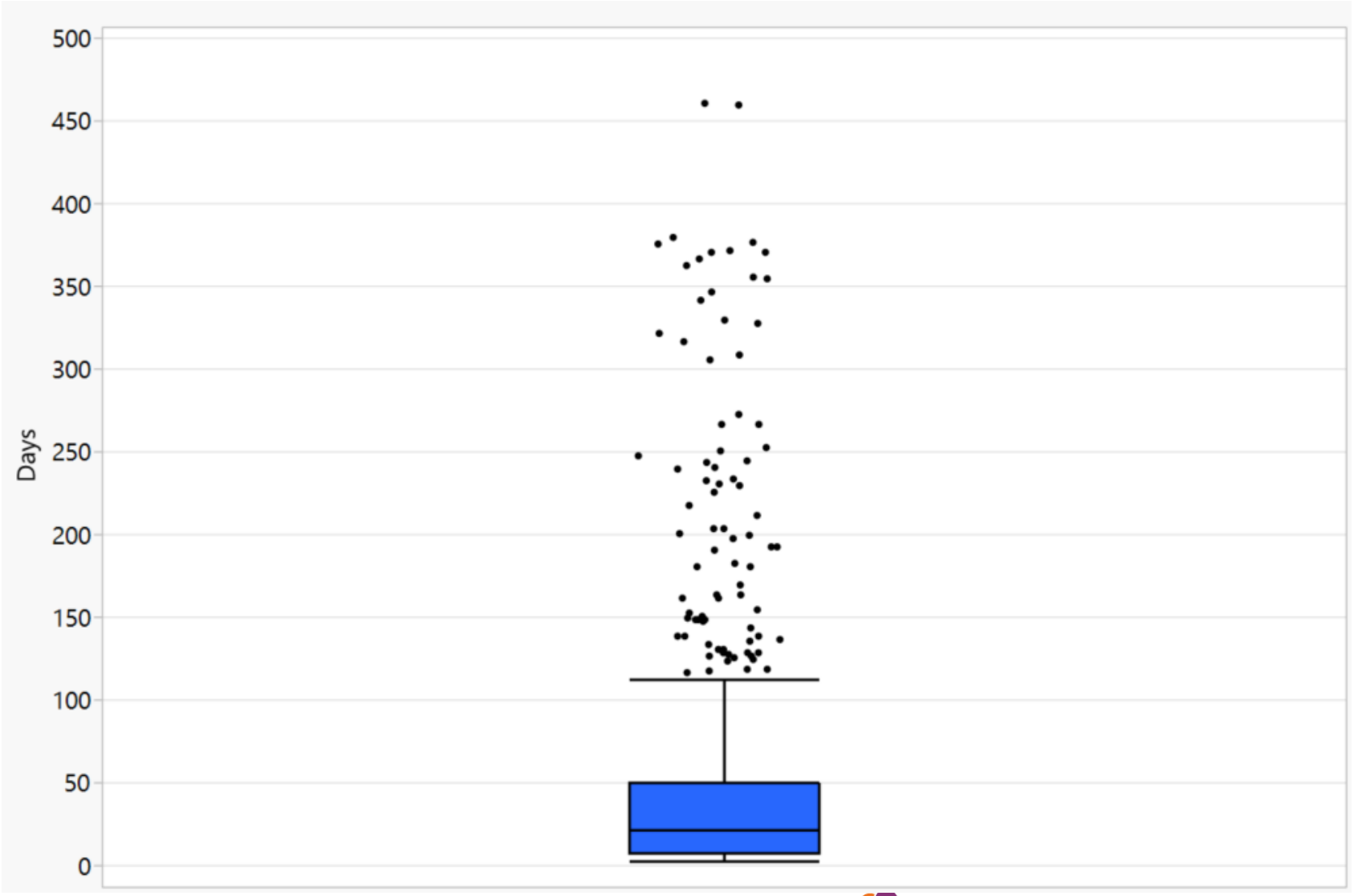
Time to Confirmed Diagnosis



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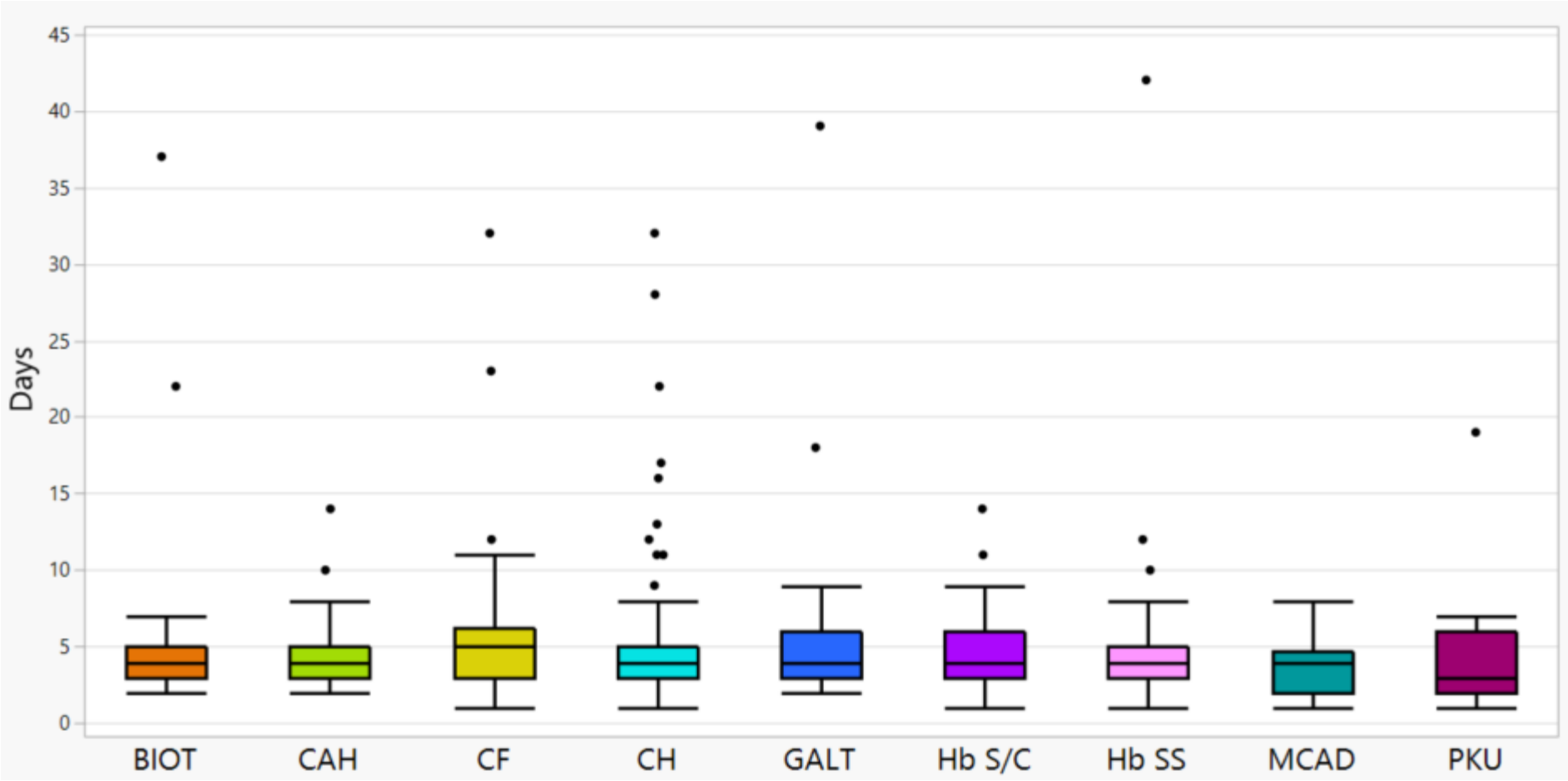
Time to Receipt by Lab by Disorder



