

Regulation 61-80

Neonatal Screening For Inborn Metabolic Errors and Hemoglobinopathies

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S.C. Department of Health and
Environmental Control

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Section A – Purpose and Scope

This regulation establishes rules implementing provisions of Section 44-37-30 of the South Carolina Code of Laws, 1976, as amended, regarding testing of newborn children for inborn metabolic errors and hemoglobinopathies. The Department of Health and Environmental Control has been given the legislative mandate to promulgate rules and regulations for screening for inborn metabolic errors and hemoglobinopathies and to ensure compliance with the screening of every child born in South Carolina. The responsibilities of the various agencies, institutions and persons involved in the screening process are defined. Procedures for storage and use of blood specimens and maintenance of confidentiality are included.

Section B – Definitions

1. Inborn Metabolic Errors—shall mean inborn errors of metabolism.
2. Hemoglobinopathy—shall mean a hematologic disorder or carrier state caused by alteration in the genetically determined molecular structure of hemoglobin which may result in overt anemia as well as clinical and other laboratory abnormalities.
3. Identifying Information—shall mean child’s legal name, sex, race, birth date, time of birth, place of birth, birth weight, current weight, feeding type; parent’s or legal guardian’s complete name, complete address and telephone number; mother’s Social Security Number.
4. Attending Physician—shall mean the physician who has entered into an agreement to provide care during and/or after delivery for the mother and/or her child. The physician listed on the laboratory form will be assumed to be the attending physician until notification to the contrary is received in accordance with Official Departmental Instructions.
5. Department—shall mean the South Carolina Department of Health and Environmental Control.
6. Laboratory—shall mean the South Carolina Department of Health and Environmental Control Bureau of Laboratories.
7. Bureau of Maternal and Child Health—shall mean an organizational unit of the South Carolina Department of Health and Environmental Control.
8. Official Departmental Instructions—shall mean detailed instructions approved by the Commissioner of the South Carolina Department of Health and Environmental Control or his designee under which the public and private health care providers, including hospitals, laboratories, clinics, physicians and their staffs screen all children born in South Carolina for designated Inborn Metabolic Errors and Hemoglobinopathies.

Section C – Testing

1. The Laboratory shall perform all screening tests for inborn metabolic errors and hemoglobinopathies using procedures compliant with the Clinical Laboratories Improvement Act of 1988, as amended, and approved by the Food and Drug Administration. If any result is abnormal, the appropriate test shall be repeated and confirmatory tests performed in accordance with Official Departmental Instructions.
2. The Laboratory, in conjunction with the Bureau of Maternal and Child Health, shall adopt standards for the quality assurance and interpretation of approved tests and for the collection of specimens.

3. Confirmation and repeat specimen testing are available from the Laboratory at no charge to patients suspected or diagnosed as having one of the diseases if the analysis is completed at the Laboratory.

4. Test results and identifying information are to be reported and recorded in accordance with Official Departmental Instructions.

Section D – Collection of Specimen

1. A specimen shall be collected from every child born in South Carolina for the purpose of screening for inborn metabolic errors and hemoglobinopathies.

2. Births in a Hospital

a. The attending physician is responsible for the collection of the specimen from every child born in the hospital in accordance with Official Departmental Instructions and is responsible for submission of the specimen to the Laboratory on the day of collection.

b. Under the direction of the attending physician, the specimen shall be collected under the most favorable conditions following the procedures specified in the Official Departmental Instructions. The brochure produced by the Department that explains newborn screening for inborn metabolic errors and hemoglobinopathies and blood specimen storage options shall be given to the parent or legal guardian of the child.

c. A specimen shall be collected from every child born in the hospital prior to release from the hospital (except when the parents object due to religious convictions) in accordance with the procedure specified in the Official Departmental Instructions. If the parent objects to the screening on the basis of religious convictions, the parent shall complete the procedure specified in the Official Departmental Instructions.

d. If for some reason the specimen is not collected at the hospital, the hospital shall then be responsible for notifying the Bureau of Maternal and Child Health as specified in the Official Departmental Instructions.

e. The Hospital shall review the patient record for each child born in the hospital no later than ten (10) days after delivery to ensure that a specimen was collected and submitted to the Laboratory.

