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

Calendar Year 2017

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*
Department of Health and Human Services

Overview

RSA 132:10-a requires that all infants born in New Hampshire (NH) be screened at birth for a panel of thirty-four (34) disorders, as determined by the State. (See Appendix A for current panel listing). Section 132:10-c allows parents or guardians to refuse this screening if they so desire. The Department of Health and Human Services, Division of Public Health Services, Bureau of Population Health and Community Services, Maternal and Child Health Section is responsible for oversight of the Newborn Screening Program, whose administration is detailed by Administrative Rules He-P 3008. 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.

With the establishment of the designated Newborn Screening Fund in August 2003 and the collection of $71 per filter paper for screening from birth hospitals, the program is self-sustaining. All of the program expenses are covered by this fund with no impact on the state budget and no use of general funds for operations. Infants who are identified with disorders through newborn screening require care, which is often covered by commercial insurance or in some cases by New Hampshire Medicaid.

RSA 132:10-a requires that the Department make an annual report to the Health and Human Services Oversight Committee on the findings of newborn screening in the prior year and any related fiscal impact. The reader is referred to the previous reports on file for additional background and historical information relative to the Newborn Screening Program operations.

2017 Activities

In 2017, New Hampshire screened 11,974 infants, which represented more than 99% of all occurring births. Appendix B contains the Newborn Screening Program statistics for calendar year 2017. This includes the number of disorders identified in that year, the number of infants who missed newborn screening, and the number of infants whose families refused newborn screening.

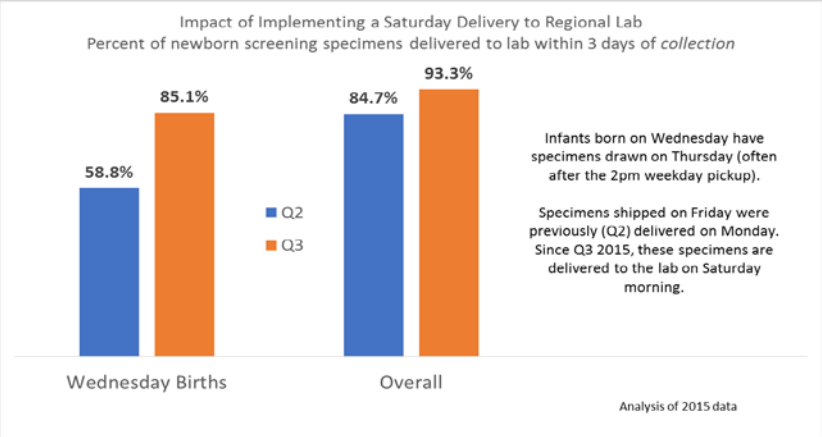
RSA 132:10-a enables a Newborn Screening Advisory Committee, which met in 2017. On its agenda were items such as a presentation on the readiness of the New England Newborn Screening Program at the University of Massachusetts Medical School, the current state laboratory contractor, to screen for Pompe, X-ALD and MPSI disorders. The Newborn Screening Advisory Committee is authorized to consider additions and then make recommendations to the Commissioner of the Department of Health and Human Services to the state's screening panel guided by the following criteria:

- 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.

The Advisory Committee is also guided by the Federal Secretary's Advisory Committee on Heritable Disorders in Newborns and Children, Recommended Uniform Screening Panel (RUSP). Pompe disease was added to the RUSP in 2013 and MPSI and X-ALD disorders were added in 2016. The University of Massachusetts New England Newborn Screening Program is currently pilot testing its screening algorithm for these disorders. Once this pilot is complete, New Hampshire Newborn Screening Advisory will review the data and scientific literature and reconsider adding the conditions to the New Hampshire panel.

The Advisory Committee also heard presentations in 2017 on the Newborn Screening Program's Timeliness Improvement Project. For the past several years, the state has participated in a national quality improvement project to improve the timeliness of newborn screening, which is a key factor in identifying and treating disorders. The Newborn Screening Program has worked in partnership with birth hospitals to improve the timeliness of specimen draw and the days lapsed between draw and arrival at the laboratory, in particular, measures primarily within the control of hospital staff such as the timely handoff to the UPS overnight courier. 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. The Newborn Screening Program provides each of the 19 birth hospitals with a quarterly report on timeliness outcomes as well as providing technical assistance for improvement. The data suggest that the birth hospitals are relatively timely in obtaining the specimen, but there are several factors that can affect the delivery to the laboratory. Through New Hampshire's contract with the New England Newborn Screening Program, UPS picks up specimens at each birth hospital on Monday through Saturday to be delivered the next day Monday through Friday. Saturday envelopes are delivered on Monday. The addition in the past few years of a Saturday pick-up significantly decreased the time from hospital to laboratory as evidenced by this analysis of 2015 data (when the Saturday pick-ups first occurred):



Improvement continues in 2017. Sixteen of 19 hospitals have reached ≥95% of specimens collected between 24 and 48 hours. Between July 1st and September 30th 2017, 97.3% of newborn specimens from birth hospitals were collected between 24 and 48 hours of birth, compared to 95.0% reported in the second quarter of 2017.

However, additional improvements need to be made in ensuring that specimens ship on time. Between July 1st and September 30th 2017, just over 92% of specimens shipped on time, with only 5 of 19 hospitals meeting or exceeding the state goal of 95% of on time shipping. While hospitals have made considerable improvements since 2015, the process of collecting a specimen from a newborn and sending by UPS to the New England Newborn Screening Laboratory requires complicated workflows. Staff at each hospital and within the DPHS Newborn Screening Program are to be commended for their continued commitment to quality improvement.

Because UPS does not offer Sunday delivery service, the Newborn Screening Program is continuing to investigate the feasibility of adding a Sunday pick-up by private courier. This would ensure that all infants, regardless of the day or time that they are born would have access to the most timely screening results.

Future Plans

Newborn screening remains a dynamic field. Advances in science and technology are ongoing and will continue to impact our state's program. The Newborn Screening Advisory Committee looks forward to reviewing the evolving research and literature and hearing from families regarding conditions such as Pompe, X-ALD, MPSI and Krabbe Leukodystrophy, among others.

The New Hampshire Newborn Screening Program will also continue to monitor and provide quarterly quality assurance reports to all birth hospitals. The program will continue the efforts around timeliness with an added emphasis on Cystic Fibrosis screening. The program is working on this particular initiative, whose objective is to get infants in earlier for definitive diagnoses, with the New Hampshire Cystic Fibrosis Center at Dartmouth Hitchcock.

Administrative Rules He-P 3008 will expire on June 30, 2018. The Newborn Screening Program revising its rules with input from the public and the Newborn Screening Advisory Committee with a final version to be considered by the Joint Legislative Committee on Administrative Rules (JLCAR) in the spring of 2018.

The role of the New Hampshire Newborn Screening Program is to improve the quality of all aspects of the Newborn Screening Process. In the coming year, the program will continue to 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, 2018	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
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
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.
* Screening for CCHD is by required RSA 132:10-a for all babies in NH regardless of birth place, effective 8/11/12.

Appendix B

2017 Newborn Screening results

TOTAL NH BIRTHS	12,039
SCREENING REFUSED	19
MISSED SCREENINGS	0
DIED	14
TRANS OUT OF STATE (Initial screening completed outside NH)	26
Other	6

Disorder	Number identified and confirmed as positive
Congenital Hypothyroidism	3
Cystic Fibrosis	4
Congenital Adrenal Hyperthyroidism	1
PKU	1
ASLD	1
Total	10

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