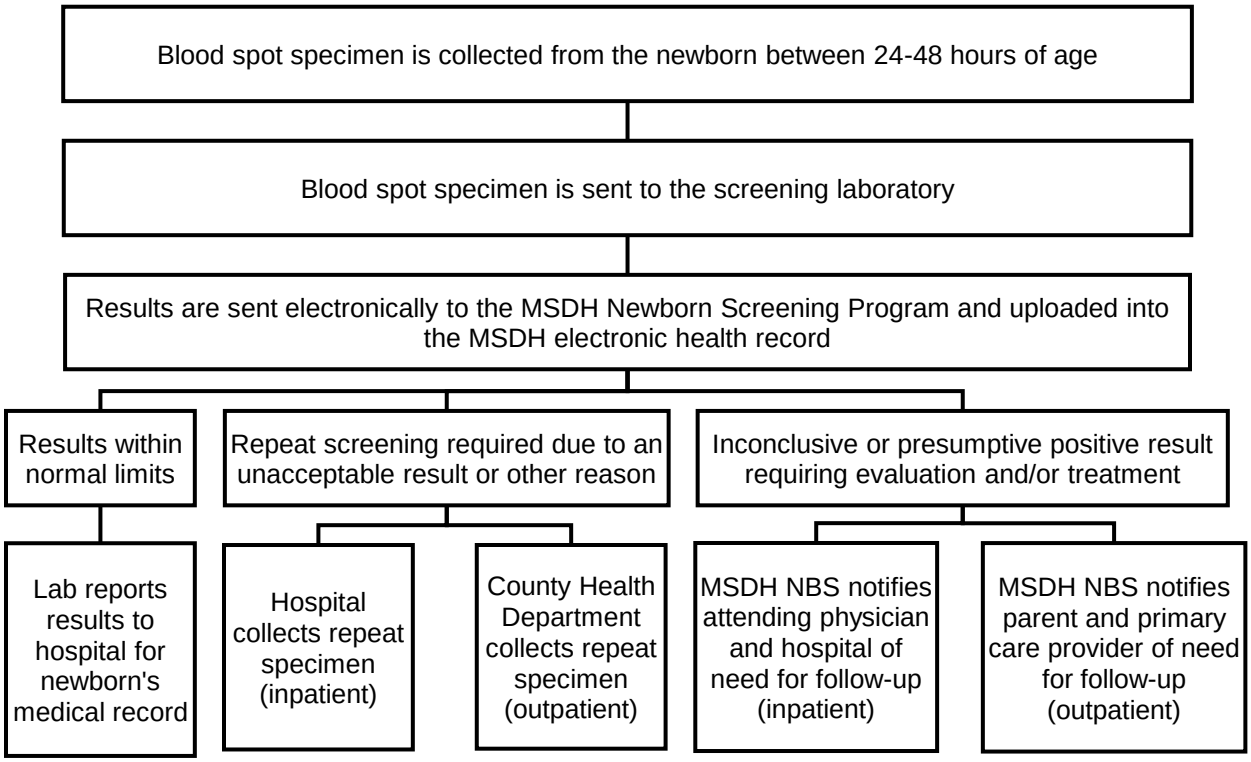
Mississippi Newborn Screening Update 2016-2020

The primary goal of the Mississippi Newborn Screening Program is to ensure every infant born in the state is screened for disorders and infants with abnormal results are referred for medical evaluation, confirmatory testing, and initiation of medical and/or nutritional treatment, if indicated. The Mississippi Newborn Screening Program includes birthing hospitals, the state screening laboratory, tertiary care centers, and public health staff. The program is housed in the MSDH Office of Child and Adolescent Health, Bureau of Genetic Services, and screens for a wide range of disorders including:

- Amino Acid Disorders
- Fatty Acid Oxidation Disorders
- Organic Acid Disorders
- Hemoglobin Disorders
- Lysosomal Storage Disorders
- Endocrine Disorders
- General Genetic Conditions, including Biotinidase Deficiency, Galactose Disorders, Cystic Fibrosis, Mucopolysaccharidosis, and Spinal Muscular Atrophy
- Severe Combined Immunodeficiencies and other T-cell related lymphocyte deficiencies

Newborn Screening Process



Through the collaboration of all stakeholders in the newborn screening process, almost all infants born in Mississippi are screened each year. Newborns may not have been screened due to transferring out of state for a higher level of care shortly after birth, death prior to specimen collection or other exceptions.

