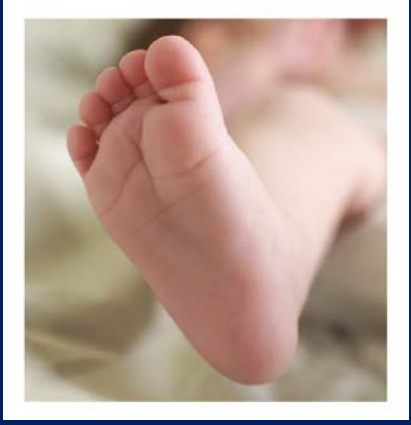
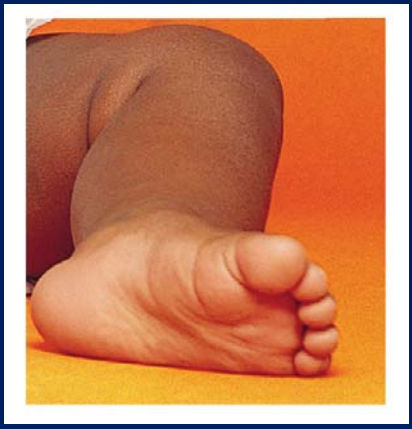
MISSISSIPPI STATE DEPARTMENT OF HEALTH


Newborn Screening Report 2003-2008

AUTHORS AND CONTRIBUTORS

Beryl W. Polk, PhD, MS, CPM
Director, Genetic Services Program

Philis Hoggatt, RN
Nurse Coordinator, Newborn Screening Program

Lynn Walker, MD
Pediatric Clinician

Patricia Terry, MSM, LSW
Program Manager

Geneva Cannon, MHS, RN
Director, Office of Child and Adolescent Health

Connie L. Bish, PhD, MPH
State MCH Epidemiologist

Ninglong Han, MS
Biostatistician, Office of Health Data and Research

Miranda Richardson, BS
Health Program Specialist, Senior

LaQuita S. Cooper, MPH, MHSA
Division Director

ACKNOWLEDGMENTS

The Newborn Screening Program wishes to thank the Mississippi Genetics Advisory Committee (GAC) for its vital role in supporting the activities of the Mississippi State Department of Health (MSDH) Newborn Screening Program and the MSDH staff for coordinating care of newborns identified with genetic disorders/diseases.

Through the committed work and dedication of health department staff and the continued involvement of the Genetics Advisory Committee, we will work together to maintain Newborn Screening as a successful public health service for our great state.

Mississippi State Department of Health, Office of Child and Adolescent Health
Bureau of Genetic Services

TABLE OF CONTENTS

EXECUTIVE SUMMARY

Newborn screening is a well-established national public health program credited with saving lives. This national program has a long history dating back to the 1960's and has made advances in both science and technology. In 1979 newborn screening in Mississippi began with two disorders and by 2003 included 40 disorders (Appendix A). Newborn screening continues to evolve with further additions anticipated in the future.

Experienced staff and detailed systems are in place to manage the daily activities of the program. MSDH Newborn Screening Program is highly organized and efficient and ensures that all infants born in Mississippi receive a complete newborn screen in a timely manner with repeat testing and follow-up as indicated.

INTRODUCTION

This Newborn Screening Report provides an overview of screening performance, diagnosis confirmation, quality assurance, and includes four appendices. Appendix A is the list of disorders included in the Mississippi Newborn Screening Panel. Appendix B describes disorders/diseases, consequences, and treatment options. Appendix C lists the Mississippi Genetics Advisory Committee members. Appendix D is an example of the dried blood spot card.

WHAT IS NEWBORN SCREENING?

Newborn screening is a public health service performed before a baby is discharged from the delivering facility to identify serious or life threatening conditions prior to symptoms. This screening provides early detection of numerous diseases and disorders so that timely treatment can be initiated and long term sequelae minimized. Long term sequelae, depending on the condition, may include organ damage, stroke, or death if left undiagnosed and untreated.

HISTORY OF NEWBORN SCREENING

In the 1960's, Dr. Robert Guthrie of Buffalo, New York developed a bacterial inhibition assay to diagnose phenylketonuria (PKU). The level of the amino acid phenylalanine in a drop of dried blood collected on filter paper was measured using a bacterial inhibition assay. Newborns could then be identified with PKU, and a diet low in phenylalanine could be started early to hopefully avoid mental retardation, behavioral problems, and other abnormalities.

Newborn screening for PKU and hypothyroidism began in Mississippi in 1979 when the State Board of Health was authorized by the Mississippi Legislature to establish a testing program for these diseases. Thus, newborn screening on a voluntary basis began. In 1984 this testing became mandatory by Mississippi law for all newborns. Further legislation in 1988 authorized the addition of two more disorders, hemoglobinopathies and galactosemia, and in 2001 congenital adrenal hyperplasia was added. In 2002, the Ben Haygood Comprehensive Newborn Screening Program was enacted, taking advantage of tandem mass spectrometry, molecular technologies, and biochemical analysis to increase the number of disease/disorders screened to 40. After education of health care providers and staff in delivering facilities, implementation of this expanded screen began in June 2003.

