NewSTEPs
2019 ANNUAL REPORT

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NewSTEPs

A Program of the Association of Public Health Laboratories™

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INTRODUCTION

The Newborn Screening Technical assistance and Evaluation Program (NewSTEPs) is funded through a cooperative agreement between the Association of Public Health Laboratories (APHL) and the Genetic Services Branch of the US Health Resources and Services Administration (HRSA).

The formation of NewSTEPs has been a critical step in ensuring that states can adequately evaluate themselves through comparisons with other states using standard data and identify areas for quality improvement in newborn screening (NBS) programs. The activities of NewSTEPs are designed to build partnerships with the ultimate goal of maintaining and improving quality in NBS. The purpose of NewSTEPs is to strengthen existing newborn and genetic screening programs by providing data, technical and educational resources to various NBS stakeholders.

NewSTEPs VISION

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

NewSTEPs MISSION

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

THE STATE OF NEWBORN SCREENING IN THE US

Newborn screening (NBS) is a state-based public health system that screens newborns for congenital and inherited disorders that may not present with clinical symptoms at birth, but can cause permanent disability or death if not detected or treated within the first few days of life. NBS is a complex system that involves various partners (NBS programs, parents, clinicians, policy makers, information management vendors, birthing hospitals, courier services, etc.) and a range of activities (education, laboratory analysis, follow-up, treatment, monitoring and evaluation, advocacy, and data and information exchange).

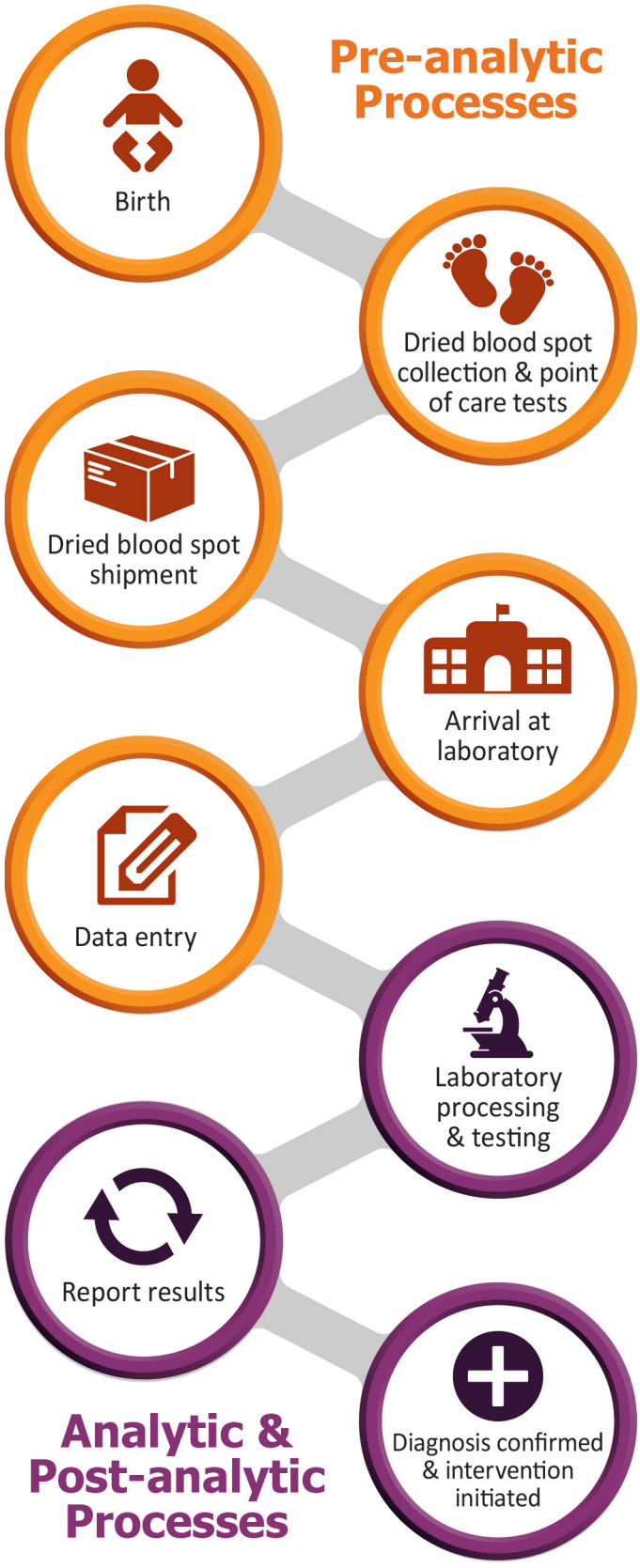
Each year in the United States, approximately four million newborns are screened and over 12,000 cases are identified with serious but treatable disorders. The Health and Human Services (HHS) Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) evaluates and recommends disorders to be included on the Recommended Uniform Screening Panel (RUSP).[1,2] Each state, however, mandates the specific disorders to be screened by their own program, implements system processes including follow-up of out-of-range results, and is responsible for quality improvement and assurance of the entire NBS system. Differences in the number and type of disorders screened in each NBS program are due to a variety of factors, including but not limited to:

- The prevalence of a disorder
- NBS programs' readiness or willingness to screen
- Infrastructure
- State legislative mandates
- Cost
- Availability of medical specialists.

This report will provide a snapshot of data collected in the NewSTEPs data repository following the NBS process of a dried blood spot (DBS) from collection to reporting of results and finally the confirmation of a diagnosis of an infant (**Figure 1**).

State profile level data, quality indicator data and case level data is represented as of September 2019.

Figure 1: Newborn Screening Process



1 CDC Grand Rounds: Newborn Screening and Improved Outcomes, 2012. MMWR Morb Moral Wkly Rep.2012;61(21):390-393.

2 HHS Recommended Uniform Screening Panel: https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html

Newborn Screening Programs Overview

There are 53 NBS programs represented in NewSTEPs, which include 36 newborn screening laboratories receiving specimens from not only their local state, but from other states as well. There are a number of NBS programs that outsource screening for specific disorders (e.g., Kentucky outsources testing of lysosomal storage disorders, and Montana outsources disorders detected using tandem mass spectrometry). For some programs, outsourcing with external laboratories to test DBS specimens may be more cost effective than performing screening in house depending on infrastructure and birth rate.[3] Number of births from 2017, type of laboratory used per state and territory and number of required screens by state and territory are outlined in **Table 1**. Details on number of required screens are provided in the Newborn Screening Specimen Collection section of this report.

Table 1: Newborn screening program overview, September 2019 (N=53)

NBS program	2017 births	Laboratory type	Number of required screens
Alabama	58,941	State public health laboratory	Two screens
Alaska	10,445	Regional laboratory	One screen
Arizona	81,872	State public health laboratory	Two screens
Arkansas	37,520	State public health laboratory	One screen
California	471,658	State public health laboratory	One screen
Colorado ◊	64,382	State public health laboratory	Two screens
Connecticut	35,221	State public health laboratory	One screen
Delaware	10,855	Private laboratory	Two screens
District of Columbia	9,560	Private laboratory	One screen
Florida	223,630	State public health laboratory	One screen
Georgia	129,243	State public health laboratory	One screen
Guam	3,297	Regional laboratory	One screen
Hawaii	17,517	Regional laboratory	One screen
Idaho	22,181	Regional laboratory	Two screens
Illinois	149,390	State public health laboratory	One screen
Indiana	82,170	Private laboratory	One screen
Iowa ◊	38,430	State public health laboratory	One screen
Kansas	36,519	State public health laboratory	One screen
Kentucky	54,752	State public health laboratory*	One screen
Louisiana	61,018	State public health laboratory	One screen
Maine	12,298	Regional laboratory	One screen
Maryland	71,641	State public health laboratory	Two screens
Massachusetts ◊	70,702	State public health laboratory	One screen
Michigan	111,426	State public health laboratory	One screen
Minnesota	68,595	State public health laboratory	One screen
Mississippi	37,357	Private laboratory	One screen
Missouri	73,034	State public health laboratory	One screen

3 Delaware Today, Dec. 19, 2017. Delaware gives $4.3 million contract for newborn screenings to Nemours. https://www.delawareonline.com/story/news/health/2017/12/20/delaware-gives-4-3-million-contract-newborn-screenings-nemours/960589001/

NBS program	2017 births	Laboratory type	Number of required screens
Montana	11,799	State public health laboratory**	One screen
Nebraska	25,821	Private laboratory	One screen
Nevada	35,756	State public health laboratory	Two screens
New Hampshire	12,116	Regional laboratory	One screen
New Jersey	101,250	State public health laboratory	One screen
New Mexico	23,767	Regional laboratory	Two screens
New York	229,737	State public health laboratory	One screen
North Carolina	120,125	State public health laboratory	One screen
North Dakota	10,737	Regional laboratory	One screen
Ohio	136,832	State public health laboratory	One screen
Oklahoma	50,214	State public health laboratory	One screen
Oregon ◊	46,361	State public health laboratory	Two screens
Pennsylvania	137,745	Private laboratory	One screen
Puerto Rico	24,310	State public health laboratory	One screen
Rhode Island	10,638	Regional laboratory	One screen
South Carolina	57,029	State public health laboratory	One screen
South Dakota	12,134	Regional laboratory	One screen
Tennessee	81,016	State public health laboratory	One screen
Texas	382,050	State public health laboratory	Two screens
Utah	48,585	State public health laboratory***	Two screens
Vermont	5,655	Regional laboratory	One screen
Virginia	100,391	State public health laboratory	One screen
Washington ◊	87,562	State public health laboratory	Two screens
West Virgina	18,675	State public health laboratory	One screen
Wisconsin	64,975	State public health laboratory	One screen
Wyoming	6,903	Regional laboratory	Two screens

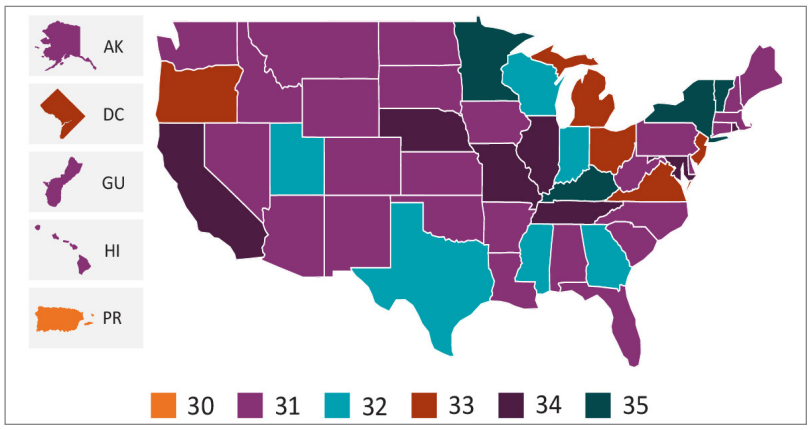
◊ Regional laboratories

* Kentucky outsources Lysosomal Storage Disorders to Mayo Clinic Laboratory
** Montana outsources MS/MS to Wisconsin State Laboratory of Hygiene
*** Utah outsources MS/MS to ARUP Laboratories

Disorders Screened

At the recommendation of ACHDNC, the US Health and Human Services (HHS) Secretary has recommended 35 NBS disorders for the RUSP.[4] As of September 2019, all NBS programs screen for at least 30 disorders on the RUSP and six NBS programs screen for all 35 NBS disorders (**Figure 2**).

