NewSTEPs
2022 Annual Report

July 2023

NewSTEPs

A Program of the Association of Public Health Laboratories™

CONTENTS

APHL gratefully acknowledges the contributions of all 53 NBS programs that submitted data to the NewSTEPs Data Repository. APHL also commends our members for their dedicated efforts to ensure that newborns are screened and treated in a timely fashion.

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INTRODUCTION

The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), a program of the Association of Public Health Laboratories, presents its 2022 Annual Report with the goal of sharing the state of newborn screening (NBS) laboratory and follow-up programs in the United States with our members and partners. Thank you to all the NBS programs contributing data to support data driven continuous quality improvement and information sharing efforts. Please direct any questions regarding this report to newsteps@aphl.org.

Note: Case data is represented as of March 2023; state profile, quality indicator data and new disorder implementation data is represented as of June 2023. While NewSTEPs utilizes numerous data collection methods and solutions for data entry, there has been a recent decrease in quality indicator and case data entered, partially due to ongoing impacts from the COVID-19 pandemic and compounded by public health workforce limitations. The contents of this report focus on noteworthy changes that occurred within the US NBS system since the publication of the 2019 NewSTEPs Report and the 2020 NewSTEPs Report.

APHL NEWBORN SCREENING

Vision

All babies have a healthier start through newborn screening in the US and globally.

Mission

Driving global NBS systems to excellence by shaping policy, promoting data- driven improvements, and pursuing innovations in public health lab practice.

STATE PROFILE DATA

NBS Programs Overview

There are 53 NBS programs represented in the NewSTEPs Data Repository, including all 50 states, the District of Columbia, Guam and Puerto Rico. There were 3,682,934 babies born in the 53 programs represented by NewSTEPs in 2022, a nonsignificant decline from 2021.[1] Number of births by state/territory, type of laboratory used for NBS, and number of required screens by state and territory are outlined in **Table 1**.

Table 1. NBS program overview (N=53)

NBS program	2022 births *(provisional)*	Laboratory type	Number of required screens
Alabama	58,079	State Public Health Laboratory	Two Screens
Alaska	9,331	Regional Laboratory	One Screen
Arizona	78,517	State Public Health Laboratory	Two Screens
Arkansas	35,380	State Public Health Laboratory	One Screen
California	418,523	State Public Health Laboratory	One Screen
Colorado◊	62,346	State Public Health Laboratory	Two Screens
Connecticut	35,323	State Public Health Laboratory	One Screen
Delaware	10,786	Private Laboratory	One Screen
District of Columbia	8,047	Private Laboratory	One Screen
Florida	224,226	State Public Health Laboratory	One Screen
Georgia	125,827	State Public Health Laboratory	One Screen
Guam	2,623 *Note: Birth data is from 2021 as 2022 data is unavailable*	Regional Laboratory	One Screen
Hawaii	15,225	Regional Laboratory	One Screen
Idaho	22,382	Regional Laboratory	Two Screens
Illinois	128,315	State Public Health Laboratory	One Screen
Indiana	79,598	Private Laboratory	One Screen
Iowa◊	36,482	State Public Health Laboratory	One Screen
Kansas	34,385	State Public Health Laboratory	One Screen
Kentucky	52,219	State Public Health Laboratory *Note: Kentucky outsources Lysosomal Storage Disorders to Mayo Clinic Laboratory*	One Screen
Louisiana	56,096	State Public Health Laboratory *Note: Louisiana outsources partial DBS screening to Revvity Omics*	One Screen
Maine	12,079	Regional Laboratory	One Screen
Maryland	68,694	State Public Health Laboratory	Two Screens
Massachusetts◊	68,613	State Public Health Laboratory	One Screen
Michigan	102,248	State Public Health Laboratory	One Screen
Minnesota	63,914	State Public Health Laboratory	One Screen

1 https://www.cdc.gov/nchs/data/vsrr/vsrr028.pdf

◊ Regional laboratories

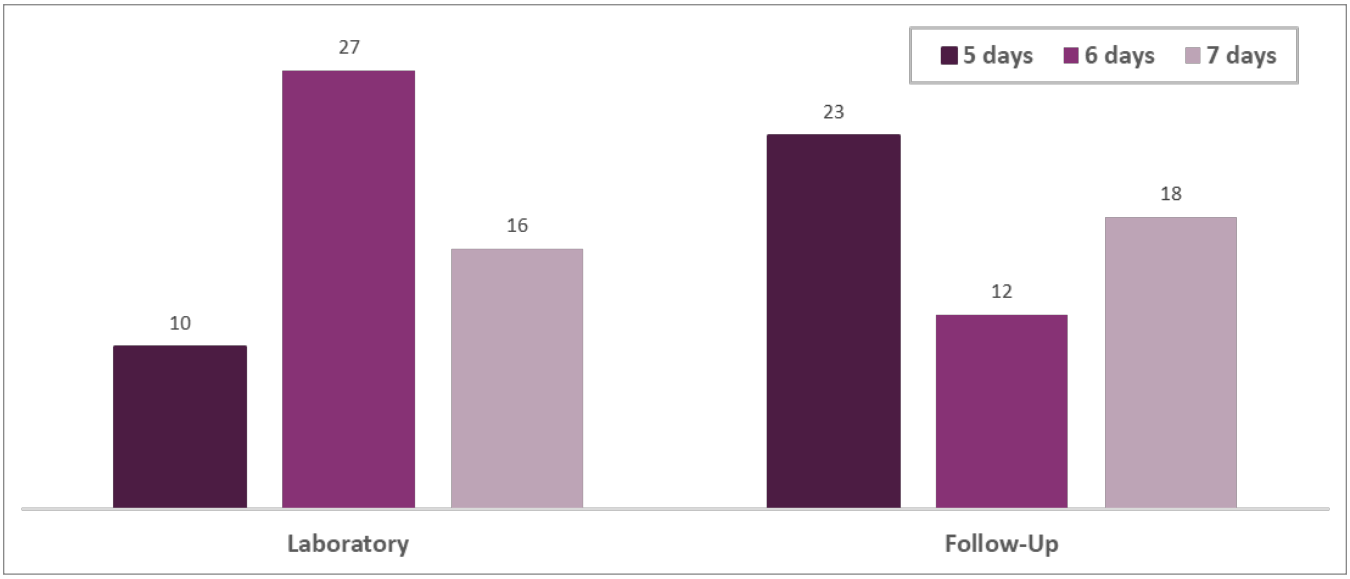
NBS program	2022 births *(provisional)*	Laboratory type	Number of required screens
Mississippi	34,609	Private Laboratory	One Screen
Missouri	68,977	State Public Health Laboratory	One Screen
Montana	11,154	State Public Health Laboratory ***Note:*** *Montana outsources MS/MS to Wisconsin State Laboratory of Hygiene*	One Screen
Nebraska	24,316	Private Laboratory	One Screen
Nevada	33,174	State Public Health Laboratory	Two Screens
New Hampshire	12,062	Regional Laboratory	One Screen
New Jersey	102,883	State Public Health Laboratory	One Screen
New Mexico	19,501	Regional Laboratory	Two Screens
New York	207,484	State Public Health Laboratory	One Screen
North Carolina	121,389	State Public Health Laboratory	One Screen
North Dakota	9,561	Regional Laboratory	One Screen
Ohio	128,221	State Public Health Laboratory	One Screen
Oklahoma	48,301	Private Laboratory	One Screen
Oregon◊	39,451	State Public Health Laboratory	Two Screens
Pennsylvania	130,003	Private Laboratory	One Screen
Puerto Rico	19,091	State Public Health Laboratory	One Screen
Rhode Island	10,214	Regional Laboratory	One Screen
South Carolina	57,775	State Public Health Laboratory	One Screen
South Dakota	11,188	Regional Laboratory	One Screen
Tennessee	82,262	State Public Health Laboratory	One Screen
Texas	389,533	State Public Health Laboratory	Two Screens
Utah	45,761	State Public Health Laboratory	Two Screens
Vermont	5,275	Regional Laboratory	One Screen
Virginia	95,405	State Public Health Laboratory	One Screen
Washington◊	83,207	State Public Health Laboratory	Two Screens
West Virginia	16,905	State Public Health Laboratory	One Screen
Wisconsin	59,930	State Public Health Laboratory	One Screen
Wyoming	6,044	Regional Laboratory	Two Screens

