2024 NewSTEPs Annual Report

NewSTEPs

A Program of the Association of Public Health Laboratories™

July 2025

Contents

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Introduction

The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), a program of the Association of Public Health Laboratories (APHL), presents its 2024 Data Annual Report with the goal of sharing the current landscape of newborn screening (NBS) in the United States (US). NewSTEPs aims to strengthen newborn screening systems by providing data and technical assistance to various partners.

NewSTEPs gratefully acknowledges the contributions of the US NBS programs that submitted data to the NewSTEPs Repository. NewSTEPs also commends our members for all their dedicated efforts to ensure that newborns are screened and treated in a timely fashion.

Please direct any questions regarding this report to newsteps@aphl.org.

Please note that quality indicator and state profile data are represented as of February 28, 2025, and case data is represented as of March 17, 2025. The contents of this report focus on noteworthy changes that occurred within the US NBS system since the publication of the 2022 NewSTEPs Annual Report.

APHL NewSTEPs

Vision

All babies have a healthier start through newborn screening.

Mission

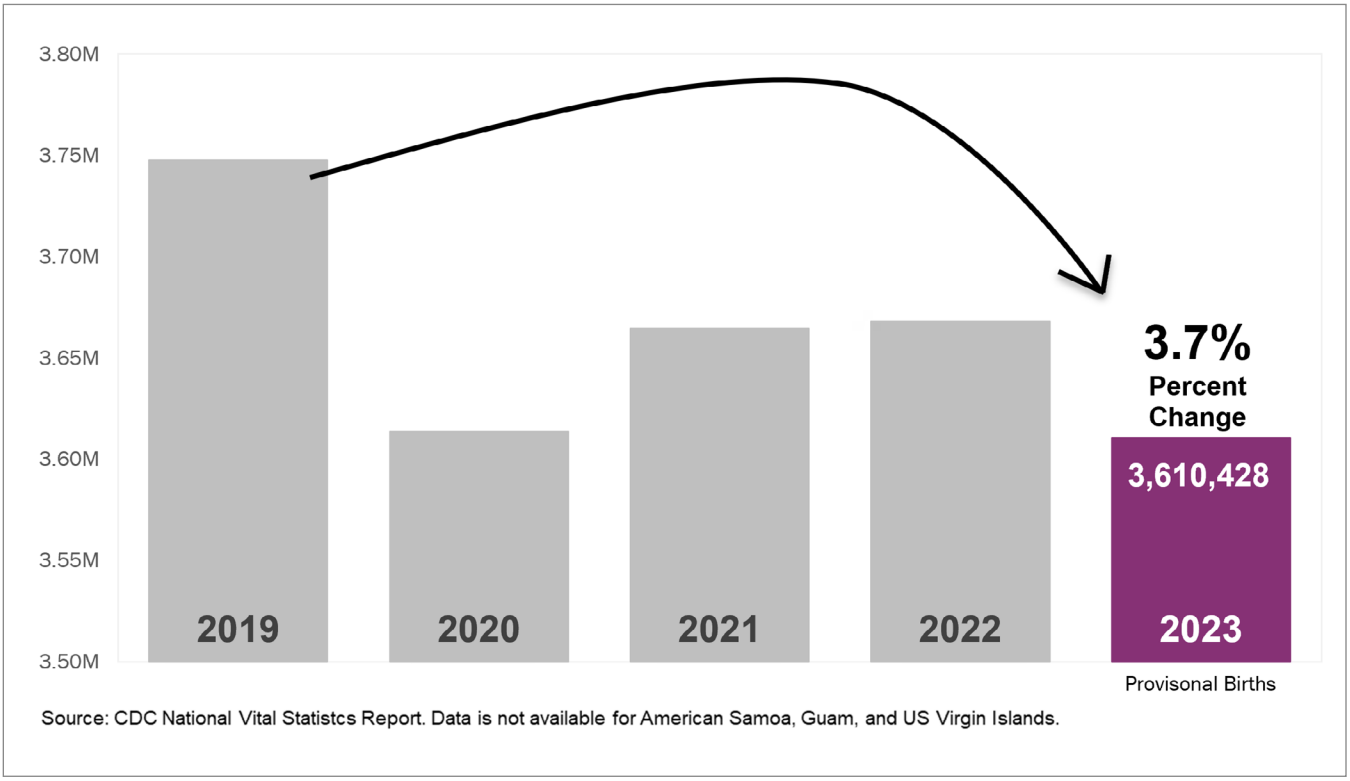
Driving newborn screening systems to excellence by shaping policy, promoting data-driven improvements and pursuing innovations in public health laboratory practices.

Profile Data

NBS Program Overviews

The NewSTEPs Repository represents 56 NBS programs, including all 50 states, American Samoa, the Commonwealth of the Northern Mariana Islands, the District of Columbia (DC), Guam, Puerto Rico and the US Virgin Islands. The 2023 provisional birth data is from the US Centers for Disease Control and Prevention (CDC) Vital Statistics.[a] Overall, the number of births in the US has declined over time, with a 3.7% change from 2019 to 2023 (**Figure 1**).

Figure 1: From 2019–2023, there was a 3.7% decrease in US births (data as of February 28, 2025).



NBS laboratory types vary. Thirty-five programs (62.5%) utilize their state public health laboratory, while 14 programs (25%) use a regional laboratory, and seven programs (12.5%) contract a private laboratory (**Table 1**).

Each state/territory mandates the screening of newborns and specifies if newborns will receive one or two screens. Currently, twelve NBS programs require two screens, recommending that a routine second dried blood spot (DBS) specimen be collected on all newborns regardless of the results of the first newborn screen. The purpose of the routine second screen is to improve the specificity and identify any disorders that were not detectable on the initial screen. The remaining 42 NBS programs require a single newborn screen, but certain circumstances may necessitate additional screens. For example, if the first specimen is collected too early, unsatisfactory, or yields a screen positive, the NBS program may request a subsequent specimen to be collected on the newborn.

a Provisional birth data is not available for American Samoa, Guam and US Virgin Islands.

Table 1: 2024 NBS program overview (data as of February 28, 2025).[b]

State	2023 births *(provisional)*	Responsible Laboratory	Number of Screens	Number of Core RUSP
Alabama	57,803	State Public Health Laboratory	Two Screen	35
Alaska	8,914	Regional Laboratory	One Screen	32
American Samoa	*No data available*	Other Laboratory *American Samoa utilizes the New Zealand Laboratory*	One Screen	*No data available*
Arizona	78,076	State Public Health Laboratory	Two Screen	37
Arkansas	35,213	State Public Health Laboratory	One Screen	35
California	399,368	State Public Health Laboratory	One Screen	37
Colorado[c]	61,475	State Public Health Laboratory	Two Screen	34
Connecticut	34,531	State Public Health Laboratory	One Screen	37
Delaware	10,396	Private Laboratory	One Screen	37
District of Columbia	7,885	Private Laboratory	One Screen	35
Florida	221,365	State Public Health Laboratory	One Screen	37
Georgia	125,046	State Public Health Laboratory	One Screen	36
Guam	*No data available*	Regional Laboratory	One Screen	33
Hawaii	14,643	Regional Laboratory	One Screen	35
Idaho	22,377	Regional Laboratory	Two Screen	35
Illinois	124,743	State Public Health Laboratory	One Screen	37
Indiana	78,856	Private Laboratory	One Screen	36
Iowa[c]	35,994	State Public Health Laboratory	One Screen	35
Kansas	34,056	State Public Health Laboratory	One Screen	36
Kentucky	51,830	State public health laboratory *Kentucky outsources Lysosomal Storage Disorders to Mayo Medical Laboratory*	One Screen	38
Louisiana	54,682	State Public Health Laboratory *Louisiana outsources partial screening to Revvity Omics*	One Screen	34
Maine	11,617	Regional Laboratory	One Screen	35
Maryland	65,561	State Public Health Laboratory	Two Screen	37
Massachusetts[c]	67,113	State Public Health Laboratory	One Screen	35
Michigan	99,055	State Public Health Laboratory	One Screen	36
Minnesota	61,671	State Public Health Laboratory	One Screen	37
Mississippi	34,449	Private Laboratory	One Screen	34
Missouri	67,058	State Public Health Laboratory	One Screen	37
Montana	11,069	State Public Health Laboratory *Montana outsources MS/MS to the Wisconsin State Laboratory of Hygiene*	One Screen	33
Nebraska	24,043	Private Laboratory	One Screen	35
Nevada	31,759	State Public Health Laboratory	Two Screen	33
New Hampshire	11,929	Regional laboratory	One Screen	35
New Jersey	100,943	State Public Health Laboratory	One Screen	36

b Source: Hamilton, B., Martin, J., & Osterman, M. (2024). Births: provisional data for 2023. Vital Statistics Rapid Release, no 35. doi.org/10.15620/cdc/151797

c Regional Laboratories

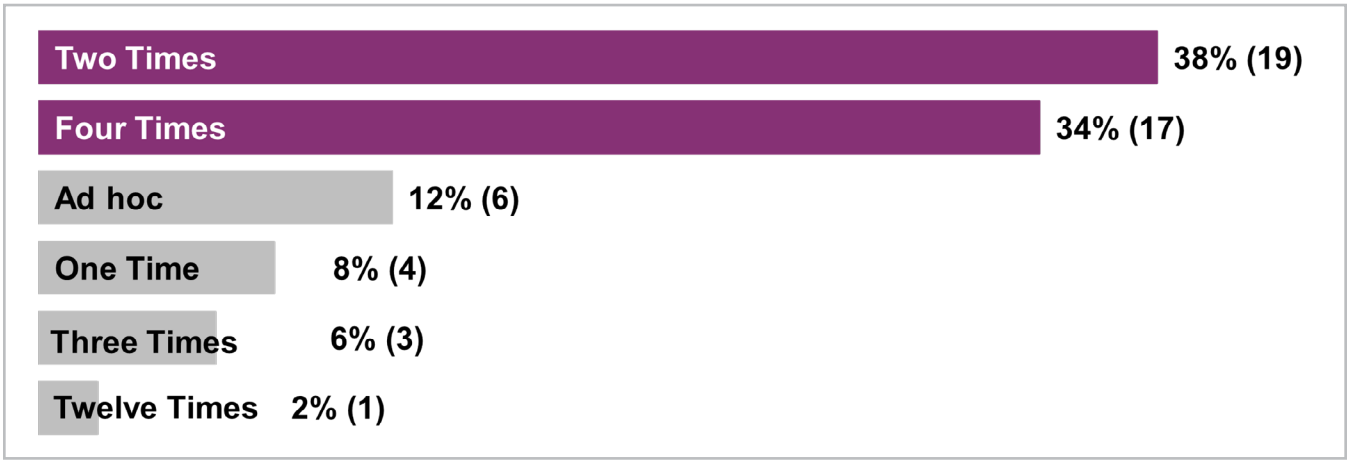
State	2023 births *(provisional)*	Responsible Laboratory	Number of Screens	Number of Core RUSP
New Mexico	20,815	Regional Laboratory	Two Screen	35
New York	203,126	State Public Health Laboratory	One Screen	37
North Carolina	119,744	State Public Health Laboratory	One Screen	35
North Dakota	9,614	Regional Laboratory	One Screen	34
Northern Marianas Islands	571	Regional Laboratory	*No data available*	*No data available*
Ohio	126,785	State Public Health Laboratory	One Screen	36
Oklahoma	47,872	State Public Health Laboratory	One Screen	35
Oregon[c]	38,225	State Public Health Laboratory	Two Screen	35
Pennsylvania	126,757	Private Laboratory	One Screen	38
Puerto Rico	18,529	State Public Health Laboratory	One Screen	31
Rhode Island	9,801	Regional Laboratory	One Screen	35
South Carolina	57,688	State Public Health Laboratory	One Screen	36
South Dakota	11,198	Regional Laboratory	One Screen	34
Tennessee	82,973	State Public Health Laboratory	One Screen	36
Texas	387,636	State Public Health Laboratory	Two Screen	33
US Virgin Islands	*No data available*	Private Laboratory	*No data available*	*No data available*
Utah	45,016	State Public Health Laboratory	Two Screen	36
Vermont	5,058	Regional Laboratory	One Screen	35
Virginia	92,512	State Public Health Laboratory	One Screen	35
Washington[c]	80,879	State Public Health Laboratory	Two Screen	35
West Virginia	16,403	State Public Health Laboratory	One Screen	36
Wisconsin	59,719	State Public Health Laboratory	One Screen	34
Wyoming	5,987	Regional Laboratory	Two Screen	35

Advisory Committees

State and territorial NBS advisory committees serve to evaluate and facilitate the addition of disorders to the screening panel and make recommendations regarding the structure of programs, such as testing algorithms, policies and standards. Their role is to help ensure that NBS programs effectively and efficiently screen, diagnose and treat newborns. Advisory committees frequently include varied representation from families, physicians, laboratory and follow-up staff, and other partners.

