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December 2016

NEWBORN SCREENING TIMELINESS

Most States Had Not Met Screening Goals, but Some Are Developing Strategies to Address Barriers

GAO-17-196

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GAO Highlights

Highlights of GAO-17-196, a report to congressional committees

Why GAO Did This Study

Each year, over 12,000 newborns are born with heritable or other conditions that require early detection and treatment. Newborn screening is a state public health activity, and includes the collection of a blood specimen from the newborn, specimen arrival at a state's lab, and results reporting. Barriers at any stage of this process can lead to delays in treatment and potential harm to the newborn. The Newborn Screening Saves Lives Reauthorization Act of 2014 included improving timeliness as an explicit goal for HRSA-supported newborn screening programs, which include technical assistance for and data collection from participating states.

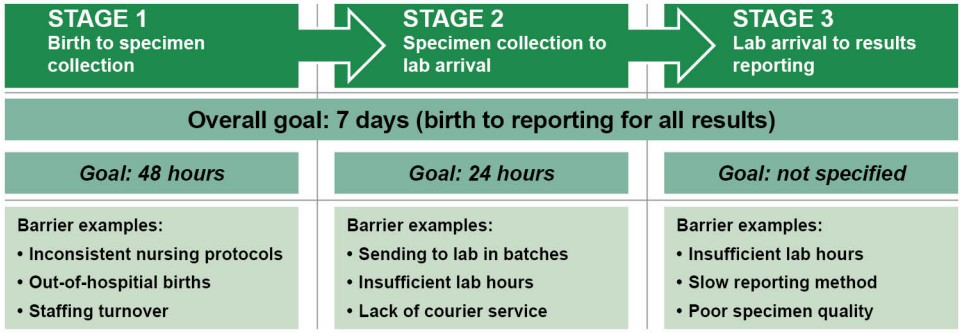
The act included a provision for GAO to review newborn screening timeliness. This report examines (1) what is known about the timeliness of newborn screening for heritable conditions; and (2) barriers identified as contributing to screening delays, and strategies used to address them. GAO reviewed time-frame goals from the advisory committee, an August 2016 report from NewSTEPs with an analysis of annual timeliness data from states for 2012 through 2015 (the most recently available data), and a 2014 report on a survey conducted for the advisory committee. GAO also reviewed relevant documents and interviewed officials from NewSTEPs, two advisory committee members who worked on timeliness issues, and newborn screening officials in four states selected because they were focusing on activities related to newborn screening timeliness.

View GAO-17-196. For more information, contact Marcia Crosse at (202) 512-7114 or crossem@gao.gov

What GAO Found

Most states that reported timeliness data had not screened newborns within recommended goals to detect conditions that may require treatment. The Department of Health and Human Services' (HHS) Advisory Committee on Heritable Disorders in Newborns and Children recommended time-frame goals in 2015 for newborn screening, such as reporting all results within 7 days of birth. Data provided by 38 states for 2012-2015 showed that states generally had not met the committee's suggested benchmark of meeting each time-frame goal for at least 95 percent of specimens, which the committee encouraged states to achieve by 2017. Missing data and variations in data collection limit a full understanding of timeliness trends, but HHS's Health Resources and Services Administration (HRSA) has supported activities to address these challenges. HRSA supports the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), which collects newborn screening data. NewSTEPs has been taking steps to improve data for future analysis, such as by clarifying data definitions and working with states to help ensure they use these definitions when submitting timeliness data.

Advisory Committee's Newborn Screening Time-Frame Goals and Barriers Identified by States



Sources: GAO analysis of information from the Advisory Committee on Heritable Disorders in Newborns and Children and selected states. | GAO-17-196

State newborn screening officials identified numerous barriers to timely newborn screening, and a variety of strategies to address them. Newborn screening officials who responded to the advisory committee's 2014 survey identified barriers, such as a lack of understanding of the importance of timely screening for out-of-hospital births, limited courier availability to transport specimens to a lab, and insufficient lab hours. Selected state newborn screening officials interviewed by GAO reported developing various strategies to address these barriers. For example, one state increased courier service so rural hospitals located far from the state's lab could shorten specimen transport time. HRSA has been providing states with technical assistance, but it is too soon to determine which strategies developed through this technical assistance, if any, will have a measurable impact on timeliness.

In commenting on a draft of this report, HHS generally agreed with the report's findings, but questioned the use of 2017 benchmark goals to measure performance and the exclusion of two conditions. GAO believes its use of the 2017 benchmark and scope were appropriate, as discussed in the report.

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Abbreviations

APHL	Association of Public Health Laboratories
HHS	Department of Health and Human Services
HL7	Health Level 7
HRSA	Health Resources and Services Administration
LIMS	laboratory information management system
MOU	memorandum of understanding
NewSTEPs	Newborn Screening Technical assistance and Evaluation Program
NICU	neonatal intensive care unit
RUSP	Recommended Uniform Screening Panel

U.S. GOVERNMENT ACCOUNTABILITY OFFICE
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December 15, 2016

The Honorable Lamar Alexander
Chairman
The Honorable Patty Murray
Ranking Member
Committee on Health, Education, Labor, and Pensions
United States Senate

The Honorable Fred Upton
Chairman
The Honorable Frank Pallone, Jr.
Ranking Member
Committee on Energy and Commerce
House of Representatives

Each year, more than 12,000 newborns are born with heritable or other conditions that require early detection and treatment to prevent serious illness or death. Early detection, diagnosis, and treatment of these conditions may prevent a child's death, serious illness, or disability. For example, newborns with galactosemia cannot properly digest a certain type of sugar and must avoid milk products to prevent organ damage or death. Newborns with maple syrup urine disease cannot properly break down certain proteins that can build up in the blood to toxic levels; if untreated, these newborns are at risk for brain damage, coma, or death.

Virtually all newborns in the United States (about 4 million each year) are screened for heritable and other conditions, but barriers experienced in any stage of the newborn screening process can lead to delays in reporting results and beginning needed treatment. Newborn screening involves three stages. First, after birth, a health care provider collects a blood specimen from the newborn on a card.[1] Next, this specimen is sent to a state lab for testing. Lastly, the results are reported to the newborn's provider. Newborn screening is a state public health activity, with each state responsible for designing and implementing its own newborn screening system.[2] As a result, states may experience varying barriers to

[1]In this report, the term provider refers to medical professionals such as physicians, nurses, and midwives.

[2]In this report, the term states refers to all 50 states, the District of Columbia, Guam, and Puerto Rico.

timely screening—such as problems with timely preparation of specimens for shipment to the lab—and use different strategies to address such barriers.

While largely a state public health activity, the federal government has a role in newborn screening. For example, the Children's Health Act of 2000 authorized the Department of Health and Human Services (HHS) to award grants to improve the ability of states to provide newborn screening for heritable conditions.[3] In addition, Congress created a clearinghouse for newborn screening information under the Newborn Screening Saves Lives Act of 2007.[4] Time-frame goals for completing newborn screening were initially identified in a 2005 report prepared for HHS's Advisory Committee on Heritable Disorders in Newborns and Children.[5] Subsequently, following news reports in late 2013 and an advisory committee survey of states in early 2014, concerns were raised that many states were not performing newborn screening in a timely manner. Soon after, the Newborn Screening Saves Lives Reauthorization Act of 2014 was enacted, incorporating timeliness improvement as an explicit goal for newborn screening efforts supported by HHS's Health Resources and Services Administration (HRSA).[6] These efforts involve newborn screening education, data collection, technical assistance, and other activities. For example, HRSA supports the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs), which involves the collection of newborn screening data from states on a voluntary basis through a data repository. In 2015, the advisory committee identified new time-frame goals and NewSTEPs updated the data repository to allow for states to submit timeliness data on the new goals.

The Newborn Screening Saves Lives Reauthorization Act of 2014 included a provision for GAO to report on the timeliness of newborn screening for heritable conditions. In this report, we examine

[3]Pub. L. No. 106-310, § 2601, 114 Stat. 1101, 1164 (2000) (codified, as amended, at 42 U.S.C. § 300b-8).

[4]Pub. L. No. 110-204 § 5, 122 Stat.705, 708-709 (2008) (codified, as amended, at 42 U.S.C. § 300b-11).

[5]The advisory committee provides information and recommendations about newborn screening to the Secretary of Health and Human Services.

[6]Pub. L. No. 113-240, § 2, 128 Stat. 2851, 2852 (2014) (codified at 42 U.S.C. § 300b-8(a)(5)).

1. what is known about the timeliness of newborn screening for heritable conditions; and
2. barriers that have been identified as contributing to delays in newborn screening for heritable conditions, and strategies being used to address them.

To examine what is known about the timeliness of newborn screening for heritable conditions, we reviewed state-reported data on newborn screening, including the percentages of specimens screened within time-frame goals recommended by the advisory committee in 2015, from an August 2016 report prepared by NewSTEPs, a program administered by the Association of Public Health Laboratories (APHL) in partnership with the University of Colorado's School of Public Health. For its report, NewSTEPs requested data from all 53 states for 2012 through 2015, and 38 states submitted annual timeliness data that were included in the report.[7] This included 20 of the states that had signed a memorandum of understanding (MOU) with APHL to enter data into the data repository under NewSTEPs and 18 other states that submitted timeliness data using an equivalent spreadsheet. We also reviewed documents provided by NewSTEPs, and interviewed officials from APHL and the University of Colorado's School of Public Health regarding the data repository, including methods for entering data into the repository, and regarding the NewSTEPs August 2016 report.

In addition, we reviewed the advisory committee's April 2015 letter to the Secretary of Health and Human Services that included the committee's time-frame goals and the benchmark that states should meet each of these time-frame goals for at least 95 percent of specimens by 2017.[8] We also interviewed APHL and University of Colorado School of Public Health officials about any plans to track newborn screening timeliness in the future. We interviewed newborn screening stakeholders identified by APHL officials and on HRSA's website to learn about efforts to track newborn screening timeliness. These stakeholders included a member of the advisory committee who co-chaired a timeliness workgroup, a

[7]Although the time-frame goals were not in place for the entire period of time captured in the NewSTEPs report (2012 through 2015), the data provide useful information about the states' past performance relative to these goals. Not all of the 38 states reported data for all years or all stages of the newborn screening process.

[8]For the April 2015 letter, see http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendations/timelynewbornscreeninggoalschainletter.pdf (accessed March 1, 2016).

member of the advisory committee who conducted research related to newborn screening barriers, and officials from associations (including the Association of Maternal & Child Health Programs, Association of State and Territorial Health Officials, Genetic Alliance, and March of Dimes).

We assessed the reliability of the annual timeliness data in the NewSTEPs August 2016 report by, for example, reviewing whether the 38 states that submitted data used common data definitions and whether there were mechanisms in place to mitigate data entry errors (such as automatic calculations of percentages), and determined that they were sufficiently reliable for our purposes. These timeliness data are not generalizable to other states, but provided valuable insight on what is known about newborn screening timeliness in the reporting states.

To examine barriers identified as contributing to delays in newborn screening for heritable conditions, and strategies being used to address them, we reviewed documents, including reports and presentations on newborn screening timeliness. Specifically, we reviewed the advisory committee's 2014 Newborn Screening Timeliness Survey Report, which identified barriers to and strategies for timely newborn screening based on survey results from newborn screening officials in 51 states.[9] From the survey report, we selected barriers identified by respondents as having a major or moderate impact on timeliness; barriers that were frequently reported by survey respondents in written responses; and barriers in the survey that were also highlighted by a number of state newborn screening officials in published presentations and reports. Through a combination of written responses and interviews, we then collected more detailed information about these barriers and strategies developed to address them from officials in four selected states: Arizona, Colorado, Minnesota, and Wisconsin. We selected these states because they were focusing on activities related to newborn screening timeliness, which would allow them to provide more detailed information on a number of barriers and strategies. The information from these states is not generalizable to other states, but provided information on strategies used to address identified barriers.

[9]This report was prepared by APHL for the advisory committee as part of a cooperative agreement with HRSA to administer NewSTEPs. Not all states responded to all questions. We assessed the reliability of information in the survey report by comparing it to related documents and interviewing stakeholders, and we determined the information was sufficiently reliable for our purposes. For more information about the survey, see https://www.aphl.org/AboutAPHL/publications/Documents/NBSTimelinessSuveyReport_10-2014.pdf (accessed April 28, 2016).

We reviewed documents provided by APHL officials, including the NewSTEPs August 2016 report, which contained information on steps some states had undertaken to improve timeliness. We interviewed officials from APHL and the University of Colorado's School of Public Health to learn about plans to share information from NewSTEPs 360, which is a program administered by these organizations to provide technical assistance focused on newborn screening timeliness to states through grants. We also interviewed these officials to clarify or elaborate on barriers to timely newborn screening and strategies to address them.[10] In addition, we reviewed documents from newborn screening stakeholders and interviewed them to elaborate on information about barriers to timely newborn screening and strategies to address such barriers. (See app. I for more information about our scope and methodology.)

We conducted this performance audit from January 2016 to December 2016 in accordance with generally accepted government auditing standards. Those standards require that we plan and perform the audit to obtain sufficient, appropriate evidence to provide a reasonable basis for our findings and conclusions based on our audit objectives. We believe that the evidence obtained provides a reasonable basis for our findings and conclusions based on our audit objectives.

Background

Newborn Screening Overview

Newborn screening for heritable and other conditions begins with a provider collecting a blood specimen from a newborn within a few days of birth.[11] The newborn's heel is pricked to obtain a few drops of blood, which are placed and dried on a specimen collection card, and then sent to a state lab for testing. (See fig. 1 for an example of a collection card.) State departments of health may use their own lab to test newborn

[10]Documents we reviewed to understand states' experiences with barriers to newborn screening timeliness included reports from an initiative called the Collaborative Improvement and Innovation Network. APHL administered this initiative as part of NewSTEPs to help improve newborn screening timeliness in eight states. The initiative involved identifying root causes of poor timeliness, creating goals and objectives to guide states' efforts, and developing and sharing strategies to improve timeliness.

[11]Some newborn screening may involve point-of-care testing instead of blood specimen testing. This report focuses on timeliness of newborn screening using a blood specimen.

