Appendix

DHEC Newborn Screening Contact Information

Children's Health

Please call the staff listed below if you have any questions about newborn screening clinical issues or about follow-up services.

Medical Consultant: Eileen Walsh, MD.....803-898-0362
email: walshem@dhec.sc.gov
Follow-up Program Manager:
Kathy Tomashitis, MNS, RD, LD.....803-898-0619
email: tomashkf@dhec.sc.gov
Follow-up Program Nurse Coordinator, Patient Results:
Dana Smith, RN.....803-898-0593
email: smithdm@dhec.sc.gov
Follow-up Program Dietitian Coordinator, Patient Results, Metabolic Formula:
Jennifer Schlub, RD, LDN……803-898-1969
email: schlubjk@dhec.sc.gov

Fax number for all Children's Health staff.....803-898-0337

Mailing address for all Children's Health staff
Children's Health
(NAME)
SC DHEC
2100 Bull Street
Columbia, SC, 29201

Bureau of Laboratories

Please call the staff listed below if you have any questions about laboratory services.

Patient Results:..... 803-896-0298/803-896-0889 (fax)*preferred
…..…… 803-896-0878 (phone)
Testing/Technical questions:
Nancy Giurato, PhD……...803-896-9725
email: giuratnl@dhec.sc.gov
Sandi Hall, MT (ASCP).....803-896-0891
email: hallss@dhec.sc.gov
Data Entry:
Evelyn Edwards, MPH.....803-896-0897
email: edwardey@dhec.sc.gov
Mailing Envelopes, Newborn Screening Collection Forms:
Carlton Atkinson.....803-896-0913
email: atkinsct@dhec.sc.gov
On-site Workshops:
Roberta Bartholdi.....803-896-3897
email: barthork@dhec.sc.gov

Fax number for all Bureau of Laboratories staff.....803-896-0991

Mailing address for Bureau of Laboratories staff

(NAME)
SC DHEC BOL
Chemistry Division
Newborn Screening Laboratory
8231 Parklane Road
Columbia, SC, 29223

Educational Materials Library

To order Newborn Screening Brochures and/or Parental Statement of Religious Objection (DHEC 1804):

Lamar O'Neil….....803-898-3539
Fax for all ERC staff……803-898-3476
http://www.scdhec.gov/Agency/EML/

Mailing address for Education Materials Library staff

SC DHEC/EML
2600 Bull Street
Columbia, SC, 29201

Criteria For Notification of Abnormal Results: Weekday/Monday Holiday

The following table outlines the methods by which the physician of record is notified by the Division of Children's Health and/or the Bureau of Laboratories of an abnormal screening result or an unacceptable specimen.

Condition/Situation	Phone call with mailed confirmation	Mailed confirmation only
PKU/other Amino acid disorders	Amino acids other than TYR: Any result	Amino acids other than TYR: None
	TYR >= 800 µM regardless of SUAC result	TYR from 330 to 800 µM with normal SUAC result
	Elevated SUAC regardless of TYR result	None
CH	TSH >= 40 µIU/mL	TSH from 19 to 40 µIU/mL for infants 7 days of age and younger
		TSH from 10 to 40 µIU/mL for infants 8 days of age and older
Galactosemia	GALT < 40 µM regardless of GAO result	GAO >= 10 mg/dL with normal GALT
		GALT from 40 to 50 µM with normal GAO
CAH	17-OH progesterone >= 48 ng/mL for infants with birth weights >= 2500 grams	17-OH progesterone from 30 to 48 ng/mL for infants with birth weights >= 2500 grams
	17-OH progesterone >= 130g/mL for infants with birth weights < 2500 grams	17-OH progesterone from 76 to 130g/mL for infants with birth weights < 2500 grams
MCADD/other Fatty acid disorders	Most abnormalities (likelihood of disease status in some abnormal acylcarnitines can be determined using the Region 4 Genetics Collaborative Post Analytical tools)	Results from selected abnormal acylcarnitines that score likely normal using the R4GC tools
	C14:1 >= 0.50 µM with elevated C14:1/C2	None
	C14:1>=0.75 µM regardless of C14:1/C2 level	None
Organic acid disorders	Acyl carnitines other than C3: Most abnormalities (likelihood of disease status in some abnormal acylcarnitines can be determined using the R4GC tools)	Results from selected abnormal acylcarnitines that score likely normal using the R4GC tools
	C3 >= 10 µM with elevated C3/C2	C3 from 5.30 to 10 µM with elevated C3/C2
	C3 >= 15 µM with normal C3/C2	C3 from 5.30 to 15 µM with normal C3/C2
Cystic Fibrosis	None	Any elevated IRT
Biotinidase Deficiency	Any result	None