Of the 53 NBS programs reporting, 94% (n=50) have an NBS advisory committee in their state/territory. Of these 50 program advisory committees, 28% (n=14) are mandatory and 72% (n=36) are voluntary. NBS advisory committee meeting frequency varies throughout the year depending on the program (**Figure 2**). See the NBS Advisory Committee Dashboard for the program-level breakdown.

Figure 2: Most NBS Advisory Committees meet two or four times per year (n=50; data as of February 28, 2025).



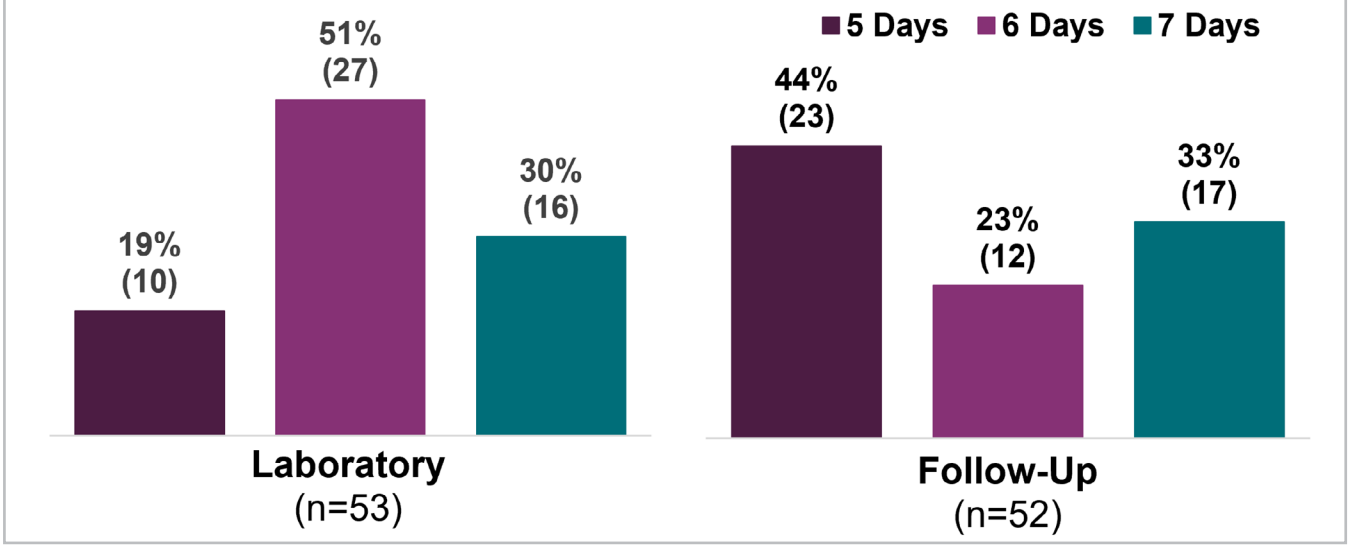
NBS Program Operating Hours

Many NBS programs have gradually expanded the number of operational days per week, performing testing and follow-up activities on weekends and holidays. These activities include but are not limited to: receiving specimens and data entry of patient demographics; performing specimen testing, including molecular and repeat testing; and notifying providers of the NBS results.

In 2015, seven NBS laboratories and 10 follow-up programs operated seven days a week. By 2024, 16 (30%) laboratories and 17 (33%) follow-up programs operated seven days a week (**Figure 3**). See the Operating Days and Hours Dashboard for the program-level breakdown.

NBS programs select which programmatic tasks are performed on weekends and holidays. Seventy-eight percent (n=39) of laboratories report out time-critical results on weekends, and 58% (n=29) of laboratories report out time-critical results on holidays. Seventy-two percent (n=37) of programs notify providers of time-critical results on weekends, and 58% (n=30) notify providers of time-critical results on holidays.[d]

Figure 3: Number of days per week NBS laboratory (n=53) and follow-up (n=52) programs operated in 2024 (data as of February 28, 2025).



d NewSTEPs does not collect data on operating hours by specific holiday.

NBS Program Fees

Fees for an initial newborn screen varies across the US, ranging from $0–259 (n=52). Twenty NBS programs charge between $101–$150 (n=20) for their initial NBS fee, four programs do not impose any fee, and seven programs charge $200 or more (**Figure 4**).

NBS programs also charge differently for the initial and subsequent screens. Thirty-seven programs only charge one fee—the initial screening fee includes the requested subsequent screen. Nine programs provide the fee for the requested subsequent screen, which ranges from $68.63–$235 (**Table 2**). See the NBS Fee Report for the program-level breakdown.

Figure 4: The majority of NBS programs charge between $101-$150 for the initial NBS fee (n=52; data as of February 28, 2025).

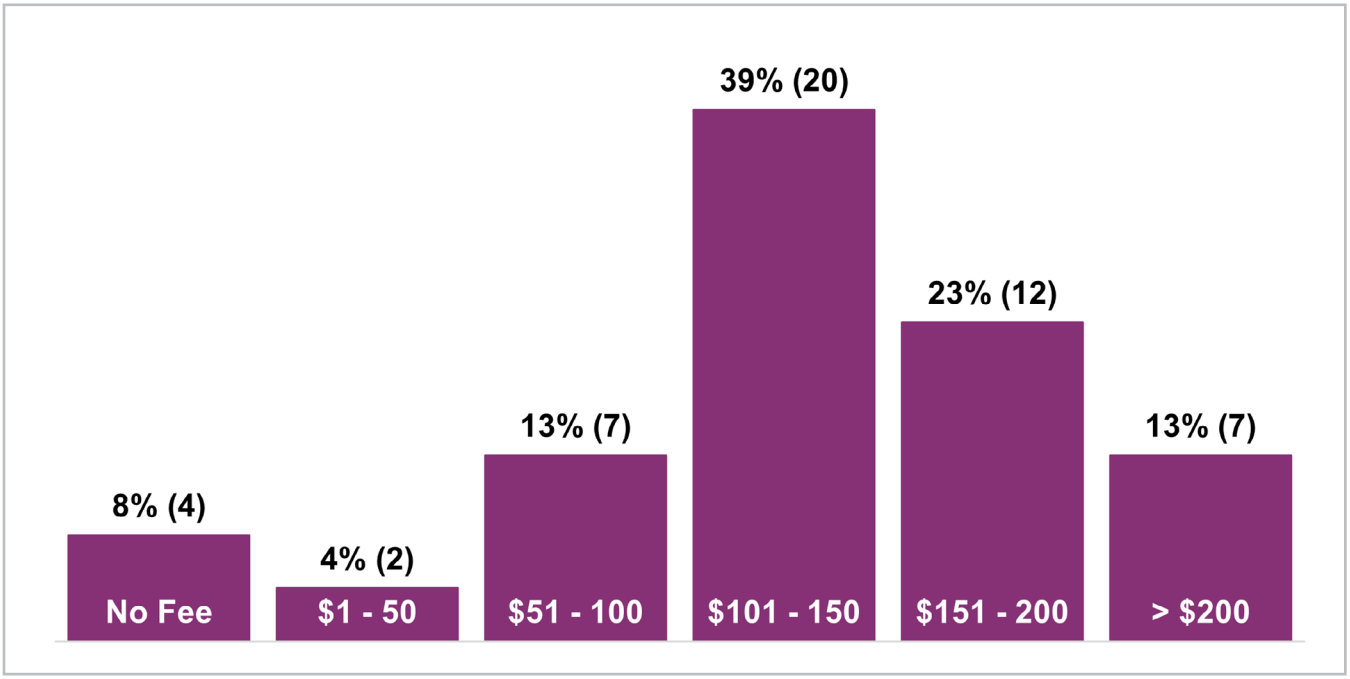


Table 2: NBS Fees for Initial and Requested Subsequent Screen in 2024 (data as of February 28, 2025)

Screen	Median	Minimum	Maximum
Initial Screen *n=52*	$133.52	$0	$258.50
Requested Subsequent Screen *n=9*	$128	$68.63	$235

Information Management Systems

Each NBS laboratory and follow-up program has an information management system (IMS) that is vital for data storage, organization and management. NBS laboratory IMS (or LIMS) vendors include: Revvity (n=24), Neometrics/Natus (n=13), internally developed (n=3), StarLIMS (n=3) and other (n=12). Similarly, follow-up systems are: Revvity (n=17), Neometrics/Natus (n=12), internally developed (n=13), StarLIMS (n=2) and other (n=10) (**Table 3**). See the NBS Information Management Systems Dashboard for the program-level breakdown.

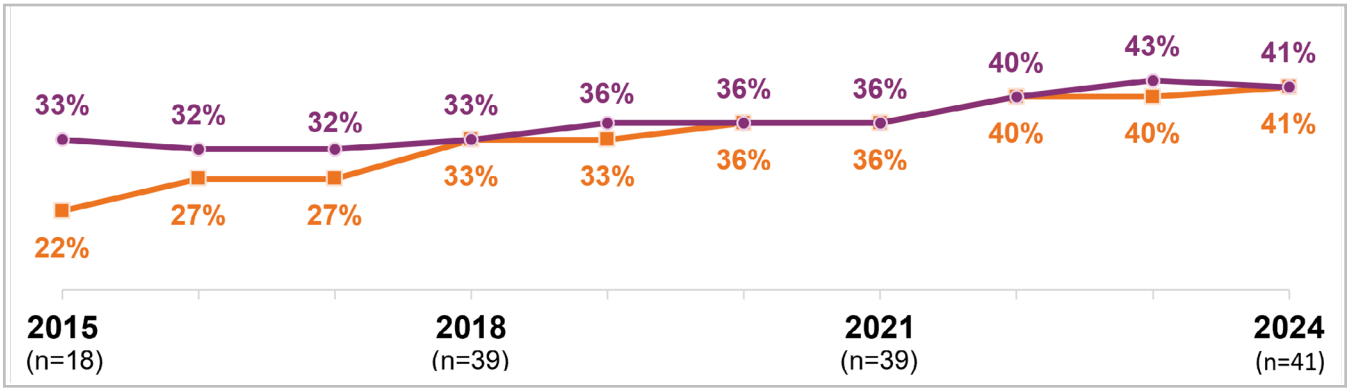
Table 3: NBS laboratory and follow-up IMS in 2024 (data as of February 28, 2025).

LIMS Vendor	Laboratory (n=56)	Follow-up (n=54)
Revvity	43% (n=24)	32% (n=17)
Neometrics/Natus	25% (n=14)	22% (n=12)
Internally Developed	5% (n=3)	24% (n=13)
StarLIMS	5% (n=3)	4% (n=2)
Other Vendors	22% (n=12) *Vendors: Citrix (1), Epic Beaker (1), Horizon (2), NeoMed (1), Omni Lab (1), OpenELIS (4), Orchard Harvest (1), Revvity and Natus (1)*	18% (n=10) *Vendors: Excel (1), Kidsnet (1), Maven (1), NeoMed (1), Omni Lab (1), OpenELIS (4), Welligent Auris (1)*

Health Information Technology

Electronic messaging (e.g., HL7 messaging) in NBS facilitates accurate data sharing and timely reporting of NBS results to healthcare providers. Since 2015, the number of NBS programs that have the capability to accept HL7 order messaging has increased from 22% to 41%. Similarly, the percentage of NBS programs that have implemented HL7 result messaging has increased from 33% to 41% (**Figure 5**).[e] See the NBS Electronic Messaging Dashboard for the program-level breakdown.