◊ Regional laboratories

NBS Program Operating Hours

NBS programs range in hours and days of operation, with varying levels of NBS programmatic activities performed on weekends and/or holidays, details of which can be found on the NewSTEPs State Profiles for each state/territory.[1] The majority of NBS laboratories are open 6 days per week (51%; n=27) and 30% (n=16) are open seven days a week. Similarly, 43% (n=3) of NBS follow-up programs are open five days a week; 23% (n=12) have follow-up open six days a week; and 34% (n=18) of follow-up programs operate seven days a week (**Figure 1**).

Figure 1. Number of days per week that NBS laboratory and follow-up programs remain open (N=53)



NBS Program Fees

Fees for initial newborn screens across the US states and territories range from $0 to $235 (N=53). Most US NBS programs charge between $51-150 (n=35) for their initial newborn screens, five programs impose no fee, four programs charge less than $50, and an additional nine programs charge greater than $151 (**Figure 2**). Analysis of NewSTEPs State Profile data reveal that there do not appear to be any correlations between initial NBS fee and either NBS program operating hours or number of disorders screened.

Figure 2. Distribution of initial NBS fees across NBS programs (N=53)

1 https://www.newsteps.org/data-resources/state-profiles

Most US NBS programs are funded by the NBS fee (n=44), while ten programs are funded by general funds, and one program is funded by both. Most NBS fees are collected via billing to hospitals/submitters (n=37), with other mechanisms of collecting fees including collection kit purchases (n=10), billing to Medicaid/insurance (n=2) or via electronic payment (n=1). Three NBS programs did not provide data to NewSTEPs on how their fees are collected.

NBS Laboratory Information Management Systems

Each NBS program has information management systems within their laboratory and follow-up programs that are vital for data storage, data organization, and data management. NBS Laboratory Information Management Systems (LIMS) vendors in US states and territories can be stratified into five categories: PerkinElmer/Revit (n=24), Neometrics/Natus (n=13), StarLIMS (n=3), internally developed/custom software (n=6) or other (n=7). Similarly, case management information systems for follow-up programs can be stratified into five categories: PerkinElmer/Revvity (n=14), Neometrics/Natus (n=10), StarLIMS (n=2), Internally Developed (n=17) or other (n=10) (**Table 2**).

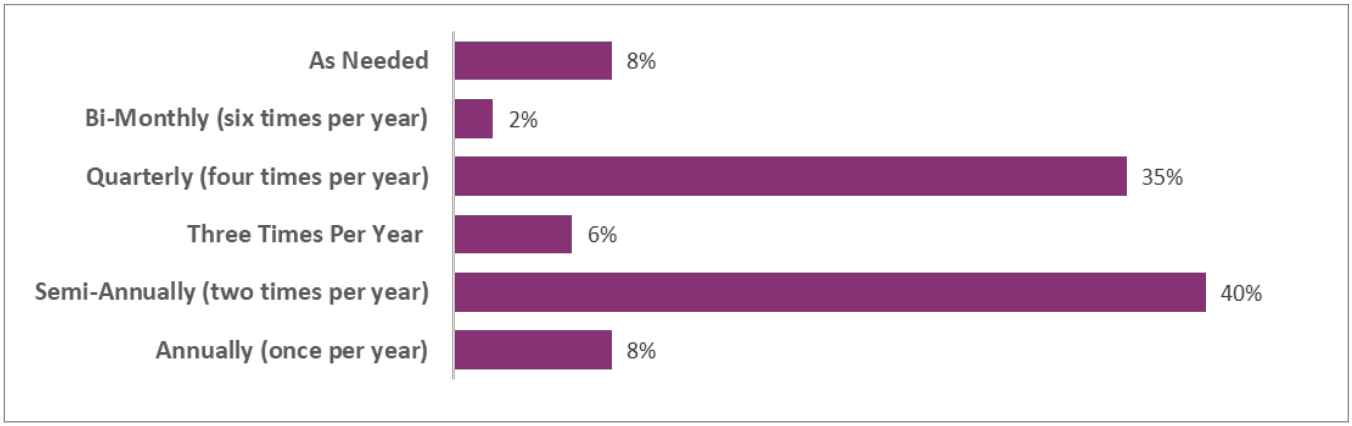
Table 2. NBS laboratory and follow-up program information management systems (N=53)

Information System Vendor	Laboratories Utilizing	Follow-Up Programs Utilizing
PerkinElmer/Revvity	24	14
Neometrics/Natus	13	10
StarLIMS	3	2
Internally Developed	6	17
Other	7	10

NBS Advisory Committees

State and territorial NBS advisory committees serve to evaluate and facilitate adding disorders to NBS panels and make recommendations regarding the structure of programs. Their role is to help ensure NBS programs effectively and efficiently screen, diagnose and treat newborns. Advisory committees frequently include varied representation from families, physicians, laboratory staff, follow-up program staff and other partners. In 2023, 91% (n=48) of NBS programs reported the existence of a NBS advisory committee in their state/territory, with 9% (n=5) NBS programs reporting that none currently exists. Of the 48 programs with an advisory committee, 25% (n=12) states/territories indicated that their advisory committee was mandatory, and the remaining 75% (n=36) programs indicated the advisory committee was voluntary. The advisory committees meet at varying frequencies throughout the year ranging from once per year to six times per year (**Figure 3**).