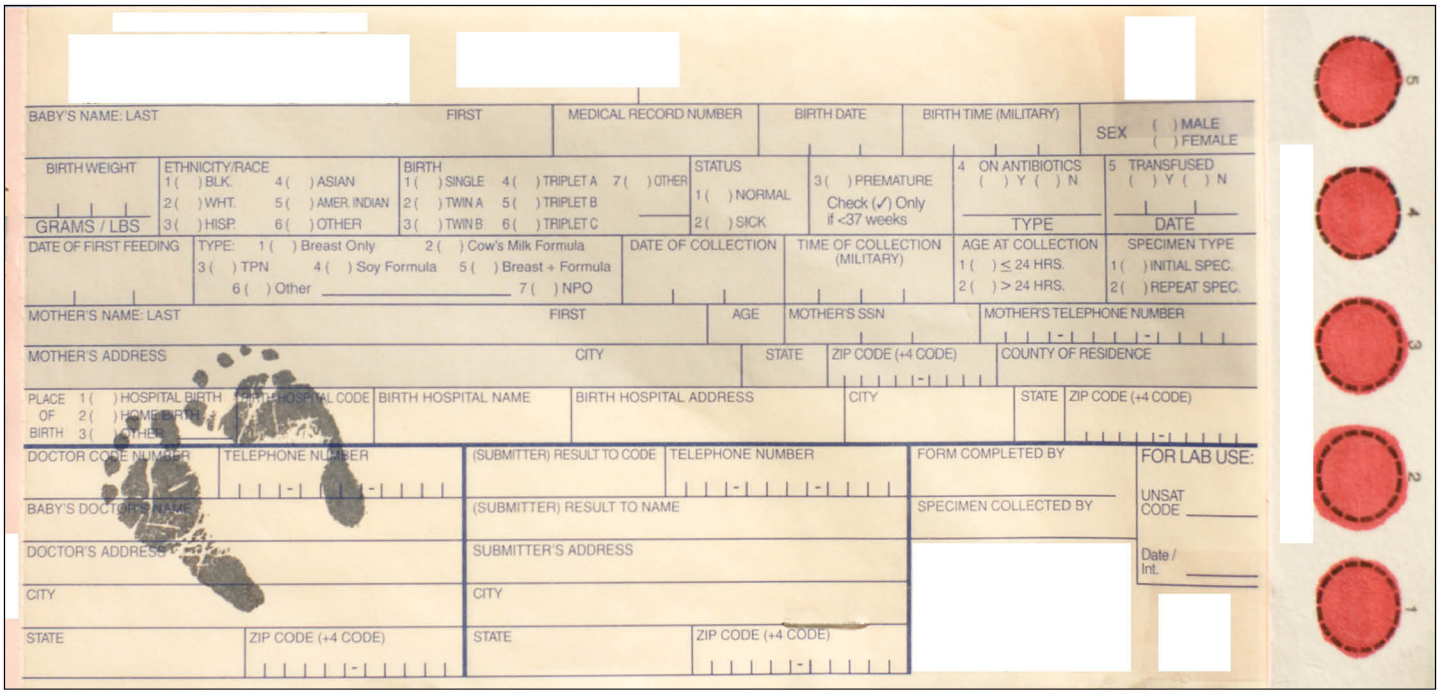
screening specimens or may contract with a private lab, a lab at a university medical school, or another state's lab.[12] After testing, lab staff notify providers of either normal results or presumptive positive results, which indicate that a newborn may have a heritable condition, subject to follow-up testing to determine if the condition is truly present.[13] Lab staff may report presumptive positive results to providers by, for example, fax or phone call before sending all normal and presumptive positive results.[14]

[12]In this report, the term state lab refers to a lab designated by a state to test newborn screening specimens.

[13]Presumptive positive results are positive screening results from an initial round of testing; they do not confirm that a newborn has a heritable or other condition. Presumptive positive results indicate that a newborn may be at risk for a heritable or other condition so that definitive follow-up testing can be offered to determine if the condition is truly present. For example, following a presumptive positive result from newborn screening for phenylketonuria, additional tests will be performed to determine whether the newborn has the condition, or a milder condition, which does not require treatment.

[14]Newborn screening may also include activities conducted after results reporting, such as screening a second specimen and patient follow-up; these additional activities are outside the scope of this report.

Figure 1: Example of a Blood Specimen Card Used for Newborn Screening



Source: © The Association of Public Health Laboratories. | GAO-17-196

Note: This figure shows an example of one state's newborn screening specimen card, with identifying information removed.

The newborn screening process involves collaboration between providers and other hospital staff, lab staff, and state newborn screening officials. Providers and other hospital staff are responsible for ensuring that newborn screening specimens are collected and sent to the state lab for testing. Lab staff and follow-up staff, such as nurses and social workers, are responsible for entering demographic data associated with the specimen into the state's laboratory information management system (LIMS), testing the newborn screening specimen, and reporting results to providers. Newborn screening officials at state departments of health support providers and labs with education, data, and resources.

Advisory Committee's Time-Frame Goals

HHS's Advisory Committee on Heritable Disorders in Newborns and Children, which was chartered to recommend newborn screening improvements in states and provide technical information and advice about newborn screening to the Secretary of Health and Human Services, established a Recommended Uniform Screening Panel (RUSP), which is

a list of conditions for which newborns should be screened.[15] A 2005 report prepared for the advisory committee to make recommendations for the RUSP also identified time-frame goals for individual stages of the newborn screening process, such as from specimen collection to arrival at the lab, for the conditions on the RUSP.[16] Subsequently, in response to a public comment during a committee meeting in September 2013, the advisory committee took additional steps to address newborn screening timeliness concerns:

- In 2014, the advisory committee designated 16 of 32 conditions on the RUSP as "time-critical" conditions. These are conditions for which acute symptoms or potentially irreversible damage could develop in the first week of life, and for which early recognition and treatment can reduce the risk of illness and death.[17]
- Also in 2014, the advisory committee, in conjunction with APHL, conducted a survey and issued its 2014 Newborn Screening Timeliness Survey Report, which included information on barriers to and strategies for newborn screening timeliness identified by newborn screening officials in 51 states.

[15]Two conditions on the RUSP—critical congenital heart disease and hearing loss—involve point-of-care testing rather than blood specimen testing. According to HRSA officials, these two conditions, as well as primary congenital hypothyroidism (which involves blood specimen testing), are not strictly considered heritable conditions, but require early screening. See appendix II for more information about conditions on the RUSP.

[16]In 2001, HRSA contracted with the American College of Medical Genetics to convene an expert group to review available information on newborn screening and to make recommendations based on an analysis of scientific evidence in order to strengthen newborn screening in states. The report on this work, which recommended a minimum uniform panel of conditions for screening in states, also included the following time-frame goals: (1) all newborn screening results from first specimens should be available for the first post-hospital discharge visit within 5 days of specimen collection; (2) most results should be available within 2 days of a specimen arriving at a lab; and (3) specimens should arrive at a lab within 3 days of specimen collection. For more information about this report, see http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/uniformscreening.pdf, page 93 (accessed February 8, 2016). In September 2005, the advisory committee communicated its endorsement of the report and recommended that the Secretary of Health and Human Services initiate action to facilitate adoption of the panel of conditions by every state.

[17]The advisory committee adopted the definition of time-critical conditions from the Society of Inherited Metabolic Conditions.

- In April 2015, the advisory committee sent a letter to the Secretary of Health and Human Services with new time-frame goals.[18] For example, the 2015 letter included time-frame goals for the full newborn screening process (from birth to results reporting) rather than from specimen collection to results reporting. Additionally, the 2015 letter added different time-frame goals for time-critical and non-time-critical conditions, and shortened the time-frame goal for a specimen arriving at the lab from 3 days after collection to 24 hours after collection.

The advisory committee's 2015 time-frame goals included recommended time frames for completing the full newborn screening process—that is, from birth to results reporting:

- All newborn screening results should be reported for all conditions to a provider as soon as possible, but no later than 7 days after birth.
- Presumptive positive results for time-critical conditions should be reported immediately to a provider, but no later than 5 days after birth.
- Presumptive positive results for all non-time-critical conditions should be reported to a provider as soon as possible, but no later than 7 days after birth.

The advisory committee's 2015 time-frame goals also include time frames for the first two newborn screening stages (from birth to specimen collection and from specimen collection to lab arrival) to help states achieve the goals for the full process; the committee did not identify a time-frame goal for the third newborn screening stage (lab arrival to results reporting):

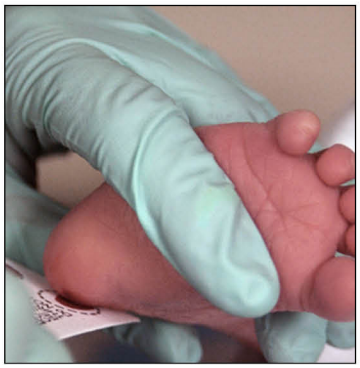
1. Newborn screening specimens should be collected in the appropriate time frame for the newborn's condition, but no more than 48 hours after birth.
2. Newborn screening specimens should arrive at the lab as soon as possible; ideally within 24 hours of collection.

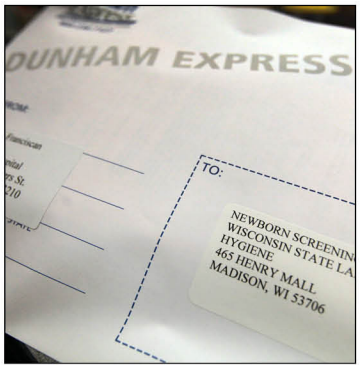
Finally, the advisory committee encouraged states to benchmark progress by meeting each of these time-frame goals for at least 95

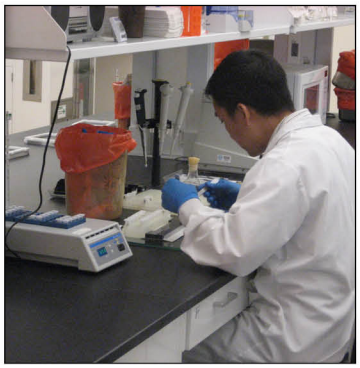
[18]These time-frame goals apply to newborn screening using blood specimen tests; they do not apply to the two conditions (critical congenital heart disease and hearing loss) on the RUSP that involve point-of-care testing.

percent of specimens by 2017. (See fig. 2 for more information about the 2015 time-frame goals.)

Figure 2: Activities Included in the Newborn Screening Process and Related Time-Frame Goals Set by the Advisory Committee on Heritable Disorders for Newborns and Children in 2015







	STAGE 1 Birth to specimen collection	STAGE 2 Specimen collection to lab arrival	STAGE 3 Lab arrival to results reporting
Newborn screening activities	• Birth date and time recorded on newborn screening card • Specimen collected (blood drawn from the newborn via heel prick and placed on newborn screening card) • *Specimen collection time recorded on newborn screening card (beginning of stage 2)*	• *Specimen collection time recorded on newborn screening card* • Specimen dries for at least 3 hours • Specimen prepared for shipping • Specimen picked up by mail or courier and transported to state lab • *Specimen lab arrival date and time recorded (beginning of stage 3)*	• *Specimen lab arrival date and time recorded* • Demographic data entered into lab's information management system • Specimen tested for conditions • Report presumptive positive results or normal results by, for example, phone, fax, mail, or lab's website
Time-frame goals	• Time-critical conditions: 5 days (birth to reporting of presumptive positive results) • Non-time-critical conditions: 7 days (birth to reporting of presumptive positive results) • All conditions: 7 days (birth to reporting of presumptive positive and normal results)		
	• 48 hours (birth to collection)	• 24 hours (collection to lab arrival)	• Not specified

Sources: GAO analysis of information from the Association of Public Health Laboratories and Advisory Committee on Heritable Disorders in Newborns and Children; (left to right) Air Force, © Milwaukee Journal Sentinel, and © the Association of Public Health Laboratories (images). | GAO-17-196

Notes: The time-frame goals shown in this figure were recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children in April 2015. Newborn screening may include additional activities that take place after stage 3, such as screening for a second specimen and patient follow-up. The activities and time-frame goals shown in this figure do not apply to newborn screening for conditions that involve point-of-care testing, such as hearing loss.

HRSA's Newborn Screening Efforts

HRSA's Maternal and Child Health Bureau has responsibility for enhancing, improving, and expanding the ability of states to provide newborn screening.[19] HRSA oversees a number of programs that provide resources to improve newborn screening quality and increase newborn screening education. Following the enactment of the Newborn Screening Saves Lives Reauthorization Act of 2014, some of these programs focused on newborn screening timeliness.

One of these programs is NewSTEPs, which is administered by APHL under a cooperative agreement.[20] NewSTEPs began in 2012 to offer a forum for collaboration among state newborn screening officials and other stakeholders; to facilitate continuous quality improvement and data-driven outcome assessments through a data repository; and to create a national newborn screening technical assistance center that provides training, addresses challenges, and supports program improvement through partnerships with newborn screening stakeholders. In 2013, NewSTEPs launched its data repository to collect annual newborn screening data from participating states. To participate in the data repository, states must sign an MOU with APHL; 35 states had signed an MOU as of November 20, 2016, according to HHS.

[19]Other HHS agencies conduct work related to newborn screening. The Centers for Disease Control and Prevention helps state labs implement testing for new conditions and ensure the quality of their newborn screening tests through a quality assurance program; the Centers for Medicare & Medicaid Services regulates lab testing performed on human specimens, including newborn screening specimens, and administers Medicaid, which covers newborn screening for eligible infants; the Food and Drug Administration reviews and clears the newborn screening cards used for specimen collection, clears or approves newborn screening diagnostic tests, and also approves safe and effective treatments for heritable conditions; and the National Institutes of Health has sponsored research on conditions identified through newborn screening.

[20]NewSTEPs was authorized by the Newborn Screening Saves Lives Act of 2007. See Pub. L. No. 110-204 § 2, 122 Stat.705, 705-706 (2008) (codified, as amended, at 42 U.S.C. § 300b-8). In June 2012, HRSA entered into a cooperative agreement with APHL to administer NewSTEPs through June 2018. NewSTEPs is a collaborative effort between APHL and the University of Colorado's School of Public Health, with both organizations operating in partnership. In this report, references to NewSTEPs represent the combined work of both organizations.

