Mississippi Newborn Screening Report

2011-2016

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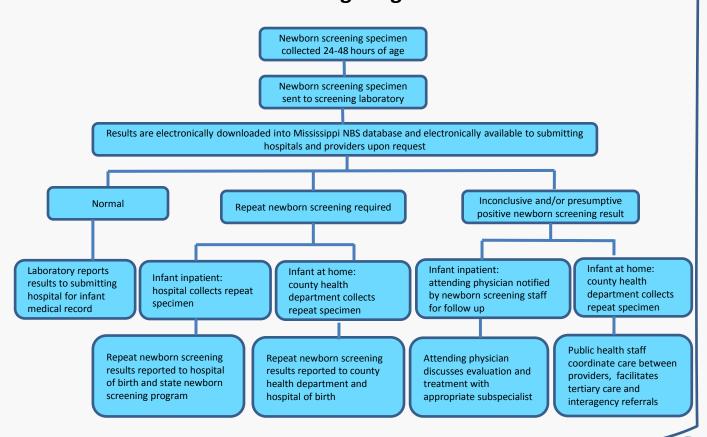
Introduction

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

- Amino Acid Disorders
- · Organic Acid Disorders
- Fatty Acid Oxidation Disorders
- · Biotinidase Deficiency
- Hypothyroidism
- Congenital Adrenal Hypoplasia

- · Cystic Fibrosis
- Galactosemia
- Hemoglobinopathies
- SCID
- Pompe
- Critical Congenital Heart Disease (CCHD)

Newborn Screening Program Flow Chart



Number and Percentage of Newborns Screened, 2011-2016

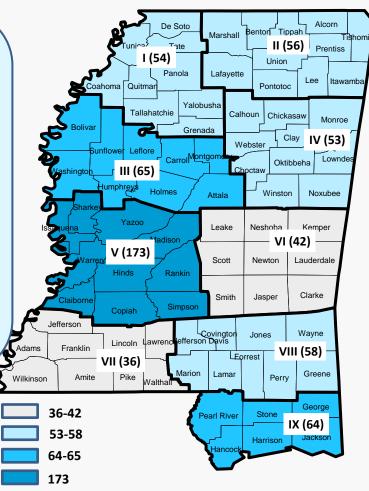
Through the collaboration of all stakeholders in the newborn screening process, almost all infants born in Mississippi are screened each year. Newborns may not have been screened due to transferring out of state for a higher level of care shortly after birth, death prior to specimen collection or other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

	2011	2012	2013	2014	2015	2016
Mississippi Occurrence Births	38,939	37,787	37,648	37,956	37,618	37,141
Number of Newborns Screened	37,979	36,759	36.607	36,961	36,495	36,061
Percentage of Newborns Screened	97.5%	97.3%	97.2%	97.4%	97.0%	97.1%

Newborns with Confirmed Genetic Disorders/Diseases by Public Health District, 2011-2016

The map displays the number of infants with confirmed disorders/diseases over a six-year period (2011-2016) according to their residence within the public health districts.

Between 2011–2016, approximately 601* newborns were identified with genetic disorders/diseases. Districts V and III had the most confirmed number of genetic disorders/diseases. The population concentration in district V compared to other districts may contribute to the increased number of confirmed disorders/diseases. It is also the location of the state's only tertiary center were the majority of children are referred for evaluation and treatment. District III has an overall lower population, however, it has the largest percentage of African Americans, who are at greater risk of Hemoglobinopathy disorders (HGBD), the largest group of disorders in the state.



^{*} The total number (601) of newborns screened with confirmed disorders/diseases does not include non-Mississippi residents

Data Source: Mississippi Newborn Screening Program Database

Confirmed Disorders/Diseases, 2011–2016*

Over the six-year period, 99.7% of the newborns screened had normal results. A total of 613 newborns with disorders/diseases were confirmed. Among them, 387 or 63% were hemoglobinopathy disorders.