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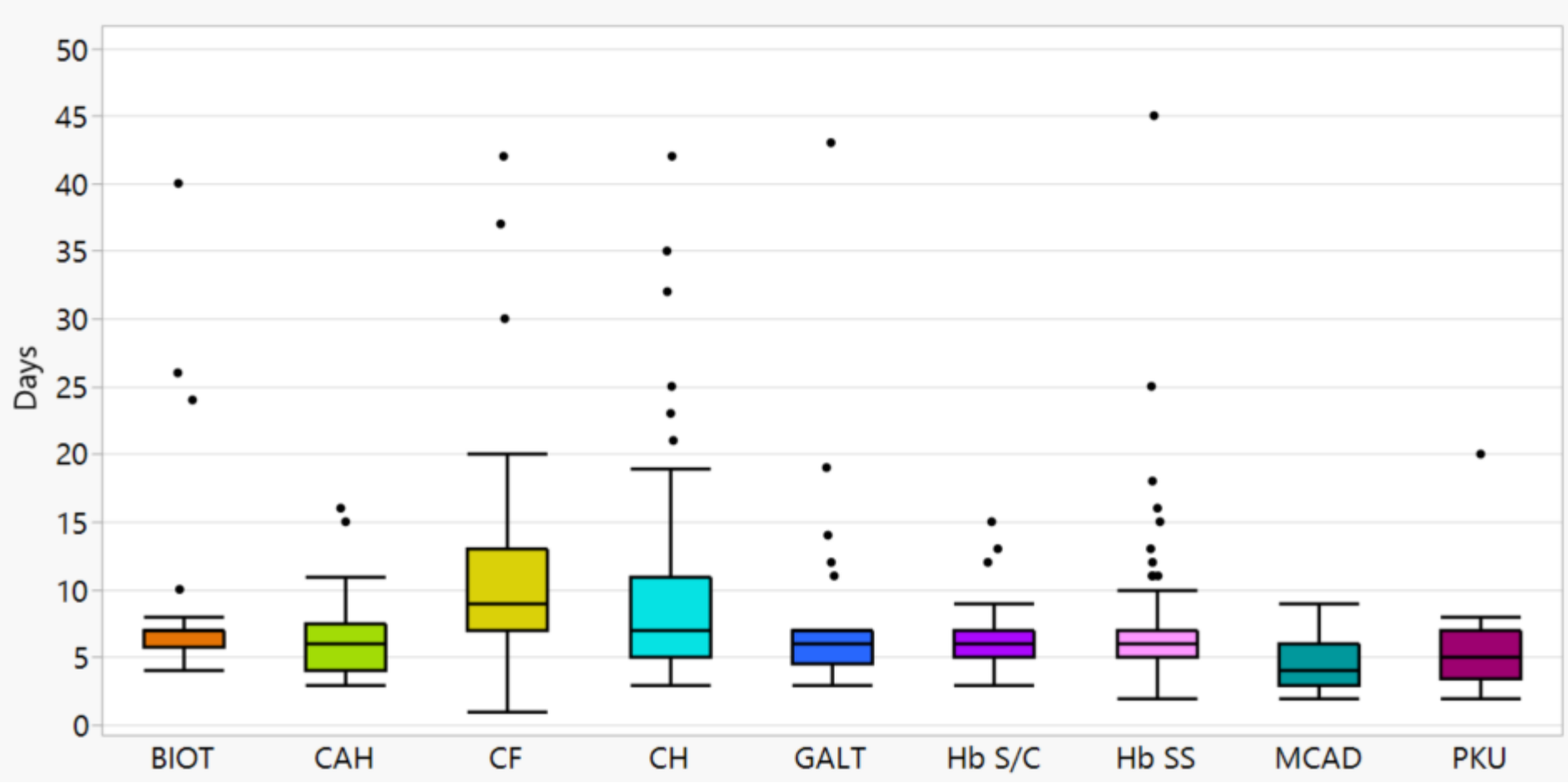
Time to Release of Out of Range Results by Disorder



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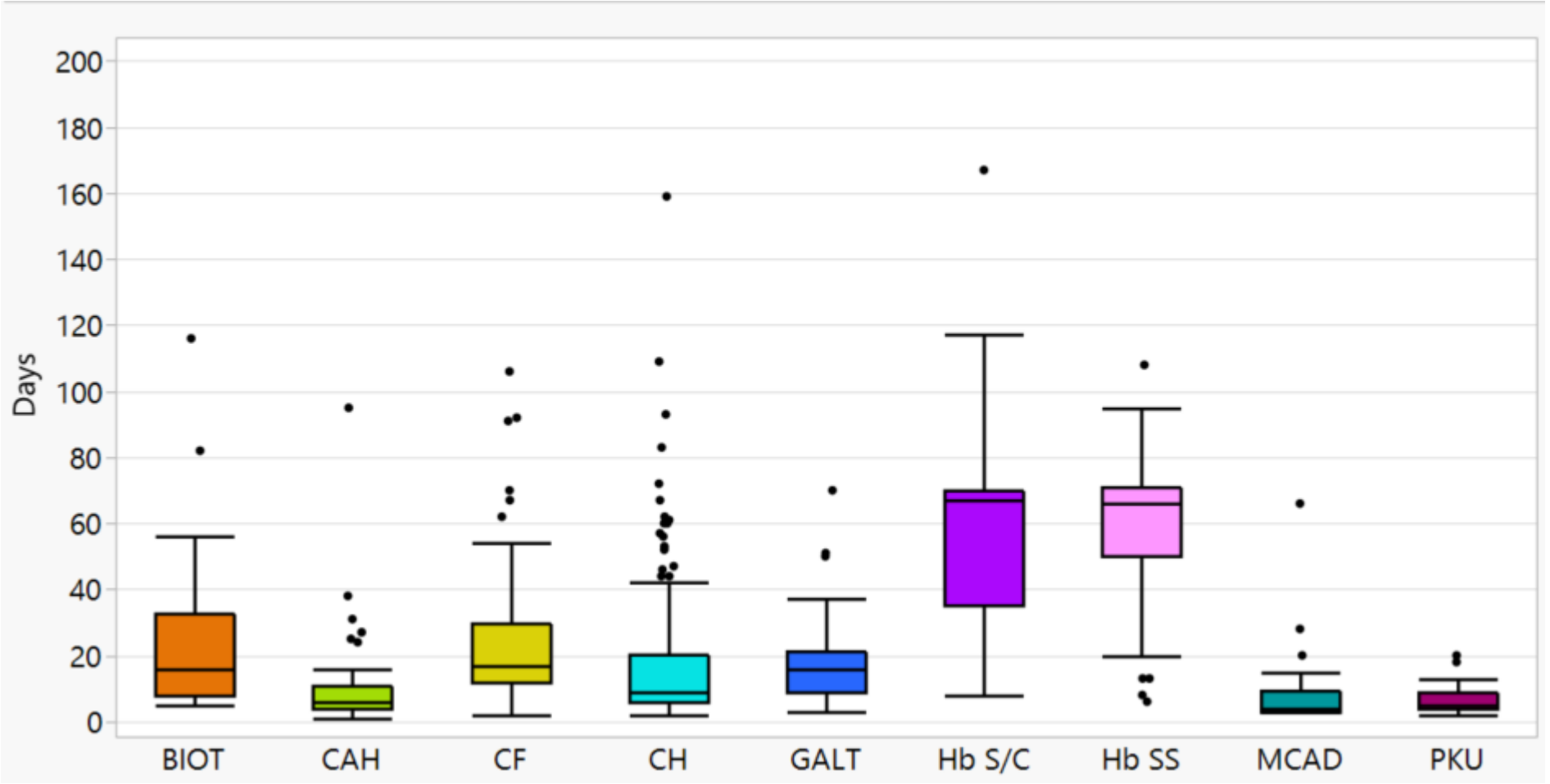
Time to Intervention by Disorder



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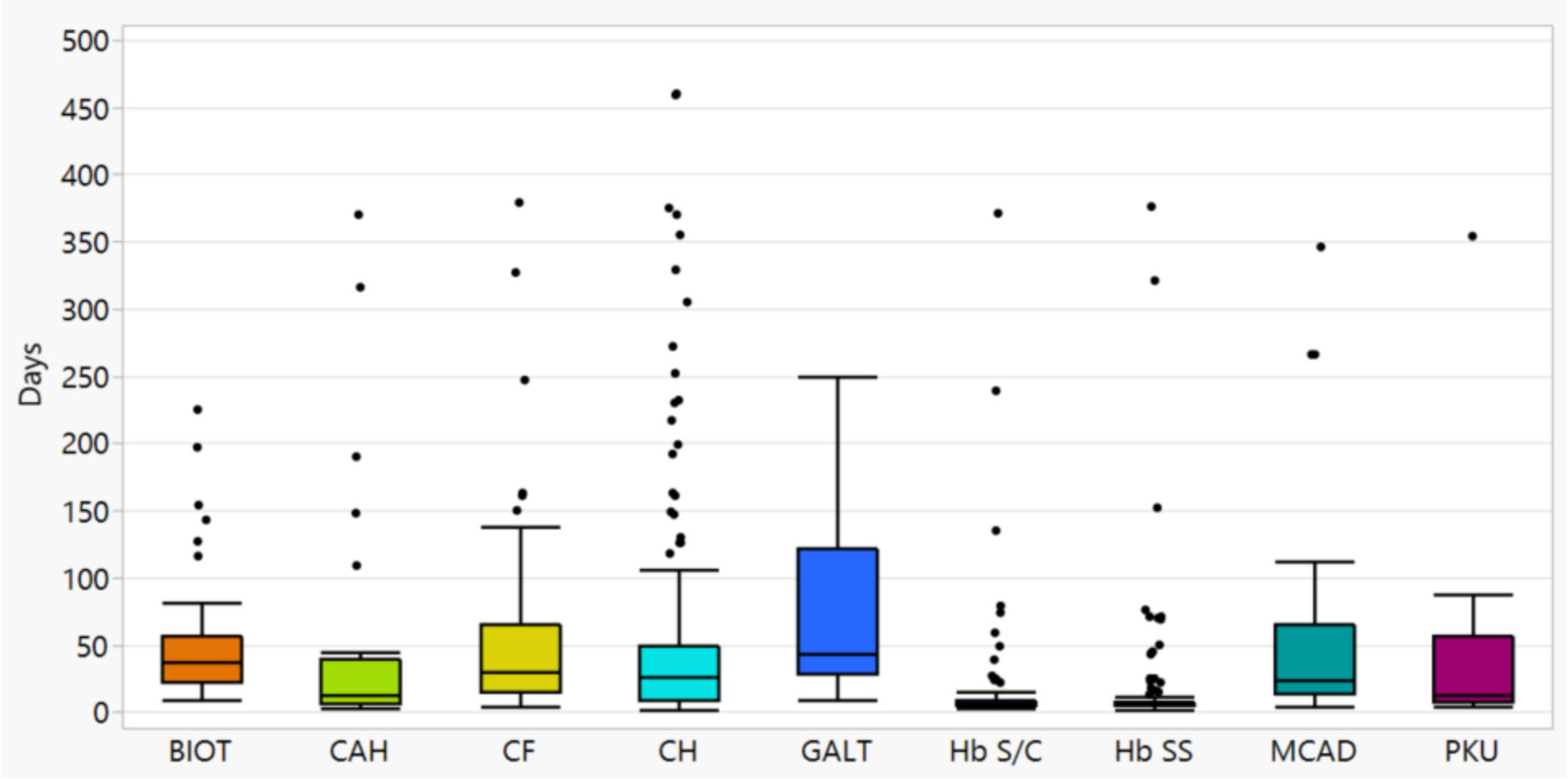
Time to Confirmed Diagnosis by Disorder



