

**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

March 1, 2019

Reporting Requirement

GS § 130A-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 the practice of testing every newborn for certain harmful or potentially fatal disorders that aren't otherwise apparent at birth. It begins with a small heel prick to collect a few drops of blood on a filter paper card within the first 24-48 hours of life. The card is sent to a laboratory where scientists look for signs of rare but serious conditions that might not be noticeable by parents or even the baby's health care provider.

Through the blood test, physicians often can determine whether newborns have certain conditions or disorders that eventually could cause health problems. Although these conditions are rare, and most babies are given a clean bill of health, early diagnosis and proper treatment sometimes can make the difference between life-long impairment and healthy development.

NBS is more than a series of tests. It's a closely coordinated system that involves maternal and child health providers, public health laboratories and geneticists, all working together to protect the baby's health.

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 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 each condition that is listed on the Recommended Universal Screening Panel (RUSP) by the Secretary of the United States Department of Health and Human Services and the Advisory Committee on Heritable Disorders of Newborns and Children.

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

Amino Acid Disorders

- Argininosuccinic Aciduria (**ASA**)
- Benign Hyperphenylalaninemia (**H-PHE**)
- Citrullinemia, Type I (**CIT**)
- Classic Phenylketonuria (**PKU**)
- Homocystinuria (**HCY**)
- Maple Syrup Urine Disease (**MSUD**)
- Tyrosinemia, Type I (**TYR I**)
- Tyrosinemia, Type II (**TYR II**)
- Tyrosinemia, Type III (**TYR III**)

Endocrine Disorders

- Congenital Adrenal Hyperplasia (**CAH**)
- Primary Congenital Hypothyroidism (**CH**)

Fatty Acid Oxidation Disorders

- Carnitine Acylcarnitine Translocase Deficiency (**CACT**)
- Carnitine Palmitoyltransferase Type II Deficiency (**CPT-II**)
- Carnitine Uptake Defect (**CUD**)
- Glutaric Acidemia, Type II (**GA-2**)
- Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (**LCHAD**)
- Medium-Chain Acyl-CoA Dehydrogenase Deficiency (**MCAD**)
- Short-Chain Acyl-CoA Dehydrogenase Deficiency (**SCAD**)
- Trifunctional Protein Deficiency (**TFP**)
- Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (**VLCAD**)

Hemoglobin Disorders

- Hemoglobinopathies (**Var Hb**)
- S, Beta-Thalassemia (**Hb S/βTh**)
- S, C Disease (**Hb S/C**)
- Sickle Cell Anemia (**Hb SS**)

Organic Acid Conditions

- 2-Methylbutyrylglycinuria (**2MBG**)
- 3-Hydroxy-3-Methylglutaric Aciduria (**HMG**)
- 3-Methylcrotonyl-CoA Carboxylase Deficiency (**3-MCC**)
- Beta-Ketothiolase Deficiency (**BKT**)
- Glutaric Acidemia, Type I (**GA-1**)
- Holocarboxylase Synthetase Deficiency (**MCD**)
- Isobutyrylglycinuria (**IBG**)
- Isovaleric Acidemia (**IVA**)
- Methylmalonic Acidemia (Cobalamin Disorders) (**Cbl A, B**)
- Methylmalonic Acidemia (Methylmalonyl-CoA Mutase Deficiency) (**MUT**)
- Methylmalonic Acidemia with Homocystinuria (**Cbl C, D, F**)
- Propionic Acidemia (**PROP**)

Other Disorders

- Biotinidase Deficiency (**BIOT**)
- Classic Galactosemia (**GALT**)
- Cystic Fibrosis (**CF**)
- Severe Combined Immunodeficiency (**SCID**)

The following conditions were recently added on the RUSP and have not yet been implemented on the NC NBS panel as of this report. However, it is anticipated these will be added to the NC NBS panel in the near future:

- Glycogen Storage Disease Type II (Pompe)
- Mucopolysaccharidosis Type 1
- X-linked Adrenoleukodystrophy

- Spinal Muscular Atrophy due to homozygous deletion of exon 7 in SMN1
- *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 screens are added, follow-up protocols are developed from 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 of 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 month 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 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 prior to 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 \$7.5 million to operate in SFY 2017-18, but it collected approximately \$5.67 million in receipts.

