

## **Title 15 - Mississippi State Department of Health**

### **Part IV – Office of Health Services**

#### **Subpart 01 – Division of Genetics**

#### **CHAPTER 38 RULES AND REGULATIONS GOVERNING NEWBORN SCREENING AND BIRTH DEFECTS REGISTRY**

##### **38 AUTHORITY**

###### **38.01 Statutory Authority**

Sections 41-21-201 and 41-21-203 of the Mississippi Code of 1972, Annotated, authorizes the State Department of Health to adopt rules and regulations to carry out the Newborn Screening and Follow-up Program for hypothyroidism, phenylketonuria (PKU), hemoglobinopathy, congenital adrenal hyperplasia (CAH), galactosemia, and other such conditions as specified by the State Board of Health as stated herein below in section B .

Section 41-24-1 of the Mississippi Code of 1972, Annotated, authorizes the State Department of Health to adopt rules and regulations to establish a program of testing to determine the presence of sickle cell trait or sickle cell anemia.

###### **38.02 Legal Requirements**

Under the statutory authority, the physician attending a newborn child, or the persons attending a newborn child who was not attended by a physician, is held responsible for ensuring that the child is tested for the newborn screening tests as described in these rules and regulations. State law exempts from these tests any child whose parents object thereto on the grounds that such tests conflict with their religious practices or tenets.

Under the statutory authority, screening for congenital hypothyroidism (TSH), phenylketonuria (PKU), hemoglobinopathies (Hgb), congenital adrenal hyperplasia (CAH), and galactosemia (GAL) will be conducted statewide. Screening for the following conditions, as determined and specified by the State Board of Health, will also be conducted:

Argininemia

Argininosuccinic Aciduria (ASA Lyase Deficiency)

Biotinidase Deficiency

Carbamoylphosphate Synthetase Deficiency (CPS Deficiency)

Carnitine Palmitoyltransferase I Deficiency (CPT I)  
Carnitine Palmitoyltransferase II Deficiency (CPT II)  
Carnitine/Acylcarnitine Translocase Deficiency (Translocase)  
Citrullinemia (ASA Synthetase Deficiency)  
Cystic Fibrosis (CF)  
Glutaric Aciduria Type I (GA I)  
Homocystinuria  
3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency (HMG)  
Hyperammoninemia, Hyperornithinemia, Homocitrullinemia  
Syndrome (HHH)  
Hypermethioninemia  
Isobutyryl-CoA Dehydrogenase Deficiency  
Isovaleric Acidemia (IVA)  
Long-Chain 3-hydroxyacyl-CoA Dehydrogenase Deficiency  
(LCHAD)  
Malonic Aciduria  
Maple Syrup Urine Disease (MSUD)  
Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)  
2-Methylbutyryl-CoA Dehydrogenase Deficiency  
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Def)  
3-Methylglutaconyl-CoA Hydratase Deficiency  
Methylmalonic Acidemia (MMA)  
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency  
Multiple Acyl-CoA Dehydrogenase Deficiency (MADD or GA II)  
Multiple CoA Carboxylase Deficiency  
5-Oxoprolinuria (Pyroglutamic aciduria)

Propionic Acidemia (PPA)

Short-Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)

Short-Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (SCHAD)

Trifunctional Protein Deficiency (TFP Deficiency)

Tyrosinemia Type I (TYR I)

Tyrosinemia Type II (TYR II)

Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

## 39 SPECIMEN COLLECTION

### 39.01 Specimen Collection Requirements

1. The specimen must be dried blood spots for screening and whole blood for confirmatory testing. Specimen should be collected according to the instructions issued by the Newborn Screening Program and as specified in the Child Health and Public Health Nursing Manuals.
2. Newborn screening should be performed prior to hospital discharge. Any specimen collected prior to 24 hours of age will require repeat specimen collection.
3. Newborn screening collection for GAL and Hgb are accepted for testing under the assumption that the infant has had a lactose feeding and has not been transfused. This statement is noted on Mississippi's newborn screening lab slip. Any alternate feeding status or most recent transfusion date must be appropriately documented on the lab slip.
4. The performing laboratory must receive the specimen within five working days. All specimens requiring repeat testing will be monitored by the Newborn Screening Program as follows:
  - a. Specimen repeated due to lack of information will be the responsibility of the originating hospital.
  - b. All other repeat specimen will be followed by the patient's local county health department.
5. A Mississippi State Department of Health newborn screening form must be completed in full and accompany the specimen. It is critical that the data on the form be accurate; the information entered must be compatible with

that recorded on the infant's birth certificate. The form must be completed according to the instructions issued by the Newborn Screening Program.