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Cystic Fibrosis

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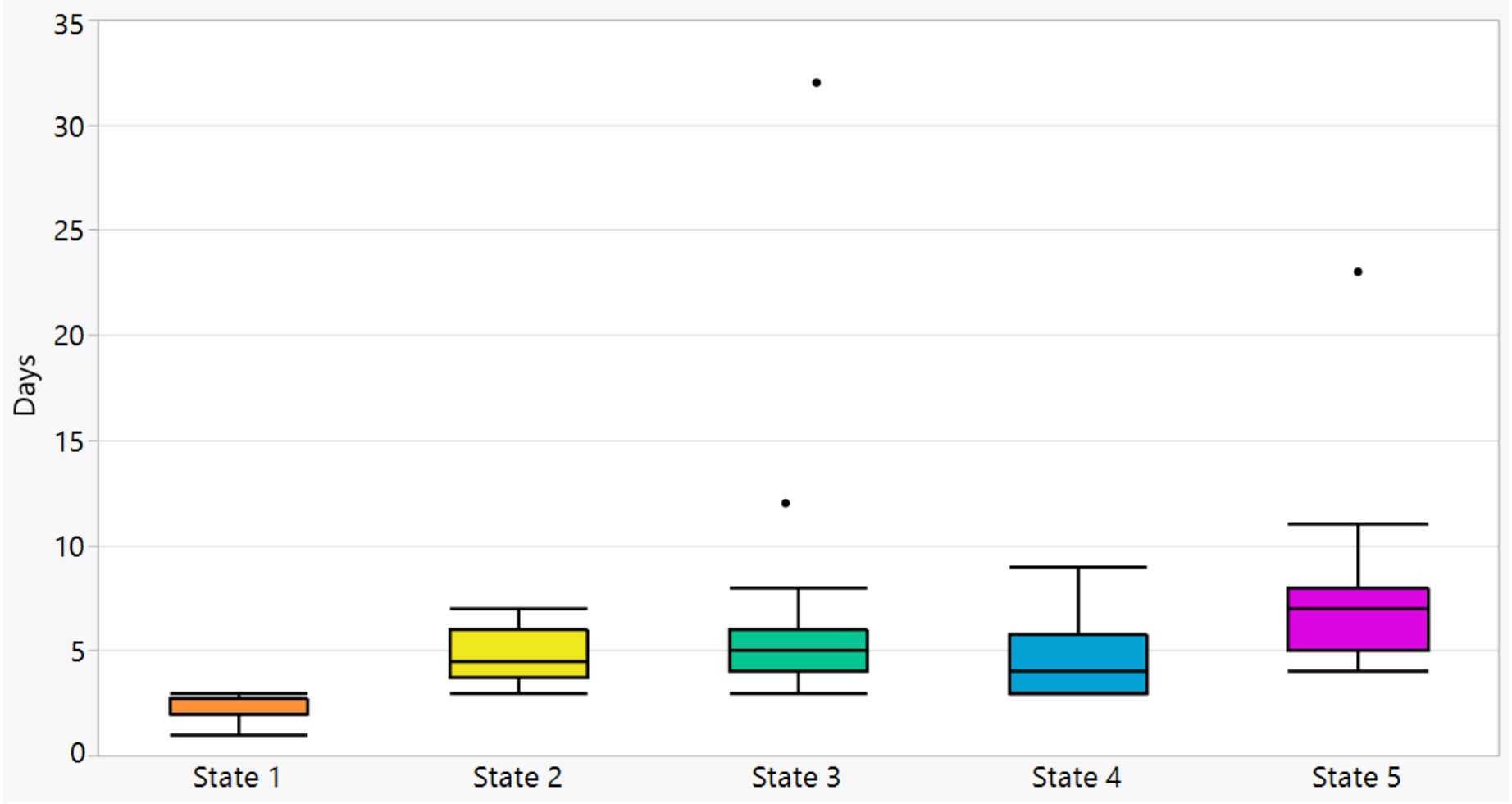
Cystic Fibrosis
Time to Receipt by Lab by State



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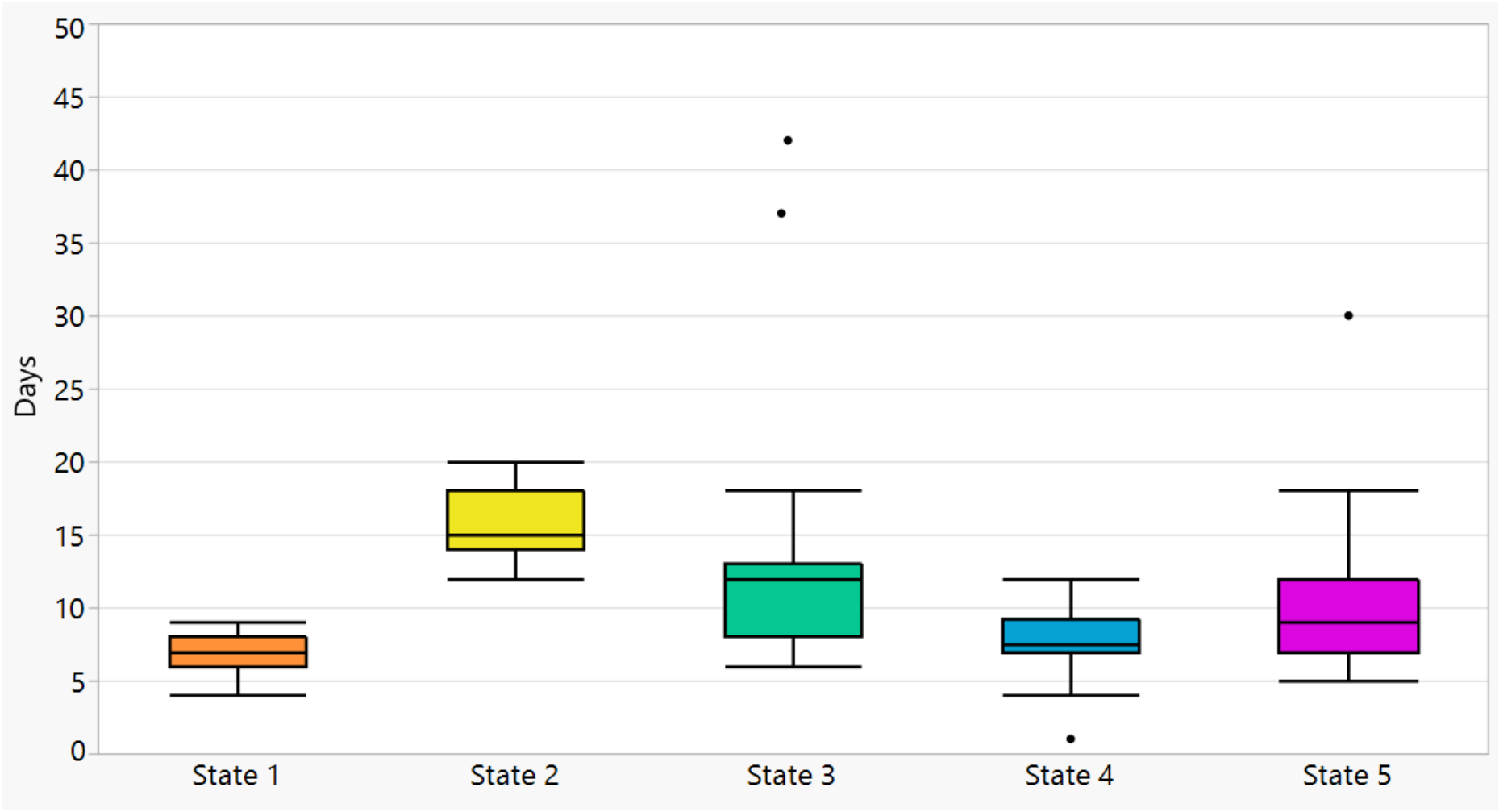
Cystic Fibrosis
Time to Release of Out of Range Results