Condition/Situation	Phone call with mailed confirmation	Mailed confirmation only
Hemoglobinopathies	None	Any disease result
SCID	Any result	None
Unacceptable specimen	None	Any situation where the specimen was unacceptable

Criteria For Notification of Abnormal Results: Saturday/Other Holiday

The following table outlines the methods by which the physician of record is notified by the Division of Children's Health when the result is indicative of immediate morbidity/mortality.

Abnormal Analyte(s)	Condition(s)	Action
PHE	PKU	Wait until Monday
VAL and/or LEU+ILE	MSUD	Contact MD and specialist
MET	HCU	Wait until Monday
CIT	Citrullinemia I, II, ASA	Contact MD and specialist
SUAC	TYR I	Contact MD and specialist
TYR	TYR II, III	Wait until Monday
GALT < 40 µM and high GAO	Classical galactosemia	Contact MD and specialist
GALT < 25 µM and normal GAO	Duarte galactosemia	Contact MD and specialist
GALT normal and GAO > 20 mg/dL	Other galactosemia	Contact MD and specialist
Repeat GALT normal and high GAO	Other galactosemia	Contact MD and specialist
C3 >15 µM or >10 µM with one or more high ratios	PA, MMA	Contact MD and specialist
C3 high with both ratios high	PA, MMA	Contact MD and specialist
C3 high not detailed above	PA, MMA	Wait until Monday
C3DC+C4OH/C10	MA	Wait until Monday
C5	IVA, 2-MBDD	Contact MD and specialist
C4DC+C5OH	3-MCC; BKT; others	Contact MD and specialist
Multiple short and medium chain AC's	GA II	Contact MD and specialist
C5DC+C6OH	GA I	Contact MD and specialist if C5DC+C6OH>1.0 µM
C8	MCADD	Contact MD and specialist
C10:2	Dieonyl reductase	Wait until Monday
C16OH	LCHADD	Contact MD and specialist if indicated by R4GC tool
C14:1	VLCADD	Contact MD and specialist if C14:1>=0.75 µM in isolation or C14:1 >=0.50 µM and ratio. Use

Abnormal Analyte(s)	Condition(s)	Action
		R4GC dual scatter plot tool.
C0	CUD	Contact MD and specialist if C3+C16<2
C0/(C16+C18)	CPT I	Contact MD and specialist if indicated by R4GC tool
C16 and C18:1	CPT II	Contact MD and specialist
Biotinidase	Biotinidase deficiency	Contact MD and specialist
Any elevation TSH	Primary CH	Wait until Monday
17OHP>=48 ng/mL in NBW or 130 ng/mL in LBW	CAH	Contact MD and specialist
17OHP lower level abnormal result	CAH	Wait until Monday
Any elevation IRT	CF	Wait until Monday
Any abnormal Hb	SS or other Hb	Wait until Monday
Any elevation Cq	SCID	Wait until Monday