3. Births Outside a Hospital

a. The attending physician is responsible for the collection of the specimen from every child in accordance with the Official Departmental Instructions and for submission of the specimen to the Laboratory on the day of collection.

b. Under the direction of the attending physician, the specimen shall be collected under the most favorable conditions following the procedure specified in the Official Departmental Instructions. The brochure produced by the Department that explains newborn screening for inborn metabolic errors and hemoglobinopathies and blood specimen storage options shall be given to the parent or legal guardian of the child.

c. If the parents object to the screening on the basis of religious convictions, the parents shall complete the procedure specified in the Official Departmental Instructions.

d. If for some reason the specimen is not collected within three (3) days of delivery by the attending physician, this physician shall notify the Bureau of Maternal and Child Health as specified in the Official Departmental Instructions.

e. If there is not an attending physician, then the person in attendance is responsible for the collection of the specimen. If there is no other person in attendance, then the parents or legal guardian shall notify the Health Department in the county in which the child resides within three (3) days of delivery so that a specimen may be collected.

Section E – Assurance of Diagnosis and Follow-up

1. Information obtained as a result of the tests conducted for screening for inborn metabolic errors and hemoglobinopathies is confidential and may be released only to the infant’s physician or other staff acting under the direction of the physician, the child’s parent or legal guardian, and the child when he/she is eighteen years of age or older.

2. Normal and abnormal test results will be forwarded by the Laboratory and/or Bureau of Maternal and Child Health to the attending physician who shall be responsible for informing the parents or legal guardian of test results.

3. If the child is not under the care of the attending physician, as specified in the Official Departmental Instructions, the person in attendance shall notify the Bureau of Maternal and Child Health. The Department will then notify the parents or legal guardian of the test results.

4. Upon notification that a specimen was insufficient or that it is necessary for a test to be repeated, the attending physician shall collect and submit a second specimen to the Laboratory in accordance with Official Departmental Instructions.

5. The attending physician shall initiate appropriate medical follow-up and diagnosis when abnormal test results occur. If that is not possible, the Bureau of Maternal and Child Health shall be notified as specified in the Official Departmental Instructions.

6. The attending physician shall notify the Bureau of Maternal and Child Health of all children born in South Carolina who are diagnosed as having inborn metabolic errors or hemoglobinopathies.

7. Appropriate genetic counseling should be offered to all families of children with abnormal test results as outlined in the Official Departmental Instructions.

Section F – Storage of Specimen

1. Hospital staff or other persons who collect blood specimens for the purpose of screening for inborn metabolic errors and hemoglobinopathies shall inform each child’s parent or legal guardian of the blood specimen storage options.

2. Hospital staff or other persons who collect these blood specimens shall give the brochure produced by the Department that explains newborn screening for inborn metabolic errors and hemoglobinopathies to the parent or legal guardian as a means of informing them of the benefits of screening and blood specimen storage. Hospital staff or other persons who collect these blood specimens shall indicate that the brochure was given to the parent or legal guardian by documenting in the appropriate space on the Blood Sample Storage Options Form.

3. The Laboratory shall store all specimens at minus 20° Centigrade and may release specimens for purposes of confidential, anonymous scientific study unless prohibited by the parents, legal guardians, or children from whom the specimens were obtained when the children are eighteen years of age or older.

4. Hospital staff or other persons who collect these specimens shall ensure that the parent's or legal guardian's storage choice is documented on the Blood Sample Storage Options form if the parent or legal guardian does not agree to have their child's blood specimen stored and potentially released for confidential, anonymous scientific study. In these instances, the Laboratory shall maintain all such specimens based upon the storage option chosen by the parent or legal guardian as documented on the Blood Sample Storage Options form.

Section G – Use of Stored Specimen

1. Stored blood specimens may be released for the purposes of confidential, anonymous scientific study unless prohibited by the parent, legal guardian, or child from whom the specimen was obtained when he/she is eighteen years of age or older.