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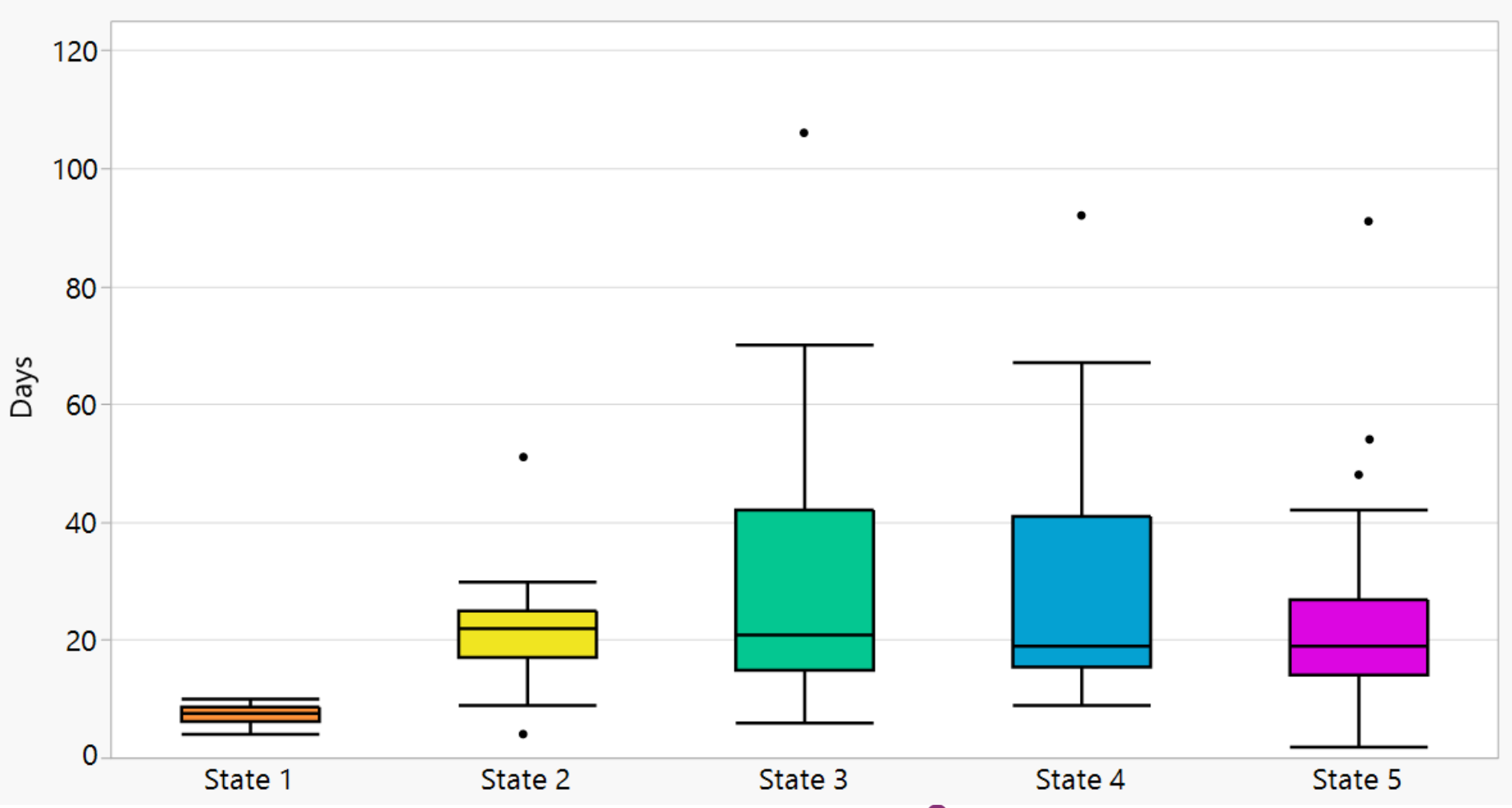
Cystic Fibrosis
Time to Intervention by State



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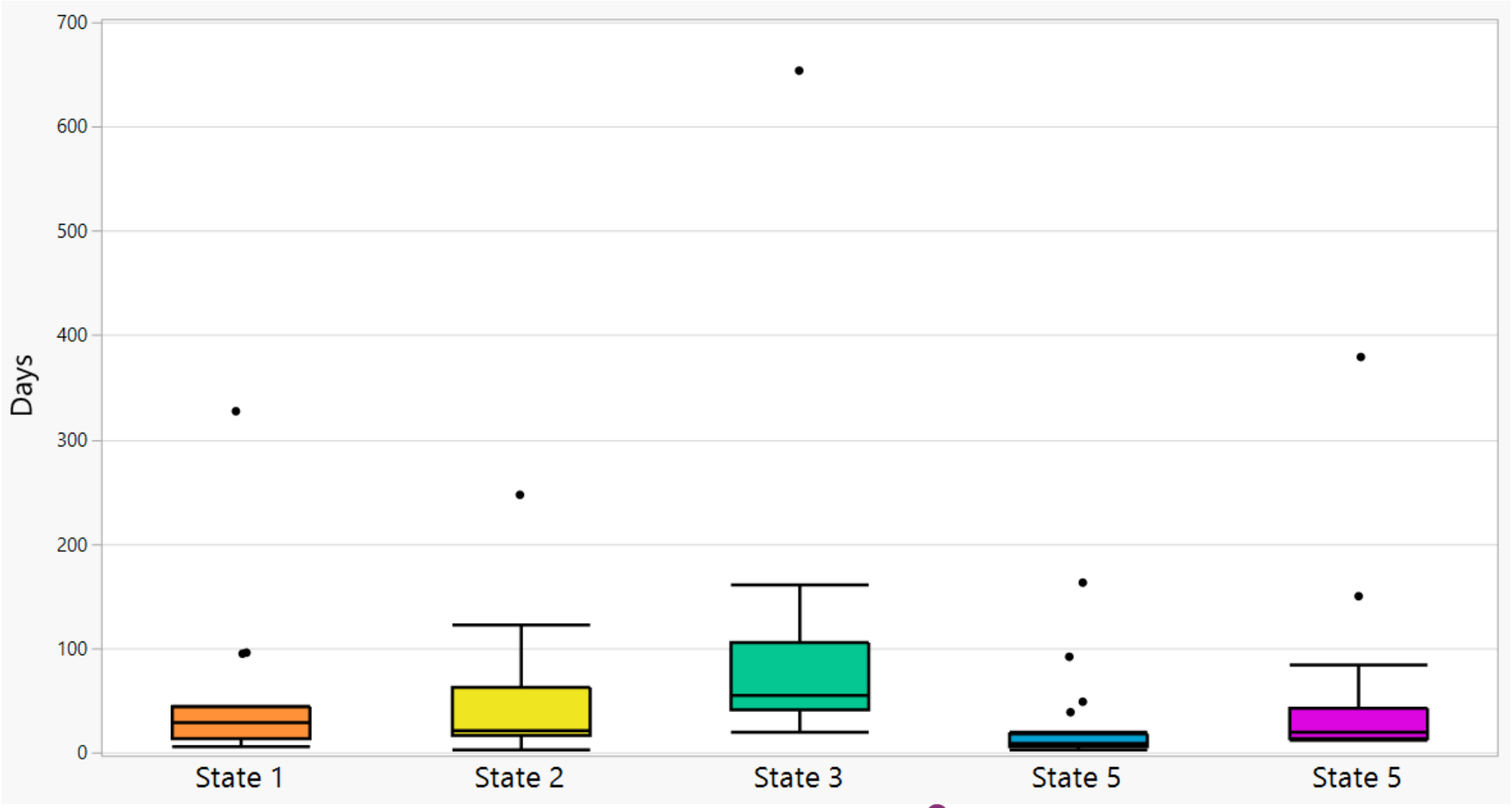
Cystic Fibrosis
Time to Confirmed Diagnosis by State



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Congenital Hypothyroidism

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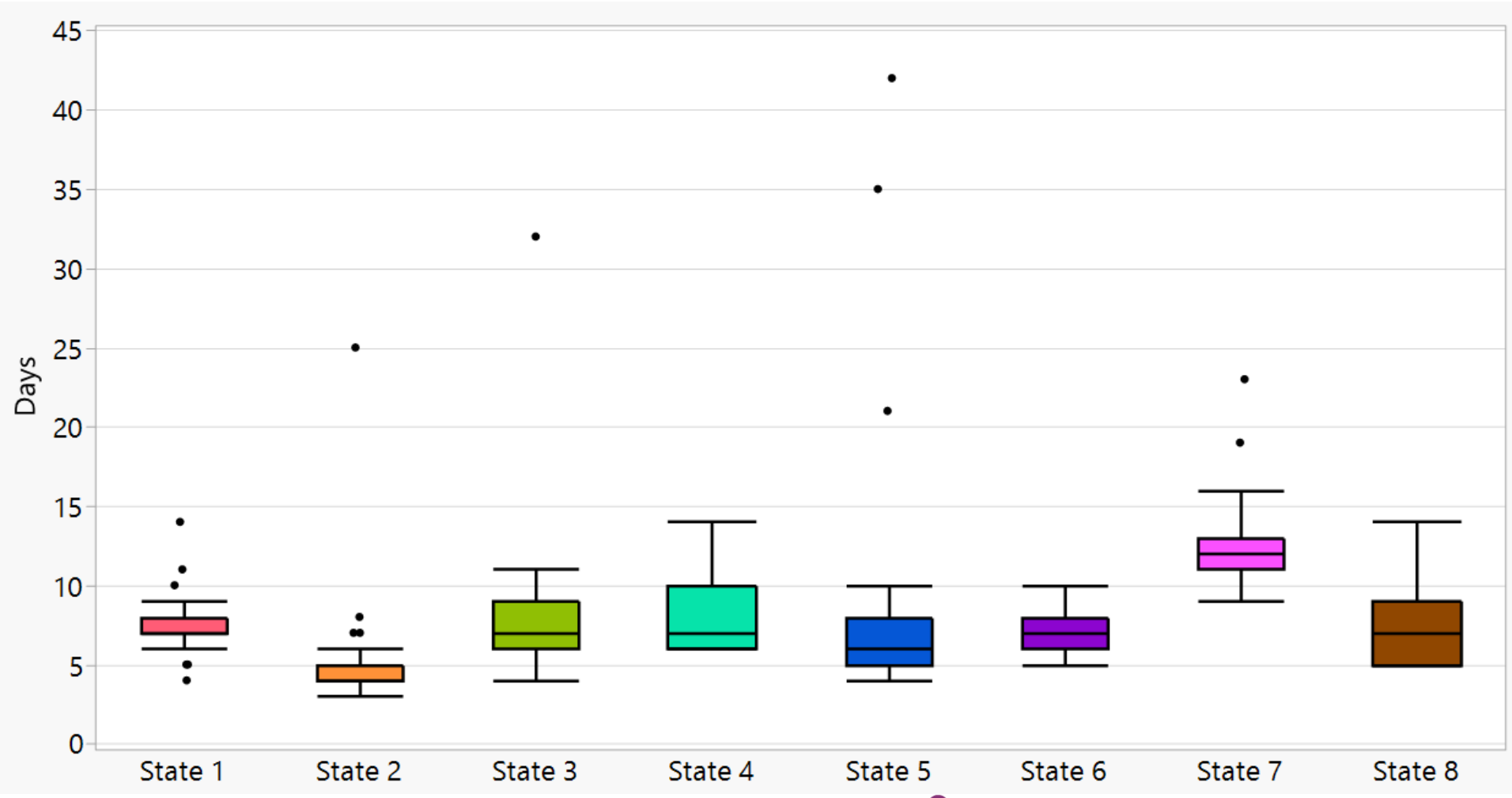
Congenital Hypothyroidism
Time to Release of Out of Range Results



