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

Calendar Year 2020

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
April 2021

NH DIVISION OF
Public Health Services
Department of Health and Human Services

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 has responsibility for oversight of the New Hampshire Newborn Screening Program (NBSP). The NBSP is responsible for assuring that all infants born in New Hampshire are screened; daily management of screening results; assuring that screening is timely and complete for each infant; and that any infants identified through this process receive timely 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 entails that all infants born in New Hampshire 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 132:10-a 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/dphs/bchs/mch/newborn.htm that provide additional background and historical information.

CALENDAR YEAR 2020

DATA

From January 1, 2020 through December 31, 2020, the NBSP screened 11,742 infants, born in New Hampshire, which represented more than 99% of all occurring births. Tables 1 and 2 contain the NBSP statistics for 2020. This includes the number of infants who were screened; declined; died; transferred out of state prior to having an initial screen; lost to follow-up; number of presumptive positives or out of range on the initial screening, disorders identified; and pending confirmation of diagnoses in that year.

Table 1: 2020 NBSP data

Total NH births	**11,829**
Number of NH births screened	11,743
Screening declined	19
Lost to follow –up	7
Died	24
Transferred out of State (Initial screening completed outside NH)	36
Missed	0

Table 2: NBSP disorders identified for 2020

Disorder	Presumptive Positive (out of range)
Argininemia	4
Biotinidase	1
Acylcarntine Disorders	30
Congenital Adrenal Hyperplasia (CAH)	57
Cystic Fibrosis	51
Galactosemia	1
Hemoglobinopathies (3 types)	151
Congenital Toxoplasmosis	2
Maple Syrup Urine Disease	25
Homocystinuria	91
Phenylketonuria	21
Aminoacidopathies disorder	5
Pompe	1
Severe Combined Immunodeficiency Disorder	53
Spinal Muscular Dystrophy	1
Congenital Hypothyroidism	113
Total Presumptive Positive (out of range)	**607**

Table 3: NBSP disorders with Conformed Diagnosis for 2020

Disorder	Confirmed Diagnosis
Congenital Hypothyroidism	7
Cystic Fibrosis	4
Biotinidase	1
Congenital Toxoplasmosis	1
Congenital Adrenal Hyperplasia (CAH)	1
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD, an Acylcarntine Disorder)	1
Other*	1
Total Confirmed Diagnosis	**16**
Pending confirmation	14

**due to NBS testing other conditions were identified through follow up testing*

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 consults with physician metabolic specialist, Dr. Amy Kritzer of Boston Children's Hospital.

The NBSP's Public Health Nurse Coordinator retired at the end of 2019 and efforts began to recruit and fill this critical job. The COVID-19 pandemic and a hiring freeze for much of 2020 impacted the posting and filling of this position. After a long search, the NBSP hired a 1.0 FTE Public Health Nurse Coordinator in early 2021. The NBSP Public Health Nurse Coordinator brings expertise in nursing care coordination and medical referrals. Shortly before, in the fall of 2020, a 0.5 FTE Nurse Coordinator was hired, who brings years of nursing experience, both in teaching and in clinical skills.

The State's Laboratory contractor for the NBSP, the New England Newborn Screening Program at the University of Massachusetts' Medical School (the Laboratory) has provided these services for more than twenty years, is in year three of the amended contract that was approved by the Governor and his Executive Council on June 20, 2018. The NBSP is currently in the final phase of developing a Request for Proposal (RFP) for a laboratory to conduct analysis utilizing standardized, approved laboratory methods for the disorders on the NH screening panel. It is anticipated that the RFP will be posted in the summer of 2021.

NEWBORN SCREENING ADVISORY COMMITTEE

The Newborn Screening Advisory Committee (NBSAC) in a non-pandemic time would meet on a bi-annual basis, also in accordance with Administrative Rule He-P 3008AND RSA 132:10-a. However, in 2020, the NBSAC only met once virtually on October 14th.

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.

Often, clinical specialists are brought in to present and answer questions before a final vote, done by Australian ballot, is made. The Laboratory also presents on test reliability and potential false positives/negatives. At the October 2019 meeting, Pompe, X-ALD and MPS-1 were voted on and were recommended for addition to the screening panel. At the

following year's NBSAC in October of 2020, discussion took place on protocol development, screening initiation, and follow up of the three disorders.