Figure 2: Number of RUSP core disorders universally screened by state, September 2019



4 HRSA. Federal Advisory Committees, Recommended Uniform Screening Panel. https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html

After the initial 29 disorders were added to the RUSP in 2005, Severe combined immunodeficiency (SCID) was added in 2010, followed by Critical congenital heart disease (CCHD) in 2011, Pompe disease in 2015, and X-linked adrenoleukodystrophy (X-ALD) and Mucopolysaccharidosis type I (MPS I) in 2016. Spinal muscular atrophy (SMA) is the most recent disorder added to the RUSP in 2018. As of September 2019, 100% of NBS programs screen for SCID,[5] 100% (N=53) screen for CCHD, 40% (n=21) screen for Pompe, 30% (n=16) screen for X-ALD, 36% (n=19) screen for MPS I and 21% (n=11) screen for SMA (**Tables 2** and **3**). A visual representation of the NBS status of most recently added RUSP core disorders is depicted in **Table 3**.

Table 2: Number and percentage of newborn screening programs universally screening for most recently added RUSP disorders, September 2019 (N=53)

Disorder	Year added to the RUSP	NBS programs offering universal screening	Newborns with access to universal screening
SCID	2010	52	98%
CCHD	2011	53	100%
Pompe	2015	21	40%
X-ALD	2016	16	30%
MPS I	2016	19	36%
SMA	2018	11	21%

Table 3: NBS status of most recently added RUSP disorders, September 2019 (N=53)

● Universally screened ◆ Offered to select populations X Not screened

State	CCHD (2010)	SCID (2011)	Pompe (2015)	MPS I (2016)	X-ALD (2016)	SMA (2018)
Alabama	●	●	X	X	X	X
Alaska	●	●	X	X	X	X
Arizona	●	●	X	X	X	X
Arkansas	●	●	X	X	X	X
California	●	●	●	●	●	X
Colorado	●	●	X	X	X	X
Connecticut	●	●	X	X	●	X
Delaware	●	●	X	X	X	X
District of Columbia	●	●	●	●	●	X
Florida	●	●	X	X	●	X
Georgia	●	●	X	X	X	●
Guam	●	●	X	X	X	X
Hawaii	●	●	X	X	X	X
Idaho	●	●	X	X	X	X
Illinois	●	●	●	●	●	X
Indiana	●	●	X	X	X	●
Iowa	●	●	X	X	X	X
Kansas	●	●	X	X	X	X
Kentucky	●	●	●	●	●	●

5 Pennsylvania is unique due to legislative rules, however each newborn has access to SCID NBS.

● Universally screened ◆ Offered to select populations X Not screened

State	CCHD (2010)	SCID (2011)	Pompe (2015)	MPS I (2016)	X-ALD (2016)	SMA (2018)
Louisiana	●	●	X	X	X	X
Maine	●	●	X	X	X	X
Maryland	●	●	●	●	X	●
Massachusetts	●	●	●	●	●	●
Michigan	●	●	●	●	X	X
Minnesota	●	●	●	●	●	●
Mississippi	●	●	●	X	X	X
Missouri	●	●	●	●	X	●
Montana	●	●	X	X	X	X
Nebraska	●	●	●	●	●	X
Nevada	●	●	X	X	X	X
New Hampshire	●	●	X	X	X	X
New Jersey	●	●	●	●	X	X
New Mexico	●	●	X	X	X	X
New York	●	●	●	●	●	●
North Carolina	●	●	X	X	X	X
North Dakota	●	●	X	X	X	X
Ohio	●	●	●	●	X	X
Oklahoma	●	●	X	X	X	X
Oregon	●	●	●	●	X	X
Pennsylvania	●	◆	●	●	●	●
Puerto Rico	●	●	X	X	X	X
Rhode Island	●	●	●	●	●	X
South Carolina	●	●	X	X	X	X
South Dakota	●	●	X	X	X	X
Tennessee	●	●	●	●	●	X
Texas	●	●	X	X	●	X
Utah	●	●	X	X	X	●
Vermont	●	●	●	●	●	●
Virginia	●	●	●	●	X	X
Washington	●	●	X	X		X
West Virginia	●	●	X	X	X	X
Wisconsin	●	●	●	X	X	●
Wyoming	●	●	X	X	X	X

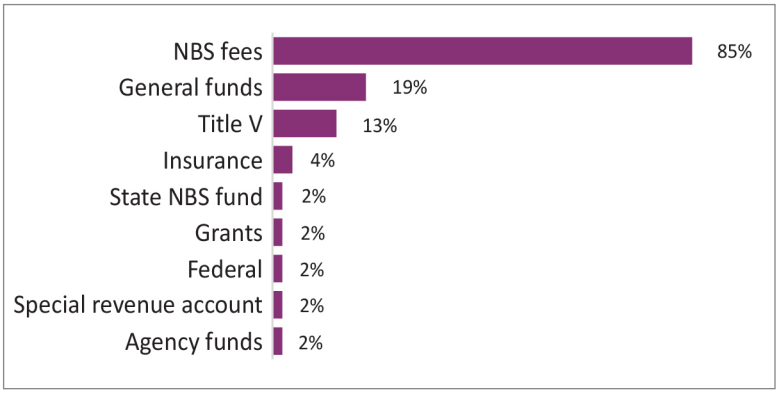
State Advisory Committees

Eighty-nine percent (n=47) of NBS programs reported having a newborn screening advisory committee, which may evaluate and facilitate adding disorders to individual state NBS panels. State advisory committees often include parents, physicians, laboratory staff and follow-up staff, among other stakeholders. They represent the interests of the state and make recommendations about the implementation and structure of NBS programs. Their role is to help ensure programs effectively and efficiently screen, diagnose and treat newborns for disorders on the state panel. The committees meet at various frequencies throughout the year. Of the 52 programs that submitted data, 75% (n=39) reported that adoption of a new disorder is pursued in that state or territory as official policy or procedure following addition of the disorder to the RUSP.

Newborn Screening Funding

The majority of NBS programs are funded through a single funding source (75%, n=39/52), which either includes the NBS Fee (63%, n=33), Title V funding (2%, n=1), general funds (6%, n=3), a state-funded NBS fund (2%, n=1) or special revenue funds (2%, n=1). Eleven programs (21%) are funded by two funding sources, with half being funded by either the NBS Fee and General Funds (6%, n=3), or the NBS Fee and Title V funding (4%, n=2). The other five remaining programs include a combination of NBS Fee, state or agency funds, general funds, insurance reimbursement, Federal or grant funding, or Title V funding. Two programs (4%) are funded by three funding sources, which include a combination of the NBS Fee, General Funds, Title V funding, insurance reimbursement and some other type of Federal or grant funding (**Figure 3**).

Figure 3: Newborn screening funding sources as reported by NBS programs, September 2019 (n=52)

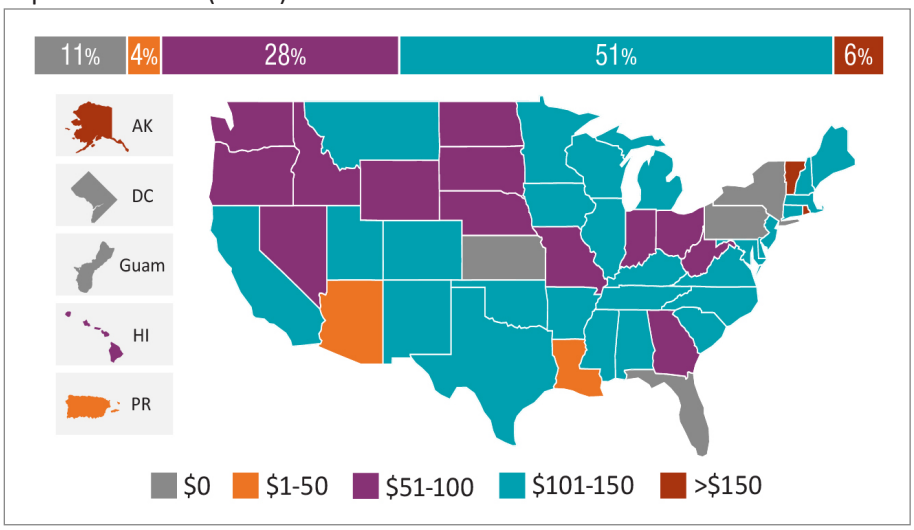


Newborn Screening Fees

Fees for initial dried blood spot (DBS) screening range from $0-$203 (mean=$98, median=$110, N=53) with a little more than half (n=27) of NBS programs having a fee between $101-$150 for the initial screen. Six (11%) programs do not have an initial NBS fee (**Figure 4a**).

The majority of NBS programs include a repeat screen in the initial fee, with the exception of ten NBS programs that charge an additional fee ranging from $55-$150 (mean=$105, median=$114, n=10 (**Table 4b**).

Figure 4a: Initial newborn screening fee, percent of total and by state, September 2019 (N=53)*



* NBS costs may also be supported by general funds or other funding sources within the state.

Table 4b: Repeat newborn screening fees, September 2019 (n=10)

Program	Repeat Fee
Texas	$55.24
New Hampshire	$71
Ohio	$74.61
Missouri	$95
Mississippi	$110
Michigan	$117.69
Illinois	$118
Arkansas	$121
Montana	$134
Minnesota	$150

Figure 5: NBS fee collection method as reported by NBS programs, September 2019 (N=53)

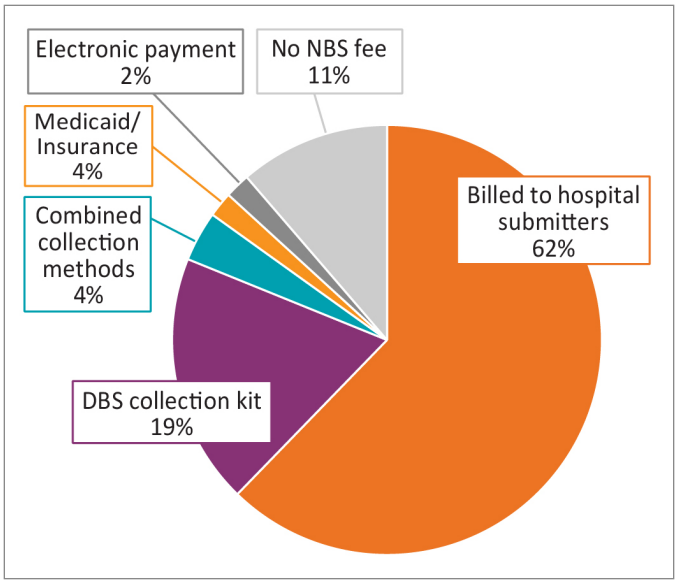
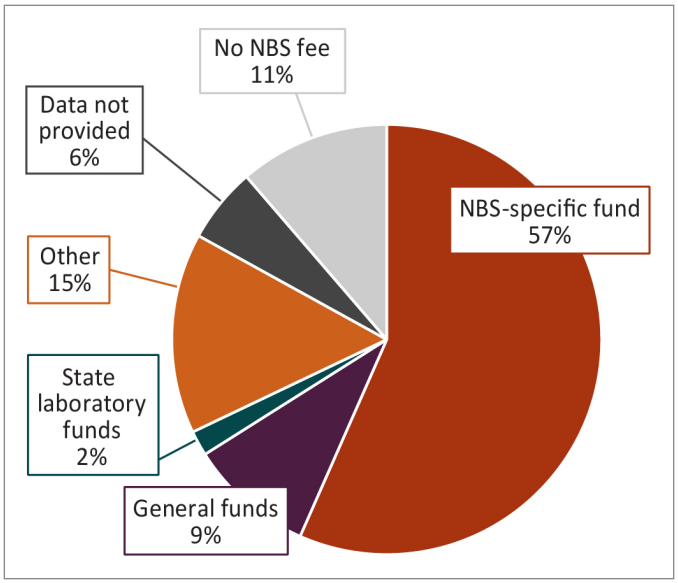