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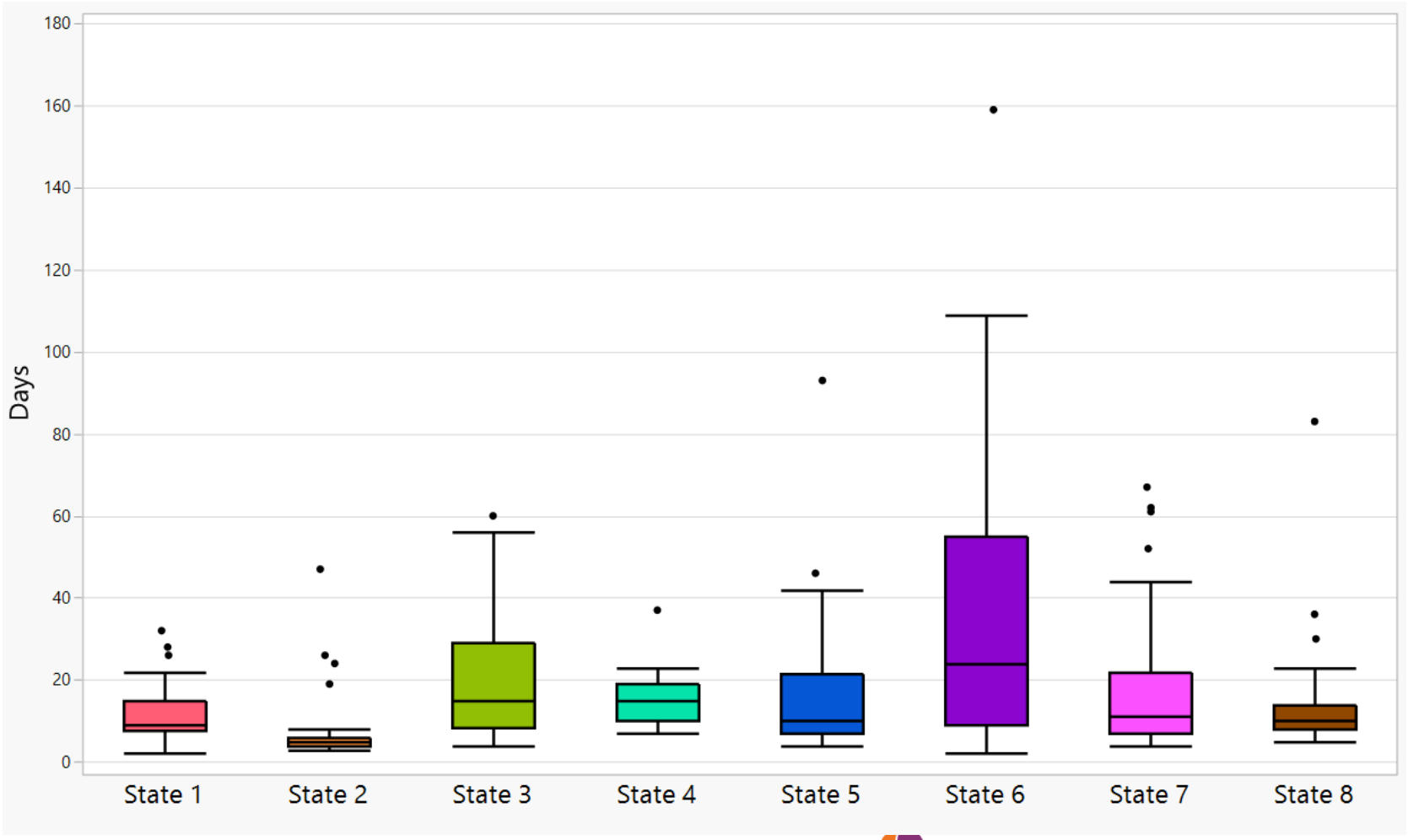
Congenital Hypothyroidism
Time to Intervention by State



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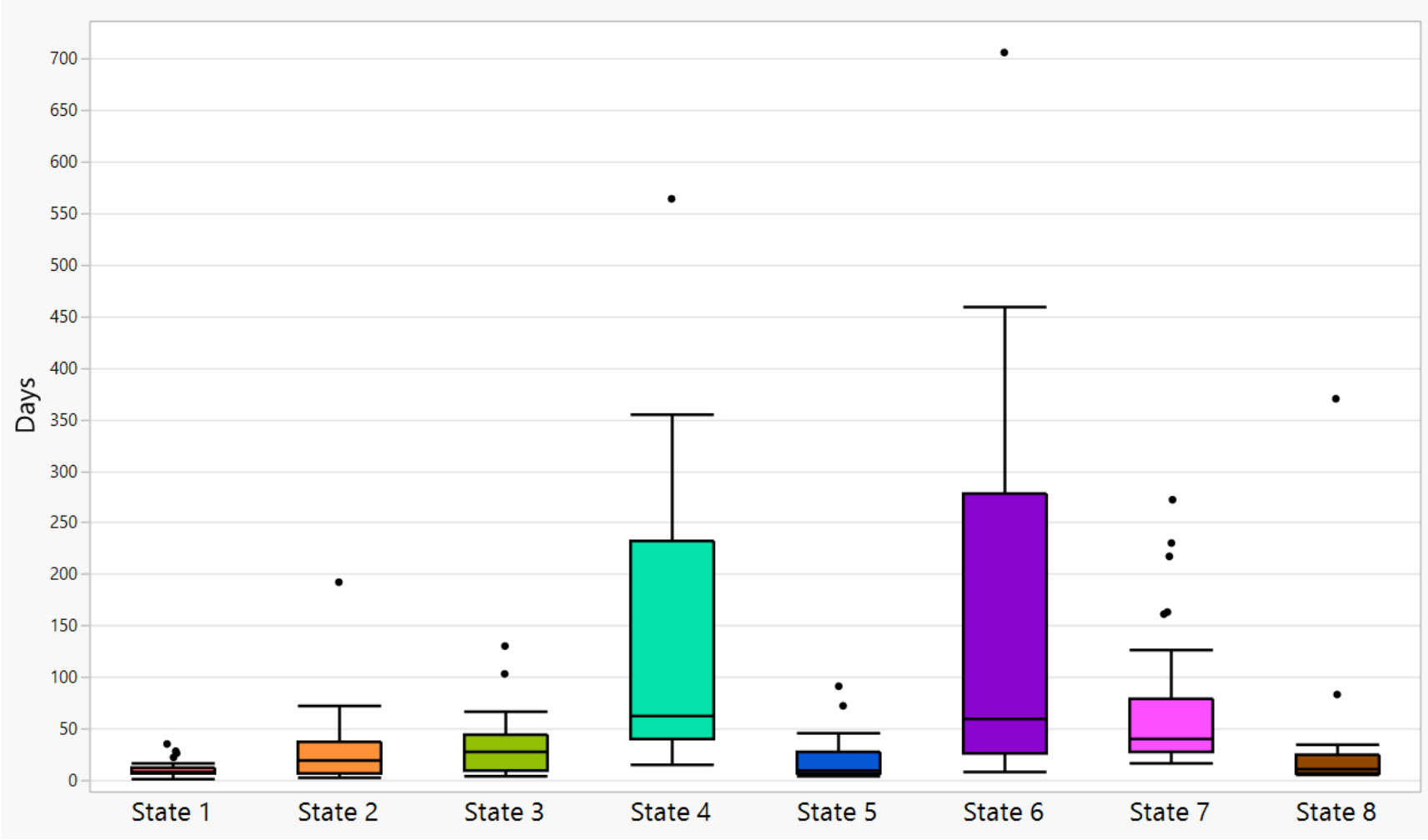
Congenital Hypothyroidism
Time to Confirmed Diagnosis by State



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Quality Indicators

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Quality Indicator Data

- Overview of indicators
- Challenges in data collection. States pull data from different sources.
- Partnering with LIMS vendors
  - Efforts to ensure data are consistent
  - Differences in local collection
- Data collection is deliberate – and will result in high quality data

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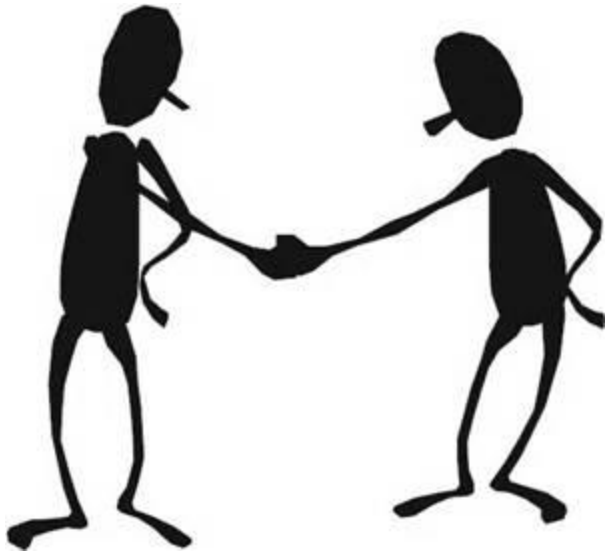
Conversations with Vendors

- Partnering with PerkinElmer and Natus to develop queries that will collect data in a systematic way from all states
- Will expand to other vendors and states with locally-developed systems



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Challenges Uncovered in Data Collection

- Example: QI 1a: **Percent of invalid dried blood spot specimens/cards due to improper collection:** Number of dried blood spot specimens/cards on which labs cannot report a complete newborn screening panel due to improper collection errors [occurring pre-analytic] divided by number of specimens submitted, multiplied by 100.