NEWBORN SCREENING

The primary goal of the Newborn Screening Program is to screen every baby born in the state and refer infants with abnormal results to appropriate centers for evaluation, confirmatory testing, and initiation of medical and/or nutritional management as needed. The newborn screening system includes birthing hospitals, a contract screening laboratory, public health staff, and tertiary care centers. The program is located in the MSDH Health Services' Office of Child and Adolescent Health in the Bureau of Genetic Services. The interactions of the components of this system are further explained below and in the flow chart that follows.

A. BIRTHING HOSPITALS

Mississippi currently has 44 hospitals that provide obstetric and newborn care. Hospital staff facilitate the screening process by completing the dried blood spot card and collecting the specimen properly (see Appendix D). An overnight courier service delivers the specimens to the screening laboratory.

B. LABORATORY

PerkinElmer Genetics in Bridgeville, Pennsylvania performs the newborn screen testing. This laboratory is accredited by Clinical Laboratory Improvement Amendments (CLIA) and is directed by Joseph M. Quashnock, Laboratory Director, PhD, FACB. The laboratory establishes reference ranges for each disorder to maximize detection rates while minimizing the rate of false positives and false negatives.

The screening laboratory generates hospital quality assurance reports which can be accessed through the laboratory's website. The report compares specimen quality for each hospital to statewide specimen quality. Quality indicators include, but are not limited to, adequate specimen collection, complete identifying information, and timely specimen transport to the screening laboratory.

C. FOLLOW-UP/CARE COORDINATION

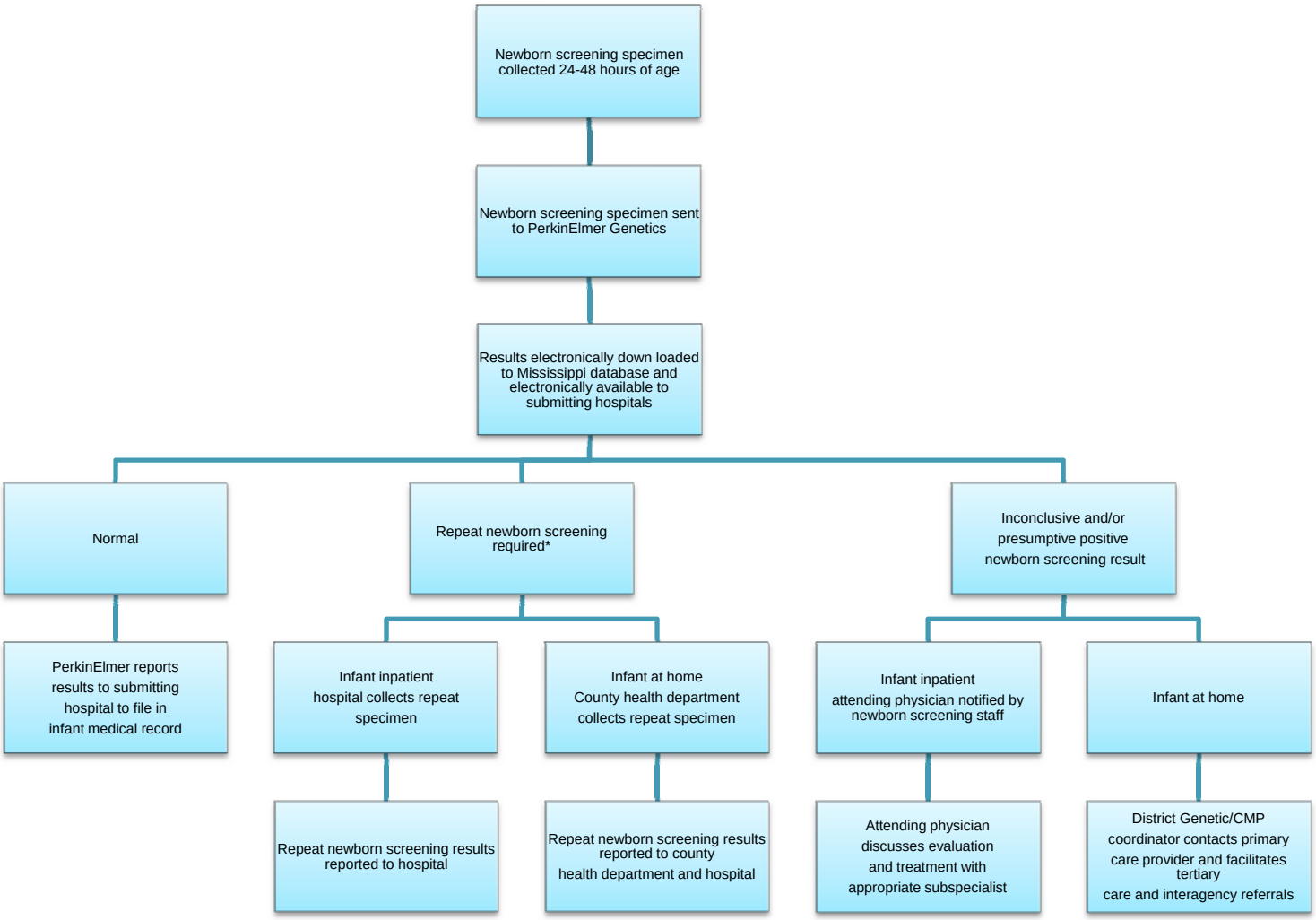
The newborn screening staff notify an infant's primary care provider of an abnormal screening result and assist as needed with referral to a tertiary care center. This follow-up and care coordination begins with all abnormal screening results. After diagnosis, care coordination continues with the tertiary care center and primary care provider so that each child has access to appropriate resources.