Figure 6: NBS fee holding location as reported by NBS programs, September 2019 (N=53)

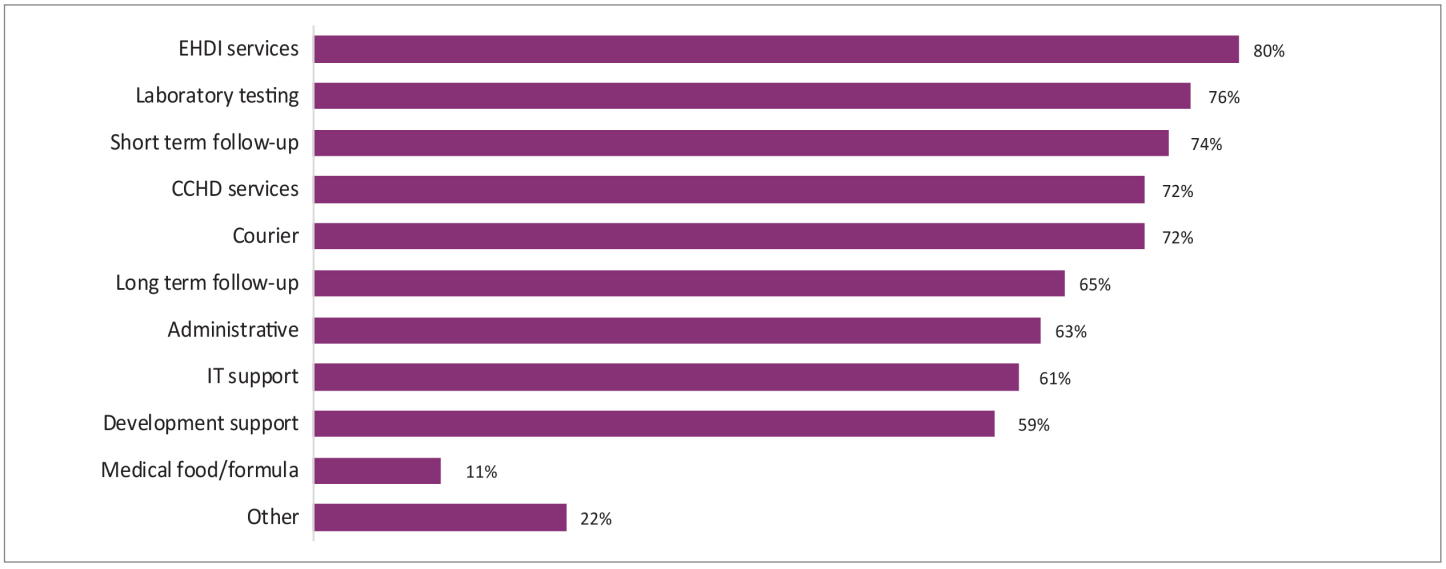


NBS programs collect fees in multiple ways and the categories are not mutually exclusive. Sixty-two percent (n=33) of NBS programs collect NBS fees by billing directly to birthing facilities or submitters, 19% (n=10) charge for DBS collection kits, 2% (n=1) bill to Medicaid or insurance, 4% (n=2) use a combined fee collection method (bill to birthing facilities or submitters and to Medicaid or insurance), 2% (n=1) accept electronic payment and 11% (n=6) have no NBS fee (**Figure 5**).

The NBS fee holding location is where NBS fees are kept after collection. The majority of NBS programs place collected fees in an NBS-specific fund; some hold fees in a general fund, in their state laboratory fund, and in another location (**Figure 6**). Examples of other locations include in the state public health services fund, general cash funds for metabolic foods, contracted laboratory funds, or not with the NBS program at all.

Fee use varies by program, but the majority of NBS programs reported using their NBS fees to support Early hearing detection and intervention, followed by laboratory testing, short-term follow-up, CCHD services and courier services. Only five NBS programs used their fees to support medical food services (**Figure 7)**. Newborn screening programs may use fees for more than one activity. Other activities funded by the NBS fee may include use for office supplies or printing, Phenylkenturia monitoring services, for the state program, biobank program or general fund budget.

Figure 7: Percent of programs that report using fees for specific NBS activities, September 2019 (n=46)

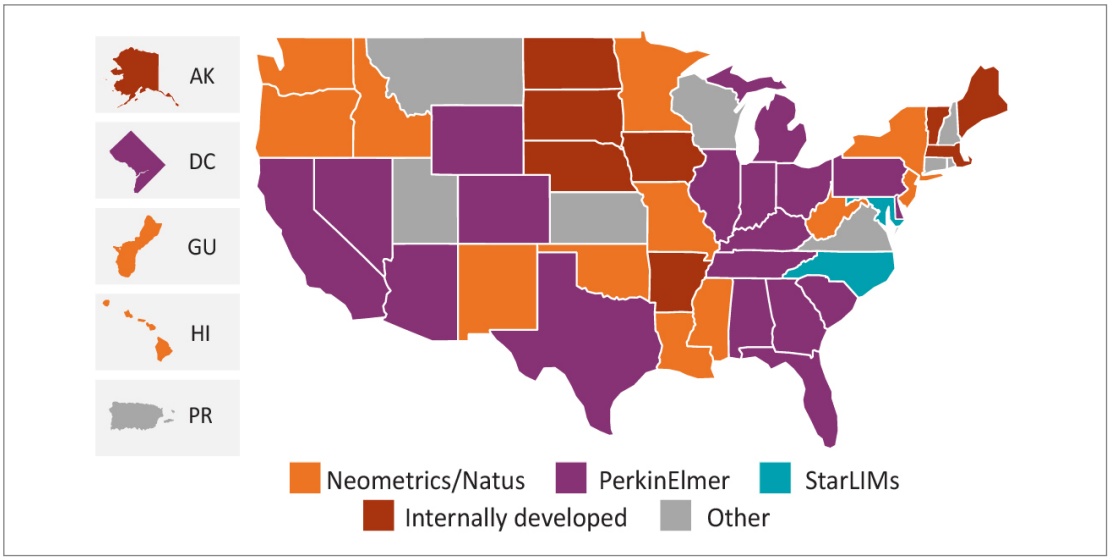


Information Management Systems

Each NBS program has information management systems within their laboratory and follow-up programs that are vital in storing, organizing and managing data generated by both the NBS laboratory and follow-up program.

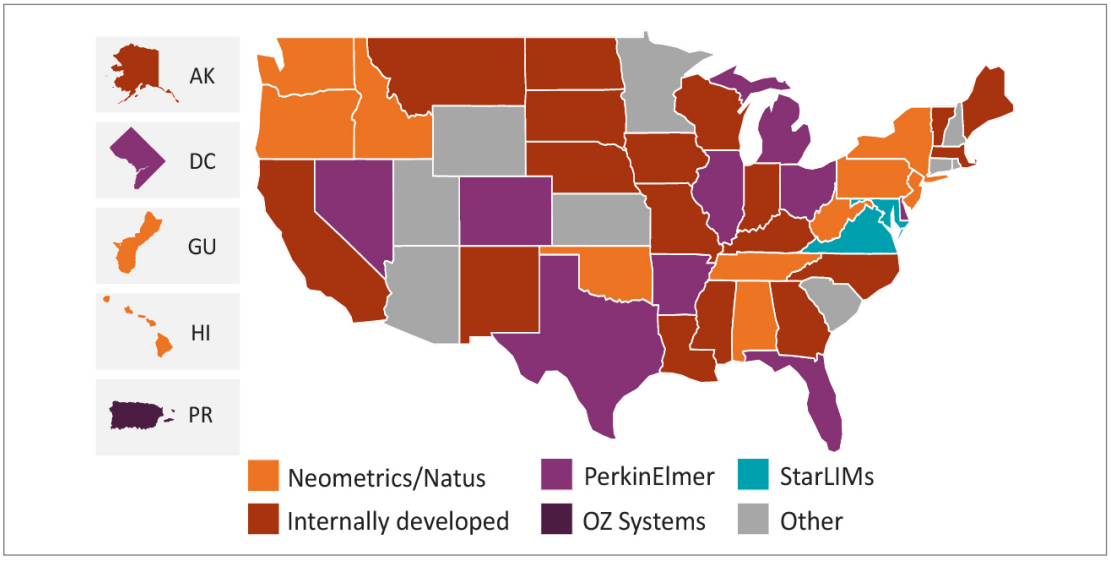
Laboratory information management systems (LIMS) vendors can be stratified into five categories: Neometrics/Natus (n=14), PerkinElmer (n=21), StarLIMS (n=3), internally developed/custom software (n=8), or Other (n=9) (Horizon, Orchard Harvest, Citrix, NeoMed, Labware and Epic Beaker) (**Figure 8**). Virginia reported using both StarLIMS and PerkinElmer.

Figure 8: Laboratory information management systems by state, September 2019 (N=53)



Similarly, case management information system (CMIS) vendors can be stratified into six categories: Neometrics/Natus (n=11), PerkinElmer (n=10), StarLIMS (n=2), Internally Developed (n=18), OZ Systems (n=1) and Other (n=11; KIDSNet, NeoMed, Labware, Citrix, Welligent Auris, Excel spreadsheet, custom written code, HiTrack) (**Figure 9**).

Figure 9: Case management information systems by state, September 2019 (N=53)



Health Information Technology in Newborn Screening

The use of electronic messaging in NBS facilitates more accurate data sharing and quicker results reporting. NBS programs are beginning to embrace health information technology (HIT) methods that promote timely submission of NBS orders and results reporting. Health Level 7 (HL7) is a widely used messaging standard to exchange electronic health data. As of September 2019, 23% (n=10/43) accept orders and send results from at least one submitter, 9% (n=4/43) accept orders but do not send results from at least one submitter, and 9% (n=4/43) send results but do not receive orders from at least one submitter. (**Figures 10** and **11**)

The use of HL7 order message acceptance and result reporting by at least one submitter in NBS programs has increased since 2012 (**Figure 12**).

