Annual Report on Newborn Screening to
New Hampshire Health and Human Services Oversight Committee

Calendar Year 2021

New Hampshire Newborn Screening Program
Maternal and Child Health Section
Bureau of Population Health and Community Services
Division of Public Health Services
Department of Health and Human Services
January 2023

INTRODUCTION

The Department of Health and Human Services (DHHS), Division of Public Health Services, Bureau of Population Health and Community Services, Maternal and Child Health Section is responsible for the New Hampshire Newborn Screening Program (NBSP). The NBSP assures that all infants born in New Hampshire are screened for heritable conditions. The program is responsible for daily management of screening results (normal and out-of-range); that the screening is timely and complete; and any infants with an out-of-range result receive a timely referral to a specialist. Many disorders identified through newborn screening require care and treatment throughout the lifespan.

RSA 132:10a and He-P 3008 legislates that all infants born in New Hampshire are eligible to be screened at birth for a panel of disorders as determined by the State through a complex decision making process also defined in statute. This statute includes a clause, 132:10-c, which allows parents or guardians to decline this screening if they so desire.

RSA 132:10a also includes a requirement that DHHS make an annual report to the New Hampshire Health and Human Services Oversight Committee on the previous year's NBSP activities. 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.

In 2021, RSA 132:10a was amended and on November 20, 2021, it became effective. The legislation requires that the fees for the filter papers, which are directly paid by the hospitals, are to be offset by commercial insurance or Medicaid for all of the tests required under the NBSP Panel (Appendix A).

CALENDAR YEAR 2021

DATA

From January 1, 2021 through December 31, 2021, the NBSP screened 12,547 infants born in New Hampshire, which represented 99% of all occurring births. Tables 1, 2 and 3 contain the NBSP statistics for 2021. This includes the number of infants who were screened; declined; died; transferred out of state prior to having an initial screen; other (including lost-to-follow-up, duplicate birth certificates, or screens obtained on out-of-state filter paper); number of abnormal specimens or samples, disorders identified; and pending confirmation of diagnoses in that year.

Table 1: 2021 NBSP Data

Total NH births	**12,662**
Number of NH births screened	12,547
Screening declined	34
Other	17
Died	30
Transferred out of State (Initial screening completed outside NH)	34
Missed	0

Table 2: NBSP Abnormal Screens 2021

Abnormal Areas	**# of Abnormal Screens**
Amino Acid or Acylcarnitines	167
Biotinidase (BIO)	1
Congenital Adrenal Hyperplasia (CAH)	74
Congenital Hypothyroidism (T4/TSH)	173
Cystic Fibrosis (CF)	44
Galactosemia (Total GAL/ GATL)	6
Maple Syrup Urine Disease (MSUD)	3
Lysosomal Storage Disorders (GAA)	4
Lysosomal Storage Disorders (IDUA)	3
Severe Combined Immunodeficiency (TREC)	45
Hemoglobinopathies (trait or disease)	202
Spinal Muscular Atrophy (SMA)	3
*Unsatisfactory Specimens	9
X-ALD (C26 HPLC)	1
Total Abnormal Samples	**662**

*Abnormal screening result on an unsatisfactory specimen which was collected too early (collected <24 hours of age)

Table 3: NBSP Confirmed Cases For 2021

Disorder	# Confirmed
Citrullinemia	1
Congenital Hypothyroidism (CH)	14
Cystic Fibrosis (CF)	5
Galactosemia	2
Maple Syrup Urine Disease (MSUD)	1
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)	1
Methyl Malonic Acidemia (MMA)	2
Mucopolysaccharidosis type I (MPS-1)	1
Classic Phenylketonuria (PKU)	1
Pompe Disease	1
Sickle Cell Disease	3
Spinal Muscular Atrophy (SMA)	2
X-linked Adrenoleukodystrophy (X-ALD)	1
Total Confirmed Diagnoses	**35**

STAFFING

The care of children who screen positive for the conditions on New Hampshire's Newborn Screening Panel is complex, involving primary care physicians, metabolic specialists, genetic counselors, and metabolic nutritionists. It is important to have consultation available from specialists to help guide pediatricians and others through the confirmation of diagnosis and long-term clinical management.

The NBSP experienced several staff transitions in 2021. The 0.5 Full Time Equivalent (FTE) Nurse Program Specialist position was vacant from October 2021 until March 2022. The 0.5 FTE Program Planner position was vacant from mid January 2021 until May 2022. During these vacancies, the program was run by one full-time Public Health Nurse Coordinator.

