State of New Hampshire
Newborn Screening Program

Annual Report
Calendar Year
2022







NEW HAMPSHIRE DHHS
DEPARTMENT OF HEALTH & HUMAN SERVICES

NH DIVISION OF Public Health Services
Improving health, preventing disease, reducing costs for all
Department of Health and Human Services

INTRODUCTION

The Department of Health and Human Services (DHHS), Division of Public Health Services, Bureau of Family Health and Nutrition, Maternal and Child Health Section has responsibility for oversight of the New Hampshire Newborn Screening Program (NBSP). The NBSP is responsible for ensuring all infants born in New Hampshire are screened for 36 heritable conditions; managing daily screening results; ensuring that screening is timely; and ensuring that any infants identified with a presumptive positive result receive timely follow up and referral to specialty care for confirmation of diagnosis and initiation of treatment. Many disorders identified through newborn screening require care and treatment throughout the lifespan.

RSA 132:10-a requires that a physician, hospital, nurse midwife, midwife, or other health care provider attending the birth of a newborn shall screen the infant for a panel of heritable disorders. This statute also includes a clause, 132:10-c, which allows parents or guardians to decline the screening if they so desire.

RSA 132:10-a also requires that DHHS make an annual report to the New Hampshire Health and Human Services Oversight Committee on the previous year's NBSP activities including but not limited to the number and type of tests performed and their fiscal impact. Previous years' reports are available on the NBSP website at https://www.dhhs.nh.gov/programs-services/population-health/maternal-child-health/newborn-screening-program .

CALENDAR YEAR 2022

DATA

From January 1, 2022, through December 31, 2022, the NBSP screened 12,030 infants out of 12,135 births in New Hampshire, which represented 99% of all occurring births. Tables 1, 2, and 3 contain the NBSP statistics for 2022. The NBSP reports 2022 data due to the length of time it takes for obtaining confirmation of disorders. Table 1 includes the number of infants who were screened; had screening declined by a parent or guardian; died; were transferred out of state prior to having an initial screen; missed screening; or other (screen not obtained and no midwife or provider listed for follow up; duplicate birth certificates; infant screened but midwife used another state's filter paper; or infant screened but family chose private lab). Table 2 includes the number of infants with abnormal specimens or samples, the potential disorders identified, and the number of unsatisfactory specimens for 2022. Table 3 includes the number of confirmed diagnoses for 2022.

Table 1: NBSP data for 2022

NH Births	
Screened	12,030
Screening declined	33
*Other	17
Died	12
Transferred out of State (Initial screening completed outside NH)	42
Missed	1
Total NH Births	**12,135**

***Other:** screen not obtained, no midwife or provider listed for follow up; duplicate birth certificates; infant screened but midwife used another state's filter paper; or infant screened but family chose private lab.

Table 2: NBSP Abnormal Screens for 2022

Abnormal Screen Result	**# of Infants with Abnormal Screens**
Amino Acid or Acylcarnitines	198
Biotinidase (BIO)	4
Congenital Adrenal Hyperplasia (CAH)	62
Congenital Hypothyroidism (T4/TSH)	177
Cystic Fibrosis (CF)	45
Galactosemia (Total GAL/ GATL)	2
Maple Syrup Urine Disease (MSUD)	5
Lysosomal Storage Disorders (GAA)	5
Lysosomal Storage Disorders (IDUA)	3
Severe Combined Immunodeficiency (TREC)	51
Hemoglobinopathies (trait or disease)	175
*Unsatisfactory Specimens	5
Total # Infants with Abnormal Screens	**732**

*Abnormal screening result on an unsatisfactory specimen (drawn prior to 24hrs of life, clotted, poor soak, etc.).

Table 3: NBSP Confirmed Cases for 2022

Disorder	# Cases Confirmed
Pompe Disease	1
Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)	2
Phenylketonuria (PKU)	2
Congenital hypothyroidism	11
Sickle cell disease	2
Duarte galactosemia	2
Biotinidase deficiency	2
Cystic Fibrosis	1
CFTR-related metabolic syndrome (CRMS)	3
*Other Disorder	4
Total Confirmed Diagnoses	**30**

*Other disorder not on the newborn screening panel but detected from screening

STAFFING

The care of children diagnosed with the conditions on New Hampshire's Newborn Screening Panel is complex, involving primary care physicians, metabolic specialists, genetic counselors, immunology specialists, endocrinology specialists, and hematology specialists. It is important to have consultation available from specialists to help guide providers caring for an infant with an abnormal screen that requires a diagnostic work up. To this end, the NBSP renewed the contract with a metabolic specialist from Boston Children's Hospital at the beginning of 2022. The contract is effective June 30, 2022, through June 30, 2025.

