

# ARKANSAS LEGISLATIVE COUNCIL

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**Room 315, State Capitol Building**  
**Little Rock, AR 72201**

**Marty Garrity**  
Executive Secretary

Tel: 501-682-1937  
Fax: 501-682-1936

March 19, 2020

Ms. Amy Fecher, Secretary  
Department of Transformation and Shared Services  
Business Operations  
501 Woodlane St, Ste 203-N  
Little Rock, Arkansas 72201

Dear Ms. Fecher:

Pursuant to ALC Rule 16, the Co-Chairs of the Legislative Council and the ALC Administrative Rules Subcommittee determined that emergency approval of the following appropriation requests was necessary to enable the agencies to perform essential services: *Arkansas Department of Health - Rules Pertaining to Newborn Screening*.

As such, the Co-Chairs of the Legislative Council and the Co-Chairs of the ALC Administrative Rules Subcommittee acted on behalf of the Legislative Council in regard to this request. This request shall be filed as approved and reported to the Legislative Council at its next regularly scheduled meeting in accordance with ALC Rule 16.

Please notify each affected agency of this emergency approval.

Sincerely,

A handwritten signature in black ink, appearing to read "Marty Garrity".

Marty Garrity  
Executive Secretary

MG:sla

Attachment

Cc: Senator Cecile Bledsoe, Co-Chair, Arkansas Legislative Council  
Representative Jeff Wardlaw, Co-Chair, Arkansas Legislative Council  
Senator Lance Eads, Co-Chair, ALC Administrative Rules  
Representative Dan Sullivan, Co-Chair, ALC Administrative Rules  
Dr. Nathaniel Smith, MD, MPH, Secretary, Arkansas Department of Health

**ALC Report  
For Emergency Review and Approval  
of Rule**

**March 18, 2020**

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On March 18, 2020, Senator Cecile Bledsoe and Representative Jeff Wardlaw, Co-Chairs of Legislative Council, and Senator Lance Eads and Representative Dan Sullivan, Co-Chairs of the Administrative Rules Subcommittee, received written notification from Amy Fecher, the Secretary of the Department of Transformation and Shared Services, requesting Emergency Review and Approval of a Rule under Rule 16(a) of the Rules of the Arkansas Legislative Council.

Pursuant to Rule 16, the Co-Chairs of the Legislative Council and the Administrative Rules Subcommittee determined that emergency review and approval of the Rule was necessary to enable the agency to perform essential services. As such, the Co-Chairs of the Legislative Council and the Co-Chairs of the Administrative Rules Subcommittee may act on behalf of the Legislative Council in regard to this request, and state herein that this request shall be filed as reviewed and approved and reported to the Legislative Council at its next regularly scheduled meeting in accordance with Rule 16.

Below is a summary of the Rule that was given emergency review and approval.

Department of Health, Public Health Lab  
Rules Pertaining to Testing of Newborn Infants

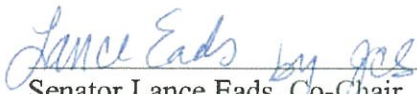
Respectfully Submitted,



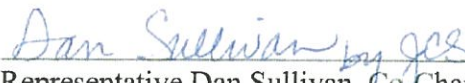
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Senator Cecile Bledsoe, Co-Chair  
Arkansas Legislative Council



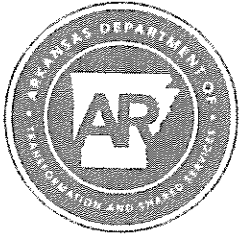
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Representative Jeff Wardlaw, Co-Chair  
Arkansas Legislative Council



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Senator Lance Eads, Co-Chair  
Administrative Rules Subcommittee



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Representative Dan Sullivan, Co-Chair  
Administrative Rules Subcommittee



Department of Transformation and Shared Services  
Governor Asa Hutchinson  
Secretary Amy Fecher

March 17, 2020

The Honorable Senator Lance Eads, Co-Chair  
The Honorable Representative Dan Sullivan, Co-Chair  
Administrative Rules Subcommittee  
Arkansas Legislative Council  
Via Email to Jessica Sutton, Administrator

**RE: Request for Immediate Consideration of Rules**

The Secretary of the Department of Transformation and Shared Services has surveyed all the Departments that have pending Rules on the March 19, 2020 agenda. The only request for immediate consideration of a rule is for the Board of Health and the Arkansas Department of Health concerning the Rules Pertaining to Newborn Screening. It is in the best interest of the State of Arkansas and essential for the Department of Health to enable the State Public Health Lab to immediately test newborns for four additional diseases, including Spinal Muscular Atrophy (SMA), mandated by Act 58 of 2019.

The proposed amendments to the Rules pertaining to Testing for Newborn Infants adds tests for newborn screening including for Spinal Muscular Atrophy, (SMA), pursuant to Act 58 of 2019. The proposed amendments to the Rules also add three additional tests for Pompe Disease, MPS 1 spectrum of disease, and childhood onset (cerebral) X-ALD. At the present time, 31 tests are performed on newborns, and 29 of the tests are performed in the Public Health Laboratory. The tests are performed on dried blood spots that are collected from the infants soon after 24 hours of birth. After passing the accreditation process, these four tests can now be performed in the State Public Health Laboratory utilizing existing blood samples without having to require new blood samples from the hospital. These tests allow for early treatment of conditions for better outcomes for children.

The current fee is \$121.00 and the proposed amendment adds an additional \$10.00 charge for the SMA test. The fee is \$131.00. This fee amount, which is an increase from the previous fee amount, was agreed upon by Arkansas Medicaid and the Arkansas Insurance Department. The Department of Human Services, Division of Medical Services, promulgated its corresponding rule, Hospital 3-19 Spinal Muscular Atrophy Newborn Screening, and its final rule has been filed.

Sincerely,

  
Amy Fecher

Office of the Secretary  
501 Woodlane, Suite 201 \* Little Rock, AR 72201 \* 501.319.6551  
TRANSFORM.AR.GOV

# ARKANSAS STATE BOARD OF HEALTH

## RULES PERTAINING TO TESTING OF NEWBORN INFANTS



Promulgated Under the Authority of  
Ark. Code Ann. § 20-15-301 et  
seq., and Act 58 of 2019

*Effective on*

Arkansas Department of Health  
Nathaniel Smith, MD, MPH,  
Secretary of Health

RULES PERTAINING TO TESTING OF NEWBORN INFANTS  
Table of Contents

I. DEFINITIONS.....	2
II. PURPOSE.....	4
III. AUTHORITY.....	5
IV. RESPONSIBILITY.....	5
V. SPECIMEN COLLECTION AND SUBMISSION.....	7
VI. ANALYSIS, INTERPRETATION, AND REPORTING OF RESULTS.....	8
VII. ARKANSAS DEPARTMENT OF HEALTH ROLE IN TREATMENT AND MONITORING.....	10
VIII. SEVERABILITY.....	11
IX. REPEAL.....	11

**SECTION I. DEFINITIONS.**

A. **Phenylketonuria (PKU), Congenital Hypothyroidism (CH) and Galactosemia** are conditions (diseases) which cause irreversible brain damage and mental retardation unless they are detected and treated at a very early stage in the life of a newborn individual. Untreated Galactosemia also results in liver disease, cataracts, and increased susceptibility to serious infection. Early diagnosis and treatment are absolutely essential in order to avoid these health problems.