39.02 **Fees**

A charge will be assessed for every infant screened to defray the cost of maintaining a central registry, lab testing and health department follow-up on positive and repeat tests, for all conditions.

40 **FOLLOW-UP**

40.01 The Newborn Screening Program will be responsible for assuring that all infants with positive, questionable, and repeat screening tests are appropriately followed. Follow-up on infants who have a primary care provider will be coordinated with the provider. The local health department will provide repeat follow-up on all specimens that have been collected too early or improperly.

40.02 If the newborn screening tests have to be repeated due to lack of information on the lab slip, the hospital will be charged with finding the newborn and repeating the newborn screening tests.

41 **LABORATORY REQUIREMENTS**

41.01 **Compliance with Standards**

Any laboratory which offers this testing must meet the standards outlined in this section and, if requested, provide the agency with a written statement that they will comply with these standards. All specimens must be tested in an approved laboratory located in the United States.

The results of hemoglobinopathies, galactosemia, and congenital adrenal hyperplasia screening are not always clear cut and this type of screening requires extensive input from a recognized reference laboratory. Screening by tandem mass spectrometry requires extensive expertise and experience in this testing methodology.

A single control laboratory is required for screening. The laboratory should be proficient in all required testing methodologies.

41.02 **Specimen Requirements**

Specimen acceptable for analysis includes only dried blood spots for newborn screening, and whole blood or serum for confirmatory testing.

41.03 **Method for Specimen Analysis**

1. **Argininemia**

Method: fluorometric assay or by tandem mass spectrometry analysis

2. **Biotinidase Deficiency**

Method: continuous flow enzyme assay

3. **Congenital Hypothyroidism**

Method: Enzyme Immunoassay (EIA)

4. **Cystic Fibrosis (CF)**

Method: Immunoreactive Trypsinogen (IRT)

5. **Congenital Adrenal Hyperplasia (CAH)**

Method: Enzyme Immunoassay (EIA)

6. **Galactosemia**

Method: continuous flow chemistry analysis and Gal 1, G-PUT Deficiency Screening Test

7. **Hemoglobinopathies**

Method: isoelectric focusing

8. **Phenylketonuria (PKU)**

Method: continuous flow chemistry analysis or tandem mass spectrometry analysis

9. **Other Disorders**

Method: tandem mass spectrometry analysis, or biochemical and other established technologies.

41.04 **Quality Control**

1. The laboratory must be successfully participating in an acceptable proficiency testing program that will monitor the performance of all testing methodologies. Acceptable testing programs include the following:
  - a. College of American Pathologists (CAP)
  - b. American Association of Clinical Chemists (AACC)

- c. Centers for Disease Control (CDC)
- 2. Reagents used by the laboratory must be FDA approved. Documentation must be provided upon request for any appropriate and necessary reagent used by the laboratory that is not FDA approved.
- 3. The laboratorian must examine the quality and integrity of blood spots and must have a written procedure for rejection of those specimen judged to be unacceptable.
- 4. The laboratory must test a minimum of 40,000 specimens per year for each disorder.
- 5. Standard curves must be done with each assay of TSH and CAH.
- 6. For TSH and CAH testing, normal, borderline, and high controls must be included in each run.
- 7. Since interpretation of 17-OHP levels for CAH is weight dependent, a current weight in grams must be documented for all specimen submitted for CAH testing.
- 8. Laboratories must be Medicare approved.
- 9. Hemoglobinopathies
  - a. Control(s) containing AFSC and FAS must be included in each assay.
  - b. All samples that are not normal (not FA or AF) must be sent to a recognized reference laboratory as liquid blood unless a diagnosis has been determined by DNA analysis or other valid means.
  - c. If transfused, a repeat blood spot specimen or a liquid blood sample will be collected and tested between two and twelve weeks post last transfusion.