In response to requirements in the Newborn Screening Saves Lives Reauthorization Act of 2014 for HRSA to support timely newborn screening, NewSTEPs updated the data repository to collect timeliness data from participating states that are consistent with the advisory committee's 2015 time-frame goals. For example, a state's data in the repository include the percentage of specimens for which all results for all conditions were reported within the advisory committee's goal of 7 days after birth. NewSTEPs can use each state's reported percentage for a given time-frame goal to monitor the state's progress toward meeting the advisory committee's 95 percent benchmark in a given year—that is, whether screening was completed within a time-frame goal (e.g., 7 days) for 95 percent of a state's specimens. Most of the states with signed MOUs began entering timeliness data into the data repository in mid-2016.

In addition to incorporating timeliness data in NewSTEPs' data repository, HRSA oversees NewSTEPs 360, a program that provides technical assistance and collects monthly data on newborn screening timeliness through grants to states. Administered by the University of Colorado's School of Public Health, in collaboration with APHL under a cooperative agreement with HRSA, NewSTEPs 360 aims to improve timeliness in newborn screening by providing quality improvement training.[21] For example, according to officials involved in administering the program, NewSTEPs 360 holds monthly quality improvement coaching calls intended to help each participating state develop innovative strategies that focus on timeliness barriers. In addition, participating states enter

[21]NewSTEPs 360 was authorized by the Newborn Screening Saves Lives Reauthorization Act of 2007, as amended by the Newborn Screening Saves Lives Reauthorization Act of 2014. See Pub. L. No. 113-204 § 2, 122 Stat.705, 705-706 (2008), as amended by Pub. L. No. 113-240, § 2, 128 Stat. 2851, 2851-2852 (2014) (codified at 42 U.S.C. § 200b-8). In June 2015, HRSA entered into a cooperative agreement with the University of Colorado's School of Public Health to administer NewSTEPs 360 through August 2018. NewSTEPs 360 is a collaborative effort between the University of Colorado's School of Public Health and APHL, with both organizations operating in partnership. In this report, references to NewSTEPs 360 represent the combined work of both organizations.

monthly timeliness data into the data repository.[22] Twenty-eight states were participating in this program, as of October 26, 2016.[23]

Most States Had Not Met the Advisory Committee's Benchmark for Newborn Screening Timeliness, but Data Challenges Limit a Full Understanding of Timeliness Trends

Most states that reported timeliness data to NewSTEPs had not met the advisory committee's 95 percent benchmark for newborn screening timeliness. Missing data for several states and variations in data collection limit a full understanding of newborn screening timeliness trends, but HRSA has been taking steps to address these challenges.

Most States that Reported Timeliness Data Had Not Met the Advisory Committee's Benchmark, but Improved Over Time

Most states that reported 2015 timeliness data (the most recent data available) to NewSTEPs had not met the advisory committee's 95 percent benchmark for newborn screening timeliness for all conditions within 7 days. However, timeliness for completing this screening process improved over time for the majority of states.

Timeliness Data for Reporting All Results for All Conditions

Most states reporting 2015 timeliness data to NewSTEPs, which collects annual newborn screening data from states, had not met the advisory committee's 95 percent benchmark for completing the full newborn screening process (stages 1 through 3) for all conditions within 7 days. According to the advisory committee's benchmark, by 2017, states should report newborn screening results for all conditions within 7 days of birth, for at least 95 percent of specimens. In 2015, 5 of the 27 states reporting

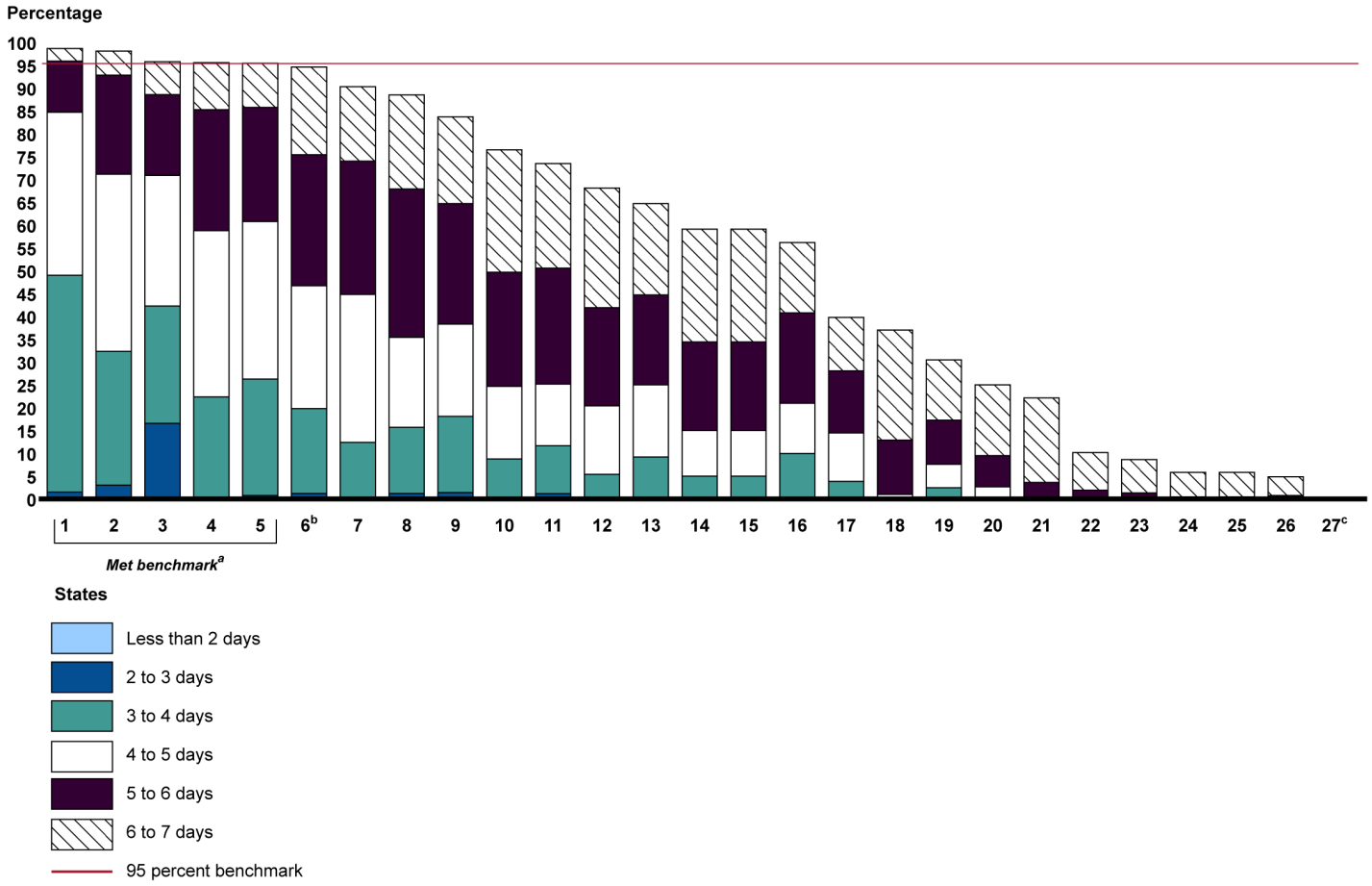
[22]These monthly timeliness data are based on the same measures used by states entering annual timeliness data into the data repository under NewSTEPs.

[23]HRSA reported that in addition to NewSTEPs and NewSTEPs 360, the agency provided funding to the Association of State and Territorial Health Officials in June 2014 to support the development of policy and programmatic approaches to improve the quality of newborn screening in six states. All six states worked on newborn screening timeliness as a part of this work, which was completed in January 2015.

timeliness data for this measure met this 95 percent benchmark. (See fig. 3.)

Figure 3: States' Percentages of Specimens for Which Results Were Reported to Providers within 7 Days of Birth, 2015 (All Newborn Screening Results for All Conditions)

In 2015, 5 of 27 states met the 95 percent benchmark for reporting all newborn screening results for all conditions within 7 days, and 1 state was within 1 percentage point of the benchmark. The Advisory Committee on Heritable Disorders for Newborns and Children encouraged states to achieve the 95 percent benchmark by 2017.



Source: GAO analysis of data from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

Notes: This figure shows the percentages of first specimens screened (birth to reporting of presumptive positive and normal results for time-critical and non-time-critical conditions) within various time categories in each of the 27 states that provided data for this time-frame goal. NewSTEPs rounded percentages to the nearest tenth of a percentage to determine the number of states that met the 95 percent benchmark. The 7-day time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in April 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

[a]These five states met the 95 percent benchmark (95.38 to 98.59 percent).

[b]This state did not meet the benchmark, but was within 1 percentage point (94.54 percent).

[c]This state reported that less than 1 percent of all newborn screening results were reported within 7 days of birth.

States' timeliness for completing the full newborn screening process for all conditions improved over time. The number of states meeting the benchmark was higher in 2015 compared to the previous 3 years. Likewise, the median percentage of specimens screened within 7 days was higher in 2015 than in the previous 3 years. (See table 1.) According to NewSTEPs, there were 21 states that demonstrated improvement from 2012 to 2015.

Table 1: Number of States Meeting the 2017 Benchmark for Reporting All Newborn Screening Results for All Conditions within 7 Days of Birth, by Year (2012-2015)

Year	Number of states			Percentage of specimens with newborn screening results reported to providers within 7 days of birth		
	Reporting data	Meeting benchmark (7-day time-frame goal met for at least 95 percent of specimens)	Not meeting benchmark (7-day time-frame goal met for less than 95 percent of specimens)	Median (half of the states reported 7-day time-frame goal met for at least this percentage of specimens)	Upper quartile (one-fourth of the states reported 7-day time-frame goal met for at least this percentage of specimens)	Range (lowest - highest percentage reported by a state)
2012	27	3	24	45.4	77.1	0.8 – 97.8
2013	27	2	25	40.1	79.8	1.2 – 98.1
2014	28	2	26	42.6	79.6	0.0 – 98.1
2015	27	5	22	59.0	90.2	0.0 – 98.5

Source: GAO analysis of information from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

Notes: This table shows the percentages of first newborn screening specimens screened (birth to reporting of presumptive positive and normal results for time-critical and non-time-critical conditions) within 7 days in the 28 states that provided data for this time-frame goal. Not all states reported data for all years. NewSTEPs rounded percentages to the nearest tenth of a percentage to determine the number of states that met the 95 percent benchmark. In 2015, 5 states met this benchmark (95.38 to 98.59 percent), 1 additional state was within 1 percentage point of the benchmark (94.54 percent), and 1 state had less than 1 percent of its specimens screened within 7 days of birth. The 7-day time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

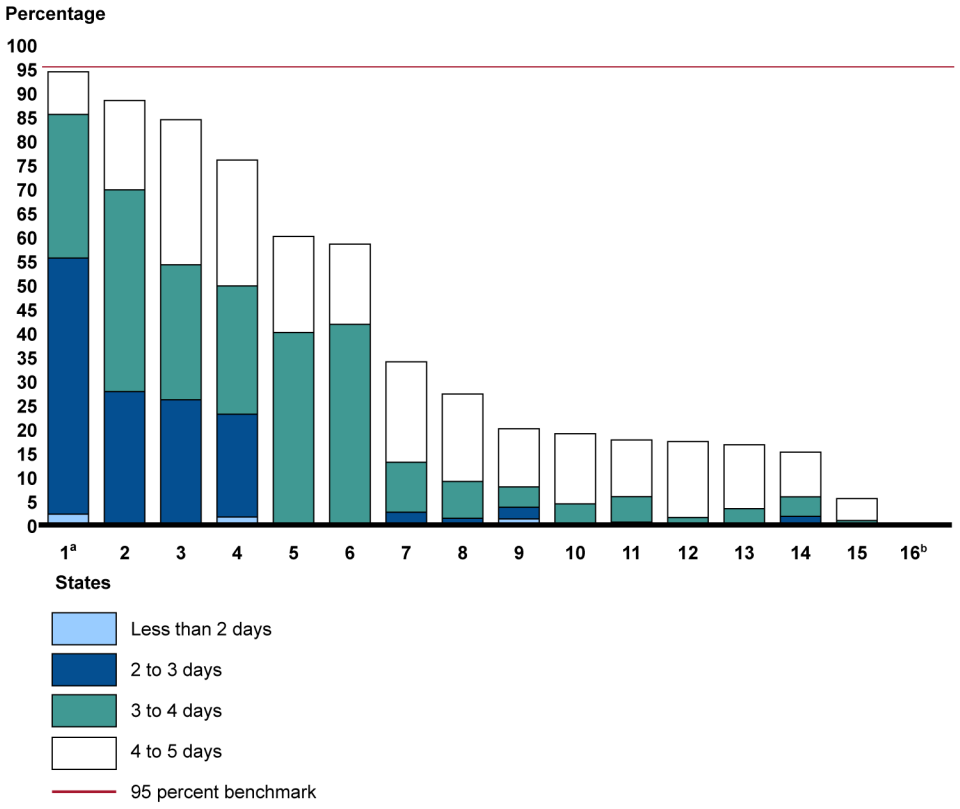
Timeliness Data for Presumptive Positive Results for Time-Critical Conditions

In 2015, states also had not met the advisory committee's benchmark for timely reporting of presumptive positive results for time-critical conditions. According to this benchmark, by 2017, states should report these results for 95 percent of specimens within 5 days of birth. In 2015, none of the 16

states that reported on this measure met the 95 percent benchmark. (See fig. 4.)

Figure 4: States' Percentages of Specimens for Which Results Were Reported to Providers within 5 Days of Birth, 2015 (Presumptive Positive Results for Time-Critical Conditions)

In 2015, none of the 16 states met the 95 percent benchmark for reporting presumptive positive newborn screening results for time-critical conditions within 5 days. The Advisory Committee on Heritable Disorders for Newborns and Children encouraged states to achieve the 95 percent benchmark by 2017.