NBS programs use web portals as another avenue for clients (e.g., birthing hospitals, clinicians) to access data. These web portals may be used in different capacities depending on programmatic needs and infrastructure. Out of the 46 programs that provided data, 65% (n=30) use a web portal in at least one capacity. Of these, 80% (n=24) use a web portal for sharing data related to the newborn screen, and 31% (n=9) use a web portal for DBS test orders and demographic data entry. Additionally, 86% (n=25) use a web portal for NBS results reporting. Twenty-four percent (n=7) of NBS programs use web portals in all three capacities (**Table 7**).

Figure 10: HL7 orders messaging status by state (HL7 orders submitted by at least one submitter), September 2019 (N=53)

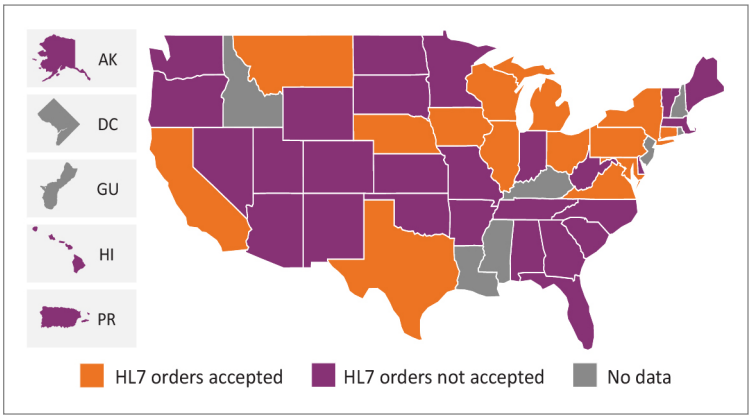


Figure 11: HL7 results messaging status by state (HL7 results received by at least one submitter), September 2019 (N=53)

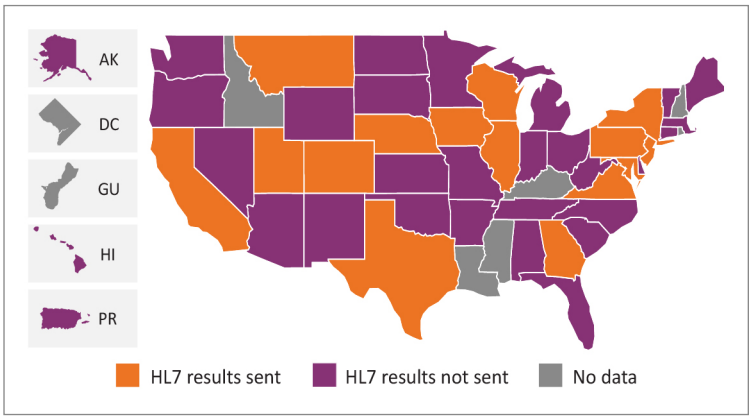


Figure 12: Comparison of NBS programs accepting HL7 orders and sending HL7 results (HL7 orders and results submitted and/or received by at least one submitter), September 2019

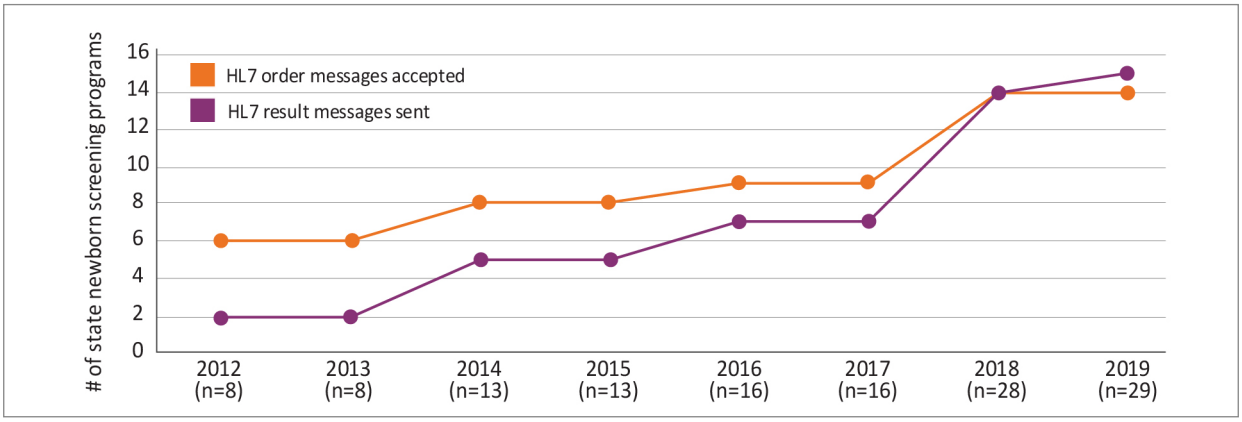


Table 7: NBS program web portal use, September 2019 (n=30)

State	Data entry portal present?	Data sharing portal present?	Result retrieval portal present?
Alabama		●	
Alaska		●	●
California			●
Colorado		●	●
Delaware		●	●
Florida	●	●	●
Georgia		●	●
Indiana		●	●
Iowa		●	●
Kentucky	●		
Louisiana		●	
Maryland	●	●	●
Michigan		●	
Minnesota		●	●
Missouri		●	●
Montana	●	●	●
Nebraska		●	●
Nevada			●
New Mexico		●	●
New York	●	●	
North Carolina	●	●	●
North Dakota		●	●
Ohio		●	●
Oklahoma			●
Oregon	●	●	●
Pennsylvania	●	●	●
South Dakota		●	●
Tennessee			●
Texas	●	●	●
Washington			●

Semantic data standards are used to standardize terminology/ vocabulary used by healthcare professionals when electronically exchanging patient health data. These universal code systems allow for the active use of standard electronic information exchanged between public health laboratories, birthing hospitals, and clinical care sites. This permits timely receipt and processing of NBS test orders by the NBS laboratories, leading to faster return of results and timely interpretation of results and medical intervention for the newborn. Out of the 19 programs that provided data, 94% (n=17) use Logical Observation Identifiers Names and Codes (LOINC), 53% (n=10) use Systemized Nomenclature of Medicine (SNOMED) and 42% (n=8) use local codes. Twenty-six percent (n=5) use all three semantic data standards (**Table 8**).

Table 8: NBS program standard code usage, September 2019 (n=19)

State	LOINC usage	SNOMED usage	Local code usage
California	●	●	●
Georgia			●
Illinois	●		
Iowa	●	●	●
Kentucky	●	●	
Maryland	●		
Minnesota	●	●	
New Jersey	●	●	
North Dakota	●	●	●
Ohio	●		
Oregon	●	●	
Pennsylvania	●		●
South Dakota	●	●	●
Tennessee	●		
Texas	●		
Utah	●		
Virginia	●	●	●
Washington	●	●	
Wisconsin			●

NEWBORN SCREENING SPECIMEN COLLECTION

Consent for Newborn Screening

Consent for newborn screening is implied in the majority of states (86%, n=46) and one program (Wyoming) indicated that parents must provide written consent for NBS. Additionally, NBS programs may have policies under which parents may opt out of NBS. Of the 52 programs that provided data, 52% (n=27) have opt-out policies based on religious beliefs, 10% (n=5) do not have an opt-out policy and 38% (n=20) have an "other" opt-out policy. The other responses were: opt-out if family education is provided coupled with a signed waiver, opt-out based on parental choice, opt-out for personal objection, opt out for any reason, opt-out with signed form, or opt-out with sworn declaration to the Department of Health.

Number of Screens

Each state has mandates to screen newborns and these mandates specify if newborns will receive one or two screens. Thirteen states (Alabama, Arizona, Colorado, Delaware, Idaho, Maryland, Nevada, New Mexico, Oregon, Texas, Utah, Washington, and Wyoming) are considered two-screen states because they require that a second dried blood spot specimen be routinely collected on all newborns regardless of the results of the first newborn screen. The purpose of the second screen is to improve the specificity and minimize missed cases (false-negatives) of conditions that are not detectable on the initial screen.[6] Newborns in the other 40 states and territories typically undergo a single newborn screen.

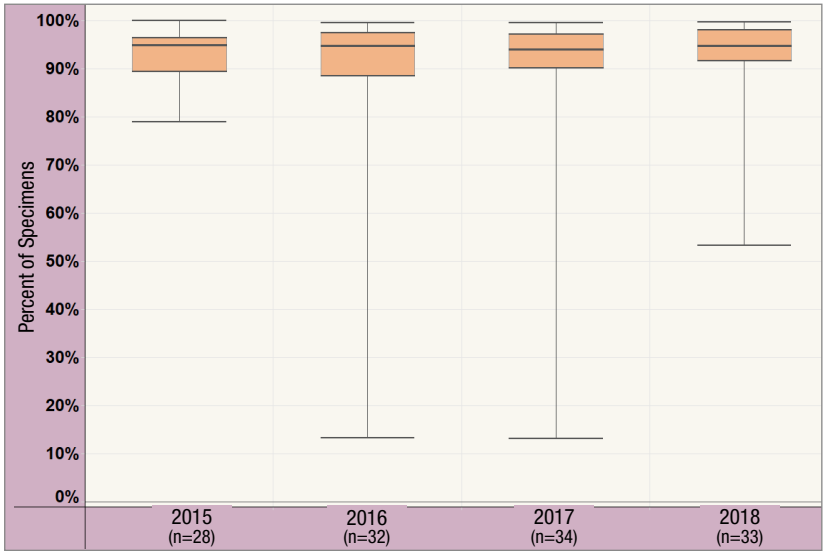
There are certain circumstances that may prompt an additional screen in one-screen states, such as when a specimen is collected too early or if there is an unsatisfactory specimen due to collection or transport errors. States and territories that require one screen (n=40) and those that require two screens (n=13) are provided in **Figure 1**.

Dried Blood Spot Specimen Collection

ACHDNC's NBS timeliness goals recommend that initial NBS specimens should be collected in the appropriate time frame for the newborn, but no later than 48 hours of life.[7] A majority of NBS programs have met this goal with 95% of NBS specimens collected no later than 48 hours of life, with a gradual improvement across the years **(Figure 13)**. By 2018, more than 90% of specimens were collected within 48 hours of life in 25 programs (76%, n=25/33) and more than 95% were collected within 48 hours in 16 programs (48%, n=16/33).

NBS laboratories flag unsatisfactory specimens for analysis and request a new specimen, which may result in delayed testing, as it requires additional time for laboratory personnel to acquire an acceptable specimen. The program median

Figure 13: Percent of first DBS specimens collected within 48 hours of birth, by year*



* NewSTEPs collects quality indicator data starting from 2012; however, due to few and disparate numbers of programs submitting data, this report focuses on data starting from 2015 as many programs did not have capability in their Laboratory Information Management Systems to capture this level of information prior to 2015.