Figure 3. NBS advisory committee meeting frequencies (n=48)

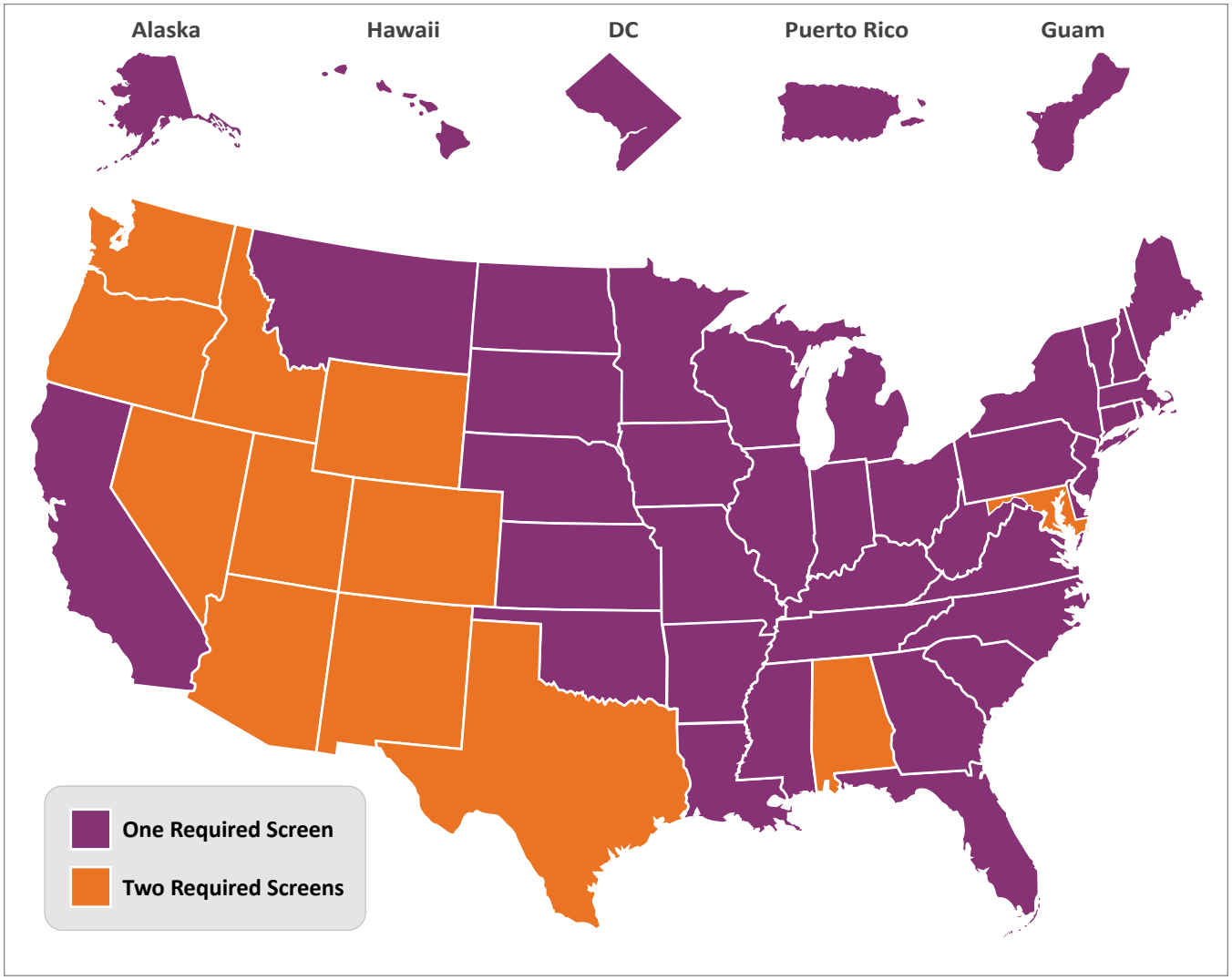


Number of Screens

Each state/territory has mandates to screen newborns, and these mandates specify if newborns will receive one or two screens. Twelve states (Alabama, Arizona, Colorado, Idaho, Maryland, Nevada, New Mexico, Oregon, Texas, Utah, Washington and Wyoming) are two-screen states recommending that a routine second dried blood spot (DBS) specimen be collected on all newborns regardless of the results of the first newborn screen (**Figure 4**). The purpose of the second screen is to improve the specificity and minimize delayed diagnoses of disorders that may not be detectable on the initial screen based on the assays performed in the two screen states. Out of the 12 two-screen states, seven have a policy that mandates a second screen on all infants for a partial panel of disorders; two have a policy that mandates a second screen on all infants for the full panel of disorders; two have a policy that recommends, but does not mandate, a second screen on all infants for a partial panel of disorders; and one program has a policy that recommends, but does not mandate, a second screen on all infants for the full panel of disorders.

Newborns in the other 41 states and territories typically undergo a single newborn screen. There are certain circumstances that may prompt an additional screen (referred to as a requested subsequent screen), such as when a specimen is collected too early or if there is an unsatisfactory specimen due to collection or transportation errors or if there is an abnormal result.

Figure 4. Number of required screens by state, June 2023 (N=53)



RECOMMENDED UNIFORM SCREENING PANEL

Core Disorders

US states and territories follow individual procedures for which disorders to add to their respective NBS panels, with many being guided by recommendations made by the Secretary of Health and Human Services for addition to the Recommended Uniform Screening Panel (RUSP). A complete list of the core RUSP disorders are displayed in **Table 3**.

According to NewSTEPs, as of June 2023 all states screen for at least 31 of 37 core disorders on the RUSP, with some states screening for an additional subset of secondary and other conditions. No states currently screen for all 37 core disorders on the RUSP (**Table 4**).

Table 3. Recommended Uniform Screening Panel Disorders

Year	Disorders Added to RUSP					
2006*	3-MCC ASA BIOT BKT CAH	Cbl A, B CF CIT CH CUD	GA1 GALT Hb S/B+ Th Hb S/C Hb SS	HCY HEAR HMG IVA LCHAD	MCAD MCD MSUD MUT PKU	PROP TFP TYR I VLCAD
2010	Severe Combined Immunodeficiency (SCID)					
2011	Critical Congenital Heart Disease (CCHD)					
2015	Pompe					
2016	Mucopolysaccharidosis Type I (MPS I) X-linked Adrenoleukodystrophy (XALD)					
2018	Spinal Muscular Atrophy (SMA)					
2022	Mucopolysaccharidosis Type II (MPS II)					
2023	Guanidinoacetate methyltransferase deficiency (GAMT)					

Table 4. Number of Core RUSP disorders screened by state/territory as of June 2023

# Core RUSP Disorders Screened	States				
31	Hawaii	Nevada	Puerto Rico		
32	Alaska	Montana	North Dakota		
33	Alabama Arkansas	Guam Mississippi	South Dakota Texas	Wisconsin	
34	Colorado District of Columbia	Iowa Kansas	Louisiana Maryland	South Carolina Utah	Wyoming
35	Arizona California Connecticut Idaho Indiana	Kentucky Maine Massachusetts Minnesota Nebraska	New Hampshire New Jersey New Mexico North Carolina Ohio	Oklahoma Oregon Pennsylvania Rhode Island Tennessee	Vermont Virginia Washington West Virginia
36	Illinois	Michigan	Missouri	New York	

* The first 29 disorders added to the RUSP are displayed here in their abbreviated form. For the full list please visit the NewSTEPs website.