In addition, MSDH Genetic/Children Medical Program (CMP) coordinators in each public health district review the hospitals' quality assurance reports, visit hospitals to evaluate the screening process, and make recommendations for improvements as indicated. Ongoing updates and training are provided to hospitals.

D. TERTIARY CARE CENTERS

Primary care providers or newborn screening staff refer infants and their families to appropriate tertiary care centers for evaluation, diagnosis, and treatment. These specialty centers include, but are not limited to, the state's only tertiary care center at the University of Mississippi Medical Center (UMMC) in Jackson. UMMC pediatric specialty care providers utilized for newborn screening follow-up and consultation include endocrinologists, hematologists, pulmonologists, and metabolic disease specialists. Additionally, satellite genetic and sickle cell clinics are located throughout the state.

MISSISSIPPI NEWBORN SCREENING FLOW CHART



The flow chart above explains the path of a screening specimen from collection to results. This process includes birthing hospitals, the newborn screening laboratory, newborn screening staff, primary care providers, and tertiary care centers.

* Reasons for a repeat newborn screening include, but are not limited to, specimen quantity insufficient for testing, specimen appears diluted or contaminated, and specimen exhibits serum rings.

MISSISSIPPI BIRTHS AND SCREEN NUMBERS

Through the collaboration of all stakeholders in the newborn screening process, a majority of Mississippi births are screened each year. Table 1 shows the number of births in Mississippi compared to the number of newborns screened from June 1, 2003, when the Ben Haygood Comprehensive Newborn Screening Program began, through December 31, 2008. MSDH Vital Statistics provides Mississippi birth numbers, while screening numbers are compiled from the newborn screening laboratory and MSDH newborn screening database. A screen may not have been performed because of the following reasons: infant transferred out of state for a higher level of care shortly after birth, infant death prior to specimen collection, or religious exemption. The total number of screens is greater than the number of births because repeat screens may be necessary due to inconclusive results or improper specimen collection or handling. Also all newborns delivered and tested out of state, but residing in Mississippi with unacceptable, inconclusive, or presumptive positive screening results are retested through the Mississippi Newborn Screening Program.

Table 1: Mississippi Births and Screens

	(6/1/2003-12/31/2003)	2004	2005	2006	2007	2008
Births in Mississippi	25,293	41,562	41,180	44,863	45,509	44,136
Number of Newborns Screened	24,966	41,480	40,726	44,444	44,738	43,573
Total Number of Screens	26,063	42,603	43,323	47,010	47,062	45,684
Percent of Newborns Screened	98.7%	99.8%	98.9%	99.0%	98.3%	98.7%

NEWBORN SCREENING DATA

Over the five and a half year period since expanded newborn screening began, a majority (99.7%) of all newborn screens have been normal. However, a total of 718 disorders/diseases have been confirmed. Table 2 on the following page delineates these confirmed disorders/diseases. Confirmation occurs after additional studies have been performed by tertiary care centers.

Table 2: Confirmed Disorders/Diseases 2003-2008

Amino Acid Disorders	2003 [1]	2004	2005	2006	2007	2008	Total
Carbamoylphosphate Synthetase Deficiency				1			1
Citrullinemia (ASA Synthetase Deficiency)					1		1
Homocystinuria			1				1
Hypermethioninemia				1			1
Phenylketonuria (PKU)		2	1	2	4		9[2]
Tyrosinemia			1	1			2
Total		**2**	**3**	**5**	**5**		**15**

Organic Acid Disorders	2003 [1]	2004	2005	2006	2007	2008	Total
2-Methylbutyryl-CoA Dehydrogenase Deficiency			1				1
3-Methylcrotonyl-CoA Carboxylase Deficiency		1					1
3-Methylglutyryl-CoA Hydratase Deficiency			1				1
Glutaric Aciduria Type				2			2
Isobutyryl-CoA Dehydrogenase Deficiency			1				1
Isovaleric Acidemia		1					1
Methylmalonic Acidemia				1			1
Propionic Acidemia			1	1			2
Total		**2**	**4**	**4**			**10**

Fatty Acid Oxidation Disorders	2003 [1]	2004	2005	2006	2007	2008	Total
Medium-Chain Acyl-CoA Dehydrogenase Deficiency		2	2	7	3	2	16
Multipe Acyl-CoA Dehydrogenase Deficiency			1			1	2
Short-Chain Acyl-CoA Dehydrogenase Deficiency		1	1	2	4	3	11
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency			1			2	3
Total		**3**	**5**	**9**	**7**	**8**	**32**