#### 41.05 **Disorders being Screened by Biochemical and Other Technologies**

##### 1. **BIOTINIDASE DEFICIENCY**

Biotinidase Deficiency is caused by the complete or partial lack of the enzyme biotinidase. This condition can lead to seizures, developmental delay, eczema, and hearing loss.

##### 2. **CONGENITAL ADRENAL HYPERPLASIA**

Congenital Adrenal Hyperplasia (CAH) is a genetic endocrine disorder caused primarily by a deficiency of enzymes needed for the adrenal glands to make the hormones cortisol and aldosterone. It can result in

masculinization of female genitalia as well as adrenal crisis and early infant death.

### 3. **CYSTIC FIBROSIS**

Cystic Fibrosis (CF) is an inherited condition that affects the glands that produce mucus, tears, sweat, saliva, and digestive juices. It causes severe lung damage and nutritional deficiencies. Respiratory failure is the most dangerous consequence.

### 4. **CONGENITAL HYPOTHYROIDISM**

Hypothyroidism is a disorder in which there is a decrease in the production of thyroid hormone, possibly resulting in brain damage and mental retardation in the absence of prompt treatment.

### 5. **GALACTOSEMIA**

Galactosemia is an inborn error of metabolism, inherited as an autosomal-recessive trait, in which the hepatic enzyme galactose-1-phosphate uridyl transferase is absent, preventing the conversion of the milk sugar galactose to glucose. If untreated death can occur in the first month of life.

### 6. **HEMOGLOBINOPATHIES**

Hemoglobinopathy, which includes sickle cell diseases, thalassemia, and other variants are blood disorders resulting from change in the structure of hemoglobin. Sickle Cell Disease, the most common hemoglobinopathy in Mississippi, is an inherited disease found primarily in African-Americans and people of Mediterranean descent. Although there is no cure for sickle cell disease, early detection is important for effective treatment and prevention of complications. Infection due to *Streptococcus pneumoniae* is a significant cause of death during the first few years of life for patients with sickle cell disease.

#### 41.06 **Disorders Screened by Tandem Mass Spectrometry**

A tandem mass spectrometer is an analytical instrument consisting of two mass spectrometers in series connected by a reaction chamber or collision cell. It can identify a compound by its mass and determine how much of the compound is present. Through tandem mass spectrometry analysis, many genetic disorders can be detected from one blood specimen.

#### 1. **MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY**

Medium Chain Acyl- CoA Dehydrogenase Deficiency (MCAD) is a hereditary condition that is caused by a lack of an enzyme required to convert fat to energy. For individuals with this condition, prolonged

fasting can lead to hypoglycemia, vomiting, lethargy, seizures, coma, apnea, cardiac arrest, or sudden unexplained death.

## 2. **PHENYLKETONURIA**

Phenylketonuria (PKU) is a genetic disorder inherited as an autosomal-recessive trait caused by the absence of an enzyme that is necessary for metabolism of the essential amino acid phenylalanine. If untreated, neurologic deterioration, seizures, and severe mental retardation will occur.

## 3. **OTHER DISORDERS**

Other less prevalent conditions are detectable by tandem mass spectrometry. They are grouped into amino acid disorders, organic acid disorders, and fatty acid disorders (See Attachment A). Many of these conditions can be life threatening if appropriate and timely interventions are not initiated.

### 41.07 **RECORD RETENTION**

Records of standardization, quality control, and patient values must be kept for at least two years. It is advisable for laboratories to retain these records until the statute of limitations regarding medical malpractice actions expires as stipulated by Mississippi state law.

### 41.08 **SPECIMEN RETENTION**

Specimen must be retained for at least 365 days. Under no circumstances will the retained specimen be used for research or purposes other than confirmation of previous test results.

## 42 **BIRTH DEFECTS REGISTRY**

### 42.01 **Authority**

#### 1. **Statutory Authority**

Section 41-21-205 of the Mississippi Code of 1972, Annotated, authorizes the State Department of Health (the department) to adopt rules and regulations to govern the operation of the Birth Defects Registry.

