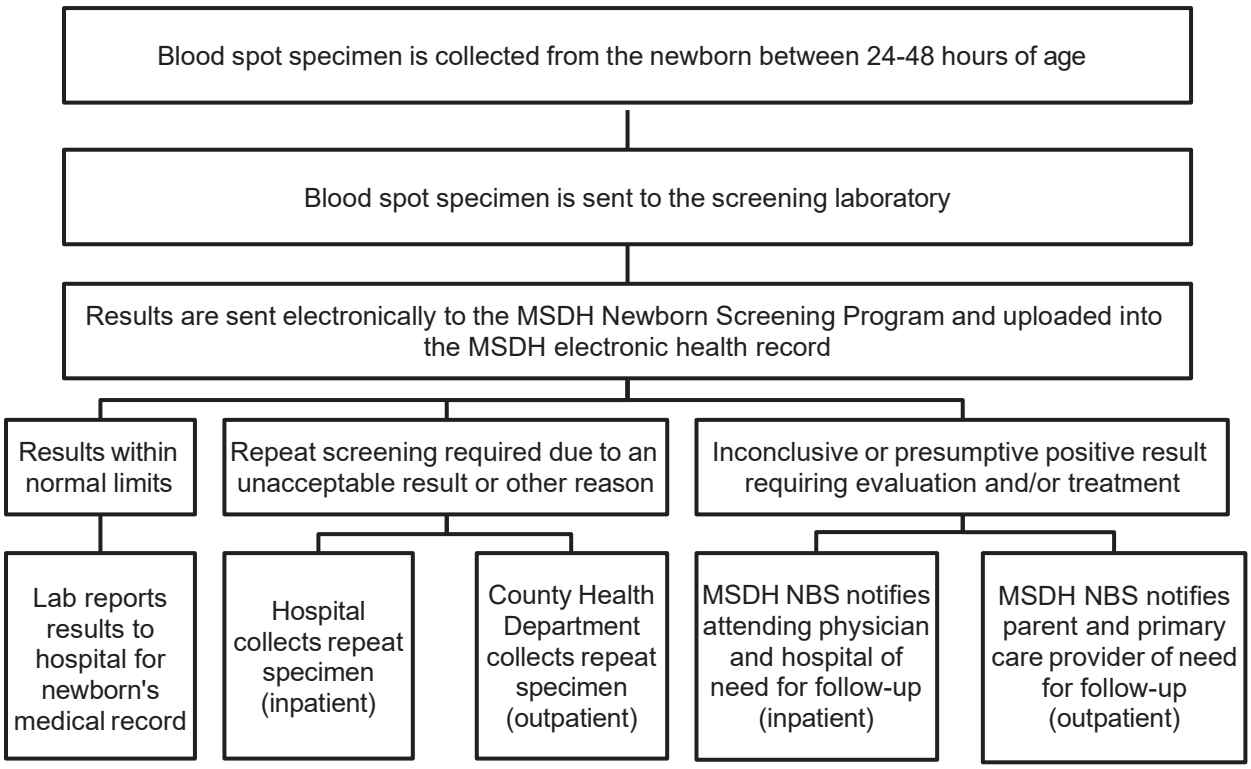
Mississippi Newborn Screening Update 2022-2025

The primary goal of the Mississippi Newborn Screening Program is to ensure that every infant born in the state is screened for disorders and that 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 such as Biotinidase Deficiency, Galactose Disorders, Cystic Fibrosis, Mucopolysaccharidosis, and Spinal Muscular Atrophy
- Severe Combined Immunodeficiencies and other T-cell related lymphocyte deficiencies

Newborn Screening Process



The Summary of Mississippi Newborns Screened and findings during the four year period 2022-2025 through the Mississippi Newborn Screening program are included in following 4 tables: Table 1, Number and Percentage of Newborns Screened through Mississippi Newborn Screening during 2022- 2025; Table 2, Confirmed Genetic Disorders/Diseases detected through Mississippi Newborn Screening 2022- 2025; Table 3, Hemoglobinopathies detected through Mississippi Newborn Screening during 2022- 2025; and Table 4 Hemoglobinopathy Traits identified through Mississippi Newborn Screening during 2022- 2025.

Table 1. Number and Percentage of Newborns Screened through Mississippi Newborn Screening, 2022 -2025[1]

	2022	2023	2024	2025
Mississippi Occurrence Births	33683	33310	32,443	31,524
Number of Newborns Screened	33612	33195	32,194	31,419
Percentage of Newborns Screened	99.8%	99.7%	99.2%	99.6%

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.

