2024 Newborn Screening Program Annual Report

As Required by

Texas Health and Safety Code

Section 33.020

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TEXAS
Health and Human Services

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Table of Contents

Executive Summary

Texas Health and Safety Code, Chapter 33, requires the Texas Department of State Health Services (DSHS) to test every newborn for certain genetic and metabolic disorders. DSHS is authorized to test for all conditions listed on the national Recommended Uniform Screening Panel (RUSP) if funds are available.[1]

Newborn screening is essential to early identification of, and timely treatment for, genetic and metabolic disorders that may have severe consequences for children. The Texas newborn screening (NBS) panel started in 1963 covering a single condition and has grown exponentially. Advances in testing and treatment lead to new conditions being added to the RUSP. As a result, expectations for the NBS program continue to grow. There are currently 63 conditions on the RUSP (37 core conditions and 26 secondary conditions).

Texas screens for 55 disorders through testing at the DSHS laboratory in Austin. Texas also screens for two conditions in the birthing facility: hearing loss and critical congenital heart disease. In 2023, the DSHS laboratory screened approximately 400,000 newborns, with nearly 1,100 testing positive for one of the 55 conditions. By volume, DSHS operates the largest NBS program in the United States.

DSHS is in the process of adding four disorders to the NBS panel: Pompe disease, mucopolysaccharidosis type I (MPS I), mucopolysaccharidosis type II (MPS II), and Krabbe disease. The implementation of these screens is expected to occur in mid-2025. Following these additions, DSHS will work to add screening for ganidinoacetate methyltransferase (GAMT).

The current DSHS Austin laboratory building is operating at full capacity. DSHS has maximized space through renovations and space reallocation. This meticulous space planning has allowed DSHS to meet demand up to now. Without more laboratory space, however, DSHS cannot add more conditions in future years. DSHS anticipates needing a minimum of 46,000 additional square feet of usable space to

[1] The RUSP is a standardized list of disorders that have been evaluated by the Advisory Committee on Heritable Disorders in Newborns and Children and recommended by the U.S. Secretary of Health and Human Services based on the net benefit of adding the screen, the ability of laboratories to screen for the disease, and the availability of treatment. See https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp.

support laboratory needs and meet testing demand over the next 5 years. Most of this growth is attributed to the needs of the NBS program.

DSHS must identify additional permanent space solutions to accommodate future newborn screens as well as other testing needs outside of the NBS program.

Introduction

Overview

Texas Health and Safety Code, Section 33.020, requires DSHS to identify each disorder included in the list of core conditions on the RUSP not included in the state's newborn screening program. DSHS must also identify any additional program capacity needed to:

- Implement the additional NBS test;
- Require each newborn to receive the additional NBS test; and
- Summarize the plan for implementing the additional test.

Further, DSHS must identify any potential barriers to implementation, and the anticipated implementation date of the tests.

Texas first implemented newborn screening in 1963 to identify treatable genetic conditions early and prevent serious complications in babies. Today, the DSHS laboratory screens each newborn for 55 rare, but potentially severe, genetic disorders or medical conditions. Providers collect bloodspot specimens by pricking the heel of a newborn and applying the blood to a special filter paper. Providers then send the specimens to the DSHS laboratory in Austin for testing.[2]

Each Texas newborn is screened twice, for an average of 800,000 screens annually. The first screen should be collected at 24-48 hours after birth, typically at the birthing facility. The second screen is collected at 7-14 days after birth, typically at a primary provider's office.

If a laboratory test is outside of normal range, DSHS clinical care coordination staff will follow up immediately with the newborn's healthcare provider to ensure appropriate follow up for the baby. For NBS conditions, treatment must begin as soon as possible since many of these disorders can cause developmental delays, illness, or even death. Care coordination staff provide diagnostic testing recommendations, next steps, condition information to medical professionals, and

[2] Texas Department of State Health Services. Newborn Screening Disorders. 2024. Accessed May 2024. https://www.dshs.texas.gov/newborn-screening-program/newborn-screening-disorders

support for families. In 2023, DSHS followed up on all 44,540 abnormal results from the laboratory. Approximately 1,100 newborns were determined to have conditions screened for in the newborn screening program. By identifying these babies quickly, the families were given the chance to access providers and life-changing treatments in time to improve the baby's well-being and health outcomes.

