Michigan Department of
Health and Human Services

MICHIGAN NEWBORN SCREENING PROGRAM

Annual Report 2020

June 2022

MDHHS
Michigan Department of Health & Human Services

Acknowledgements

State of Michigan
Governor, Gretchen Whitmer

Michigan Department of Health and Human Services
Director, Elizabeth Hertel

Chief Medical Executive
Dr. Natasha Bagdasarian, MD

Public Health Administration
Senior Deputy Director, Dr. Alexis Travis, PhD

Bureau of Epidemiology and Population Health,
Director, Sarah Lyon-Callo, PhD, MA, MS

Bureau of Laboratories
Director, Sandip Shah, PhD

Our thanks to **Patricia McKane, DVM, MPH**, Director, Division of Lifecourse Epidemiology and Genomics, and **Chris Fussman, MS,** Manager, Maternal and Child Health Epidemiology Section, for guidance with this report.

We would like to thank everyone at the State Laboratory for their review of this report.

We would also like to thank all of our newborn screening follow-up coordinating centers and advisory committees for their efforts.

Authors

Isabel Hurden, MPH
Newborn Screening Epidemiologist, Maternal and Child Health Epidemiology Section,
Division of Lifecourse Epidemiology and Genomics

Rebecca Shaulis, BS
Quality Assurance Coordinator, Newborn Screening Follow-up Section,
Division of Lifecourse Epidemiology and Genomics

Mary Kleyn, MSc
Manager, Newborn Screening Follow-up Section,
Division of Lifecourse Epidemiology and Genomics

Table of Contents

Executive Summary

The Newborn Screening (NBS) Annual Report provides an overview of the Michigan NBS Program, screening performance metrics and quality assurance information.

Since the program began in 1965 with screening for phenylketonuria, more than 50 disorders have been added to the screening panel. Through 2020, more than 7.5 million infants have been screened with more than 7,400 diagnosed with diseases included in the NBS blood spot panel.

Of the 102,236 infants screened in 2020, the vast majority were Michigan residents and 307 (0.3 %) were diagnosed with a disease. Overall, one infant out of 333 screened was diagnosed with one of the disorders included in the NBS panel (see Table 1 for list of disorders).

Developments occurring in 2020:

Michigan continued to disseminate findings at both the state and national level:

- The findings from different studies and analyses related to NBS were presented at the:
    - ◊ Association of Public Health Laboratories (APHL) Newborn Screening and Genetic Testing Symposium (virtual).

Michigan continued to conduct NBS-related trainings:

- The NBS Follow-up Program held a virtual educational conference for hospital staff.

NBS follow-up staff presented or participated as an exhibitor at numerous educational events:

- Kalamazoo Baby Fair.
- Baby bag distribution in Lansing at the Cristo Rey Community Center in Lansing, NBS educational packets distributed to expecting parents.
- Lansing Maternal Infant Health Summit (virtual).
- Lansing Baby Fair (virtual).
- A presentation to Grand Valley State University (GVSU) Master's of Public Health (MPH) students (virtual).
- Two Michigan Department of Health and Human Services (MDHHS) Virtual Baby Fairs.

NBS laboratory personnel and follow-up staff continued to serve on national NBS committees, including:

- Education and Training workgroup for the Advisory Committee on Heritable Disorders in Newborns and Children.
- The Clinical Laboratory Standards Institute Document Development Committee.
- Critical Congenital Heart Disease (CCHD) Technical Assistance Workgroup.
- Health Information Technology (HIT) Association of Public Health Laboratories (APHL) work group.
- Quality assurance/Quality control APHL Subcommittee work group.

New screening developments:

- The NBS Program started screening for spinal muscular atrophy (SMA) in March 2020

Continuing work:

- The NBS Program continued working towards implementing screening for guanidinoacetate methyltransferase (GAMT). deficiency

Listing of Figures & Tables

Figures

Tables

Acronym Key

Acronym	Name
CCHD	Critical Congenital Heart Disease
EBC	Electronic Birth Certificate
FPR	False Positive Rate
MCIR	Michigan Care Improvement Registry
MDHHS	Michigan Department of Health and Human Services
MS/MS	Tandem Mass Spectrometry
NBS	Newborn Screening
NICU	Neonatal Intensive Care Unit
PCP	Primary Care Physician
PPV	Positive Predictive Value
QA	Quality Assurance
SCN	Special Care Nursery

I. Introduction

The Newborn Screening (NBS) Annual Report provides an overview of Michigan's NBS Program, screening performance metrics related to disorders included in the NBS panel and quality assurance information. This report does not contain any appendices as they have not changed since the last version of this report. All appendices, including the NBS research guidelines, supportive legislation and NBS advisory committees, can be found in previous reports.

This report is intended to provide:

- An introduction and historical account of the development of NBS in Michigan.
- Michigan screening performance metrics.
- Quality assurance information.

What is Newborn Screening?

NBS is the process of early identification of health conditions followed by their subsequent treatment before the onset of disease processes. Successfully screening, confirming and treating newborns with disorders in a timely manner minimizes the risk of long-term sequelae. Depending on the condition, potential outcomes of disorders in the NBS panel include, but are not limited to, brain/neurological damage, intellectual disability, damage to the liver, eyes or spleen or death if not detected early. To prevent these outcomes from occurring, NBS programs test blood spots collected from infants during the first few days of life and refer infants with abnormal screens for appropriate confirmatory testing and medical management.

NBS began in the 1960s when Dr. Robert Guthrie developed the bacterial inhibition assay to diagnose phenylketonuria (PKU) by determining the level of the amino acid phenylalanine in a drop of a baby's blood placed on a strip of filter paper. In 1965, Dr. K. Stanley Read at the Michigan Department of Public Health and Dr. Richard Allen at the University of Michigan introduced NBS for PKU to Michigan and almost immediately turned what had been a devastating, untreatable, genetic disorder into a condition readily manageable by a low protein diet (Figure 1).[1] In 1977, a test for congenital hypothyroidism (CH) was added to the NBS panel, and screening for galactosemia was initiated in 1985. Public Act 14 of 1987 mandated further expansion of screening with the addition of three disorders: biotinidase deficiency, maple syrup urine disease (MSUD), and hemoglobinopathies such as sickle cell disease (SCD). The Act also permitted MDHHS's state laboratory to be the sole testing site and mandated a fee to fund the program. In 1993, another endocrine disorder, congenital adrenal hyperplasia (CAH), was added to the screening panel.

The introduction of tandem mass spectrometry (MS/MS) in 2003 enabled the state laboratory to efficiently screen for a large number of disorders using a single blood spot. This technology replaced Dr. Guthrie's bacterial inhibition assays previously used to detect PKU and MSUD. The first additional disorder screened with this method was medium chain acyl-CoA dehydrogenase deficiency (MCAD), a disorder of fatty acid oxidation that can result in sudden death during periods of fasting. MS/MS technology allowed further expansion of the NBS screening panel in 2004 to include an additional three amino acid disorders: homocystinuria (HCY), citrullinemia (CIT), and argininosuccinic aciduria (ASA).

[1]For more information on the history of PKU and PKU-related NBS Program evaluations conducted in Michigan, see Chapter IV of the 2009 NBS Annual Report.

In 2005, a pilot project was initiated to expand the screening panel to 48 disorders by including the 31 additional MS/MS disorders recommended by the American College of Medical Genetics and Genomics (ACMG) and the March of Dimes. Screening for cystic fibrosis began in Michigan on October 1, 2007, meeting another ACMG recommendation. Hearing screening was also added to the NBS panel in 2007, but this report does not include hearing screening results.[1] Screening for severe combined immunodeficiency (SCID) began on October 1, 2011. Screening for Hemoglobin H disorder began in 2012. Pulse oximetry screening for critical congenital heart disease (CCHD) began in 2014. Information on pulse oximetry screening can be found in a separate annual report[2]. Pompe disease and Mucopolysaccharidosis Type 1 (MPS 1) were added to the panel in 2014 and 2015, respectively, and screening began in 2017. Screening for X-linked adrenoleukodystrophy began in October 2019. Screening for spinal muscular atrophy began in March 2020.