Amino Acid Disorders	2011	2012	2013	2014	2015	2016	Total	
Hypermethioninemia	0	0	0	0	0	1	1	
Maple Syrup Urine Disease	0	0	0	0	0	0	0	
Phenylketonuria	2	1	1	2	4	2	12	
Tyrosinemia	0	0	0	0	0	0	0	
Isobutryrl-CoA dehydrogenase deficiency	0	0	0	0	0	1	1	
Total	2	1	1	2	4	4	14	
Organic Acid Disorders	2011	2012	2013	2014	2015	2016	Total	
3-methylcrotonyl-Coa Carboxylase Deficiency	0	0	1	1	0	1	3	
Glutaric Aciduria Type I	0	0	1	0	0	0	1	
Isovaleric Acidemia	1	0	0	0	0	1	2	
Propionic Acidemia	0	0	0	0	1	0	1	
Total	1	0	2	1	1	2	7	
	•		1	1	•			
Fatty Acid Oxidation Disorders	2011	2012	2013	2014	2015	2016	Total	
Carnitine Palmitoryltransferase II	1	0	0	0	0	0	1	
Long-chain L-3 hydroxyacyl-CoA dehyrogenase deficiency	0	1	0	0	0	0	1	
Medium-Chain Acyl-CoA Dehydrogenase Deficiency	2	1	2	0	0	2	7	
Short-Chain Acyl-CoA Dehydrogenase Deficiency	1	0	3	0	1	0	5	
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	0	0	0	1	1	1	3	
Multipe Acyl-CoA Dehydrogenase Deficiency	0	0	0	0	0	1	1	
Total	4	2	5	1	2	4	18	
General Disorders	2011	2012	2013	2014	2015	2016	Total	
Biotinidase Deficiency	3	1	5	1	5	2	17	
Hypothyroidism	23	12	15	12	9	14	85	
Congenital Adrenal Hypoplasia (Included CAH-SW)	2	5	3	3	3	1	17	
Cystic Fibrosis	7	9	6	8	4	5	39	
Galactosemia	1 74	3	1	3	2	7	17	
Hemoglobinopathies (HGBD)	71	60	65	74	61	56	387	
Pompe Total	0 107	0 90	9 5	0 101	0 84	1 86	5 63	
Total	107	30	95	101	04	00	303	
Severe Combined Immunodeficiency (SCID) / Others	2011	2012	2013	2014	2015	2016	Total	
SCID	0	0	1	2	0	1	4	
Congenital Hypoparathyroidism & DiGeorge Syndrome	0	0	1	0	0	0	1	
DiGeorge Syndrome	0	0	1	3	0	0	4	
	0	1	0	0	0	0	1	
DiGeorge Velocardiofocial Syndrome								
Partial Di-George Syndrome	0 0	0 1	0 3	1 6	0	0 1	1 11	
Total	U	1	3	0	U	1	11	
Grand Total	114	94	106	111	91	97	613**	
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[•] This does not include hemoglobinopathy traits

^{**} The total number (613) of newborns screened with confirmed disorders/diseases includes non-Mississippi residents

Hemoglobinopathy Disorders (HGBD) Identified through Newborn Screening 2011-2016

HGBD is a group of blood disorders or diseases that affect red blood cells. These disorders include both sickle cell disease and thalassemia.

During the six-year period (2011-2016) in Mississippi, 387 HGBD cases were confirmed, which represented the largest group of genetic disorders or diseases identified through newborn screening. Among them, 99% were African Americans and 1% were whites. The incidence of HGBD for African American newborns screened in Mississippi was 3.9 per 1,000.