Source: GAO analysis of data from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

Notes: This figure shows the percentages of first newborn screening specimens reporting presumptive positive newborn screening results for time-critical conditions within various time categories in each of the 16 states that provided data for this time-frame goal. NewSTEPs rounded percentages to the nearest tenth of a percentage to determine the number of states that met the 95 percent benchmark. The 5-day time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in April 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

[a]This state was within 1 percentage point of the 95 percent benchmark (94.26 percent).

[b]This state reported that no presumptive positive newborn screening results for time-critical conditions were reported within 5 days of birth.

The states' data for reporting presumptive positive results for time-critical conditions did not indicate consistent improvement over time. The median percentage of specimens screened for time-critical conditions within 5 days increased from about 23 percent in 2012 to 28 percent in 2014, but decreased to about 24 percent in 2015. NewSTEPs noted that reporting results for time-critical conditions within 5 days of birth may be the most important time-frame goal, and while the data indicate that states had difficulty meeting this goal in 2015, the data from 2014 indicate that achieving timely reporting for a high percentage of specimens is possible. For example, in 2014, two states reported meeting the 95 percent benchmark for time-critical conditions. (See table 2.) NewSTEPs also noted that time-frame goals specifically for time-critical conditions were not in place before April 2015 (when the advisory committee recommended the current time-frame goals).

Table 2: Number of States Meeting the 2017 Benchmark for Reporting Presumptive Positive Results for Time-Critical Conditions within 5 Days of Birth, by Year (2012-2015)

Year	Number of states			Percentage of specimens with newborn screening results reported to providers within 5 days of birth		
	Reporting data	Meeting benchmark (5-day time-frame goal met for at least 95 percent of specimens)	Not meeting benchmark (5-day time-frame goal met for less than 95 percent of specimens)	Median (half of the states reported 5-day time-frame goal met for at least this percentage of specimens)	Upper quartile (one-fourth of the states reported 5-day time-frame goal met for at least this percentage of specimens)	Range (lowest - highest percentage reported by a state)
2012	14	0	14	22.7	46.9	0.0 – 88.4
2013	14	0	14	22.9	70.1	0.0 – 89.0
2014	15	2	13	28.1	61.5	0.0 – 100.0
2015	16	0	16	23.6	68.0	0.0 – 94.2

Source: GAO analysis of information from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

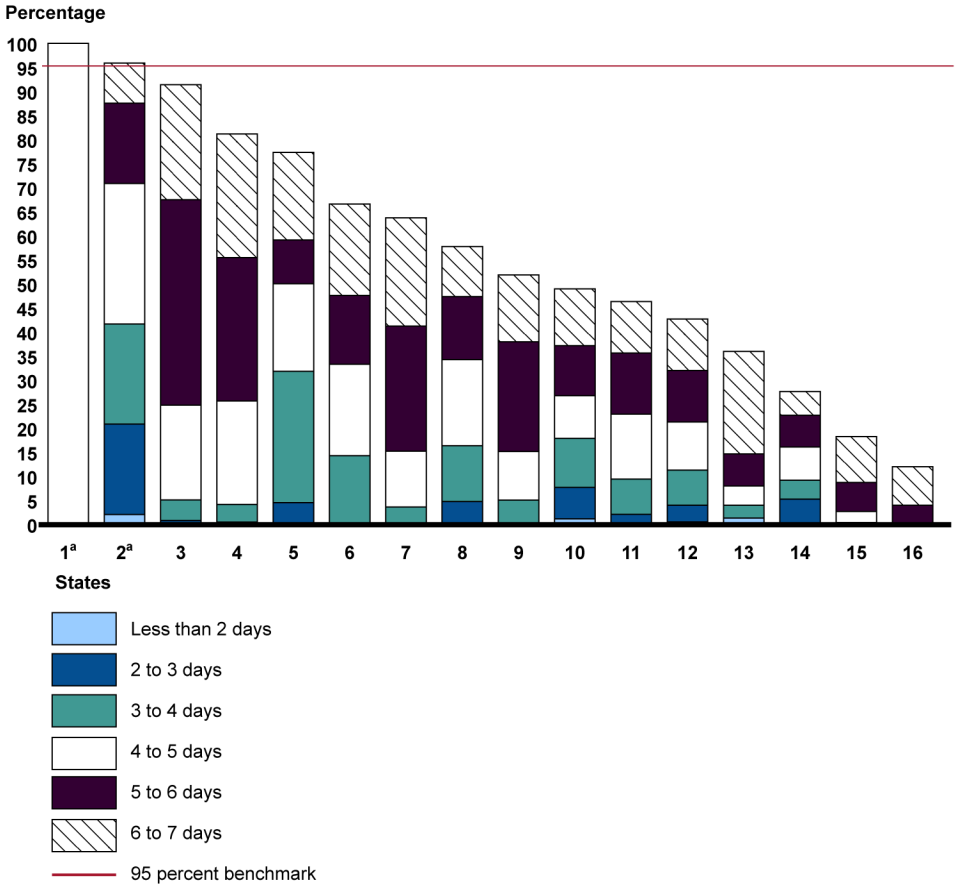
Notes: This table shows the percentages of first newborn screening specimens screened (birth to reporting of presumptive positive results for time-critical conditions) within 5 days in the 16 states that provided data for this time-frame goal. Not all states reported data for all years. NewSTEPs rounded percentages to the nearest tenth of a percentage to determine the number of states that met the 95 percent benchmark. In 2015, none of the states met the benchmark, 1 state was within 1 percentage point of the benchmark (94.26 percent), and 1 state had none of its specimens screened within 5 days of birth. The 5-day time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in April 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

Timeliness Data for Presumptive Positive Results for Non-Time-Critical Conditions

For non-time critical conditions in 2015, 2 of 16 states reporting on this measure had met the benchmark of reporting presumptive positive results within 7 days of birth for at least 95 percent of specimens. (See fig. 5.)

Figure 5: States' Percentages of Specimens for Which Results Were Reported to Providers within 7 Days of Birth, 2015 (Presumptive Positive Results for Non-Time-Critical Conditions)

In 2015, 2 of 16 states met the 95 percent benchmark for reporting presumptive positive newborn screening results for non-time-critical conditions within 7 days. The Advisory Committee on Heritable Disorders for Newborns and Children encouraged states to achieve the 95 percent benchmark by 2017.



Source: GAO analysis of data from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

Notes: This figure shows the percentages of first newborn screening specimens reporting presumptive positive newborn screening results for non-time-critical conditions within various time categories in each of the 16 states that provided data for this time-frame goal. The 7-day time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in April 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

[a]These two states met the 95 percent benchmark (95.91 and 100.00 percent).

The states' data for reporting presumptive positive results for non-time-critical conditions did not indicate consistent improvement over time. The median percentage of specimens screened for non-time-critical conditions within 7 days decreased from about 52 percent in 2012 to about 49 percent in 2013, but then increased to about 52 percent in 2014 and about 55 percent in 2015. (See table 3.)

Table 3: Number of States Meeting the 2017 Benchmark for Reporting Presumptive Positive Results for Non-Time-Critical Conditions within 7 Days of Birth, by Year (2012-2015)

Year	Number of states			Percentage of specimens with newborn screening results reported to providers within 7 days of birth		
	Reporting data	Meeting benchmark (7-day time-frame goal met for at least 95 percent of specimens)	Not meeting benchmark (7-day time-frame goal met for less than 95 percent of specimens)	Median (half of the states reported 7-day time-frame goal met for at least this percentage of specimens)	Upper quartile (one-fourth of the states reported 7-day time-frame goal met for at least this percentage of specimens)	Range (lowest - highest percentage reported by a state)
2012	14	0	14	51.7	75.0	13.8 – 93.8
2013	14	0	14	48.5	75.2	5.6 – 94.9
2014	15	2	13	51.9	92.6	0.0 – 100.0
2015	16	2	14	54.8	79.2	12.0 – 100.0

Source: GAO analysis of information from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

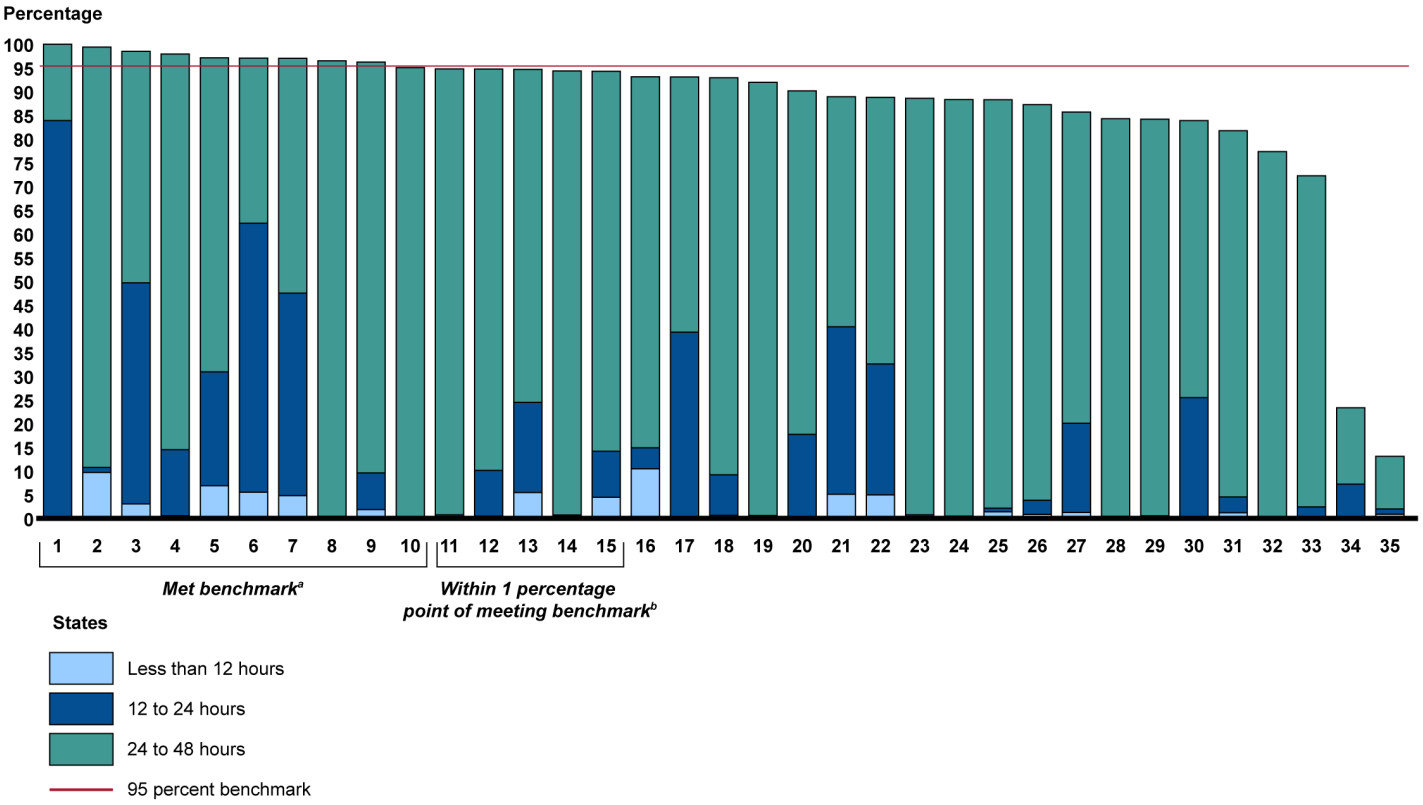
Notes: This table shows the percentages of first newborn screening specimens screened (birth to reporting of presumptive positive results for non-time-critical conditions) within 7 days in the 16 states that provided data for this time-frame goal. Not all states reported data for all years. The 7-day time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in April 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

Timeliness Data for Stage 1 of the Newborn Screening Process

States that reported timeliness data to APHL generally had not met the advisory committee's 95 percent benchmark for stage 1. In 2015, 10 out of 35 states that reported timeliness data for this stage had 95 percent of specimens collected within 48 hours of birth—a stage 1 goal. An additional 5 states did not meet the benchmark, but were close. (See fig. 6.) The median percentage of specimens collected within 48 hours of birth was about 93 percent, meaning that half of the states reported having about 93 percent or more of the specimens collected within 48 hours. (See table 4.)

Figure 6: States' Percentages of Newborn Screening Specimens Collected within 48 Hours of Birth, 2015

In 2015, 10 of 35 states met the 95 percent benchmark for collecting specimens within 48 hours of birth, and 5 additional states were within 1 percentage point of the benchmark. The Advisory Committee on Heritable Disorders for Newborns and Children encouraged states to achieve the 95 percent benchmark by 2017.



Source: GAO analysis of data from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

Notes: This figure shows the percentages of first newborn screening specimens collected within various time categories in each of the 35 states that provided data for this time-frame goal. NewSTEPs rounded percentages to the nearest tenth of a percentage to determine the number of states that met the 95 percent benchmark. The 48-hour time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in April 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

[a]These 10 states met the 95 percent benchmark (94.98 to 99.90 percent).

[b]These five states were within 1 percentage point of the benchmark (94.22 to 94.70 percent).