Best Specimen Collection Timing by Disorder

Condition	Best Age to Screen	Factors Affecting Tests	Consequences to Infant If Not Identified
Biotinidase deficiency	Birth-72 hours	False positive-premature/jaundiced False negative-red cell transfusion/ECLS	Hypotonia, seizures, developmental delay, abnormal movements, breathing problems, hair loss and hearing loss
Congenital Adrenal Hyperplasia	12-48 hours and 2-4 weeks	False positive-sick/stressed infant False negative-maternal steroids; infant dexamethasone	Acute crisis with failure to thrive, dehydration and shock, early puberty, virilization of females Can cause death in the newborn period
Cystic Fibrosis	24 hours-7 days	False positive-hypoxia, respiratory stress, hypoglycemia, trisomies (13, 18, 21), preterm, collect <12 hours False negative-meconium ileus, ? other GI	Failure to thrive, malnutrition, severe respiratory disease
Primary Congenital Hypothyroidism	12-72 hours and 2-6 weeks	False positive-sick/stressed infant, preterm, topical iodine False negative-delayed TSH rise	Prolonged jaundice, lethargy, poor muscle tone, mental retardation, abnormal movements, motor delays
Fatty Acid Oxidation Disorders	Birth-48 hours	False positive-Carnitine or MCT supplementation False negative-Carnitine supplementation	Hypoketotic hypoglycemia, metabolic decompensation/crisis, seizures Can cause death in the newborn period
Galactosemia	Birth-48 hours	False positive-liver disease False negative-red cell transfusion/ECLS	Hypoglycemia, jaundice, sepsis, failure to thrive, mental retardation Can cause death in the newborn period
Hemoglobinopathies	Birth-72 hours	False positive-none reported False negative-red cell transfusion/ECLS	Chronic hemolysis, intermittent vaso-occlusive pain episodes, splenic dysfunction which can lead to life-threatening infection
Urea Cycle, Amino	24-48	False positive-PN, liver	Seizures, lethargy, poor feeding,

Condition	Best Age to Screen	Factors Affecting Tests	Consequences to Infant If Not Identified
Acid, and Organic Acid Disorders	hours	disease, immature liver enzymes False negative-collect <24 hours	metabolic decompensation/crisis, coma, developmental delay, mental retardation Some forms can cause death in the newborn period

Adapted from *Newborn Screening for Preterm, Low Birth Weight, and Sick Newborns; Approved Guideline,* Clinical and Laboratory Standards Institute, October 2009

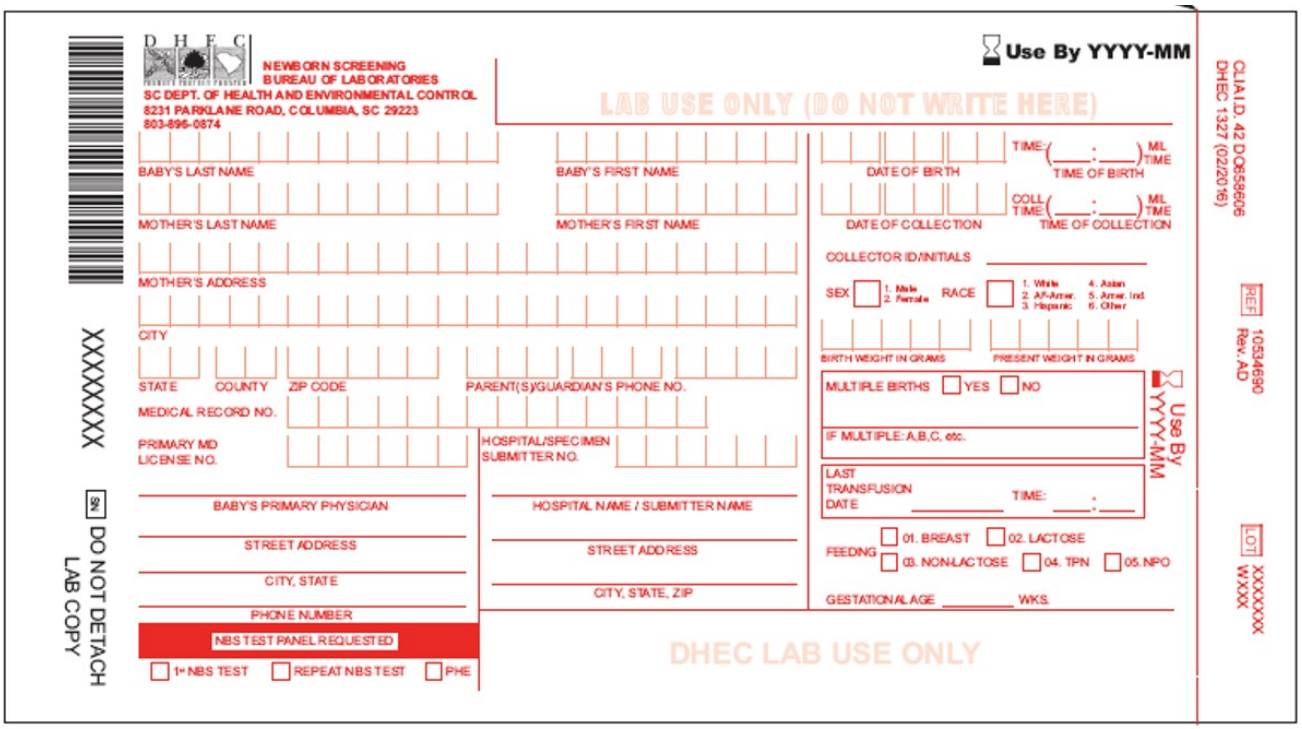
Sickle Cell Foundations in South Carolina