B. **Sickle Cell Disease (SS)** is the most common inherited abnormality of a red blood cell protein called hemoglobin. It is caused by a genetic abnormality that must be inherited from both parents. Sickle Cell Disease may cause serious health problems even in the first few months of life. It occurs much more commonly in people of African American, Asian and Mediterranean descent. In addition to the anemia, it lowers resistance to infection, and is associated with increased morbidity and mortality unless diagnosed and treated early. Sickle Cell Disease is one of a handful of related hemoglobinopathies each of which can cause similar health problems with varying severity.

C. **Sickle Cell Trait (AS)** and other hemoglobinopathy traits differ from their corresponding diseases. Traits occur when the genetic abnormality is inherited from only one parent, the other parent contributing a normal gene. Hemoglobinopathy traits cause only minor health issues that show up occasionally in life. They are associated with normal life spans. Sickle Cell Trait (AS) is the most common, occurring in 8 to 10% of African Americans.

D. **Biotinidase Deficiency (BIOT)** is caused by the lack of an enzyme called biotinidase, resulting in an inability of the body to use Vitamin B substances absorbed by the intestines. Without sufficient biotin, several other critical enzyme systems are unable to function properly. Biotinidase deficiency can lead to seizures, developmental delay, skin rash, and hearing loss. Newborns with the disorder appear normal, but develop critical symptoms after the first weeks or months of life. Symptoms include floppiness, seizures, developmental delay, hair loss, rashes, hearing loss and vision loss. Metabolic acidosis can result in coma and death. A daily biotin dietary supplement can prevent all symptoms.

E. **Congenital Adrenal Hyperplasia (CAH)** is a group of disorders caused by the deficiency of an adrenal enzyme resulting in decreased production of important hormones called cortisol and aldosterone. Cortisol helps the body respond to stressful events. Aldosterone helps the body maintain its fluids and salts. Without enough of these hormones, the affected newborn may appear normal, but can quickly develop symptoms including lethargy, vomiting, muscle weakness and dehydration. In severe cases death may occur within a few weeks if left untreated. One kind of CAH may show up first as ambiguous genitalia in the newborn. Infants with milder forms of the disorder are still at risk for reproductive and growth difficulties. If detected early and maintained on appropriate doses of medication, infants diagnosed with CAH can have normal growth and development.

F. **Cystic Fibrosis (CF)** is a disorder in which the body cannot make an important protein involved in using chloride ions, an ingredient in table salt. The major clinical consequences are the production of abnormally thickened mucous secretions in the lungs and digestive systems of affected newborns. With early detection and lifelong comprehensive treatment plans, infants diagnosed with CF can be expected to live longer and in a better state of health than in the past.

G. **Amino Acid Disorders** make up a group of inherited conditions in which protein metabolism is disrupted. Onset of symptoms may occur shortly after birth or after an apparently normal neonatal period. The symptoms may occur in episodes with normal periods in between. The clinical onset may include unusual odors in the urine, irritability, poor feeding, changes in muscle tone, lightened pigmentation, failure to thrive, jaundice, or liver enlargement. Other symptoms include intoxication-like symptoms such as vomiting, lethargy, seizures, and coma. Treatment of amino acid metabolism disorders includes a low-protein diet strictly controlling intake of specific amino acids.

H. **Fatty Acid Oxidation Disorders** make up a group of genetic metabolic deficiencies in which the body is unable to oxidize (break down) fatty acids to make energy. An enzyme is either missing or not working correctly. The main source of energy for the body is a sugar called glucose. Normally when the glucose runs out, fat is broken down into energy. However, that energy is not readily available to children and adults with a fatty acid oxidation disorder. If undiagnosed and untreated, these disorders can lead to serious complications affecting the liver, heart, and eyes; general muscle development; and possibly death. Symptoms of a metabolic “crisis” include vomiting, diarrhea, lethargy and difficulty breathing.

I **Organic Acid Disorders** make up a group of inherited metabolic diseases that lead to accumulation of organic acids in biological fluids (e.g., blood and urine). The accumulation produces disturbances in the acidity of the body and causes alterations in metabolic chemical reactions. These disorders can cause intoxication-like symptoms such as vomiting, metabolic acidosis, ketosis, dehydration, or coma. Some patients may have too little sugar in the blood, or too much lactic acid or ammonia. Chronic symptoms include recurrent vomiting, failure to thrive, floppiness and general developmental delay. Symptoms of these disorders can be diminished by restricting protein in the diet and, in some cases, supplementation with vitamins or a nutrient called carnitine.

J. **Severe Combined Immunodeficiency (SCID)** is a group of disorders characterized by severe defects in the T-lymphocyte and B-lymphocyte systems. Affected babies are susceptible to multiple types of life-threatening bacterial, viral, and fungal infections. Early diagnosis of SCID is imperative as SCID is curable with hematopoietic stem cell transplantation. Infants with SCID die of infections by age two (2) years unless immunity is reconstituted by treatment. SCID is commonly known as the “bubble boy” disease.

K. The **Collector** is the person or party responsible for collecting and submitting the blood specimen for testing. The persons or parties who are collectors under these Rules and Regulations are described in Section IV.A.

L. **The Department** is the Arkansas Department of Health.

M. **Spinal muscular atrophy (SMA)** is a genetic disease affecting the central nervous system, peripheral nervous system, and voluntary muscle movement (skeletal muscle). Most of the nerve cells that control muscles are located in the spinal cord, which accounts for the word spinal in the name of the disease.

N. **Pompe disease** is an inherited disorder caused by the buildup of a complex sugar called glycogen in the body’s cells. The accumulation of glycogen in certain organs and tissues, especially muscles, impairs their ability to function normally.

O. **Mucopolysaccharidosis (MPS1)** refers to a group of inherited conditions in which the body is unable to properly breakdown mucopolysaccharides (long chains of sugar molecules that are found throughout the body). As a result, these sugars buildup in cells, blood and connective tissue which can lead to a variety of health problems.

P. **Adrenoleukodystrophy (X-ALD)** is a disease linked to the X chromosome. It is a result of



fatty acid buildup caused by the relevant enzymes not functioning properly, which then causes damage to the myelin sheath of the nerves, resulting in seizures and hyperactivity. Other symptoms include problems with speaking, listening, and understanding verbal instructions.

## **SECTION II. PURPOSE.**

The purpose of these Rules is to assure that all infants born in Arkansas have the opportunity to be screened for genetic illnesses.

These Rules provide a method to assure that:

1. All newborn infants are tested for Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia, Sickle Cell Disease (SS), Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, ~~and~~ Severe Combined Immunodeficiency (SCID), Spinal Muscular Atrophy (SMA), Pompe Disease, MPS 1 spectrum of disease, and childhood onset (cerebral) X-ALD.
2. All newborns with an abnormal screen receive appropriate medical follow-up.

