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

Calendar Year 2019

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

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NH DIVISION OF Public Health Services
Improving health, preventing disease, reducing costs for all

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). This includes daily management of screening results; assuring that all infants born in New Hampshire are screened; 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 requires 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 Health and Human Services DHHS 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 2019

DATA

From January 1, 2019 through December 31, 2019, the NBSP screened 11,743 infants, born in New Hampshire, which represented more than 99% of all occurring births. Tables 1 and 2 contain the NBSP statistics for 2019. This includes the number of disorders identified in that year; the number of infants who missed newborn screening; the number of infants whose families refused newborn screening; and information on the usage of the metabolic medical consultant services.

Table 1: 2019 NBSP data

Total NH births	11,807
Number of NH births screened	11,743
Screening declined	16
Missed screening	1
Died	14
Transferred out of State (Initial screening completed outside NH)	20

Table 2: NBSP disorders identified for 2019

Disorder	Number positively identified
Congenital Hypothyroidism	3
Cystic Fibrosis	2
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	1
Isovaleric Acidemia (IVA)	1
Hemoglobin E (HbE) disease	1
Sickle Cell	1
Glutaric Aciduria Type 1 (GA I)	1
Total	10

**Other findings include infant carrier status, false positives, transient findings and maternal disorders.*

NEWBORN SCREENING ADVISORY COMMITTEE

The Newborn Screening Advisory Committee (NSAC) meets on a bi-annual basis, also in accordance with RSA 132:10-a and Administrative Rule He-P 3008. The NSAC 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 is made. The New England Newborn Screening Program at the University of Massachusetts' Medical School (the Laboratory), the State's contracted laboratory, also presents on test reliability and potential false positives/negatives. At the October 2018 meeting, Spinal Muscular Atrophy (SMA) was voted in as a recommendation for addition to the panel. According to Administrative Rule He-P 3008, there are then several steps necessary before the DHHS Commissioner can weigh in and make a final decision on the addition. 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.

Protocols were developed for the potential addition of SMA. In this case, it was determined that the filter paper fee would need to increase from $71 to $104 based on the screening for the new disorder as well as an additional Sunday courier pickup (discussed in the forthcoming timeliness section). Since this was the first significant increase in filter paper costs in seven years, financial discussions were held with the New Hampshire Hospital Association and its members. According to statute and administrative rule, hospitals must have at least 30 days-notice before a rate change is to take place.

Also during this this time period, a sole source amendment to the Laboratory contract was being negotiated and scheduled for a hearing at the Governor and his Executive Council meeting.

After all the steps were completed and with the DHHS Commissioner's approval, the NBSP began testing for SMA late in 2019.

The NBSAC met twice during calendar year 2019. During those meetings, Pompe, X-ALD and MPS-1 were discussed in detail as potential additions to the screening panel. During 2018 and part of 2019, the laboratory was pilot testing the equipment needed for confirmation of the aforementioned disorders. The NBSAC voted to recommend all three disorders at the meeting in October of 2019. The development of protocols, etc. before the submission to the DHHS Commissioner for review is currently taking place.

STAFFING

Because 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's longtime medical consultant, Dr. Harvey Levy of Boston Children's Hospital, retired this past year. A procurement process soliciting a new highly specialized medical consultant took place and Dr. Amy Kritzer, also of Boston Children's Hospital was selected.

The NBSP's 1.0 FTE Coordinator also retired after more than a decade in the position. Efforts are currently underway to recruit and fill this this critical job. A 0.6 FTE Nurse Coordinator was hired into the NBSP bringing many years of nursing experience, both in teaching and in clinical skills. The NBSP also has a 0.5 FTE Program Assistant who fulfills many of the clerical and administrative duties necessary in such a program.

As was stated previously, the State's Laboratory contractor for the NBSP, the New England Newborn Screening Program at the University of Massachusetts' Medical School had their contract amended and approved by the Governor and his Executive Council for another two years. The Laboratory recently changed their on-line data system, which required the technical assistance of DHHS's information technology and data security staff to help the NBSP get back online and functioning with minimal disruption to the daily workflow.

QUALITY IMPROVEMENT

The NBSP relies on timeliness for optimal results. The age of an 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; and the staff who are responsible for sending the dried specimens to the laboratory. 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.