Table 1 provides the complete list of disorders currently screened for in Michigan. The highlighted disorders are those that are screened for in Michigan, but no cases have ever been identified and confirmed through NBS. Detailed information about the disorders included in the screening panel, confirmation of diagnoses, and follow-up of positive tests can be found in the Michigan NBS Guide, which is available by clicking on the "Resources for Hospitals and Health Professionals" link on the NBS website.(Michigan.gov/NewbornScreening).

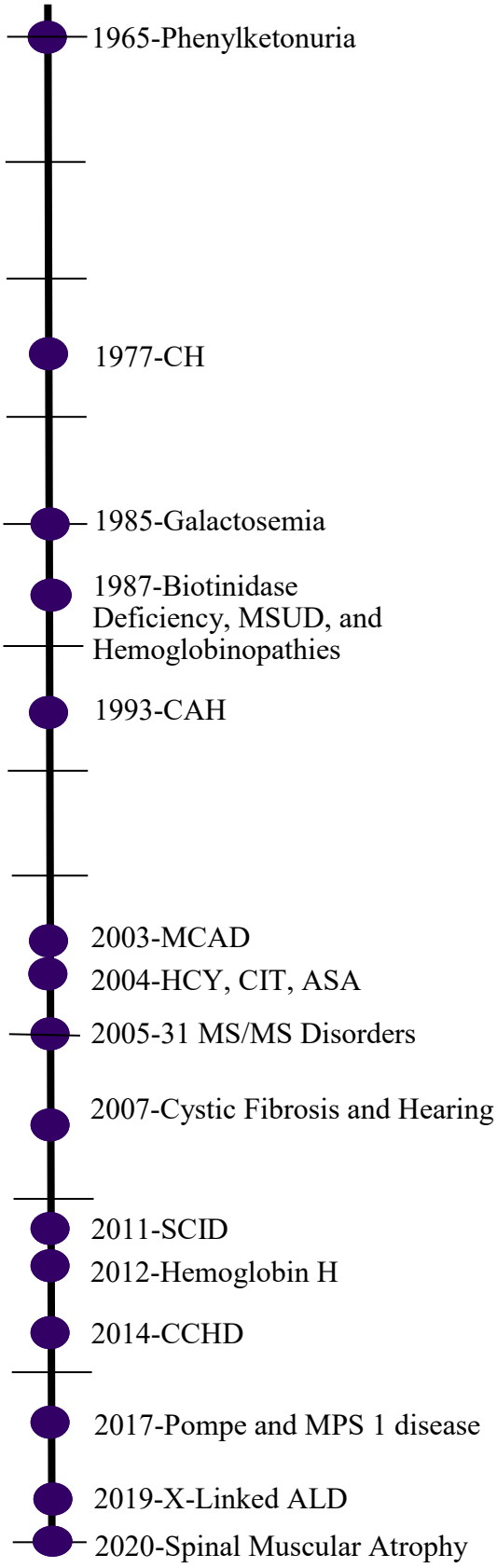

Figure 1. Implementation of Disorders on the NBS Panel, Michigan, 1965-2020

[1]More information about the newborn hearing screening program can be found at Michigan.gov/Ehdi.
[2] The critical congenital heart disease annual reports can be found here.

Table 1. Disorders included in the Newborn Screening Blood Spot Panel, Michigan, 2020

Amino Acid Disorders	Organic Acid Disorders
1. Argininemia	30. 2-Methyl-3-hydroxy butyric aciduria
2. Argininosuccinic acidemia	31. 2-Methylbutyryl-CoA dehydrogenase deficiency
3. Citrullinemia	32. 3-Hydroxy 3-methylglutaric aciduria
4. Citrullinemia Type II	33. 3-Methylcrotonyl-CoA carboxylase deficiency
5. Homocystinuria	34. 3-Methylglutaconic aciduria
6. Hypermethioninemia	35. Beta-ketothiolase deficiency
7. Maple syrup urine disease	36. Glutaric acidemia Type I
8. Phenylketonuria	37. Isovaleric acidemia
9. Benign hyperphenylalaninemia defect	38. Methylmalonic acidemia (Cbl A, B)
10. Biopterin cofactor biosynthesis defect	39. Methylmalonic acidemia (Cbl C, D)
11. Biopterin cofactor regeneration defect	40. Methylmalonic acidemia (mutase deficiency)
12. Tyrosinemia Type I	41. Multiple carboxylase deficiency
13. Tyrosinemia Type II	42. Propionic acidemia
14. Tyrosinemia Type III	**Hemoglobinopathies**
Fatty Acid Oxidation Disorders	43. S/Beta thalassemia
15. Carnitine acylcarnitine translocase deficiency	44. S/C disease
16. Carnitine palmitoyltransferase I deficiency	45. Sickle cell anemia
17. Carnitine palmitoyltransferase II deficiency	46. Variant hemoglobinopathies
18. Carnitine uptake defect	47. Hemoglobin H disease
19. Dienoyl-CoA reductase deficiency	**Endocrine Disorders**
20. Glutaric acidemia Type II	48. Congenital adrenal hyperplasia
21. Long-chain L-3-hydroxyl acyl-CoA dehydrogenase deficiency	49. Congenital hypothyroidism
22. Medium/short-chain L-3-hydroxyl acyl-CoA dehydrogenase deficiency	**Other Disorders**
23. Medium-chain acyl-CoA dehydrogenase deficiency	50. Biotinidase deficiency
24. Medium-chain ketoacyl-CoA thiolase deficiency	51. Galactosemia
26. Trifunctional protein deficiency	52. Cystic fibrosis
27. Very long-chain acyl-CoA dehydrogenase deficiency	53. Severe combined immunodeficiency
Lysosomal Storage Disorders	54. T-cell related lymphocyte deficiencies
28. Pompe Disease	55. X-Linked Adrenoleukodystrophy
29. Mucopolysaccharidosis I (MPS I)	56. Spinal Muscular Atrophy

Notes: Highlighted disorders have never been detected in Michigan through NBS. The following disorders are reported together because the same analyte(s) is used for screening: #3-4, #5/#6, #8-11, #13/#14, #15/#17, #21/#26, #31/#37,#32-34/ #41, #43/#39/42, , #30/#35.