## **SECTION III. AUTHORITY.**

These Rules are promulgated pursuant to the authority conferred by Arkansas Code Annotated § 20-15-301 et seq. and Act 58 of 2019.

## **SECTION IV. RESPONSIBILITY.**

- A. Collection and Submission.
  1. Medical Facilities/Medical Staff: In all cases where the birth of an infant occurs in a medical facility licensed by the Board of Health, it shall be the responsibility of the governing body and medical staff of the facility to adopt and enforce policies and procedures which ensures that blood test for Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia, Sickle Cell Disease (SS), Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, ~~and~~ Severe Combined Immunodeficiency (SCID), Spinal Muscular Atrophy (SMA), Pompe Disease, MPS 1 spectrum of disease, and childhood onset (cerebral) X-ALD are conducted and processed in accordance with these rules. The licensed facility shall also be responsible for submission of the usable blood specimen in cases where an infant less than six months of age is admitted (i.e., born out of hospital, neonatal transfer, etc.), and it is brought to the attention of the facility or the attending physician that the infant is untested. If an infant is discharged from a licensed medical facility without collection and submission of a usable specimen for testing, it shall be the responsibility of the



discharging facility and the attending physician to arrange for the testing. The discharging facility and attending physician shall notify the Arkansas Department of Health (“Department”) within one week of discharge if their efforts fail to arrange for testing.

2. Physicians: Physicians assuming care of infants who are under six months of age and who come to their attention as being untested or inadequately tested for Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia, Sickle Cell Disease (SS), Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis(CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, and Severe Combined Immunodeficiency (SCID), Spinal Muscular Atrophy (SMA), Pompe Disease, MPS 1 spectrum of disease, and childhood onset (cerebral) X-ALD, shall also be responsible for assuring collection and submission of usable blood specimens for these infants.
3. Licensed Midwives: In cases where the birth occurs outside a licensed medical facility or in the home, it shall be the responsibility of an attending licensed midwife to advise the parents of this law and the procedure for conducting newborn screening, and documenting that a blood sample is obtained after 24 hours and no later than 72 hours after birth. If the blood sample is not obtained for any reason, an attending licensed midwife must document the incident in the patient’s chart.
4. The Department: The Department’s Local Health Unit shall collect and submit usable blood specimens on all infants under six months of age who come to their attention as being tested or inadequately tested. This responsibility shall not be in lieu of that of the preceding individuals and facilities.

B. Payment

1. The Collector will be charged a fee of one hundred and thirty-one dollars (\$131.00) for the processing and testing of newborn screening specimens by the Department.
2. The Board of Health may determine the amount of this fee based on the Department’s cost to process and test the specimens.

C. Laboratory Analysis

1. The Department shall be responsible for provision of forms and instructions for the blood specimen collection; processing and recording of the specimen received; analysis of specimen; determination of abnormal results; and reporting of lab results within a time period which would allow preventive medical intervention.

D. Follow-Up

1. The Department shall be responsible for the interpretation of laboratory results and the reporting of abnormal results to the attending physician or birth attendant. If the screening result is suggestive of Classical or Variant

PKU, Galactosemia, Sickle Cell Disease (SS), Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, ~~or~~ Severe Combined Immunodeficiency (SCID), Spinal Muscular Atrophy (SMA), Pompe Disease, MPS 1 spectrum of disease, or childhood onset (cerebral) X-ALD, the Department shall consult with specialist physicians who are retained by contract to provide clinical advice on these conditions. The Department shall notify the Collector of the specimen and enter the infant's information in a tracking system maintained to evaluate program operations and infants' medical outcomes.

2. Attending Physician/Medical Attendant:

- a) Upon receipt of a notice of an abnormal test result the physician or medical attendant shall be responsible for the appropriate medical treatment, referral, and/or retesting within the timeframe specified by the Department for that particular disorder. It is strongly recommended that consultation be obtained with a physician who has special competence in the management of these disorders.
- b) The attending physician or other responsible health care provider who conducts testing in follow-up to abnormal screens shall report subsequent test results (whether negative or positive) to the Department. To provide for long term follow up the Department will collect data on affected infants each year for five years to determining health care maintenance and health status, especially the presence of mental retardation or permanent disability.

The Department will establish protocols for follow-up of all screened disorders in collaboration with medical specialists under contract. For infants with abnormal test results, the physician will be notified of the results and informed of the recommended protocols for follow-up of the specific order.

## **SECTION V. SPECIMEN COLLECTION AND SUBMISSION**

- A. The blood specimen for PKU, CH, Galactosemia, Sickle Cell Anemia, Biotinidase (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, ~~or~~ Severe Combined Immunodeficiency (SCID), or Spinal Muscular Atrophy (SMA), Pompe Disease, MPS 1 spectrum of disease, and childhood onset (cerebral) X-ALD testing must be collected and submitted as described below:
- B. Timing of Specimen Collection
  1. For all healthy infants born in medical facilities, the specimen shall be collected before the time of discharge from the facility. Optimum time for collection is 24 to 72 hours after birth, and all Collectors should strive to comply with that time frame. If any infant is discharged or specimen

collected prior to 24 hours of age, a repeat test shall be arranged by the medical facility and the attending physician. This repeat specimen shall be collected by the infant's seventh day of life. A repeat test for Sickle Cell Disease shall not be required if specimen was collected prior to 24 hours of age.

2. Specimens from ill or premature infants shall be obtained as soon as possible after their condition has sufficiently stabilized.
3. Specimens from infants not born in medical facilities shall be collected between 24 and 72 hours after birth.

Infants under six months of age who are known to be untested or inadequately tested shall have blood specimens collected and submitted by the responsible authority as soon as possible.

C. Specimen Collection and Submission

1. Specimens shall be dispatched to the Arkansas Department of Health Public Health Laboratories, Little Rock, Arkansas, no later than one (1) business day from collection. Specimens are submitted only on forms provided by the Public Health Laboratory. The Collector is responsible for supplying complete and accurate identifying information on the collection form to be used for tracking infants with abnormal screening results.

D. Forms

1. Submission: forms may be obtained by writing to the Public Health Laboratories at:

Arkansas Department of Health  
201 South Monroe Street  
Little Rock, AR 72205

The county health units will not supply these forms.

E. Unsatisfactory Specimens

1. Inadequate, contaminated, or otherwise unusable specimens shall be reported to the Collector after laboratory determination of an unsatisfactory specimen. The Collector shall be responsible for assuring recollection and resubmission within seven calendar days of notification.

## **SECTION VI. ANALYSIS, INTERPRETATION, AND REPORTING OF RESULTS**

A. Laboratory Analysis

1. All specimens received by the laboratory shall be initially examined within five working days of receipt. Abnormal results shall be reported to the Collector within two working days of determination.