Figure 5: The percent of NBS programs that accept and send HL7 result messages has increased since 2015 (data as of February 28, 2025).[e]

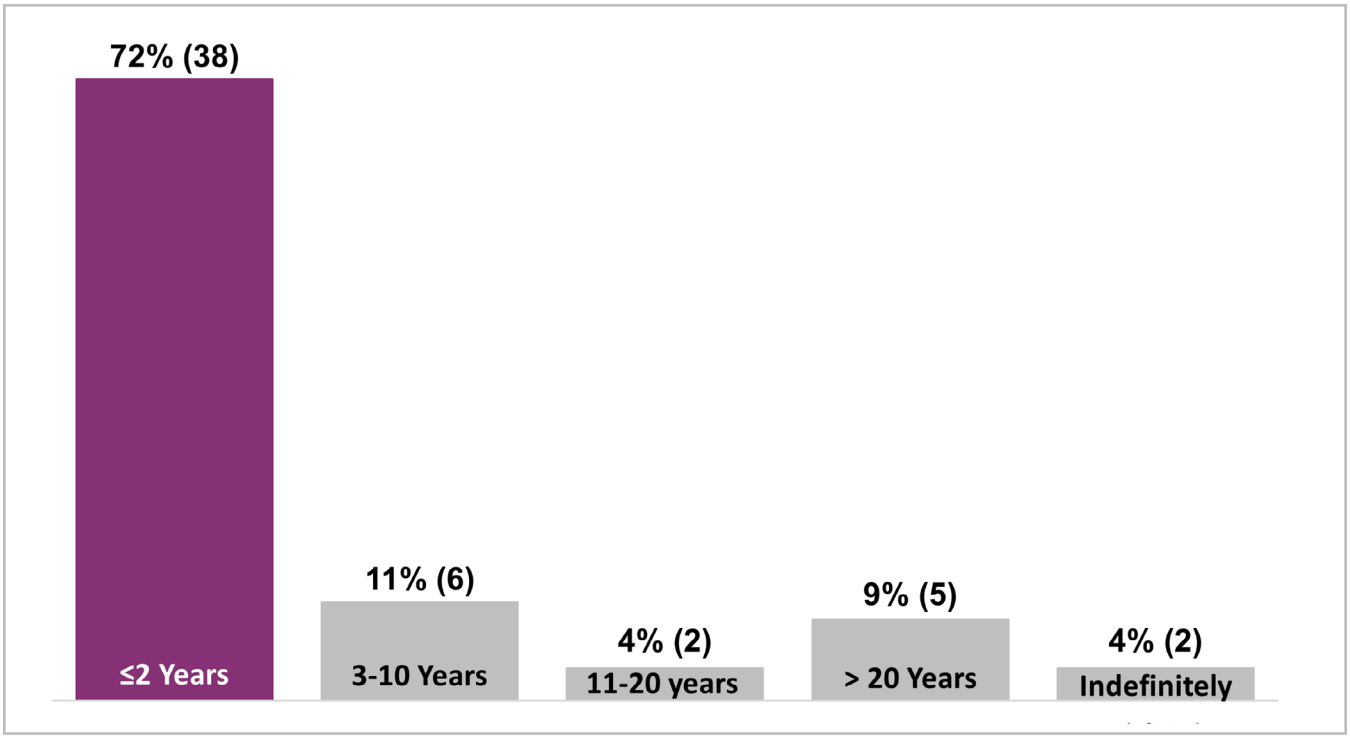


Dried Blood Spot and Data Retention

NBS DBS specimen retention is based on local, state and federal regulations, and programs may retain the physical specimens differently based on the screening result and confirmed diagnosis. NewSTEPs does not currently stratify DBS specimen retention based on the screening result. Fifty-three NBS programs reported DBS specimen retention time: two years or less (72%), three to ten years (11%), 11 to 20 years (4%), greater than 20 years (9%) and indefinitely (4%) (**Figure 6**). See the DBS Retention Report for the program-level breakdown.

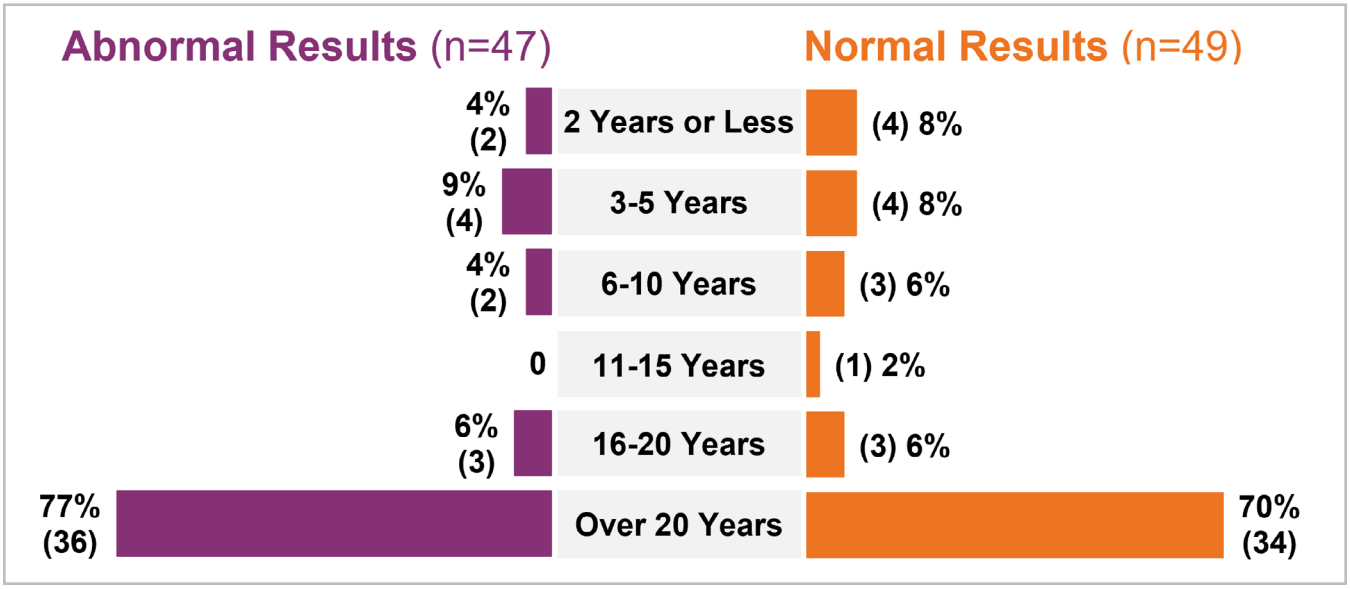
e In 2024, one NBS program accepts HL7 order messages but does not send results using HL7. Additionally, one NBS program sends results using HL7 but does not accept HL7 order message. The remaining 16 NBS programs accept order messages and send result messages using HL7.

Figure 6: In 2024, most NBS programs retain physical DBS specimens for two years or less (n=53) (data as of February 28, 2025).



Similarly, each NBS program maintains their own retention policies for DBS data. Data retention duration may differ based on the screening result and confirmed diagnosis. Forty-seven NBS programs reported the retention of abnormal results: two years or less (4%, n=2), three to five years (9%, n=4), six to 10 years (4%, n=2), 16 to 20 years (6%, n=3) and greater than 20 years (77%, n=36). Furthermore, 49 NBS programs reported on the retention of normal results: two years or less (8%, n=4), three to five years (8%, n=4), six to 10 years (6%, n=3), 11 to 15 years (2%, n=1), 16 to 20 years (6%, n=3) and greater than 20 years (70%, n=34) (**Figure 7**). See the Data Retention Report for the program-level breakdown.

Figure 7: In 2024, the majority of NBS programs retain DBS data for over 20 years for abnormal results (n=47) and normal results (n=49; data as of February 28, 2025).



New Disorders Implementation

Recommended Uniform Screening Panel

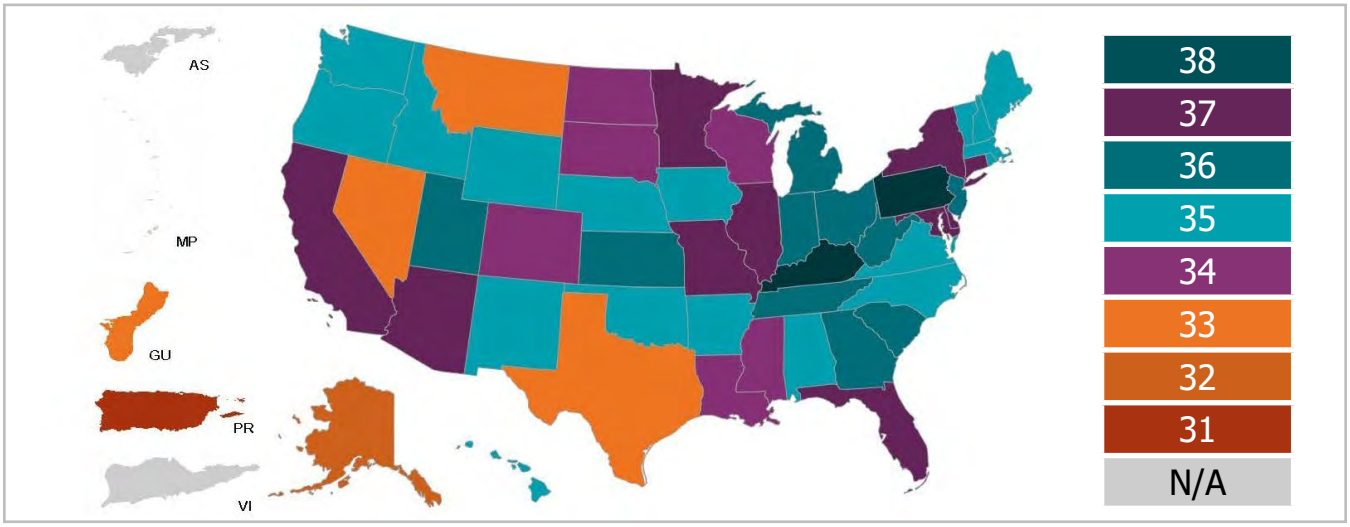
The US states and territories follow individual procedures for which disorders to add to their respective NBS panel, with many being guided by the recommendations made by the US Secretary of Health and Human Services (HHS) for addition to the Recommended Uniform Screening Panel (RUSP), which currently consists of 38 core RUSP disorders (**Table 4**).

All reporting US jurisdictions screen for at least 31 of the 38 core disorders, with some programs screening for an additional subset of secondary disorders. As of February 2025, two states—Kentucky and Pennsylvania—screen for all 38 core disorders on the RUSP (**Figure 8**). See the NBS Status for All Disorders Dashboard to view the program-level breakdown.

Table 4: Core disorders added to the RUSP, by year.

Year	Disorder Added to the RUSP					
2006[f]	3-MCC	Cbl A,B	GA I	HCY	MCAD	PROP
	ASA	CF	GALT	HEAR	MCD	TFP
	BIOT	CH	Hb S/B+Th	HMG	MSUD	TYR I
	BKT	CIT	Hb S/C	IVA	MUT	VLCAD
	CAH	CUD	Hb SS	LCHAD	PKU	
2010	Severe Combined Immunodeficiency (SCID)					
2011	Critical Congenital Heart Disease (CCHD)					
2015	Glycogen Storage Disease Type II (Pompe)					
2016	X-linked Adrenoleukodystrophy (x-ALD); Mucopolysaccharidosis Type I (MPS I)					
2018	Spinal Muscular Atrophy (SMA)					
2022	Mucopolysaccharidosis Type II (MPS II)					
2023	Guanidinoacetate methyltransferase deficiency (GAMT)					
2024	Infantile Krabbe Disease					

Figure 8: US Jurisdictions Screen Between 31 and 38 Core Disorders (data as of February 28, 2025).



f The first 29 disorders added to the RUSP are displayed in their abbreviated form. For a complete list visit NewSTEPs.org.

Implementation Timeline

The initial 29 core RUSP disorders involved laboratory analysis using single-analyte Tandem Mass Spectrometry (MS/MS). In contrast, the disorders added in the past decade have increasingly complex phenotypes, screening methodologies and evolving treatment regimens. The implementation summary (**Table 5**) for the newest RUSP disorders describes when the disorder was added to the RUSP, the percentage of newborns with access to universal screening and the average number of years to implement after the disorder was added to the RUSP.