General Disorders	2003 [1]	2004	2005	2006	2007	2008	Total
Biotinidase Deficiency	1	8	4	2	6	7	28[3]
Congenital Hypothyroidism	13	28	18	16	15	16	106
Congenital Adrenal Hyperplasia			2	3	2		7
Cystic Fibrosis	1	12	5	5	12	11	46
Galactosemia	7	13	5	5	8	11	49[4]
Hemoglobinopathies	48	71	62	87	79	78	425
Total	**70**	**132**	**96**	**118**	**122**	**123**	**661**
GRAND TOTAL	**70**	**139**	**108**	**136**	**134**	**131**	**718**

[1] June 1, 2003 through December 31, 2003
[2] 5 Classic PKU
[3] 14 Complete Biotinidase Deficiency
[4] 8 Classic Galactosemia

Hemoglobinopathies represent the largest group of disorders/diseases identified through newborn screening. Table 3 provides further details of abnormal hemoglobin types.

Table 3: Hemoglobinopathy Disorders identified through Newborn Screening, 2003-2008

Disorders	2003 [1]	2004	2005	2006	2007	2008	Grand Total
Hemoglobin Sickle Cell Anemia	26	42	37	36	31	27	199
Hemoglobin FS+Barts				1	2		3
Hemoglobin Sickle C Disease	12	16	13	34	34	21	130
Hemoglobin S/Beta + Thalassemia	1	5	4	6	4	14	34
Hemoglobin S/ Beta O Thalassemia		2	1	5	1	3	12
Hemoglobin C Disease	9	2	3	2	4	4	24
Hemoglobin C/Beta + Thalassemia		3	4	2	3	7	19
Hemoglobin C/Beta O Thalassemia						1	1
Hemoglobin B/Thal						1	1
Hemoglobin D-LA+C		1					1
Hemoglobin E Disease				1			1
Grand Total	**48**	**71**	**62**	**87**	**79**	**78**	**425**

[1] June 1, 2003 through December 31, 2003

Hemoglobin trait is an important finding in newborn screening. Table 4 shows the breakdown of hemoglobin traits identified during the five and a half year period.

Table 4: Hemoglobin Traits detected through Newborn Screening, 2003-2008

Trait	2003 [1]	2004	2005	2006	2007	2008	Grand Total
Hemoglobin S Trait (Sickle Cell Trait)	858	1424	1538	1688	1637	1584	8729
Hemoglobin FAS+Barts			1	39	54	72	166
Hemoglobin FAS+possible alpha variant					1		1
Hemoglobin AS + Variant					1	2	3
Hemoglobin C Trait	315	503	464	548	497	483	2810
Hemoglobin FAC + Barts				13	11	20	44
Hemoglobin D Trait	6	18	15	15	24	18	96
Hemoglobin D Los Angeles Trait		1	1	1		2	5
Hemoglobin AD or AG Trait					1	1	2
Hemoglobin AFD or AFG Trait						1	1
Hemoglobin G Philadelpia Trait	5	11	6	7	5	1	35
Hemoglobin E Trait	3	2	5	5	11	6	32
Hemoglobin FAE+Barts	1	2			1	1	5
Hemoglobin O Arab Trait		3	3	1	3	1	11
Hemoglobin Beta Thal Trait						1	1
Hemoglobin J-Baltimore Trait		1					1
Hemoglobin Variant [2]					2	8	10
Grand Total	**1188**	**1965**	**2033**	**2317**	**2248**	**2201**	**11952**

[1] June 1, 2003 through December 31, 2003

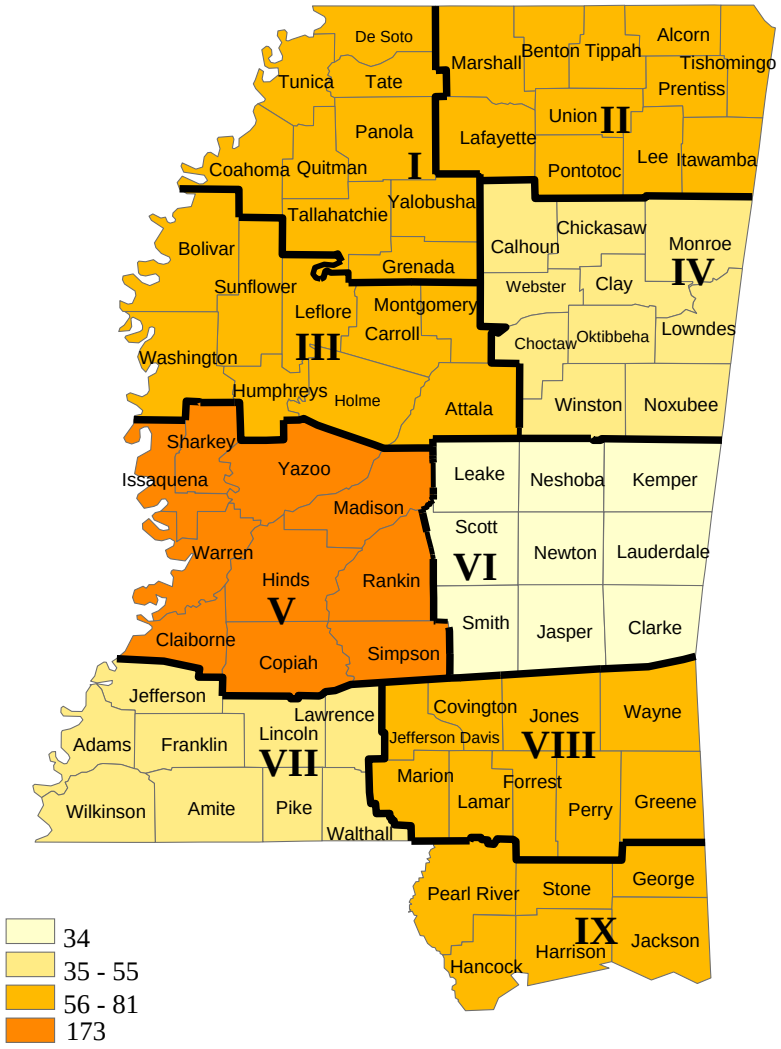
[2] Further testing for hemoglobin identification as clinically indicated by health care provider