Newborn Screening SFY 2017-18								
Expenditure Description	State Lab		Follow up Program Children's Health		Indirect Cost		Total Newborn Screening Program	
	Budget	Expended	Budget	Expended	Budget	Expended	Budget	Expended
Personal Services*	\$1,885,448.00	\$1,576,739.77	\$155,703.00	\$97,654.75	\$1,306,691.29	\$1,306,691.29	\$3,347,842.29	\$2,981,085.81
Purchased Services**	\$1,272,630.00	\$725,026.32	\$18,371.00	\$372.40			\$1,291,001.00	\$725,398.72
Supplies	\$1,920,306.00	\$1,730,157.17	\$2,000.00	\$887.11	\$1,857,672.07	\$1,857,672.07	\$3,779,978.07	\$3,588,716.35
Property, Plant & Equipment	\$319,713.00	\$209,625.75	\$25.00	\$-	\$-	\$-	\$319,738.00	\$209,625.75
Other Expenses & Adjustments	\$-	\$(1.40)	\$-	\$-	\$-	\$-	\$-	\$(1.40)
Total Expenditures	\$5,398,097.00	\$4,241,547.61	\$176,099.00	\$98,914.26	\$2,945,254.61	\$3,164,363.36	\$8,738,559.36	\$7,504,825.23
Revenue Description	Budget	Collected	Budget	Collected	Budget	Collected	Budget	Collected
Other Licenses, Fees and Permits***	\$24,424.00	\$-	\$-	\$-	\$-	\$-	\$24,424.00	\$-
Newborn Screening Fees****	\$4,560,974.00	\$5,279,625.55	\$110,413.00	\$98,914.26	\$-	\$-	\$4,671,387.00	\$5,378,539.81
Accounts Received Interest*****	\$-	\$7,649.09	\$-	\$-	\$-	\$-	\$-	\$7,649.09
Accounts Received Penalty*****	\$-	\$77,717.47	\$-	\$-	\$-	\$-	\$-	\$77,717.47
DMA Medicaid Title X*****	\$812,699.00	\$204,804.51	\$-	\$-	\$-	\$-	\$812,699.00	\$204,804.51
Total Revenues	\$5,398,097.00	\$5,569,796.62	\$110,413.00	\$98,914.26	\$-	\$-	\$5,508,510.00	\$5,668,710.88
* Expenditures incurred for services rendered by permanent and temporary employees and the related fringe benefits								
** Expenditures incurred for services required to ensure the ongoing operation of State government facilities and government services; for example, operational services, maintenance agreements, service contracts, travel, postage, telecom, etc.								
*** Fees from workshops								
**** Fees from hospitals and birthing centers								
***** Collected per NC General Statute 147-86.23								
***** Medicaid receipts from non-hospital repeat NBS samples								