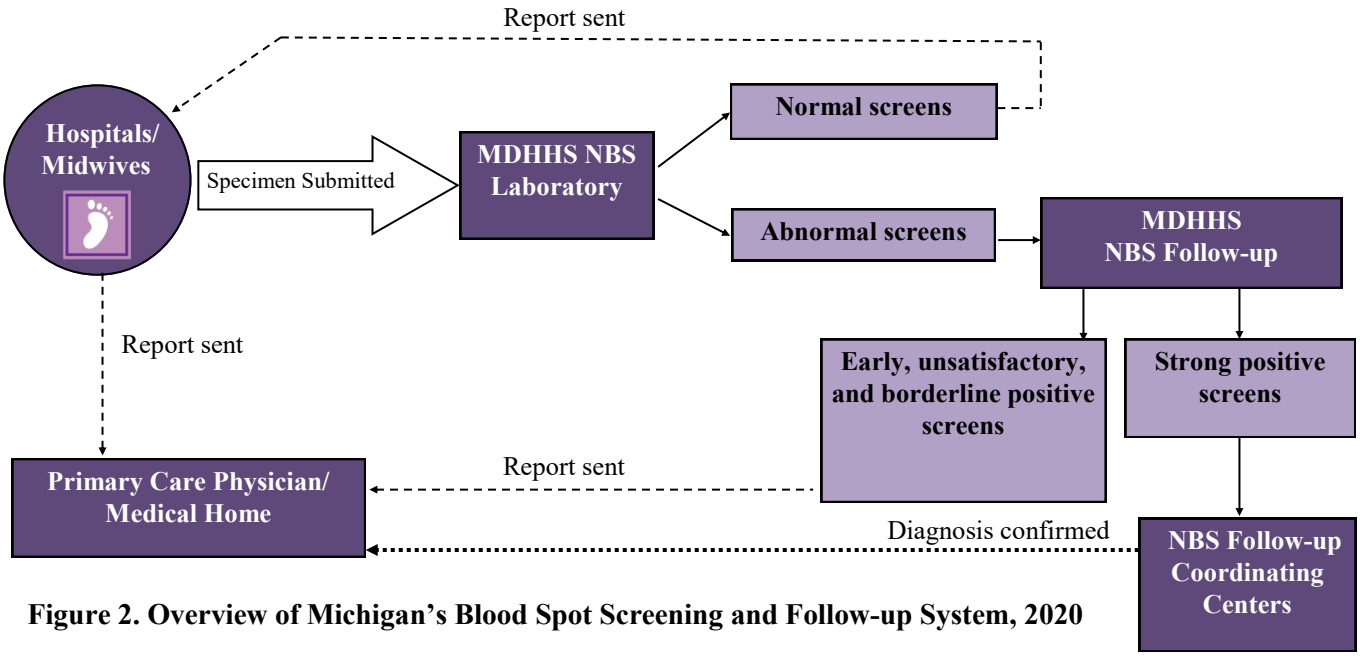

Figure 2. Overview of Michigan's Blood Spot Screening and Follow-up System, 2020

Abbreviations: MDHHS-Michigan Department of Health and Human Services; NBS-Newborn Screening.

HOSPITALS

In 2020, 80 Michigan hospitals had birthing units. Each hospital has a designated NBS Coordinator who helps facilitate the screening process. Hospital coordinators receive a quarterly quality assurance report from the NBS Follow-up Program that includes information on hospital-specific performance indicators compared to the state overall. Hospitals receive periodic site visits by the NBS Nurse Consultant to evaluate the screening process and make recommendations for improvement.

MIDWIVES AND HOME BIRTH ATTENDANTS

There are approximately 83 midwives registered with the NBS Program. Midwives are provided with individual assistance in meeting program standards. Although the number of midwife deliveries is small, they are more prevalent in the Amish and Mennonite communities, which have a higher incidence of several disorders included in the NBS panel.

MICHIGAN DEPARTMENT OF HEALTH AND HUMAN SERVICES

The MDHHS NBS Program includes the NBS Laboratory, the Follow-up Program, and medical management coordinating centers. More detailed descriptions of each entity are included in previous reports available on the NBS website (michigan.gov/newbornscreening).

II. Methods

This section describes the methods used to calculate: a) total number of newborns eligible for screening, b) total number of newborns diagnosed through the NBS process, c) the demographic characteristics of screened newborns, d) screening performance metrics, and e) quality assurance indicators.

TOTAL NUMBER OF NEWBORNS ELIGIBLE FOR SCREENING

Vital statistics data collected by the Vital Records & Health Data Development Section within the Division for Vital Records and Health Statistics at MDHHS was used to determine the total number of live births statewide that were eligible for screening. The number of live births in 2020 (n=103,239) is a preliminary estimate based on the number of births reported by December 2021.

TOTAL NUMBER OF NEWBORNS DIAGNOSED BY NEWBORN SCREENING

The MDHHS laboratory information system (PerkinElmer Life Sciences, Inc.) was used to identify positive cases. Data collected at the coordinating centers and managed by the NBS Follow-up Program was used to determine the total number of cases identified by NBS and to describe the population screened. Cases referred to in this report have the following characteristics: a) they were identified by NBS, b) they were Michigan residents, and c) they were identified and diagnosed through established laboratory and clinical protocols.

DEMOGRAPHIC CHARACTERISTICS OF SCREENED NEWBORNS

The demographic characteristics of screened newborns are presented for Michigan residents screened in Michigan. This report focuses on cases and screening results among Michigan residents only since out-of-state infants born within the state are followed up and diagnosed elsewhere.

Table 2. Screening Performance Indicator Descriptions

Indicator	Description
Newborns (N)	The total number of screened live births among in-state residents.
Total + (% NICU)	Total number of positive screens among in-state residents (the percentage of infants with positive screens who were admitted to the NICU among all infants with positive screens).
Positive	Screening value exceeds cutoff.
Strong +	Strong positive screen (in most cases considered a medical emergency and referred immediately for diagnostic testing).
Borderline +	Borderline positive screen (not a medical emergency and repeat screen requested).
Confirmed +	A diagnosis of a disorder that has been confirmed.
False +	A positive screen that is not confirmed as a case of a disease included in the NBS panel.
Detection Rate	The number of infants having a confirmed disorder out of the total number of infants screened, depicted as a ratio. One case per 'X' number of infants screened depicted as 1: 'X'.
FPR	False positive rate: the number of infants with false positive screens divided by the total number of infants screened, expressed as a percentage.
PPV	Positive predictive value: the number of infants confirmed with a disorder divided by the number of infants having positive screens, expressed as a percentage.

SCREENING PERFORMANCE METRICS

Table 2 provides a description of screening performance metrics included in subsequent tables. These indicators are commonly used to assess the performance of screening tests and allow for comparisons both over time and with other screening programs. Ideal screening tests have a high positive predictive value (perfect = 100%) and a low false positive rate (perfect = 0%). A perfect screening test correctly identifies all cases of a disorder with no false positives. Detection rates, the total number of cases identified out of the total number of newborns screened, are based on the total number of screens for in-state residents. Cases are defined as newborns identified with disorders via NBS. Maternal disorders and carriers identified by NBS are not included as confirmed cases in the performance metrics, though they are presented in separate tables in this report.

QUALITY ASSURANCE INDICATORS

Quality assurance (QA) data were obtained from NBS cards and information recorded by the state NBS laboratory and coordinating centers. Hospital quarterly reports contain QA indicators that focus on: a) time from birth to specimen collection, b) specimen arrival at the state NBS laboratory by the appropriate day, c) number of specimens that are unsatisfactory, d) number of birth certificates with NBS kit number recorded, and e) number of screened births with BioTrust consent form returned that is completed appropriately, and f) number of errors on the NBS card. Time from birth to start of treatment is another QA indicator used by the NBS Program.

III. Screening Results

DEMOGRAPHIC CHARACTERISTICS OF SCREENED NEWBORNS

This section describes the population of screened infants born in 2020 in terms of race, birth weight, gestational age and birth place (hospital regular nursery, NICU/SCN or non-hospital). These data are helpful in understanding the epidemiology (distribution of disease cases among the population) of the disorders covered in subsequent sections of this report. For example, sickle cell disease is predominantly found in Black populations, so the number of cases will fluctuate with the birth rate of Black populations.

The Michigan NBS Program screened 99.1% of the live births occurring in Michigan in 2020, as determined by the linkage of NBS records to preliminary live births records received from the Vital Records & Health Data Development Section and follow-up of unmatched records (Figure 3). Of the 103,239 live births that occurred in 2020, 331 were listed as deceased on their birth certificate. Many of these infants are not screened due to their short life spans, so they are excluded from the linkage calculations. Of the 102,908 remaining live births, the linkage algorithm successfully matched newborn screens for 101,739 infants (98.9%). The 1,169 unmatched records were sent to NBS Follow-up Program technicians for further investigation. This more in-depth follow-up revealed that 267 (22.8%) of the unmatched records were screened in Michigan. For these infants, the linkage algorithm failed to create the match for a variety of reasons, including data recording errors, data entry errors, or name changes due to adoptions. Overall, 901 infants (0.7%) born in the state were not screened in Michigan. Of those 901 not screened in Michigan, 106 were screened out of state. Of the remaining 795 infants not screened, 511 (64.1%) were born in a non-hospital setting.