Growth in the Last 20 Years

The NBS program grew exponentially in the past twenty years with the advancement of testing equipment and treatment options. In 2003, the NBS program began operations at the recently completed DSHS Austin laboratory building. At the time, the program screened for seven conditions.

Advances in laboratory science have rapidly accelerated the ability of laboratory testing to identify babies who have a genetic condition. One such advance is tandem mass spectrometry, a laboratory technique that identifies the chemical makeup of biomolecules. Tandem mass spectrometry also allows high-volume testing for multiple disorders at once, which has been critical to the addition of disorders to the panel.

At the same time, advances in treatment and therapies have increased the number of conditions that can be treated and, therefore, considered for addition to the RUSP. While some of the original NBS panel conditions triggered critical changes to an infant's diet, more sophisticated treatment types are now receiving federal approval for genetic disorders. Examples of emerging treatment types are:

- Gene therapy, which modifies a person's genes to treat or prevent disease.
- Exon-skipping drugs, which help genes fix themselves by skipping over nonfunctional exons (portions of the genome that are involved in protein synthesis) to improve the body's ability to produce proteins necessary for development and many bodily functions.

In 2006, DSHS increased the number of conditions screened from 7 to 26 using tandem mass spectrometry. DSHS then added 24 more conditions in 2015. Through steady laboratory improvements and infusions of legislative funding, DSHS has built the NBS panel up to 55 conditions screened through laboratory testing.

DSHS also continued to optimize operations of the newborn screening program during this twenty-year period. For example, DSHS moved to a 6-day workweek in 2007. To facilitate more Monday testing, the laboratory initiated Sunday courier services for specimen collection in 2014. Additionally, DSHS increasingly conducted

second-tier testing (confirmatory tests when primary screens have unclear results) to deliver more targeted results for providers and families. DSHS has also continued quality assurance activities to refine the process and prevent false negatives unnecessarily impacting families.

Funding Mechanisms

In 1998, the NBS program implemented testing fees and Medicaid reimbursements to support the construction of the Austin laboratory building. Fees now cover ongoing testing and care coordination costs. Historically, startup funds to add new tests required specific appropriations, which could delay adding new screens by several years.

In 2019, the legislature created the Newborn Screening Preservation Account through Senate Bill 748 (86th Legislature, Regular Session) and through the 2020-2021 General Appropriations Act. When certain fee amounts exceed the amount appropriated in a given fiscal year, DSHS can use those additional funds to add new screens to the screening panel. Funding uses are limited to:

- Costs of offering additional newborn screening tests; and
- Capital assets, equipment, and renovations for the laboratory to ensure the continuous operation of the program.

The Newborn Screening Preservation Account allows DSHS to add screens more quickly and without the need for a specific appropriation to do so. DSHS received the first deposits into the Newborn Screening Preservation Account in 2022. DSHS received approval to use these funds to begin infrastructure planning and implementation of four additional screens.

RUSP Conditions and Planned Implementation

Initial Steps for Adding Conditions

The RUSP is a list of primary and secondary conditions that the federal Health and Human Services (HHS) Secretary recommends states should offer as part of a newborn screening program. The RUSP also serves as the basis for required coverage by certain insurance providers for those screens. The federal HHS Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) analyzes the potential benefit of screening for a particular condition. ACHDNC makes recommendations to the HHS Secretary about whether a condition should be included on the RUSP. Each state then determines which disorders are tested.