Positions Funded SFY 2017-18	
State Lab	Budgeted Salary
Med Lab Technologist I	\$40,178
Chemistry Technician I	\$30,308
Chemistry Supervisor III	\$71,835
Med Lab Technologist II	\$45,557
Med Lab Technologist I	\$40,288
Processing Unit Superviso V	\$35,326
Chemist II	\$55,000
Med Lab Technologist I	\$40,000
Med Lab Technologist I	\$40,178
Chemistry Technician I	\$33,960
Med Lab Technologist I	\$41,000
Laboratory Improvement Consultant	\$51,972
Med Lab Technologist II	\$39,962
Medical Lab Supervisor II	\$64,702
Med Laboratory Supervisor IV	\$93,797
Med Lab Technologist II	\$44,917
Med Lab Technologist I	\$40,333
Chemist II	\$55,000
Med Lab Technologist I	\$40,580
Chemist II	\$55,678
Med Lab Technologist II	\$46,000
Medical Laboratory Technologist II	\$44,095
Laboratory Improvement Consultant	\$56,767
Laboratory Medical Specialist	\$50,513
Laboratory Medical Specialist	\$51,654
Laboratory Medical Specialist	\$50,326
Medical Laboratory Supervisor II	\$64,640
Childrens Health	Budgeted Salary
Public Health Genetic Counselor	\$60,955
Public Health Educator III	\$45,576
New Positions SFY 2018-19	
Information Technology	Budgeted Salary
Application Systems Specialist I	\$98,302
State Lab	Budgeted Salary
Public Health Scientist	\$70,780
Chemist II	\$56,941
Chemistry Tech II	\$41,218
Childrens Health	Budgeted Salary
Social Worker	\$52,477
Social Worker	\$52,477
Social Worker	\$52,477

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 2018-19 Fees & Other Receipts Received Year-to-Date (as of 1/9/2019)	Estimated SFY 2018-19 Fees & Other Receipts	Projected SFY 2019-20 Fees & Other Receipts
NBS	\$4,504,061	\$9,008,122	\$11,640,000
NBS Equipment Replacement and Acquisition Fund	\$1,083,822	\$2,167,644	\$3,700,000
Total	\$5,587,883	\$11,175,766	\$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 increase will now fully cover routine equipment replacement as well as associated costs to implement, onboard, and purchase equipment for new tests.

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

Spinal Muscular Atrophy was added to the RUSP this fiscal year. Resources anticipated to bring this test online in NC are:

- Equipment: Real-time PCR (RT-PCR) detection; 5 instruments at a cost of approximately \$40,000 each (total of \$200,000)
- Positions: 1 Full Time Equivalent Medical Laboratory Specialist; approximately \$75,000 annual salary and fringes

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

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

The Fund was established in SFY 2018-19, so there was no balance in the Fund as of June 30, 2018.

Amounts credited to the Fund

As of January 9, 2019, for SFY 2018-2019, \$1,083,822 has been credited to the Fund.

Amounts expended from the Fund and the purposes of the expenditures

No funds have been expended from the Equipment Replacement and Acquisition Fund as of the writing of this report.

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

The State Laboratory of Public Health anticipates the following expenditures and approximate costs from the NBS Equipment Replacement and Acquisition Fund:

- **SFY 2018-2019**
Scientific and Information Technology equipment - \$1,881,700
- **SFY 2019-2020**
Scientific and Information Technology equipment - \$3,700,000

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 today are well documented and may be summarized as follows:

- The expansion of NBS screening in NC as new RUSP conditions are added will encounter the following challenges:
 - Updating NBS administrative rules to include new conditions added to the RUSP and make appropriate, corresponding increases to the NBS fee is a lengthy process.
 - Some hospitals and birthing centers could struggle with funding regularly increasing NBS fees.
 - Once instrumentation, equipment, supplies, and personnel are available and onsite, there must be adequate time to perform validations to ensure the State Lab is meeting all federal, state, and local requirements to screen for the new condition(s).
 - It will take time to build the new Newborn Screening Equipment Replacement and Acquisition Fund to ensure there are adequate funds to acquire instrumentation and equipment. It will also take time after a fee increase is implemented to build funds to acquire program supplies and hire program personnel to add a new screening test.
- Advances in technology, biology, and medicine mean that the pipeline of conditions that meet the criteria for newborn screening could grow more quickly over time. This could raise challenging questions at some point in the future about how to support growth (both financially and functionally) in newborn screening while maintaining the highest standards of quality in both screening and follow-up. This is an issue that state

public health laboratories throughout the country are wrestling with, many of which already feel stretched adding one new screening per year.