Infants were not screened due to parental refusal of screening (n=409), infant expired or receiving palliative care (n=46) or some other reason for not being screened or reason is unknown (n=328). For all infants without a newborn screen, NBS Follow-up staff either contact the NBS coordinator for hospital births or send a parental notification and midwife notification letter for home births. In 2020, 24 infants born in hospitals are known to have missed being screened, and those hospitals were contacted. Twelve of the 24 babies were screened after NBS follow up program staff contacted the hospitals.

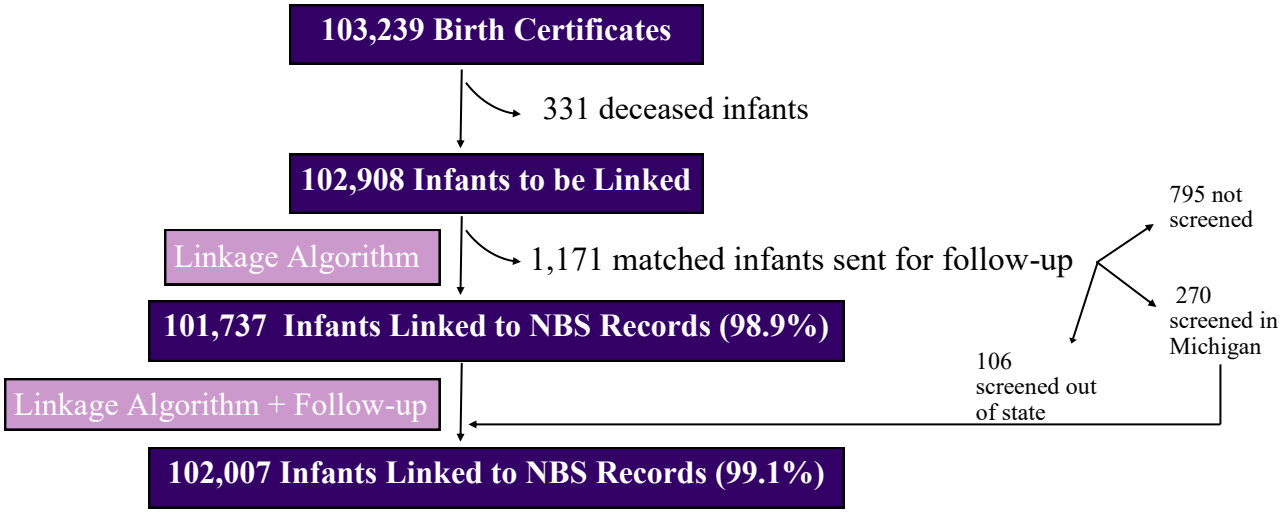

Figure 3. Newborn Screening and Live Births Records Linkage, Michigan, 2020

In total, 102,236 NBS samples were received from infants born in 2020. Of those, 287 (0.3% of screens) belonged to out-of-state residents or were collected out-of-state. Table 3 reports the demographic and perinatal characteristics by race of screened in-state residents born in 2020. This report details the screening results for in-state residents only since non-residents are typically followed in their home state. As indicated in Table 3, the majority of in-state infants screened were white, born in hospital nurseries, term (≥37 weeks gestational age), and of normal birth weight (>2,500 g). Overall, 10.9% of in-state infants screened were admitted to the NICU or special care nursery (SCN), 8.5% were low birthweight (<2,500 grams), and 9.6% were born preterm (<37 weeks gestational age). Black infants were over-represented among NICU, preterm and low birthweight births.

Table 3: Demographics of Infants Screened by Race, Michigan, 2020, Excluding Out-of-State Residents

Race	Column Total		Nursery Type						Birth Weight (g)		Gestational Age (wks.)	
			Regular Hospital		NICU/SCN		Non-Hospital		<2500		<37	
	N	%	N	%	N	%	N	%	N	%	N	%
White	62,335	61.1	55,189	88.5	6,075	9.8	1,071	1.7	4,122	6.7	5,263	8.5
Black	18,704	18.4	15,388	82.3	3,250	17.4	66	0.4	2,749	14.9	2,658	14.4
Multi-Racial	6,988	6.9	6,297	90.1	616	8.8	75	1.1	540	7.8	661	9.5
Other	7,839	7.7	7,165	91.4	659	8.4	15	0.2	664	8.6	573	7.4
Missing	6,083	6.0	5,501	90.4	538	8.8	44	0.7	489	8.3	532	9.1
Column Total:	101,949	100.0	89,540	87.8	11,138	10.9	1,271	1.3	8,564	8.5	9,687	9.6

Notes: All percentages are row percentages except for Column Total which is a column percentage. All characteristics are as recorded on the newborn screening card. A total of 1,378 and 1,149 newborns were missing birth weight and gestational age on the card, respectively. Non-hospital nursery type includes home births, births that occurred at birthing centers, and all other births that did not occur at a hospital.

SCREENING OUTCOME INFORMATION

In the following subsections, outcome information is provided for the disorders included in the NBS panel in 2020. The total numbers of cases detected both in and through 2020 are presented along with screening performance metrics.

CUMULATIVE DETECTION RATE

Table 4 reports the cumulative detection rate of disorders identified via NBS by classification both in and through 2020. The metabolic disorders detected by MS/MS are grouped by category (amino acid, organic acid and fatty acid oxidation disorders). Two metabolic disorders, galactosemia and biotinidase deficiency, are detectable by enzyme assay screening rather than MS/MS and are listed separately. The galactosemia cumulative detection rate includes both Duarte compound heterozygotes (D/G) and classic galactosemia (G/G). However, only D/G cases that have been detected since 2004, the year that CHMMC began short-term treatment of this disorder, are included in the cumulative detection rate. Similarly, the biotinidase deficiency cumulative detection rate includes both partial and profound biotinidase deficiency. Treatment of partial biotinidase deficiency did not begin until 2000.

Table 4: Disorders Identified in Newborns via Newborn Screening, 1965-2020

Type of Disorder Classification (Year Screening Began)	Cases in 2020 (N)	Cases Through 2020 (N)	Cumulative Detection Rate
Galactosemia (1985)	3	222	1:20,620
Biotinidase Deficiencies (1987)	17	377	1:11,130
Amino Acid Disorders (1965)	11	803	1:9,253
Organic Acid Disorders (2005)	8	108	1:15,895
Fatty Acid Oxidation Disorders (2003)	7	294	1:6,725
Congenital Hypothyroidism (1977)	121	2,706	1:1,551
Congenital Adrenal Hyperplasia (1993)	9	184	1:18,052
Sickle Cell Disease (1987)	73	2,167	1:1,935
Hemoglobin H Disease (2012)	1	17	1:51,641
Cystic Fibrosis (2007)	26	348	1:3,843
Primary Immunodeficiencies (2011)	13	134	1:7,389
Lysosomal Storage Disorders (2017)	7	24	1:10,675
X-Linked Adrenoleukodystrophy (2019)	1	2	1:63,830
Spinal Muscular Atrophy* (2020)	10	12	1:9,898
Total	307	7,401	-

*Two cases of SMA were detected in 2019 during population studies

Figure 4 displays detection rates in 2020 and overall. CH, the most commonly diagnosed disorder, accounted for 39.4% of cases diagnosed in 2020 and 36.6% cumulatively. Sickle cell disease, the second most commonly diagnosed disorder, accounted for 23.8% of cases detected in 2020 and 29.3% detected cumulatively. Cystic fibrosis (CF) was the third most commonly diagnosed disorder in 2020 and accounted for 8.5% of cases detected in 2020 and 4.7% of cases detected cumulatively.