SCREENING DATA BY PUBLIC HEALTH DISTRICT

The Mississippi map below shows the state broken into the nine public health districts. The number of confirmed disorders/diseases in relationship to the newborn's residence is noted by a graded color scheme. Public Health District V includes Hinds County, which includes the state's capital city, Jackson, and the tertiary care center at UMMC. This district has the highest population concentration for this largely rural state, which likely accounts for the highest number of confirmed disorders/diseases.

Confirmed Disorders/Diseases by Public Health District

Mississippi, 2003-2008



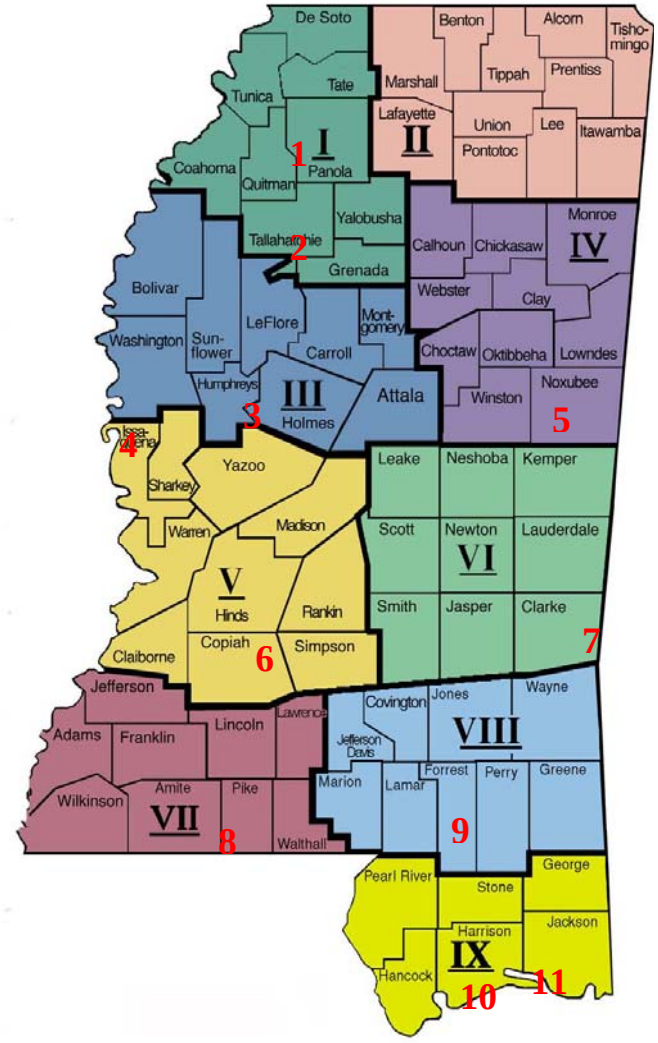
GENETIC AND SICKLE CELL CLINICS
BY PUBLIC HEALTH DISTRICT

Because the disorders/diseases identified through newborn screening are found in every public health district in the state, Genetic and Sickle Cell Satellite Clinics have been established to address the special health care needs of these patients and improve access to care. A compounding factor for our largely rural state is that UMMC is the state's only tertiary care center located in Jackson, Hinds County. Access to health care, especially medical specialty care, directly impacts the health outcomes of newborns and children with special health care needs such as those diagnosed through newborn screening. The map below shows the location of these clinics.

The District Genetic/CMP staff assist specialty providers with these satellite clinics by providing nursing, social work and nutritional support, and help patients and families connect with local community resources.