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.

Table 2 below contains all genetic core and secondary diseases and disorders in RUSP that were detected through the Mississippi Newborn Screening program during 2022-2025.

[1] Data obtained from Mississippi State Department of Health EPIC and The Office of Vital Records.

Table 2. Confirmed Genetic Disorders/Diseases detected through Mississippi Newborn Screening 2022-2025[2]

Core/second RUSP	Confirmed Disease/disorder	2022	2023	2024	2025	Total
Core	X-linked adrenoleukodystrophy	0	1	1	2	4
Core	3-Hydroxy-3-Methyglutaric Aciduria	0	0	0	0	0
Core	3-methylcrotonyl-Coa Carboxylase Deficiency	0	2	0	0	2
Core	Argininosuccinic Aciduria (ASA Deficiency)	0	1	0	0	1
Core	Biotinidase Deficiency	2	3	2	4	11
Core	Carnitine Uptake/Transport Defect	0	0	0	0	0
Core	Citrullinemia type I	0	0	0	1	1
Core	Classic Galactosemia	4	3	1	1	9
Core	Classic phenylketonuria	0	0	0	0	0
Core	Congenital Adrenal Hypoplasia	4	0	0	0	4
Core	Cystic Fibrosis	0	0	0	0	0
Core	Glutaric acidemia type I	0	0	0	0	0
Core	Glycogen Storage Disease Type II (Pompe)	3	5	4	4	16
Core	Holocarboxylase Synthase Deficiency; Multiple carboxylase deficiency	0	0	0	0	0
Core	Homocystinuria	0	0	0	0	0
Core	Isovaleric Acidemia	1	1	1	1	4
Core	Long-chain L-3 hydroxy acyl-CoA dehydrogenase deficiency	0	0	0	0	0
Core	Maple Syrup Urine Disease	0	0	0	0	0
Core	Methylmalonic Acidemia (Cobalamin disorders)	0	2	0	0	2
Core	Methylmalonic Acidemia (Methyl malonyl-CoA Mutase)	0	0	0	0	0
Core	Mucopolysaccharidosis Type I	0	0	1	0	1
Core	Primary Congenital Hypothyroidism	17	23	17	23	80
Core	Propionic Acidemia	0	0	0	0	0

Core/second RUSP	Confirmed Disease/disorder	2022	2023	2024	2025	Total
Core	Severe Combined Immunodeficiencies (SCID)	**0**	**2**	**0**	1	**4**
Core	Spinal Muscular Atrophy (SMA)	3	3	2	1	9
Core	ß-Ketothiolase Deficiency	0	0	0	0	0
Core	Trifunctional Protein Deficiency	0	0	0	0	0
Core	Tyrosinemia type I	1	1	0	0	2
Core	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	1	0	0	0	1
Core	S, βeta-Thalassemia	2	6	4	2	14
Core	S,C Disease	8	10	15	8	41
Core	S,S Disease (Sickle Cell Anemia)	15	21	29	15	80
Core	Medium-Chain Acyl-CoA Dehydrogenase Deficiency	1	1	1	0	3
Secondary	2,4 Dienoyl-CoA reductase deficiency	0	0	0	0	0
Secondary	2-Methyl-3-hydroxybutyric aciduria	0	0	0	0	0
Secondary	2-Methylbutyrylglycinuria	0	0	0	0	0
Secondary	3-Methylglutaconic aciduria	0	0	0	0	0
Secondary	Argininemia	0	0	0	0	0
Secondary	Carnitine acylcarnitine translocase deficiency	0	0	0	0	0
Secondary	Carnitine palmitoyl transferase type I deficiency	0	0	0	0	0
Secondary	Carnitine palmitoyl transferase type II deficiency	0	0	0	0	0
Secondary	Citrullinemia type II	0	0	0	0	0
Secondary	Biopterin defect in cofactor biosynthesis	0	0	0	0	0
Secondary	Biopterin defect in cofactor regeneration	0	0	0	0	0
Secondary	Galactoepimerase deficiency (uridine diphosphate galactose 4-epimerase deficiency)	0	0	1	0	1
Secondary	Galactokinase deficiency	0	0	0	0	0
Secondary	Isobutyrylglycinuria	0	0	1	0	1
Secondary	Medium-chainketoacyl-CoA thiolase deficiency	0	0	0	0	0
Secondary	Methylmalonic acidemia with homocystinuria	0	0	0	0	0