Disorders detected by MS/MS (amino acid, organic acid, and fatty acid oxidation disorders) accounted for 8.5% of cases in 2020 and 16.3% cumulatively. However, PKU, the first disorder screened in Michigan, is now screened by MS/MS, meaning the overall proportion of cases detected by MS/MS is an overestimate because it includes cases detected prior to 2003 when MS/MS screening was initiated. The cumulative detection rate for fatty acid oxidation disorders (4.0%) is an underestimate because MCAD screening began in 2003, while screening for other conditions did not begin until 2005. This means that births included in the denominator from 2003-2005 were not eligible for diagnosis of a fatty acid oxidation disorders other than MCAD leading to an artificially low cumulative detection rate.

Screening for lysosomal storage disorders started in August 2017 and accounted for 2.3% of cases diagnosed in 2020, but only 0.3% of cases detected cumulatively. Screening for X-linked adrenoleukodystrophy (X-ALD) began in October 2019. X-ALD this accounts for 0.3% of cases diagnosed in 2020, but only 0.03% of cases detected cumulatively. Screening for SMA began in March 2020. SMA cases account for 3.3% of cases detected in 2020, and 0.2% of cases detected cumulatively.

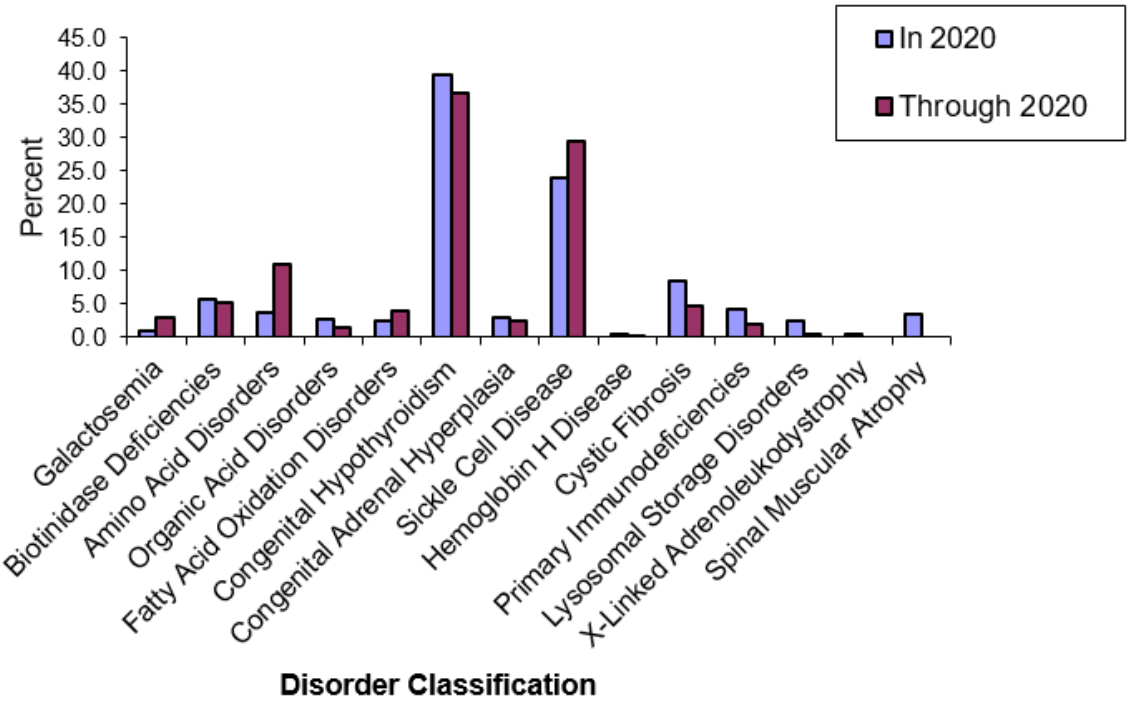

Figure 4. Percent Distribution of Disorders Identified in Newborns via Newborn Screening, Michigan Residents, in 2020 and through 2020

SCREENING PERFORMANCE METRICS

Screening performance metric targets are available in previous annual reports. Screening performance metrics include the detection rate, false positive rate (FPR), and positive predictive value (PPV). Table 5 reports screening performance metrics for all disorders in 2020.

Performance metrics for individual MS/MS disorders are provided in separate tables (see Tables 7-9).

GALACTOSEMIA

Three cases of classic galactosemia were detected in 2020, resulting in a FPR of 0.003% and PPV of 75%.

BIOTINIDASE DEFICIENCY

Seventeen cases of biotinidase deficiency were detected in 2020, one was profound and 16 were partial biotinidase deficiency cases. The detection rate was 1:6,372. The FPR and PPV were 0.03% and 34%, respectively.

CYSTIC FIBROSIS

Twenty-six cases of CF were detected in 2020 (detection rate-1:3,921). The associated FPR and PPV were 0.3% and 7.8%, respectively. Additionally, eight cases of CFTR-related metabolic syndrome were detected. Chapter IV of the 2008 Annual Report provides more detailed information describing CF screening in Michigan.

X-LINKED ADRENOLEUKODYSTROPHY

One infant was identified with X-ALD in 2020 resulting in a FPR of 0.01% and PPV of 7.1%.

SPINAL MUSCULAR ATROPY

Ten infants were identified with SMA in 2020 resulting in a FPR of 0.0% and a PPV of 100%

ENDOCRINE DISORDERS-CH AND CAH

A total of 121 cases of CH were detected in 2020. The CH screening FPR was 1.9%, and the PPV was 6.0%. The overall detection rate for CH was 1:843. Chapter IV of the 2007 Annual Report provides more detailed information describing CH screening in Michigan.

Nine cases of CAH were detected in 2020, eight were non-salt wasting and one was salt-wasting, The CAH screening FPR was 0.1%, and the PPV was 9.8%. The overall detection rate for CAH was 1:11,328.

HEMOGLOBINOPATHIES

Hemoglobinopathies include sickle cell disease (SCD) and Hemoglobin H disease. One case of Hemoglobin H were detected in 2020, resulting in a detection rate of 1:101,949. The Hemoglobin H disease FPR was 0.02% and the PPV was 4.8%.

Additional SCD screening outcome information is reported in Table 6. SCD screening differs from screening for the other disorders because the purpose is to identify the presence or absence of abnormal hemoglobin and not to quantify selected analytes. There is no screening reference range, and the results of screening are essentially considered a confirmatory diagnosis. Confirmatory testing is primarily for differentiating sickling genotypes.

As depicted in Table 6, SCDs are quite common among Black populations. While the overall incidence of SCD is one case per 1,367 screened, the incidence in Black populations is one in 317 screened in Michigan.

In addition, two cases of hemoglobin C thalassemia plus and one case of hemoglobin C disease was also identified.

