North Carolina Department of Health and Human Services' Newborn Screening Program

G.S. 130A-125(e)

Report to the

House Appropriations Committee on Health and Human Services

Senate Appropriations Committee on Health and Human Services

Fiscal Research Division

By

North Carolina Department of Health and Human Services

February 28, 2020

Reporting Requirement

North Carolina General Statute130A-125(e) requires the Department of Health and Human Services (DHHS) to annually report on March 1 to the House Appropriations Committee on Health and Human Services, the Senate Appropriations Committee on Health and Human Services, and the Fiscal Research Division on the DHHS Newborn Screening Program.

The report shall include the following information for the preceding fiscal year:

(1) A description of the services funded by the Newborn Screening Program, including a description of the Department's activities with respect to each of the services listed in GS § 130A-125(a).
(2) A detailed budget and list of expenditures for the Newborn Screening Program, including all positions funded.
(3) Fees and other receipts collected for the Newborn Screening Program.
(4) Projected fees and other receipts for the Newborn Screening Program for the current and upcoming fiscal year.
(5) Any condition the Department anticipates will be listed on the RUSP within the current or upcoming fiscal year and a description of the following:
    a. Any laboratory instruments or equipment the Department will need to purchase in order to perform screening for that condition.
    b. Any additional positions the Department will need to establish in order to perform screening for that condition.
(6) The balance in the Newborn Screening Equipment Replacement and Acquisition Fund as of the preceding June 30.
(7) Amounts credited to the Fund.
(8) Amounts expended from the Fund and the purposes of the expenditures.
(9) Proposed expenditures of the monies in the Fund for the current and upcoming fiscal year.
(10) Any other information the Department deems relevant to maintaining the Newborn Screening Program as a fee-supported program.

Introduction and Background

Newborn screening (NBS) is a comprehensive, coordinated system consisting of education, screening, follow-up contact, diagnosis, treatment and management, and program evaluation designed to identify newborns at risk for rare and potentially fatal conditions that aren't otherwise apparent at birth. NBS for biochemical and genetic disorders involves a small heel prick to collect a few drops of blood on a filter paper card within the first 24-48 hours of life. This card is sent to a laboratory where scientists look for signs of these serious conditions.

In North Carolina, the newborn screening program began in 1966 as a voluntary effort, testing for only one disorder, phenylketonuria (PKU). The state’s program became a legislative mandate in 1991 with the passage of "An Act to Establish a Newborn Screening Program."

Through the screening test and subsequent diagnostic evaluation, physicians can determine whether newborns have certain conditions or disorders that eventually could cause health problems. Although these conditions are rare, and most babies have normal screening results, early diagnosis and proper treatment can make the difference between life-long impairment and healthy development.

Services Funded by the Newborn Screening (NBS) Program

The following are services funded by the NBS Program and activities completed by the DHHS Division of Public Health (DPH) for each service as defined in General Statute 130A-125 (a):

- *Development and distribution of educational materials regarding the availability and benefits of newborn screening*

    - The brochure "A Test to Save Your Baby's Life" describes the newborn metabolic screening program, and is translated into English, Spanish, Hmong, Mandarin, and Arabic. It is available online and is distributed at the time of birth.
    - The flyer "North Carolina Newborn Screening Program" describes the NBS program. It is available online and is disseminated via targeted distribution to obstetric/prenatal practices.
    - Syndrome-specific information is provided in conjunction with the reporting of abnormal results to parents and providers. Examples include education pamphlets for Cystic Fibrosis and Severe Combined Immunodeficiency (SCID).
    - The brochure "My Baby's Hearing Screening" describes what to expect from newborn hearing screening, and is translated into English, Spanish, Arabic, Chinese, French, Hmong, Korean and Vietnamese. It, too, is available online and distributed by birthing facilities.

- *Provision of laboratory testing*

The State Laboratory of Public Health (SLPH) provides all laboratory services for the newborn screening (NBS) program for conditions on the North Carolina Newborn Screening Program Panel, in accordance with the Recommended Universal Screening Panel (RUSP) from the Secretary of the United States Department of Health and Human Services.