Each program's implementation of universal NBS screening for Glycogen Storage Disease Type II (Pompe), Mucopolysaccharidosis Type I (MPS I), X-linked Adrenoleukodystrophy (x-ALD) and Spinal Muscular Atrophy (SMA) can be found in the **Appendix (page 26)**, and the status of universal screening by program and year for MPS II, Guanidinoacetate Methyltransferase Deficiency (GAMT) and Infantile Krabbe can be found in **Table 6**.

Table 5: Implementation summary for the newest RUSP disorders (n=53; data as of February 28, 2025)

Conditions	POMPE	MPS I	x-ALD	SMA	MPS II	GAMT	Infantile Krabbe
Year Added to RUSP	2015	2016	2016	2018	2022	2023	2024
Number of States Performing Population Screening	48	45	48	51	12	12	12
Percent of newborns with access to universal screening[g]	87%	84%	97%	99%	35%	38%	34%
Average number of years to implement after addition to the RUSP *For programs that completed implementation by February 2025*	5.0	4.1	4.8	2.2	0.7	.02	-6.3[h]

Table 6: Jurisdictions that have implemented screening for MPS II, GAMT and Infantile Krabbe Disease (data as of February 28, 2025).

Year	MPS II	GAMT	Infantile Krabbe
2006			New York
2012			Missouri
2015		Utah	
2016			Kentucky Ohio
2017	Illinois		Illinois Tennessee
2018		New York	
2019	Missouri		New Jersey
2021			Georgia Indiana Pennsylvania
2022		Michigan	
2023	Pennsylvania West Virginia	Connecticut	South Carolina

g Percent of newborns with access to universal screening for the newly added RUSP disorder is calculated by summing the annual births for the jurisdictions that are universally screening for the disorder divided by all US births. Annual births are pulled from the CDC Vital Statistics 2022 Final Births which includes Guam.

h Programs have implemented Infantile Krabbe Disease before its addition to the core RUSP; therefore, the average number of years to implementation is a negative number.

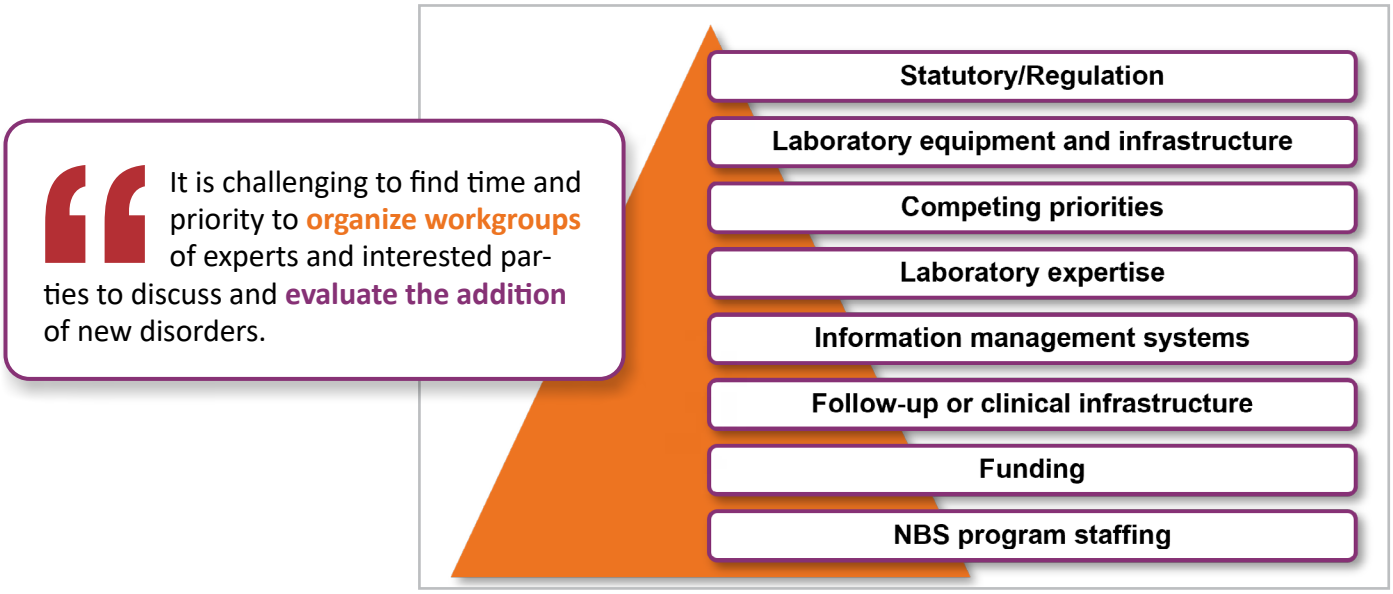
Year	MPS II	GAMT	Infantile Krabbe
2024	Arizona California Connecticut Delaware Florida Kansas Kentucky Maryland	California Delaware Kentucky Maryland Pennsylvania	Minnesota
2025		Arizona Florida Minnesota	

Implementation Challenges

The 2024 NewSTEPs Needs Assessment asked NBS laboratory and follow-up programs (n=80) to identify their top challenges when implementing a new disorder in the last two years. Respondents indicated staffing as the most significant challenge in onboarding new disorders (55%, n=44), followed by funding (39%, n=31). Additionally, 29% (n=23) mentioned the need for follow-up or clinical infrastructure , 24% (n=19) pointed to information management system (IMS) issues, 21% (n=17) highlighted laboratory expertise and 21% (n=17) noted competing priorities (**Figure 9**).

One major concern of several NBS laboratories is the lack of physical space for the new equipment and infrastructure needed to expand their screening panel. Programs often face challenges when executing contracts necessary for purchasing equipment, supplies or outsourcing testing. Many programs find it difficult to obtain quality assurance materials and samples from true positives to perform validation testing. Additionally, US Food and Drug Administration (FDA)-cleared testing kits are often not available for all disorders; as a result, programs are often required to develop their own laboratory-developed tests (LDTs).

Figure 9: New disorder implementation challenges.



NBS Performance Metrics

There are eight quality indicators that serve as standardized performance metrics and are used to provide longitudinal evaluation of NBS programs (**Table 7**). The quality indicators support data-driven assessments and help inform national and programmatic quality improvement initiatives. Please refer to the Quality Indicator Source Document for complete definitions and recent changes made to the metrics.

NBS programs report quality indicator data each spring for the previous calendar year. Due to this delay, this report highlights 2023 quality indicator data, as NBS programs have not yet reported 2024 data at the time of publication. Further, due to ongoing validation, this report may not reflect previously-reported data and is a snapshot of data provided as of February 28, 2025. Dashboards can be viewed in real-time by visiting the NewSTEPs Dashboards and Reports; permission to access these dashboards is role-based.

Table 7: NewSTEPs Quality Indicators (QI)

QI	Definition
QI 1	**Unsatisfactory Specimens:** Percent of DBS specimens that were unacceptable due to improper collection and/or transport
QI 2	**Missing Essential Information:** Percent of DBS specimens with at least one missing state-defined essential data field upon receipt at the laboratory
QI 3	**Unscreened Newborns:** Percent of newborns not receiving a newborn screen
QI 4	**Lost to Follow-up:** Percent of infants that have no recorded final resolution with the NBS program
QI 5	**Timeliness of NBS activities**
QI 6	**Screen Positives:** Percent of infants with an out-of-range newborn screen result requiring clinical diagnostic workup[i]
QI 7	**Confirmed Cases:** Birth prevalence of disorders detected by NBS with a confirmed diagnosis by an appropriate medical professional (see **Number of Cases Identified Through NBS on page 25**)
QI 8	**Missed Cases:** Number of infants that have a confirmed diagnosis by a physician but did not have an out-of-range newborn screen[j]

QI 1 & 2 | Unsatisfactory Specimens and Missing Essential Information

DBS specimens received at NBS laboratories are inspected for specimen quality and missing state-defined essential information. Specimens may be unacceptable for testing due to collection or transportation errors, resulting in the need for additional specimen collection. Missing essential information impacts result interpretation and/or impedes the ability to identify and locate the infant in an emergent situation, causing delays and potential harm to the newborn. Both issues delay NBS testing and reporting.

Starting in 2023, NewSTEPs stratified first specimens and requested subsequent (including routine second) specimens as some NBS programs reported more unsatisfactory specimens and specimens missing essential information on the second or subsequent screen, biasing the data. Due to this metric change, we cannot compare 2023 data to previous years.

In 2023, the median percent of unsatisfactory first specimens is 1.76% (n=34) and 3.85% (n=21) for requested subsequent and routine second specimens. The majority of unsatisfactory specimens are a result of collection issues rather than transportation issues (**Table 8**). In 2023, 17 NBS programs fell below the national median for unsatisfactory first specimens (**Figure 10**). NBS DBS specimens may be deemed unsatisfactory upon receipt at the NBS laboratory for a variety of reasons (e.g., insufficient quantity of blood, smearing, poor saturation, oversaturation, layering blood drops, contamination, etc.). Additionally, NBS programs differ in how they characterize which specimens are deemed unsatisfactory for screening.

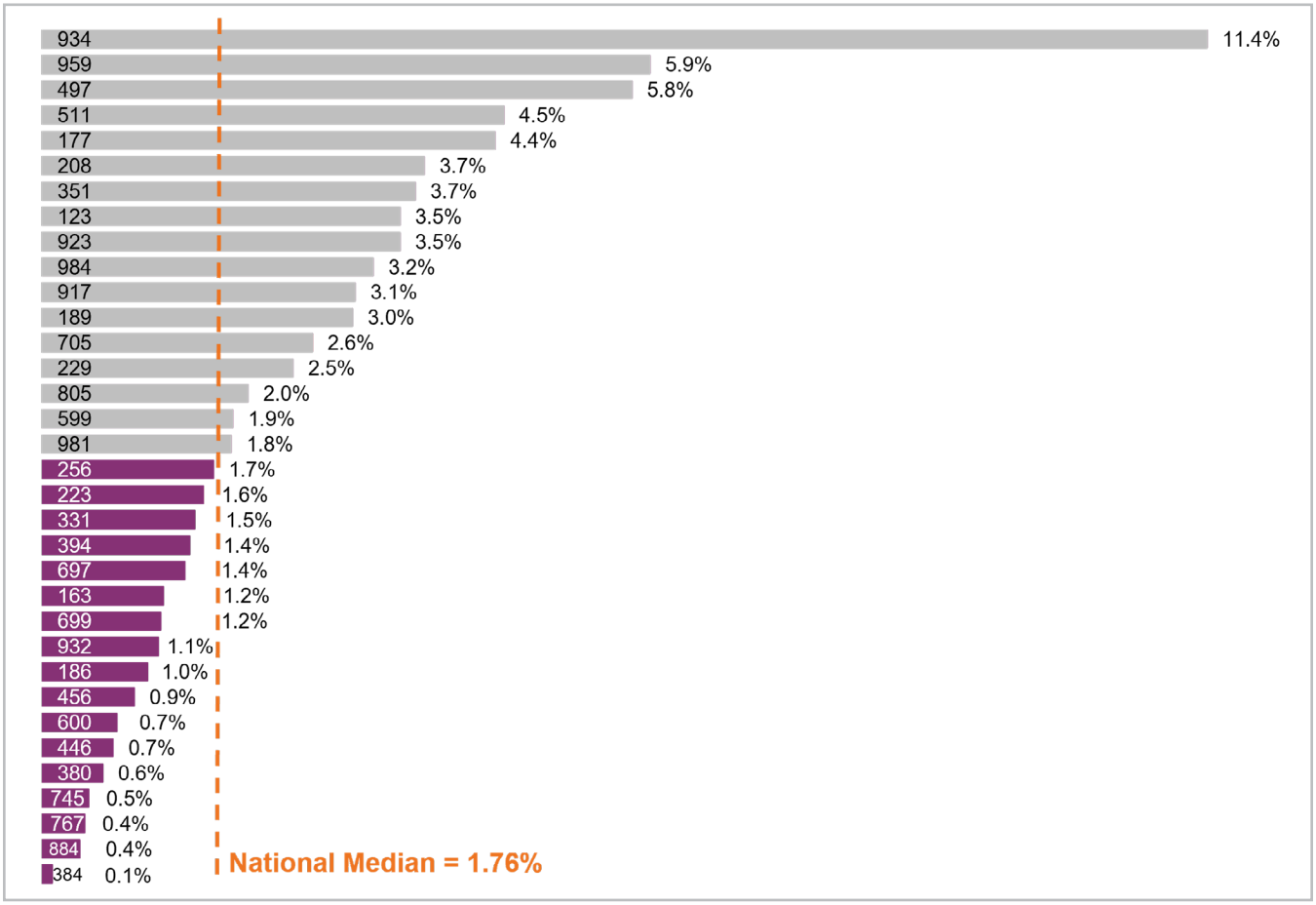
i Data is incomplete and will not be reported on at this time.

j This is the number of missed cases known by the state/territory. A missed case is only considered to be missed if the state/territory was screening for the disorder at the time of birth. Missed cases are not included in the report due to the data being incomplete, as many programs do not enter individual case-data.