Table 5: Screening Results and Performance Metrics, Michigan, 2020

Disorder Type	Total N	Total + N (% NICU)	Confirmed + N	Positive Detection Rate	FPR %	PPV %
Galactosemia	101,949	6 (50.0)			0.003	50.0
Classic (GG)			3	1:33,983		
Duarte (DG)			0	-		
Total			3	1:33,983		
Biotinidase Deficiency		50 (8.0)			0.03	34.0
Profound			1	1:101,949		
Partial			16	1:6,372		
Total			17	1:5,997		
Cystic Fibrosis (CF)		334 (18.9)	26	1:3,921	0.3	7.8
Congenital Hypothyroidism (CH)		2,021 (19.0)	121	1:843	1.9	6.0
Congenital Adrenal Hyperplasias (CAH)		92 (97.9)			0.1	9.8
Salt wasting			8	1:12,744		
Non-Salt wasting			1	1:101,949		
Total			9	1:11,328		
Sickle Cell Disease (SCD)		95 (9.5)	73	1:1,397	0.02	76.8
Hemoglobin H Disease		21 (14.3)	1	1:101,949	0.02	4.8
Primary Immunodeficiencies		65 (86.2)			0.05	20.0
SCID			1	1:101,949		
Syndromes with T-cell Impairment			7	1:14,564		
Non-preterm Secondary T-cell Lymphopenias			3	1:33,983		
ADA SCID/ADA deficiency			2	1:50,974		
Total			13	1:7,842		
Lysosomal Storage Disorders		17 (58.8)	7	1:14,564	0.01	41.2
X-Linked Adrenoleukodystrophy (X-ALD)		14 (35.7)	1	1:101,949	0.01	7.1
Spinal Muscular Atrophy (SMA)		10 (10.0)	10	1:10,195	0.0	100.0
Amino Acid Disorders		25 (12.0)	11	1:9,268	0.01	44.4
Organic Acid Disorders		19 (15.8)	8	1:12,744	0.01	42.1
Fatty Acid Oxidation Disorders		109 (8.3)	7	1:14,564	0.1	6.4
MS/MS Disorders Total		153 (9.8)	26	1:3,921	0.1	17.0

Notes: Maternal cases and carriers identified following an abnormal newborn screen are not included as confirmed cases in the screening performance indicators (i.e., considered false positives).

PRIMARY IMMUNODEFICIENCIES

In total, 13 cases of primary immunodeficiencies (PID) were identified, resulting in FPR of 0.05% and PPV of 20%. Of the 13 cases, seven had syndromes with T-cell impairment, three had non-preterm secondary T-cell lymphopenias, one had true severe combined immunodeficiency (SCID) and two had ADA SCID/ADA deficiency. Chapter IV of the 2011 Annual Report provides more detailed information about PID screening in Michigan.

MS/MS DISORDERS

In total, 26 disorders were detected by MS/MS. The overall FPR for MS/MS disorders was 0.1%. The PPV was 17%, and the detection rate was 1:3,921.

SCREENING PERFORMANCE METRICS-INDIVIDUAL MS/MS DISORDERS

AMINO ACID DISORDERS

Eleven infants were identified with amino acid disorders (Table 7) by MS/MS. Phenylketonuria (PKU) was the most frequent amino acid disorder identified, found in one of every 20,390 newborns screened (5 newborns total). Chapter IV of the 2009 Annual Report provides more detailed information about PKU screening in Michigan. Two cases of ASA, two case of MSUD, one case of citrullinemia, and one case of TYR I was also identified.

ORGANIC ACID DISORDERS

Eight infants were diagnosed with an organic acid disorder in 2020. Seven infants were diagnosed with Propionic acidemia/Methylmalonic acidemia (PA/MMA), and one was diagnosed with 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC).

FATTY ACID OXIDATION DISORDERS

Seven newborns were identified with organic acid disorders (Table 9) by MS/MS. Three were diagnosed with medium-chain acyl-CoA dehydrogenase deficiency (MCAD), three were diagnosed with Carnitine uptake defect (CUD), and one was diagnosed with Carnitine palmitoyltransferase II deficiency (CPT II).

LYSOSOMAL STORAGE DISORDERS

Seven infants were identified with lysosomal storage disorders (Table 10); all seven had Pompe disease. The overall PPV for LSDs was 41.2% and the false positive rates was 0.01%. For Pompe disease, the PPV was 77.8% and for MPS 1 the PPV was 0%, since no positives confirmed with MPS I. The false positive rate was 0.002% for Pompe and 0.008% for MPS 1.

Table 6: Hemoglobinopathy Screening Performance Metrics, Michigan, 2020

Disorder	Newborns (N)	Confirmed + (N)		Positive Detection Rate	
		Total	Among Black Populations	Total	Among Black Populations
Sickle Cell Anemia	101,949	40	30	1:2,549	1:623
SC Disease		18	17	1:5,664	1:1,100
Sickle β thalassemia		14	11	1:7,282	1:1,700
Sickle beta thalassemia zero		1	1	1:101,949	1:18,704
Total		73	59	*1:1,397*	1:317

Notes: Out of the number of Michigan resident infants screened, total N=105,810, among black populations N=18,704

Table 7: Amino Acid Disorders Detected by Tandem Mass Spectrometry, Screening

Disorder	Newborns N	Total + N	Con-firmed + (N)	Positive Detection Rate	FPR (%)	PPV (%)
Phenylketonuria	101,949	8			0.003	62.5
Medically treated (PKU)			3	1:33,983		
Benign Hyperphenyl-alaninemia (H-PHE)			2	1:50,975		
Total			5	1:20,390		
Citrullinemia (CIT)/CIT II		3	1	1:101,949	0.002	33.3
Tyrosinemia I (TYR I)		2	1	1:101,949	0.001	50.0
Tyrosinemia II/III (TYR II/III)		3	0	-	0.003	0.0
Maple Syrup Urine Disease (MSUD)		7	2	1:50,975	0.005	28.6
Argininosuccinic acidem-ia (ASA)		2	2	1:50,975	0.0	100.0

Table 8: Organic Acid Disorders Detected by Tandem Mass Spectrometry, Screening Performance Metrics, Michigan, 2020

Disorder	Newborns N	Total + N	Confirmed + (N)	Positive Detection Rate	FPR (%)	PPV (%)
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)	101,949	2	1	1:101,949	0.001	50.0
Glutaric Acidemia Type I (GA I)		1	1	1:101,949	0.0	100.0
Propionic Acidemia (PA)/ Methylmalonic Acidemia (MMA)		16	7	1:14,564	0.01	43.8

Notes: Maternal cases and carriers identified following an abnormal newborn screen are not included as confirmed cases in the screening performance indicators (i.e., considered false positives).

Table 9: Fatty Acid Oxidation Disorders Detected by Tandem Mass Spectrometry, Screening Performance Metrics, Michigan, 2020

Disorder	Newborns N	Total + N	Confirmed + (N)	Positive Detection Rate	FPR (%)	PPV (%)
Carnitine Uptake Defect (CUD)	101,949	90	3	1:33,983	0.1	3.3
Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)		4	3	1:33,983	0.001	75.0
Very Long-chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)		1	0	-	0.001	0.0
Carnitine palmitoyltransferase II deficiency (CPT II)		6	1	1:101,949	0.005	16.7
Glutaric acidemia Type II (GA II)		1	0	-	0.001	0.0
Carnitine Palmitoyltransferase I Deficiency (CPT I)		1	0	-	0.001	0.0
Long-chain L-3-hydroxyl acyl-CoA dehydrogenase deficiency (LCHAD)		4	0	-	0.004	0.0

Notes: Maternal cases and carriers identified following an abnormal newborn screen are not included as confirmed cases in the screening performance indicators (i.e., considered false positives).

Table 10: Lysosomal Storage Disorders, Screening Performance Metrics, Michigan, 2020

Disorder	Newborns N	Total + N	Confirmed + (N)	Positive Detection Rate	FPR (%)	PPV (%)
Pompe disease	101,949	9	7	1:14,564	0.002	77.8
Mucopolysaccharidosis Type 1 (MPS 1)		8	0	-	0.008	0.0

Notes: Maternal cases and carriers identified following an abnormal newborn screen are not included as confirmed cases in the screening performance indicators (i.e., considered false positives).

SCREENING PERFORMANCE METRICS AMONG STRONG POSITIVE SCREENS

This section provides screening performance metrics (FPR and PPV) among strong positive screens relative to those among total positive screens (strong and borderline positives). Disorders lacking a borderline positive category are not reported in Table 11 because their performance metrics have been previously reported in Tables 5-10. Disorders not detected in 2020 and detected disorders with no borderline positive screens are also excluded from Table 11, as there would be no change in screening performance overall compared to strong positive screens only.