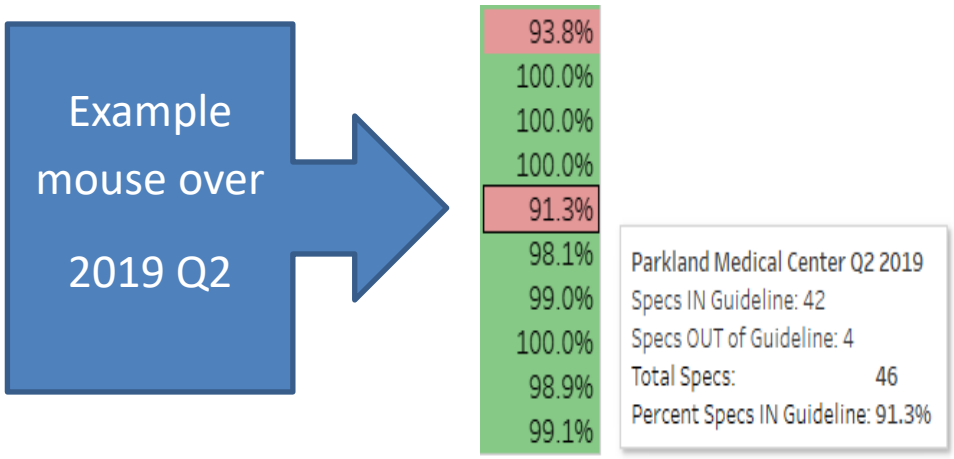
Beginning in early 2015, the NBSP has provided a quarterly timeliness progress report to all birthing hospitals focused on measures primarily within the control of hospital staff.

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. Statewide, more than 97% of specimens collected in hospitals in 2019 were collected between 24 and 48 hours (Table 1A).