In 2022, there were a series of staff transitions. The 0.5 Full Time Equivalent (FTE) Nurse Program Specialist position was vacant from October 2021 to March 2022. In March 2022, the NBSP filled this position with a registered nurse who brings experience working in public health and epidemiology at the state level. The 0.5 FTE Program Planner position was vacant from the end of January 2021 to May 2022. In May 2022, the NBSP filled this position with a medical assistant who brings experience working in many healthcare settings, including primary care and urgent care. At the end of 2022, the NBSP was at full staffing capacity.

NEWBORN SCREENING ADVISORY COMMITTEE

The Newborn Screening Advisory Committee (NBSAC) makes recommendations to the NBSP on clinical, educational, and operational aspects of the program. The NBSAC held bi-annual meetings in May 2022 and October 2022. in accordance with RSA 132:10-a and Administrative Rule He-P 3008 Section 132:10-a Newborn Screening Tests Required; Newborn Screening Advisory Committee. (state.nh.us). The NBSAC meetings are open to the public and are posted to the DHHS Newborn Screening website. Often a parent of a child with disorder will come and present. This past year we had a parent of a child with Angelman Syndrome present to the members of the NBSAC.

The New England Newborn Screening Program director also updated the Newborn Screening Advisory Committee on their ongoing efforts to assess readiness to screen for new conditions added to the national Recommended Universal Screening Panel in 2022, Guanidinoacetate Methyltransferase (GAMT) Deficiency and Mucopolysaccharidosis Type II (MPS II). It has yet to be determined if and when the contracted lab will be able to provide screening for either of these disorders. No disorders were added to the NH screening panel in 2022.

The Newborn Screening Advisory Committee votes on approving or denying additions to the New Hampshire screening panel based on the following criteria for each disorder:

- The disorder is well-defined with a known incidence.
- The disorder is associated with significant morbidity and/or mortality.
- The disorder can be detected with a screening test that is ethical, safe, accurate, and cost-effective.
- Effective treatment exists for the disorder, and that early treatment, meaning before the onset of symptoms, is more effective in improving health outcomes than later treatment.

QUALITY IMPROVEMENT

The NBSP relies on timeliness and specimen quality for optimal results. The age of the infant at specimen draw, time between specimen collection and receipt at the laboratory, and the interval between the blood draw and screening result are all important factors that can impact the quality of screening results.

In addition, the process of screening a newborn involves several steps and often multiple individuals within a facility. They include staff who fill out the filter paper, staff who obtain the specimen from the infant, staff who are responsible for sending the dried specimens to the laboratory, and the laboratory staff who conduct the analysis. Delays or errors in any one of these steps can impact the entire process and result in delayed identification of infants who need follow-up.

The NBSP provides a quarterly timeliness progress report to all birthing facilities focused on measures primarily within the control of hospital staff, including whether specimens were collected between 24 and 48 hours of birth. Minimizing this time can lead to quicker reporting of screening results to healthcare providers and save infant lives through early detection and intervention.

Historical data demonstrates significant progress in meeting the target of ≥95% of specimens collected within the 24 to 48-hour guideline. The target was set at ≥95% because there are rare instances where a specimen may appropriately be taken outside of the 24 to 48-hour window (prior to treatments, early discharge, or transfer to higher level of care). Among the 16 current birthing hospitals, the percentage of newborn screening specimens taken between 24 to 48 hours improved from 78.1% in Quarter 1 of 2015 to 98.0% in Quarter 4 of 2022. A majority of hospitals, as seen in Appendix B, met the ≥95% target throughout 2022. Those that didn't were only slightly below the target threshold. This successful screen rate can be attributed to the collaborative, data-driven quality improvement efforts undertaken between the NBSP and the state's birthing hospitals.