Performance metrics among strong positive screens are particularly useful clinically in that they report the risk of a strong positive being a true case (PPV) or a false positive (FPR). When evaluating the significance of a strong positive screen, the performance metrics below should be considered. As indicated in Table 10, the FPRs and PPVs among strong positive screens are significantly improved relative to the overall screening performance metrics among all positive screens. Maternal cases and carriers identified through NBS are not included in Table 11.

Table 11: Screening Performance Metrics (FPR and PPV) among Strong Positive Screens compared to All Positive Screens, Michigan, 2020

Disorder Type	Among All +		Among Strong +	
	FPR	PPV	FPR	PPV
	%	%	%	%
Congenital Hypothyroidism (CH)	1.8	6.0	0.2	22.9
Congenital Adrenal Hyperplasia (CAH)	0.1	9.8	0.01	26.1
Phenylketonuria (PKU)	0.003	62.5	0.001	80.0
Galactosemia	0.003	50.0	0.002	60.0
Cystic Fibrosis (CF)	0.3	7.8	0.005	76.0
Primary Immunodeficiency (PID)	0.05	20.0	0.02	31.4
Biotinidase Deficiency	0.03	34.0	0.002	75.0
Carnitine uptake defect (CUD)	0.1	3.3	0.001	75.0

The FPR for CH is reduced approximately nine-fold for strong positive screens, and the PPV is increased approximately four-fold compared to all positives. Although cystic fibrosis does not have a strong positive category, children with compound heterozygote or homozygote DNA mutations were considered “strong positive” in Table 11. The FPR decreased 51-fold and the PPV increased from 8% to 76% when excluding children with a heterozygote DNA mutation. For PID, the PPV increased from 20% to 31% for strong positive screens compared to all positive screens. Overall, strong positive screens are far less likely to be false positives and far more likely to be indicative of true disease compared to positive screens overall (i.e., both strong and borderline).

CARRIERS, MATERNAL DISORDERS AND OTHER DISORDERS DETECTED

Although the overarching goal of NBS is to detect disorders in newborns, carriers and maternal disorders are also identified. For disorders in the NBS panel, carriers have one normal gene and one gene variant and typically do not display any clinical symptoms. On a routine basis, the NBS Follow-up Program refers all newborns with positive screens to the appropriate medical management coordinating center that will follow-up to determine the final diagnosis: no disease, disease, carrier, or maternal disorder. NBS will only detect carriers or maternal disorders following an abnormal screen. Thus, NBS will not identify all carriers or all maternal disorders.

In 2020, a total of 3,025 infants were identified as carriers of a disease included in the NBS panel, following an abnormal screen (Table 12). The majority of these infants (n=2,760) had a hemoglobin trait. There were 259 infants who were identified as cystic fibrosis carriers, two were identified as Pompe carriers, one was identified as a GA1 carrier, one was identified as a CAH carrier, one was identified as a heterozygote X-ALD case (female X-ALD carrier) and one was identified as a LCHAD/TFP carrier.

Table 12: Carriers Identified from Newborn Screening, Michigan, 2020

Disorder	N
Hemoglobin Traits	2,760
Cystic fibrosis (CF)	259
Pompe disease	2
Glutaric Acidemia 1 (GA1)	1
Congenital Adrenal Hyperplasias (CAH)	1
X-Linked Adrenoleukodystrophy (X-ALD)	1
Long-chain L-3-hydroxyl acyl-CoA dehydrogenase deficiency (LCHAD)/ Trifunctional protein deficiency (TFP)	1

Nes: All of these infants were identified following an abnormal screen. Not all carriers will have abnormal screens, so not all carriers will be detected through newborn screening

MATERNAL DISORDERS IDENTIFIED FROM NEWBORN SCREENING

Besides confirmatory diagnostic testing for infants, medical management centers also offer diagnostic testing for mothers. Since mothers may have the disease rather than the infant, they could possibly be identified through NBS for a few disorders. No maternal cases were detected in 2020.

IV. Quality Assurance Information

This section includes quality assurance (QA) information about NBS specimen characteristics and indicators included in the quarterly reports that are distributed to hospitals.

SPECIMEN CHARACTERISTICS

Table 13 reports specimen characteristics by nursery type where the specimen was collected. Although 11% of infants were admitted to the NICU or SCN, 50% and 18% of strong and borderline positive screens were received from infants in the NICU, respectively. Isolated elevations of one or more amino acids and/or acyl-carnitines were also more prevalent among specimens received from infants in the NICU; these elevations are commonly associated with infants receiving total parenteral nutrition or transfusions, or low birth weight or preterm infants. While the overall number of unsatisfactory specimens was greatest among hospital nurseries, the proportion of unsatisfactory specimens was greatest among non-hospital samples (2.8%). Early (collected before 24 hours of life) and transfused specimens were more common among infants from the NICU (7.7%, 0.5%, respectively), while late specimens (collected after six days of life) were most common among non-hospital deliveries (8.7%). The NBS Follow-up Program tracks all strong and borderline positive, isolated elevation, unsatisfactory, early, and transfused specimens; approximately 4,800 specimens required follow-up in 2020.

Table 13: Specimen Characteristics by Nursery Type, Michigan, 2020

Indicator	Type of Birth					
	Regular Nursery		NICU/SCN		Non-Hospital	
	N	%	N	%	N	%
Strong Positive Specimens	198	0.2	202	1.8	8	0.1
Borderline Positive Specimens	1,640	1.8	372	3.3	7	0.6
All Positive Specimens*	2,201	2.5	662	5.9	17	1.3
Isolated elevations of amino acids and acyl-carnitines	7	0.0	482	4.3	1	0.1
Unsatisfactory Specimens	899	1.0	222	1.9	35	2.8
Late (>6 days) Specimens	39	0.0	10	0.1	109	8.7
Early (<1 day) Specimens	253	0.3	854	7.7	18	1.4
Transfused Specimens	0	0.0	55	0.5	1	0.1
Specimens Missing Demographics **	1,242	1.4	119	1.1	44	3.5
Total Births Screened	89,540	87.8	11,138	10.9	1,271	1.3

**Includes all strong and borderline specimens plus specimens positive for cystic fibrosis or hemoglobinopathies.*
***Defined as missing race, specimen collection time, or birth weight.*
Notes: Percentages expressed in the above table are column percentages, except for Total Births Screened which is a row percentage.

PERFORMANCE INDICATORS

During 2020, the hospital quarterly reports included six indicators related to blood spot screening. Table 14 lists the indicators and the performance goal for each indicator.

Table 14: Indicators and Performance Goals for Newborn Screening, Michigan, 2020

Measure	Performance Goal
Late Screens	Less than 2% of screens collected greater than 36 hours after birth.
Appropriate Day	Greater than 90% of screens arrive in state laboratory on or before the appropriate day.
Unsatisfactory Screens	Less than 1% of screens are unsatisfactory.
NBS Card Number	Greater than 95% of electronic birth certificates have the NBS card number recorded.
Returned BioTrust for Health Consent Forms	At least 90% of specimens have a returned BioTrust for Health consent form that is completed appropriately.
NBS card with incorrect demographics	Less than 1% of specimens have errors in the birth date/ time and/or specimen collection date/time on the NBS card.