COBRA Human Services Agency Sickle Cell Program
3962 Rivers Ave
PO Box 71473
Charleston, SC 29415
Toll Free (800) 354-4704
(843) 225-4866, Service Line
(843) 225-4869, Fax
cobraagency@bellsouth.net

Orangeburg Area Sickle Cell Foundation
825 Summers Ave
PO Box 892
Orangeburg, SC 29116
(803) 534-1716, Phone
(803) 531-2422, Fax
orangeburgsickle@aol.com

James R Clark Memorial Sickle Cell Foundation
1420 Gregg St
Columbia, SC 29201
Toll Free (800) 506-1273
(803) 765-9916, Phone
(803) 799-6471, Fax
www.jamesrclarksicklecell.org
office@jamesrclarksicklecell.org

Louvenia Barksdale Sickle Cell Anemia Foundation
645 S Church St
PO Box 191
Spartanburg, SC 29304
(864) 582-9420, Phone
(864) 582-9421, Fax
www.barksdalesicklecell.org
ldbarksdale@charter.net



SOUTH CAROLINA DEPARTMENT OF HEALTH AND ENVIRONMENTAL CONTROL
BUREAU OF LABORATORIES

Newborn Screening (Instructions for Completing DHEC-1327) Revised 02/2016.

PURPOSE

This form is used to provide identification and essential information and a means of submitting blood specimens for newborn screening. Due to the makeup of this form and the information needed, it cannot be pre-addressed by the Bureau of Laboratories. It has to be filled out completely by the sender. NOTE: There is a space for two senders. Both senders will receive a copy of the results.

A completed form must be submitted with the circles on the filter paper filled with the newborn's blood. The instructions for specimen collection and handling of blood specimen are on the back of the form. Never place the form in plastic bags to submit to the laboratory. Plastic bags can cause false laboratory results. Always check expiration date of the filter paper. This information is on the face of the form. The laboratory will not accept blood on expired filter paper forms. Follow the general instructions for the patient and sender information. Further instructions are below.

BABY'S LAST NAME: Enter baby's legal last name

BABY'S FIRST NAME: Enter baby's legal first name

MOTHER'S LAST NAME: Enter mother's last name, adoption agency or lawyer's office (if considered baby's legal guardian).

MOTHER'S FIRST NAME: Enter mother's first name.

MOTHER'S ADDRESS: Enter mother's complete mailing address, city, state, county code, and zip code. (See back of the sender copy for county codes)

PARENT(S)/GUARDIAN'S PHONE NO.: Enter telephone number of parent(s) or guardian.

MEDICAL RECORD NO.: Enter hospital medical record number or DHEC Patient Number.

PRIMARY MD LICENSE NO. (BABY'S PRIMARY PHYSICIAN): Enter the number assigned by the Board of Medical Examiners of South Carolina preceded by the letter "M". If in a group of physicians enter the number assigned by the Bureau of Laboratories preceded by the letter "G".

BABY'S PRIMARY PHYSICIAN: Enter physician's name, Street Address, City, State, and phone number.

HOSPITAL/SPECIMEN SUBMITTER NO.: Enter the sender number. If a hospital, enter the number assigned by the Bureau of Laboratories preceded by the letter "H".

HOSPITAL NAME/SUBMITTER NAME.: Enter name of hospital or medical group/MD that is submitting the specimen.

STREET ADDRESS: Enter hospital or medical group/MD street address.

CITY, STATE, ZIP: Enter hospital's or medical group/MD's city, state and zip code.