Disorders	2011	2012	2013	2014	2015	2016	Total
Hemoglobin Sickle Cell Anemia	36	30	37	34	29	15	181
Hemoglobin FS + Barts	1	4	2	6	1	2	16
Hemoglobin Sickle "C " Disease	0	1	0	0	1	1	3
Hemoglobin S/ Beta + Thalassemia	6	1	3	5	2	6	23
Hemoglobin S/Beta o Thalessemia	1	2	0	0	0	0	3
Hemoglobin Sickle C Disease (FSC)	19	17	18	22	17	18	111
Hemoglobin Sickle "C" Disease+Barts	0	0	0	0	1	0	1
Hemoglobin C/ Beta + Thalassemia	0	0	1	1	2	4	8
Hemoglobin C/Beta o Thalassemia	0	0	0	0	1	0	1
Hemoglobin B/Thal	0	0	0	0	1	1	2
Hemoglobin ASF + Other Hemoglobin	1	0	0	0	0	0	1
Hemoglobin Beta Thalassemia Intermedia	0	0	0	0	0	0	0
Hemoglobin C Disease	3	4	2	3	1	1	14
Hemoglobin E Disease + Barts	0	0	0	0	0	0	0
Hemoglobin F only/disease	0	1	0	0	0	1	2
Hemoglobin FCA	1	0	0	0	0	4	5
Hemoglobin FSA	0	0	1	3	3	2	9
Hemoglobin SC Disease + Barts	3	0	0	0	1	1	5
Hemoglobin S/HPFH	0	0	0	0	0	0	0
Hemoglobin SD Disease	0	0	1	0	0	0	1
Hemoglobin SE Disease	0	0	0	0	1	0	1
Total	71	60	65	74	61	56	387

Hemoglobinopathy Traits (HGBT) detected through Newborn Screening 2011-2016

HGBT is commonly known as sickle cell trait, hemoglobinopathy trait results when a baby receives a gene for hemoglobin A from one parent and a gene for a different hemoglobin type (S, C, or D) from the other parent.

During the six-year period (2011-2016) in Mississippi, 10,863 HGBT cases were found. The incidence of HGBT was 48 per 1,000 newborns screened overall. Out of the overall incidence rate of HGBT, 101 per 1,000 screened were African American newborns and 6.3 per 1,000 screened white newborns.

Trait	2011	2012	2013	2014	2015	2016	Total
Hemoglobin S Trait	1,358	1,324	1,237	1,246	1,293	1218	7,676
Hemoglobin FAS Trait+BART	87	79	80	61	75	56	438
Hemoglobin FAS + Fast	0	0	1	0	0	0	1
Hemoglobin FAVBAR	0	1	1	0	1	1	4
Hemoglobin AS Trait+ Variant	0	0	0	0	0	0	0
Hemoglobin C Trait	432	392	366	379	405	381	2,355
Hemoglobin FAC Trait+BART	23	21	19	23	20	17	123
Hemoglobin D Trait	7	16	20	19	11	12	85
Hemoglobin D Los Angeles Trait	6	2	0	0	0	0	8
Hemoglobin D or G Trait	0	0	0	0	0	0	0
Hemoglobin AD or AG Trait	7	10	7	4	4	8	40
Hemoglobin AD or AG w/some F	2	0	0	0	0	0	2
Hemoglobin ADF or Hb AGF Tra	11	6	5	1	5	3	31
Hemoglobin AG or AD T+ Fast	0	0	0	0	0	0	0
A + Fast Hemoglobin	0	1	0	0	0	0	1
AF + Fast Hemo + Pos	0	1	0	0	0	0	1
AF Hemoglobin + Fast	0	1	3	5	4	3	16
AF Hgb/Poss other va	0	1	0	0	0	1	2
Hemoglobin AFD or AFG Trait	1	2	0	0	0	1	4
Hemoglobin ASF	1	2	0	2	2	0	7
Hemoglobin E Trait	7	14	6	12	5	11	55
Hemoglobin AE with some F	0	1	0	0	0	0	1
Hemoglobin FAE Trait+BART	0	0	0	3	0	2	5
Hemoglobin ACF	0	0	1	0	0	0	1
Hemoglobin O Ar Trait	0	0	3	1	2	1	7
Grand Total	1,942	1,874	1,749	1,756	1,827	1715	10,863