Table 15 lists the statistics for each performance measure and whether the goal was met, by nursery type. For late screens, regular nurseries and NICU/SCN nurseries met the goal. Of note, almost 50% of non-hospital births had screens collected more than 36 hours after birth. Timely collection of specimens is critical for ensuring prompt screening and referral to medical management. Receipt by appropriate day is a measure based on specimen collection time and each hospital's courier pickup days and times. Any specimen collected more than five hours before the designated pickup time for that day should be sent out the same day and received in the state laboratory the next day. For appropriate day, regular nurseries met the goal with 92.4% of specimens arriving on or before the appropriate day. NICUs had 85.6% of specimens arrive by the appropriate day and thus did not meet the goal of 90%.

None of the groups met the goal for unsatisfactory specimens for 2020, although regular nurseries were very close with 1% of screens unsatisfactory. None of the groups met the goal for recording of the NBS card number on birth certificates. Although none of the nursery types met the BioTrust for Health measure, regular nurseries were very close; 89.8% of regular nursery births had a BioTrust for Health consent form returned that was appropriately completed compared to approximately 65% of NICUs/SCNs and 71% of non-hospital births.

The metric regarding data accuracy aims for less than 1% of specimens to have errors in the birth or collection date/time fields or any other demographic on the card such as birthweight or gestational age. When an error is suspected the NBS lab calls hospitals or the birth attendants to verify the information on the NBS card. Non-hospital births had the highest percentage of errors with 10.7% of cards having errors, while regular baby nurseries had 2.5% of cards with errors and NICUs/SCNs had 3.6% of cards with errors.

Table 15: Measures for Newborn Screening, by Nursery Type, Michigan, 2020

Measure	Nursery Type	N	%	Met Goal?
Late Screens: Less than 2% of screens collected greater than 36 hours after birth.	Regular	521	0.6	Yes
	NICU/SCN	139	1.3	Yes
	Non-hospital	627	49.8	No
Appropriate Day: Greater than 90% of screens arrive in state laboratory on or before the appropriate day.	Regular	83,369	92.4	Yes
	NICU/SCN	9,632	85.6	No
	Non-hospital*	NA		
Unsatisfactory Screens: Less than 1% of screens are unsatisfactory.	Regular	899	1.0	No
	NICU/SCN	222	2.0	No
	Non-hospital	35	2.8	No
NBS Card Number: Greater than 95% of electronic birth certificates have the NBS card number recorded.	Regular	89,342	90.0	No
	NICU/SCN**	-	-	-
	Non-hospital	1249	59.0	No
Returned BioTrust for Health Consent Forms Completed Appropriately: At least 90% of specimens have a returned consent form that is completed appropriately.	Regular	80,382	89.8	No
	NICU/SCN	7,213	64.8	No
	Non-hospital	902	71.0	No
NBS card with incorrect dates/times: Less than 1% of specimen have errors in their birth date/ time and/or collection date/time on the NBS card.	Regular	2,237	2.5	No
	NICU/SCN	405	3.6	No
	Non-hospital	136	10.7	No

*Receipt by appropriate day is not calculated for non-hospital births because they do not have a designated courier pick-up time for each day like birthing facilities have.
**Recording of NBS card number is not a performance measure for NICUs since the birth hospital is asked to draw the NBS specimen before transferring the infant to the NICU. Infants transferred to NICUs (as recorded on the birth certificate) are not included in the performance measure for regular nurseries.

SCREENING TURN-AROUND TIME

Turn-around time in NBS refers to the time from birth to initiation of treatment. The target turn-around time for initiating treatment for the early-onset life-threatening disorders (CAH, galactosemia and disorders detected by MS/MS) is no later than the seventh day of life. The goals for other disorders vary.

TIME TO TREATMENT

Table 16 reports the time to treatment for disorders other than hemoglobinopathies and cystic fibrosis. Penicillin prophylaxis, the treatment for hemoglobinopathies, is initiated later than treatment for other disorders and is reported in a separate table (Table 17). As indicated in Table 16, time to treatment ranged from zero to 184 days among all disorders. Since borderline positive screens require one or more retests before being referred for confirmatory testing, CH is presented separately by initial screening result (strong or borderline) in the table.

GALACTOSEMIA

All three classic galactosemia cases were treated within the first week of life.

BIOTINIDASE DEFICIENCY

One case of profound Biotinidase was identified in 2020 and that infant was treated in the first week of life. The treatment start date ranged from five to 73 days after birth for the 16 partial Biotinidase deficiency cases.

MS/MS DISORDERS

All eight infants identified with an amino acid disorder began treatment in the first week of life.

Of the eight infants identified with an organic acid disorder, two started treatment within the first week of life, the other six cases began treatment in the second week of life.

Of the seven infants identified with an fatty acid disorder, six began treatment within the first week of life and one began treatment in their second week of life.

ENDOCRINE DISORDERS

The salt-wasting form of CAH is life-threatening in the first few weeks of life. There were eight infants diagnosed with salt-wasting CAH, six were treated in the first week of life, one was treated in the second week of life and one was treated in the third week of life.

The target for CH is treatment by 14 days of life for newborns with initial TSH values greater than 50 (i.e., strong positives). Of the 58 CH cases with a strong positive screen, 43 (74.1%) were treated by the 14th day of life.

Table 16: Time to Treatment of Amino Acid Disorders, Organic Acid, Fatty Acid Oxidation, and Endocrine Disorders, Michigan, 2020

Disorder		Total	Treatment Time (days from birth)			Treatment Time Range (days)
			N			
		N	1-7	8-14	>14	
Spinal Muscular Atrophy	SMA	10	0	2	8	8-184
Galactosemia	Classic (GG)	3	3			2-4
Biotinidase Deficiency	Profound	1	1			5
	Partial	16	4	9	4	5-73
Amino Acid Disorders	PKU-medically treated	3	3			3-6
	ASA	2	2			0-2
	MSUD	2	2			2
	TYR 1	1	1			4
	Total	8	8			0-6
Organic Acid Disorders	3MCC	1	0	1		11
	PA/MMA	7	2	5		2-10
	Total	8	2	6		2-11
Fatty Acid Oxidation Disorders	MCAD	3	3			2-4
	CUD	3	2	1		5-9
	CPT II	1	1			4
	Total	7	6	1		2-9
Endocrine Disorders	CH– Strong	58	27	16	15	3-37
	CH– Borderline	63	8	12	43	4-129
	CAH– Salt Wasting	8	6	1	1	3-16
Total		182	65	47	71	0-184

Notes: Disorders that do not require treatment are excluded from the table. Time to treatment categories for CH are based on the results of the first screen. Infants who screened borderline or inconclusive on their first screened required a repeat screen before referral to medical management.

HEMOGLOBINOPATHIES

Table 17 reports the time to treatment among newborns with hemoglobinopathies. The target is to initiate penicillin prophylaxis by four months of life (120 days). Of the 69 cases with a known penicillin initiation date, 94.0% were treated with penicillin within the first four months, 2.9% began treatment between four and five months of life and 2.9% began treatment after five months of life.

Table 17: Time to Penicillin Initiation for Sickle Cell Disorders, Michigan, 2020

Disorder	Penicillin Prophylaxis Initiation Time		
	< 120 days	**120-149 days**	**> 150 days**
Sickle Cell Disorders*	65 (94.0%)	2 (2.9%)	2 (2.9%)

*Four cases were missing a penicillin initiation date.

V. Conclusions

NBS is a critical public health program that protects the lives of our state's newest residents. The NBS Laboratory screened 102,236 infants born in 2020, and the NBS Follow-up Program tracked approximately 4,800 strong and borderline positive, isolated elevation, unsatisfactory, early, and transfused specimens; newborns with strong positive screening results were immediately referred to the appropriate NBS follow-up coordinating center for evaluation. A total of 307 newborns were identified with a disorder by NBS in 2020, as well as 3,025 carriers. Since blood spot screening began in Michigan in 1965, 7,401 newborns have been diagnosed and treated. We are continuing to both expand and refine the NBS Program in order to better protect the health of infants born in Michigan.

MDHHS-Pub-1445 (3-22)