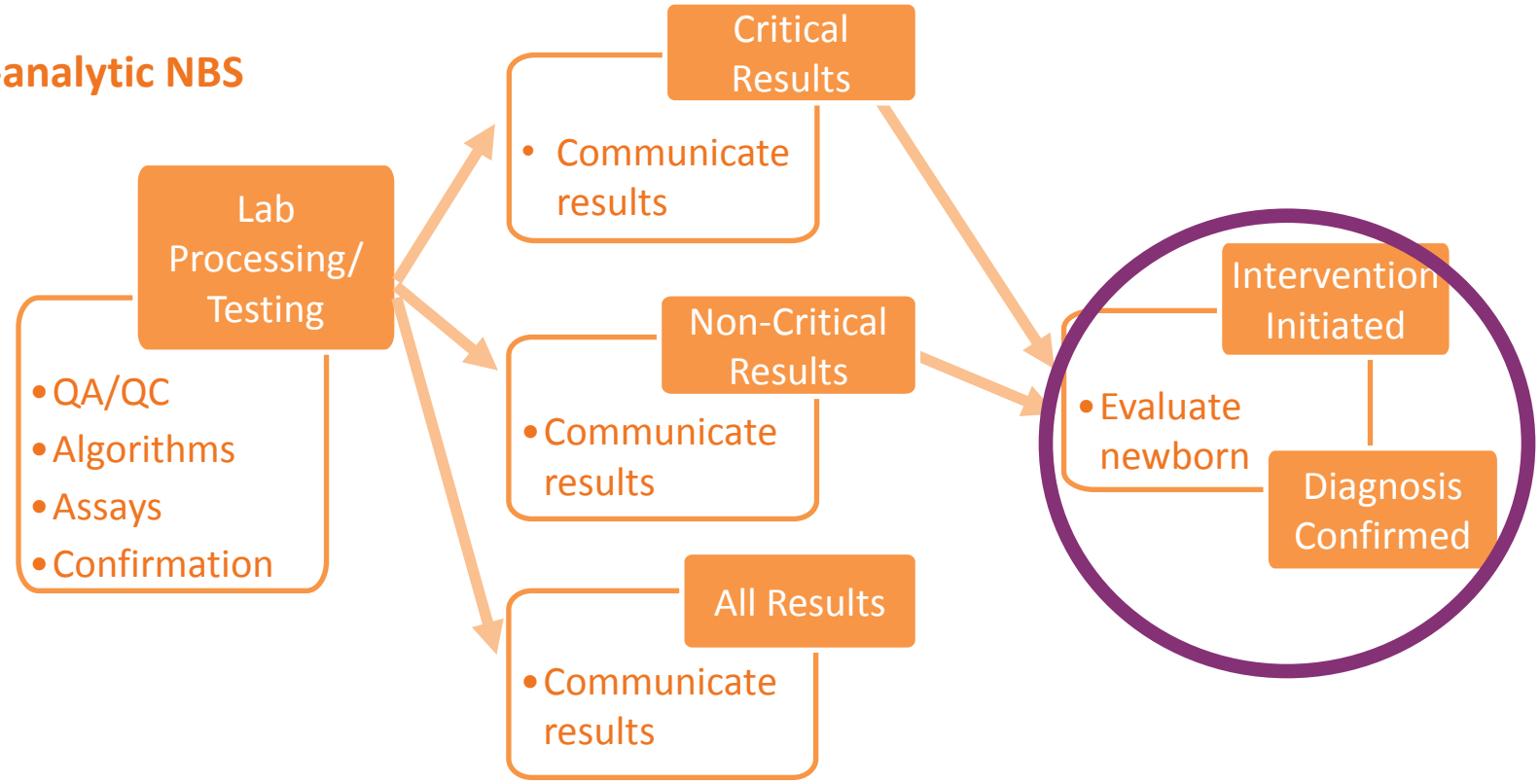
Colorado's adaptation of the LIMS system

- Potential reasons for unsatisfactory due to collection
  - No blood applied
  - No blotter with slip
  - Incorrect form
  - Clots or uneven
  - Serum separation
  - Contaminated
  - Multiple application
  - Incomplete submission
  - Quantity not sufficient

Next steps

- Timelines
- Where do we go from here with the data
  - Natus
  - PerkinElmer
  - Others…not all states are covered by these LIMS vendors. How do we extend the lessons learned?

Analytic -> Post-analytic NBS



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Newborn level data collected within NewSTEPs

Purpose

"**To** provide an accurate characterization of the frequency of newborn screening disorders in the U.S., along with timing of screening and diagnostic activities"

Systematic definitions helpful at local AND national levels

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Cystic Fibrosis Example

- Newborn with abnormal newborn screen:
  - IRT 105 ng/ml (normal range < 60 ng/ml)
  - NBS DNA analysis revealed F508/R117H; 7T/9T
  - Referred to CF Center for Sweat Test
  - Sweat test results: 25 mmol/L (diagnostic > 60mmol/L)

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CF Diagnosis can vary by clinician

- Dr. Smith: Baby likely has **CF**. Follow monthly and repeat sweat test; tell family baby has CF.
- Dr. Jones: Baby has **CRMS** (Cystic Fibrosis Related Metabolic Syndrome). Not CF, we should follow this baby every 6 months to see if baby develops CF symptoms
- Dr. Garcia: Baby is fine, **no CF, no CRMS**. No diagnosis, baby does not need to be seen.

Surveillance case definitions

- This newborn would be classified as CRMS using the case definitions

- The burden of CRMS in the U.S. is not well understood

Two Cases from the Repository

- Case 1
  - Elevated IRT
  - 2 mutations known to be disease causing on NBS
  - Sweat test >60mmol/L
  - Repeat sweat test 30-59mmol/L
  - No repeat DNA test
- Diagnosed as CRMS

- Case 2
  - Elevated IRT
  - 2 mutations known to be disease causing on NBS
  - Sweat test 30-59mmol/L
  - Repeat sweat test quantity not sufficient
  - No repeat DNA test
- Diagnosed as CF

Challenges and Solutions

- Culture change
  - Time commitment
  - Developing communication avenues
- Case Definition Implementation Workgroup
  - Marketing
  - Communication
- New Disorders on RUSP
  - CCHD
  - SCID
  - Pompe
  - MPSI
- Manuscript in preparation

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Efforts to Support NBS Programs and Timeliness

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Collaborative Improvement and Innovation Network: CoIIN

- Eight states participating in continuous quality improvement activities to address challenges in timeliness:
  - Arizona, California, Colorado, Iowa, New Hampshire, Tennessee, Texas, Wyoming
- Fifteen-month project, teams of 5 individuals from states, comprised of laboratory, follow-up, hospital staff
- Sharing ideas and collaborating to find solutions

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NewSTEPs 360

A Partnership between The Colorado School of Public Health and The Association of Public Health Laboratories™

- Funding starting September 1
- Will support at least 20 state newborn screening programs to improve timeliness over three years
- Build on success of CoIIN
- Competitive funding opportunity

NewSTEPs 360 and related activities are funded through a cooperative agreement (#UG8MC28554) to ColoradoSPH by the Genetic Services Branch of the Health Resources and Services Administration (HRSA).

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Project Instant Gratification (PIGs)

- Giving tools back to states that help them to do their jobs
  - Did You Know E-mails
  - Run Charts
  - Personalized QI Reports