6 Shapira, S. K., Hinton, C. F., Held, P. K., Jones, E., Harry Hannon, W., & Ojodu, J. (2015). Single newborn screen or routine second screening for primary congenital hypothyroidism. Molecular genetics and metabolism, 116(3), 125–132. doi:10.1016/j.ymgme.2015.08.003

7 Advisory Committee on Heritable Disorders in Newborns and Children Newborn Screening Timeliness Goals: https://www.hrsa.gov/advisory-committees/heritable-disorders/newborn-screening-timeliness.html

of unsatisfactory specimens has remained relatively consistent across the years (**Table 9**). Furthermore, DBS cards that are submitted without complete essential demographic information delay testing and reporting of results. The participating program median of reported specimens with missing essential information has also remained relatively consistent across the years (Table 9). One caveat of this data is that some NBS programs cannot separate first and subsequent samples, creating potential biases in reporting. Newborn screening programs have been actively working on educational initiatives to reduce unsatisfactory rates, including developing hospital report cards for NBS, one on one technical assistance and site visits.

Table 9: Unsatisfactory specimens and specimens without complete demographic information reported by year

	2015	2016	2017	2018
	Median % (n) Interquartile range			
Unsatisfactory specimens	1.6% (n=21) 1.0% - 2.1%	1.5% (n=32) 1.1% - 2.4%	1.5% (n=33) 0.9% - 2.4%	1.6% (n=32) 0.9% - 2.2%
Specimens missing essential information	2.0% (n=15) 0.3% - 8.7%	1.5% (25) 0.5% - 3.6%	2.2% (n=26) 0.5% - 4.1%	1.9% (n=25) 0.9% - 3.3%

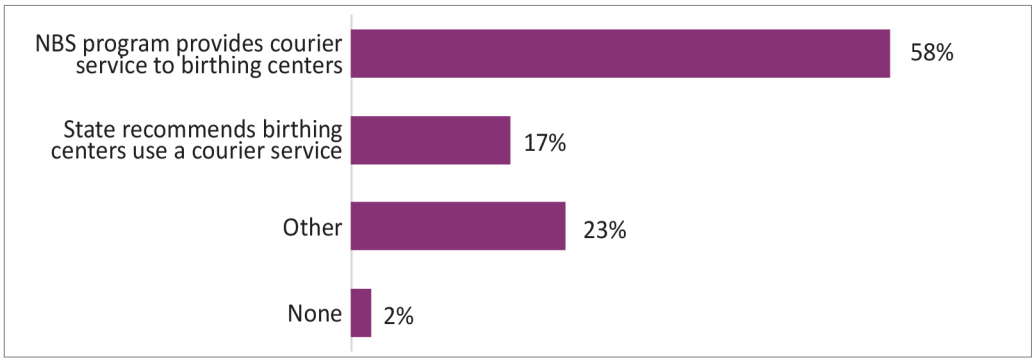
NEWBORN SCREENING SPECIMEN TRANSPORT

Courier Usage

The DBS specimen is transported to the NBS laboratory once it has been collected and adequately dried.[8] Courier usage status is defined in the NewSTEPs Data Repository as the method for transportation of specimens from birthing centers to the NBS laboratory for testing. Fifty-eight percent (n=31) of programs reported that the NBS program provides a courier service for birthing centers, 17% (n=9) reported that their state recommends birthing centers to use a courier service, 23% (n=12) have some other courier service usage, and 2% (n=1) reported no courier usage (**Figure 14**). States offer recommended couriers services, which hospitals can use as they see fit.

Twenty-five of the 53 NBS programs use UPS (n=11) and/or FedEx (n=9) to transport specimens from birthing centers to NBS laboratories, and four NBS programs use both. Eight NBS programs reported the use of a local courier and five use a regional courier. Only two NBS programs reported USPS as a service used to transport specimens (**Figure 15**). Local couriers are those particular to specific states or counties, whereas regional couriers may be shared amongst different states, and may operate across state lines.

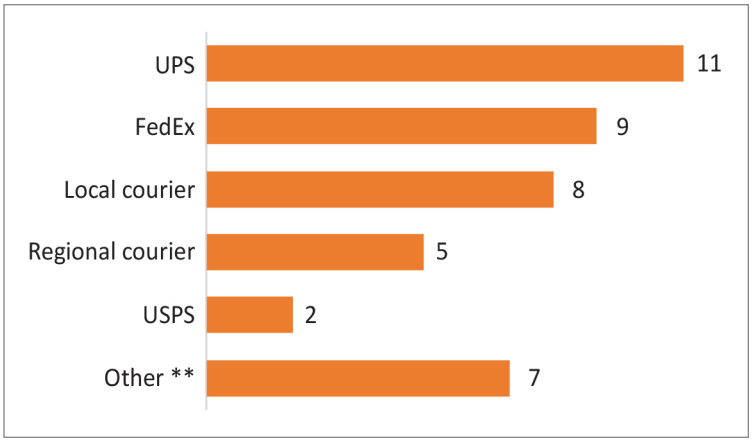
Figure 14: Courier usage status reported by NBS programs, September 2019 (N=53)



8 Clinical and Laboratory Standards Institute (CLSI). Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard—Sixth Edition. CLSI document NBS01-A6 (ISBN 1-56238-884-3). Clinical and Laboratory Standards Institute, 950 West Valley Road, Suite 2500, Wayne, Pennsylvania 19087-1898 USA, 2013).

Courier pickup and delivery vary by day of the week. Sixty-eight percent of NBS programs (36) have a weekend courier (Saturday, Sunday or both), 17% (n=9) have a Monday through Friday courier only, and 15% (8) did not provide data (**Table 10**). Additionally, 21 out of 45 NBS programs (47%) reported having a courier that operates on holidays. One caveat of this data is that it does not capture the percentage of birthing centers that benefit from a weekend or holiday courier in the state. For example, one program provides a weekend courier to 38 of 48 of their birthing facilities, and the remaining ten receive a courier Monday through Friday due to a small number of births at the birthing facilities. NewSTEPs collects this data at the state-level and these specific details are not apparent in aggregate data.

Figure 15: Type of services used* to transport specimens from birthing centers to the NBS laboratory, September 2019 (n=25)



* NBS programs were able to select multiple courier providers in the NewSTEPs Data Repository

**Other responses indicated specimens include: transported by air, or that hospitals or contracted laboratories are responsible for transporting specimens to the laboratory.

Courier pickup from birthing facilities does not equate to receipt of specimens at the NBS laboratory. For example, courier services may pick up specimens from a hospital on a Sunday but not deliver them to the NBS lab until Monday. In addition, it may take several days for the NBS laboratory to receive specimens from birthing facilities located in remote areas, or if screening is outsourced and requires specimens to be transported out of state. Seventy-five percent (n=40) of NBS programs receive specimens on weekends (Saturday, Sunday or both), 17% (9) receive specimens Monday through Friday only, and 8% (n=4) did not provide data (**Table 10**). Additionally, 19 out of 49 NBS programs (39%) reported specimen receipt on holidays.

Table 10: Number of days of courier pickup and delivery, and specimen receipt, September 2019 (N=53)

State	Courier pickup and delivery (per week)			Specimen receipt (per week)		
	Five days	Six days	Seven days	Five days	Six days	Seven days
Alabama	●			●		
Alaska			●			●
Arizona		●			●	
Arkansas	●				●	
California		●			●	
Colorado		●			●	
Connecticut						●
Delaware		●			●	
District of Columbia						
Florida			●		●	
Georgia	●			●		
Guam						
Hawaii		●		●		
Idaho	●				●	
Illinois		●			●	
Indiana		●			●	

State	Courier pickup and delivery (per week)			Specimen receipt (per week)		
	Five days	Six days	Seven days	Five days	Six days	Seven days
Iowa		●			●	
Kansas			●	●		
Kentucky		●			●	
Louisiana		●			●	
Maine			●			●
Maryland		●			●	
Massachusetts		●			●	
Michigan			●		●	
Minnesota		●			●	
Mississippi		●			●	
Missouri		●			●	
Montana			●		●	
Nebraska		●			●	
Nevada	●			●		
New Hampshire						
New Jersey		●			●	
New Mexico					●	
New York	●			●		
North Carolina		●			●	
North Dakota	●					●
Ohio			●		●	
Oklahoma			●			●
Oregon					●	
Pennsylvania		●			●	
Puerto Rico	●			●		
Rhode Island						
South Carolina		●			●	
South Dakota			●			●
Tennessee			●		●	
Texas			●		●	
Utah		●			●	
Vermont		●			●	
Virginia			●			●
Washington					●	
West Virginia	●			●		
Wisconsin		●		●		
Wyoming		●			●	

Receipt of Specimens at the NBS Laboratory

NBS laboratories have varying definitions of specimen receipt at the laboratory. This definition is of value when calculating the transit time of the DBS from the birthing hospital/submitter. Of the 37 NBS programs that completed this field in the NewSTEPs Data Repository, 32 defined specimen receipt as manually or electronically recorded by laboratory staff (86%), one by courier drop-off (3%), one by when testing is initiated (3%) and three by "other" (8%), which include specific designated receipt times by the laboratory (**Figure 16**).

Additionally, NBS programs provided information on how they record when a specimen is received at the NBS laboratory. Of the 37 programs that responded, the most common method was date and time stamp (**Figure 17**). "Other" methods included when specimens are accessioned for testing, a date stamp with batched time stamp, or recording of entry in the LIMS or other electronic registry.

Figure 16: NBS program definitions of specimen receipt at NBS laboratory, September 2019 (n=37)

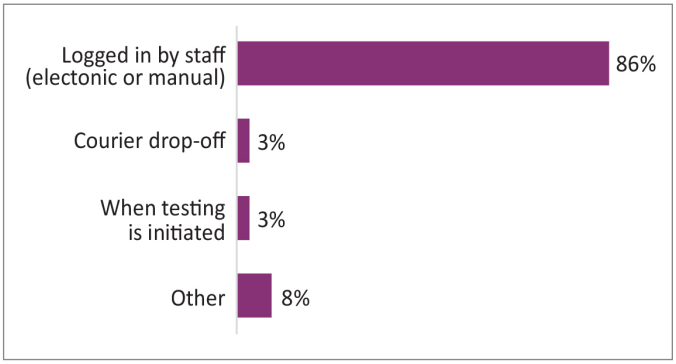
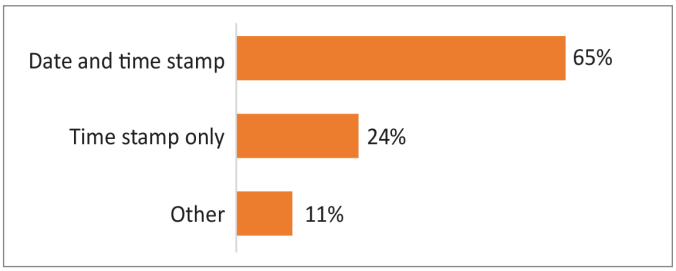