The State contracts with the New England Newborn Screening Program (NENSP) at the University Of Massachusetts Chan Medical School, which is currently in the first year of a three year contract with an option to renew for an additional three years. There was a Request For Proposals released during the summer of 2021, from which NENSP was selected. The contract was approved by the Governor and his Executive Council on June 29, 2022. The NENSP provides filter paper collection kits to birth facilities and midwives, provides newborn screening laboratory services, and provides results and recommendations for out-of-range results. They also contract with a courier service to

provide timely pickup and delivery of specimens from birth hospitals to the NENSP laboratory.

The Department contracts with a metabolic specialist, Dr. Amy Kritzer, who provides medical consultation and recommendations related to clinically significant screening results to the NBSP and to the NH medical community, as needed. The NBSP is in its second year of a three year contract with the metabolic specialist, with the ability to renew for an additional three years.

NEWBORN SCREENING ADVISORY COMMITTEE

During 2021, The Newborn Screening Advisory Committee (NBSAC) resumed its bi-annual meeting schedule in accordance with RSA 132:10-a and Administrative Rule He-P 3008. The first meeting was held virtually and the second convened in-person once the Governor terminated the State of Emergency for the COVID 19 pandemic. NBSAC meetings are open to the public and are posted to the DHHS Newborn Screening website: https://www.dhhs.nh.gov/programs-services/population-health/maternal-child-health/newborn-screening-program.

The NBSAC determines recommended additions to the screening panel based on lengthy discussions focused on the following 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.

During 2021, no disorders were added to the national Recommended Universal Screening Panel (RUSP) or to the NH screening panel. The NBSAC reviewed Cystic Fibrosis Newborn Screening Guidelines, NBSP current protocols and practices, and NH data on timeliness and unsatisfactory specimens. The NBSAC also reviewed NENSP testing updates on newly added disorders and reviewed challenges encountered with supplies, equipment and staffing related to the COVID-19 Pandemic.

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 very important. The process of screening a newborn involves a number of critical steps and often involves multiple individuals within a facility. These include staff who complete the demographic information on the filter paper, staff who obtain the specimen from the infant, staff who are responsible for sending the dried specimens to the laboratory, and finally the laboratory staff themselves. 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

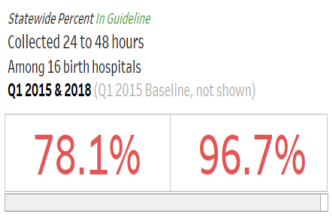
quarterly timeliness progress report to all birthing hospitals focused on measures primarily within the control of hospital staff.

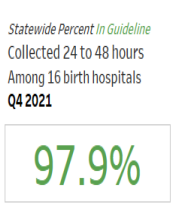
Specimens should be collected between 24 and 48 hours of life. This is one of the factors that impacts the overall timeline between birth and reporting of screening results to the healthcare provider. Minimizing this time can save infant lives through early detection and intervention. Historical data clearly demonstrates significant progress in meeting the target of ≥95% of specimens collected within the 24-48 hour guideline. The target was set at 95% because there are rare instances where a specimen may appropriately be taken outside of the 24-48 hour window. As seen in Table 4 below, among the 16 current birth hospitals, the percentage of newborn screening specimens taken between 24-48 hours improved from 96.7% to 97.9% from Quarter 1 2018 to Quarter 4 2021. This is a vast improvement from Q 1 2015 where only 78.1% of specimens were collected between 24-48 hours after birth. In 2021, the majority of hospitals, as seen in Table 4, met the 95% target throughout the year. Those that didn't were only slightly below. This can be attributed to the collaborative data driven quality improvement efforts between the NBSP and the State's birthing hospitals.