Satellite Clinics
By Public Health Districts

1. Senatobia	▲
2. Batesville	★
3. Greenwood	▲ ★
4. Greenville	★
5. West Point	▲ ★
6. Jackson	▲ ◆
7. Meridian	▲
8. McComb	▲ ★
9. Hattiesburg	▲ ★
10. Gulfport	▲ ★
11. Ocean Springs	▲
Genetic Clinic	▲
Sickle Cell Clinic	★
UMMC	◆



NEWBORN SCREENING PROGRAM

Economic Impact of Newborn Screening in Mississippi

Newborn screening saves lives and improves health outcomes. Specialty care for the management of the disorders identified through newborn screening is required to improve health outcomes and possibly reduce long-term healthcare costs. Early identification of newborn screening disorders results in earlier treatment, often before symptoms occur to prevent the consequences of delayed identification.

The Newborn Screening Program is supported by a $100.00 fee per screen, which is paid by the birthing hospitals. These funds are used to pay for laboratory testing, program administrative costs, and follow-up efforts.

Regional Activities

Mississippi is an active participant in the Southeastern Regional Genetics Group (SERGG). SERGG is a network of genetics and newborn screening providers in Alabama, Florida, Georgia, Louisiana, Mississippi, North Carolina, South Carolina, Tennessee, Puerto Rico, and the U.S. Virgin Islands. The network is funded in part by a grant from the Federal Health Resources and Service Administration (HRSA) Maternal and Child Health Bureau (MCHB).

The goals of SERGG are to address the inequities in genetic services and resources in the region and to expand existing regional capabilities and resources to address these gaps. SERGG facilitates information sharing among providers of genetic services and consumers and establishes collaborative partnerships with other professional organizations.

Future Plans

Newborn Screening is an impressive public health achievement and is an essential component of Mississippi child health services. The future depends on our responses to new advances in genetic science and changes in health care delivery and financing. The Mississippi Genetics Advisory Committee meets annually to stay informed of current national trends in newborn screening to facilitate timely recommendations for our state program. The Mississippi Newborn Screening Program will implement a hemoglobinopathy surveillance registry to track health outcomes of patients identified with certain hemoglobinopathies. The Newborn Screening Program also plans to conduct a survey of providers and parents on their knowledge of newborn screening. Information from this survey will be used for training and other programmatic activities.

For questions regarding this report contact the MSDH Newborn Screening Program at 601-576-7619.

APPENDIX A
MISSISSIPPI GENETIC NEWBORN SCREENING PANEL

Effective June 1, 2003

Disorders detected by tandem mass spectrometry (MS/MS)

Amino Acid Disorders

Argininemia (ARG)
Argininosuccinic Aciduria (ASA Lyase Deficiency)
Carbamoylphosphate Synthetase Deficiency (CPS Deficiency)
Citrullinemia (ASA Synthetase Deficiency)
Homocystinuria (HCys)
Hyperammoninemia, Hyperornithinemia, Homocitrullinemia Syndrome (HHH)
Hypermethioninemia (MGT)
DNA - Maple Syrup Urine Disease (MSUD)
5-Oxoprolinuria (Pyroglutamic aciduria)
Phenylketonuria (PKU)
Tyrosinemia Type I (TYR I)
Tyrosinemia Type II (TYR II)

Organic Acid Disorders

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG Co-A Lyase Def)
DNA - Glutaric Aciduria Type I (GA I)
Isobutyryl-CoA Dehydrogenase Deficiency
DNA - Isovaleric Acidemia (IVA)
Malonic Aciduria
2-Methylbutyryl-CoA Dehydrogenase Deficiency (2MBCD Deficiency)
DNA - 3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)
3-Methylglutaconyl-CoA Hydratase Deficiency (3MGA Deficiency)
DNA - Methylmalonic Acidemia (MMA)
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
Multiple CoA Carboxylase Deficiency (MCCD)
DNA - Propionic Acidemia (PPA)

Fatty Acid Oxidation Disorders

Carnitine/Acylcarnitine Translocase Deficiency (Translocase)
Carnitine Palmitoyltransferase I Deficiency (CPT I)
Carnitine Palmitoyltransferase II Deficiency (CPT II)
DNA - Long-Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
DNA - Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or GA II)
Carnitine Palmitoyltransferase II Deficiency (CPT II)
Short-Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
Short-Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD)
Trifunctional Protein Deficiency (TFP Deficiency)
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

General Disorders detected by biochemical and other technologies

DNA - Biotinidase Deficiency (BIO)
Congenital Hypothyroidism (TSH)
Congenital Adrenal Hyperplasia (CAH)
DNA - Cystic Fibrosis (CF)
DNA - Galactosemia (GAL)
DNA - Hemoglobinopathies (HGB)

DNA: Denotes DNA confirmatory testing for some disorders with common mutations.