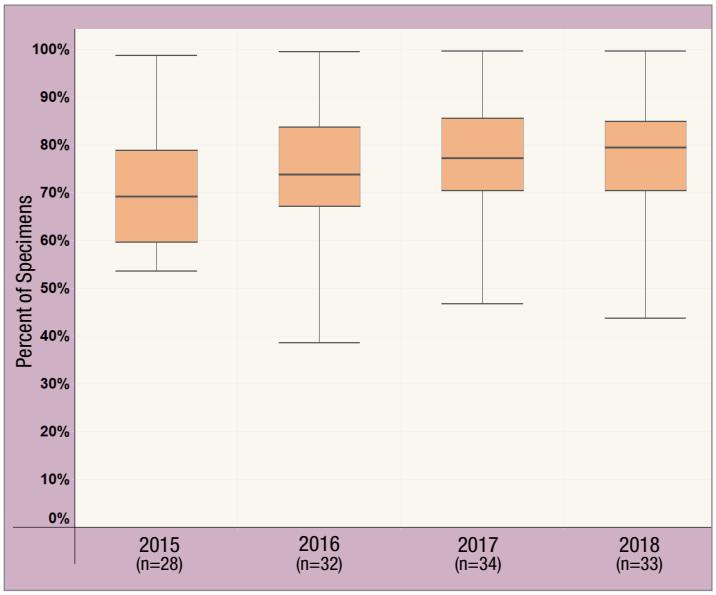
Figure 17: Method of recording specimen receipt, September 2019 (n=37)



Timeliness of Specimen Receipt

Most NBS programs are still working to achieve the ACHDNC specimen delivery goal. In 2018, only one of the 33 programs (3%) received at least 95% of specimens within two calendar days, and for 15 NBS programs, more than 80% of specimens were received within two calendar days (45%, n=15/33). NBS programs have successfully improved utilization of courier systems demonstrated by a stepwise increase of timely specimen receipt in each year. The program median for the time from first specimen collection to receipt at the NBS laboratory on the next calendar day increased from 28% in 2015 to 41% in 2018. Allowing two calendar days after collection to receipt, the program median increased from 69% in 2015, to 74% in 2016, to 77% in 2017, and to 80% in 2018 (**Figure 18**). The best potential for timeliness gains are increasing the number of days of courier operations as well as increasing the number of days that NBS laboratories are open to accept specimens.

Figure 18: Percent of first dried blood spot specimens received within two days after specimen collection, by year



NEWBORN SCREENING PROGRAM PROCESSES

Newborn Screening Program Operating Hours And Activities

The timely implementation of NBS activities and providing timely NBS results is critical for the early identification and treatment of affected infants. Expanding laboratory and follow-up operating hours and activities is one method NBS programs are taking to improve timeliness. Starting in 2015, the Missouri NBS program received additional funding from the legislature that allowed for the addition of a Sunday courier and to add eight more birthing facilities to pickup routes. By expanding the courier to operate one day before the laboratory meant that weekend newborns were able to be screened on Monday instead of Tuesday. Additional state NBS program case narratives are including in the NewSTEPs 360 Toolkit for Expanding Newborn Screening Services.[9]

Newborn screening laboratories range in hours and days of operation. Sixteen (30%) NBS programs have laboratories that are open five days a week, 26 (49%) are open six days a week, and 11 (21%) are open seven days a week (**Figure 19**). Additionally, 20 (38%) reported that their NBS laboratories are open on holidays.

Various NBS laboratory activities are performed on weekends and/or holidays. Reporting time-critical results was the most frequently reported laboratory activity on weekends (n=33) and holidays (n=24) followed by testing for time-critical disorders on weekends (n=32) and holidays (n=23). Both molecular testing and reporting non-time-critical results are the two least frequent NBS laboratory activities performed on weekends (n=20 and 15, respectively) and holidays (n=11). Overall, NBS programs perform fewer laboratory activities on holidays compared to weekends (**Figure 20**).

Figure 19: Days per week NBS laboratory is open, September 2019 (N=53)

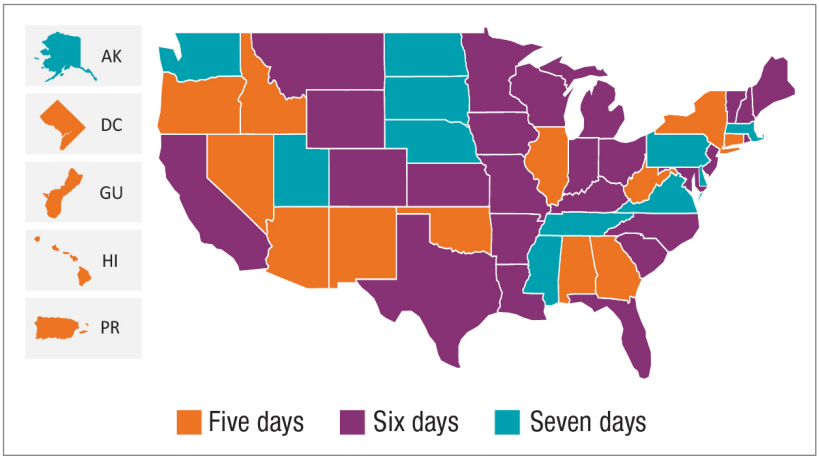
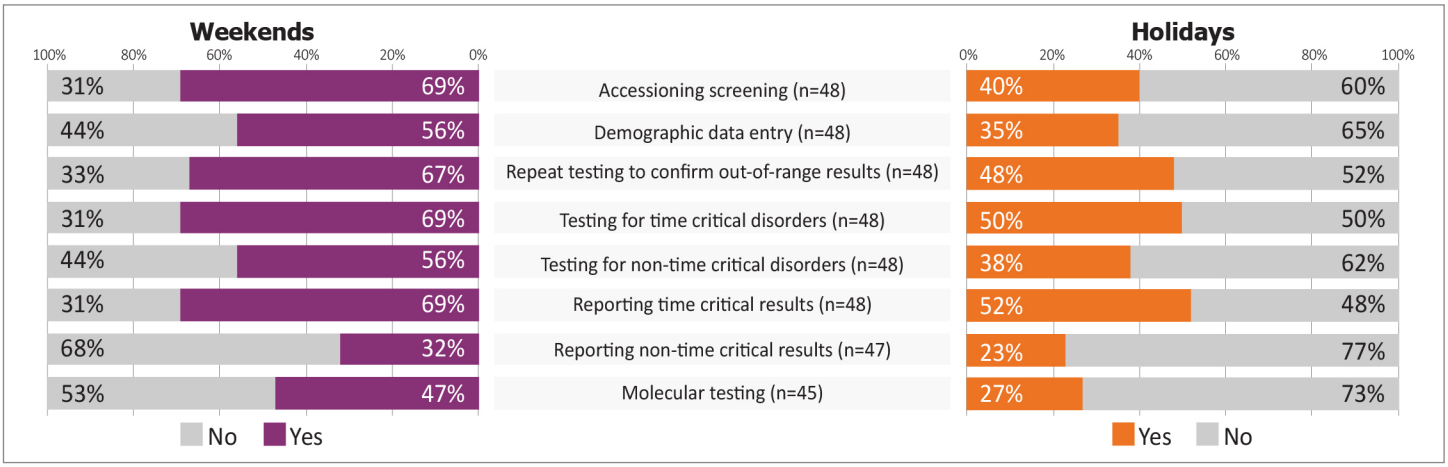


Figure 20: NBS programs performing laboratory activities on weekends and holidays, September 2019

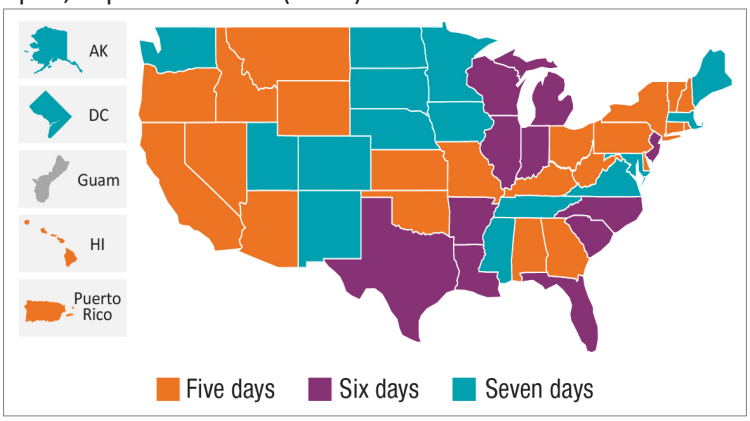


9 Timeliness Toolkit for Expanding Newborn Screening Services: https://www.newsteps.org/toolkits/timeliness-toolkit-expanding-newborn-screening-services

In the NBS follow-up program, 26 (49%) perform follow-up services five days per week, 10 (19%) six days per week, and 16 (31%) perform follow-up services seven days per week (**Figure 21**). Furthermore, 15 (29%) follow-up programs are open on holidays, while 37 (71%) programs do not have follow-up services on holidays.

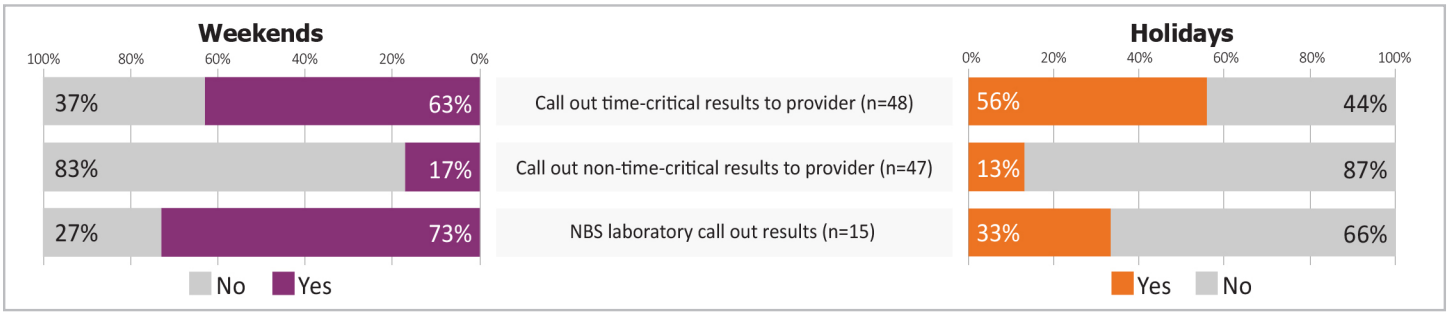
Sixty-three percent (30/48) call out time-critical results to the provider on weekends and 56% (27/48) call out critical results on holidays. Further, 17% (n=8/47) call out non-time-critical results to providers on weekends compared to 13% (n=6/47) that call out non-time-critical results on holidays. Lastly, 73% (n=11/15) stated that their NBS laboratory is responsible for calling out results on weekends and 33% (n=5/15) reported that their laboratories call out results on holidays (**Figure 22**).