Table 4: Number of States Meeting the 2017 Benchmark for Collecting Newborn Screening Specimens within 48 Hours of Birth, by Year (2012-2015)

Year	Number of states			Percentage of specimens collected within 48 hours of birth		
	Reporting data	Meeting benchmark (48-hour time-frame goal met for at least 95 percent of specimens)	Not meeting benchmark (48-hour time-frame goal met for less than 95 percent of specimens)	Median (half of the states reported 48-hour time-frame goal met for at least this percentage of specimens)	Upper quartile (one-fourth of the states reported 48-hour time-frame goal met for at least this percentage of specimens)	Range (lowest - highest percentage reported by a state)
2012	33	3	30	86.3	90.4	13.5 – 99.8
2013	33	5	28	87.8	92.8	13.2 – 99.9
2014	35	7	28	89.6	94.5	13.0 – 100.0
2015	35	10	25	92.8	96.6	13.0 – 99.9

Source: GAO analysis of information from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

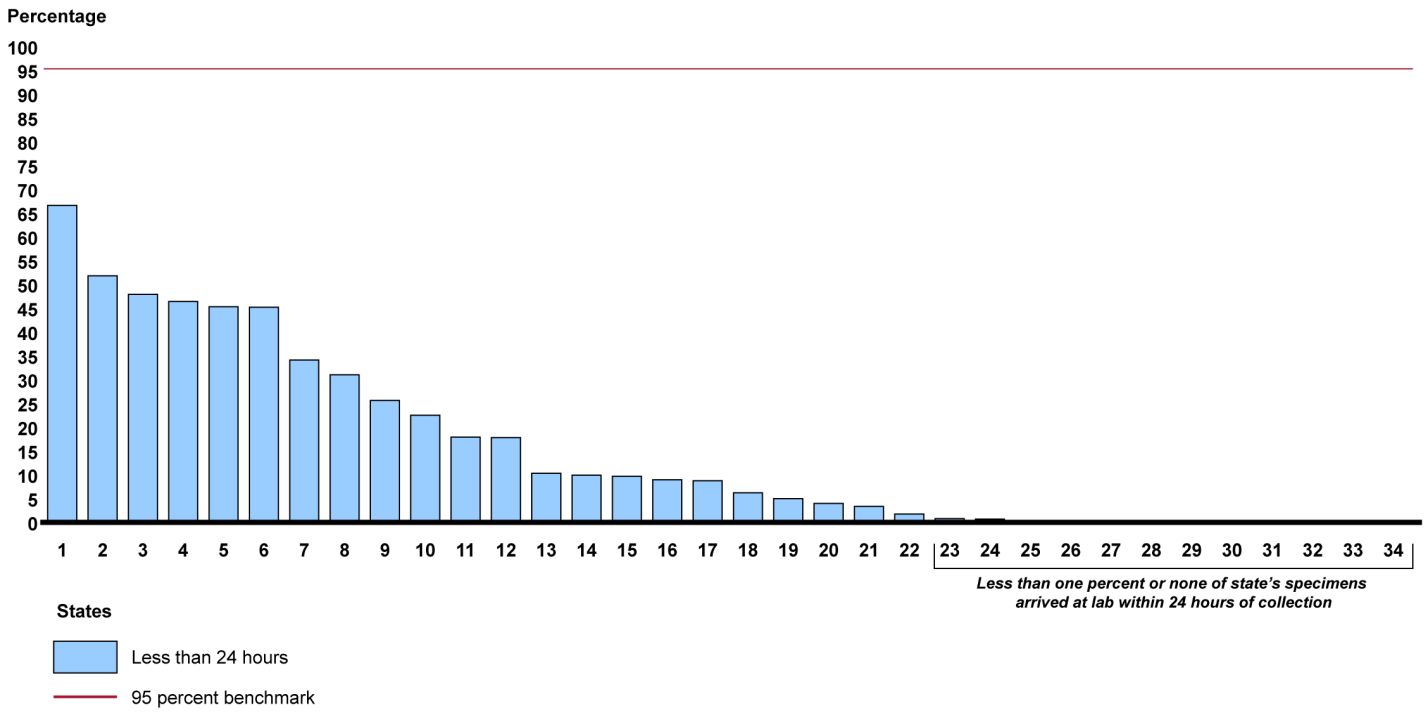
Notes: This table shows the percentages of first newborn screening specimens collected within 48 hours of birth in the 35 states that provided data for this time-frame goal. Not all states reported data for all years. NewSTEPs rounded percentages to the nearest tenth of a percentage to determine the number of states that met the 95 percent benchmark. In 2015, 10 states met the benchmark (94.98 to 99.90 percent) and 5 additional states were within 1 percentage point of the benchmark (94.22 to 94.70 percent). The 48-hour time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in April 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

Timeliness Data for Stage 2 of the Newborn Screening Process

In contrast to stage 1, none of the 34 states that reported stage 2 data for 2015 approached the 95 percent benchmark. (See fig. 7.) The median percentage of specimens arriving at the lab within 24 hours of collection was about 7 percent, meaning that half of the states reported having 7 percent or fewer of the specimens arriving at the lab within 24 hours of collection. (See table 5.) The NewSTEPs August 2016 report noted that the advisory committee's 24-hour goal for specimen arrival is ambitious. NewSTEPs also measured the percentage of specimens each state reported arriving at that lab within 48 hours of collection and found that the median percentage of specimens arriving at the lab within that more generous time-frame goal was higher (about 53 percent).

Figure 7: States' Percentages of Newborn Screening Specimens Arriving at the Lab within 24 Hours of Collection, 2015

In 2015, none of the 34 states met the 95 percent benchmark for specimens arriving at a state lab within 24 hours of specimen collection. The Advisory Committee on Heritable Disorders for Newborns and Children encouraged states to achieve the 95 percent benchmark by 2017.



Source: GAO analysis of data from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

Notes: This figure shows the percentages of first newborn screening specimens arriving at the lab within 24 hours of collection in each of the 34 states that provided data for this time-frame goal. Twelve of the states had less than 1 percent or none of its specimens arriving at the lab within 24 hours of collection. The 24-hour time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in April 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

Table 5: Number of States Meeting the 2017 Benchmark for Arrival of Newborn Screening Specimen at Lab within 24 Hours of Collection, by Year (2012-2015)

Year	Number of states			Percentage of specimens arrived at lab within 24 hours		
	Reporting data	Meeting benchmark (24-hour time-frame goal met for at least 95 percent of specimens)	Not meeting benchmark (24-hour time-frame goal met for less than 95 percent of specimens)	Median (half of the states reported 24-hour time-frame goal met for at least this percentage of specimens)	Upper quartile (one-fourth of the states reported 24-hour time-frame goal met for at least this percentage of specimens)	Range (lowest - highest percentage reported by a state)
2012	32	0	32	3.4	12.1	0.0 – 52.8
2013	32	0	32	3.5	12.9	0.0 – 50.7
2014	33	0	33	3.4	23.5	0.0 – 54.7
2015	34	0	34	7.4	25.6	0.0 – 66.6

Source: GAO analysis of information from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). | GAO-17-196

Notes: This table shows the percentages of first newborn screening specimens arriving at a newborn screening lab within 24 hours of specimen collection in the 34 states that provided data for this time-frame goal. Not all states reported data for all years. In 2015, 12 states had less than 1 percent or none of its specimens arriving at the lab within 24 hours of birth. The 24-hour time-frame goal was recommended by the Department of Health and Human Services' Advisory Committee on Heritable Disorders for Newborns and Children in April 2015. The committee encouraged states to achieve this goal for 95 percent of specimens by 2017.

Missing Data and Variations in Data Collection Limit a Full Understanding of Newborn Screening Timeliness Trends, but HRSA Has Been Addressing These Challenges

Missing data for a number of states limit a full understanding of newborn screening timeliness trends. The NewSTEPs August 2016 report included annual timeliness data for 38 states, but did not include any data for 15 states.[24] According to APHL officials, none of the states expressly declined to provide data to NewSTEPs for the August 2016 report; however, some states did not respond to the data request, and some states' officials indicated that they were willing to provide data, but could not do so in time, citing resource constraints. In addition, the 38 states that provided data did not do so for all time-frame goals or all years (2012 through 2015). APHL officials told us that the lack of data for certain time-frame goals or years was due to factors such as competing priorities or limitations in states' information systems, specifically in LIMS. For example, APHL reported that two states do not electronically capture the date and time that test results are reported to providers, and newborn screening officials in those states could not search paper records in time

[24]The 15 states that were not included in the NewSTEPs August 2016 report were: Connecticut, District of Columbia, Guam, Idaho, Kansas, Kentucky, Maryland, Massachusetts, Michigan, Mississippi, Montana, Nevada, Oklahoma, Utah, and West Virginia.

to provide the data. Additionally, a few states had recently changed their LIMS, which resulted in limited access to data for some years.

Variations in data collection also limit a full understanding of newborn screening timeliness trends. According to APHL officials, the data in the NewSTEPs August 2016 report generally represent with accuracy the time taken to screen specimens in reporting states, but there are a number of limitations, including the following examples:

- Although the advisory committee's time-frame goals apply to first specimens only, some states' data did not distinguish a lab's receipt of a first specimen from receipt of a subsequent specimen, which can result in the appearance of longer screening times (that is, longer times from birth to specimen collection and birth to reporting results) for such states.
- Variation exists in how state labs define specimen arrival at the lab, which can be the time a specimen is delivered by a courier, the time lab staff record receipt of the specimen in LIMS, or the time lab staff initiate testing of the specimen. This variation can affect the data reported for stage 2 (specimen collection to lab arrival).
- Many states' LIMS do not allow lab staff to record separate dates for when results for time-critical conditions and results for non-time-critical conditions from the same specimen card were reported to providers, even though time-critical results may be reported earlier. These systems typically include data entry fields that capture only the date that all results (presumptive positive and normal) for all conditions (time-critical and non-time-critical) were reported to providers, which can result in the appearance of longer newborn screening times for states with such systems.

With HRSA's support, NewSTEPs has been taking steps to improve the completeness and consistency of the annual newborn screening timeliness data that states submit to the data repository. APHL officials told us that they expect to have all 53 states sign the MOU and enter data into the data repository. As participation in the data repository increases and data definitions are used more consistently across states, NewSTEPs can more accurately assess timeliness in states across the country. Steps taken by NewSTEPs include

- *Increasing participation in the data repository*. According to HHS, as of November 20, 2016, 35 out of 53 states had signed an MOU with APHL to provide annual data to the data repository for future analysis, and, therefore, receive data-related technical assistance from

NewSTEPs.[25] APHL officials told us they have been working with the remaining 18 states to address issues, such as confidentiality concerns, in an effort to have the MOUs signed. APHL officials say they have been reaching out to achieve buy-in from the remaining states through a variety of methods, including sending emails; making phone calls; conducting in-person meetings; incorporating reminders in webinars on newborn screening; and engaging with organizations, such as the American Academy of Pediatrics. APHL officials told us that while the data in the NewSTEPs August 2016 report provide a meaningful understanding of timeliness in a large number of states, they expect this understanding to improve as more states sign MOUs and submit data to the data repository.

- *Clarifying data definitions*. NewSTEPs has also reviewed the data definitions used for the data repository to address variability in data collection and reporting among states. APHL officials said that, as a result of this review, NewSTEPs revised guidance documents for its data dictionary to, for example, more clearly separate screening timeliness data for first specimens from data for subsequent specimens. NewSTEPs is working with states participating in the program to help ensure they use these revised definitions consistently when submitting timeliness data to the data repository.

APHL officials told us they plan to publish state-specific reports on the NewSTEPs website by early 2017 to promote continuous quality improvement by allowing states and others an opportunity to review states' progress toward meeting the advisory committee's benchmarks. According to these officials, each state will be able to track its progress on a specific time-frame goal over time, as well as examine how its timeliness compares to that of other states.

[25]Not all of the 35 states with signed MOUs have started entering data into the data repository. APHL officials told us they expect all states with signed MOUs to be entering annual timeliness data on a yearly basis by April 2017.

States Identified Numerous Barriers Affecting Timeliness, and a Variety of Strategies Have Been Developed to Address Them

State newborn screening officials identified numerous barriers to timeliness in each of the three stages of the newborn screening process and developed a variety of strategies to address these barriers. HRSA, through its cooperative agreement for NewSTEPs 360, has been funding activities to provide technical assistance to states to address barriers and improve the timeliness of newborn screening.

States Identified Numerous Barriers to Timeliness throughout the Newborn Screening Process and Some States Have Developed Strategies to Address Them

Newborn screening officials from 51 states who responded to the advisory committee's 2014 survey identified numerous barriers to timeliness in each of the three stages of the newborn screening process. Examples of barriers include a lack of understanding of the importance of timely screening among providers performing out-of-hospital births (stage 1), limited courier availability (stage 2), and insufficient lab operating hours (stage 3). Newborn screening officials in selected states told us they developed a variety of strategies to address these barriers.

Stage 1: Birth to Specimen Collection

Newborn screening officials who responded to the 2014 survey identified barriers to timely collection of newborn screening specimens. Barriers included nursing protocols that are not always consistent with advisory committee time-frame goals, lack of feedback to hospitals on timeliness performance, and lack of understanding of the importance of timely screening for out-of-hospital births. (See table 6.) Newborn screening officials in the four selected states we interviewed reported developing strategies to address the barriers.

Table 6: Selected Barriers and Examples of Strategies for Timely Newborn Screening from Birth to Specimen Collection

Newborn screening officials who responded to a 2014 survey identified barriers to timely newborn screening from birth to specimen collection, and selected states involved in newborn screening activities developed strategies to address barriers.

Barrier	Strategy
Hospital nursing protocols are not always consistent with advisory committee goals.	Change hospital nursing protocols to be consistent with timeliness goals, and provide education to providers on such changes.
Lack of feedback from newborn screening officials at the state level to hospitals on timeliness performance.	Provide feedback to hospitals regularly, including information on performance related to timeliness goals and cases where the timeliness goals were not met.

Barrier	Strategy
Premature infants and infants transferred shortly after birth.	Provide hospital feedback with a subset of information specific to neonatal intensive care units (NICU) and targeted educational presentations to NICUs.
Lack of understanding of the importance of timely screening among providers performing out-of-hospital births.	Conduct focused outreach and training to midwives on the importance of timely screening.
Staffing issues, such as high turnover, result in some providers being unfamiliar with newborn screening timeliness initiatives.	Provide ongoing and reinforced education through, for example, an educational video for new hospital staff.

Source: GAO analysis of information from the Advisory Committee on Heritable Disorders in Newborns and Children, and newborn screening officials in selected states. | GAO-17-196

Note: The selected barriers shown in this table include barriers identified by respondents to the advisory committee's 2014 survey as having a major or moderate impact on timeliness; barriers that were frequently reported by survey respondents in the written responses; or barriers in the survey that were highlighted by a number of state newborn screening officials in published presentations and reports.

Newborn screening officials from selected states reported that following nursing protocols that are inconsistent with the advisory committee's time-frame goals can cause delays. For example, according to newborn screening officials in one state, nursing protocols often dictate that specimen collection be performed as late as possible prior to the baby's discharge from the hospital. According to these officials, this protocol can result in late collection of some specimens; for example, among newborns born via Caesarean-section, who often have longer hospital stays. To improve the timeliness of specimen collection, this state recommended that hospitals make nursing protocols consistent with the advisory committee goal to collect specimens within 24 to 48 hours of birth, and has developed educational strategies to advise providers to aim for collection to take place within 24 hours of birth.