As of this report, SLPH provides screening for the following core conditions on the RUSP:

Amino Acid Disorders

- Argininosuccinic Aciduria
- Citrullinemia, Type I
- Classic Phenylketonuria
- Homocystinuria
- Maple Syrup Urine Disease
- Tyrosinemia, Type I

Endocrine Disorders
- Congenital Adrenal Hyperplasia
- Primary Congenital Hypothyroidism

Fatty Acid Oxidation Disorders
- Carnitine Uptake Defect
- Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency
- Medium-Chain Acyl-CoA Dehydrogenase Deficiency
- Trifunctional Protein Deficiency
- Very Long-Chain Acyl-CoA Dehydrogenase Deficiency

Hemoglobin Disorders
- S, Beta-Thalassemia
- S, C Disease
- Sickle Cell Anemia

Organic Acid Conditions
- 3-Hydroxy-3-Methylglutaric Aciduria
- 3-Methylcrotonyl-CoA Carboxylase Deficiency
- Beta-Ketothiolase Deficiency
- Glutaric Acidemia, Type I
- Holocarboxylase Synthetase Deficiency
- Isovaleric Acidemia
- Methylmalonic Acidemia (Cobalamin Disorders)
- Methylmalonic Acidemia (Methymalonyl-CoA Mutase Deficiency)
- Propionic Acidemia

Other Disorders
- Biotinidase Deficiency
- Classic Galactosemia
- Cystic Fibrosis
- Severe Combined Immunodeficiency

As of this report, SLPH provides screening for the following secondary conditions on the RUSP:

Amino Acid Disorders
- Argininemia
- Benign Hyperphenylalaninemia
- Biopterin defect in cofactor biosynthesis
- Biopterin defect in cofactor regeneration
- Citrullinemia, type II
- Hypermethioninemia
- Tyrosinemia, Type II
- Tyrosinemia, Type III

Fatty Acid Oxidation Disorders

- Carnitine Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyltransferase Type II Deficiency
- Glutaric Acidemia, Type II
- Short-Chain Acyl-CoA Dehydrogenase Deficiency
- Medium-chain ketoacyl-CoA thiolase deficiency

Hemoglobin Disorders

- Various other hemoglobinopathies

Organic Acid Conditions

- 2-Methylbutyrylglycinuria
- 2-Methyl-3-hydroxybutyric aciduria
- 3-Methylglutaconic aciduria
- Isobutyrylglycinuria
- Malonic acidemia
- Methylmalonic Acidemia with Homocystinuria

Other Disorders

- Galactoepimerase deficiency
- Galactokinase deficiency
- T-cell related lymphocyte deficiencies

NC NBS is working towards implementation of the following core conditions on the RUSP for which SLPH does not currently screen:

- Glycogen Storage Disease Type II (Pompe)
- Mucopolysaccharidosis Type 1
- X-linked Adrenoleukodystrophy
- Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1

The process to onboard screening for additional disorders is complex as newborn screening is more than a simple test. The public health laboratory testing is a critical, core component, but it is just one piece of a broader public health system working for families. Every state newborn screening program has six essential parts: screening, follow-up, diagnosis, management, evaluation, and education, and the Department must still ensure that the full system is in place before screening can begin.

- *Development of follow-up protocols to assure early treatment for identified children, and the provision of genetic counseling and support services for the families of identified children*

  - As disorders are added to the NC NBS panel, follow-up protocols are developed from a literature review with subspecialist input and are approved by the NC NBS Advisory Committee.
  - Subspecialist and genetics resource lists are faxed to the provider at the time of an abnormal result report and include test results and recommendations.

- *Provision of necessary dietary treatment products or medications for identified children as medically indicated and when not otherwise available*

    - Orders for specialized metabolic formulas are initiated by a tertiary care center in North Carolina caring for individuals without third-party coverage of these products and sent to the DHHS Division of Public Health Nutrition Services Branch.
    - Specialized metabolic formulas are ordered by Nutrition Services Branch directly from the manufacturer, shipped to local WIC agencies, and picked up by families or individuals for whom the products were ordered.

- *For each newborn, provision of physiological screening in each ear for the presence of permanent hearing loss*

    - Medical facilities that provide birthing or inpatient neonatal services are required to physiologically screen each newborn in each ear for the presence of permanent hearing loss before the infant is discharged from the medical facility, unless medical complications prevent screening.
    - Any physician that attends a newborn within 30 days of birth and determines that the newborn has not been physiologically screened in each ear for the presence of permanent hearing loss is required to refer the patient for such screening within 30 days of birth or as soon as is practical.
    - All persons performing physiologic hearing screenings for infants less than six months of age and/or diagnostic auditory evaluations and amplification selections for infants less than twelve months of age are required to report the results of these screenings, evaluations, or selections to DPH within five days.
    - Infants who do not pass the initial newborn hearing screening should receive an outpatient re-screening by one month of age, unless there are known risk factors for permanent hearing loss, and they are referred directly for a diagnostic auditory evaluation.
    - Infants who do not pass the outpatient re-screening should be referred to a pediatric audiologist for diagnostic auditory evaluation no later than three months of age.
    - Infants who are diagnosed with permanent hearing loss should be fitted with amplification devices (if appropriate) and referred to early intervention services no later than six months of age.