B. Interpretations of Results

1. Phenylketonuria (PKU)

- a) The Department shall define the phenylalanine level which constitutes a positive screening result for PKU.
  - b) An infant whose phenylalanine level is determined by the Department to be negative for PKU requires no action to be taken. However, attending physicians shall give special consideration when testing circumstances or infant evaluation/family history suggests the possibility of need for prescreening in cases where PKU or PKU variants may actually exist in spite of initial negative screening results.
2. Congenital Hypothyroidism (CH)
- a) The Department shall define the thyroxine and thyroid stimulating hormone levels which constitute positive screening results for CH.
  - b) Occasionally test results suggestive of CH may be reported which, upon retesting, will be found within normal limits. Likewise it is possible that test results which are reported as normal in the neonatal period could mask the delayed onset of CH. While an infrequent occurrence, in the face of clinical findings, this possibility must be considered by the attending physician.
3. Galactosemia
- a) The Department shall define the galactose-1-phosphate uridyl transferase (GALT) levels which constitute positive screening results for Galactosemia.
  - b) It is possible that an infant affected with Galactosemia could have normal initial screening results. This situation is most likely to occur in infants who have received no or insufficient feedings with lactose-containing milk or formula prior to testing, or who have received blood transfusions prior to testing.
  - c) The medical caretaker shall give special consideration to retesting any infant whose case findings, testing circumstances, or family history seems to medically warrant it.
4. Sickle Cell Anemia or Trait
- a) The Department shall define the laboratory value which constitutes a positive screening result for Sickle Cell Disease (SS), Sickle Cell Trait (AS) or other related hemoglobinopathy.
  - b) An infant whose hemoglobin is determined by the Department to be negative for SS or other related serious hemoglobinopathies requires no special consultation; however, infants with trait conditions should be followed for mild anemias and urinary tract infections.
  - c) The medical caretaker shall give special consideration to re-testing any infant whose case findings, testing circumstances, or family history seems to medically warrant it.
5. Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia

(CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, Severe Combined Immunodeficiency (SCID).

- a) The Department shall define the laboratory value which constitutes a positive screening result for Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders Severe Combined Immunodeficiency (SCID).

C. Reporting of Results

1. Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders Severe Combined Immunodeficiency (SCID).
  - a) Immediately upon obtaining the initial positive screening result, the Department shall notify the attending physician or medical attendant, who shall be responsible for ensuring that prompt follow-up diagnostic testing is conducted.
  - b) Appropriate, expectant medical management shall not be withheld pending the confirmatory test results. A non-physician Collector shall immediately refer the infant for appropriate medical intervention. It is recommended that a pediatric geneticist, endocrinologist, or pulmonologist consultant be utilized in the management of these infants.
2. Sickle Cell Disease (SS) and other serious Hemoglobinopathies
  - a) Immediately upon obtaining the initial positive screening result, presumptive of SS or other serious hemoglobinopathy, the Department shall notify the Collector, who shall be responsible for insuring that prompt follow-up diagnostic testing is conducted.
  - b) Appropriate, expectant medical management shall not be withheld pending the confirmatory test results for either SS or other related hemoglobinopathy. Therefore, non-physician Collector shall immediately refer the infant for appropriate medical intervention. It is recommended that a pediatric hematologist consultant be utilized in the management of these infants.
  - c) Immediately upon obtaining an initial positive screening, presumptive of trait, the Department shall notify the Collector in writing. The parent shall be notified in writing by the Department.

**SECTION VII. ARKANSAS DEPARTMENT OF HEALTH ROLE IN TREATMENT AND MONITORING**

- A. Listing of Consultants
  - 1. For Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia, Sickle Cell Disease and other hemoglobinopathies, Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, Severe Combined Immunodeficiency (SCID), the Department shall maintain a list of pediatric consultants having special competence in these disorders, and shall make the names of such consultants known to the attending physicians of infants with abnormal screening test results.
  
- B. Registry
  - 1. For Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemias, Sickle Cell Disease (SS), and other hemoglobinopathies, Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, Severe Combined Immunodeficiency (SCID), the Department shall maintain a registry to record laboratory results and diagnoses of all tested infants, and to track referral for those infants in whom abnormal findings were noted during the screening process.
  
- C. Nutritional Therapy
  - 1. Phenylketonuria (PKU)
    - a) Nutritional therapy with low phenylalanine formula and/or foods shall be instituted after the diagnosis of PKU.
  
  - 2. Galactosemia
    - a) Nutritional therapy with lactose-free formula and/or foods shall be instituted after the diagnosis of Galactosemia.
  
  - 3. Other genetic conditions
    - a) Other genetic conditions discovered by the laboratory testing done pursuant to these regulations may require nutritional therapy as recommended by specialist consultants.

**SECTION VIII. SEVERABILITY**

If any provision of these Rules, or application thereof to any person or circumstance is held invalid, such invalidity shall not affect other provisions or applications of these Rules which give effect without the invalid provisions or applications, and to this end the provisions here to are declared to be severable.

**SECTION IX. REPEAL**

All Rules and parts of Rules in conflict here with are hereby repealed.

## CERTIFICATION

This will certify the foregoing Rules Pertaining to Newborn Screening were adopted by the Arkansas State Board of Health at a regular session of the Board held in Arkansas on the 24<sup>th</sup> day of October, 2019. The effective date of this rule shall be \_\_\_\_\_.

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Nathaniel Smith, MD, MPH  
Secretary of Health  
Secretary, Arkansas State Board of Health



# ARKANSAS STATE BOARD OF HEALTH

## RULES PERTAINING TO TESTING OF NEWBORN INFANTS



Promulgated Under the Authority of  
Ark. Code Ann. § 20-15-301 et  
seq., and Act 58 of 2019

*Effective on*

Arkansas Department of Health  
Nathaniel Smith, MD, MPH,  
Secretary of Health

RULES AND REGULATIONS PERTAINING TO TESTING OF NEWBORN  
INFANTS

Table of Contents

I. DEFINITIONS.....	2
II. PURPOSE.....	4
III. AUTHORITY.....	5
IV. RESPONSIBILITY.....	5
V. SPECIMEN COLLECTION AND SUBMISSION.....	7
VI. ANALYSIS, INTERPRETATION, AND REPORTING OF RESULTS.....	8
VII. ARKANSAS DEPARTMENT OF HEALTH ROLE IN TREATMENT AND MONITORING.....	10
VIII. SEVERABILITY.....	11
IX. REPEAL.....	11

**SECTION I. DEFINITIONS.**

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B. **Sickle Cell Disease (SS)** is the most common inherited abnormality of a red blood cell protein called hemoglobin. It is caused by a genetic abnormality that must be inherited from both parents. Sickle Cell Disease may cause serious health problems even in the first few months of life. It occurs much more commonly in people of African American, Asian and Mediterranean descent. In addition to the anemia, it lowers resistance to infection, and is associated with increased morbidity and mortality unless diagnosed and treated early. Sickle Cell Disease is one of a handful of related hemoglobinopathies each of which can cause similar health problems with varying severity.

C. **Sickle Cell Trait (AS)** and other hemoglobinopathy traits differ from their corresponding diseases. Traits occur when the genetic abnormality is inherited from only one parent, the other parent contributing a normal gene. Hemoglobinopathy traits cause only minor health issues that show up occasionally in life. They are associated with normal life spans. Sickle Cell Trait (AS) is the most common, occurring in 8 to 10% of African Americans.