Newborn Disorder Implementation

The initial 29 RUSP disorders involved single analyte Tandem Mass Spectrometry (MS/MS) laboratory analysis, while disorders added in the past decade have increasingly complex phenotypes, screening methodologies, and evolving treatment regimens. All states/territories currently offer near universal screening for most of the core RUSP disorders except for the following: Pompe, Mucopolysaccharidosis Type I (MPS I), x-linked adrenoleukodystrophy (x-ALD), Spinal Muscular Atrophy (SMA), Mucopolysaccharidosis Type II (MPS II), and Guanidinoacetate methyltransferase deficiency (GAMT). **Table 5** offers an implementation summary for these newest RUSP disorders, as of June 2023.

Table 5. Implementation summary for newest RUSP disorders, as of June 2023 (N=53)

Conditions	SCID	CCHD	POMPE	MPS I	x-ALD	SMA	MPS II	GAMT
Year Added to RUSP	2010	2011	2015	2016	2016	2018	2022	2023
Number of States Performing Population Screening	53	53	43	40	35	49	3	3
Percent of newborns with access to universal screening*	100%	100%	83%	80%	85%	98%	6%	10%
Average number of years to implement after addition to the RUSP	4.3	2.7	4.5**	3.6**	3.8**	2.1**	***	***

Tables 6–11 provide disorder-specific stratification of the year in which each state implemented universal NBS for Pompe, MPS I, x-ALD, SMA, MPS II and GAMT, respectively, as of June 2023. Orange shading within tables below represents the year a specific disorder was added to the RUSP.

Table 12 summarizes the data of the prior tables, offering a snapshot view of how many states have implemented each new disorder per year as of June 2023. This data is rapidly evolving and real time updates can be found on NewSTEPs data visualizations and reports online.

* Calculated using 2022 provisional births

** Of states offering universal screening

*** Insufficient data

Table 6. Pompe implementation dates, as of June 2023 (n=43)

2013	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023
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

Table 7. Mucopolysaccharidosis Type I implementation dates, as of June 2023 (n=40)

2013	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023
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

Table 8. x-linked Adrenoleukodystrophy implementation dates, as of June 2023 (n=35)

2013	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023
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

Table 9. Spinal Muscular Atrophy implementation dates, as of June 2023 (n=49)

2013	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023
					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	Alaska Alabama Arizona Idaho Louisiana New Jersey New Mexico Ohio Oregon South Carolina Virginia	District of Columbia

Table 10. Mucopolysaccharidosis Type II implementation dates, as of June 2023 (n=3)

2013	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023
				Illinois	Missouri					West Virginia

Table 11. Guanidinoacetate Methyltransferase Deficiency implementation dates, as of June 2023 (n=3)

2013	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023
		Utah			New York				Michigan	

Table 12. Number of states implementing screening for each new disorder, stratified by year as of June 2023

Condition	2013	2014	2015	2016	2017	2018	2019	2020	2021	2022	2023	Total
POM-PE	1	1	1	3	4	6	5	4	7	7	4	43
MPS I	1	0	1	1	5	7	5	4	7	5	4	40
XALD	1	0	0	2	2	8	4	4	5	6	3	35
SMA	-	-	-	-	-	5	10	14	8	11	1	49
MPS II	-	-	-	-	1	1	0	0	0	0	1	3
GAMT	-	-	1	0	0	1	0	0	0	1	0	3

NUMBER OF CASES IDENTIFIED THROUGH NBS

NewSTEPs collects individual-level case data for infants, which includes demographics, NBS information (including timeliness metrics) and diagnostic information to inform case classification. While not all programs provide this level of data due to a variety of reasons, including the voluntary nature of this data entry, participating programs reported individual-level cases to NewSTEPs from 2015–2021 (**Table 13**).

These include all core RUSP disorders. De-identified data sets are available upon request from NewSTEPs and these data have been analyzed in reports elsewhere on time to diagnosis and intervention stratified by race and ethnicity, with additional analyses forthcoming.[1]

Table 13. Individual core RUSP disorder cases reported to NewSTEPs, 2015-2021

Year	Individual Cases Reported
2015	4,124
2016	3,971
2017	4,691
2018	5,234
2019	4,706
2020	3,550
2021	3,508

NewSTEPs also collects aggregate confirmed case data for infants diagnosed through NBS each year. Confirmed case data is reported to NewSTEPs on a two-year lag time (i.e., for an infant with a confirmed case who is born in 2020, their case data is submitted by 2022), thereby allowing adequate time for the program to gather and record a final diagnosis on the infant. **Table 14** reflects data reported in aggregate for core RUSP disorders in 2018, 2019 and 2020, with the following notes and caveats:

- **Table 14** does not include CCHD or hearing loss cases.
- **Table 14** does not include GAMT cases due to the lack of data in the reporting period.
- The subtypes of sickle cell disease listed as separate diseases on the RUSP were combined into one category of "Presence of Hb S" to mirror collection terminology in the NewSTEPs Data Repository. These diseases are collectively referred to as sickling hemoglobinopathies and do not include cases with an identified hemoglobinopathy trait.
- **Table 14** includes data from 51 US NBS programs, however, not all states and territories were offering universal screening for all disorders (**Table 12**) during the reporting period and therefore these aggregate case counts are not always representative of the true national birth prevalence.
- Mississippi and the District of Columbia did not report aggregate cases for 2018-2020.
- Case counts are self-reported by NBS programs; variations may exist depending on the criteria utilized within the program and the clinical knowledge of entering data (e.g., some states might have counted other Hb S diseases in their case count for sickle cell disease, or some might have included cases identified with hemoglobinopathy trait).
- Some states may face challenges classifying certain diseases, especially those with milder or later-onset phenotype. As a result, case counts may change.