Table 8: Unsatisfactory specimens by first and requested subsequent/ routine second screen specimens in 2023 (data as of February 28, 2025).[k, l]

Specimen Type	Category	Number of Programs Reporting Data	Percent of Unsatisfactory Specimens		
			Median	Min	Max
First Specimens	Collection	34	1.69%	0.08%	11.34%
	Transport	25	0.05%	0.00%	0.95%
	Total	**34**	**1.76%**	**0.10%**	**11.40%**
Requested Second Specimens	Collection	21	3.63%	0.43%	10.46%
	Transport	19	0.20%	0.00%	1.55%
	Total	**21**	**3.85%**	**0.97%**	**10.66%**

Figure 10: In 2023, 17 NBS programs were below the national median for unsatisfactory first specimens (data as of February 28, 2025).[m]



k Percent of unsatisfactory specimens and missing information reported before 2023 combined first, requested subsequent, and routine second specimens. Therefore, we cannot compare 2023 data to previous years, as the data has been stratified by first and requested subsequent specimens.

l If it is unknown whether unacceptable specimens were due to improper collection or transport, programs are instructed to only count specimens under improper collection. Limitations in LIMS queries may prevent programs from collecting or separating unacceptable specimens due to improper transport.

m Programs represented in the bar charts have been randomly assigned numbers to de-identified programs that have submitted data.

Each NBS program defines their own essential information data elements that are critical for testing and follow-up activities. Essential information examples include but are not limited to: patient identification number, infant's first and last name, mother's first and last name, newborn's weight, date and time of birth, date and time of specimen collection, and physician's name and phone.

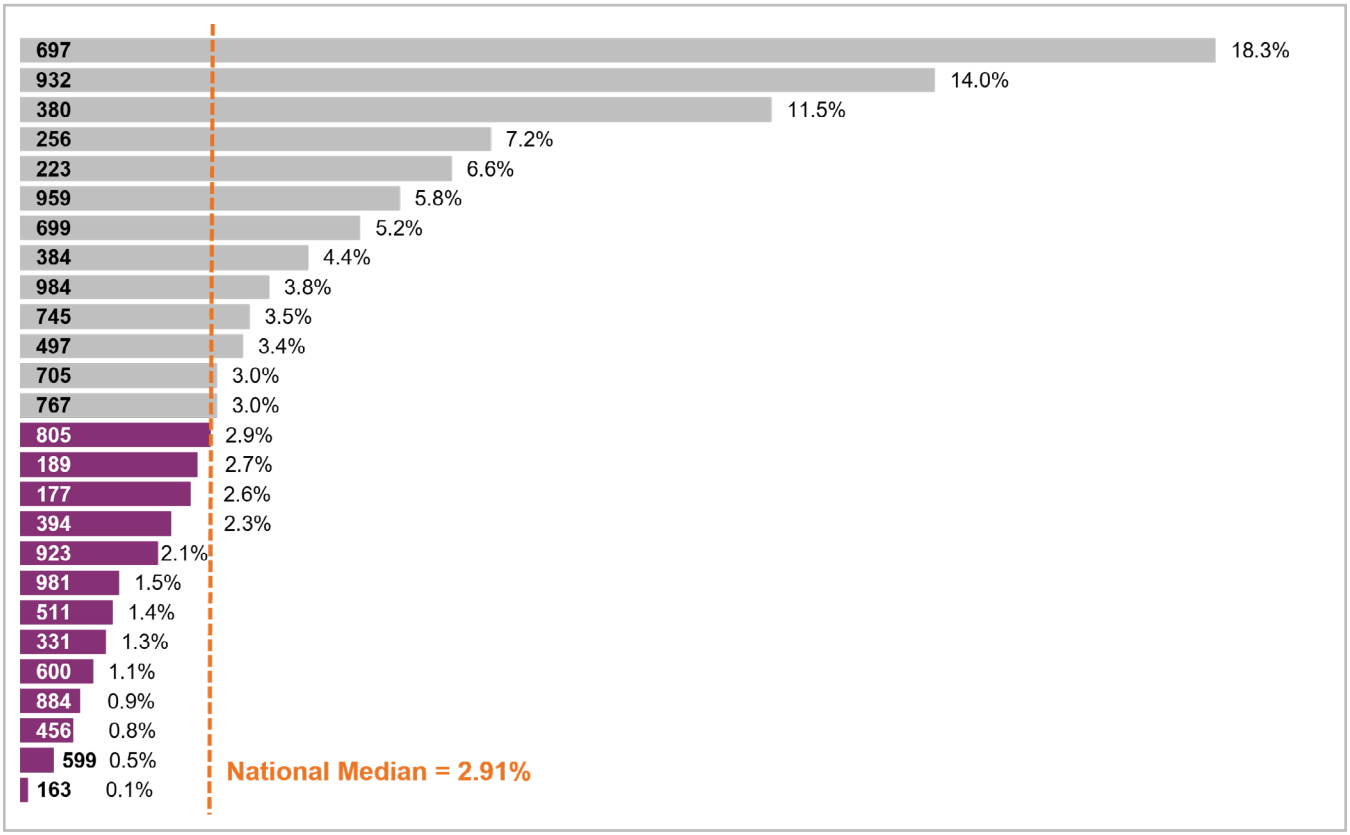
In 2023, the median percent of missing essential information for first specimens was 2.91% (n=26) and 5.42% (n=15) for requested subsequent and routine second specimens (**Table 9**). Thirteen NBS programs fell below the national median for this metric—reflecting strong performance in completion of demographic entry (**Figure 11**).

It is important to track unsatisfactory specimens and specimens missing essential information to determine if additional education and training are required to limit specimen quality issues and, therefore, limit delays in the screening process. By stratifying the first and requested subsequent specimens, NBS programs can determine where focused education and training may be needed.

Table 9: Missing essential information for first and requested subsequent/routine second screen specimens in 2023 (data as of February 28, 2025).[n]

Specimen Type	Number of Programs Reporting Data	Percent of Specimens Missing Essential Information		
		Median	Min	Max
First Specimens	26	2.91%	0.14%	18.27%
Requested Subsequent Specimens	15	5.42%	0.86%	25.72%

Figure 11: In 2023, 13 NBS programs were below the national median for missing essential information (data as of February 28, 2025)[o]



n Percent of specimens missing essential information reported before 2023 combined first, requested subsequent, and routine second specimens. Therefore, we cannot compare 2023 data to previous years, as the data has been stratified by first and requested subsequent specimens.

o Programs represented in the bar charts have been randomly assigned numbers to de-identified programs that have submitted data.

QI 3 | Unscreened Newborns

A key to ensuring access across the NBS system is to support screening for all newborns . NewSTEPs collects data on the proportion of total proportion of unscreened newborns, due to parental refusals, pre-analytic errors and missing/unmatched screens for two-screen states. The denominator is the annual births, which is pulled from the CDC Vital Statistics.

Of the NBS programs that can report this information,[p] the median percent of newborns that did not receive a valid DBS newborn screen in 2023 is 0.44%, with a maximum of 1.77% (**Table 10**). The number of unscreened newborns reported since 2018 is shared in **Table 11**.

A common limitation for state and territory public health departments determining if all newborns in their jurisdictions have received a screen is the lack of uniform linkage to vital records or birth certificate data. Infant-level matching with vital records or birth certificate data is the only way to compare the number of infants born to the number of infants screened in a jurisdiction. However, programs may be able to report on the number of parental refusals when documentation is sent to the program. Additionally, programs may experience increased challenges in tracking unscreened babies born outside of birthing facilities (i.e., home births).

Table 10: Newborns not receiving a valid DBS screen in 2023 (data as of February 28, 2025).[p]

Missed Screen Type	Number of Programs Reporting Data	Percent of Unscreened Newborns		
		Median	Min	Max
Newborns without a valid DBS screen[q]	20	0.44%	0.05%	1.77%
Newborns without a valid DBS screen due to parental refusal	16	0.09%	0.00%	0.68%
Newborns without a valid DBS screen due to pre-analytical errors (QI 3c)[r]	11	0.03%	0.00%	0.14%

Table 11: The median percent of unscreened newborns since 2018 (data as of February 28, 2025).

Birth Year	Number of Programs Reporting Data	Percent of Unscreened Newborns		
		Median	Min	Max
2023	20	0.44%	0.05%	1.77%
2022	21	0.44%	0.05%	2.30%
2021	22	0.29%	0.06%	3.37%
2020	20	0.31%	0.03%	3.65%
2019	15	0.32%	0.03%	1.99%
2018	13	0.32%	0.02%	1.92%

p NBS programs experience limitations in reporting the total number of newborns that did not receive a valid screen. This is due to the lack of uniform linkage to vital records or birth certificate data. NBS programs are only able to report this metric if there is a system in place to match NBS screening information to vital records or birth certificate data.

q Counts of unscreened newborns due to parental refusal (QI 3b) and pre-analytic error (QI 3c) should be included in the total number of unscreened newborns (QI 3a). QI 3a should be greater than or equal to QI 3b + QI 3c.

r This includes any pre-analytic event—except parental refusal—that would prevent the newborn from receiving a complete screen. For DBS screens, some examples include unacceptable specimens that never had a subsequent specimen received at the laboratory, specimens lost in transit or specimens for which hospital personnel forgot to either collect or ship the specimen.

QI 4 | Lost to Follow-up

Quality practices for NBS programs include tracking the percent of newborns that are lost to follow-up (i.e., have no final resolution with the NBS program by 12 months of age). In 2023, the median percent lost to follow-up following the receipt of an unsatisfactory specimen was 4.25% (n=18). The median percent lost to follow-up following a borderline result was 1.77% (n=17) and the median percent lost to follow-up following an out-of-range result was 2.94% (n=18) (**Table 12**). Of note, programs ranged from no lost to follow-up (0%) to roughly 25-30% of infants without a final resolution with the NBS program, presenting opportunities for improvement.

NBS programs have indicated that this metric is challenging to collect because they may not receive reliable information or have a mechanism to track it. Importantly, having no final resolution within the NBS program does not necessarily equate harm to an infant, but could be a function of other priorities across the surveillance and clinical system where feedback loops for closing out cases in the follow-up systems remain incomplete.

Table 12: Infants lost to follow-up in 2023 (data as of February 28, 2025).[s]

No Final Resolution by 12 Months of Age Following:	Number of Programs Reporting Data	Percent Lost to Follow-up[r]		
		Median	Min	Max
The receipt of an unsatisfactory specimen	18	4.25%	0.00%	29.55%
Borderline result for which a sub-sequent DBS specimen was re-quested for a repeat screening	17	1.77%	0.00%	24.06%
Out-of-range result requiring clinical diagnostic workup	18	2.94%	0.00%	32.18%

QI 5 | Timeliness

Timeliness quality indicators are broken into various metrics to identify the components of the NBS system that can be shortened to decrease the risk of potential harm to infants who may be identified with a NBS disorder. The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommends that 95% of specimens should follow these timeliness goals:

- Collection of first DBS specimens within 48 hours from birth (QI5a.i)
- Receipt of first DBS specimens at the NBS laboratory within one day of collection (QI5b.i)[t]
- Reporting time-critical disorders by five days from birth (QI5d.i)
- Reporting first DBS specimens with normal and out-of-range results for all disorders by seven days from birth (QI5d.iii)

Time from Birth to Collection

The majority of NBS programs submitting data to NewSTEPs collect DBS specimens within the recommended timeframe of 48 hours from birth, with a gradual improvement each year. The median percent of first DBS specimens collected within 48 hours of birth has increased from 94.75% (n=26) in 2018 to 97.56% (n=37) in 2023. In 2023, 28 programs reporting on this metric reached the recommended benchmark, compared to only 17 programs reporting this metric in 2018 (**Table 13**, **Figure 12**).