Number and Percentage of Newborns Screened, 2016-2020[1]

	2016	2017	2018	2019	2020
Mississippi Occurrence Births	37141	37370	37009	36634	35480
Number of Newborns Screened	36061	36428	35949	35505	34363
Percentage of Newborns Screened	97.1%	97.5%	97.1%	96.9%	96.9%

RUSP CONDITIONS

The Mississippi Newborn Screening Panel aligns with the Recommended Uniform Screening Panel (RUSP) which contains core and secondary conditions as recommended by the Advisory Committee on Heritable Disorders in Newborns and Children and recommended by the Secretary of the U.S. Department of Health and Human Services.

During the five-year period (2016-2020), 99.8% of the newborns screened in Mississippi had normal results. A total of 445 newborns were confirmed with disorders/diseases. Of these, 248 newborns were confirmed as having hemoglobinopathies, representing the largest group of disorders or diseases identified through newborn screening.

Confirmed Disorders/Diseases detected through Newborn Screening 2016-2020[2]

RUSP Core and Secondary Conditions	2016	2017	2018	2019	2020	Total
2,4 Dienoyl-CoA reductase deficiency	0	0	0	0	0	**0**
2-Methyl-3-hydroxybutyric aciduria	0	0	0	0	0	**0**
2-Methylbutyrylglycinuria	0	0	0	0	0	**0**
3-Hydroxy-3-Methyglutaric Aciduria	0	0	0	0	0	**0**
3-methylcrotonyl-Coa Carboxylase Deficiency	1	0	0	0	0	**1**
3-Methylglutaconic aciduria	0	0	0	0	0	**0**
Argininemia	0	1	0	0	0	**1**
Argininosuccinic Aciduria (ASA Deficiency)	0	0	0	0	0	**0**
Biotinidase Deficiency	2	4	0	1	3	**10**

[1] Data obtained from Mississippi State Department of Health (MSDH), Newborn Screening Database and The Office of Vital Records website.

[2] Data obtained from Mississippi State Department of Health (MSDH), Newborn Screening Database. All tabular data represents the number of confirmed conditions in newborns. As newborns may have more than one condition confirmed and may be counted in more than one cell for a given year, the column totals may exceed the total number of newborns with confirmed conditions for that year. Row totals represent the total number of newborns with the respective confirmed condition for the five-year span.

RUSP Core and Secondary Conditions	2016	2017	2018	2019	2020	Total
Carnitine acylcarnitine translocase deficiency	0	0	0	0	0	**0**
Carnitine palmitoyl transferase type I deficiency	0	0	0	0	0	**0**
Carnitine palmitoyl transferase type II deficiency	0	0	0	0	0	**0**
Carnitine Uptake/Transport Defect	0	0	0	0	0	**0**
Citrullinemia type I	0	0	1	1	0	**2**
Citrullinemia type II	0	0	0	0	0	**0**
Classic Galactosemia	7	3	4	1	3	**18**
Congenital Adrenal Hypoplasia (includes non-classical, salt-wasting, and simple virilizing)	1	1	1	0	3	**6**
Cystic Fibrosis	5	13	6	4	4	**32**
Disorders of biopterin biosynthesis	0	0	0	0	0	**0**
Disorders of biopterin regeneration	0	0	0	0	0	**0**
Galactoepimerase deficiency (uridine diphosphate galactose 4-epimerase deficiency)	0	0	0	0	0	**0**
Galactokinase deficiency	0	0	0	0	0	**0**
Glutaric acidemia type II	0	0	0	2	0	**2**
Glutaric Aciduria Type I	0	0	0	2	0	**2**
Glycogen Storage Disease Type II (Pompe)	1	2	3	1	1	**8**
Hemoglobinopathies (HGBD)	56	67	51	42	32	**248**
Holocarboxylase Synthase Deficiency; Multiple carboxylase deficiency	1	0	1	0	0	**2**
Homocystinuria	0	0	0	0	0	**0**
Hypermethioninemia	1	1	1	0	0	**3**
Hyperphenylalaninemia (includes variant and benign)	1	1	0	0	0	**2**
Isobutyrylglycinuria	1	0	0	0	0	**1**
Isovaleric Acidemia	1	0	0	0	0	**1**
Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency	0	0	0	0	0	**0**
Malonic acidemia	0	1	0	0	0	**1**
Maple Syrup Urine Disease	0	0	0	0	0	**0**
Medium/Short-chain L-3-Hydroxy acyl-CoA dehydrogenase	3	1	0	0	0	**4**
Medium-Chain Acyl-CoA Dehydrogenase Deficiency	2	1	1	0	1	**5**
Medium-chain ketoacyl-CoA thiolase deficiency	0	0	0	0	0	**0**
Methylmalonic Acidemia (Cobalamin disorders)	0	1	0	0	0	**1**
Methylmalonic Acidemia (Methylmalonyl-CoA Mutase)	0	1	0	0	0	**1**
Methylmalonic acidemia with homocystinuria	0	0	0	0	0	**0**
Mucopolysaccharidosis Type I[3]	*	*	*	0	0	**0**

[3] Condition not added to the screening panel until November 2019.