2. The Department's Institutional Review Board shall approve all scientific studies that use stored blood specimens before the specimens are released.

3. Blood specimens released for scientific study shall not contain information that may be used to determine the identity of the children from whom they were obtained by the person(s) to whom the specimens are released. The Department shall code the specimens before releasing them so that the Department can identify the children from whom the blood specimens were obtained if necessary.

4. If any such scientific study identifies genetic or other information that may benefit the children from whom the specimens were obtained, the Department may confidentially provide this information to the parents, legal guardians or children from whom the specimens were obtained when the children are eighteen years of age or older.

Section H – Forms

1. Religious Objection Form: The Religious Objection Form, Appendix A of this regulation, shall be completed if the parents refuse newborn screening for inborn metabolic errors and hemoglobinopathies for their child based upon religious convictions.

2. Information Release Form: The Information Release Form, Appendix B of this regulation, may be completed as needed for release of information regarding newborn screening for inborn metabolic errors and hemoglobinopathies to persons other than those specified elsewhere in this regulation.

3. Blood Sample Storage Options Form: The Blood Sample Storage Options Form, Appendix C of this regulation, shall be completed if the parents or legal guardians do not agree to have their child's specimen stored and potentially released for confidential, anonymous scientific study.

Section I – Enforcement Provision

1. Constitutionality

If any part or provision of these regulations is legally declared unconstitutional or if the application thereof to any persons or circumstances is held invalid, the validity and constitutionality of the remainder of these regulations shall not be affected thereby.

2. Penalties

Violation of these regulations shall be punishable in accordance with Section 44-37-30 of the Code of Laws of South Carolina, 1976, as amended.

APPENDIX A: Religious Objection Form: DHEC 1804, Newborn Screening Program, Parental Statement of Religious Objection

I am the parent or legal guardian of _____, a child born _____ in South Carolina. I request that my child not be tested by blood spot screening in order to detect silent, deadly metabolic diseases and hemoglobinopathies. I certify that this refusal is based on religious grounds. Religious grounds are the only permitted reason for refusal under South Carolina law, Section 44-37-30 (C).

I understand that my child may suffer brain damage, other bodily harm or death if a disease that can be detected by blood spot screening is not diagnosed. I understand that such harm can be lessened or prevented by early diagnosis and treatment. I understand that these diseases are usually silent, and may be present in a child that looks healthy. I understand that the blood spot screening test is the best way to detect these disorders early, and that testing is routinely done for every child. I understand that this testing is quick, easy and that the results are confidential. I understand that this testing has been the standard of care for all children born in South Carolina and the rest of the United States for many years.

I have been fully informed of, and fully understand, the possible devastating consequences to my child's health if blood spot screening is not done. I have been fully informed of, and fully understand the benefits of testing and blood specimen storage. I have been given the brochure produced by the South Carolina Department of Health and Environmental Control that describes the conditions for which testing is currently available and explains the benefits of testing and blood specimen storage. I also understand that my child would have been tested for these conditions except for my objection. I have been given the opportunity to ask questions concerning this testing and these codifications, and all of my questions have been fully answered to my satisfaction.

I release and hold harmless the South Carolina Department of Health and Environmental Control, the hospital or other facility at which the birth occurred, the person(s) responsible for the collection of the blood spots, and any other person or entity relying on this objection, for any injury, illness and/or consequences, including the death of my child, which may result to my child as the result of my refusal of blood spot screening.

Parent: _____ Date: _____
Witness: _____

NOTE TO PROVIDERS: This form is only necessary if the parent or legal guardian refuses testing for inborn metabolic errors and hemoglobinopathies.

APPENDIX B: Information Release Form: DHEC 1878, Authorization to Release Information Relative to Newborn Screening for Inborn Metabolic Errors and Hemoglobinopathies

Please check all boxes that apply.