D. **Biotinidase Deficiency (BIOT)** is caused by the lack of an enzyme called biotinidase, resulting in an inability of the body to use Vitamin B substances absorbed by the intestines. Without sufficient biotin, several other critical enzyme systems are unable to function properly. Biotinidase deficiency can lead to seizures, developmental delay, skin rash, and hearing loss. Newborns with the disorder appear normal, but develop critical symptoms after the first weeks or months of life. Symptoms include floppiness, seizures, developmental delay, hair loss, rashes, hearing loss and vision loss. Metabolic acidosis can result in coma and death. A daily biotin dietary supplement can prevent all symptoms.

E. **Congenital Adrenal Hyperplasia (CAH)** is a group of disorders caused by the deficiency of an adrenal enzyme resulting in decreased production of important hormones called cortisol and aldosterone. Cortisol helps the body respond to stressful events. Aldosterone helps the body maintain its fluids and salts. Without enough of these hormones, the affected newborn may appear normal, but can quickly develop symptoms including lethargy, vomiting, muscle weakness and dehydration. In severe cases death may occur within a few weeks if left untreated. One kind of CAH may show up first as ambiguous genitalia in the newborn. Infants with milder forms of the disorder are still at risk for reproductive and growth difficulties. If detected early and maintained on appropriate doses of medication, infants diagnosed with CAH can have normal growth and development.

F. **Cystic Fibrosis (CF)** is a disorder in which the body cannot make an important protein involved in using chloride ions, an ingredient in table salt. The major clinical consequences are the production of abnormally thickened mucous secretions in the lungs and digestive systems of affected newborns. With early detection and lifelong comprehensive treatment plans, infants diagnosed with CF can be expected to live longer and in a better state of health than in the past.

G. **Amino Acid Disorders** make up a group of inherited conditions in which protein metabolism is disrupted. Onset of symptoms may occur shortly after birth or after an apparently normal neonatal period. The symptoms may occur in episodes with normal periods in between. The clinical onset may include unusual odors in the urine, irritability, poor feeding, changes in muscle tone, lightened pigmentation, failure to thrive, jaundice, or liver enlargement. Other symptoms include intoxication-like symptoms such as vomiting, lethargy, seizures, and coma. Treatment of amino acid metabolism disorders includes a low-protein diet strictly controlling intake of specific amino acids.

H. **Fatty Acid Oxidation Disorders** make up a group of genetic metabolic deficiencies in which the body is unable to oxidize (break down) fatty acids to make energy. An enzyme is either missing or not working correctly. The main source of energy for the body is a sugar called glucose. Normally when the glucose runs out, fat is broken down into energy. However, that energy is not readily available to children and adults with a fatty acid oxidation disorder. If undiagnosed and untreated, these disorders can lead to serious complications affecting the liver, heart, and eyes; general muscle development; and possibly death. Symptoms of a metabolic “crisis” include vomiting, diarrhea, lethargy and difficulty breathing.

I **Organic Acid Disorders** make up a group of inherited metabolic diseases that lead to accumulation of organic acids in biological fluids (e.g., blood and urine). The accumulation produces disturbances in the acidity of the body and causes alterations in metabolic chemical reactions. These disorders can cause intoxication-like symptoms such as vomiting, metabolic acidosis, ketosis, dehydration, or coma. Some patients may have too little sugar in the blood, or too much lactic acid or ammonia. Chronic symptoms include recurrent vomiting, failure to thrive, floppiness and general developmental delay. Symptoms of these disorders can be diminished by restricting protein in the diet and, in some cases, supplementation with vitamins or a nutrient called carnitine.

J. **Severe Combined Immunodeficiency (SCID)** is a group of disorders characterized by severe defects in the T-lymphocyte and B-lymphocyte systems. Affected babies are susceptible to multiple types of life-threatening bacterial, viral, and fungal infections. Early diagnosis of SCID is imperative as SCID is curable with hematopoietic stem cell transplantation. Infants with SCID die of infections by age two (2) years unless immunity is reconstituted by treatment. SCID is commonly known as the “bubble boy” disease.

K. The **Collector** is the person or party responsible for collecting and submitting the blood specimen for testing. The persons or parties who are collectors under these Rules and Regulations are described in Section IV.A.

L. **The Department** is the Arkansas Department of Health.

M. **Spinal muscular atrophy (SMA)** is a genetic disease affecting the central nervous system, peripheral nervous system, and voluntary muscle movement (skeletal muscle). Most of the nerve cells that control muscles are located in the spinal cord, which accounts for the word spinal in the name of the disease.

N. **Pompe disease** is an inherited disorder caused by the buildup of a complex sugar called glycogen in the body’s cells. The accumulation of glycogen in certain organs and tissues, especially muscles, impairs their ability to function normally.

O. **Mucopolysaccharidosis (MPS1)** refers to a group of inherited conditions in which the body is unable to properly breakdown mucopolysaccharides (long chains of sugar molecules that are found throughout the body). As a result, these sugars buildup in cells, blood and connective tissue which can lead to a variety of health problems.

P. **Adrenoleukodystrophy (X-ALD)** is a disease linked to the X chromosome. It is a result of

fatty acid buildup caused by the relevant enzymes not functioning properly, which then causes damage to the myelin sheath of the nerves, resulting in seizures and hyperactivity. Other symptoms include problems with speaking, listening, and understanding verbal instructions.

## SECTION II. PURPOSE.

The purpose of these Rules ~~and Regulations~~ is to assure that all infants born in Arkansas have the opportunity to be screened for genetic illnesses.

These Rules ~~and Regulations~~ provide a method to assure that:

1. All newborn infants are tested for Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia, Sickle Cell Disease (SS), Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, ~~and~~ Severe Combined Immunodeficiency (SCID), Spinal Muscular Atrophy (SMA), Pompe Disease, MPS 1 spectrum of disease, and childhood onset (cerebral) X-ALD.
2. All newborns with an abnormal screen receive appropriate medical follow-up.

## SECTION III. AUTHORITY.

These Rules ~~and Regulations~~ are promulgated pursuant to the authority conferred by Arkansas Code Annotated § 20-15-301 et seq. and Act 58 of 2019.

## SECTION IV. RESPONSIBILITY.

- A. Collection and Submission.
  1. Medical Facilities/Medical Staff: In all cases where the birth of an infant occurs in a medical facility licensed by the Board of Health, it shall be the responsibility of the governing body and medical staff of the facility to adopt and enforce policies and procedures which ensures that blood test for Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia, Sickle Cell Disease (SS), Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, ~~and~~ Severe Combined Immunodeficiency (SCID), Spinal Muscular Atrophy (SMA), Pompe Disease, MPS 1 spectrum of disease, and childhood onset (cerebral) X-ALD are conducted and processed in accordance with these rules ~~and regulations~~. The licensed facility shall also be responsible for submission of the usable blood specimen in cases where an infant less than six months of age is admitted (i.e., born out of hospital, neonatal transfer, etc.), and it is brought to the attention of the facility or the attending physician that the infant is untested. If an infant is discharged from a licensed medical facility without collection and submission of a usable specimen for testing, it shall be the responsibility

of the discharging facility and the attending physician to arrange for the testing. The discharging facility and attending physician shall notify the Arkansas Department of Health ("Department") within one week of discharge if their efforts fail to arrange for testing.