According to the Health Resources Services Administration, disorders on the RUSP are chosen based on:

- Evidence that supports the potential net benefit of screening;
- The ability of states to screen for the disorder; and
- The availability of effective treatments.[3]

There are currently 64 conditions on the RUSP, 37 core conditions and 26 secondary conditions. Krabbe disease is the latest addition to the RUSP, effective July 1, 2024.

Texas Health and Safety Code, Section 33.011, requires screening for conditions on the RUSP, as funding allows. Section 33.011 includes two exceptions to this requirement: galactoepimerase deficiency and galactokinase deficiency.

Using Newborn Screening Preservation Account funds, DSHS is in the process of implementing four additional screens for lysosomal storage disorders currently on the RUSP:

- Pompe disease,
- Mucopolysaccharidosis type I (MPS I), and
- Mucopolysaccharidosis type II (MPS II or Hunter syndrome)

[3] See https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp.

- Krabbe disease.

Implementation Process for Adding New Screens

The addition of disorders to the newborn screening panel is a long and complicated process. A variety of factors impact implementation timelines.

Before a condition can be added, the NBS program must:

- Modify space and systems within the existing laboratory;
- Install instruments, which includes verification and validation studies;
- Make updates to the Laboratory Information Management System, or LIMS, which is an IT system that uses screening algorithms to identify abnormal screens for follow up; and
- Hire and train additional staff for both laboratory testing and clinical care coordination.

Typically, this labor and time-intensive process limits DSHS to adding one disorder to the newborn screening panel at a time. However, Pompe, MPS I, MPS II, and infantile Krabbe disease are all lysosomal storage disorders that use the same screening technology. This will allow DSHS to implement the new screens simultaneously.

Timeline for Implementing Recommended Screens

DSHS initially estimated adding Pompe, MPS I, and MPS II conditions to the panel by May 2024. Overall, the DSHS Austin laboratory building needs more retrofitting than originally anticipated to facilitate implementing these conditions. The revised timeline for adding the four conditions is now mid-2025. DSHS intends to add GAMT following this initial implementation.

Several factors contributed to the delay. Existing mass spectrometers had to be replaced first due to the machines nearing end of useful life. This requires removing old equipment and adding new equipment in its place. Chemical fume hoods must be removed and replaced. Only then can the laboratory accommodate additional mass spectrometers to support the new lysosomal storage disorder screens.

The laboratory also needs to add auxiliary equipment for the tandem mass spectrometers supporting the new lysosomal storage disorders before implementing GAMT screening. Before this can be done, sufficient power must be rewired for that area of the building. These activities are all expected to be complete in mid-2025.

Future Additions to the RUSP

DSHS actively evaluates conditions that could be added to the RUSP in the future. This monitoring has allowed DSHS to proactively plan for space repurposing, reconfiguration, and renovations. DSHS also monitors for whether current staff levels and the six-day work week will remain sufficient if specific screenings are added to the recommended panel.

The ACHDNC is considering recommendations to add metachromatic leukodystrophy and Duchenne muscular dystrophy. Additionally, other conditions are earlier in the pipeline for the committee's consideration.

Metachromatic leukodystrophy is an autosomal recessive lysosomal disorder, which can lead to progressive loss of function to the brain and nervous system. This disorder can present in infants, children, and adults, with the potential for more severe and immediate outcomes for younger patients. Current incidence estimates range from 1 in every 40,000 to 1,160,000 individuals.

Duchenne muscular dystrophy is a genetic condition that causes progressive muscular degeneration and weakness. Duchenne most commonly occurs in boys, at an incidence of about 1 in 3,600 baby boys.

If these or other conditions are added to the RUSP, the DSHS laboratory cannot accommodate any additional testing equipment needed to implement the new screens. Given the current trajectory, DSHS will need additional laboratory space to keep Texas newborn screening on par with the national recommendations and other states.

Barriers to Additional Implementation

DSHS works to add all RUSP conditions to the newborn screening program, and the Newborn Screening Preservation Account has been a key component in facilitating this effort. However, DSHS needs to address obstacles that prevent the NBS program from growing and adapting to the current list of RUSP conditions. The foremost limitation is lack of laboratory space. At the same time, DSHS has identified opportunities to further harness the Newborn Screening Preservation Account.