Figure 21: Number of days per week NBS follow-up program is open, September 2019 (N=53)*



* Guam did not provide data

Figure 22: NBS programs performing follow-up activities on weekends and holidays, September 2019



SHORT- AND LONG-TERM FOLLOW-UP

Once the NBS laboratory completes testing of the specimen, the NBS follow-up program receives the results (in most NBS programs). Depending on the nature of the results, the NBS follow-up program (or NBS laboratory) shares the results with the primary care physician of record, parent, specialist and/or referral center. NewSTEPs collects definitions used to best describe short-term follow-up (STFU) among NBS programs, along with a description of long-term follow-up (LTFU) activities, if any (**Table 11**). NBS programs indicated that short-term follow-up is defined as: until confirmatory testing is performed (n=2), other (n=7), until the infant is on treatment (n=11), or until diagnosis is made or ruled out (n=33). Twenty-nine programs said that LTFU activities exist for their NBS program. NBS programs also provided information on how long they follow-up with inconclusive diagnoses. Responses included six months (n=3), one year (n=5), other (n=7) or until diagnosis is made or ruled out (n=33).

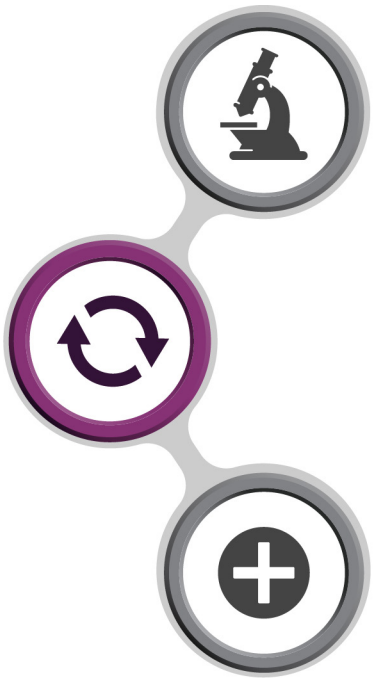


Table 11: Follow-up definitions and activities by NBS programs (N=53), September 2019

NBS program	Short-term follow-up definition	Follow-up period for inconclusive diagnosis	Long-term follow-up activities exist?
Alabama	Until the infant is on treatment	Until diagnosis is made/ruled out	●
Alaska	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	
Arizona	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	
Arkansas	Until the infant is on treatment	Until diagnosis is made/ruled out	●
California	Until diagnosis is made/ruled out and on treatment if needed	Until diagnosis is made/ruled out	●
Colorado	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	
Connecticut	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●
Delaware	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●
District of Columbia	Until diagnosis is made/ruled out		
Florida	Until diagnosis is made/ruled out	6 months	
Georgia	Until the infant is on treatment	Until diagnosis is made/ruled out	
Guam	Until diagnosis is made/ruled out		●
Hawaii	Until the infant is on treatment	Until diagnosis is made/ruled out	●
Idaho	Until the infant is on treatment	Until diagnosis is made/ruled out	
Illinois	Until diagnosis is made/ruled out	6 months	●
Indiana	Until confirmatory testing is performed	Up to 3 years	●
Iowa	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●
Kansas	Until confirmatory testing is performed	1 year	
Kentucky	Until diagnosis is made/ruled out	Laboratory reports to primary care physician and calls that a repeat is needed; 10 days later a letter is sent to parent; 10 days later a certified letter sent to parent; then close as lost to follow-up 10 days later if no response	
Louisiana	Until diagnosis is made/ruled out and/or infant is under treatment	On a case-by-case basis; do an administrative review and make the decision to close based on information. Most of the time information is found on babies via Women Infant and Children (WIC) program	●
Maine	Until the infant is on treatment	Until diagnosis is made/ruled out	●
Maryland	Until diagnosis is made/ruled out		●
Massachusetts	Until the infant is on treatment	Until diagnosis is made/ruled out	●
Michigan	Until diagnosis is made/ruled out		●
Minnesota	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●

NBS program	Short-term follow-up definition	Follow-up period for inconclusive diagnosis	Long-term follow-up activities exist?
Mississippi	Until diagnosis is made/ruled out	1 year	●
Missouri	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●
Montana	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	
Nebraska	Until diagnosis is made/ruled out and if treatment is indicated when that starts	Until diagnosis is made/ruled out	●
Nevada	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●
New Hampshire	Until the infant is on treatment	Until diagnosis is made/ruled out	
New Jersey	Until diagnosis is made/ruled out or 3 months, whichever comes first	Until diagnosis is made/ruled out OR 1 year, whichever comes first	●
New Mexico	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●
New York	Until confirmatory testing is performed and/or Until diagnosis is made/ruled out depending on the condition	There is a diagnosis in the system of "possible" disease, which allows the program to close cases to short-term follow-up	●
North Carolina	Until confirmatory testing is nor-mal or infant has seen a specialist and is on treatment	Until diagnosis is made/ruled out	●
North Dakota	Until diagnosis is made/ruled out	On a case-by-case basis	●
Ohio	Until diagnosis is made/ruled out	1 year	
Oklahoma	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●
Oregon	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●
Pennsylvania	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	
Puerto Rico	Until confirmatory testing is per-formed and the patient is referred to the corresponding specialist	Until diagnosis is made/ruled out	
Rhode Island	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	
South Carolina	Until diagnosis is made/ruled out	Do not have this category of inconclusive diagnosis- follow until ruled out	
South Dakota	Until diagnosis is made/ruled out	1 year	
Tennessee	Until diagnosis is made/ruled out		
Texas	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	●
Utah	Until diagnosis is made/ruled out	1 year	
Vermont	Until the infant is on treatment	Until diagnosis is made/ruled out	
Washington	Until the infant is on treatment	Until diagnosis is made/ruled out	●
West Virginia	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	
Wisconsin	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	
Wyoming	Until diagnosis is made/ruled out	Until diagnosis is made/ruled out	

Number of Cases Identified Through Newborn Screening

NewSTEPs collects aggregate confirmed case data for infants diagnosed with a disorder identified through NBS each year. **Table 12** reflects data reports for 2015, 2016 and 2017. The table does not include CCHD or Hearing loss cases and includes data from 51 US Newborn Screening Programs, unless otherwise indicated.

Table 12: Number of cases on the Recommended Uniform Screening Panel identified by newborn screening, 2015-2017

Disorder	Year		
	2015	2016	2017
3-Hydroxy-3-methyglutaric aciduria - HMG	<5	<5	<5
3-Methylcrotonyl-CoA carboxylase deficiency - 3-MCC	99	94	105
Argininosuccinic aciduria - ASA	16	21	22
Beta-Ketothiolase deficiency - BKT	<5	<5	6
Biotinidase deficiency - BIOT	183	144	157
Carnitine uptake defect/carnitine transport defect - CUD	53	42	49
Citrullinemia, type I - CIT	20	30	26
Classic galactosemia - GALT	98	76	78
Classic PKU & Hyperphe	241	228	229
Congenital adrenal hyperplasia - CAH	272	272	282
Congenital hypothyroidism – CH*	2,224	2,252	2,179
Cystic fibrosis – CF*	755	692	682
Glutaric acidemia type I - GA1	31	29	44
Holocarboxylase synthase deficiency - MCD	<5	None	<5
Homocystinuria - HCY	6	5	7
Isovaleric acidemia - IVA	31	28	25
Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency - LCHAD	12	8	7
Malonic acidemia - MAL	<5	<5	None
Maple syrup urine disease - MSUD	23	19	22
Medium-chain acyl-CoA dehydrogenase deficiency - MCAD	253	231	206
Methylmalonic acidemia (cobalamin disorders) - Cbl A,B	17	12	14
Methylmalonic acidemia (methylmalonyl-CoA mutase) - MUT	10	<5	8
Mucopolysaccharidosis I - MPS I**	<5	<5	6
Pompe**	14	17	31
Presence of Hb S	1,957	1,836	2,026
Propionic acidemia - PROP	19	21	23
Severe Combined Immunodeficiencies - SCID	71	78	71
Trifunctional protein deficiency - TFP	<5	<5	<5
Tyrosinemia, type I - TYR I	6	5	11
Very long-chain acyl-CoA dehydrogenase deficiency - VLCAD	79	66	61
X-linked Adrenoleukodystrophy**	5	17	61
Total	**6,507**	**6,237**	**6,543**

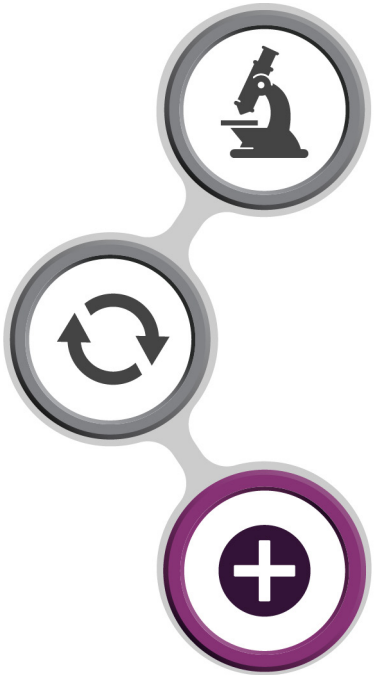
* Data is from 50 US NBS programs
** Data is only from those NBS programs universally screening for Pompe, MPS I or X-linked Adrenoleukodystrophy

Timeliness Data For Cases With A Confirmed Diagnosis

The 40 participating programs reported 9,528 cases with a confirmed diagnosis of a disorder identified by newborn screening for the years 2015-2017. (**Table 13**).

NBS programs are meeting the national recommended time frames as described below:

- Median time from birth to specimen collection in all three years is within the recommended time frame of 24–48 hours.
- Median report time for time-critical disorders was five days in all three years, and is within the recommended time frame of reporting results in five days of life.



A Wilcoxon signed-rank test showed that the there is a statistical decrease in the mean days from birth to medical intervention for time-critical disorders, between 2015 and 2017 ($p = 0.0002$). This metric illustrates that NBS programs are improving processing and reporting results out in an efficient timely manner. Medical intervention is defined as when care of the infant changed (i.e., the earliest point at which a clinical action was rendered based on follow-up on the newborn screening results and is inclusive of date therapy was initiated or a decision was made to defer therapy based on current presentation). Medical intervention may occur before a diagnosis is determined and is therefore a critical step in ensuring the newborn is under medical supervision as soon as possible. NewSTEPs revised the definitions for medical intervention to be specific for each disorder. This may have introduced a bias in that all states may not have been reporting in a standardized way.

Table 13: Timeliness metrics for newborns identified with a disorder on the newborn screening panel by year, September 2019; Median (Interquartile Range)

	2015			2016			2017		
	All	**Time-critical**	**Non-time-critical**	**All**	**Time-critical**	**Non-time-critical**	**All**	**Time-critical**	**Non-time-critical**
Total Number	3,318	478	2,840	3,022	428	2,594	3,188	438	2,750
Collection (hours)	29 (25-39)	29 (25-39)	29 (25-39)	28 (24-38)	31 (24-31)	28 (24-38)	29 (25-40)	31 (25-44)	29 (24-40)
Receipt at lab (days from birth)	3 (3-4)	3 (2-5)	3 (3-4)	3 (3-4)	3 (3-4)	3 (3-4)	3 (3-4)	3 (3-4)	3 (3-4)
Result release (days from birth)	6 (4-7)	5 (3-6)	6 (4-7)	5 (4-7)	5 (3-7)	6 (4-7)	5 (4-7)	5 (3-6)	6 (4-7)
Intervention (days from birth)	14 (7-31)	6 (4-11)	17 (8-36)	13 (7-29)	6 (4-11)	15 (8-33)	11 (6-28)	5 (4-7)	14 (7-30)
Diagnosis (days from birth)	23 (9-50)	14 (6-41.5)	24 (10-52)	22 (10-47)	13 (6-30)	24 (11-50)	20 (9-43)	13 (7-25)	22 (9-46)

SUMMARY

As NBS celebrates over 55 years since its inception in the US, all programs screen for at least 30 disorders on the core RUSP as of September 2019. Approximately four million newborns each year are screened to determine risk of developing NBS disorders, enabling receipt of timely medical intervention.