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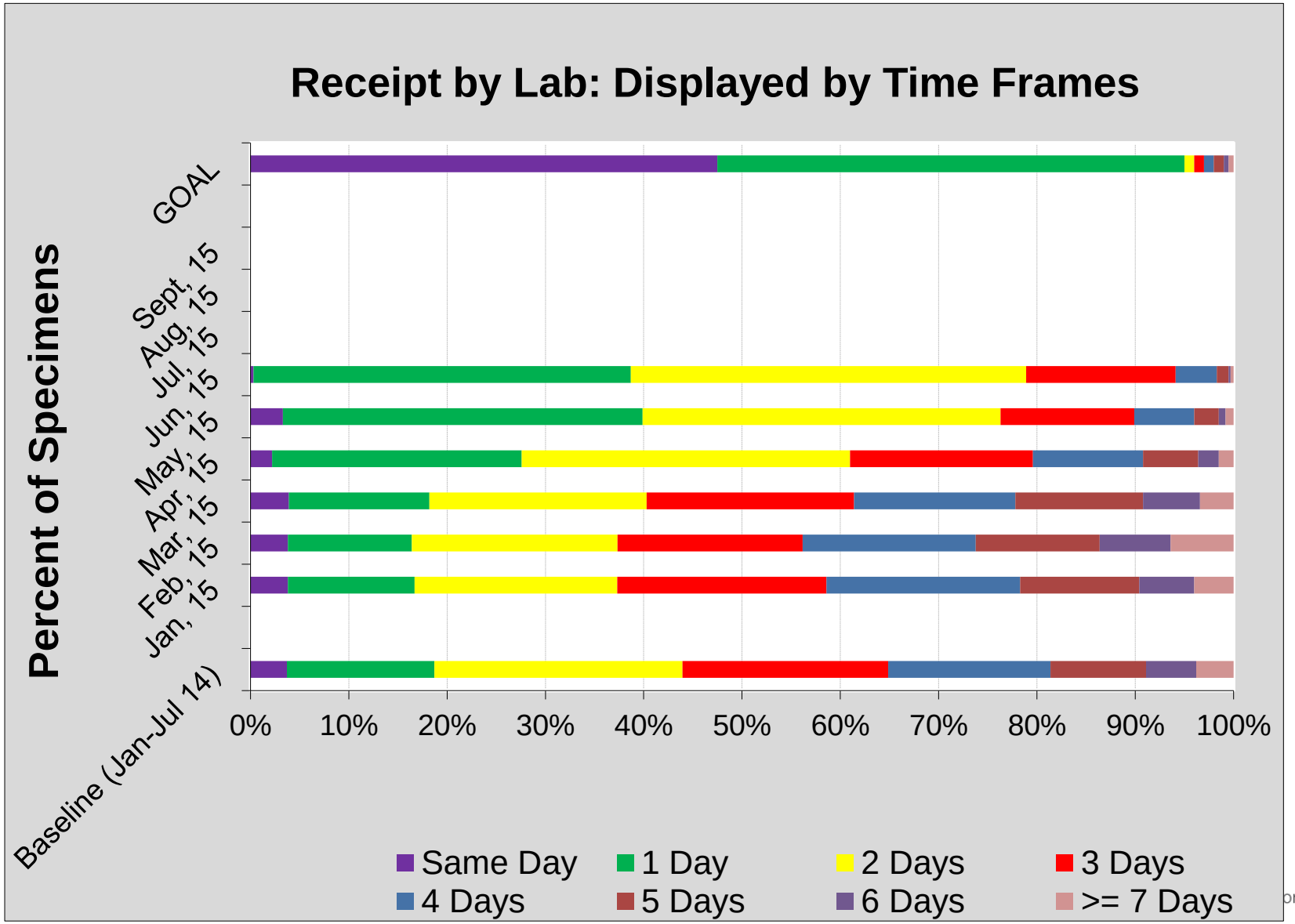
Did you know...

> *That the following real-time queries are now available on the NewSTEPs website to answer your questions about newborn screening programs?*

- **Screened Conditions Report** - the status of NBS conditions that are screened in each state.
- **Conditions By Query Report** - details (e.g., equipment used) on the screened NBS conditions.
- **NBS Fees Report** - the NBS fees charged by each state.
- **DBS Retention Report** - the dried blood spot (DBS) specimen storage /retention times and storage conditions for each state NBS program.
- **Courier System Report** - the courier system used by each state NBS program.

Tennessee: Tracking improvement in timeliness



Partnerships and Collaborations

- Steering Committee and Workgroups
- Newborn Screening Programs
- Regional Collaboratives
- Federal Partners
- Private Partners
- Vendors

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What have we learned?
Where do we go from here?

- NewSTEPs is partnering with state newborn screening programs to develop solutions for strengthening the NBS System
  - Quality data
  - Technical assistance
  - Bringing people together to share ideas and expertise

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NewSTEPs Team

APHL ASSOCIATION OF PUBLIC HEALTH LABORATORIES

- Jelili Ojodu, MPH
- Sikha Singh, MHS, PMP
- Careema Yusuf, MPH
- Thalia Wood, MPH
- Ruthanne Salsbury
- Guisou Pineyro, MPH

colorado school of public health

- Marci Sontag, PhD
- Yvonne Kellar-Guenther, PhD
- Joshua Miller, MPH

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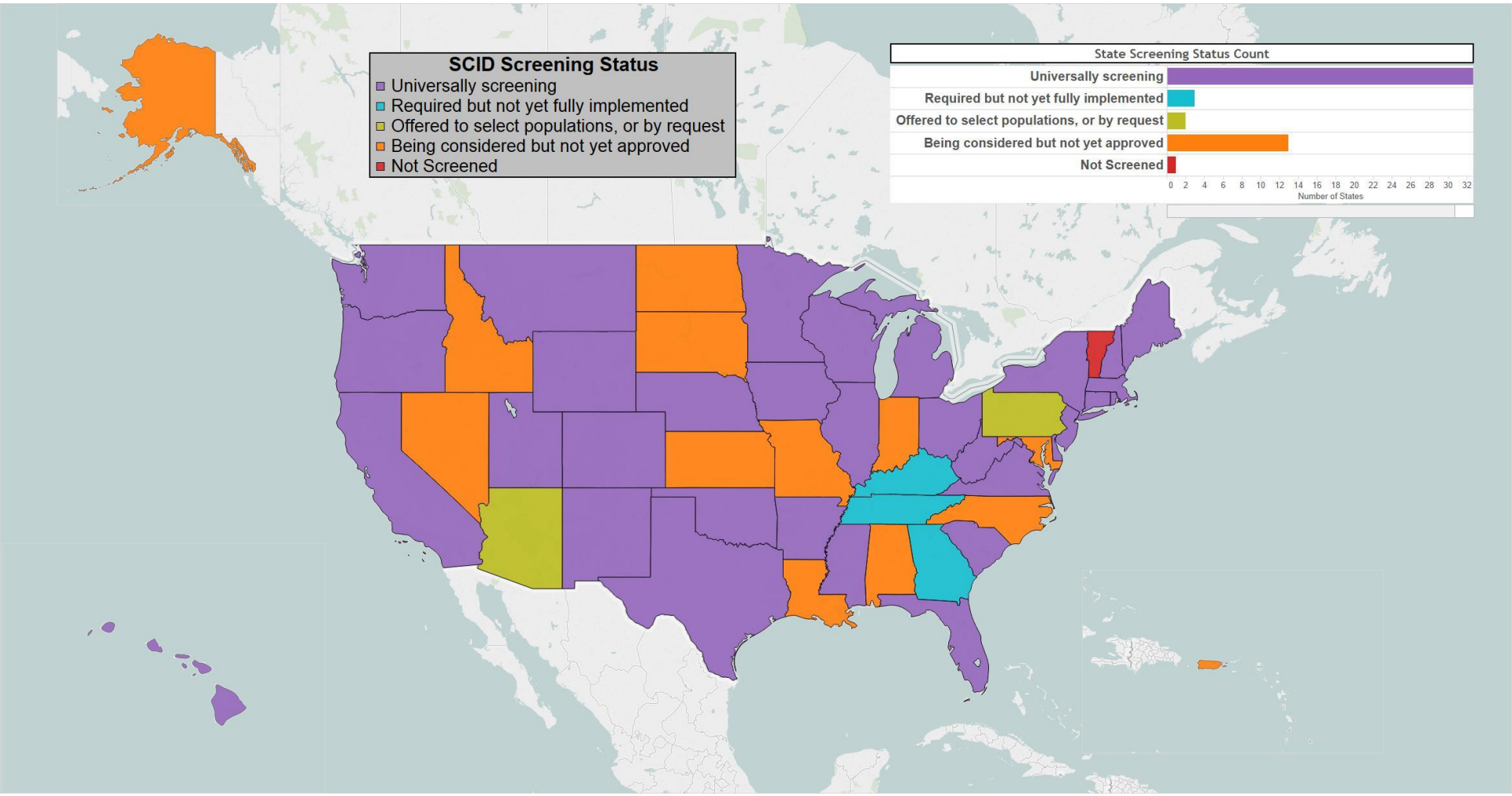
SCID Screening Status



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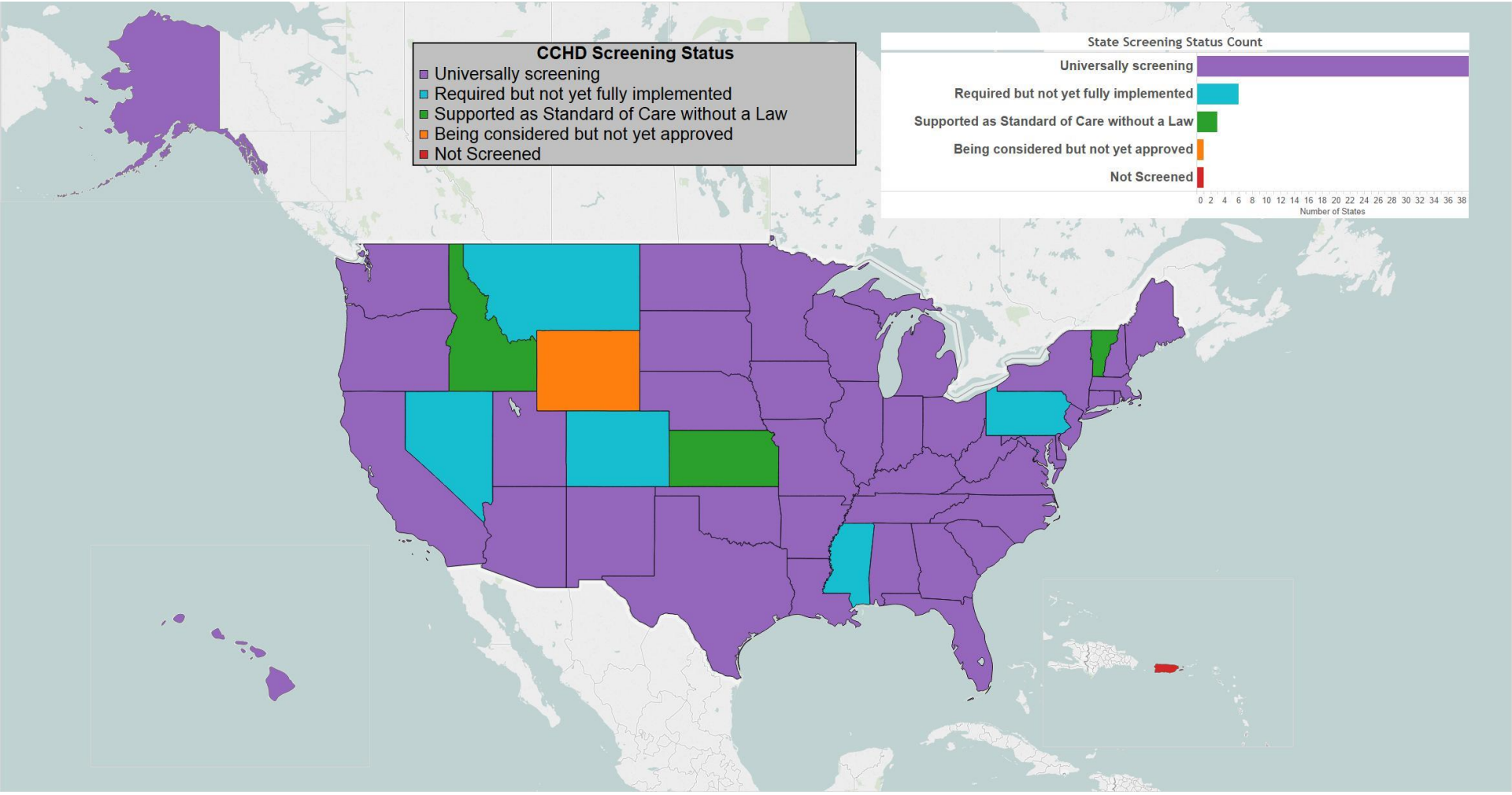
CCHD Screening Status