Additional opportunities for quality improvement include decreasing the number of unsatisfactory filter paper specimens, which require repeat specimens. To obtain a repeat specimen, an infant must return to a facility where staff have the expertise to draw a filter paper specimen. This can lead to a delay in newborn screening results and a potential delay in diagnostic workup, follow up, and treatment if there is an abnormal result. A repeat screen also incurs additional costs to the facility. From January 1, 2022, through December 31, 2022, spending on filter papers due to initial unsatisfactory specimens totaled $30,368. This represents a $2,628 decrease in spending on filter papers for repeat screens as compared to 2021, which totaled $32,996.

In October 2022, the NBSP began sending a quarterly data report to birthing hospitals informing them of unsatisfactory specimens. The report includes a breakdown of how many unsatisfactory specimens were received at the lab per quarter, along with the reason the specimen was unsatisfactory. A specimen may be deemed unsatisfactory if it is layered or clotted; improperly dried; of an insufficient quantity; poorly soaked; contaminated; transfused within 48 hours of specimen collection; or drawn on expired filter paper. From January 1, 2022, through December 31, 2022, the NBSP had 208 unsatisfactory specimens submitted to the NENSP. Table 4 contains the NBSP statistics for 2022 for unsatisfactory specimens.

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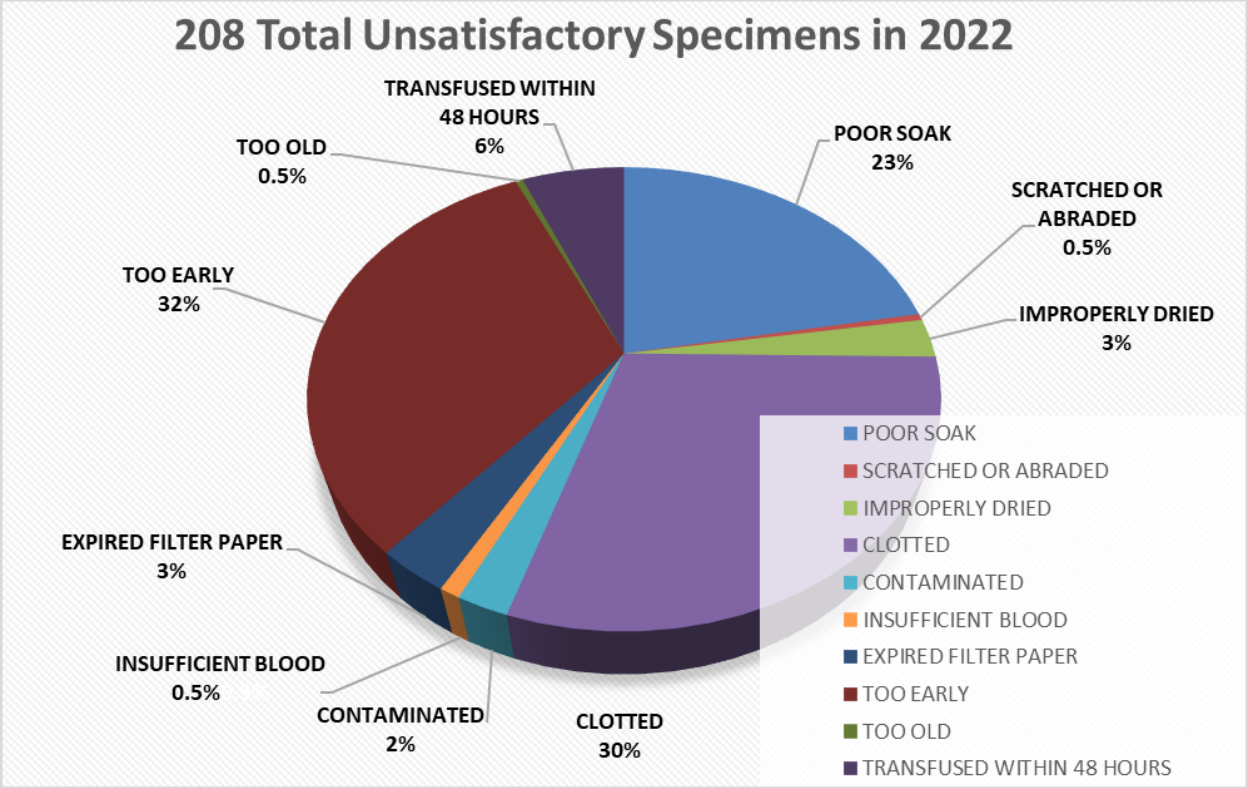
Table 4: Unsatisfactory Specimens in 2022



DATA SYSTEM

Throughout 2022, the NBSP has worked with a contracted vendor, OZ Systems, to report newborn screening results. Over the past year, OZ Systems and DHHS have been working with birth facilities across the state to establish a secure interface for linking hospitals' electronic medical records to the OZ System using established health data exchange standards for newborn screening results.