APPENDIX B
DISEASE/DISORDER POSSIBLE TREATMENT CONSIDERATIONS

Disease/Disorder Group	Clinical Signs/ Consequences	Possible Treatment Considerations
Amino Acids/Organic Acids/Nitrogen	Acute progressive metabolic encephalopathy: poor feeding, emesis, seizures, irritability, hypotonia, lethargy, coma and death. Clinical signs include: anorexia, emesis, tachypnea or apnea, hypertonia or hypotonia, lethargy or irritability, dermatitis (occasional alopecia), developmental delay and seizures.	Reduce the accumulation of the substrate that is toxic, e.g., amino acid-restricted diet; use supplements to help excrete toxic substrates and provide alternative metabolic pathways; provide conditionally essential nutrients; supply products of blocked primary pathways, and replace deficient cofactors *
Fatty Acid Oxidation Disorders	Encephalopathy, vomiting, hypoketotic hypoglycemia, muscle weakness, cardiomyopathy, rhabdomyolysis, metabolic acidosis, abnormal liver function tests, lactic acidosis	Avoid high fat diet, avoid fasting, provide L-carnitine supplementation when appropriate, provide essential fatty acids, use of MCT Oil, use uncooked cornstarch, continued night feedings *
Classic Galactosemia	CNS, GI, liver, renal, eye and ovarian failure, infertility, failure to thrive in almost all, emesis and diarrhea within days of exposure, jaundice, onset 4-10 days, lasting >6 weeks, hepatomegaly, ascites, cirrhosis progressing to liver failure, *Escherichea coli* sepsis; neonatal death, cerebral edema (increased intracranial pressure), cataracts, verbal dyspraxia	Galactose restricted diet *
Biotinidase Deficiency	Poor feeding, lethargy, hypotonia, developmental delay, seizures, alopecia, hearing deficits	Biotin Replacement therapy
Congenital Hypothryoidism	Development delay, mental retardation, poor growth	Thyroid Replacement Therapy
Congenital Adrenal Hyperplasia	Lethargy, life-threatening adrenal crises, coma, shock, death, failure to thrive, ambiguous genitalia in females, genitourinary plastic therapy	Cortisol Replacement Therapy
Cystic Fibrosis	Failure to thrive, pulmonary infections, chronic lung disease, pancreatic insufficiency, possible sterility in males, clubbing, rectal prolapse	Pancreatic Enzymes Replacement Therapy, Airway Clearance Techniques (ACTs), inhaled medications, antibiotics, exercise
Hemoglobinopathies	Vaso-occlusive complications, hepatosplenomegaly hemolytic anemia, splenic sequestration, gallstones, blood transfusions, splenectomy, chronic organ damage, pneumonia, acute chest syndrome, pain episodes, aplastic crisis, stroke, life threatening infections, death	Family education, immunizations, penicillin prophylaxis, prompt treatment of acute illness

***** Treatments are based on the disorder and clinical situation as determined by the biochemical geneticist.

APPENDIX C
GENETICS ADVISORY COMMITTEE MEMBERS

Hans-Georg O. Bock, M.D.
University of Mississippi Medical Center
Division of Medical Genetics
Department of Preventive Medicine
2500 North State Street
Jackson, MS 39216

Office: 601- 984-1900 **Chair**
Fax: 601-984-1916
Home: 601-992-0736
hbock@prevmed.umsmed.edu

Tami H. Brooks, M.D.
University of Mississippi Medical Center
North Pediatric Clinic
5965 I-55 North
Jackson, MS 39213

Office: 601-815-5300 **Vice Chair**
Fax: 601-978-3549
tbrooks@njpeds.umsmed.edu

Victoria (Vickie) Murdy, LCSW
Institute for Disability Studies/USM
17 Annapolis
Hattiesburg, Mississippi 39402

Office: 601-266-5163
Fax: 601-543-8445
victoria.murdy@usm.edu

Omar Abdul-Rahman, M.D.
University of Mississippi Medical Center
Division of Medical Genetics
Department of Preventive Medicine
2500 North State Street
Jackson, MS 39216

Office: 601- 984-1900
Fax: 601-984-1916
Home: 601-992-0736
oabdulrahman@prevmed.umsmed.edu

Gail Megason, M.D.
University of Mississippi Medical Center
Department of Pediatrics-Hematology
2500 North State Street
Jackson, MS 39216

Office: 601- 984-5220
Fax: 601-984-1916
Home: 601-992-0736
gmegason@ped.umsmed.edu

Michael Bailey, PhD
Mississippi Hospital Association
116 Woodgreen Crossing
Madison, Mississippi 39110

Office: 601-982-3251
Fax: 601-368-3200
Mbailey@mhanet.org

Chris Friedrich, M.D.
University of Mississippi Medical Center
Division of Medical Genetics
Department of Preventive Medicine
2500 North State Street
Jackson, Mississippi 39216

Office: 601-984-1900
Fax: 601-984-1916
cfriedrich@prevmed.umsmed.edu

Randall Henderson, M.D.
Southern Mississippi Neonatology
117 Carrie Road
Hattiesburg, MS 39402

Office: 601-288-3440
Fax: 601-288-3451
erhender@aol.com

George Moll, M.D.
University of Mississippi Medical Center
Department of Pediatric Endocrinology
2500 North State Street
Jackson, MS 39216

Office: 601-984-5217
Fax: 601-815-3672
vred2@ped.umsmed.edu

March of Dimes
Mississippi Chapter
10 River Bend Place, Suite 110
Flowood, MS, 39232

Office: 601-933-1071
Fax: 601-933-1152

Robin Wilkerson, Ph.D., R.N., B.C.
University of Mississippi Medical Center
School of Nursing
2500 North State Street
Jackson, MS 39216