PROTOCOL DEVELOPMENT

After disorders are recommended for addition to the NBS panel by the NBSAC, there are then several steps necessary before the DHHS Commissioner can weigh in and make a final decision on the addition, according to Administrative Rule He-P 3008. This can and often takes several months and includes:

- The development of process and clinical protocols; e.g. where is an "out of range" infant referred to for a second confirmatory test; does the infant need the services of the NBSP physician consultant? If treatment is needed, where can this infant be referred to for timely attention?
- Determination if the filter paper fee needs to increase. The purchase of the filter papers by the birthing hospitals are used to collect the specimen from the infant. These fees in accordance with RSA 132:10-a and Administrative Rule He-P 3008 supports the NBSP through a revolving fund. All of the program expenses are covered by this fund with no use of general funds or federal funds for operations.

During the first half of 2020, protocols were developed for the potential addition of Pompe, X-ALD and MPS-1. This involved the collaboration of many colleagues included the Laboratory, clinical colleagues including the NBSP's physician metabolic consultant and several members of the NBSAC, in particular the Co-Chair, who is one of Dartmouth Hitchcock Medical Center's Genetic Counselors and specializes in newborn screening. The latter is often the first referral of newborns with positive screening results.

Like with the addition of SMA in the previous year, it was determined that the filter paper fee would need to increase from $104 to $146 per filter paper. Discussions with the New Hampshire Hospital Association previous to the addition of SMA at the end of 2019 had alerted birthing hospitals to this potential second rate increase earlier than normal. According to statute and administrative rule, birthing hospitals must have at least 30 days-notice before a rate change is to take place.

After all the steps were completed and with the DHHS Commissioner's approval, the NBSP began testing for Pompe, MSP-1, and X-ALD on August 26, 2020. Screening for SMA began on December 26, 2019. During 2020, seven presumptive positives were identified with the added disorders; protocols were effectively followed up so that the affected infants could receive timely care and further diagnostics.