Core/second RUSP	Confirmed Disease/disorder	2022	2023	2024	2025	Total
Secondary	Short-Chain Acyl-CoA Dehydrogenase Deficiency	1	1	0	0	2
Secondary	Tyrosinemia type II	0	0	0	0	0
Secondary	Tyrosinemia type III	0	0	0	0	0
Secondary	22q11.2 deletion syndrome (T-cell related lymphocyte deficiencies)	1	0	6	4	11
Secondary	DiGeorge anomaly (HCC) (T-cell related lymphocyte deficiencies)	2	1	4	1	8
Secondary	Ataxia-telangiectasia (T-cell related lymphocyte deficiencies)	0	0	0	0	0
Secondary	Idiopathic T-cell lymphopenia (T-cell related lymphocyte deficiencies)	0	0	0	0	0
Secondary	Glutaric acidemia type II	0	0	0	0	0
Secondary	Hypermethioninemia	0	0	0	0	0
Secondary	Medium/Short-chain L-3-Hydroxy acyl-CoA dehydrogenase	0	0	0	0	0
Secondary	Malonic acidemia	0	0	0	0	0
Secondary	Hb C-disease[3]	7	5	3	1	16

[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.

[3] According to federal advisory committee on Heritable Disorders in Newborns and Children recommendation, (Newborn Screening for HbH Disease), not adding Hemoglobin H disease (Barts) to the RUSP, so Hemoglobin H is not included in here.

HEMOGLOBINOPATHIES

Hemoglobinopathies are a group of inherited (genetic) conditions that affect the hemoglobin in blood. Hemoglobinopathies include SS disease, S beta-thalassemia, and SC disease. There are also other abnormal hemoglobin "Various other hemoglobinopathies", which include Hb C beta-thalassemia, Hb C-disease, Hb D beta-thalassemia, Hb E beta-thalassemia, Hb E-disease, Hb H-disease, Hb S O-Arab disease, Hb S Other than A, C, D, E, O-Arab, Hb SD-disease, Hb SE-disease, Hb beta zero-thalassemia, and Hb disease other than A, C, D, E, F, H, O-Arab,S.

Table 3. Hemoglobinopathies detected through Mississippi Newborn Screening 2022-2025[4]

Hemoglobinopathy Disorders	2022	2023	2024	2025	Total
Hb beta zero-thalassemia	0	0	0	0	0
Hb C beta-thalassemia	0	0	0	0	0
Hb C-disease	7	5	3	1	16
Hb D beta-thalassemia	0	0	0	0	0
Hb E beta-thalassemia	0	0	0	0	0
Hb E-disease	0	1	0	0	1
Hb F, and other than A, C,D,E,F,H,O-Arab,S	0	0	0	0	0
Hb H-disease (BARTs)[5]	500	516	377	280	1673
Hb S O-Arab disease	0	0	0	0	0
Hb S Other than A,C,D,E,O-Arab	0	0	0	**0**	0
Hb SD-disease	0	0	0	0	0
Hb SE-disease	0	0	0	0	0
S, βeta-Thalassemia	2	6	4	2	14
S,C Disease	8	10	15	8	41
S,S Disease (Sickle Cell Anemia)	15	21	29	15	80

[4] Data obtained from Mississippi State Department of Health EPIC. Newborns may have more than one genetic condition confirmed.

[5] Hb H-diseases (Barts) are counted here, although they aren't included in RUSP.

Hemoglobinopathy Traits

Hemoglobin traits are a group of inherited (genetic) conditions that affect some of the hemoglobin in the blood. In hemoglobin trait, some of the body's normal hemoglobin is replaced with hemoglobin that is changed slightly, which is also called a variant hemoglobin[6]. commonly known as sickle cell trait. Having a sickle cell trait means a person has inherited the sickle cell gene from only one parent. Table 4 below contains hemoglobinopathy traits that were identified through the Mississippi Newborn Screening program during four-year period (2022- 2025).

Table 4. Hemoglobinopathy Trait and Barts identified through Mississippi Newborn Screening 2022-2025[7]

Hemoglobinopathy Traits	2022	2023	2024	2025	Total
Hb C-carrier	292	300	347	327	1266
Hb D-carrier	17	19	11	17	64
Hb E-carrier	9	11	10	6	36
Hb O-Arab carrier	0	2	0	2	4
Hb S (sickle)-carrier	1078	990	959	851	3878
Hb carrier other than C,D,E,S,O-Arab	0	0	0	1	1

[6] HRSA Hemoglobin Trait | Newborn Screening

[7] Data obtained from Mississippi State Department of Health EPIC. Newborns may have more than one condition Confirmed