RUSP Core and Secondary Conditions	2016	2017	2018	2019	2020	Total
Phenylketonuria	2	2	1	2	0	**7**
Primary Congenital Hypothyroidism	14	11	12	14	13	**64**
Propionic Acidemia	0	0	0	0	0	**0**
Severe Combined Immunodeficiencies (SCID)	1	0	0	0	0	**1**
Short-Chain Acyl-CoA Dehydrogenase Deficiency	0	1	0	0	0	**1**
Spinal Muscular Atrophy (SMA)[4]	*	*	*	1	2	**3**
ß-Ketothiolase Deficiency	0	0	0	0	0	**0**
T-cell related lymphocyte deficiencies	0	0	0	0	0	**0**
Trifunctional Protein Deficiency	0	0	0	0	0	**0**
Tyrosinemia type I	0	0	0	0	1	**1**
Tyrosinemia type II	0	0	0	0	0	**0**
Tyrosinemia type III	0	0	0	0	0	**0**
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	1	0	0	0	0	**1**

HEMOGLOBINOPATHIES

Hemoglobinopathy Disorders (HGBD) are blood disorders or diseases affecting red blood cells. These disorders include both sickle cell disease and thalassemia. Over the five-year period, 248 newborns screened had hemoglobinopathy disorders.

Hemoglobinopathy Disorders (HGBD)
detected through Newborn Screening 2016-2020 [5]

Hemoglobinopathy Disorders	2016	2017	2018	2019	2020	Total
Hb beta zero-thalassemia	1	1	1	0	0	**3**
Hb C beta-thalassemia	8	2	2	0	2	**14**
Hb C-disease	1	6	2	2	4	**15**
Hb D beta-thalassemia	0	0	0	0	0	**0**
Hb E beta-thalassemia	0	0	0	0	0	**0**
Hb E-disease	0	0	0	0	0	**0**
Hb F, and other than A,C,D,E,F,H,O-Arab,S	1	0	0	0	0	**1**
Hb H-disease (BARTs)	0	0	0	0	0	**0**
Hb S O-Arab disease	0	0	0	0	0	**0**
Hb S Other than A,C,D,E,O-Arab	3	2	0	14	1	**20**

[4] Condition not added to the screening panel until November 2019.
[5] Data obtained from Mississippi State Department of Health (MSDH), Newborn Screening Database and The Office of Vital Records website. All tabular data represents the number of confirmed conditions in newborns. As newborns may have more than one condition confirmed and may be counted in more than one cell for a given year, the column totals may exceed the total number of newborns with confirmed conditions for that year. Row totals represent the total number of newborns with the respective confirmed condition for the five-year span.

Hemoglobinopathy Disorders	2016	2017	2018	2019	2020	Total
Hb SD-disease	0	0	0	0	0	**0**
Hb SE-disease	0	0	0	0	0	**0**
S, βeta-Thalassemia	8	9	5	3	1	**26**
S,C Disease	19	15	22	6	10	**72**
S,S Disease (Sickle Cell Anemia)	15	32	19	17	14	**97**

Hemoglobinopathy trait (HGBT), commonly known as sickle cell trait, results when a baby receives a gene for hemoglobin A (normal hemoglobin) from one parent and a gene for a different, abnormal hemoglobin type (S, C, or D) from the other parent. During the five-year period (2016-2020), 8,402 HGBT cases were identified through newborn screening in Mississippi. The incidence of HGBT was 47.1 per 1,000 newborns screened overall.

Hemoglobinopathy Traits (HGBT)
detected through Newborn Screening 2016-2020 [6]

Hemoglobinopathy Traits	2016	2017	2018	2019	2020	Total
Hb C-carrier	398	379	405	390	370	**1942**
Hb D-carrier	12	10	9	15	14	**60**
Hb E-carrier	13	6	7	9	8	**43**
Hb O-Arab carrier	1	0	1	1	2	**5**
Hb S (sickle)-carrier	1218	1165	1229	1213	1202	**6027**
Hb carrier other than C,D,E,S,O-Arab	73	75	57	61	59	**325**

[6] Data obtained from Mississippi State Department of Health (MSDH), Newborn Screening Database and The Office of Vital Records website. All tabular data represents the number of confirmed conditions in newborns. As newborns may have more than one condition confirmed and may be counted in more than one cell for a given year, the column totals may exceed the total number of newborns with confirmed conditions for that year. Row totals represent the total number of newborns with the respective confirmed condition for the five-year span.