#### 2. **Legal Requirements**

Under the statutory authority, the Board of Health (the board) shall:

- a. establish in the department a birth defects surveillance program to:
  - i. identify and investigate birth defects, and

- ii. maintain a central registry of cases of birth defects
  - b. design a birth defects data system that will:
    - i. provide information to identify risk factors and causes of birth defects,
    - ii. provide information on other possible causes of birth defects,
    - iii. provide for the development of strategies to prevent birth defects,
    - iv. provide for interview studies about the causes of birth defects, and
    - v. provide for the collection of birth defect information
  - c. adopt rules, regulations and procedures to govern the operation of the registry program and to carry out the intent of this action
  - d. specify the types of information to be provided to the birth defects registry and the persons and entities who are required to provide such information to the birth defects registry
  - e. prescribe the manner in which records and other information are made available to the department
  - f. obtain records and/or test results of individuals with birth defects not previously reported or observed for inclusion in the central registry
  - g. collect, analyze and place data in the central registry to facilitate epidemiological studies/reviews and to maintain security
  - h. use the registry to:
    - i. investigate the causes of birth defects and other health conditions as authorized by statute,
    - ii. design and evaluate measures to prevent the occurrence of birth defects, and other conditions, and
    - iii. conduct other investigations and activities necessary for the board and the department to fulfill their obligation to protect the public health
3. **The Newborn Screening and Birth Defects Advisory Committee**

The State Health Officer may appoint or delegate his/her authority for the purposes of this section to an advisory committee, not to exceed (13) persons, to assist in the design and implementation of this central registry with representation from relevant groups including, but not limited to,

hospitals, physicians, board-certified clinical geneticists, personnel of the department, personnel of other appropriate state agencies, disabled persons and parents of disabled children (resulting from a birth defect). If a central registry advisory committee is created by the State Health Officer, the board shall consult and be advised by the committee on the promulgation of rules, regulations and procedures for the purposes of this section.

#### 42.02 **Identifying Reportable Cases**

##### 1. **Definition of Birth Defect**

A birth defect is an abnormality of structure, function or metabolism, whether genetically determined or a result of environmental influences during embryonic or fetal life. A birth defect may present from the time of conception through one year after birth, or later in life.

- a. From birth to one year of age certain principal birth defects shall be reported.
- b. Other birth defects found later in life may be reported at any time up to age twenty-one.

##### 2. **Reportable Birth Defects**

Live Births and Reportable Fetal Deaths with birth defects (fetal death of 20 completed weeks of gestation or more, or a weight of 350 grams or more) shall be reported. Birth Defects of the following categories must be reported:

Craniofacial	GI/GU
Neural Tube	Teratogen
Cardiac	Skeletal
Genetic Disorders	Skin
Congenital Tumors	Central Nervous System

##### 3. **Persons and Entities Required to Provide Information to the Registry**

- a. The physician must report every birth defect case the first time the patient is seen, for individuals born on or after January 1, 2000. A reporting form (See Attachment B) or its equivalent as determined by the Mississippi State Department of Health is required when reporting a suspected or diagnosed birth defect. If the patient is seen for another birth defect on another occasion, that defect shall also be reported.
- b. Appropriate birth certificate data will be reported.

- c. Appropriate data from other department registries such as the Cancer Registry, Newborn Hearing Registry will be reported.
- d. The state's tertiary care center and other hospitals will report data through newborn discharge summaries or by completing and submitting individual reporting forms.
- e. Appropriate data on specified disorders detected through newborn screening will be reported.

**4. Criteria for Inclusion as a Case**

- a. The infant/fetus must have a reportable structural defect, newborn screening disorder, functional or metabolic disorder, genetically determined or a defect resulting from an environmental influence during embryonic or fetal life.
- b. The defect optimally should be diagnosed or its signs and symptoms recognized within the first year of life, but defects can be recognized and included up to twenty-one years of age.
- c. An infant must have been born alive or a fetus must have gestational age of at least 20 weeks or a birth weight of at least 350 grams to be included in the Birth Defects Registry.