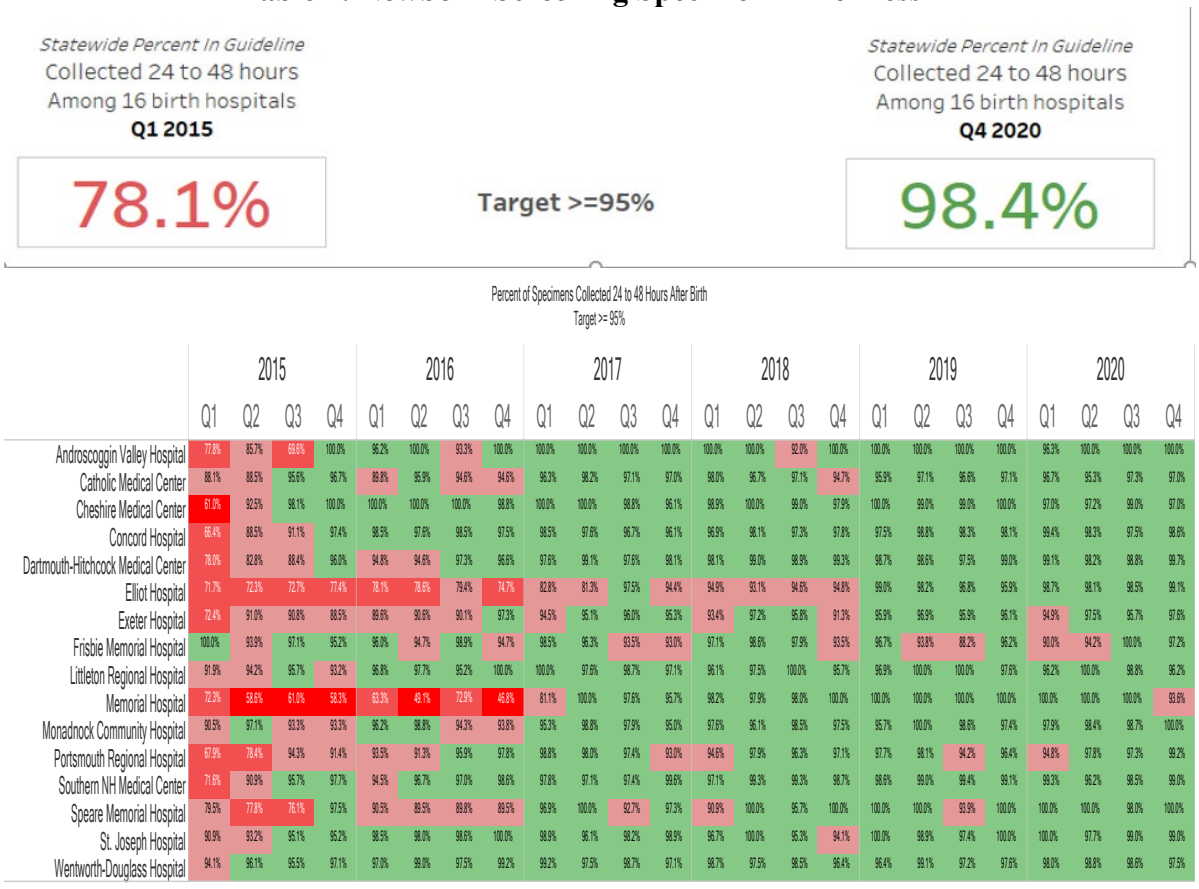
QUALITY IMPROVEMENT

The NBSP relies on timeliness and specimen quality for optimal results. The age of the infant at specimen draw, days lapsed between draw and arrival at the laboratory and the days between the blood draw and a 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 including the staff who complete the demographic information on the filter paper; the staff who obtain the specimen from the infant; the 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 demonstrate 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. Among the 16 current birth hospitals, the percentage of newborn screening specimens taken between 24-48 hours improved from 78.1% to 98.4% from Quarter 1 in 2015 to Quarter 4 in 2020. 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.

Table 4: Newborn Screening Specimen Timeliness



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 Laboratory (responsible for courier service within its State contract) utilizes the United Parcel Service (UPS) as the courier. During 2020, a quality improvement process focused on this metric in conjunction with the Laboratory and UPS. Prior years' delayed overnight deliveries made this timeliness metric unreliable and it was dropped from the quarterly reports given to the birthing hospitals. It is hoped with the additional focus that reliable data can again be provided by mid-2021.

Additional measures for quality improvement include the issue 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 who have the expertise to draw a repeat filter paper specimen. By having a repeat drawn results will be delayed.

Not only is the NBSP looking at unsatisfactory specimen for timeliness, the NBSP aims to be cost effective. Each additional repeat increases the cost to the facility. As mentioned above filter paper costs have increased to $146 per filter paper. From January 1, 2020 through December 31, 2020 the unsatisfactory specimen cost was $36,792.

From January 1, 2020 through December 31, 2020, the NBSP had 252 unsatisfactory specimen. The reasons for unsatisfactory specimens include; layer or clotted (126); poor soak (48); improperly dried (9); quantity not sufficient (5); contaminated (10); no blood (6); and expired filter papers (48).

DATA SYSTEM

With input from stakeholders, the Maternal and Child Health Section lead 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 currently have independent data collection systems. An RFP was released in November 2019; an extensive review process of received proposals was conducted and the Oz System was selected. On December 16, 2020, the Governor and his Executive Council approved the contract with the Oz System. The Oz System will interface with all birthing hospitals systems, state data systems, special screening devices, electronic medical records and the laboratory using established health data exchange standards. It is anticipated that all birthing hospitals will be utilizing the Oz System by the winter of 2022.

UPCOMING PLANS

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 and provide quality assurance reports to all locations of birth in the state. In the coming year, the program will maximize the use of data to ensure no infants are missed; ensure that specimens are tested and followed up in a timely manner; and that New Hampshire infants are screened for the appropriate conditions (Appendix A).

The NBSP has begun the initial phase for the pilot project for the deployment of the Oz System at birth hospitals. A presentation to the NH Hospital Association was done to

provide high level operations of the system and the time and effort each birth hospital would need to dedicate to the project. In the upcoming months, the NBSP will work with one to two birth hospitals on rolling out the Oz system for real time data collection.

In addition, the NBSP will be writing a Request for Proposals for its laboratory contract to be released fall/winter 2021/2022 with a starting date of July 1st 2022.

Appendix A

New Hampshire Newborn Screening Panel as of January 1, 2021	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.