A. I agree that information about _____, born _____, obtained as a result of tests conducted for screening for inborn metabolic errors and hemoglobinopathies may be released or exchanged with the following providers:

B. In cases where this information is immediately needed for continuity of health care, I authorize the South Carolina Department of Health and Environmental Control to provide this information to the providers listed above by fax.

C. I authorize my signed form to be faxed to the providers listed above.

I understand that my confidentiality cannot be guaranteed when sending this information by fax. I understand that the copy of my signature below may be treated as an original signature.

I am the client, parent or legal guardian. I understand that I am responsible for this information if it is released to me and that my records are protected generally under state laws as well as statutes governing specific types of information and cannot be disclosed without my authorization. I also understand that I may revoke this authorization at any time except to the extent that action has been taken on it.

Signature: _____ Date: _____
Witness: _____ Date: _____
Revoked: _____ Date: _____

Some babies are born with diseases of the blood or body function. A baby with one of these diseases looks healthy. However, these diseases can cause intellectual disability, abnormal growth, infections, or death. Some of these diseases can be found by early testing. This testing, called newborn screening, is important so that your baby is not harmed by one of these diseases. During newborn screening, a small sample of your baby's blood is taken from the heel. The blood is tested. The blood shows if your baby has any of the "newborn screening" diseases. If your baby has one of these diseases, your doctor can treat your baby.

DHEC can store your baby's blood sample for special study. Studies help DHEC find out new information about diseases. If a study finds something in your child's blood sample that can help your child, DHEC can confidentially notify you (or your child if he/she is 18 years or older).

APPENDIX C: Blood Sample Storage Options Form: DHEC 1812, Blood Sample Storage Options, Screening for Inborn Metabolic Errors and Hemoglobinopathies

Child's complete legal name: _____
Child's date of birth: _____
Parent or legal guardian's complete name: _____
Parent or legal guardian's complete address: _____

South Carolina law requires the Department of Health and Environmental Control to store your child's blood sample in a manner required by law. The blood sample is collected on a special piece of filter paper. This is called "newborn screening." The blood is tested to see if your child has one of the "newborn screening" diseases that can cause intellectual disability, abnormal growth or even death. After the tests are done, the filter paper is stored in a freezer at the state laboratory. This storage is highly protected, and each sample is held under strict confidentiality. A child's blood sample can only be released for approved research, without any identifying information, to learn new information about diseases. The law allows you to choose one of the options below, if you do not want your child's blood sample handled this way. **However, you are not required to check one of the boxes below.**

I want my child's blood sample stored by the South Carolina Department of Health and Environmental Control, but I do not want my child's blood sample to be used for research.

I want my child's blood sample destroyed by the South Carolina Department of Health and Environmental Control two years after the date of testing.

I want my child's blood sample to be returned to me two years after the date of testing. I understand that it is my responsibility to notify the South Carolina Department of Health and Environmental Control, 2600 Bull Street, Columbia, SC, 29201, of address or name changes.

I have been given the brochure produced by the South Carolina Department of Health and Environmental Control that describes the conditions for which testing is currently available and explains the benefits of testing and blood sample storage.

Parent: _____ Date: _____

I have given the brochure produced by the South Carolina Department of Health and Environmental Control to the parent/legal guardian of the child named above.

Name: _____ Date: _____

DHEC can store your baby's blood sample for special study. Studies help DHEC find out new information about diseases. If a study finds something in your child's blood sample that can help your child, DHEC can confidentially notify you (or your child if he/she is 18 years or older).

IF THIS FORM IS NOT SIGNED BY A PARENT/LEGAL GUARDIAN AND/OR NONE OF THE ABOVE BOXES ARE CHECKED, THE BLOOD SAMPLE WILL BE STORED AS REQUIRED BY SC CODE ANN. SECTION 44-37-30 AT -20 DEGREES CENTIGRADE AND MAY BE RELEASED ONLY FOR CONFIDENTIAL, ANONYMOUS SCIENTIFIC STUDY.

NOTE TO PROVIDERS: The parent or legal guardian is not required to sign this form. However, the person who gives the brochure that explains neonatal testing and blood sample storage to the parent or legal guardian must sign this form.