**5. Process for Making Records and Other Information Available to The Birth Defects Registry**

- a. Hospitals, physicians, and other health care professionals may submit records and birth defect information electronically or by completing and submitting individual reporting forms.
- b. The following persons who act in compliance with this section are not civilly or criminally liable for furnishing the information required under this section:
  - i. A hospital, clinical laboratory, genetic treatment center or other health care facility;
  - ii. An administrator, officer or employee of a hospital, clinical laboratory, genetic treatment center or other health care facility; and
  - iii. A physician or employee of a physician.
- c. The department field staff will visit health care facilities to gather medical and other required information of children with birth defects. This information will be recorded on registry data report forms (See

Attachment B) on potentially reportable conditions to be added to the registry.

- d. The department may obtain records and/or test results of individuals with known or potential birth defects not previously reported.

#### 6. **Confidentiality and Security**

- a. Information collected and analyzed by the department under this section shall be placed in the central registry to facilitate epidemiological studies/reviews and to maintain security.
  - i. Data obtained under this section directly from the medical records of a patient is for the confidential use of the department and the persons or public or private entities that the department determines are necessary to carry out the intent of this section. The data is privileged and may not be divulged or made public in a manner that discloses the identity of an individual whose medical records have been used for obtaining data under this section.
  - ii. Information that may identify an individual whose medical records have been used for obtaining data under this section is not available for public inspection under the Mississippi Public Records Act of 1993.
  - iii. Statistical information collected under this section is public information.
- b. Misuse of the Registry Data:

Any person or entity who misuses the information provided to the registry shall be subject to a civil penalty of Five Hundred Dollars (\$500.00) for each such failure or misuse. Such penalty shall be assessed and levied by the board after a hearing, and all such penalties collected shall be deposited into the State General Fund.

#### 7. **Policies and Procedures**

The department will maintain written policies and procedures to guide the operations of the Birth Defects Registry.

# CONCISE SUMMARY OF ECONOMIC IMPACT STATEMENT

## STATE OF MISSISSIPPI Mississippi State Department of Health

MS State Department of Health  
c/o Daniel R. Bender  
P.O. Box 1700  
Jackson, MS 39215-1700  
601.576.7472  
[daniel.bender@msdh.state.ms.us](mailto:daniel.bender@msdh.state.ms.us)

Specific Legal Authority Authorizing the promulgation of  
Rule: Section 41-21-201 of MS code

Reference to Rules repealed, amended or suspended by the  
Proposed Rule: Chapter 38 and 42 of the Rules and  
Regulations governing the Newborn Screening Program and  
Birth Defects Registry 39.02 (Fees) and 42.01 (Authority) # 3  
Newborn Screening and Birth Defects Advisory Committee

An Economic Impact Statement is required for this proposed rule by Section 25-43-3.105 of the Administrative Procedures Law. This is a Concise Summary of the Economic Impact Statement which must be filed with the Notice of Proposed Rule Adoption in the Secretary of State's Office. The full text of the Economic Impact Statement may be obtained from the agency contact person at the above address.

Persons may present their views by submitting written comments on the proposed rule adoption to the agency contact person at the above address. Additional information on where, when and how persons may present their views or demand an oral proceeding on the proposed rule are included in the Notice of Proposed Rule Adoption to which this is attached.

a. Description of the need for and the benefits of the proposed rule:

This proposed rule will allow the MS State Department of Health to increase fees to support the operation of the State Newborn Screening Program. Currently the program screens approximately 45,000 infants and provides follow-up for infants and families identified with genetic diseases and disorders.

b. Cost estimate to the agency and other state or local government entities:

The proposed increase of \$30 per screen will be assessed to delivering hospitals on each newborn screened. Insurance companies and other third party payors of newborn/maternity care will also be impacted. The proposed increase of \$30 per screen is estimated to cost an increase of \$1.35 million to birthing hospitals in the state. Other agencies who may be impacted include insurance companies that pay for maternity services.

c. Estimate of the cost or economic benefit to all persons:

The increase is estimated at \$1.35 million. The economic benefits of this program and its services are timely screening of newborns and coordination of follow-up to medical subspecialty providers for children identified with special health care needs. Early identification of genetic conditions may result in the improved quality of life and decrease disabilities or even the death of a child.