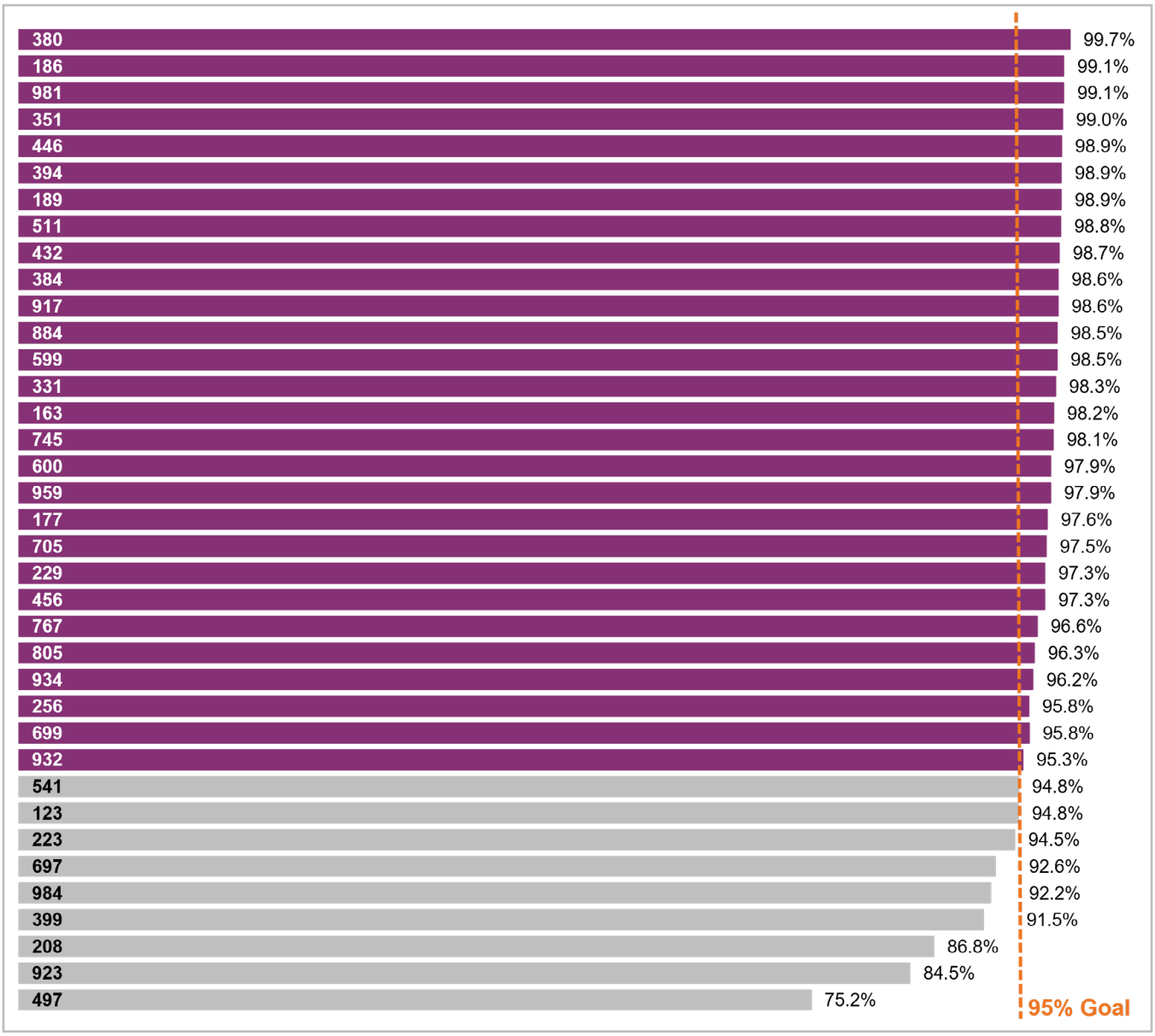
s Lost to follow-up (QI 4) is still pending for 2023 as it is defined as having "no recorded final resolution by 12 months of age." Therefore, in some instances, infants lost to follow-up in 2023 might not be reported until 2025 to account for this 12-month period.

t Although ACHDNC recommends 24 hours for receipt at the NBS laboratory, NewSTEPs uses a two-day benchmark.

Table 13: First DBS specimens collected within 48 hours of birth increased from 2018 to 2023 (data as of February 28, 2025).

Year	Number of Programs Reporting Data	Percent DBS Collected within 48 Hours of Birth			Programs above the 95% Benchmark	
		Median	Min	Max	Number	Percent
2023	37	97.58%	75.16%	99.72%	28	75.68%
2022	39	97.49%	40.01%	99.85%	30	76.92%
2021	39	97.04%	78.54%	99.62%	28	71.18%
2020	38	97.46%	83.31%	99.62%	29	76.32%
2019	33	96.37%	20.12%	99.55%	22	66.67%
2018	36	94.75%	46.10%	99.61%	17	47.22%

Figure 12: In 2023, 28 NBS programs reached the 95% benchmark for first DBS collection within 48 hours of birth (data as of February 28, 2025).



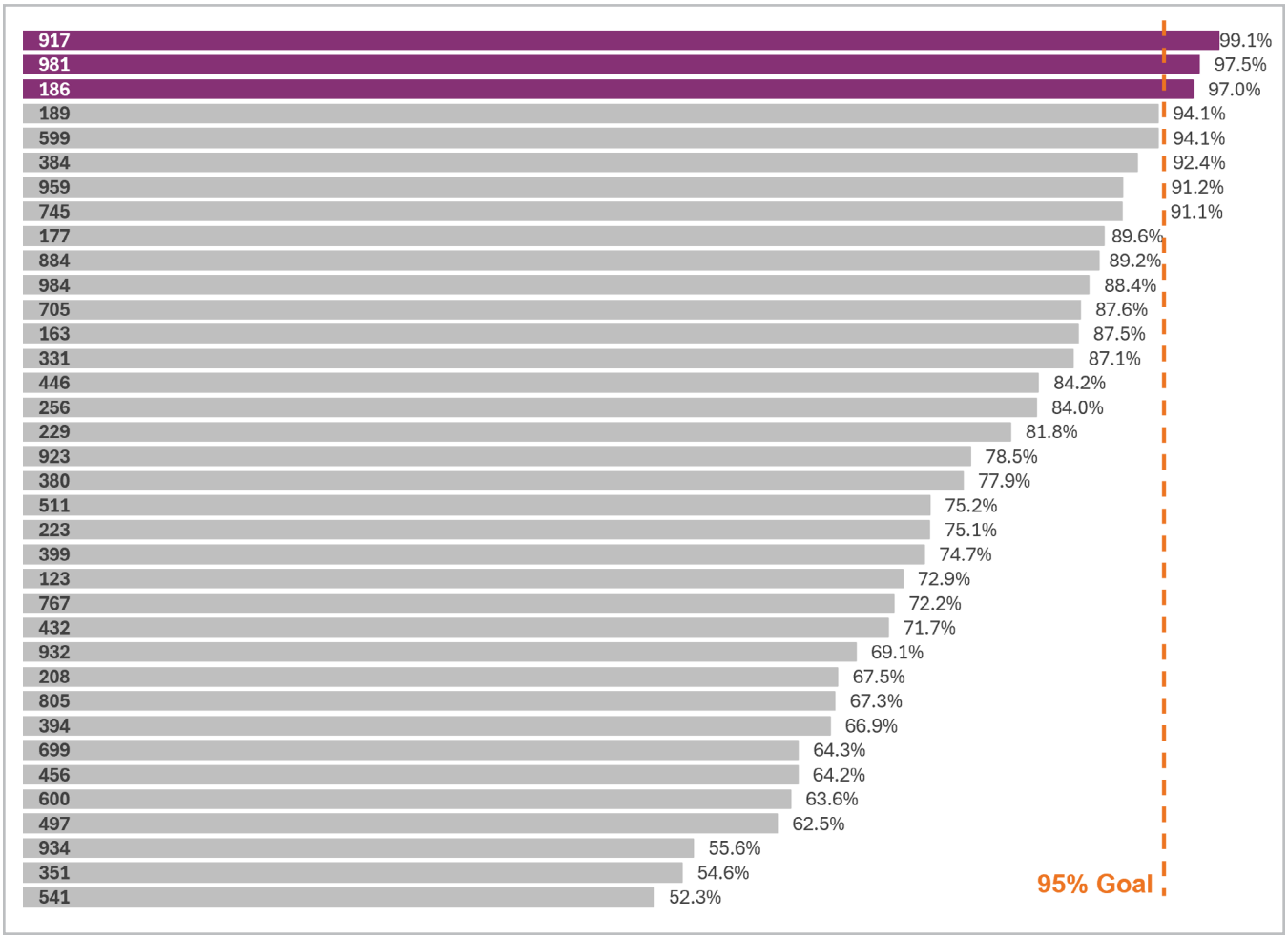
Time from Collection to Receipt

The ACHDNC recommends that 95% of DBS specimens be received at the NBS laboratory as soon as possible–ideally, within 24 hours of collection. NewSTEPs utilizes the benchmark of two calendar days. In 2023, the median percent for specimen receipt within two calendar days was 77.92% (n=37); only three programs met the NewSTEPs benchmark (**Table 14**, **Figure 13**) and one program met the ACHDNC one-day benchmark.

Table 14: First DBS specimens received at the NBS laboratory within two days of collection, 2018 to 2023 (data as of February 28, 2025).

Year	Number of Programs Reporting Data	Percent DBS Received within Two Days of Collection			Programs above the 95% Benchmark	
		Median	Min	Max	Number	Percent
2023	37	77.92%	0.0%	99.13%	3	8.11%
2022	36	74.82%	31.67%	99.07%	2	5.56%
2021	39	79.35%	53.57%	99.50%	3	7.69%
2020	38	76.21%	57.33%	99.24%	4	10.53%
2019	33	80.78%	54.55%	99.34%	1	3.03%
2018	36	78.48%	43.60%	99.55%	1	2.77%

Figure 13: In 2023, three NBS programs reached the 95% benchmark for specimen receipt within two days of collection (data as of February 28, 2025).



NBS laboratories have varying definitions of specimen receipt; it ranges from when the specimen is dropped off by the courier (n=2), when the specimen is recorded by laboratory staff (n=34), when testing is initiated (n=3) or other (n=3).[u] Overall, there has been little improvement in the time of specimen receipt at the NBS laboratory. Receipt within two days—let alone one day—of collection continues to be a challenge for NBS programs due to limited courier days, lack of courier service in remote areas and the number of days the laboratory is open to receive specimens.

Time from Birth to Reporting Out Results

Upon testing completion, NBS programs share results with the appropriate medical providers, which include hospitals, birthing centers, pediatricians and specialist centers. NewSTEPs separates reporting of results by time-critical disorders, non-time-critical disorders, and all normal and out-of-range results for all disorders.

NBS programs have indicated some variations in reporting results due to the limitations of different LIMS and processes. For out-of-range results, the report-out date/time is when a medical provider is notified of the actionable result, and for normal results, the report out date/time is when the final report is sent to the submitter (e.g., made available via a portal, HL7, etc.).[v]

Time-critical Disorders

The time-critical disorders were created based on a Society for Inherited Metabolic Disorders' position statement and expert opinions from various specialists. The list of time-critical disorders was updated in April 2023 to include secondary conditions (i.e., GA II, CPT II and CACT),[w] and Pompe and Infantile Krabbe Disease were added in 2024. The ACHDNC recommends that presumptive positive results for time-critical disorders should be communicated to the newborn's healthcare provider immediately, but no later than five days of life.

In 2023, the median percent of specimens with time-critical results reported within five days of birth is 50.52% (n=33); two programs met the 95% benchmark (**Table 15, Figure 14**).[x]

Table 15: Specimens with out-of-range results for time-critical disorders reported within five days of birth in 2018-2023 (data as of February 28, 2025).