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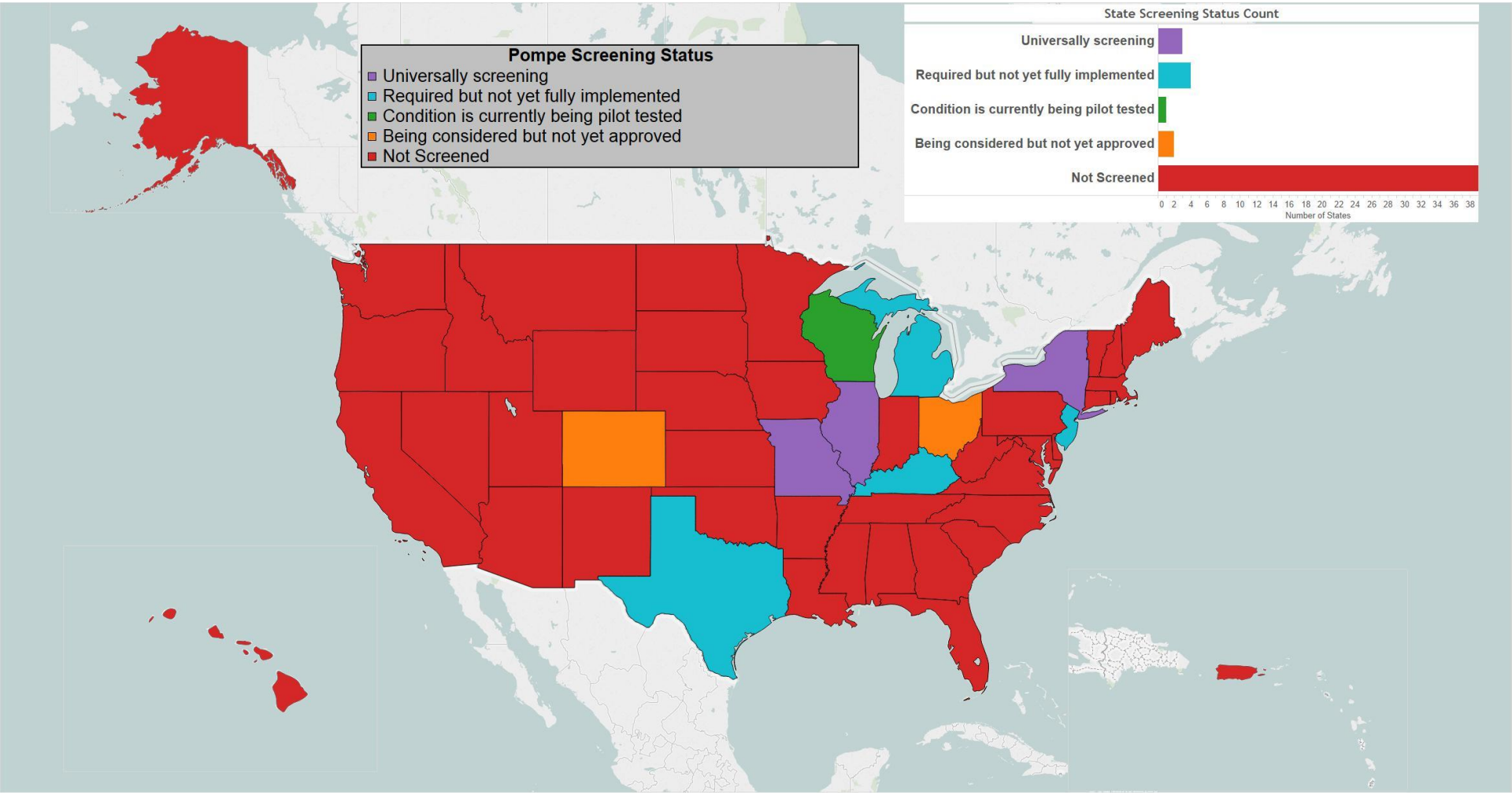
Pompe Screening Status



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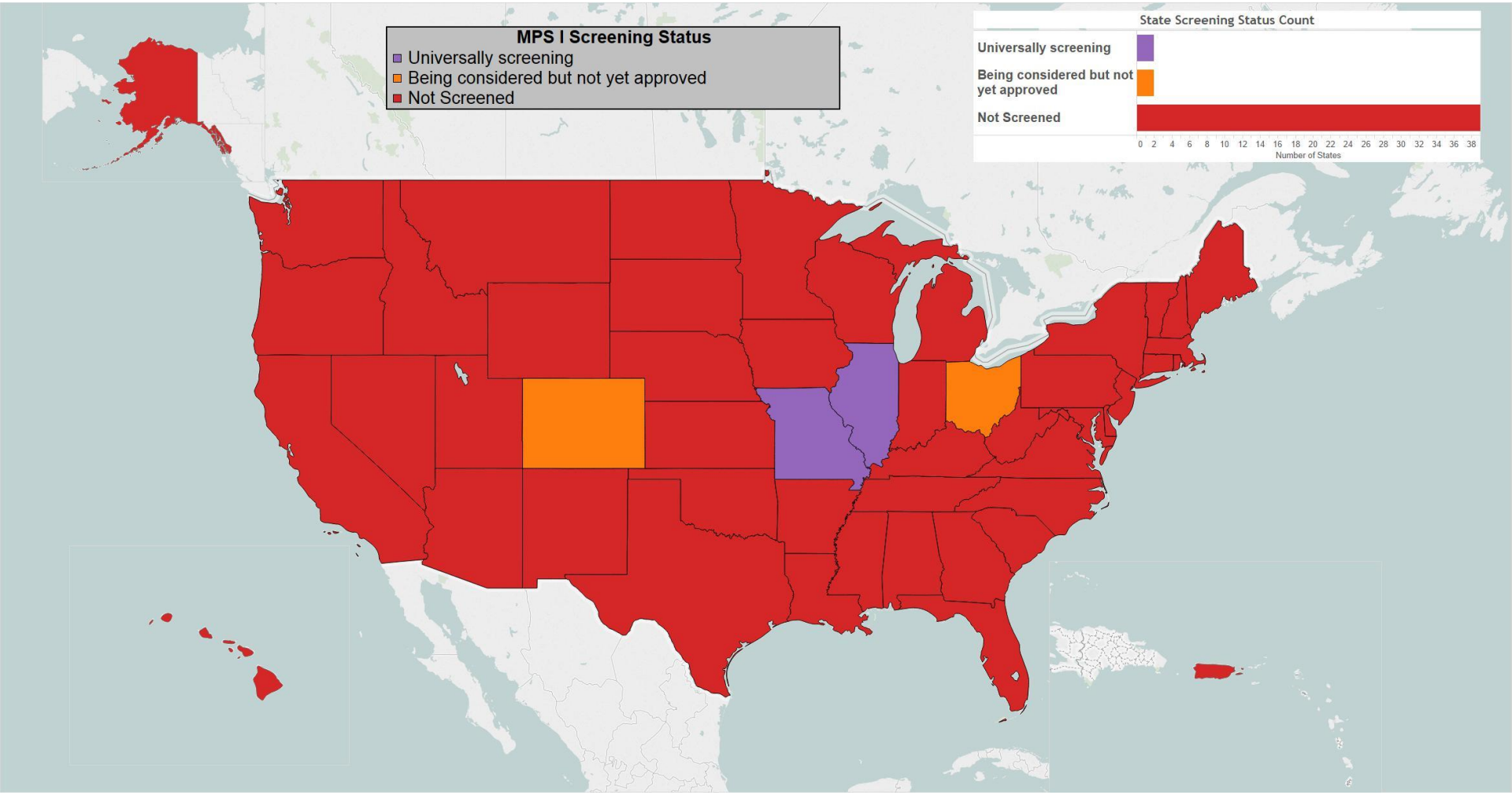
MPS-I Screening Status



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