Office: 601-984-6253
Fax: 601-984-6206
rwilkerson@umc.edu

Kelly Hersey, M.D.
Department of Pediatrics
University of Mississippi Medical Center
2500 North State Street
Jackson, MS 39216

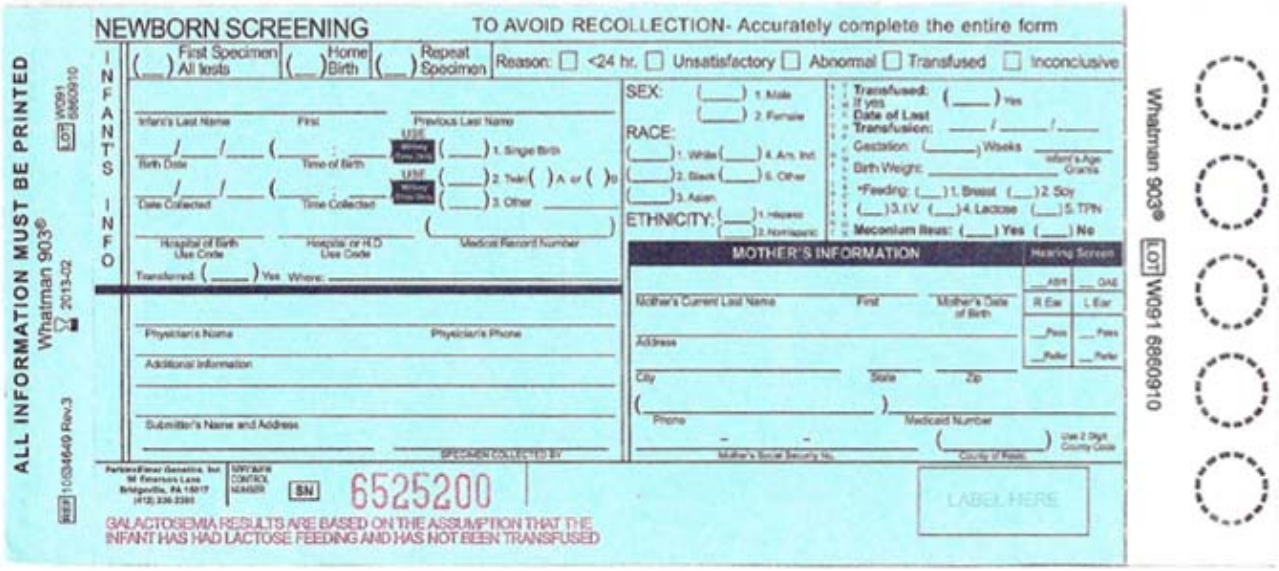
Office: 601-815-7155
Office: 601-815-2005
khersey@ped.umsmed.edu

Mrs. Julie Manley (Parent)
P.O. Box 1700
Jackson, MS 39215

Home: 601-824-1541
Cell Phone: 601-906-5442
manley_family@comcast.net

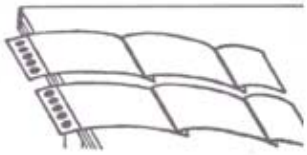
Revised March 3, 2011

APPENDIX D
DRIED BLOOD SPOT CARD



INSTRUCTIONS

1. Hold infant's limb in a dependent position to increase blood flow.
2. Clean heel thoroughly. Wipe with alcohol and dry before puncturing.
3. Puncture heel with sterile lancet deep enough to assure free flow of blood.
4. Wipe away first drop and discard.
5. Allow a large drop of blood to form on the infant's heel. Apply the back side of the filter paper directly to the puncture site where the drop of blood has formed. **The drop of blood should be large enough to approximately fill one circle.**
   DO NOT: a) Apply more than one drop of blood per circle.
   DO NOT: b) Apply blood to both front and back of filter paper.
6. Apply blood to all circles.
7. Allow blood spots to completely dry in a horizontal position at room temperature for a minimum of 4 hours (see diagram). Do not stack specimens while specimen is exposed. After drying, rewrap this cover sheet to its original position to protect specimen.



8. Send by Pre-Paid Overnight Courier within 24 hours of collection to:
   PerkinElmer Genetics, Inc.
   90 Emerson Lane
   Bridgeville, PA 15017
   (412) 220-2300
9. If you have questions please call the Mississippi State Department of Health Genetic Screening Program at (601) 576-7619.

The picture on the previous page shows both sides of the dried blood spot card. All information must be completed on the colored side of the card while instructions for specimen collection are noted on the opposite side.

To further improve specimen collection the following instructions are also provided:

- Specimen information must be legibly printed and accurate to avoid recollection.
- Specimen collection occurs regardless of age prior to discharge from the birthing facility or transfer to a higher level of care.
- Optimum specimen collection occurs between 24-48 hours of life or prior to blood transfusion.
- The first specimen collected is the INITIAL specimen; all subsequent specimens are REPEAT specimens.
- If the INITIAL specimen is collected before 24 hours of life, a REPEAT specimen should be obtained between 24-72 hours of life.
- After collection each specimen dries a minimum of 4 hours prior to shipping to the laboratory.
- Specimens must be shipped to the laboratory within 24 hours of collection.