This de-identified aggregate data has been valuable in the national public domain, enhancing the body of literature around infants with congenital disorders identified through NBS in the United States. Based on a recent analysis conducted by NewSTEPs utilizing this data, approximately 8,180 infants with a disorder on the core RUSP will be detected annually through DBS based NBS (based on number of live births in 2021), assuming universal screening for all core RUSP disorders.[2] This represents a notable change in prevalence previously reported in literature.[3]

1 https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/meetings/newsteps-data-equity-analysis.pdf

2 https://www.mdpi.com/2409-515X/9/2/23

3 http://dx.doi.org/10.15585/mmwr.mm6936a6

Table 14. Number of aggregate cases of core RUSP disorders identified by NBS, 2018–2020 (n=51)*

Disorders		2018	2019	2020
Amino Acid Disorders	ASA	12	30	15
	CIT I	26	26	16
	PKU & Hyperphe	316	271	260
	MSUD	17	18	21
	TYR I	20	4	14
	HCY	8	3	5
Endocrine Disorders	CAH	272	241	260
	CH	2,402	2,430	2,542
Fatty Acid Oxidation Disorders	CUD	39	33	35
	LCHAD	8	9	15
	MCAD	237	227	180
	TFP	1	4	3
	VLCAD	57	75	71
Hemoglobin Disorders	Presence of Hb S**	1,891	1,885	1,759
Lysosomal Storage Disorders	Pompe	79	98	134
	MPS I	15	29	27
	MPS II	0	0	0
Organic Acid Disorders	HMG	4	3	1
	3-MCC	87	72	64
	BKT	3	7	5
	GA I	40	39	27
	MCD	2	3	2
	IVA	19	25	23
	Cbl A,B	8	6	8
	MUT	19	13	13
	PROP	21	17	24
Other Disorders	BIOT	215	235	202
	GALT	73	65	77
	CF	917	809	768
	SCID	57	55	68
	SMA	11	79	135
	XALD	90	106	149
Total* **		**6,966**	**6,917**	**6,923**

* Does not include CCHD, hearing loss cases or GAMT cases.

** The subtypes of sickle cell disease listed as separate diseases on the RUSP were combined into one category of "Presence of Hb S"

*** Includes data from 51 US NBS programs, however, not all states and territories were offering universal screening for all disorders during the reporting period and therefore these aggregate case counts are not always representative of the true national birth prevalence.

NEWSTEPS NBS PERFORMANCE METRICS

NewSTEPs utilizes quality indicators to track quality practices within and across NBS programs in the US to support data driven performance assessments and, ultimately, use these metrics to inform national and program specific quality improvement initiatives. The eight quality indicators span the NBS process from specimen collection through confirmation of a screened condition (**Table 15**).

Unsatisfactory Specimens

NBS DBS specimens may be deemed unsatisfactory for a variety of reasons upon receipt at the NBS laboratory. Additionally, NBS programs differ in which specimens are deemed unsatisfactory for screening. Examples of unsatisfactory specimens include specimens with insufficient quantity of blood, clotting, smearing or contamination, inadequately filled circles, oversaturation of blood, blood layering due to improper collection or incomplete drying.

According to the NewSTEPs state profile data, 52.8% (n=28) of NBS programs test all specimens and report results when possible; 20.8% (n=11) do not test unsatisfactory specimens, instead requesting repeat samples; 26.4% (n=14) report utilizing other unsatisfactory screening policies.

It is important to track the percentage of unsatisfactory specimens to determine if additional education is required at birthing centers to limit specimen quality issues and, therefore, limit delays in the screening process. During the global COVID-19 pandemic, which has left enduring impacts to the entire public health system, the healthcare system was burdened with an excess of patients, impacting quality practices resulting from workforce and resource shortages. NewSTEPs data shows that the median percent of unsatisfactory specimens across reporting programs has remained below 2%, but that the variation in range remains significant in 2022, between 0.2% and 5.3% (**Table 16**).

Table 15. NBS 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 essential data field upon receipt at the lab
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
QI 7	**Confirmed Cases:** Disorders detected by NBS with a confirmed diagnosis by an appropriate medical professional
QI 8	**Missed Cases:** Reported by disorder

Table 16. Percent of unsatisfactory DBS specimens, 2015–2022

Year	Number of Programs Reporting Data	Percent Unsatisfactory DBS Specimens		
		Median	Min	Max
2015	25	1.6%	0.1%	4.4%
2016	32	1.5%	0.1%	4.3%
2017	35	1.4%	0.1%	3.3%
2018	34	1.6%	0.2%	4.1%
2019	34	1.6%	0.2%	5.3%
2020	36	1.5%	0.2%	6.0%
2021	33	1.6%	0.2%	5.2%
2022	18	1.7%	0.2%	5.3%

Specimens with Missing Essential Information

In addition to checking for the quality of DBS specimens, NBS laboratories also check to see there are any missing state-defined essential information on the collection cards. Missing essential information may delay testing and reporting of results, causing potential harm to the newborn and requiring additional work for laboratory personnel to acquire the missing information. The national median for missing essential information remained at or below 3% across the years (**Table 17**), however, the range has consistently varied significantly across years. A subset of NBS programs maintain electronic demographic data entry capabilities, while others rely on manual and hand-written processes, a likely contributor to this range.

A potential bias in reporting for this and the previous (unsatisfactory specimens) quality metric is that two-screen states (n=12) include both first and second screen specimens in these counts, and all programs may be including subsequent repeat screen requests as well. Starting in 2023 NewSTEPs will be stratifying this data collection by first and routine second specimens, which will allow for a more accurate assessment over time.

Table 17. Percent of specimens with missing essential information, 2015-2022

Year	Number of Programs Reporting Data	Percent Specimens Missing Essential Information		
		Median	Min	Max
2015	18	2.1%	0.0%	33.8%
2016	24	2.6%	0.0%	21.5%
2017	28	3.0%	0.1%	21.6%
2018	26	2.9%	0.2%	20.8%
2019	26	2.5%	0.1%	16.8%
2020	27	2.5%	0.0%	19.9%
2021	29	2.9%	0.1%	19.0%
2022	19	2.3%	0.0%	22.0%

Unscreened Newborns

A key to ensuring health equity across the NBS system is to support access to screening for all newborns. NewSTEPs collects data on the proportion of newborns not screened due to parental refusals, pre-analytic errors, or missing/unmatched screens. A limitation to state and territorial public health departments determining if all newborns in their jurisdiction have received screening is the lack of uniform linkages to vital records and birth census data. Of the NBS programs that can report, the median percent of newborns not receiving a DBS newborn screen has remained at or below 0.3%, with the range varying from 0% to just under 4% in recent years (**Table 18**).