A lack of feedback from state newborn screening officials to hospitals was also identified as a barrier to timely specimen collection, according to newborn screening officials who responded to the 2014 survey, because providers may be unaware that they are not meeting timeliness goals. Officials we interviewed from three states reported developing or improving methods of providing feedback to hospitals through online quality reports or report cards. For example, newborn screening officials in one state we interviewed said they provide feedback to hospitals through report cards that evaluate hospital performance based on the advisory committee's goal for timely specimen collection. The report cards are disseminated monthly and include an outlier report that alerts facilities when specific specimens do not meet the timeliness goal. According to the officials, these outliers showed problems with timeliness at neonatal intensive care units (NICU). As a result, the state began

reporting NICU timeliness separately on the report cards; subsequently, newborn screening officials reported that NICU timeliness has improved.

Arizona's Efforts to Address Barriers to Timely Screening for Out-of-Hospital Births

Newborn screening officials in Arizona have focused on improving the timeliness of newborn screening for out-of-hospital births. Generally, according to newborn screening officials, babies born outside of a hospital—such as in birthing centers (freestanding facilities separate from hospitals) and home births—have higher rates of delayed specimen collection. Newborn screening officials in Arizona attributed this to a variety of causes, including a lack of understanding among providers about the importance of timely screening, and a lack of standardization of protocols for birthing centers and home births. For example, for healthy, low-risk deliveries outside of hospitals, midwives often leave 4 hours after the baby is born and may not follow up during the period when specimen collection should occur. Since 2011, Arizona has provided education and basic background training on newborn screening to midwives individually and through the state's midwifery association to help address this barrier. Newborn screening officials explained that following the increased outreach and training, newborn screening timeliness for out-of-hospital births has improved, but noted that midwives continue to have problems seeking reimbursement for newborn screening services.

Source: GAO analysis of information from the Advisory Committee on Heritable Disorders in Newborns and Children, and newborn screening officials in Arizona. | GAO-17-196

Stage 2: Specimen Collection to Lab Arrival

Newborn screening officials who responded to the 2014 survey identified a variety of barriers that may delay the arrival of newborn screening specimens at the state lab, including hospitals and other providers waiting to send specimens to the lab in batches, insufficient lab operating hours, and a lack of courier services for transporting specimens. (See table 7.)

Table 7: Selected Barriers and Examples of Strategies for Timely Newborn Screening from Specimen Collection to Lab Arrival

Newborn screening officials who responded to a 2014 survey identified barriers to timely newborn screening from specimen collection to lab arrival, and selected states involved in newborn screening activities developed strategies to address barriers.

Barrier	Strategy
Waiting to send specimens to the lab in batches.	Conduct site visits and provide education on the importance of timely transport of specimens.
Operating hours of the lab may result in specimens not arriving in a timely way.	Increase lab operating hours to include Saturday hours for receipt and testing of specimens.
Transport of specimens by mail due to lack of courier service.	Expand courier service to all providers and offer transport services 6 days per week.
Geographic distance from the lab.	Expand courier service to all providers, including transport of specimens further from the lab by flight.
Lack of understanding of the importance of timely specimen transport for out-of-hospital births.	Provide courier service to all midwives, and provide education on the importance of timely transport of specimens.

Barrier	Strategy
Problems tracking date and time specimen was sent to or arrived at the lab, and lack of feedback to hospitals on their performance.	Update laboratory information management systems to include date and time specimen was sent and arrived at the lab, and provide report cards to hospitals on transit times.

Source: GAO analysis of information from the Advisory Committee on Heritable Disorders in Newborns and Children, and newborn screening officials in selected states. | GAO-17-196

Note: The selected barriers shown in this table include barriers identified by respondents to the advisory committee's 2014 survey as having a major or moderate impact on timeliness; barriers that were frequently reported by survey respondents in the written responses; or barriers in the survey that were highlighted by a number of state newborn screening officials in published presentations and reports.

One barrier to timely completion of stage 2 (collection to lab arrival) identified by officials responding to the 2014 survey was providers waiting to send specimens to the lab in batches.[26] To address this practice, known as batching, officials from selected states reported employing strategies that involved providing feedback and training to providers at hospitals. According to newborn screening officials in one state, when specimens take more than 3 days from birth to arrive at a lab—which corresponds with the combined time-frame goals for stage 1 and stage 2—hospitals are asked to review those cases and avoid batching in the future.

Another barrier identified by officials responding to the 2014 survey is that lab staff are not always available to receive newborn screening specimens, because the lab's operating hours do not align with courier service, mail, or other delivery service times. In three of the selected states, state officials told us that they addressed this barrier by having lab staff available on Saturday to receive and test specimens or to ensure they can be tested first thing Monday morning. Officials in one of these states also reported developing a process for cases in which a geneticist believes a baby's specimen is likely positive for a time-critical condition. Under this process, the baby's physician calls the state newborn screening program, and a courier or a state health official will pick up the specimen within 2 hours for transport to the lab for immediate testing.

[26]According to officials in one selected state, batching has occurred for a variety of reasons, including when lab staff thought they were saving the state money by sending larger shipments of specimens every few days, rather than sending specimens to the lab daily. Batching has also occurred unintentionally, when, for example, there was no designated provider to collect specimen cards from the drying rack to prepare them for transport.

Colorado's Efforts to Expand Courier Service to Mitigate Geographic Challenges

Newborn screening officials in Colorado told us that they expanded courier service to all hospitals in 2015 to address barriers to timely arrival of specimens at the state lab after specimen collection. According to these officials, prior to 2015, rural hospitals facing geographic challenges, such as long distances to the state lab, relied on mail services to transport newborn screening specimens to the lab. Beginning in April 2015, the newborn screening program's courier service was expanded to all hospitals in the state, including these rural hospitals. Newborn screening officials explained that courier service is particularly beneficial for hospitals located long distances from the state lab, because it can include direct transport from the hospital to a nearby airport, a flight, and direct transport from the airport to the lab. In addition, courier service was expanded to have pick-up 6 days per week for all hospitals. This increase in courier service provides additional opportunities for timely specimen pick up from the hospital for transporting to the lab. According to Colorado newborn screening officials, these efforts have reduced specimen transport time for some facilities by up to 3 days.

Source: GAO analysis of information from the Advisory Committee on Heritable Disorders in Newborns and Children, and newborn screening officials in Colorado. | GAO-17-196

Stage 3: Results Reporting

Newborn screening officials who responded to the 2014 survey identified barriers to timely results reporting, such as insufficient lab operating hours and labs' reliance on the mail to communicate results. (See table 8.)

Table 8: Selected Barriers and Examples of Strategies for Timely Newborn Screening from Lab Arrival to Results Reporting

Newborn screening officials who responded to a 2014 survey identified barriers to timely newborn screening from lab arrival to results reporting, and selected states involved in newborn screening activities developed strategies to address barriers.

Barrier	Strategy
Lab operating hours limit times that test results can be reported.	Expand operating hours to include reporting 6 days per week.
Reliance on mail limits how quickly test results are shared with provider.	Implement more timely reporting methods, such as fax or electronic reporting.
Lack of standardization of laboratory information management systems (LIMS) hinders feedback and slows reporting.	Update LIMS to align with relevant quality indicators to provide more accurate feedback, and to allow electronic messaging, reducing the burden on lab staff.
Delays in lab processes, such as some tests being run only twice per week.	Evaluate lab hours and staffing levels and increase the frequency of some testing procedures.
Specimen arrival at the lab past the time-frame goal, which may indicate problems with specimen quality for testing (e.g., specimen stored outside room temperature).	If specimen arrives outside the time frame, notify sender to request a new specimen.[a]

Source: GAO analysis of information from the Advisory Committee on Heritable Disorders in Newborns and Children, and newborn screening officials in selected states. | GAO-17-196

Note: The selected barriers shown in this table include barriers identified by respondents to the advisory committee's 2014 survey as having a major or moderate impact on timeliness or barriers that were frequently reported by survey respondents in the written responses.

[a]Officials told us that they request an additional specimen if the first specimen is received more than 7 days after collection, because the age of the first specimen and unknown storage conditions while delayed in transit may compromise test results. However, the first specimen is still tested to help ensure timely results. Officials stated that this type of delay is an infrequent occurrence.

In addition to affecting timely specimen arrival at the lab, a lab's operating hours may also affect how quickly staff are available to test specimens and report results. Officials from one state told us that their strategy to address this barrier included expanding lab operating hours to 6 days a week: specimens are processed Monday through Saturday, allowing the lab to report results for time-critical conditions to providers on Sunday instead of waiting until Monday.

Additionally, according to some state newborn screening officials we interviewed, another barrier to timely reporting is that some labs report results to providers via mail; as a result, providers could wait up to a week to receive results after they are sent. Newborn screening officials in one state told us that they updated provider records to include fax numbers and began faxing newborn screening results to providers. Newborn screening officials in two other states told us that they are beginning or planning to report presumptive positive results electronically prior to sending them by mail; for example, seven hospitals in one of these states are piloting a program that allows providers to electronically access results as soon as screening tests are completed at the lab.

Wisconsin's Efforts to Improve Its Laboratory Information Management System

Newborn screening officials in Wisconsin reported updating their laboratory information management system (LIMS) to align with newborn screening quality indicators to provide better feedback on newborn screening timeliness to hospitals, and to allow electronic messaging between hospitals and labs in the future, increasing record accuracy and reducing the need for manual entry. State newborn screening officials told us that to align LIMS with newborn screening quality indicators, they added new fields to LIMS to accurately measure time taken to complete the stages of the newborn screening process. For example, they explained that by adding a field in LIMS to record the time a specimen was received at a lab they can more accurately measure the amount of time between specimen collection and receipt at the lab. Newborn screening officials said that with this feedback, hospitals should be able to better identify changes needed to improve timeliness. Wisconsin also reported updating its LIMS to meet Health Level 7 standards, known as HL7, which provide a framework for health information retrieval and exchange from one information system to another (in this case from hospitals' information systems to LIMS). Wisconsin newborn screening officials are working with hospitals to standardize information in their electronic health information systems so that newborn screening tests can be ordered electronically and LIMS can automatically retrieve and exchange demographic and other information from hospital systems, reducing the need for manual entry of information and increasing accuracy. Finally, Wisconsin officials told us that they are creating a web portal that allows providers to access the newborn screening results in LIMS for their patients online in real-time, reducing the amount of time taken to report newborn screening test results.

Source: GAO analysis of information from the Advisory Committee on Heritable Disorders in Newborns and Children, and newborn screening officials in Wisconsin. | GAO-17-196

HRSA Has Started Supporting States through Technical Assistance to Help Address Timeliness Barriers

HRSA, through its cooperative agreement for NewSTEPs 360, has recently focused on improving newborn screening timeliness by funding activities to provide technical assistance to and information sharing among states. According to officials involved in administering NewSTEPs 360, since January 2016, they have held telephone calls to provide coaching to 20 of the 28 states participating in the program; these officials said they expect to begin coaching calls with the remaining 8 states by January 2017. The goal of these coaching calls is to help states achieve the advisory committee's benchmark of timely reporting of newborn screening results for 95 percent of newborn screening specimens by 2017. According to newborn screening officials from two states that participate in NewSTEPs 360, the coaching calls help states prioritize their efforts to improve newborn screening timeliness and help states hold themselves accountable for meeting milestones, because they report on progress made during each monthly call. In addition, these officials said that participating in NewSTEPs 360 allows them to learn about strategies developed by other participating states. For example, officials in one state told us that they formed a small group of officials from states working to update their LIMS to meet Health Level 7 standards (known as HL7) for electronic health information exchange. (These standards provide a framework for health information retrieval and exchange from one information system to another—in this case from hospital information systems to LIMS.) According to these officials, participation in this small group helped them identify milestones to break their project into manageable pieces. The officials participating in the small group also said that they compared and shared strategies for meeting both these short-term milestones and their overall goal for updating LIMS.

Officials involved in administering NewSTEPs 360 told us the program has started taking steps to analyze and share information about barriers and strategies gathered from states that enter monthly timeliness data, and receive technical assistance through the program in order to help identify and promote the use of successful strategies. According to program officials, these steps include the following examples:

- Sharing NewSTEPs' August 2016 report on newborn screening timeliness, which contained information on activities that some NewSTEPs 360 states had undertaken to improve timeliness. The report included, for example, strategies for improving newborn screening education for providers.
- Sharing information on lessons learned (such as factors that may predict newborn screening timeliness in hospitals) through an online

video and presenting strategies at the 2016 APHL Newborn Screening and Genetic Testing Symposium.

- Sharing information in messages sent to states through a listserv. For example, in September 2016, NewSTEPs 360 sent a message summarizing a new cystic fibrosis newborn screening timeliness initiative. For this initiative, NewSTEPs 360, in collaboration with the Cystic Fibrosis Foundation, convened stakeholders to identify strategies for reporting test results for cystic fibrosis in a more timely way.
- Analyzing monthly timeliness data from states participating in NewSTEPs 360. According to APHL officials, as of November 16, 2016, 20 of 28 states participating in NewSTEPs 360 had entered monthly timeliness data into the data repository and the program had begun analyzing these data to track any progress in these states. These officials told us they expect that the remaining states will begin submitting monthly timeliness data by mid-December 2016.
- Coding transcripts from the monthly coaching calls for states participating in NewSTEPs 360 to categorize and track barriers experienced by states and strategies developed to address them. As calls are completed over time, officials involved in administering NewSTEPs 360 believe this will allow them to compare the resulting data with the monthly timeliness data to measure the impact of developing a given strategy. These officials told us that they expect to present results from this analysis to all states (regardless of participation in NewSTEPs 360) in 2018.