2. Physicians: Physicians assuming care of infants who are under six months of age and who come to their attention as being untested or inadequately tested for Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia, Sickle Cell Disease (SS), Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis(CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, and Severe Combined Immunodeficiency (SCID), Spinal Muscular Atrophy (SMA), Pompe Disease, MPS 1 spectrum of disease, and childhood onset (cerebral) X-ALD, shall also be responsible for assuring collection and submission of usable blood specimens for these infants.
3. Licensed Midwives: In cases where the birth occurs outside a licensed medical facility or in the home, it shall be the responsibility of an attending licensed midwife to advise the parents of this law and the procedure for conducting newborn screening, and documenting that a blood sample is obtained after 24 hours and no later than 72 hours after birth. If the blood sample is not obtained for any reason, an attending licensed midwife must document the incident in the patient's chart.
4. The Department: The Department's Local Health Unit shall collect and submit usable blood specimens on all infants under six months of age who come to their attention as being tested or inadequately tested. This responsibility shall not be in lieu of that of the preceding individuals and facilities.

B. Payment

1. The Collector will be charged a fee of one hundred and ~~twenty-one~~ thirty-one dollars (~~\$121.00~~) (\$131.00) for the processing and testing of newborn screening specimens by the Department.
2. The Board of Health may determine the amount of this fee based on the Department's cost to process and test the specimens.

C. Laboratory Analysis

1. The Department shall be responsible for provision of forms and instructions for the blood specimen collection; processing and recording of the specimen received; analysis of specimen; determination of abnormal results; and reporting of lab results within a time period which would allow preventive medical intervention.

D. Follow-Up

1. The Department shall be responsible for the interpretation of laboratory results and the reporting of abnormal results to the attending physician or birth attendant. If the screening result is suggestive of Classical or Variant

PKU, Galactosemia, Sickle Cell Disease (SS), Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, or Severe Combined Immunodeficiency (SCID), Spinal Muscular Atrophy (SMA), Pompe Disease, MPS I spectrum of disease, or childhood onset (cerebral) X-ALD, the Department shall consult with specialist physicians who are retained by contract to provide clinical advice on these conditions. The Department shall notify the Collector of the specimen and enter the infant's information in a tracking system maintained to evaluate program operations and infants' medical outcomes.

2. Attending Physician/Medical Attendant:

- a) Upon receipt of a notice of an abnormal test result the physician or medical attendant shall be responsible for the appropriate medical treatment, referral, and/or retesting within the timeframe specified by the Department for that particular disorder. It is strongly recommended that consultation be obtained with a physician who has special competence in the management of these disorders.
- b) The attending physician or other responsible health care provider who conducts testing in follow-up to abnormal screens shall report subsequent test results (whether negative or positive) to the Department. To provide for long term follow up the Department will collect data on affected infants each year for five years to determining health care maintenance and health status, especially the presence of mental retardation or permanent disability.

The Department will establish protocols for follow-up of all screened disorders in collaboration with medical specialists under contract. For infants with abnormal test results, the physician will be notified of the results and informed of the recommended protocols for follow-up of the specific order.

## **SECTION V. SPECIMEN COLLECTION AND SUBMISSION**

- A. The blood specimen for PKU, CH, Galactosemia, Sickle Cell Anemia, Biotinidase (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, or Severe Combined Immunodeficiency (SCID), or Spinal Muscular Atrophy (SMA), Pompe Disease, MPS I spectrum of disease, and childhood onset (cerebral) X-ALD testing must be collected and submitted as described below:
- B. Timing of Specimen Collection
  1. For all healthy infants born in medical facilities, the specimen shall be collected before the time of discharge from the facility. Optimum time for collection is 24 to 72 hours after birth, and all Collectors should strive to comply with that time frame. If any infant is discharged or specimen



collected prior to 24 hours of age, a repeat test shall be arranged by the medical facility and the attending physician. This repeat specimen shall be collected by the infant's seventh day of life. A repeat test for Sickle Cell Disease shall not be required if specimen was collected prior to 24 hours of age.

2. Specimens from ill or premature infants shall be obtained as soon as possible after their condition has sufficiently stabilized.
3. Specimens from infants not born in medical facilities shall be collected between 24 and 72 hours after birth.

Infants under six months of age who are known to be untested or inadequately tested shall have blood specimens collected and submitted by the responsible authority as soon as possible.

C. Specimen Collection and Submission

1. Specimens shall be dispatched to the Arkansas Department of Health Public Health Laboratories, Little Rock, Arkansas, no later than one (1) business day from collection. Specimens are submitted only on forms provided by the Public Health Laboratory. The Collector is responsible for supplying complete and accurate identifying information on the collection form to be used for tracking infants with abnormal screening results.

D. Forms

1. Submission: forms may be obtained by writing to the Public Health Laboratories at:

Arkansas Department of Health  
201 South Monroe Street  
Little Rock, AR 72205

The county health units will not supply these forms.

E. Unsatisfactory Specimens

1. Inadequate, contaminated, or otherwise unusable specimens shall be reported to the Collector after laboratory determination of an unsatisfactory specimen. The Collector shall be responsible for assuring recollection and resubmission within seven calendar days of notification.

## **SECTION VI. ANALYSIS, INTERPRETATION, AND REPORTING OF RESULTS**

A. Laboratory Analysis

1. All specimens received by the laboratory shall be initially examined within five working days of receipt. Abnormal results shall be reported to the Collector within two working days of determination.

B. Interpretations of Results

1. Phenylketonuria (PKU)

- a) The Department shall define the phenylalanine level which constitutes a positive screening result for PKU.
  - b) An infant whose phenylalanine level is determined by the Department to be negative for PKU requires no action to be taken. However, attending physicians shall give special consideration when testing circumstances or infant evaluation/family history suggests the possibility of need for prescreening in cases where PKU or PKU variants may actually exist in spite of initial negative screening results.
2. Congenital Hypothyroidism (CH)
    - a) The Department shall define the thyroxine and thyroid stimulating hormone levels which constitute positive screening results for CH.
    - b) Occasionally test results suggestive of CH may be reported which, upon retesting, will be found within normal limits. Likewise it is possible that test results which are reported as normal in the neonatal period could mask the delayed onset of CH. While an infrequent occurrence, in the face of clinical findings, this possibility must be considered by the attending physician.
3. Galactosemia
    - a) The Department shall define the galactose-1-phosphate uridylyl transferase (GALT) levels which constitute positive screening results for Galactosemia.
    - b) It is possible that an infant affected with Galactosemia could have normal initial screening results. This situation is most likely to occur in infants who have received no or insufficient feedings with lactose-containing milk or formula prior to testing, or who have received blood transfusions prior to testing.
    - c) The medical caretaker shall give special consideration to retesting any infant whose case findings, testing circumstances, or family history seems to medically warrant it.
4. Sickle Cell Anemia or Trait
    - a) The Department shall define the laboratory value which constitutes a positive screening result for Sickle Cell Disease (SS), Sickle Cell Trait (AS) or other related hemoglobinopathy.
    - b) An infant whose hemoglobin is determined by the Department to be negative for SS or other related serious hemoglobinopathies requires no special consultation; however, infants with trait conditions should be followed for mild anemias and urinary tract infections.
    - c) The medical caretaker shall give special consideration to re-testing any infant whose case findings, testing circumstances, or family history seems to medically warrant it.
5. Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia

(CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxygenation Disorders, Organic Acid Disorders, Severe Combined Immunodeficiency (SCID).