- *For each newborn, provision of pulse oximetry screening to detect congenital heart defects*

    - Critical Congenital Heart Defect screening and evaluation of positive or abnormal screens are required before discharge from the care of the attending provider of the neonate or infant.
    - All medical facilities and attending providers of the neonate or infant are required to have and implement a written protocol for evaluation and follow up of positive screenings.
    - Evaluation and follow up should occur as soon as possible (but no later than 24 hours after obtaining a positive screening) and should follow most current

published recommendations from the American Academy of Pediatrics and the American Heart Association.

Detailed budget and list of expenditures for the Newborn Screening Program, including all positions funded

The North Carolina Newborn Screening Program cost approximately $8 million to operate in SFY 2018-19 and collected approximately $13.67 million in receipts.

Newborn Screening SFY 2018-19										
	State Lab/IT		Follow up Program Children		Equipment Fund		Indirect Cost		Total Newborn Screen Program	
Expenditure Description	Budget	Expended	Budget	Expended	Budget	Expended	Budget	Expended	Budget	Expended
Personal Services*	$2,289,670.00	$1,464,193.18	$ 378,833.00	$ 152,621.87	$ -	$ -			$ 2,668,503.00	$ 1,616,815.05
Purchased Services**	$2,737,474.00	$1,178,064.83	$ 24,855.00	$ -	$ -	$ -	$ 2,691,993.00	$ 1,802,229.66	$ 5,454,322.00	$ 2,980,294.49
Supplies	$3,527,868.00	$2,034,661.44	$ 2,936.00	$ 19.99	$ -	$ -			$ 3,530,804.00	$ 2,034,681.43
Property, Plant & Equipment	$ 185,265.00	$ 25,840.50	$ 7,605.00	$ 3,837.00	$ 3,700,000.00	$ 329,285.66			$ 3,892,870.00	$ 358,963.16
Other Expenses & Adj	$1,001,902.00	$1,001,901.98	$ -	$ -	$ -	$ -			$ 1,001,902.00	$ 1,001,901.98
Aid & Public Assistance	$ -	$ -	$ 225,000.00	$ -	$ -	$ -			$ 225,000.00	$ -
Total Expenditures	**$9,742,179.00**	**$5,704,661.93**	**$639,229.00**	**$156,478.86**	**$3,700,000.00**	**$ 329,285.66**	**$2,691,993.00**	**$1,802,229.66**	**$ 16,773,401.00**	**$ 7,992,656.11**
Revenue Decription	Budget	Collected	Budget	Collected	Budget	Collected	Budget	Collected	Budget	Collected
DPS Transfer for Hurrican Florence***		$ 291.05							$ -	$ 291.05
Other License, Fees/Permits****	$ 24,424.00	$ -							$ 24,424.00	$ -
Newborn Screening Fees*****	$8,118,809.00	$8,322,779.81	$ 572,326.00	$ 156,478.86	$3,700,000.00	$2,926,351.20	$2,691,993.00	$1,802,229.66	$ 15,083,128.00	$ 13,207,839.53
Accts Receivable Interest	$ -	$ 10,135.43							$ -	$ 10,135.43
Accts Receivable Penalry	$ -	$ 150,895.39							$ -	$ 150,895.39
Prior State Carryforward	$1,000,000.00	$ 100,000.00							$ 1,000,000.00	$ 100,000.00
DMA Medicaid Title X******	$ 598,682.00	$ 209,094.76	$ 66,903.00						$ 665,585.00	$ 209,094.76
Total Revenues	**$9,741,915.00**	**$8,793,196.44**	**$639,229.00**	**$ 156,478.86**	**$3,700,000.00**	**$2,926,351.20**	**$2,691,993.00**	**$1,802,229.66**	**$ 16,773,137.00**	**$ 13,677,965.11**
*Expenditures Incurred for services rencered by permanent employees and the related fringe benefits										
**Expenditures incurred for services required to ensure the ongoing operation of State government facilities and government services, ie. Operational services, maintenance agreements, travel, etc.										
***Receipts received from FEMA for Hurricane Florence related overtime										
****Fees form workshops										
*****Collected per NC General Statute 147-86.23										
******Medicaid receipts from non-hospital repeat NBS samples										

There are 36 positions in the NBS Program funded in SFY 2019-2020.