Current Laboratory Space Limitations

The DSHS Austin laboratory building serves as a crucial component to responding to public health needs. It processes over 1.3 million specimens each year. That number is expected to grow due to new federal water testing requirements and increased testing volumes. The demand for next generation sequencing, which provides confirmatory testing for newborn screening and advanced testing to detect variants and antibiotic resistance will also continue to grow, further increasing the number of specimens processed annually.

Completed in 2002, the Austin laboratory building was partially finished when DSHS staff moved in. The seventh floor was eventually completed, allowing for greater space use. The building now facilitates testing across various public health needs, including infectious disease, foodborne illness, biological and chemical threats, federal water testing, and core sequencing (see Appendix A). Certain tests require the use of the Biosafety Level 3 lab areas for potentially highly infectious specimens.[4]

Through meticulous space planning, DSHS has fully maximized 100 percent of the 119,600 square feet of usable space in the Austin laboratory building. DSHS has extensively reconfigured the building, with equipment and supplies spilling over into walkways. The newborn screening program currently occupies 25 percent of the

[4] There are four biosafety lab levels, with Level 4 reserved for the highest-risk specimens. Biosafety Level 3 laboratories require specific facility construction, laboratory precautions, and airflow requirements to maintain the safety of the testing environment and surrounding community.

Austin laboratory building while accounting for over 60 percent of specimens processed each year.

Testing areas, including newborn screening, must serve as multipurpose spaces, juggling the high-volume intake of specimens and testing supplies, including reagents that may require safe handling. Testing areas are also subject to constant upheaval in the form of equipment repairs, workarounds for building maintenance issues, and losing space due to other more pressing space reallocation needs. For example, DSHS has had to reallocate space from other areas to support the newborn screening panel. Combined, these space issues pose a challenge to ensuring testing areas are safe for laboratory staff.

DSHS has worked to identify projected laboratory space needs over the next five years across all testing disciplines. DSHS needs at least 165,000 square feet of usable laboratory space to keep up with testing demand, including for newborn screening. This equates to 46,000 square feet, or 39 percent more space than the existing laboratory footprint. (See Appendix A).

Much of the exponential growth in laboratory space is driven by anticipated needs for the NBS program. DSHS estimates the space needs for the program will grow by 65 percent beyond the existing available space in the next five years. As federal HHS adds more conditions to the RUSP due to advancements in treatment availability, space needs will increase.

The DSHS 2026-2027 Legislative Appropriations Request will include an exceptional item for identifying a permanent solution for additional laboratory space. The item includes a placeholder amount for construction costs; DSHS is working to refine final cost estimates. Of note, the NBS program otherwise remains self-sufficient. Once a screen is implemented, the program is self-supporting through Medicaid revenue and private pay fees.

Opportunities for the Newborn Screening Preservation Account

In fiscal year 2022, the Comptroller of Public Accounts (CPA) provided a finding of fact allowing $26.8 million to be deposited in the Newborn Screening Preservation Account.

In November 2023, DSHS requested an additional finding of fact from the CPA allowing for the reallocation of previously obligated funds related to newborn

screening. The obligated funds were related to a lapsed contract with unused funds from a previous fiscal year. The current Newborn Screening Preservation Account statute does not allow for obligating funds other than for excess fee revenue generated during the most recent fiscal year. Therefore, the lapsed contract funds did not meet the statutory definition and were inaccessible to DSHS to support activities related to adding new conditions to the newborn screening panel. Resolving this limitation will require an amendment to the account's statute, Chapter 33 of the Texas Health and Safety Code, Chapter 33, Subchapter D. Additionally, the statute limits capital uses of the account to renovations of existing laboratory space.