- a) The Department shall define the laboratory value which constitutes a positive screening result for Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders Severe Combined Immunodeficiency (SCID).

C. Reporting of Results

- 1. Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders Severe Combined Immunodeficiency (SCID).
  - a) Immediately upon obtaining the initial positive screening result, the Department shall notify the attending physician or medical attendant, who shall be responsible for ensuring that prompt follow-up diagnostic testing is conducted.
  - b) Appropriate, expectant medical management shall not be withheld pending the confirmatory test results. A non-physician Collector shall immediately refer the infant for appropriate medical intervention. It is recommended that a pediatric geneticist, endocrinologist, or pulmonologist consultant be utilized in the management of these infants.
- 2. Sickle Cell Disease (SS) and other serious Hemoglobinopathies
  - a) Immediately upon obtaining the initial positive screening result, presumptive of SS or other serious hemoglobinopathy, the Department shall notify the Collector, who shall be responsible for insuring that prompt follow-up diagnostic testing is conducted.
  - b) Appropriate, expectant medical management shall not be withheld pending the confirmatory test results for either SS or other related hemoglobinopathy. Therefore, non-physician Collector shall immediately refer the infant for appropriate medical intervention. It is recommended that a pediatric hematologist consultant be utilized in the management of these infants.
  - c) Immediately upon obtaining an initial positive screening, presumptive of trait, the Department shall notify the Collector in writing. The parent shall be notified in writing by the Department.

**SECTION VII. ARKANSAS DEPARTMENT OF HEALTH ROLE IN TREATMENT AND MONITORING**

- A. Listing of Consultants
  - 1. For Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemia, Sickle Cell Disease and other hemoglobinopathies, Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, Severe Combined Immunodeficiency (SCID), the Department shall maintain a list of pediatric consultants having special competence in these disorders, and shall make the names of such consultants known to the attending physicians of infants with abnormal screening test results.
  
- B. Registry
  - 1. For Phenylketonuria (PKU), Congenital Hypothyroidism (CH), Galactosemias, Sickle Cell Disease (SS), and other hemoglobinopathies, Biotinidase Deficiency (BIOT), Congenital Adrenal Hyperplasia (CAH), Cystic Fibrosis (CF), Amino Acid Disorders, Fatty Acid Oxidation Disorders, Organic Acid Disorders, Severe Combined Immunodeficiency (SCID), the Department shall maintain a registry to record laboratory results and diagnoses of all tested infants, and to track referral for those infants in whom abnormal findings were noted during the screening process.
  
- C. Nutritional Therapy
  - 1. Phenylketonuria (PKU)
    - a) Nutritional therapy with low phenylalanine formula and/or foods shall be instituted after the diagnosis of PKU.
  
  - 2. Galactosemia
    - a) Nutritional therapy with lactose-free formula and/or foods shall be instituted after the diagnosis of Galactosemia.
  
  - 3. Other genetic conditions
    - a) Other genetic conditions discovered by the laboratory testing done pursuant to these regulations may require nutritional therapy as recommended by specialist consultants.

**SECTION VIII. SEVERABILITY**

If any provision of these Rules ~~and Regulations~~, or application thereof to any person or circumstance is held invalid, such invalidity shall not affect other provisions or applications of these Rules ~~and Regulations~~ which give effect without the invalid provisions or applications, and to this end the provisions here to are declared to be severable.

**SECTION IX. REPEAL**

All ~~Rules Regulations~~ and parts of ~~Rules Regulations~~ in conflict here with are hereby repealed.

## CERTIFICATION

This will certify the foregoing Rules and Regulations Pertaining to Newborn Screening were adopted by the Arkansas State Board of Health at a regular session of the Board held in Paris, Arkansas on the 24<sup>th</sup> day of October, 2019 ~~22nd day of January, 2015~~. The effective date of this rule shall be May 1, 2015.

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Nathaniel Smith, MD, MPH  
Secretary of Health  
Secretary, Arkansas State Board of Health

**Arkansas Department of Health  
Rules Pertaining to Testing of Newborn Infants  
Summary of Public Comments**

1. Commenter's Name: *Meredith Woodruff*  
Commenter's Business/Agency: *parent advocate for SMA*  
  
Summary of Comment: *Lost infant to SMA. Respectfully asks ADH to adopt the proposed rule and quickly begin screening*  
  
Agency's response to Comment: *Thank you for your comments.*  
  
Were any changes made to the Proposed Rule as a result of this Comment? If so, please describe. N/A
  
2. Commenter's Name: *Aravindhyan Veerapandiyan, MD*  
Commenter's Business/Agency: *Neurology, Arkansas Children's Hospital*  
Summary of Comment: *Requests swift action by ADH to adopt these rules and begin screening for SMA*  
  
Agency's response to Comment: *Thank you for your comments.*  
  
Were any changes made to the Proposed Rule as a result of this Comment? If so, please describe. N/A
  
3. Commenter's Name: *Zdenek Berger, Ph.D.*  
Commenter's Business/Agency: *medical director, Biogen*  
  
Summary of Comment: *Supports proposed changes*  
  
Agency's response to Comment: *Thank you for your comments*  
  
Were any changes made to the Proposed Rule as a result of this Comment? If so, please describe. N/A
  
4. Commenter's Name: *Kenneth Hobby*  
Commenter's Business/Agency: *President, Cure SMA*  
Summary of Comment: *Requests swift action by ADH to adopt these rules and begin screening for SMA*  
  
Agency's response to Comment: *Thank you for your comments.*  
  
Were any changes made to the Proposed Rule as a result of this Comment? If so, please describe. N/A

## **DEPARTMENT OF HEALTH, PUBLIC HEALTH LAB**

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**SUBJECT:** Rules Pertaining to Testing of Newborn Infants

**DESCRIPTION:** The proposed amendments to the Rules Pertaining to Testing for Newborn Infants add tests for newborn screening including for Spinal Muscular Atrophy, pursuant to Act 58 of 2019. The proposed amendments to the Rules also add three additional tests for Pompe Disease, MPS 1 spectrum of disease, and childhood onset (cerebral) X-ALD.

At the present time, 31 tests are performed on newborns, and 29 of the tests are performed in the Public Health Laboratory. The tests are performed on dried blood spots that are collected from the infants soon after birth. Of the four additional tests, three of them have FDA-approved procedures, while Spinal Muscular Atrophy does not at this time. The four tests can be performed in our local health laboratory utilizing existing blood samples without having to require new blood samples from the hospital.

The current fee is \$121.00 and the proposed amendment adds an additional \$10.00 charge for the Spinal Muscular Atrophy test.