Newborn Screening Specimen Timeliness
Table 4

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

	2018				2019				2020				2021			
	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4
Androscoggin Valley Hospital	100.0%	100.0%	92.0%	100.0%	100.0%	100.0%	100.0%	100.0%	96.3%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%
Catholic Medical Center	98.0%	96.7%	97.1%	94.7%	95.9%	97.1%	96.6%	97.1%	96.7%	95.3%	97.3%	97.0%	97.6%	96.6%	96.8%	96.1%
Cheshire Medical Center	98.9%	100.0%	99.0%	97.9%	100.0%	99.0%	99.0%	100.0%	97.0%	97.2%	99.0%	97.0%	96.1%	97.3%	96.8%	99.2%
Concord Hospital	96.9%	98.1%	97.3%	97.8%	97.5%	98.8%	98.3%	98.1%	99.4%	98.3%	97.5%	98.6%	98.0%	98.1%	99.0%	96.6%
Dartmouth-Hitchcock Medical Center	98.1%	99.0%	98.9%	99.3%	98.7%	98.6%	97.5%	99.0%	99.1%	98.2%	98.8%	99.7%	97.9%	98.7%	99.0%	98.0%
Elliot Hospital	94.9%	93.1%	94.6%	94.8%	99.0%	98.2%	96.8%	95.9%	98.7%	98.1%	98.5%	99.1%	99.8%	97.3%	99.0%	99.6%
Exeter Hospital	93.4%	97.2%	95.8%	91.3%	95.9%	96.9%	95.9%	96.1%	94.9%	97.5%	95.7%	97.6%	98.4%	97.3%	98.8%	96.4%
Frisbie Memorial Hospital	97.1%	98.6%	97.9%	93.5%	96.7%	93.8%	88.2%	96.2%	90.0%	94.2%	100.0%	97.2%	93.1%	97.6%	97.4%	88.7%
Littleton Regional Hospital	96.1%	97.5%	100.0%	95.7%	96.9%	100.0%	100.0%	97.6%	96.2%	100.0%	98.8%	96.2%	98.7%	99.0%	98.8%	98.6%
Memorial Hospital	98.2%	97.9%	98.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	93.6%	94.7%	100.0%	98.0%	96.1%
Monadnock Community Hospital	97.6%	96.1%	98.5%	97.5%	95.7%	100.0%	98.6%	97.4%	97.9%	98.4%	98.7%	100.0%	98.7%	98.9%	98.8%	97.6%
Portsmouth Regional Hospital	94.6%	97.9%	96.3%	97.1%	97.7%	98.1%	94.2%	96.4%	94.8%	97.8%	97.3%	99.2%	98.1%	97.7%	97.0%	99.1%
Southern NH Medical Center	97.1%	99.3%	99.3%	98.7%	98.6%	99.0%	99.4%	99.1%	99.3%	96.2%	98.5%	99.0%	99.7%	98.7%	99.7%	99.0%
Speare Memorial Hospital	90.9%	100.0%	95.7%	100.0%	100.0%	100.0%	93.9%	100.0%	100.0%	100.0%	98.0%	100.0%	100.0%	98.0%	100.0%	100.0%
St. Joseph Hospital	96.7%	100.0%	95.3%	94.1%	100.0%	98.9%	97.4%	100.0%	100.0%	97.7%	99.0%	99.0%	97.0%	98.6%	99.3%	97.6%
Wentworth-Douglass Hospital	98.7%	97.5%	98.5%	96.4%	96.4%	99.1%	97.2%	97.6%	98.0%	98.8%	98.6%	97.5%	98.4%	97.7%	97.8%	97.8%