In addition, HRSA also funded targeted technical assistance to help nurses improve newborn screening timeliness through NewSTEPs 360. Under a sub-award from NewSTEPs 360, the Genetic Alliance started providing training to nurses on the importance of timely screening. This includes, for example, free education on newborn screening specimen collection through an online training portal. According to an official at Genetic Alliance, the organization is also involved in identifying barriers that contribute to newborn screening delays and strategies to address such barriers. Based on information gathered in focus groups with nursery and NICU nurses held in June 2016, Genetic Alliance drafted a number of recommendations for hospitals and nurses to help address barriers to timely newborn screening. These recommendations include working with nurses to better integrate updated newborn screening guidance—such as the advisory committee's 2015 time-frame goal for

collecting newborn screening specimens—into nurses' protocols for newborn screening.[27]

It is too soon to determine which strategies, if any, developed through HRSA-supported technical assistance have a measurable impact on improving timeliness in states participating in NewSTEPs 360, and whether these strategies could be effective in additional states. The program began collecting monthly timeliness data from participating states in January 2016, and not all states have started entering data; eight states that were selected to participate in NewSTEPs 360 in October 2016 have not yet started participating in monthly coaching calls. In addition, Genetic Alliance has not yet issued its recommendations to hospitals and nurses. According to HRSA officials, the agency will be conducting annual monitoring of NewSTEPs 360, and a final report that includes performance measures for NewSTEPs 360 is required to be completed by late 2018. HRSA officials told us that the report will capture the extent to which states' timeliness improved as a result of technical assistance received through the program.

Agency Comments and Our Evaluation

We provided a draft of this report to HHS for review and comment. In its written comments, reproduced in appendix III, HHS generally agreed with our data-supported findings, but noted two concerns about the conclusions we have drawn from the findings.

First, the department noted concern with our use of the advisory committee's benchmark, which it encouraged states achieve to by 2017, to assess whether states screened newborns in a timely manner. We report that data provided by 38 states for 2012-2015 showed that states generally had not met the advisory committee's recommended 2017 benchmark of meeting each time-frame goal for at least 95 percent of specimens. Our analysis is of the most recent data available, and our report states clearly that the advisory committee recommended states achieve these goals by 2017. Time-frame goals for completing newborn screening were initially identified in 2005, and concerns about the timeliness of screening date back to at least late 2013. Our analysis indicates that substantial work remains for the majority of states to

[27]As of October 31, 2016, these recommendations had not been finalized. According to a Genetic Alliance official involved in developing the recommendations, the organization plans to present its findings and recommendations from the focus groups at association conferences in 2017.

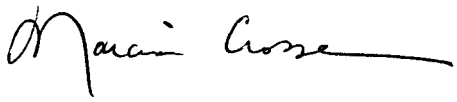
achieve the recommended benchmark by 2017, based on the latest available information.

HHS also commented that our findings were limited by not including point-of-care screening within the definition of newborn screening. In the report, we include information on the 2 conditions—critical congenital heart disease, and hearing loss—that use point-of-care screening, and note that these are 2 of the 32 conditions on the RUSP. However, since these two conditions are not subject to the advisory committee's time-frame goals (which apply to newborn screening using a blood specimen), and NewSTEPs' August 2016 report did not include data on timeliness for these two conditions, we did not include them in the timeliness data in our report or in the description of barriers and strategies.

In addition, HHS provided technical comments, which we incorporated as appropriate.

We are sending copies of this report to the Secretary of Health and Human Services, and appropriate congressional committees. In addition, the report will be available at no charge on GAO's website at http://www.gao.gov.

If you or your staff have any questions about this report, please contact me at (202) 512-7114 or at crossem@gao.gov. Contact points for our Office of Congressional Relations and Office of Public Affairs can be found on the last page of this report. Other major contributors to this report are listed in appendix IV.



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Appendix I: Scope and Methodology

To examine what is known about the timeliness of newborn screening for heritable conditions, we reviewed timeliness data from states included in an August 2016 report from the Newborn Screening Technical assistance and Evaluation Program (NewSTEPs). NewSTEPs is administered by the Association of Public Health Laboratories (APHL), in collaboration with the University of Colorado's School of Public Health, through a cooperative agreement with the Health Resources and Services Administration (HRSA), an agency within the Department of Health and Human Services (HHS).[1] The August 2016 report included data collected from states through a data repository maintained by APHL under NewSTEPs.[2]

The data repository includes (but is not limited to) annual timeliness data collected from states participating in NewSTEPs.[3] These annual timeliness data are based on time-frame goals recommended by HHS's Advisory Committee on Heritable Disorders in Newborns and Children in April 2015 and data definitions developed by a workgroup composed of newborn screening experts and stakeholders convened by APHL.[4] For individual newborn screening stages (e.g., specimen collection to lab arrival) or the full newborn screening process, the data measure the percentages of a state's specimens screened within the advisory committee's time-frame goals. For example, the data measure the percentage of specimens for which all results for all conditions were

[1]NewSTEPs is a collaborative effort between APHL and the University of Colorado's School of Public Health, with both organizations operating in partnership. Timeliness data we obtained from NewSTEPs in the August 2016 report reflect the combined work of both organizations.

[2]In this report, the term states refers to all 50 states, the District of Columbia, Guam, and Puerto Rico. States are required to have a signed memorandum of understanding (MOU) with APHL in order to enter timeliness data into the data repository. According to HHS, as of November 20, 2016, 35 of 53 states had a signed MOU.

[3]The data repository also includes, for example, equivalent monthly timeliness data collected from states participating in another program called NewSTEPs 360 and public health surveillance case data from states participating in NewSTEPs. The case data measure timeliness for each newborn, including hours from birth to specimen collection, days from birth to specimen receipt at the lab, days from birth to reporting results, days from birth to intervention, and days from birth to a confirmed diagnosis. This allows NewSTEPs and states entering data to track continuous timeliness measures for confirmed cases throughout the newborn screening process by condition, by condition category, and by the time-critical nature of treating the condition.

[4]Although the time-frame goals were not in place for the entire period of time captured in NewSTEPs August 2016 report (2012 through 2015), the data provide useful information about the states' past performance relative to these goals.

reported within the advisory's committee's goal of 7 days after birth. For the August 2016 report, NewSTEPs requested annual timeliness data from all 53 states, and 38 states submitted data that were included in this report. (See table 9.) Of the 38 states reporting timeliness data included in the report, 20 states entered data directly into the data repository and 18 additional states did not have a signed memorandum of understanding (MOU) and submitted equivalent data using a spreadsheet provided by NewSTEPs. For all 38 states included in the report, NewSTEPs determined the percentages of specimens screened within each of the committee's 2015 time-frame goals (for the full newborn screening process or individual stages) in 2012, 2013, 2014, and 2015.[5] The NewSTEPs report also included the median, quartiles, minimum, and maximum percentage meeting the 2015 time-frame goal in each year.

Table 9: States Represented in the Newborn Screening Technical Assistance and Evaluation Program (NewSTEPs) August 2016 Report on Newborn Screening Timeliness

States represented in report (total=38)	States not represented in report (total=15)
Alabama, Alaska, Arizona, Arkansas, California, Colorado, Delaware, Florida, Georgia, Hawaii, Illinois, Indiana, Iowa, Louisiana, Maine, Minnesota, Missouri, Nebraska, New Hampshire, New Jersey, New Mexico, New York, North Carolina, North Dakota, Ohio, Oregon, Pennsylvania, Puerto Rico, Rhode Island, South Carolina, South Dakota, Tennessee, Texas, Vermont, Virginia, Washington, Wisconsin, and Wyoming.	Connecticut, District of Columbia, Guam, Idaho, Kansas, Kentucky, Maryland, Massachusetts, Michigan, Mississippi, Montana, Nevada, Oklahoma, Utah, West Virginia

Source: GAO analysis of information from NewSTEPs. | GAO-17-196

Notes: In this table, states include all 50 states, the District of Columbia, Guam, and Puerto Rico. This table shows the 38 states for which annual timeliness data were represented in NewSTEPs' August 2016 report. Not all of the reporting states provided data for all time-frame goals or all years (2012-2015).

[5]In addition to calculating percentages based on the advisory committee's 2015 time-frame goals, NewSTEPs calculated percentages for stage 3 of the newborn screening process (specimen lab arrival to reporting of results) based on a 4-day time-frame goal. The advisory committee, however, does not have a time-frame goal for stage 3. NewSTEPs used the 4-day time-frame goal, because it is the difference between the combined time-frame goals for stages 1 and 2 (48 hours for specimen collection and 24 hours for lab arrival) and the 7-day goal for the full newborn screening process. In our review, we only included data based on advisory committee time-frame goals.

We assessed the reliability of the annual data in the NewSTEPs August 2016 report for the purposes of examining what is known about the timeliness of newborn screening by taking several steps. For example, we reviewed spreadsheets sent to states without an MOU and confirmed that the spreadsheets were based on the same definitions as the data repository (used by states with signed MOUs). We also confirmed that the spreadsheets had a built-in mechanism to mitigate data entry errors, such as automatic calculation of percentages. We interviewed officials from APHL and the University of Colorado's School of Public Health to confirm that the data repository also had mechanisms to reduce risks of error. For example, these officials said the data repository can automatically calculate percentages and identify obvious data errors, such as values over 100 percent. These officials also noted that the data combined from the data repository and spreadsheets were based on common data definitions and that the data were carefully reviewed and searched for outliers before being reported. Based on these steps, we determined that these data were sufficiently reliable for the purposes of our report. The timeliness data for the states providing data for the NewSTEPs August 2016 report are not generalizable to other states, but provided valuable insight on what is known about newborn screening timeliness in the reporting states.

In addition to reviewing the annual timeliness data in the August 2016 report, we reviewed time-frame goals from the advisory committee included in an April 2015 letter to the Secretary of Health and Human Services, as well as results from the committee's 2014 survey, which analyzed timeliness for specimens screened from January through May 2014. We also reviewed documents obtained from APHL officials that describe the data repository, quality indicators, and methods for entering data. We interviewed officials from APHL and the University of Colorado's School of Public Health to learn about their activities related to timeliness, including how they manage the data repository, and to discuss the August 2016 report. We also interviewed these officials to describe any plans to track newborn screening timeliness in the future. We interviewed newborn screening stakeholders identified by APHL and on HRSA's website to learn about efforts to track newborn screening timeliness. These stakeholders included a member of the advisory committee who co-chaired a timeliness workgroup, a member of the advisory committee who conducted research related to newborn screening barriers, and officials from associations (Association of Maternal & Child Health Programs, Association of State and Territorial Health Officials, Genetic Alliance, and March of Dimes). Stakeholders stated that NewSTEPs' work in tracking timeliness through the data repository represented the most

comprehensive source of information available for describing what is known about newborn screening timeliness.

To examine the barriers identified as contributing to delays in newborn screening for heritable conditions, and strategies being used to address them, we reviewed the advisory committee's 2014 Newborn Screening Timeliness Survey Report, which included findings from a survey of states intended to assist with assessing policies and practices related to the timeliness of newborn screening. The committee, in conjunction with APHL, fielded the survey in the summer of 2014 to identify barriers to and strategies for timely newborn screening, among other things.[6] The survey asked respondents to identify (1) the extent to which certain barriers (identified by newborn screening experts prior to the survey) impacted the newborn screening timeliness in their state, and (2) the strategies that were ongoing in their state to help the newborn screening system meet the recommendations for timely newborn screening. State officials indicated whether a number of barriers for each stage of the newborn screening process identified in the survey had a "major impact," "moderate impact," "minor impact," or "no impact" on timeliness in their state. State officials could also include in written responses additional barriers impacting timeliness not previously identified. The advisory committee obtained survey responses from newborn screening officials in 51 states, although not all states responded to all questions.

We selected certain barriers for which we collected more detailed information from states. To select these barriers, we reviewed state responses in the advisory committee's 2014 survey report, published presentations from APHL's 2016 Newborn Screening and Genetic Testing Symposium, and a 2015 report by the Association of State and Territorial Health Officials. We selected the barriers most frequently indicated by survey respondents as having a major or moderate impact on timeliness, as well as additional barriers that were frequently reported in the survey's written responses, which were grouped into categories in the survey report. We also selected barriers from the survey report that fewer respondents indicated as having a major or moderate impact on

[6]This report was prepared by APHL for the advisory committee as part of a cooperative agreement with HRSA to administer NewSTEPs. We assessed the reliability of information in the survey report by comparing it to related documents and interviewing stakeholders, and we determined the information was sufficiently reliable for our purposes. For more information about the survey, see https://www.aphl.org/AboutAPHL/publications/Documents/NBSTimelinessSuveyReport_10-2014.pdf (accessed April 28, 2016).

timeliness, but which were highlighted by a number of state newborn screening officials in published presentations and reports. We combined barriers that were similar into broader topics. For example, for stage 1, we included barriers related to staff training and turnover into one topic related to staffing issues.

We interviewed officials from four selected states to collect more detailed information on the barriers we selected from the survey report. These selected states were Arizona, Colorado, Minnesota, and Wisconsin. We selected these states because, according to our review of documents from APHL and the Association of State and Territorial Health Officials, they were focusing on activities related to newborn screening timeliness, which would allow them to provide in depth information on barriers and strategies. In addition, these four states' activities related to a range of barriers and strategies. For example, one state focused on improving timeliness for out-of-hospital births, while another state focused on improving its laboratory information management system to provide better feedback to hospitals. Through a combination of written responses and interviews, officials from these states provided more detailed information on how the identified barriers may have contributed to delays in their states, and described strategies they had developed or planned to develop to address these barriers. The results of our review of states are not generalizable to other states, but provided insights on these issues.

In addition to collecting information from states on barriers and strategies for timely newborn screening, we reviewed documents from APHL and the University of Colorado's School of Public Health involved in NewSTEPs and NewSTEPs 360, including NewSTEPs' August 2016 report (which contained information on activities that some states had undertaken to improve timeliness) and interviewed these officials to clarify or elaborate on information about barriers to timely newborn screening and strategies to address such barriers.[7] Similarly we also reviewed information and interviewed newborn screening stakeholders identified by APHL officials and on HRSA's website to elaborate on barriers and strategies. These stakeholders included a member of the advisory

[7]Documents we reviewed to understand states' experiences with barriers to newborn screening timeliness included reports from an initiative called the Collaborative Improvement and Innovation Network. APHL administered this initiative to help improve newborn screening timeliness in eight states. The initiative involved identifying root causes of poor timeliness, creating goals and objectives to guide states' efforts, and developing and sharing strategies to improve timeliness.

committee who co-chaired a timeliness workgroup, a member of the advisory committee who conducted research related to newborn screening barriers, and officials from associations involved in efforts to improve newborn screening (such as the Association of State and Territorial Health Officials and Genetic Alliance).