Positions Funded SFY 2019-2020	
State Laboratory	**Budgeted Salary**
Chemistry Manager II	$95,673
Public Health Scientist	$79,950
Medical Laboratory Supervisor I	$74,411
Public Health Scientist	$74,128
Chemistry Supervisor II	$72,775
Chemist II	$64,932
Laboratory Medical Specialist	$59,405
Chemist I	$58,212
Laboratory Improvement Consultant	$56,629
Chemist I	$56,375
Laboratory Improvement Consultant	$53,011
Laboratory Medical Specialist	$52,616
Laboratory Medical Specialist	$51,250
Chemist I	$47,358
Medical Laboratory Technologist II	$46,101
Medical Laboratory Technologist II	$45,100
Medical Laboratory Technologist II	$44,946
Medical Laboratory Technologist II	$44,837
Medical Laboratory Technologist II	$44,130
Medical Laboratory Technologist I	$42,866
Medical Laboratory Technologist I	$42,427
Chemistry Technician II	$42,248
Medical Laboratory Technologist I	$42,007
Medical Laboratory Technologist I	$41,295
Medical Laboratory Technologist I	$41,295
Medical Laboratory Technologist I	$40,805
Administrative Supervisor	$36,934
Medical Laboratory Technologist I	$36,830

Chemistry Technician I	$35,505
Medical Laboratory Technician	$31,200
Children's Health	**Budgeted Salary**
Public Health Genetic Counselor	$63,728
Public Health Educator III	$47,650
Social Worker	$45,000
Social Worker	$52,477
Social Worker	$52,000
Information Technology	**Budgeted Salary**
Application Systems Specialist	$100,748

Fees and other receipts collected for the Newborn Screening Program, and projected fees and other receipts for the Newborn Screening Program for the current and upcoming fiscal year

Fund	SFY 2019-20 Fees & Other Receipts Received Year-to-Date (as of February 14, 2020)	Estimated SFY 2019-20 Fees & Other Receipts	Projected SFY 2020-21 Fees & Other Receipts
NBS	**$ 7,157,269.86**	**$11,486,383.88**	**$11,640,000**
NBS Equipment Replacement and Acquisition Fund	**$ 2,285,067.58**	**$3,666,551.00**	**$3,700,000**
Total	**$9,442,337.44**	**$15,152,934.88**	**$15,340,000**

Increased receipts reflect the change in the NBS fee approved by the General Assembly to keep the Newborn Screening Program receipt supported. The authorized fee increase in advance of new disorder implementation was critical to address a historical structural budget deficit in newborn screening funding and build the infrastructure to initiate and ultimately support screening for new conditions. The Department has used the additional funds authorized by the 2018 legislation to:

- Hire essential staff
  - In the SLPH to develop, validate, and implement testing strategies to identify the three new disorders (Pompe, MPS-I, and X-ALD),
  - In the follow-up program to enhance follow-up services for existing disorders and provide additional services for the three new disorders, and
  - On the information technology team to configure the program's information management system to include the three new disorders, enable electronic

reporting, and perform data migration to the latest version of the existing software,

- Acquire instrumentation that will support new disorder testing, and
- Upgrade software and equipment for program quality and process improvement initiatives.

In addition, DHHS is procuring the necessary equipment and supplies to implement testing for Pompe, MPS-I, and X-ALD through an open-competitive bid process. It is anticipated that onboarding the testing for additional RUSP conditions will utilize the majority of the projected receipts that are greater than prior year expenditures for the NBS Program. Once procured, SLPH will begin the regulatory-required validation processes to identify the new disorders, finalize protocols for ensuring newborns who screen positive are linked to appropriate care, and configure the program's information management system to collect data and electronically report results. The Department anticipates this process will result in implementation of screening for the three new disorders in mid-2021.

North Carolina's progress in implementing these screenings is on pace with the timeline of rollouts in other states. Data from the national Newborn Screening Technical Assistance and Evaluation Program (NewSTEPs) denotes that states who had already onboarded screening for Pompe, MPS-I, and X-ALD required, on average, 26 to 45 months to implement screening. This range in time-to-implementation is based on the specific condition(s) for which the individual states began screening. DHHS is coordinating with colleagues in other states and using national resources like those available from the Association of Public Health Laboratories (APHL) to learn from those states who have already implemented screening and assure our implementation process for the same three disorders is on the lower end of these national time-to-implementation statistics.

Conditions the Department anticipates will be listed on the RUSP within the current or upcoming fiscal year

The Department is not aware of additional conditions that are likely to be added to the RUSP during the current or upcoming fiscal year.