Year	Number of Programs Reporting Data	Percent of Time Critical Specimens Reported within Five Days of Birth			Programs above the 95% Benchmark	
		Median	Min	Max	Number	Percent
2023	33	50.52%	1.70%	100.00%	2	6.06%
2022	34	42.04%	0.90%	100.00%	3	8.82%
2021	33	39.83%	0.00%	97.44%	3	9.09%
2020	32	45.77%	0.00%	97.30%	3	9.38%
2019	28	54.87%	7.20%	98.44%	3	10.71%
2018	26	51.07%	3.89%	98.62%	3	11.54%

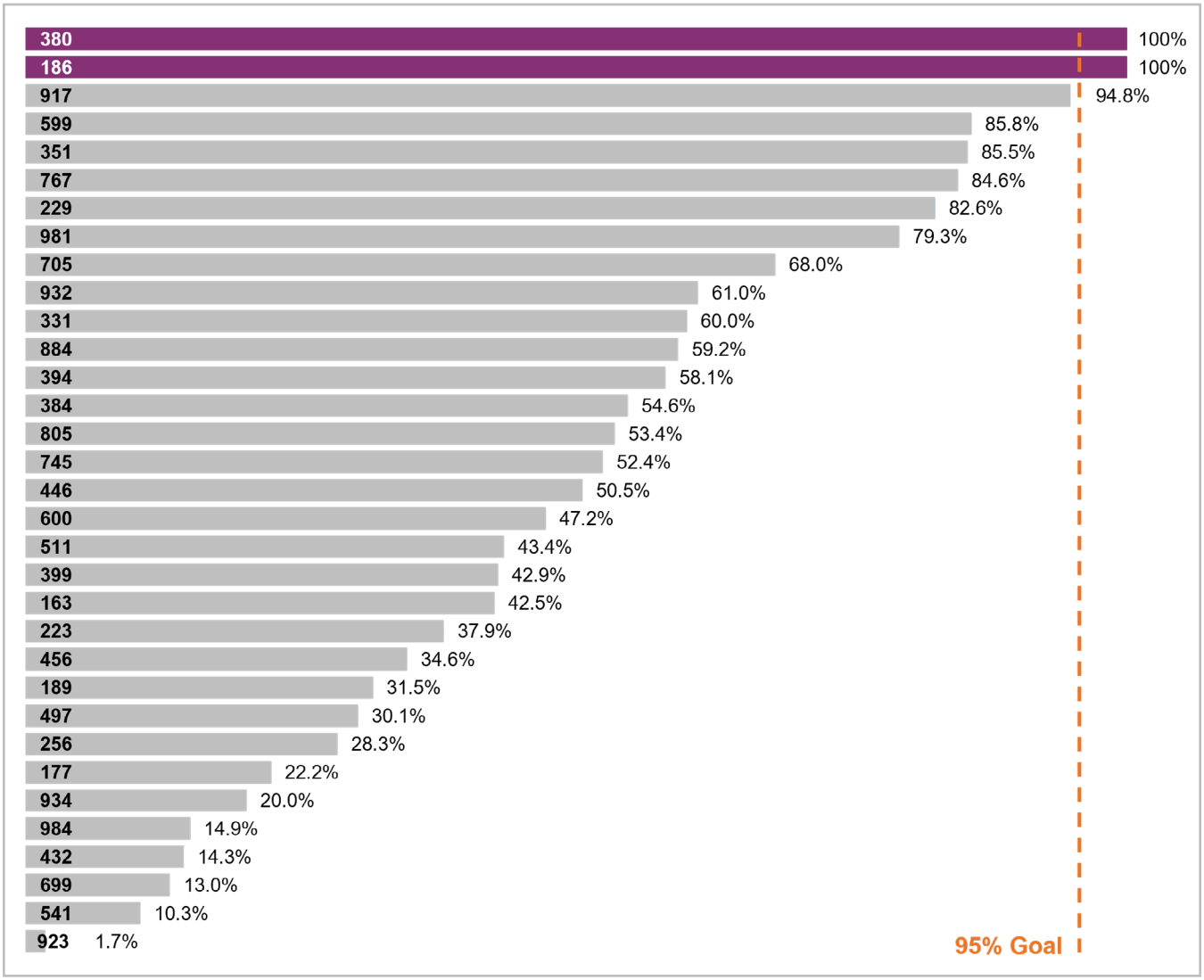
u Other definitions provided by programs are time-specific for specimen receipt.

v NBS programs likely capture the time of reporting out results differently, impacting the birth to report out metrics. Some programs may report when follow-up is notified, whereas other programs may use the report date for medical providers. NewSTEPs has clarified this in the definitions, but endpoints could still vary program to program.

w The addition in April 2023 of three secondary conditions (GA II, CPT II and CACT) to the list of time-critical disorders will impact the number of time-critical disorders reported across time, as most programs did not include these secondary disorders prior to the 2023 submission deadline.

x NewSTEPs does not specify whether this measure should include first specimens, requested subsequent specimens or routine second specimens. The purpose of this metric is to capture the time it takes to interpret the actionable result or act on the result. For this measure, time intervals should be calculated using the earliest specimen tested that led to the infant seeking diagnostic work-up.

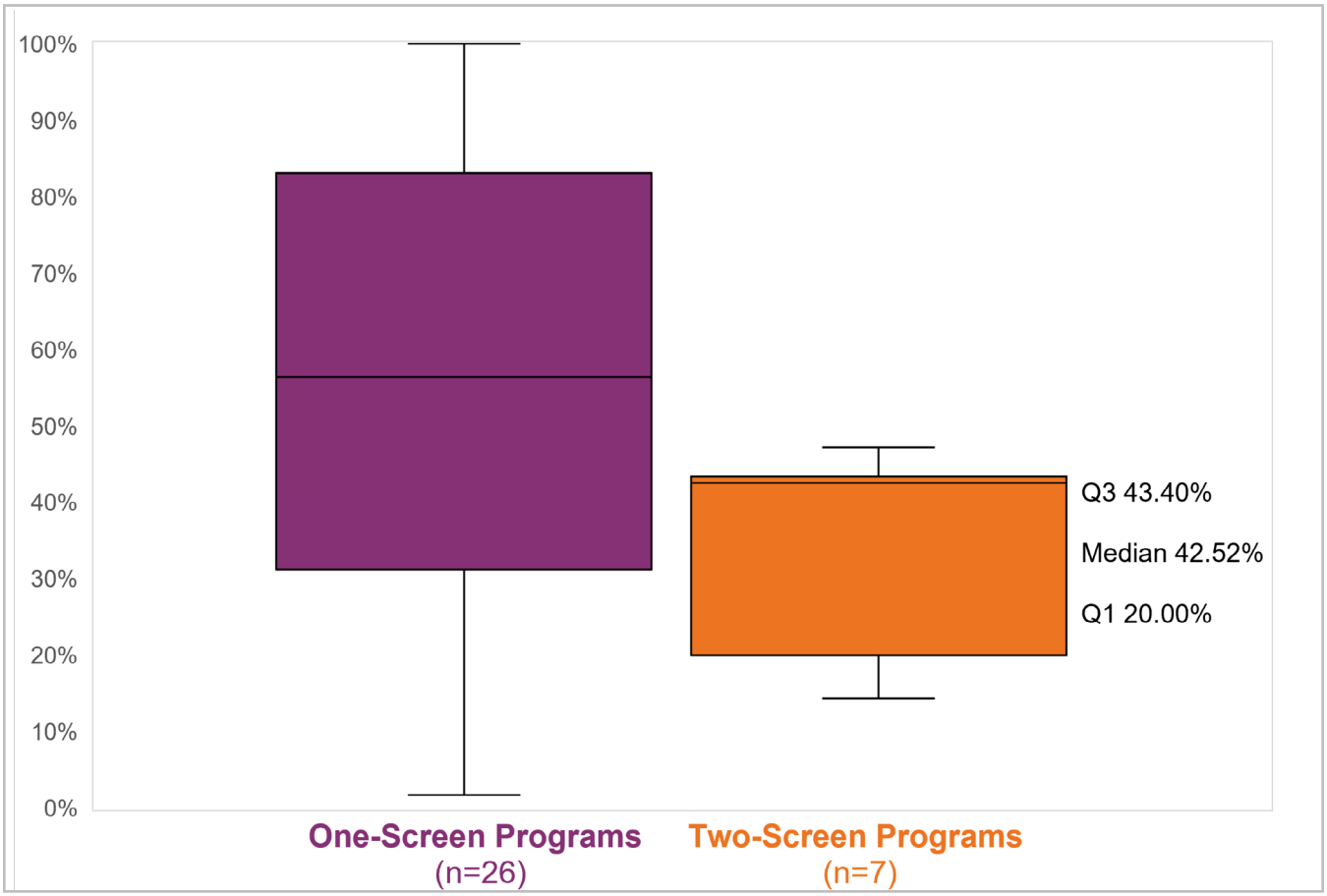
Figure 14: In 2023, two NBS programs reached the 95% benchmark for reporting time-critical results within five days of birth (data as of February 28, 2025).



Data show that despite significant investments in timeliness quality improvement, it remains difficult for NBS programs to reach the five-day benchmark for reporting time-critical results due to transportation and testing delays. For instance, some two-screen programs reported that it is difficult to meet the five-day benchmark as they are still waiting on the routine second specimen before they report the final NBS results. Most recently, NBS programs expressed concern about the difficulty of meeting the five-day benchmark for time-critical disorders that require a long incubation period during the analytic processes (i.e., Infantile Krabbe and Pompe).

NewSTEPs analyzed birth to reporting results for time-critical disorders within five days of birth by one-screen and two-screen programs. One-screen programs demonstrate higher overall performance, with a median of 56.36% (n=26) compared to 42.52% (n=7) for two-screen programs. The top 25% of one-screen programs reach 83%, while two-screen programs peaked at 43%—indicating that one-screen programs are more successful in reporting time-critical results within the first five days of life. However, one-screen programs exhibit greater variability, with a standard deviation of 29% compared to 13% for two-screen programs—showing that two-screen programs are more consistent but have lower performance (**Figure 15**). Statistical testing has not yet been conducted, so it is unclear whether these differences are statistically significant or not.

Figure 15: One-screen programs generally achieve higher performance for reporting time critical results within five days of birth, but with greater variation (data as of February 28, 2025).



Normal or Out-of-range Results for All Disorders

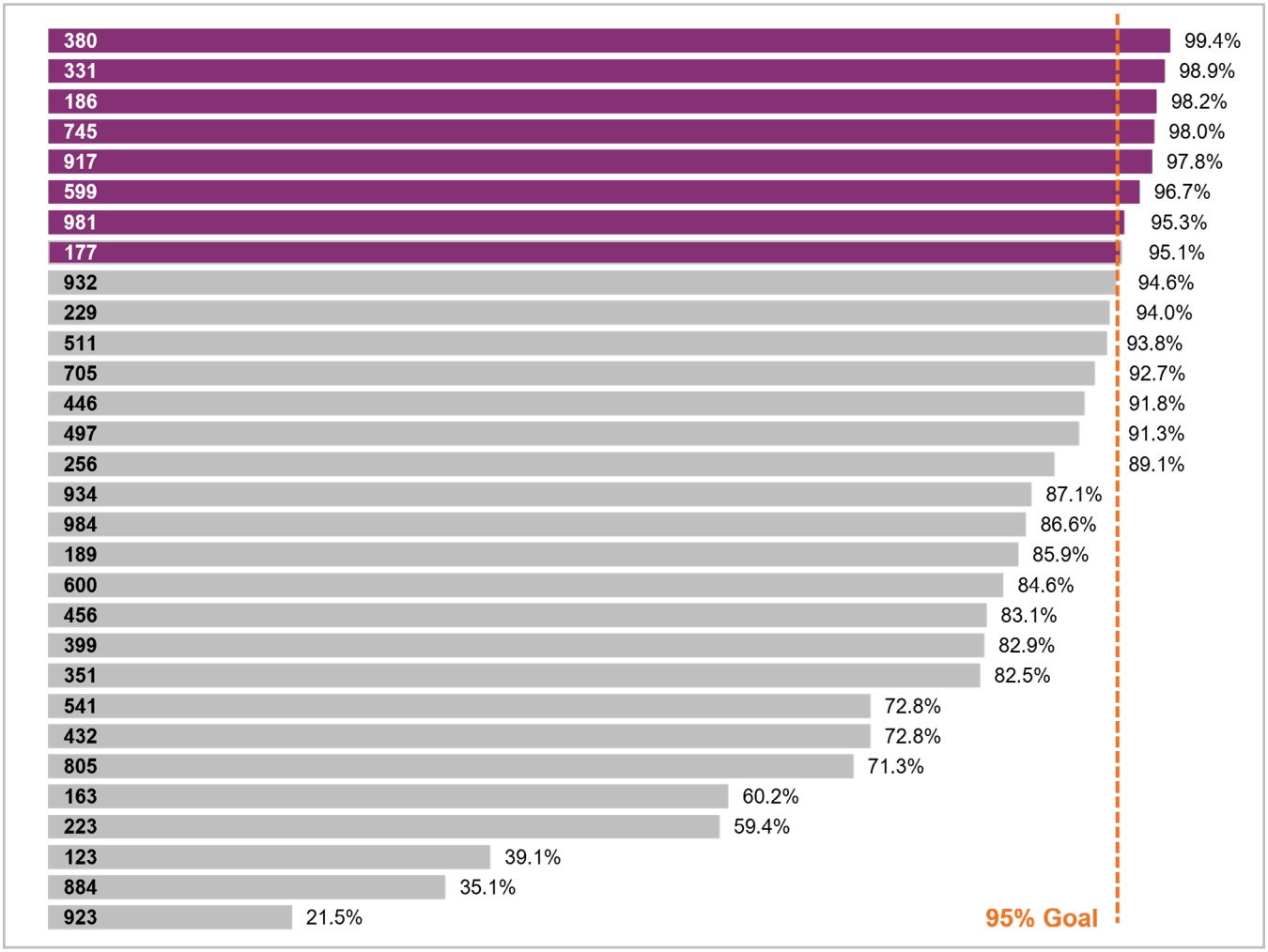
In addition to time of reporting for time-critical disorders, the ACHDNC recommends that all normal and out-of-range results for any NBS disorder be reported to an appropriate healthcare provider within seven days of life.