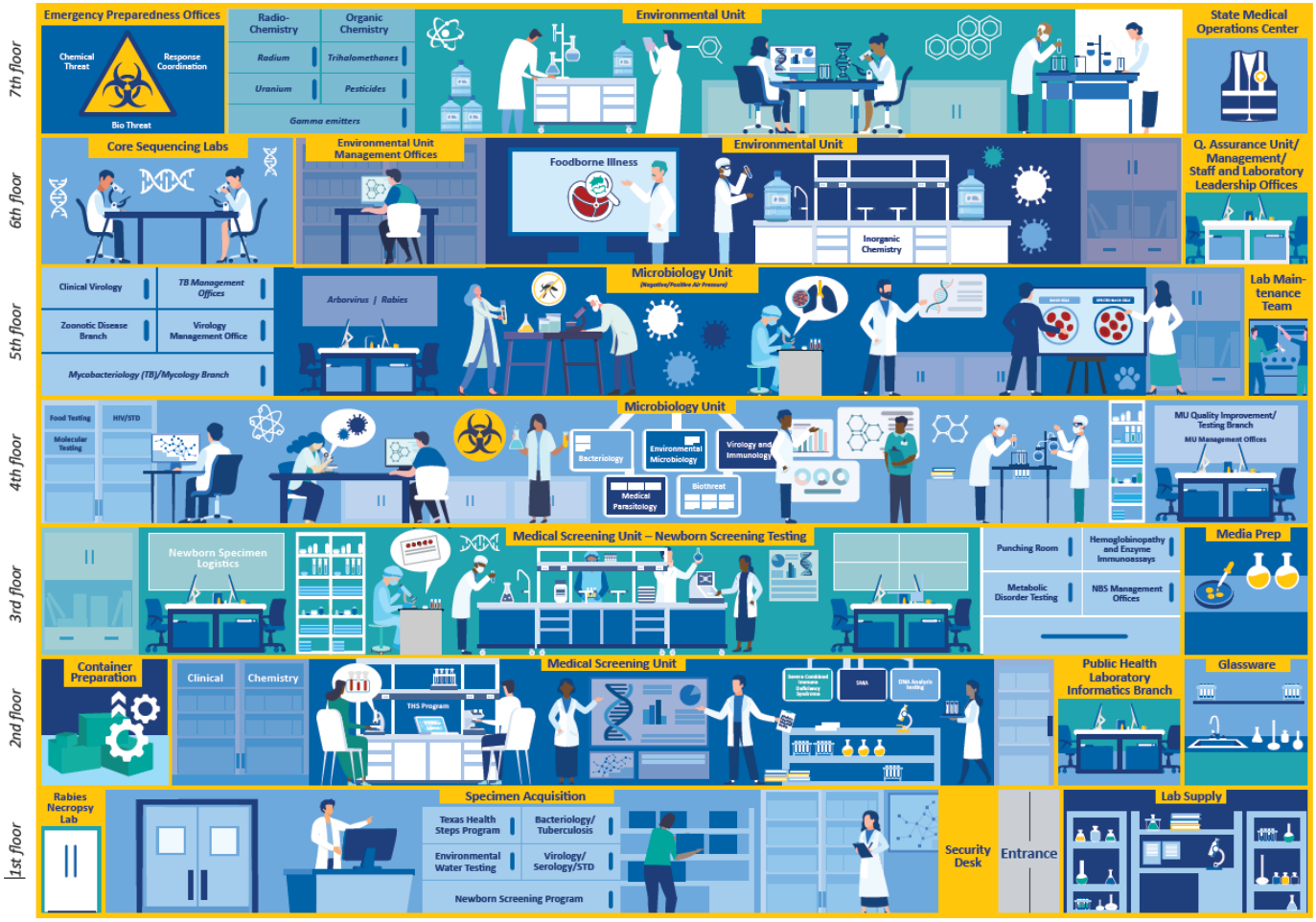
Conclusion

As DSHS works to implement additional newborn screening tests, challenges will continue to arise with laboratory space needs. DSHS is including an exceptional item in its 2026-2027 Legislative Appropriations Request for additional laboratory space. In the meantime, DSHS continues refining its understanding of the long-term physical space needs for the newborn screening program and the other critical public health testing needs provided by the DSHS Austin laboratory.