**PUBLIC COMMENT:** A public hearing was held on this rule on February 12, 2020. The public comment period expired on February 12, 2020. The agency provided the following summary of the public comments it received and its responses to those comments:

Written comments were received from four individuals representing families, advocacy groups and medical specialists. Each individual provided letters of support for adoption of the proposed rules and swift action by the Arkansas Department of Health officials to begin screening all newborns born in the state of Arkansas.

**Commenter's Name:** Meredith Woodruff, parent of child affected by SMA and Cure SMA Supporter

**COMMENT:** Mrs. Woodruff respectfully asks that the Arkansas Department of Health adopt the proposed rule and quickly begin screening Arkansas newborns for SMA. She states the addition of SMA to the proposed rules will have an enormous impact for families across Arkansas. **RESPONSE:** The Arkansas Department of Health thanks these individuals for their comments and letters of support for the proposed changes to the Rules Pertaining to Testing of Newborn Infants.

**Commenter's Name:** Dr. Aravindhan Veerapandiyan, Pediatric Neurologist at Arkansas Children's Hospital

**COMMENT:** Dr. Veerapandiyan supports the adoption of these rules and would like to see swift action by the Arkansas Department of Health to join the growing number of states who screen for SMA. Early diagnosis and treatment of spinal muscular atrophy



can lead to improved, long-lasting developmental outcomes for individuals living with SMA. Clinical data shows treatments are most effective when delivered early and pre-symptomatically. **RESPONSE:** The Arkansas Department of Health thanks these individuals for their comments and letters of support for the proposed changes to the Rules Pertaining to Testing of Newborn Infants.

**Commenter's Name:** Dr. Zdenek Berger, Medical Director, Late Stage Neuromuscular and Rare Disease, Biogen

**COMMENT:** Dr. Berger supports the proposed changes to Rules with specific backing for the elements adding spinal muscular atrophy to the state's newborn screening panel. Now that treatment options are available data indicate that initiation of treatment in the pre-symptomatic period improves clinical outcomes in newborns. **RESPONSE:** The Arkansas Department of Health thanks these individuals for their comments and letters of support for the proposed changes to the Rules Pertaining to Testing of Newborn Infants.

**Commenter's Name:** Kenneth Hobby, President, Cure SMA

**COMMENT:** Mr. Hobby and Cure SMA fully supports the revisions to the newborn screening rule to add SMA. He states that the rulemaking to add SMA to the state's newborn screening program comes at a critical and promising time for the SMA community. Treatments are available however, promising outcomes are only possible with early diagnosis and timely treatment. Cure SMA and the entire Arkansas SMA community strongly supports finalizing the proposed newborn screening rule change. **RESPONSE:** The Arkansas Department of Health thanks these individuals for their comments and letters of support for the proposed changes to the Rules Pertaining to Testing of Newborn Infants.

Lacey Johnson, an attorney with the Bureau of Legislative Research, asked the following question and received the following response:

**QUESTION:** Where do the definitions of Pompe disease, MPS1, and X-ALD come from? **ANSWER:** The assistant director at the Public Health Laboratory responded that the definitions are consistent with standard medical literature.

The proposed effective date is pending legislative review and approval.

**FINANCIAL IMPACT:** The agency indicated that this rule has a financial impact.

According to the agency, which stated that it received a cost estimate from the Department of Human Services, Division of Medical Services, the additional cost of the state rule for the current fiscal year will be \$32,421 in general revenue and \$80,114 in federal funds, for a total of \$112,535. The estimated cost for the next fiscal year will be \$63,987 in general revenue and \$161,083 in federal funds, for a total of \$225,070.

The agency estimated the total estimated cost by fiscal year to state, county, and municipal government to implement this rule at \$32,421 for the current fiscal year and \$63,987 for the next fiscal year. Per the agency, these estimates come from the Department of Human Services, Division of Medical Services.

The agency indicated that there is a new or increased cost or obligation of at least \$100,000 per year to a private individual, private entity, private business, state government, county government, municipal government, or two or more of those entities combined. It stated that Act 58 of 2019 requires insurers to cover testing for spinal muscular atrophy. Per the agency, testing currently costs \$121 per sample and this rule would increase what ADH Public Health Lab charges hospitals by \$10 for all four tests. The agency provided the following written findings:

*(1) a statement of the rule's basis and purpose*

To comply with Act 58 of 2019 and detect disorders early for treatment

*(2) the problem the agency seeks to address with the proposed rule, including a statement of whether a rule is required by statute*

Rule required by statute. Disorders need to be detected soon after birth for treatment to be most effective.

*(3) a description of the factual evidence that*

*(a) justifies the agency's need for the proposed rule; and*

*(b) describes how the benefits of the rule meet the relevant statutory objectives and justify the rule's costs*

Early detection for the four conditions decreases medical costs as early treatment is more effective.

*(4) a list of less costly alternatives to the proposed rule and the reasons why the alternatives do not adequately address the problem to be solved by the proposed rule*

There are no alternatives.

*(5) a list of alternatives to the proposed rule that were suggested as a result of public comment and the reasons why the alternatives do not adequately address the problem to be solved by the proposed rule*

There are no proposed alternatives.

*(6) a statement of whether existing rules have created or contributed to the problem the agency seeks to address with the proposed rule and, if existing rules have created or contributed to the problem, an explanation of why amendment or repeal of the rule creating or contributing to the problem is not a sufficient response*

Previous newborn screenings did not include these tests.

*(7) an agency plan for review of the rule no less than every ten years to determine whether, based upon the evidence, there remains a need for the rule including, without limitation, whether:*

- (a) the rule is achieving the statutory objectives;*
- (b) the benefits of the rule continue to justify its costs; and*
- (c) the rule can be amended or repealed to reduce costs while continuing to achieve the statutory objectives.*

ADH constantly monitors CDC guidelines, state and federal laws and regulations for opportunities to reduce and control costs.

**LEGAL AUTHORIZATION:** The Department has authority to prescribe tests to be administered to newborn infants. *See* Ark. Code Ann. § 20-15-304(2). It also has authority to promulgate rules relating to “[w]hat persons and institutions shall be required to obtain specimens from newborn infants . . . ; [t]he amount to be charged by the central laboratory for processing the specimens; and [t]he method of billing the charges to the persons and institutions[.]” Ark. Code Ann. § 20-15-304(3)(A)-(C).

Some of these rule changes implement Act 58 of 2019, sponsored by Representative Julie Mayberry, which required newborn screening for spinal muscular atrophy and mandated that insurance policies cover newborn screening for spinal muscular atrophy.

Along with spinal muscular atrophy, the Department of Health is specifically responsible for administering newborn testing for phenylketonuria, hypothyroidism, galactosemia, cystic fibrosis, and sickle-cell anemia. *See* Ark. Code Ann. § 20-15-302(a)(1)(A), as amended by Act 58, § 1 (2019); Ark. Code Ann. § 20-15-304. “In addition, if reliable and efficient testing techniques are available, all newborn infants shall be tested for other genetic disorders by employing procedures approved by the State Board of Health.” Ark. Code Ann. § 20-15-302(a)(1)(B). Per the agency, Pompe disease, MPS 1, and X-ALD all have FDA-approved testing procedures.