Appendix II: Heritable and Other Conditions on the Recommended Uniform Screening Panel (RUSP) as of March 2015

Condition	Frequency	Description
Time-critical conditions		
3-Hydroxy-3-Methyglutaric Aciduria (HMG)	<1 in 100,000	Inability to process the amino acid leucine, leading to low blood sugar and accumulations of several organic acids, especially after illness or missed meals. Untreated, can lead to brain damage, mental retardation, coma, and death. Treatment includes a diet low in protein and fat, and high in carbohydrates.
Argininosuccinic Aciduria (ASA)	<1 in 100,000	Buildup of argininosuccinic acid and ultimately ammonia, leading to brain swelling, coma, and sometimes death. Treatment consists of a low-protein diet, frequent meals, medications to prevent ammonia buildup, nutritional supplements, and sometimes a liver transplant.
Beta-Ketothiolase Deficiency (BKT)	<1 in100,000	Periodic episodes of acid buildup, often triggered by illness, which can lead to coma, brain damage, and death. Intravenous treatment to regulate blood sugar and blood acid levels can permit normal development.
Citrullinemia, Type I (CIT)	<1 in 100,000	Buildup of citrulline and ultimately ammonia, which untreated can lead to seizures, coma, brain damage, and death. Treatment with low-protein diet, medications to prevent ammonia buildup, and nutritional supplements to allow normal development.
Classic Galactosemia (GALT)	>1 in 50,000	Lack of the liver enzyme needed to convert galactose, a major sugar in milk, into glucose (blood sugar). Galactose then accumulates in and damages vital organs, leading to blindness, severe mental retardation, infection, and death. Milk and other dairy products must be eliminated from the baby's diet for life. This greatly improves the outlook for affected infants, but risk of mild developmental delays remains.
Congenital adrenal hyperplasia (CAH)	>1 in 25,000	A group of inherited disorders resulting from deficiencies of hormones produced by the adrenal gland. Severe forms of CAH, if undetected and untreated, cause life-threatening salt loss via urine. Treatment includes hormone replacement.
Glutaric Acidemia Type 1 (GA1)	>1 in 75,000	Inadequate levels of an enzyme that helps break down the amino acids lysine, hydroxyl-lysine, and tryptophan, which are building blocks of protein. Often unrecognized for up to 18 months until childhood illness triggers onset of symptoms. Without early diagnosis and prompt treatment when needed, can lead to brain damage, low muscle tone, cerebral palsy-like symptoms, and death.
Holocarboxylase Synthase Deficiency (MCD)	1 in 87,000	A condition in which the body is unable to break down proteins and carbohydrates. People with this condition have trouble using biotin, a vitamin that helps turn certain carbohydrates and proteins into energy for the body. It can lead to a harmful buildup of organic acids and toxins in the body. Early detection and treatment with biotin supplements can prevent the severe outcomes of MCD.
Isovaleric Acidemia (IVA)	<1 in 100,000	Inability to process the amino acid leucine. Can cause coma, brain damage, or death in infancy, or emerge later in childhood after infectious illness. Early diagnosis and treatment with low-protein diet and nutritional supplements allow most children to develop normally.

Condition	Frequency	Description
Long-chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)	>1 in 75,000	Inability to convert certain fats to energy. Symptoms such as feeding difficulties, low blood sugar, and lack of energy can begin soon after birth, and people with this condition may experience heart problems, difficulty breathing, liver failure, and sudden death. Treatment includes a high-carbohydrate, low-fat diet, nutritional supplements, and frequent meals.
Maple Syrup Urine Disease (MSUD)	<1 in 100,000	Genetic metabolic disorder with mild to severe symptoms, which can lead to mental retardation or death. Treatment consists of a special diet, continued indefinitely.
Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	>1 in 25,000	Seemingly well infants suddenly develop seizures due to low blood sugar. People with this condition are at risk of seizures, breathing difficulties, liver problems, brain damage, coma, and sudden death. Treatment includes nutritional supplements and frequent meals.
Methylmalonic Acidemia or methylmalonyl-CoA mutase (MUT)	>1 in 75,000	Defect in processing four amino acids, resulting in illness in first week of life. Severity varies, but death during first month and lifelong brain damage are common. Treatment includes low-protein diet, vitamin B12 injections, and nutritional supplements.
Propionic Acidemia (PROP)	>1 in 75,000	Defect in the processing of four amino acids leading to illness in newborns including brain damage, coma, and death. Even with treatment, which includes a low-protein diet and nutritional supplements, some children have development delays, seizures, increased muscle tone, frequent infections, and heart problems.
Trifunctional Protein Deficiency (TFP)	<1 in 100,000	Seemingly healthy infants can die of what appears to be sudden infant death syndrome. Other infants may develop low muscle tone, seizures, heart failure, and coma, often following illness. Treatment based on frequent meals, a low-fat diet, and nutritional supplements.
Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	>1 in 75,000	Inability to convert certain fats to energy. Unless treated, infants often develop heart and liver failure, dying before age one. Treatment includes a high-carbohydrate, low-fat diet, nutritional supplements, frequent meals, and limiting exercise.
Non-time-critical conditions		
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	>1 in 75,000	Defect in processing the amino acid leucine, leading to brain damage, seizures, liver failure, and infant death, or sometimes no symptoms until adulthood. Symptoms may develop after childhood illness. Treatment includes a low-protein diet.
Biotinidase Deficiency (BIOT)	>1 in 75,000	An inherited disorder resulting in lack of the enzyme that recycles the vitamin biotin. May cause frequent infections, uncoordinated movement, hearing loss, seizures, and mental retardation. Undiagnosed and untreated, can lead to coma and death. If condition is detected soon after birth, problems can be prevented with oral high-dose biotin.
Carnitine Uptake Defect/Carnitine Transport Defect (CUD)	<1 in 100,000	Cells cannot readily absorb carnitine, needed to transfer fatty acids into mitochondria (which supply cells with energy). Results include low blood sugar and sudden death in infancy. Older children may present with progressive heart failure. High-dose carnitine permits normal development.

Condition	Frequency	Description
Classic Phenylketonuria (PKU)	>1 in 25,000	Inability to process the essential amino acid phenylalanine, which accumulates and damages the brain. Can lead to severe mental retardation unless detected soon after birth. Treatment includes a special formula and a low-protein diet, continued indefinitely.
Critical Congenital Heart Disease (CCHD)[a]	18 in 10,000	A group of seven heart defects. Babies born with CCHD are at significant risk of disability or death if not diagnosed soon after birth.
Cystic Fibrosis (CF)	>1 in 5,000	A common inherited disorder, resulting in lung and digestive problems, and death by age 35, on average. Early diagnosis and treatment may improve the growth of babies and children with CF.
Glycogen Storage Disease Type II – Pompe (GSDII)	~1 in 40,000	Babies with Pompe disease have trouble breaking down a large sugar called glycogen. Too much glycogen can keep certain organs and tissues, like the heart and muscles, from working properly. Treatment includes enzyme replacement therapy, physical therapy, and respiratory therapy.
Hb S/Beta-Thalassemia (Hb S/BTh)	>1 in 50,000	Sickle cell disease is an inherited disease of red blood cells. Individuals with sickle cell disease have abnormal hemoglobin, the protein inside red blood cells that carries oxygen to every part of the body. Hb S/BTh is a form of sickle cell anemia, in which the child inherits one sickle cell gene and one gene for beta thalassemia, another inherited anemia. Symptoms are milder than for Hb SS, though severity varies. Routine treatment with penicillin may not be recommended for all affected children
Hb S/C Disease (Hb S/C)	>1 in 25,000	Sickle cell disease is an inherited disease of red blood cells. Individuals with sickle cell disease have abnormal hemoglobin, the protein inside red blood cells that carries oxygen to every part of the body. Hb S/C is another form of sickle cell disease, in which the child inherits one sickle cell gene and one gene for another abnormal type of hemoglobin. Hb S/C tends to be milder than Hb SS; therefore, treatment with penicillin may not be recommended.
Hearing Loss[a]	>1 in 5,000	Without early testing, most babies with hearing loss are not diagnosed until age two or three. By then, they often have delayed speech and language development. Early diagnosis allows use of hearing aids by six months, helping prevent serious speech and language problems.
Homocystinuria (HCY)	<1 in 100,000	Lack of an enzyme that converts the amino acid homocysteine into cystathionine, needed for normal brain development. Untreated, leads to mental retardation, eye problems, skeletal abnormalities, and stroke. Treatment consists of a special diet, one or more vitamins (B6 or B12), and other supplements.
Methylmalonic Acidemia – Cobalamin disorders (Cbl A, B)	<1 in 100,000	Inherited vitamin metabolism defect. Can lead to buildup of acids in blood, brain damage, seizures, paralysis, coma, and death. Treatment includes B12 injections and a low-protein diet.
Primary Congenital Hypothyroidism (CH)	>1 in 5,000	Thyroid hormone deficiency that severely retards growth and brain development. Treatment includes thyroid hormone replacement therapy with dietary restrictions.
Severe Combined Immunodeficiencies (SCID)	>1 in 100,000	A group of rare inherited disorders characterized by defects in two critical immune system cells that are normally mobilized by the body to combat infections. SCID has also been referred to in the popular media as the “bubble boy disease.” Without treatment, infants with SCID are more susceptible to and can develop recurrent infections, leading to failure to thrive and often death.
Sickle Cell Anemia – S, S Disease (Hb SS)	>1 in 5,000	Sickle cell disease is an inherited disease of red blood cells. Individuals with sickle cell disease have abnormal hemoglobin, the protein inside red blood cells that carries oxygen to every part of the body.

Condition	Frequency	Description
Tyrosinemia, Type I (TYR I)	<1 in 100,000	Lack of an enzyme that causes the byproducts of the amino acid tyrosine, particularly a very toxic compound (succinylacetone), to build up in the liver. Fatal liver and kidney failure may result. Treatment includes dietary restrictions and medication to help protect the brain, liver, and kidneys.

Source: GAO analysis of information from the Advisory Committee on Heritable Disorders in Newborns and Children, the American College of Medical Genetics, and March of Dimes. | GAO-17-196

Notes: The Department of Health and Human Services' (HHS) Advisory Committee on Heritable Disorders in Newborns and Children established the RUSP, which is a list of conditions for which newborns should be screened. Nearly all of the conditions on the RUSP are heritable conditions. According to officials from HHS's Health Resources and Services Administration, three conditions—critical congenital heart disease, hearing loss, and primary congenital hyperthyroidism—are not strictly considered heritable conditions.

In 2014, the advisory committee identified 16 of 32 conditions as "time-critical" conditions. These are conditions in which acute symptoms or potentially irreversible damage could develop in the first week of life, and for which early recognition and treatment can reduce the risk of illness and death.

[a]Newborn screening for critical congenital heart disease and hearing loss are performed using point-of-care tests instead of the blood specimen testing used for other conditions.

Appendix III: Comments from the Department of Health & Human Services

DEPARTMENT OF HEALTH & HUMAN SERVICES

OFFICE OF THE SECRETARY

Assistant Secretary for Legislation
Washington, DC 20201

DEC 0 2 2016

Marcia Crosse
Director, Health Care
U.S. Government Accountability Office
441 G Street NW
Washington, DC 20548

Dear Ms. Crosse

Attached are comments on the U.S. Government Accountability Office's (GAO) report entitled, "*Newborn Screening Timeliness: Most States Have Not Met Screening Goals, but Some Are Developing Strategies to Address Barriers*" (GAO-17-196).

The Department appreciates the opportunity to review this report prior to publication.

Sincerely,

Jim R. Esquea
Assistant Secretary for Legislation

Attachment

GENERAL COMMENTS OF THE DEPARTMENT OF HEALTH AND HUMAN SERVICES (HHS) ON THE GOVERNMENT ACCOUNTABILITY OFFICE'S DRAFT REPORT ENTITLED: NEWBORN SCREENING TIMELINESS: MOST STATES HAVE NOT MET SCREENING GOALS, BUT SOME ARE DEVELOPING STRATEGIES TO ADDRESS BARRIERS (GAO-17-196)

The U.S. Department of Health and Human Services (HHS) appreciates the opportunity from the Government Accountability Office (GAO) to review and comment on this draft report. While HHS generally agrees with the data-supported findings in the report, HHS has several significant concerns about the conclusions that GAO has drawn from the findings.

GAO's assessment of whether states screened newborns in a timely manner is based on the targets recommended by the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) in 2015. In a letter to the HHS Secretary, dated April 16, 2015, ACHDNC stated that newborn screening programs would have until 2017 to benchmark their progress in achieving the timeliness recommendations. However, GAO's conclusion that states have not met timeliness goals, per the ACHDNC's recommended target for 2017, is based on analysis of data from 2012 – 2015. Application of this 2017 target to 2012-2015 data is therefore not an accurate assessment of states' efforts to meet the ACHDNC's recommendations. Although this data provides important baseline data, it should not be used as the basis to conclude that most states have not screened newborns in a timely manner. It would be more accurate to conclude that many states are not meeting the ACHDNC's recommendations at this time, rather than reporting that most states do not screen newborns in a timely manner.

The report findings are also limited by not having included point-of-care screening within the definition of newborn screening. For example, there are two conditions on the Recommended Uniform Screening Panel that are point-of-care screening – screening for hearing and critical congenital heart disease. These two conditions follow different screening processes than the ones described on page 6 and pages 24-29, applicable only to testing of newborn bloodspots in laboratories. A report on newborn screening timeliness should include data on point-of-care screening as well.

Appendix IV: GAO Contact and Staff Acknowledgments

GAO Contact

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Staff Acknowledgements

In addition to the contact named above, Kim Yamane (Assistant Director), Hernan Bozzolo (Analyst-in-Charge), Emily Binek, Jazzmin Cooper, Drew Long, Vikki L. Porter, and Jennifer Whitworth made key contributions to this report.