The previously utilized method of continuous space reallocation within the DSHS Austin laboratory, and the newborn screening area in particular, cannot counteract the projected need for space. DSHS anticipates needing a minimum of 46,000 square feet in additional testing space, or 39 percent more than what is currently available within the existing laboratory footprint, in the next five years. Most of this growth is attributed to the needs of the newborn screening program.

As the state continues to grow, the volume of work needed to ensure timely newborn screening and coordinated follow up will continue to increase. DSHS will need the resources necessary to keep up with this demand.

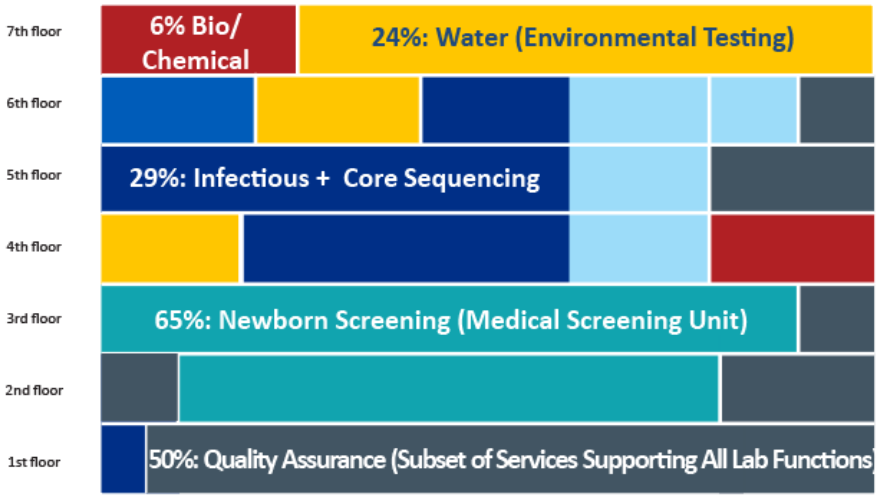
Appendix A. Austin Laboratory Footprint and Projected Growth



The newborn screening program utilizes the majority of the 2nd and 3rd floors of the Austin Laboratory building (Medical Screening Unit areas). The program also utilizes support services located on the 1st, 2nd, 3rd, 5th, and 6th floors.

Current and Projected Space in the Austin Laboratory Building

Projected Percentage Space Growth Over the Next 5 Years



- **Infectious Disease:** *Floors 1, 4, 5, and 6*
- **Foodborne Illness:** *Floors 4, 5, and 6*
- **Water:** *Floors 4, 6, and 7*
- **Newborn Screening:** *Floors 2 and 3*
- **Biological & Chemical Threats:** *Floors 4 and 7*
- **Core Sequencing:** *Floor 6*

Newborn Screening:
46.7% growth in number of instruments to implement existing and current RUSP conditions

Core Sequencing:
Rapid increase in utilization – projecting **212% increase in next 5 years** – will need 100% more space to meet this need

Water:
New EPA PFAS requirements – **24% expansion in water testing over the next 5 years**

Quality Assurance:
50% projected growth in the next 5 years – outpacing current operations that are split between on-site & remote

Space Utilization Overview

Austin Laboratory usable space:
119,600 sq. ft.

Current usable space utilization:
119,600 sq. ft.

5-year projected usable space need:
165,000 sq. ft. for equipment and some storage

Other Considerations

- Additional analysis is needed for efficient use of space for
  - storage for reagents, supplies, and specimens
  - HVAC, IT, and maintenance needs
- Some staff are already housed off-site or work remotely: operations, quality assurance, etc.

Current Estimate*

$328.3 Million
for new laboratory space

**As of September 2024*