Spinal Muscular Atrophy (SMA) was added to the RUSP last fiscal year and implementation is beginning. The Department is preparing the requisite documents for required rulemaking to add SMA to the state's newborn screening panel. DHHS expects to present the draft rule and fiscal note to the Commission for Public Health at its May 6, 2020 meeting for approval to proceed with formal rulemaking. DHHS anticipates the screening for SMA will begin in North Carolina in 2021. Resources anticipated to bring this test online are:

- Equipment: Detailed in the tables below
- Positions: 2 Full-Time Equivalent Medical Laboratory Specialist; approximately $75,000 annual salary and fringes for each

Balance in the Newborn Screening Equipment Replacement and Acquisition Fund as of the preceding June 30

The beginning balance in the Fund from June 30, 2019, was $2,597,065.54.

Amounts credited to the Fund

As of February 14, 2020, $2,285,067 had been credited to the Fund.

Amounts expended from the Fund and the purposes of the expenditures

In SFY 2019, $329,285.66 from the Fund was expended on items in the following chart:

Instrument upgrades to support screening for Severe Combined Immunodeficiencies	$319,584.53
Laboratory temperature/humidity monitors required for regulatory compliance	$701.13
Instrument upgrades to support screening for Cystic Fibrosis	$9,000.00

For SFY 2020, as of January 3, 2020, $515,184.32 has been either expended or encumbered on items in the following chart:

Small equipment to support molecular biology testing	$430.00
Instrument upgrades to support screening for Sickle Cell Disease and other hemoglobinopathies	$26,800.00
Upgrades to laboratory infrastructure to provide adequate environmental humidification required for regulatory compliance	$132,500.00
New instrumentation in support of future addition of Spinal Muscular Atrophy screening	$152,971.32
New instrumentation to expand genetic screening for Cystic Fibrosis	$202,483.00

Proposed expenditures of the monies in the Fund for the current and upcoming fiscal year

The SLPH anticipates the following expenditures and approximate costs totaling $5,745,000 from the NBS Equipment Replacement and Acquisition Fund over the current and subsequent state fiscal years (see below).

SFY 2020

Additional instrumentation in support of screening for multiple endocrine and metabolic disorders	$310,000
Tandem mass spectrometry instrumentation in support of future addition of screening for three disorders required by G.S. 130A-125	$1,350,000

Instrument upgrades to support screening for amino acid, organic acid, and fatty acid oxidation disorders	$310,000
Infrastructure upgrades to support tandem mass spectrometry instrumentation	$590,000
Information Technology upgrades to support genetic screening for Cystic Fibrosis	$150,000
TOTAL PROPOSED EXPENDITURES	**$2,710,000**

SFY 2021

Instrument replacements to support screening for amino acid, organic acid, and fatty acid oxidation disorders	$2,100,000
Infrastructure upgrades to support new instrumentation	$185,000
New instrumentation in support of future addition of Spinal Muscular Atrophy screening	$350,000
Information Technology upgrades to support new disorder expansion	$400,000
TOTAL PROPOSED EXPENDITURES	**$3,035,000**

Other information relevant to maintaining the Newborn Screening Program as a fee-supported program

Newborn screening resides in an ever-changing and complicated environment that forces states to constantly adapt to new circumstances, ranging from advances in technology and treatments to new socio-political, economic, or ethical developments. The challenges NC and other states are facing in newborn screening are well documented and may be summarized as follows:

- New technologies will radically change our ability to cost-effectively screen for many conditions at once. Basic science discoveries will continue to identify markers of new disorders. These advances could further add to the list of thousands of known and potentially screenable disorders.
- Disorder therapies will continue to evolve, transforming previously “untreatable” disorders into conditions that can be treated or cured. New treatments are likely to be highly effective but very expensive, raising fundamental cost-benefit and cost-efficacy questions. States will have to figure out how to establish care protocols, networks, and reimbursement mechanisms so they can meet the ethical obligation to treat every baby identified through newborn screening.
- Updating NBS administrative rules to include new conditions, establishing new positions and hiring personnel to implement screening for new conditions, and increasing the NBS fee to support screening for new conditions is a lengthy process.

Newborn screening is a highly successful public health program. Despite this success, many challenges are emerging that question the viability of newborn screening as we know it today. Fortunately, North Carolina has a wealth of public and private partners committed to advancing newborn screening policy and practice, an objective that will require systematic collaboration with the goal of maximizing the health of young children and the well-being of families.