Table 18. Percent of newborns not receiving a DBS newborn screen, 2015–2021

Year	Number of Programs Reporting Data	Percent Newborns Without DBS Screen		
		Median	Min	Max
2015	6	0.2%	0.1%	2.6%
2016	10	0.2%	0.0%	2.4%
2017	10	0.1%	0.0%	1.7%
2018	12	0.3%	0.0%	1.9%
2019	13	0.3%	0.0%	2.0%
2020	17	0.3%	0.0%	3.6%
2021	19	0.3%	0.0%	3.4%

Infants Lost to Follow-Up

Another valuable metric for NBS programs to ensure quality practices is to track the percent of newborns that are lost to follow-up (i.e., have no final recorded resolution with the NBS program by 12 months of age). NewSTEPs collects three metrics relative to becoming lost to follow-up:

1. Infants with no final resolution following the receipt of an unsatisfactory DBS specimen.
2. Infants with no final resolution following a borderline result.
3. Infants with no final resolution following an out-of-range result.

NBS programs have indicated that this quality metric is difficult to collect because they may not receive reliable information, or they may not have a mechanism to collect this information. In 2021 and 2022 the maximum end of the range for percent of newborns lost to follow-up increased substantially from prior years. The marked increase in this upper range may correlate with the anecdotal data that fewer parents were returning for repeat or subsequent screens due to COVID-19 restrictions at hospitals or clinics and the post-COVID healthcare system burdens to reporting data back to the public health system. Importantly, having no recorded final resolution within the NBS program does not necessarily equate to harm to the 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 19**).

Table 19. Percent of infants that have no recorded final resolution within the NBS program, 2015–2022

Lost to Follow-up After...	Year	Number of Programs Reporting Data	Percent Infants Lost to Follow-up		
			Median	Min	Max
Receipt of an unsatisfactory specimen	2015	5	5.5%	2.3%	11.1%
	2016	5	6.1%	1.7%	9.5%
	2017	10	3.1%	0.0%	13.2%
	2018	12	3.3%	0.0%	14.5%
	2019	12	2.8%	0.0%	14.3%
	2020	15	3.3%	0.0%	31.5%
	2021	19	4.2%	0.0%	29.1%
	2022	8	4.0%	0.0%	17.1%
Receipt of a borderline result	2015	3	1.0%	0.0%	3.9%
	2016	3	0.8%	0.1%	9.7%
	2017	7	0.8%	0.0%	13.3%
	2018	8	1.2%	0.0%	6.8%
	2019	10	0.8%	0.0%	4.9%
	2020	17	1.4%	0.0%	11.7%
	2021	14	1.0%	0.0%	26.8%
	2022	8	1.8%	0.1%	23.3%
Receipt of an out-of-range result	2015	6	2.0%	0.7%	8.6%
	2016	6	2.1%	0.9%	7.9%
	2017	8	1.5%	0.0%	7.0%
	2018	10	1.5%	0.0%	6.4%
	2019	12	1.4%	0.0%	8.8%
	2020	18	2.3%	0.0%	14.3%
	2021	17	1.3%	0.0%	31.8%
	2022	9	2.1%	0.0%	20.1%

NBS TIMELINESS METRICS

Timeliness related quality indicators quantify time components across the NBS system that may be optimized with the goal of decreasing the time to identification of infants at risk for NBS disorders.

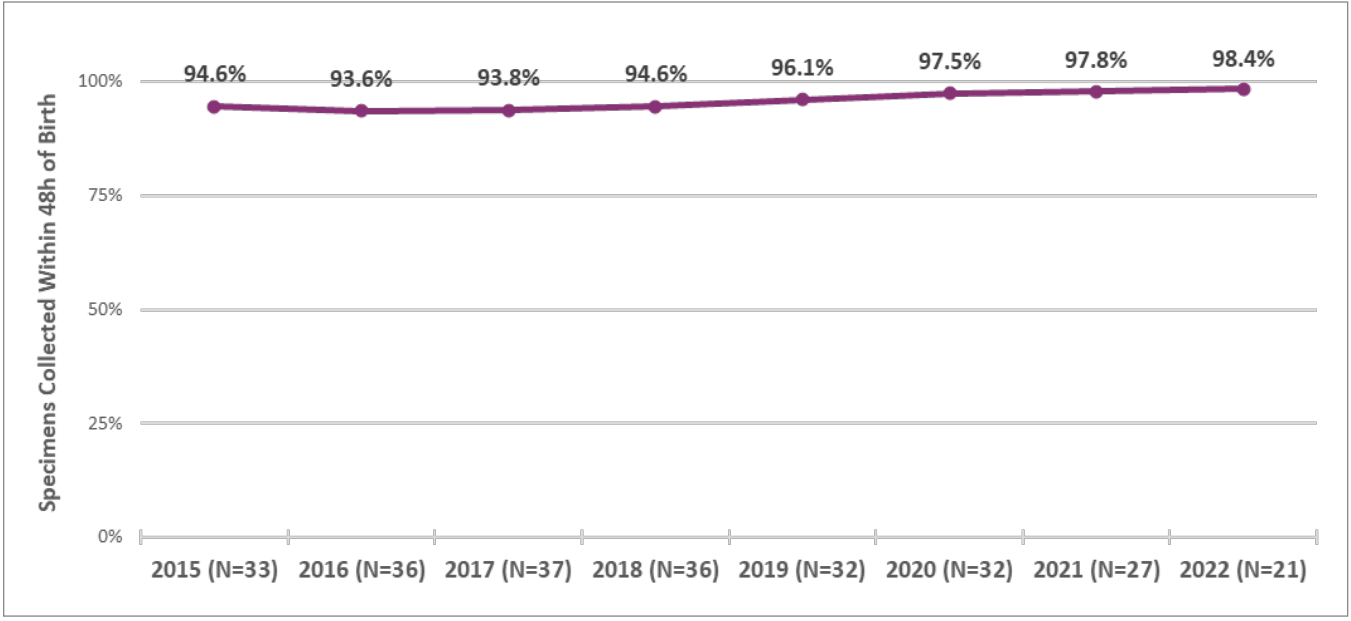
Specimen Collection

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) recommends that initial DBS specimens should be collected no later than 48 hours of life. The majority of NBS programs that submit data to NewSTEPs collect DBS specimens within this timeframe, with a gradual improvement each year (**Table 20, Figure 5**). In 2022, among the 21 NBS programs submitting data to NewSTEPs, the median percent of first DBS specimens collected within 48 hours from birth was 98.4%. Nineteen NBS programs reported that that over 95% of first DBS specimens were collected within 48 hours of birth. In the previous year (2021), 21 programs reported that over 95% of first DBS specimens were collected within 48 hours of birth.