UPCOMING PLANS

Newborn screening remains a dynamic field. Advances in science and technology are ongoing and continue to impact the state's newborn screening program. The NBSP will continue to monitor data quality and provide unsatisfactory specimen and timeliness reports to all birth facilities in the state. In the coming year, the program will maximize the use of data and quality improvement recommendations to ensure no infant is missed for newborn screening (excluding those that refuse the screening) and that all infant specimens are tested and followed up on in a timely manner.

The NBSP also plans to decrease the time between courier pick up and drop off at the UMass Laboratory, which contributes to overall timeliness of results and referral to specialty care for confirmation of diagnosis and initiation of treatment. Plans include working with birth hospitals to confirm specimen pick up and drop off locations. The UMass Laboratory is responsible for coordinating the courier services, for which it utilizes United Parcel Service (UPS) and City

Express. The New England Newborn Screening Program will continue to monitor the data and identify any challenges associated with pick up and drop off.

The NH Newborn Screening Program applied for the Health Resources and Services Administration's State Newborn Screening System Priorities Program (funding opportunity number HRSA-23-065) and was successfully awarded a grant. The purpose of this funding is to support newborn screening programs to address state-specific challenges and implement quality improvement activities such as improving and expanding the newborn screening management information system, implementing screening for new conditions added to the RUSP, and increasing access to treatment and follow-up activities for individuals and families identified with a heritable condition through newborn screening.

In 2022, the NBSP amended the Administrative Rules, He-P 3008, to allow for information disclosure to the NBSP to allow for follow up of abnormal screening results, updated specimen collection guidance to align with national recommendations, and updated term limits for the Newborn Screening Advisory Committee Co-Chairperson. Rule amendments were passed by the Joint Legislative Committee on Administrative Rules (JLCAR) in early 2023.

APPENDIX A

Table 5: New Hampshire Newborn Screening Panel as of January 1, 2022

Disorder Name	Acronym
Argininosuccinic Aciduria	ASA
Argininemia	ARG
Biotinidase	BIOT
Carnitine Uptake Defect	CUD
Carnitine Palmitoyltransferase II Deficiency	CPT II
Citrullinemia I (ASA Synthetase Def)	CIT
Cobalamin A, B	Cbl A, B
Congenital Adrenal Hyperplasia	CAH
Congenital Hypothyroidism	CH
Congenital Toxoplasmosis	TOXO
Critical Congenital Heart Disorder	CCHD
Cystic Fibrosis	CF
Galactosemia	GALT
Glutaric Aciduria Type I	GA I
Hemoglobinopathies (3 types)	Hb SS +, Hb S/BTh, +Hb S/C
3-Hydroxy-3-Methylglutaryl-CoA Lysase Deficiency	HMG
Hyperornithinemia Hyperammoninemia, Homocitrullinemia Syndrome	HHH
Homocystinuria	HCY
Isovaleric Acidemia	IVA
Long Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD
Maple Syrup Urine Disease	MSUD
Medium Chain Acyl-CoA Dehydrogenase Deficiency	MCAD
3-Methylcrotonyl-CoA Carboxylase Deficiency	3MCC
Methylmalonic Acidemia	MUT
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency	BKT
Multiple Acyl-CoA Dehydrogenase Deficiency	GA2
Multiple Carboxylase Deficiency	MCD
Phenylketonuria	PKU
Propionic Acidemia	PROP
Severe Combined Immunodeficiency Disorder	SCID
Spinal Muscular Dystrophy	SMA
Trifunctional Protein Deficiency	TFP
Tyrosinemia type I	TYR I
Very Long Chain Acyl-CoA Dehydrogenase Deficiency	VLCAD
Mucopolysaccharidosis	MPS1
X-Linked Adrenoleukodystrophy	X-ALD
Pompe	POMPE