NBS TEST PANEL REQUESTED: Check whether it is the 1st NBS TEST PANEL or a REPEAT NBS TEST PANEL. *PHE is ONLY used to request monitoring PHE and TYR analysis for persons diagnosed with PKU.*

DATE OF BIRTH: Enter baby's date of birth. Enter month, day, and year. Precede all numbers less than ten (10) with a zero (0). Example: September 1, 2013 would be 09/01/13.

TIME: Enter time of birth (hour and minute). USE MILITARY (24 HOUR CLOCK) TIME. Precede all numbers less than ten (10) with a zero (0). Examples: 9:20 am would be 09:20. 9:20 pm would be 21:20.

DATE OF COLLECTION: Enter month, day, and year specimen was collected. Precede all numbers less than ten (10) with a zero (0). Example: September 1, 2013 would be 09/01/13.

TIME OF COLLECTION: Enter time of collection (hour and minute). USE MILITARY (24 HOUR CLOCK) TIME. Precede all numbers less than ten (10) with a zero (0). Examples: 9:20 am would be 09:20. 9:20 pm would be 21:20.

COLLECTOR ID/INITIALS: Enter ID or Initials as directed by collector's supervisor.

SEX: Enter 1 for Male and 2 for Female in the block.

RACE: Insert appropriate number in block as outlined below:

1. White	4. Asian
2. African-American	5. American Indian
3. Hispanic	6. Other

BIRTH WEIGHT IN GRAMS: Enter weight of baby at birth in GRAMS.

PRESENT WEIGHT IN GRAMS: Enter weight of baby at time of specimen collection in GRAMS.

MULTIPLE BIRTHS: Mark an "X" in the appropriate box Yes or No.

IF MULTIPLE: A, B, C, etc.: If multiple birth – YES, write in baby's birth order (i.e., A, B, C, etc.)

LAST TRANSFUSION DATE: If baby has received any blood product containing red cells (including in utero transfusions), write in the date of the last transfusion (month, day, and year).

LAST TRANSFUSION TIME: If baby has received any blood product containing red cells (including in utero transfusions), enter the time of the last transfusion. USE MILITARY (24 HOUR CLOCK) TIME. Precede all numbers less than ten (10) with a zero (0). Examples: 9:20 am would be 09:20. 9:20 pm would be 21:20.

FEEDING: Check the appropriate box

GESTATIONAL AGE: Write in the approximate GA at the time of specimen collection.

THE FORM: The form is made up of three parts.
Part 1: Lab copy. DO NOT detach.
Part 2: Sender's copy can be retained by the sender.
Part 3: The Cover, along with Part 1, must stay attached and be returned to the laboratory. The flap over the dried blood spots must cover the spots when the NBS form is placed in the envelope for mailing.

DO NOT USE TAPE or STAPLES on the form. DO NOT USE an addressograph on the form. The addressograph can compress the filter paper and mar the blood.

DO NOT write or place labels in the top area of the form that is designated "LAB USE ONLY."

OFFICE MECHANICS AND FILING: After processing in the laboratory, a computer-generated report will be mailed to the sender(s) and the laboratory will retain the original paperwork.

D H E C

PROMOTE PROTECT PROSPER

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 conditions, 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 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.

Instructions
DHEC 1804, Parental Statement of Religious Objection

PURPOSE: This form is used by hospital, health department and other health care provider staffs to document a religious objection to newborn screening for inborn errors of metabolism and hemoglobinopathies.

ITEM BY ITEM INSTRUCTIONS:

Top Section: Print parents or guardians' names on the line indicated. Print child's name and date of birth on the lines indicated.

Bottom Section: The parent or guardian signs his/her name and indicates the date in the appropriate space. The witness signs his/her name and indicates the date in the appropriate space.

OFFICE MECHANICS AND FILING: Mail the original to: Newborn Screening Follow-up Program, Division of Women and Children's Services, SC DHEC, Mills/Jarrett Complex, Box 101106, Columbia, SC 29211. One copy can be given to the parent or guardian. One copy is filed under consents at the health department/facility where the form was signed. The form should be retained according to the medical records retention schedule.