Table 20. Percent of first DBS specimens collected within 48 hours of birth, 2015–2022

Year	Number Programs Reporting Data	Percent DBS Collected Within 48h			Programs Above 95%	
		Median	Min	Max	Number	Percent
2015	33	94.6%	61.0%	99.9%	13	39%
2016	36	93.6%	13.3%	99.4%	16	44%
2017	37	93.8%	13.2%	99.5%	16	43%
2018	36	94.6%	46.1%	99.6%	17	47%
2019	32	96.1%	20.1%	99.6%	20	63%
2020	32	97.5%	88.9%	99.7%	24	75%
2021	27	97.8%	83.8%	99.2%	21	78%
2022	21	98.4%	88.5%	99.2%	19	91%

Figure 5. Median of the percent of specimens collected within 48 hours of birth, 2015–2022



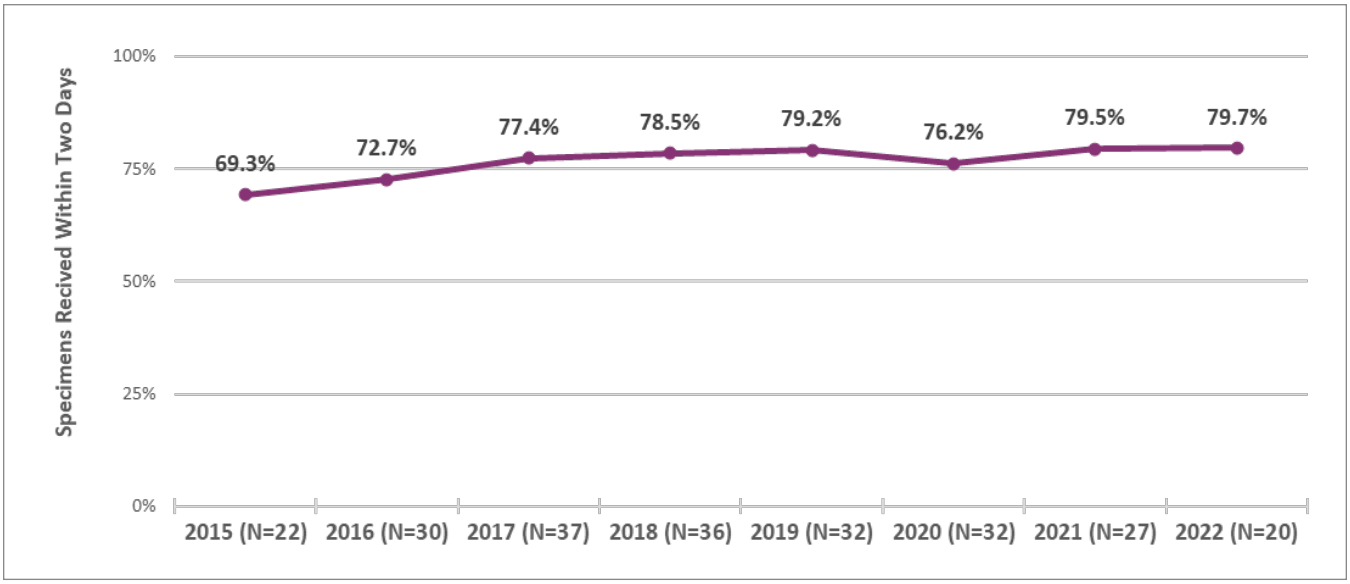
Specimen Transport

The ACHDNC recommends that NBS specimens should be received at the laboratory as soon as possible; ideally within 24 hours of collection. NewSTEPs utilizes the benchmark of specimen receipt of 95% of specimens within two calendar days. In 2022, three of the 20 reporting programs data received at least 95% of specimens within two days. In the previous year (2021), four programs (n=27) met this benchmark (**Table 21, Figure 6**). NBS laboratories have varying definitions of specimen receipt; it ranges from when the specimen is dropped off by the courier, to when the specimen is recorded by laboratory staff, through when testing is initiated. Therefore, interventions that would improve timeliness of specimen transport would involve increasing the number of days that NBS laboratories are open to accept specimens or increasing the number of days that a courier operates.

Table 21. Percent of DBS specimens received at the laboratory within two days of specimen collection, 2015–2022

Year	Number Programs Reporting Data	Percent DBS Received Within Two Days			Programs Above 95%	
		Median	Min	Max	Number	Percent
2015	22	69.3%	32.2%	98.7%	1	5%
2016	30	72.7%	38.6%	99.5%	1	3%
2017	37	77.4%	46.7%	99.6%	1	3%
2018	36	78.5%	43.6%	99.5%	1	3%
2019	32	79.2%	54.5%	99.3%	1	3%
2020	32	76.2%	57.3%	99.2%	4	13%
2021	27	79.5%	56.4%	99.5%	4	15%
2022	20	79.7%	57.4%	99.1%	3	15%

Figure 6. Median of the percent of specimens received at the laboratory within two days of specimen collection, 2015–2022



Birth to Reporting Out Results

Following testing of DBS specimens, NBS programs share results with their respective follow-up programs and distribute them to the appropriate medical providers (including hospitals/birthing centers). For reporting purposes, NewSTEPs separates reporting of results by time critical disorders, non-time critical disorders, and all results (normal and out-of-range), to be consistent with national recommendations.[1] The ACHDNC recommends that time critical results be reported within five days of life; and, that all results (normal and out-of-range) be reported within seven days of life.[2]

Data shows that despite significant investments in timeliness quality improvement, it remains difficult for NBS programs to reach the five-day benchmark for reporting of time critical results. Some of the challenges to meeting this recommendation include geographic limitations in states and territories with populations served across large areas.

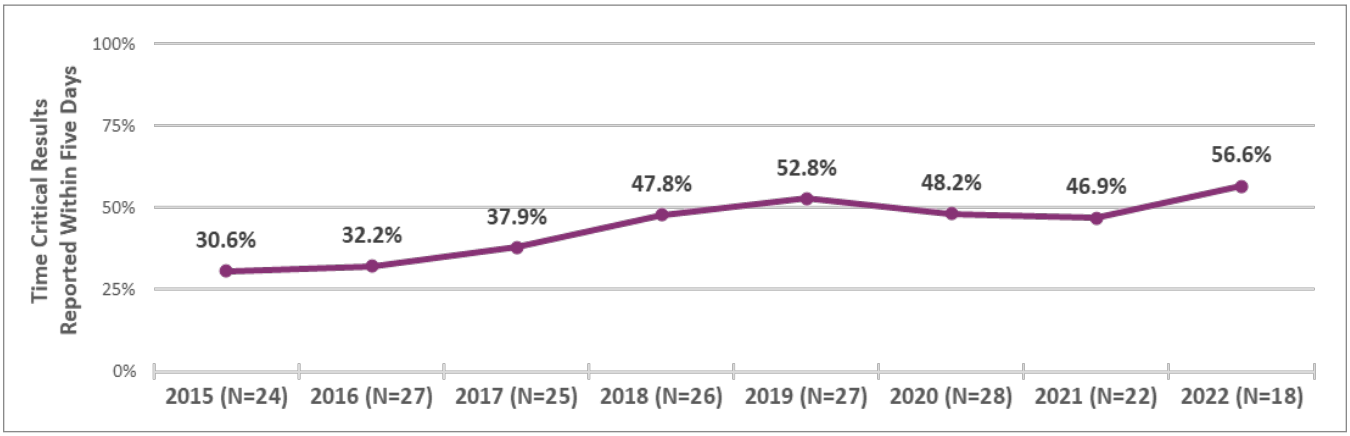
Time Critical Results

In 2022, the median percent of time critical results that were reported within five days of birth from 18 participating NBS programs was 56.6%, and in 2021 this median percent reported by 22 programs was 46.9% (**Figure 7**). In 2022 of the 18 programs reporting, two programs met the ACHDNC benchmark of 95% of time-critical results reported out within five days of birth. In 2021 four programs met this benchmark (**Table 22, Figure 7**).