Table 1a
Percent of Specimens Collected 24 to 48 Hours After Birth
Target >= 95%

	2015				2016				2017				2018				2019			
	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4	Q1	Q2	Q3	Q4
Androscoggin Valley Hospital	77.8%	85.7%	69.6%	100.0%	96.2%	100.0%	93.3%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	100.0%	92.0%	100.0%	100.0%	100.0%	100.0%	100.0%
Catholic Medical Center	88.1%	88.5%	95.6%	96.7%	89.8%	95.9%	94.6%	94.6%	96.3%	98.2%	97.1%	97.0%	98.0%	96.7%	97.1%	94.7%	95.9%	97.1%	96.6%	97.1%
Cheshire Medical Center	61.0%	92.5%	98.1%	100.0%	100.0%	100.0%	100.0%	98.8%	100.0%	100.0%	98.8%	96.1%	98.9%	100.0%	99.0%	97.9%	100.0%	99.0%	99.0%	100.0%
Concord Hospital	66.4%	88.5%	91.1%	97.4%	98.5%	97.6%	98.5%	97.5%	98.5%	97.6%	96.7%	96.1%	96.9%	98.1%	97.3%	97.8%	97.5%	98.8%	98.3%	98.1%
Dartmouth-Hitchcock Medical Center	78.0%	82.8%	88.4%	96.0%	94.8%	94.6%	97.3%	96.6%	97.6%	99.1%	97.6%	98.1%	98.1%	99.0%	98.9%	99.3%	98.7%	98.6%	97.5%	99.0%
Elliot Hospital	71.7%	72.3%	72.7%	77.4%	78.1%	78.6%	79.4%	74.7%	82.8%	81.3%	97.5%	94.4%	94.9%	93.1%	94.6%	94.8%	99.0%	98.2%	96.8%	95.9%
Exeter Hospital	72.4%	91.0%	90.8%	88.5%	89.6%	90.6%	90.1%	97.3%	94.5%	95.1%	96.0%	95.3%	93.4%	97.2%	95.8%	91.3%	95.9%	96.9%	95.9%	96.1%
Frisbie Memorial Hospital	100.0%	93.9%	97.1%	95.2%	96.0%	94.7%	98.9%	94.7%	98.5%	96.3%	93.5%	93.0%	97.1%	98.6%	97.9%	93.5%	96.7%	93.8%	88.2%	96.2%
Littleton Regional Hospital	91.9%	94.2%	95.7%	93.2%	96.8%	97.7%	95.2%	100.0%	100.0%	97.6%	98.7%	97.1%	96.1%	97.5%	100.0%	95.7%	96.9%	100.0%	100.0%	97.6%
Memorial Hospital	72.3%	58.6%	61.0%	58.3%	63.3%	49.1%	72.9%	46.8%	81.1%	100.0%	97.6%	95.7%	98.2%	97.9%	98.0%	100.0%	100.0%	100.0%	100.0%	100.0%
Monadnock Community Hospital	90.5%	97.1%	93.3%	93.3%	96.2%	98.8%	94.3%	93.8%	95.3%	98.8%	97.9%	95.0%	97.6%	96.1%	98.5%	97.5%	95.7%	100.0%	98.6%	97.4%
Parkland Medical Center	81.4%	87.2%	82.9%	82.1%	92.3%	96.0%	87.0%	84.6%	97.3%	88.3%	97.8%	97.4%	91.2%	85.7%	96.2%	86.8%	97.3%	91.3%	95.5%	91.7%
Portsmouth Regional Hospital	67.9%	78.4%	94.3%	91.4%	93.5%	91.3%	95.9%	97.8%	98.8%	98.0%	97.4%	93.0%	94.6%	97.9%	96.3%	97.1%	97.7%	98.1%	94.2%	96.4%
Southern NH Medical Center	71.6%	90.9%	95.7%	97.7%	94.5%	96.7%	97.0%	98.6%	97.8%	97.1%	97.4%	99.6%	97.1%	99.3%	99.3%	98.7%	98.6%	99.0%	99.4%	99.1%
Speare Memorial Hospital	79.5%	77.8%	76.1%	97.5%	90.5%	89.5%	89.8%	89.5%	96.9%	100.0%	92.7%	97.3%	90.9%	100.0%	95.7%	100.0%	100.0%	100.0%	93.9%	100.0%
St. Joseph Hospital	90.9%	93.2%	95.1%	95.2%	98.5%	98.0%	98.6%	100.0%	98.9%	96.1%	98.2%	98.9%	96.7%	100.0%	95.3%	94.1%	100.0%	98.9%	97.4%	100.0%
Wentworth-Douglass Hospital	94.1%	96.1%	95.5%	97.1%	97.0%	99.0%	97.5%	99.2%	99.2%	97.5%	98.7%	97.1%	98.7%	97.5%	98.5%	96.4%	96.4%	99.1%	97.2%	97.3%

In 2019, hospitals not achieving the 95% target tended to be lower volume (example, Parkland had just four specimens out of guideline in Q2 2019). The percentage alone can be misleading, so the Tableau version sent to all the birthing hospitals of the table provides mouse over functionality to provide important context. Overall, the statewide statistics show that the ≥95% target was consistently met throughout 2019.



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. United Parcel Service (UPS) overnight delivery failures beginning in the holiday season of 2017-2018 made

this metric unreliable and it was dropped from the reports given to the birthing hospitals until such time that reliable data become available.

A "What-If" analysis demonstrated the potential impact of dropping the Saturday morning courier pickup and adding a Sunday evening pickup. Specimens picked up Saturday morning are delivered to the lab on Monday because it is not open on Sundays. A Sunday evening pickup was included in the Laboratory contractual amendment and began in October of 2019. These specimens are also delivered to the lab on Monday morning, but include a significant number of weekend specimens obtained after the Saturday pickup took place. In the absence of reliable courier data, empirical evaluation of this change has not been possible. Access to courier data is still being negotiated. The NBSP continues to work on improving this service with the goal of improving timeliness.

In the upcoming year, additional measures for quality improvement will be looked at including the issue of unsaturated filter papers, which require a re-test involving the infant coming back in with accurate results being delayed. This also adds hours into the daily workflow of the NBSP.

DATA SYSTEM

With significant input from the NSAC and the IT departments of all of the birthing hospitals, the Maternal and Child Health Section lead the development of a Request for Proposals (RFP) for a scalable, secure web-based health 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 the summer of 2019, an extensive review process of received proposals was conducted and currently, a contractual process for the selected company is in process, hopefully to end in approval by the Governor and his Executive Council with a potential start date of July 1, 2020. The new web based health data management 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.

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

New Hampshire Newborn Screening Panel as of January 1, 2020	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

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