In 2023, the median percent of all first specimens with normal or out-of-range results for any disorder reported within seven days of birth was 87.10% (n=31), with eight programs reaching the 95% benchmark. This is a slight improvement from 2022 (86.75%, n=31), where six programs reached the benchmark (**Table 16**, **Figure 16**).

Table 16: First specimens with normal and out-of-range results for all disorders reported within seven days of birth, 2018–2023 (data as of February 28, 2025).

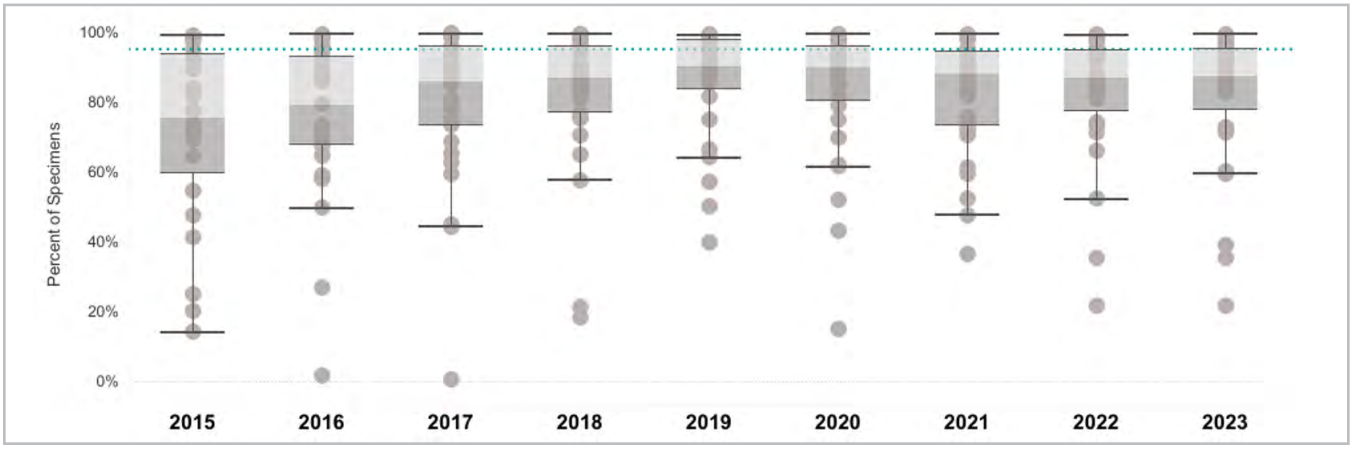
Year	Number of Programs Reporting Data	Percent of First Specimens with Normal or Out-of-range Results Reported within Seven Days of Birth			Programs above the 95% Benchmark	
		Median	Min	Max	Number	Percent
2023	31	87.10%	21.49%	99.34%	8	25.81%
2022	31	86.75%	21.59%	99.21%	6	19.35%
2021	33	87.99%	36.51%	99.47%	7	21.21%
2020	32	89.95%	14.76%	99.40%	9	28.13%
2019	27	90.17%	39.74%	99.27%	9	33.33%
2018	29	87.00%	18.12%	99.35%	8	27.59%

Figure 16: In 2023, eight NBS programs met the 95% benchmark for reporting all results within seven days (data as of February 28, 2025).



NBS programs are actively pursuing quality improvement initiatives to reduce the time between birth and result reporting. Enhancements in collection, transportation and testing have led to faster overall reporting. From 2015 to 2023, there is less variability and increasing consistency—reflecting improved performance and sustained progress toward reporting all results within seven days of life (**Figure 17**).

Figure 17: Decreased variability in first specimens reported with normal or out-of-range results within seven days of birth, 2015–2023 (data as of February 28, 2025).



Number of Cases Identified Through NBS

NewSTEPs collects de-identified case data at the individual level and aggregate confirmed case data for each birth year. Case data is reported to NewSTEPs on a two-year delay, allowing adequate time for NBS programs to gather and record the final diagnosis on the infant.

Aggregate cases reflect the total confirmed case counts per disorder by birth year, with primary congenital hypothyroidism (CH) the most prevalent NBS disorder in 2020–2022 (**Table 17**). Aggregate counts are self-reported by NBS programs; although NewSTEPs program staff do our best to validate this data, variations may exist depending on the case definitions utilized within the program and the clinical knowledge of entering data (e.g., for PKU some programs may have included cases of hyperphe, while others only include classical PKU). Additionally, some programs may face challenges classifying certain diseases, especially those with milder or later onset phenotypes. As a result, case counts may change as more diagnostic information is received; prior analyses[y] have shown that each year over 14,000 newborns are detected for the core RUSP disorders. NewSTEPs is currently working on standardizing case definitions to improve the accuracy of this data collection effort.[z]

Table 17: Total Case Counts Reported to NewSTEPs for Core RUSP Disorders, 2020-2022 (data as of March 17, 2025).*

Disorders		2022[aa] n=42	2021 n=44	2020 n=51
Amino Acid Disorders	ASA	18	11	15
	CIT I	19	17	16
	HCY	●[ab]	●	●
	MSUD	28	18	21
	PKU	170	206	240
	TYR I	7	15	16
Fatty Acid Disorders	CUD	25	25	34
	LCHAD	11	●	15
	MCAD	157	176	178
	TFP	●	●	●
	VLCAD	54	55	76
Organic Acid Disorders	3-MCC	69	69	68
	BKT	●	●	●
	Cbl A, B	8	7	●
	GA I	16	31	27
	HMG	●	●	●
	IVA	26	31	22
	MCD	●	●	●
	MUT	6	9	16
	PROP	20	19	24
Endocrine Disorders	CAH	163	134	267
	CH	2,105	2,052	2,512
Hemoglobin Disorders	Presence of Hb S[ac]	1,327	1,247	1,675
Lysosomal Storage Disorders	MPS I	23	25	24
	MPS II	9	NA	NA
	Pompe	119	109	132
Other Disorders	BIOT	162	186	198
	CF	607	655	761
	GALT	109	99	69
	SCID	56	48	71
	SMA	164	156	141
	X-ALD	131	132	145
Total		**5,617**	**5,543**	**6,781**

*** Additional Table Notes**

- Aggregate case counts do not include CCHD and hearing screens, or the newest disorders added to the RUSP in 2023-2024 (i.e., GAMT and Infantile Krabbe).
- Not all states and territories offered universal screening for all disorders during the reporting period; therefore, case counts may not be representative of the true national birth prevalence.

y Source: pubmed.ncbi.nlm.nih.gov/37092517/

z Case data presented in this report may change. The survey, *Outcomes Following a Screen Positive,* contains standardized case definitions which will eventually be integrated into the NewSTEPs case collection. Further, NewSTEPs case data will be updated to reflect the survey data received.

aa Aggregate case data for 2022 is pending.

ab NewSTEPs follows the "Rule of Five," which prevents data sharing if there are five newborns or fewer for a given category. The ● denotes disorder aggregate counts that are not being reported.

ac Subtypes of Sickle Cell Disease are combined into one category of "Presence of Hb S" in the NewSTEPs Repository. These are collectively referred to as sickling hemoglobinopathies (i.e., S,S Disease, S, beta thalassemia, and S,C disease). This should not include cases with an identified hemoglobinopathy trait, but some programs may have included trait in their reported case counts.

Appendix

Table 18: State and territory implementation of disorders, by year (orange cells denote the year the disorder was added to the core RUSP).

Disorder	2013	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	2024
Pompe *n=48* *Added to RUSP: March 2015*	Missouri	New York	Illinois	Kentucky Mississippi Pennsylvania	Michigan Minnesota Ohio Tennessee	California District of Columbia Massachusetts Nebraska Oregon Rhode Island	Maryland New Jersey Vermont Virginia Washington	Delaware Florida Indiana New Hampshire	Connecticut Georgia Kansas Maine New Mexico Oklahoma South Carolina	Colorado Guam Idaho Louisiana South Dakota Wisconsin Wyoming	Arizona Iowa North Carolina West Virginia	Alabama North Dakota
MPS I *n=45* *Added to RUSP: Feb. 2016*	Missouri		Illinois	Kentucky	Michigan Minnesota Pennsylvania Ohio Tennessee	District of Columbia Massachusetts Nebraska New York Oregon Rhode Island	Maryland New Jersey Vermont Virginia Washington	Delaware Florida Indiana New Hampshire	Connecticut Georgia Kansas Maine New Mexico Oklahoma South Carolina	Colorado Guam Idaho Louisiana Wyoming	Arizona Iowa North Carolina West Virginia	Alabama North Dakota
x-ALD *n=48* *Added to RUSP: Feb. 2016*	New York			California Connecticut	Minnesota Pennsylvania	District of Columbia Florida Kentucky Massachusetts Nebraska Rhode Island Tennessee Washington	Illinois Michigan Texas Vermont	Delaware Georgia New Hampshire Utah	Arizona Indiana Maine Missouri Oklahoma	Alaska Idaho New Jersey North Carolina Ohio Virginia	Alabama New Mexico Oregon	Hawaii Kansas Montana Nevada South Dakota
SMA *n=51* *Added to RUSP: 2018*						Indiana Massachusetts Minnesota New York Utah	Georgia Kentucky Maryland Missouri Mississippi New Hampshire Pennsylvania Texas Vermont West Virginia Wisconsin	Arkansas California Colorado Connecticut Delaware Florida Illinois Kansas Michigan Nebraska Rhode Island Tennessee Washington Wyoming	Iowa Maine Montana North Carolina North Dakota Oklahoma South Dakota Texas	Alabama Alaska Arizona Idaho Louisiana New Jersey New Mexico Ohio Oregon South Carolina Virginia	District of Columbia Nevada	Hawaii

Newborn Screening Technical Assistance and Evaluation Project

The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs) is a national newborn screening project designed to provide data, technical assistance, quality improvement resources and training to newborn screening programs. NewSTEPs functions with the goal of improving outcomes for newborns by facilitating newborn screening initiatives and programmatic outcomes, thus improving the overall quality of the newborn screening system.

Association of Public Health Laboratories

The Association of Public Health Laboratories (APHL) works to strengthen laboratory systems serving the public's health in the US and globally. APHL's member laboratories protect the public's health by monitoring and detecting infectious and foodborne diseases, environmental contaminants, terrorist agents, genetic disorders in newborns and other diverse health threats.

Contact Us

7700 Wisconsin Avenue, Suite 1000, Bethesda, MD 20814 | P: 240.485.2745 | aphl.org | newsteps.org