d. Analysis of the impact on small business:

This fee increase affects all birthing hospitals in the state. Hospitals who deliver babies have the capability to bill insurance programs and other third party payors for this cost associated with maternity/newborn services.

e. Comparison of the costs and benefits of the proposed rule to the probable costs and benefits of not adopting the rule:

The benefits of approving this rule include the availability of funds to maintain operations of the program which may result in improved quality of life and decrease in life long disabilities or even the death of affected children. The cost of not adopting the rule to increase fees may result in less qualified staff and the decrease in services offered which are covered by fees which are used to improve programmatic operations.

f. Determination as to whether less costly or less intrusive methods exist to achieve the purpose of the rule:

In order to provide on-going surveillance of the Newborn Screening Program, an increase in the fees will support and maintain a successful screening and follow-up program. Additional funding is needed to maintain a quality and effective Newborn Screening Program.

g. Description of reasonable alternative methods and reasons for rejection of the alternative methods:

The Newborn Screening Program operates financially on the fees collected from screening. It is important to increase fees to maintain an effective screening and follow-up program. The alternative might include the Newborn Screening Program requesting funds from other sources, including state general funds, to support operations. Without this increase, the level of services may also be impacted.

h. Data and methodology in making the estimates in the economic impact statement: The methodology for determining the increase in fees includes taking the following associated costs into consideration: laboratory contract for testing salaries and travel expenses for public health staff who provide follow-up, contracts with medical subspecialty providers, training and materials necessary to maintain a credible screening program for the infants in Mississippi.

**Date Rule Proposed: January 29, 2010**

**Proposed Effective Date of Rule: July 1, 2010**



**Signature and Title of Person Submitting Rule  
for Filing**

SOS FORM APA 004  
Effective Date 07/29/2005

# ATTACHMENT A

LIST OF CONDITIONS INCLUDED IN MISSISSIPPI NEWBORN SCREENING

# Mississippi Genetic Newborn Screening

Effective June 1, 2003

## Disorders detected by tandem mass spectrometry (MS/MS)

### Amino Acid Disorders

Argininosuccinic Aciduria (ASA Lyase Deficiency)  
Carbamoylphosphate Synthetase Deficiency (CPS Deficiency)  
Citrullinemia (ASA Synthetase Deficiency)  
Homocystinuria (HCys)  
Hyperammoninemia, Hyperornithinemia, Homocitrullinemia Syndrome (HHH)  
Hypermethioninemia (MGT)  
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)  
**DNA** - Glutaric Aciduria Type I (GA I)  
Isobutyryl-CoA Dehydrogenase Deficiency  
Isovaleric Acidemia (IVA)  
Malonic Aciduria  
2-Methylbutyryl-CoA Dehydrogenase Deficiency  
3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC)  
3-Methylglutaconyl-CoA Hydratase Deficiency (3MGH)  
**DNA** - Methylmalonic Acidemia (MMA)  
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency  
Multiple CoA Carboxylase Deficiency (MCCD)  
Propionic Acidemia (PPA)

### Fatty Acid Oxidation Disorders

Carnitine/Acylcarnitine Translocase Deficiency (Translocase)  
Carnitine Palmitoyltransferase I Deficiency (CPT I)  
**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)  
**DNA** - Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

### General Disorders detected by biochemical and other technologies

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

**DNA** = DNA testing also done on presumptive positive screens for these disorders

# Mississippi State Department of Health

Birth Defects Registry Reporting Form  
Genetics Services  
Post Office Box 1700  
Jackson, MS 39215-1700  
Phone: 601-576-7619

The physician must report every birth defect case the first time the patient is seen, for individuals born on or after January 1, 2000. A reporting form is required when reporting a suspected or diagnosed birth defect. If the patient is seen for another birth defect on another occasion, that defect shall also be reported.