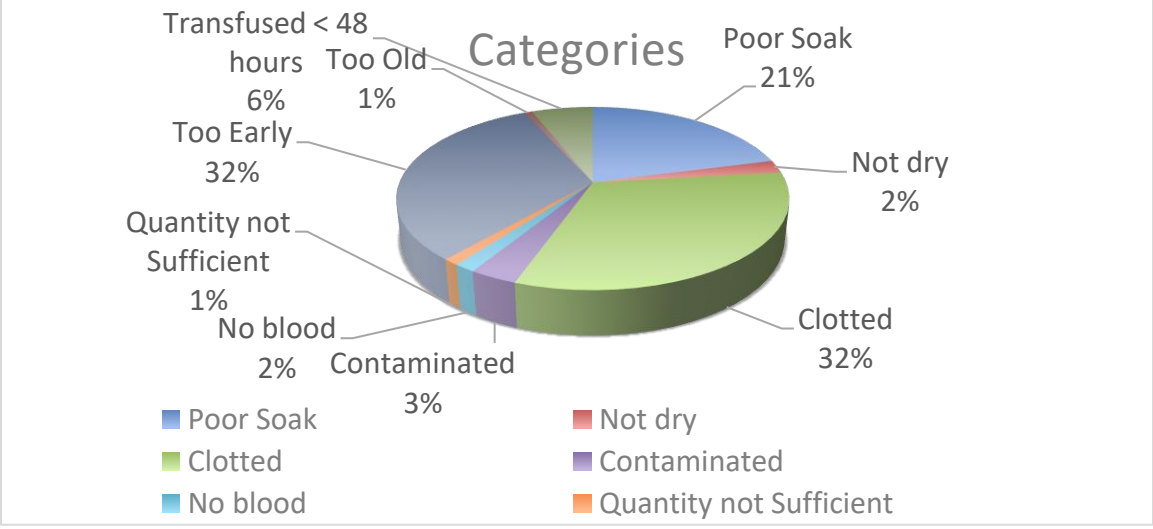
Target >=95%

The timely handoff to the courier is another factor impacting the overall timeline between birth and reporting of screening results to the healthcare provider. Minimizing this time can save infant lives through early detection and intervention. The New England Newborn Screening Program (NENSP) utilizes the United Parcel Service (UPS) as its courier for all specimens. During 2020, a quality improvement process focused on this metric in conjunction with the laboratory and UPS. During 2021, the NENSP did not have the capability to fully collect this data. The NENSP continues to monitor this data and will soon incorporate this metric into its data sets which will be able to be generated on demand by the NBSP.

Additional measures for quality improvement include decreasing the number of unsatisfactory filter paper specimens which require a repeat specimen. A repeat specimen involves the infant coming back to a facility in which appropriate staff have the expertise to draw a repeat filter paper specimen. This leads to a delay in newborn screening results and a potential delay in diagnostic workup/treatment for a condition. Not only is the NBSP looking at unsatisfactory specimens for timeliness, the NBSP aims to be cost effective. Each additional repeat screen increases the cost to the facility, with each filter paper costing $146. From January 1, 2021 through December 31, 2021 the total expenditure of filter papers due to initial unsatisfactory specimens was $32,996.

From January 1, 2021 through December 31, 2021, the NBSP had 226 initial unsatisfactory specimens. This is a decrease from the previous year which had 252 initial unsatisfactory specimen. Table 5 contains the NBSP statistics for 2021 for unsatisfactory specimens. The reasons for unsatisfactory specimens include; layer or clotted; improperly dried; quantity not sufficient; poor soak; contaminated; transfused within 48 hours of specimen collection; and expired filter papers.

Cause of Unsatisfactory Specimens 2021
Table 5



DATA SYSTEM

With input from stakeholders, the Maternal and Child Health Section led the development of a Request for Proposals (RFP) for a scalable, secure web-based integrated data management system for the NBSP, the Birth Conditions Program and the Newborn Hearing Screening Program, all of which previously had independent data collection systems. The OZ System was selected and in December 2020, the Governor and his Executive Council approved the contract. Once the vendor was selected, the NBSP worked with OZ to carry out deliverables to get the system operational. Deliverables included set up of the system for New Hampshire, reviewing security protocols, UAT testing, and moving from development to production. On July 1, 2021, the OZ system went live and birth hospitals began entering demographic data, Critical Congenital Heart Disease (CCHD) results, hearing screen results and birth conditions. The NBSP continues to work with the vendor on the integration of a vital records into OZ, which will allow the NBSP to track that all infants born in the state receive CCHD and hearing screening along with their newborn screen. The OZ System has the capacity to interface with all birthing hospitals' electronic medical record (EMR) systems using established health data exchange standards.

UPCOMING PLANS

In the winter of 2021, the NSBP began discussions for updating the Administrative Rules, He-P 3008, to allow for information disclosure for follow up of abnormal screening results, updated specimen collection guidance to align with national recommendations, and updated term limits for the Advisory Committee Co-Chairperson. Rule amendments are anticipated to go to the Joint Legislative Committee on Administrative Rules (JLCAR) in early 2023.

Upcoming plans are to develop protocols and processes for short term and long term newborn screening follow up. The NBSP currently tracks infants with abnormal screens through confirmation or rule out of disorder, though has encountered challenges in obtaining follow up information. In 2021, certain providers and specialists had difficulty sharing diagnostic information with the NBSP due to data sharing concerns. Future amendments to the Administrative Rules HeP 3008 will allow for collection of information to facilitate NBSP follow up activities.

Newborn screening remains a dynamic field. Advances in science and technology are ongoing and will continue to impact the state's program. The NBSP will continue to monitor quality assurance and plans to resume providing Unsatisfactory Specimen & Timeliness reports to all birth facilities in the state. In the coming year, the program will maximize the use of data to ensure no infant screens are missed; ensure that specimens are tested and followed up in a timely manner; and that all New Hampshire infants are screened for the approved screening panel conditions.

APPENDIX A

New Hampshire Newborn Screening Panel as of January 1, 2022	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

*Newborn hearing screening is also offered at all NH hospitals with birth facilities.