Newborn Screening Law and Regulation

Law

Neonatal Screening for Inborn Metabolic Errors and Hemoglobinopathies Section 44-37-30, of the South Carolina Code of Laws

(A) A child born in this State, except a child born of a parent who objects on religious grounds and indicates this objection before testing on a form promulgated in regulation by the Department of Health and Environmental Control, shall have neonatal testing to detect inborn metabolic errors and hemoglobinopathies.

(B) Information obtained as a result of the tests conducted pursuant to this section is confidential and may be released only to a parent or legal guardian of the child, the child's physician, and the child when eighteen years of age or older when requested on a form promulgated in regulation by the department.

(C) A blood sample obtained pursuant to this section is confidential and may be released only as the parent or legal guardian of the child from whom a blood sample was obtained, or the child when eighteen years of age or older, directs the department at the time of testing or at any time after that on a form promulgated in regulation by the department.

(D)(1) Unless otherwise directed pursuant to this subsection, a blood sample obtained pursuant to this section must be stored by the department at minus 20º centigrade and may be released for purposes of confidential, anonymous scientific study. The release of a blood sample must conform with regulations promulgated by the department. At the time of testing or at any time after that, on a form promulgated in regulation by the department, the parent or legal guardian of the child from whom a blood sample was obtained, or the child when eighteen years of age or older, may direct the department to:

(a) return a blood sample in its entirety and any test results not less than two years after the date of testing;

(b) destroy a blood sample in a scientifically acceptable manner not less than two years after the date of the testing; or

(c) store a blood sample at minus 20º centigrade but not release the blood sample for confidential, anonymous scientific study.

(2) A blood sample released for confidential, anonymous study pursuant to this section must not contain information which may be used to determine the identity of the donor. A blood sample released pursuant to this section may contain demographic or other statistical information. If scientific study identifies genetic information that may benefit the child, the department may notify confidentially the parent or legal guardian, or the child if eighteen years of age or older, of this information.

(E)(1) A blood sample that has not been stored at minus 20° centigrade before the effective date of this section must be destroyed in a scientifically acceptable manner six months from the effective date of this section unless a parent or legal guardian of a child from whom a blood sample was obtained, or the child if eighteen years of age or older, requests return of the blood sample on a form provided by the department.

(2) A blood sample stored at minus 20° centigrade pursuant to this section before the effective date of this section must be retained as prescribed in subsection (D) unless directed by the parent or legal guardian of the child from whom a blood sample was obtained to destroy or return the blood sample.

(F) The department shall promulgate regulations necessary for the implementation of this section. All forms must include information concerning the benefits of neonatal testing and storage of a blood sample.

(G) A person who violates this section or the regulations promulgated pursuant to this section or who provides or obtains or otherwise tampers with a blood sample collected pursuant to this section is guilty of a misdemeanor and, upon conviction, may be fined not more than fifty thousand dollars or imprisoned for not more than three years."

Severability

SECTION 3. If any section, subsection, paragraph, subparagraph, sentence, clause, phrase, or word of this act is for any reason held to be unconstitutional or invalid, such holding shall not affect the constitutionality or the validity of the remaining portions of this act, the General Assembly hereby declaring that it would have passed this act, and each and every section, subsection, paragraph, subparagraph, sentence, clause, phrase, and word thereof, irrespective of the fact that any one or more other sections, paragraphs, subparagraphs, sentences, clauses, phrases, or words thereof may be declared to be unconstitutional, invalid, or otherwise ineffective.

Proviso 34.37. (DHEC: Metabolic Screening) The department may suspend any activity related to blood sample storage as outlined in Section 44-37-30 (D) and (E) of the 1976 Code, if there are insufficient state funds to support the storage requirements. In that event, the samples may be destroyed in a scientifically appropriate manner after testing. The department shall notify providers of the suspension within thirty days of its effective date.

Regulation

South Carolina Department of Health and Environmental Control
REGULATION 61-80
Neonatal Screening For Inborn
Metabolic Errors and Hemoglobinopathies

Contents:

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 conditions, 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 mental retardation, 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 mental retardation, 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

Acknowledgements

The metabolic disorders information pages were adapted from the following sources:

Fact Sheets, Oregon State Public Health Laboratory

Health Professionals Guide to Newborn Screening, Wisconsin Newborn Screening Laboratory

New England Consortium of Metabolic Programs at Children's Hospital Boston

ACMG ACT Sheets and Confirmatory Algorithms