## 1. Patient's Information

Patient's name: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_  
Last First MI Suffix Date of Birth

Sex:  Male  Female Race:  American Indian  Asian  Black  Hispanic  White  Other \_\_\_\_\_  
Specify

Admission date: \_\_\_\_\_ Discharge date: \_\_\_\_\_ Medical Record #: \_\_\_\_\_

Mississippi Resident at Birth:  Yes  No

## 2. Birth Information (If Known)

Delivery status:  Fetal Death  Induced Term  Live Birth  Stillborn

Birth Multiplicity:  Single  Twin  More than two Birth Weight \_\_\_\_\_  
Grams

Birth Facility: \_\_\_\_\_

Current Medical Provider: \_\_\_\_\_

## 3. Birth Mother (or Other Responsible Party if Mother Unknown)

Name: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_  
First Middle Last Relationship to patient

Address: \_\_\_\_\_

City, State, Zip: \_\_\_\_\_ County: \_\_\_\_\_

Date of Birth: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

Caregiver Name: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_  
(If different from above) First Middle Last

## 4. Diagnosis (ICD 9/ICD 10 and brief description)

	_____
	_____
	_____
	_____
	_____
	_____
	_____
	_____
	_____
	_____
	_____

## 5. Contact Information

Hospital: \_\_\_\_\_

Reporting Physician: \_\_\_\_\_

Date reported: \_\_\_\_\_

Submitter's name: \_\_\_\_\_

Submitter's phone #: \_\_\_\_\_

*Hospital staff to contact if additional information is needed*

## Additional information

\_\_\_\_\_

## 6. Death Information (If applicable)

Death Date: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

**MSDH Genetics Services use only**

Received date: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

Entered into BDRS: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

By: \_\_\_\_\_

District: \_\_\_\_\_

Confidential Information

# ATTACHMENT B

MANDATED BIRTH DEFECTS REPORTING FORM

**BIRTH DEFECTS REGISTRY REPORTING FORM**  
**FORM No. 272**

**PURPOSE**

This form is designed for documentation and reporting of all infants and children with birth defects from birth to 21 years of age to the State's Birth Defects Registry.

**INSTRUCTIONS**

**1. Patient's Information**

Last - Enter last name at birth, along with new last name (if name has changed since birth).

First - Enter first name (if known).

Middle - Enter middle name (if known).

Suffix - Enter suffix (if known).

Date of Birth - Enter infant's date of birth.

Sex - Check Male or Female.

Race - Check race.

Admission Date - Enter month, day and year of hospital admission.

Discharge date - Enter month, day and year infant was discharged from hospital.

Medical Record - Enter number assigned to the record by the facility to track medical records.

Mississippi Resident Birth - Check whether infant's mother was a Mississippi resident at birth.

**2. Birth Information (If Known)**

Delivery Status - Check fetal delivery status.

Birth Multiplicity - Check if a single birth, a twin or more than two.

Birth Weight - Enter infant's weight at birth in grams.

Birth Facility - Enter name of hospital where infant was born.

Current Medical Provider - Enter name of pediatrician or primary care provider.

**3. Birth Mother (or Other Responsible Party if Mother Unknown)**

Name - Enter first name, middle name and last name.

Relationship to Patient - Enter relationship to infant.

Address - Enter address (street name and house or apartment number, or P.O. Box)

City/State/Zip - Enter city, state and zip code.

County - Enter county where mother lives.

Mother's Date of Birth - Enter month, day and year of mother's birth.

Caregiver Name - Enter name of caregiver if different from birth mother.

#### **4. Diagnosis**

Enter ICD 9/ICD 10 code that corresponds to the condition and a brief description of diagnosis/defect.

#### **5. Contact Information**

Hospital - Enter name of hospital submitting the report.

Reporting Physician - Enter name of the physician reporting birth defect.

Date reported - Enter date of report.

Submitted by Name/Phone - Enter name of person submitting report and complete telephone number.

#### **6. Death Information**

Date - Enter date of death (if applicable).

### **OFFICE MECHANICS AND FILING**

The completed forms are sent to:

Genetics Services/Birth Defects Surveillance Registry  
Mississippi State Department of Health  
P.O. Box 1700  
Jackson, MS 39215-1700

The information is entered into the Birth Defects Surveillance Registry (BDSR) database.  
The form is shredded after data is entered.