APPENDIX B

Table 6: Newborn Screening Specimen Timeliness 2019-2022

*** There were no births occurring at Frisbie Hospital in Quarter 4 (Q4) of 2022.**

Percent of Specimens Collected 24 to 48 Hours After Birth
Among 16 currently active birth hospitals in New Hampshire
Target >= 95%

	2019				2020				2021				2022			
	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4
Androscoggin Valley Hospital	100.0%	100.0%	100.0%	100.0%	96.3%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%
Catholic Medical Center	95.9%	97.1%	96.6%	97.1%	96.7%	95.3%	97.3%	97.0%	97.6%	96.6%	96.8%	96.1%	97.7%	96.5%	97.8%	96.7%
Cheshire Medical Center	100.0%	99.0%	99.0%	100.0%	97.0%	97.2%	99.0%	97.0%	96.1%	97.3%	96.8%	99.2%	100.0%	100.0%	100.0%	98.1%
Concord Hospital	97.5%	98.8%	98.3%	98.1%	99.4%	98.3%	97.5%	98.6%	98.0%	98.1%	99.0%	96.6%	99.0%	97.4%	99.0%	98.2%
Dartmouth-Hitchcock Medic..	98.7%	98.6%	97.5%	99.0%	99.1%	98.2%	98.8%	99.7%	97.9%	98.7%	99.0%	98.0%	97.0%	98.6%	99.2%	98.2%
Elliot Hospital	99.0%	98.2%	96.8%	95.9%	98.7%	98.1%	98.5%	99.1%	99.8%	97.3%	99.0%	99.6%	99.2%	98.9%	99.0%	97.0%
Exeter Hospital	95.9%	96.9%	95.9%	96.1%	94.9%	97.5%	95.7%	97.6%	98.4%	97.3%	98.8%	96.4%	97.3%	98.7%	99.3%	97.9%
Frisbie Memorial Hospital	96.7%	93.8%	88.2%	96.2%	90.0%	94.2%	100.0%	97.2%	93.1%	97.6%	97.4%	88.7%	100.0%	96.2%	100.0%	
Littleton Regional Hospital	96.9%	100.0%	100.0%	97.6%	96.2%	100.0%	98.8%	96.2%	98.7%	99.0%	98.8%	98.6%	100.0%	100.0%	100.0%	97.5%
Memorial Hospital	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	93.6%	94.7%	100.0%	98.0%	96.1%	94.4%	93.1%	100.0%	97.6%
Monadnock Community Hosp..	95.7%	100.0%	98.6%	97.4%	97.9%	98.4%	98.7%	100.0%	98.7%	98.9%	98.8%	97.6%	100.0%	100.0%	98.2%	100.0%
Portsmouth Regional Hospital	97.7%	98.1%	94.2%	96.4%	94.8%	97.8%	97.3%	99.2%	98.1%	97.7%	97.0%	99.1%	97.9%	95.5%	95.5%	98.3%
Southern NH Medical Center	98.6%	99.0%	99.4%	99.1%	99.3%	96.2%	98.5%	99.0%	99.7%	98.7%	99.7%	99.0%	100.0%	100.0%	98.4%	98.2%
Speare Memorial Hospital	100.0%	100.0%	93.9%	100.0%	100.0%	100.0%	98.0%	100.0%	100.0%	98.0%	100.0%	100.0%	97.9%	96.4%	100.0%	100.0%
St. Joseph Hospital	100.0%	98.9%	97.4%	100.0%	100.0%	97.7%	99.0%	99.0%	97.0%	98.6%	99.3%	97.6%	97.2%	97.4%	99.3%	98.4%
Wentworth-Douglass Hospital	96.4%	99.1%	97.2%	97.6%	98.0%	98.8%	98.6%	97.5%	98.4%	97.7%	97.8%	97.8%	96.5%	99.2%	99.1%	99.1%

Statewide Percent In Guideline
Collected 24 to 48 hours
Among 16 birth hospitals
Q1 2015 (not shown)

78.1%

Statewide Percent In Guideline
Collected 24 to 48 hours
Among 16 birth hospitals
Q4 2022

98.0%

Target >=95%