Table 22. Percent of time critical results reported within five days of birth, 2015–2022

Year	Number Programs Reporting Data	Percent Time Critical Results Reported Within Five Days of Birth			Programs Above 95%	
		Median	Min	Max	Number	Percent
2015	24	30.6%	3.6%	99.0%	2	8%
2016	27	32.2%	0.0%	99.2%	3	11%
2017	25	37.9%	0.0%	99.5%	3	12%
2018	26	47.8%	3.9%	98.6%	3	12%
2019	27	52.8%	7.2%	98.4%	3	11%
2020	28	48.2%	0.0%	97.3%	3	11%
2021	22	46.9%	12.4%	100%	4	18%
2022	18	56.6%	3.3%	100%	2	11%

Figure 7. Median of the percent of specimens with time critical results reported out within five days of birth, 2015–2022



1 https://www.newsteps.org/media/8/download?inline=

2 https://www.hrsa.gov/advisory-committees/heritable-disorders/newborn-screening-timeliness

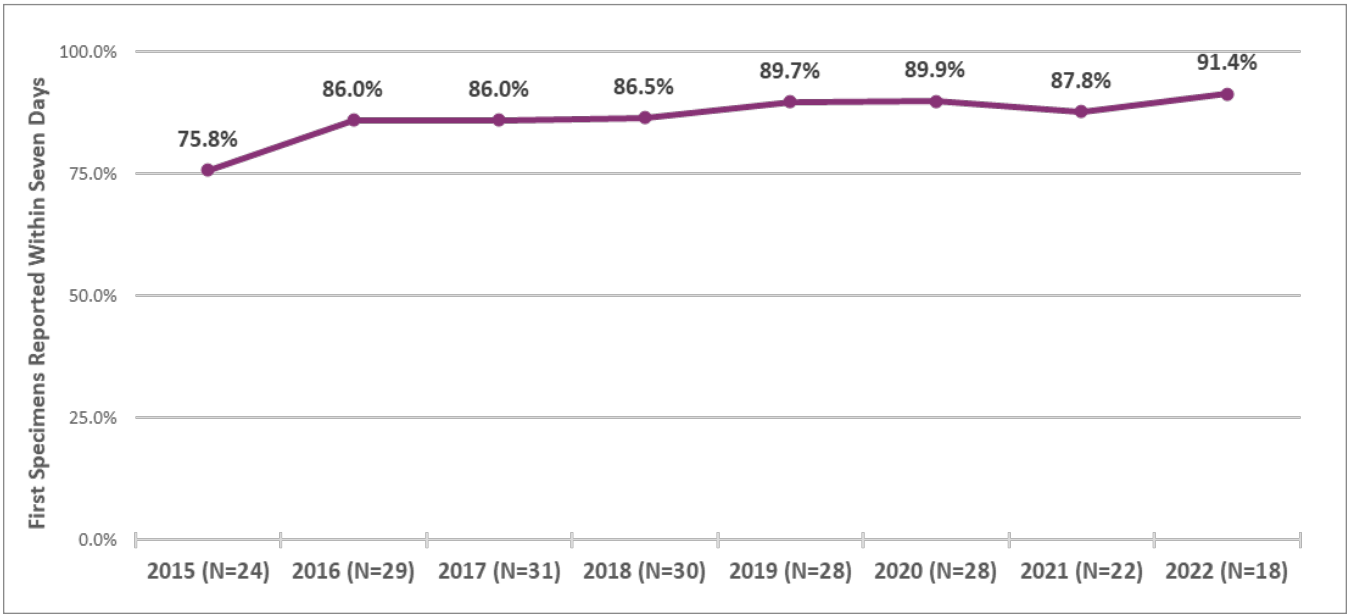
Normal and Out-of-Range Results

In 2022 the median percent of all results reported within seven days of life was 91.4% (n=18) and in 2021 this median was 87.8% (n=22) (**Table 23, Figure 8**). Of the 18 NBS programs submitting this data to NewSTEPs in 2022, four NBS programs met the ACHDNC benchmark stipulating that of 95% of all results (normal and out-of-range) should be reported out within seven days of life. In 2021 five programs met this benchmark (**Table 23**).

Table 23. Percent of all first specimen results (normal and out-of-range) reported within seven days of birth, 2015–2022

Year	Number Programs Reporting Data	Percent of First Specimens Received Within Seven Days of Birth			Programs Above 95%	
		Median	Min	Max	Number	Percent
2015	24	75.8%	14.1%	99.1%	6	25%
2016	29	86.0%	1.4%	99.4%	8	28%
2017	31	86.0%	0.6%	99.6%	8	26%
2018	30	86.5%	18.1%	99.3%	8	27%
2019	28	89.7%	39.7%	99.3%	9	32%
2020	28	89.9%	14.8%	99.4%	7	25%
2021	22	87.8%	36.5%	99.0%	5	23%
2022	18	91.4%	21.6%	99.0%	4	22%

Figure 8. Median of the percent of first specimen results (normal and out-of-range) reported within seven days of birth, 2015–2022



SUMMARY

This report illustrates the continued efforts of NBS programs and their dedicated staff in pursuing continual quality improvements, despite challenges lingering from public health disruptions including the effects of the COVID-19 pandemic, workforce shortages and resource limitations.

Importantly, this report presents data in a static moment of time, while data entry within the NewSTEPs Data Repository occurs on an ongoing basis. NewSTEPs provides real-time data visualizations of all the metrics described in this report, available online. NBS programs across the US are continuously working on quality improvement through implementation of new disorders, by enhancing utilization of electronic data exchange and automated data transfers, by improving timeliness of reporting results and more.

NewSTEPs serves as the national technical assistance resource center that collects, collates and shares successes and quality improvement practices in NBS, as well as offers continual technical assistance resources to enhance the national NBS system.

Newborn Screening Technical Assistance and Evaluation Project

The Newborn Screening Technical assistance and Evaluation Project (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

8515 Georgia Avenue, Suite 700, Silver Spring, MD 20910 | P: 240.485.2745 | www.aphl.org | www.newsteps.org