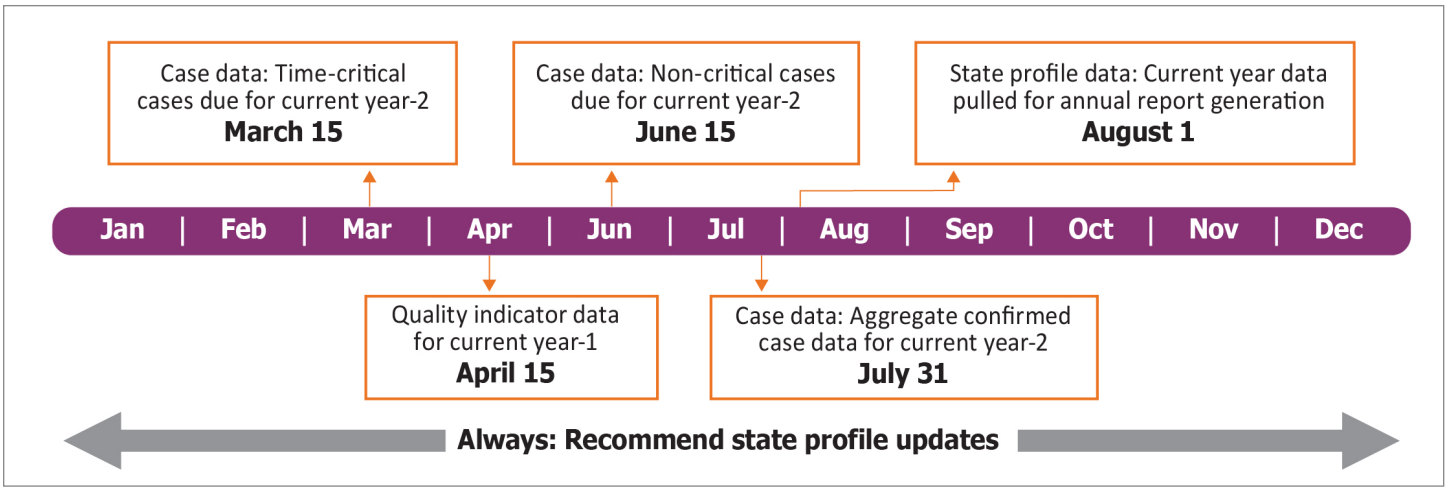
This report illustrates that NBS programs are continuously working on quality improvement by:

- Expanding their screening panels to implement the most recent NBS disorders on the RUSP
- Expanding courier services to improve time of delivery of DBS to NBS laboratories
- Expanding operating hours of NBS programs to improve laboratory processing times and timely follow-up
- Prioritizing activities for those time-critical disorders (testing, reporting out results) during weekends and holidays, and
- Utilizing standard electronic data exchange methods for ordering tests and reporting results for timely medical intervention.
- Improving timeliness of specimen collection. For example, more than 90% of specimens were collected within 48 hours of life in 25 out of 33 programs, and more than 95% were collected within 48 hours in 16 out of 33 programs.

NewSTEPs in collaboration with NBS programs will continue to collect and analyze data, and report on their activities to identify capabilities and capacities, and increase NBS visibility. NewSTEPs will continue to cultivate collaborative relationships and facilitate information exchange among the broader NBS community.

APPENDIX A: DATA COLLECTION TIMELINE

All data is collected in accordance with the data entry timeline displayed in the figure below.



NBS programs are encouraged to provide:

- NBS programs are encouraged to update this information for the current year by September 1 of the current year.
- Annual quality indicator data for the current year-1 by April 15 of the current year. For example, 2018 quality indicators were submitted by April 15, 2019.
- Time-critical case data for the current year – 2 by March 15 of the current year and non-time-critical case data for the current year -2 by June 15, of the current year. For example, for 2017 time-critical and non-time-critical case data were submitted by March 15, 2019 and June 15, 2019 respectively.
- Aggregate confirmed case data for current year – 2 by July 31 of the current year.

APPENDIX B: NEWBORN SCREENING PROGRAMS AND DATA COLLECTION METHODS

Data Collection

There are 53 newborn screening (NBS) programs in the US, consisting of all 50 states, the District of Columbia, Puerto Rico, and Guam. NewSTEPs collaborates with each NBS program to improve the quality of their program through a variety of activities including reviewing each program's data in the NewSTEPs Data Repository. The NewSTEPs Data Repository is a centralized and secure database that can be accessed by authorized users from anywhere. It allows each NBS program to explore data to meet local evaluation needs.

Both the Colorado Institutional Review Board (COMIRB) and HHS' Office of Human Research Protection ("OHRP") have deemed the data collected in the NewSTEPs Data Repository Non-Human Subject Research. Each NBS program is required to enter into a Memorandum of Understanding (MOU) in order to submit quality indicator data and case data. The MOU includes information around data ownership, data reporting, and data security. It establishes the framework in which the NBS program will share elements of its NBS data with NewSTEPs and identifies each party's roles and responsibilities. Newborn screening programs that enter data into the NewSTEPs Data Repository have access to their own data as well as aggregate data from other participating NBS programs who have an MOU with APHL. As of September 2019, 48 states have an MOU with NewSTEPs. The NewSTEPs Data Repository collects three levels of data:

State Profiles: Publicly available data that describes the NBS program and its activities. State profile data encompasses the following: an overview of the NBS program, such as annual births, number of required screens and responsible laboratory; disorders screened, including method, method's target and equipment used; policies in place, such as opt out policies, consent policies and courier service usage; processes for adding to the NBS panel; fees, such as funding sources and fee use details; program structure; contacts; advisory committee data; information technology (IT) support data; and health information technology (HIT) elements.

Quality Indicators: Eight performance metrics utilized to provide longitudinal comparisons within an NBS program, as well as comparisons to aggregate data across programs. These quality indicators have undergone careful, iterative evaluation by stakeholders to assure agreement on definitions. A quality indicator source document was developed that outlines purpose, definitions and general considerations.[10] Quality indicator data is secure and only accessible to authorized users. Quality indicators are as follows:

1. Percent of dried blood spot specimens that were unacceptable due to improper collection and/ or transport
2. Percent of dried blood spot specimens with at least one missing state-defined essential data field upon receipt at the laboratory
3. Percent of eligible newborns not receiving a newborn screen, reported by dried blood spot or point of care screen(s)
4. Percent of infants that have no recorded final resolution (confirmed diagnosis or diagnosis ruled out by an appropriate medical professional) with the newborn screening program
5. Timeliness of newborn screening activities
6. Percent of infants with an out-of-range newborn screen result requiring clinical diagnostic workup by an appropriate medical professional, reported by disorder category
7. Percent of disorders detected by newborn screening with a confirmed diagnosis by an appropriate medical professional
8. Percent of missed cases, reported by disorder

Confirmed Cases: Infant level data, including demographics and diagnostic criteria to facilitate common classifications for diagnoses across programs for all of the core newborn screening disorders. Case data is secure and only accessible to authorized users.

10 NewSTEPs Quality Indicator Source Document: https://www.newsteps.org/sites/default/files/quality-indicators/quality_indicator_source_document_july_17_2018_se.pdf

Data collection follows a data collection timeline each year as described in Appendix A. NewSTEPs has established formal processes for parties and individuals to request data from the NewSTEPs Data Repository. Each data sharing request is directed to the Data Review Workgroup, which is charged with providing expertise to make recommendations to NewSTEPs staff and the NewSTEPs Steering Committee on any requests made for data collected within the NewSTEPs Data Repository.

Challenges and Solutions to Data Collection

Barriers to data entry that NewSTEPs staff have become aware of include inability to differentiate between specimen level and case level data, lack of dedicated personnel to enter data, lack of expertise to query information management systems, no incorporation of NewSTEPs data entry into general workflow and lack of prioritization of NewSTEPs data entry in comparison to other laboratory activities (i.e. onboarding screening of new disorders).

To address these barriers, NewSTEPs continues to engage with NBS programs and encourages data submission for all of the data categories collected. NewSTEPs has provided:

- Customized technical assistance to access, collate, upload, analyze and interpret data
- A Quality Indicator Source Document is available online that defines each QI, provides a glossary of terms; quick tips, Laboratory Information Management Systems hints, calculation examples and scenarios
- Extract, Transform, Load (ETL) pilot projects that facilitate automated data extraction and transformation (calculations as needed), and uploads into the NewSTEPs Data Repository
- Data request page available on the NewSTEPs Website with form to request data (vetted through NewSTEPs' Data Review Workgroup) to further incentivize entry
- Interactive data visualizations utilizing data pulled from the Data Repository via secure Tableau sign-ins
- Regular reminders of data entry timeline; targeted and repeated outreach via phone and email coinciding with data entry timeline
- Customized tutorials of the NewSTEPs Website and Data Repository elements with states who have signed an MOU, with new staff or upon request
- Reports of frequently asked questions
- Import templates (CSV files) to facilitate the automation of data submission
- Engagement of Information Management Vendors (IMS) vendors
- State Administrator and General User Guides available on the Data Repository that include detailed information about data entry timelines and user permissions.

ACRONYM GLOSSARY

ACHDNC	Advisory Committee on Heritable Disorders in Newborns and Children
APHL	Association of Public Health Laboratories
CCHD	Critical Congenital Heart Disease
CDC	US Centers for Disease Control and Prevention
COOP	Continuity of Operations Plan
DBS	Dried Blood Spot
DC	District of Columbia
EHDI	Early Hearing Detection and Intervention
FIMS	Follow-Up Information Management System
GU	Guam
HHS	US Department of Health and Human Services
HL7	Health Level 7
HRSA	Health Resources and Services Administration
IT	Information Technology
LIMS	Laboratory Information Management System
LOINC	Logical Observation Identifiers Names and Codes
LTFU	Long-Term Follow-Up
MPS I	Mucopolysaccharidosis Type I
MOU	Memorandum of Understanding
NBS	Newborn Screening
NewSTEPs	Newborn Screening Technical assistance and Evaluation Program
PR	Puerto Rico
RUSP	Recommended Uniform Screening Panel
SCID	Severe Combined Immunodeficiency
SMA	Spinal Muscular Atrophy
SNOMED	Systemized Nomenclature of Medicine
STFU	Short-Term Follow-Up
X-ALD	X-linked Adrenoleukodystrophy

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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.

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