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

Calendar Year 2018

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

March 22nd, 2019

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

INTRODUCTION

The Department of Health and Human Services (the Department), 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 (NBS) Program. 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 35 disorders, as determined by the State. (See Appendix A for current panel listing). This statute includes a clause, 132:10-c, which allows parents or guardians to refuse this screening if they so desire.

RSA 132:10-a also includes a requirement 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 previous reports that are available on the Department's NBS program website (https://www.dhhs.nh.gov/dphs/bchs/mch/newborn.htm) provide additional background and historical information relative to the New Hampshire NBS Program operations.

SUMMARY OF ACTIVITIES FOR CALENDAR YEAR 2018

2018 Newborn Screening Program Data

For calendar year 2018, the NBS Program screened 11,897 infants, born in New Hampshire, which represented more than 99% of all occurring births. Tables1 and 2 contains the Newborn Screening Program statistics for calendar year 2018. 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.

Table1: Newborn Screening Program Data for 2018

TOTAL NH BIRTHS		11,962
Number of NH Births Screened		11,897
SCREENING REFUSED		9
MISSED SCREENINGS		0
DIED		14
TRANSFER OUT OF STATE (Initial screening completed outside NH)		28
Other		7

Table 2: Newborn Screening Program Findings, 2018

DISORDER	NUMBER IDENTIFIED
Congenital Hypothyroidism	3
Cystic Fibrosis	8
Tyrosinemia	1
Duarte Galactosemia	1
Total	13

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

Administrative Rules Update

New administrative rules, He-P 3008, were approved by the Joint Legislative Committee on Administrative Rules (JLCAR) and were effective with an amendment on September 28th, 2018. Approved changes to the Administrative Rule include: adding Critical Congenital Heart Disorder (CCHD) screening and screen results as performed via pulse oximetry to be reported to the Department; changing the expectation for the initial dried blood spot screen to 24-48 hours of life from 48-72 hours of life; adding Severe Combined Immunodeficiency Disorder (SCID) to required screening; and an update to require reporting results to the infant's medical record.

The newly adopted Administrative Rules will expire on June 30, 2028. There were no changes to the fee of $71, which is charged to the hospitals for the purchase of the filter papers used to collect the specimen from the infant. A fee increase to cover the cost of new screenings to be added to the panel will be considered in 2019 to make sure the program remains self-supporting with the designated Newborn Screening Revolving Fund. All of the program expenses are covered by this fund with no use of general funds or federal funds for operations.

Program Updates

The Newborn Screening Advisory Committee met in October of 2018. At this meeting, it voted to recommend Spinal Muscular Atrophy (SMA) to the screening panel. As per RSA 132:10-a, the Commissioner of the Department of Health and Human Services approved the addition in November 2018. The program is currently working on an amendment to the contract with the New England Newborn Screening Program (UMASS), the screening laboratory, for SMA as well as establishing specific protocols for referral.

The NBS Program 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. The NBS Program works closely with all 14 birthing hospitals, birthing centers and home birth providers to improve timeliness. All of the birth hospitals are provided with a confidential quarterly report on how they individually and in comparison to their de-identified peers are doing on hospital controlled outcomes.

The Critical Congenital Heart Disorder(CCHD) was added to the mandated panel in October 2016, authorizing the State to collect the data for surveillance. Because of the complex data transfer requirements for CCHD screening, New Hampshire has not yet begun data collection. Information and feedback has been solicited from hospitals so that a data solution will be acceptable within their IT infrastructure. The Maternal and Child Health Section is in the process of developing a Request for Proposals for such a data system. It is expected that the Request for Proposals will be posted in July 2019.

FUTURE PLANS

Newborn screening remains a dynamic field. Advances in science and technology are ongoing and will continue to impact the state's program.

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 Newborn Screening Program currently has a contract with Dr. Harvey Levy of Boston Children's Hospital to act as a Medical Consultant for New Hampshire providers. Dr. Levy will be retiring and the program has begun the procurement process to solicit for a new, highly specialized Medical Consultant.

The Newborn Screening Advisory Committee is planning to meet in the Spring of 2019. At that meeting Advisory Committee will be updated on the program's timeliness improvements and progress on establishing a data collection system for the reporting of CCHD (a request for proposals is slated to be released Spring/Summer of 2019). The addition of Pompe, X-ALD and MPS-1 will be considered per RSA 132:10-a as soon as UMASS New England Newborn Screening Laboratory has the capability of screening for these three conditions.

The Newborn Screening Program 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, 2019	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.