SC Newborn Screening (NBS) Contact Information

Clinical Follow-up

If you have any questions about newborn screening clinical follow-up or patient case management services, please contact a team member listed below:

Pediatric Medical Consultant: Sylvia Brook, MD.....803-898-0362
Email: brookax@dph.sc.gov

Pediatric Medical Consultant: Stephanie Gibson, MD.....803-898-0362
Email: gibsonsl@dph.sc.gov

NBS Follow-up Program Director:
Tanya Spells, MS, MLS(ASCP).....803-898-0619
Email: spellsty@dph.sc.gov

NBS Administrative Assistant:
Karla Clyde……..803-898-3192
Email: clydeka@dph.sc.gov

NBS Metabolic Dietitian, Metabolic Formula, Abnormal Results:
Marcelina Jackson, RD, LDN……..803-898-1969
Email: jacksom@dph.sc.gov

NBS Follow-up Program Coordinator, Abnormal Results:
Katrina L. Davis …….....803-898-0593
Email: daviskl@dph.sc.gov

NBS Follow-up Program Coordinator, Abnormal Results:
Kimberly C. Adams, MA.....803-898-0696
Email: adamskc@dph.sc.gov

NBS Follow-up Program Coordinator, Abnormal Results:
VACANT.....803-898-3845
email:

Fax number for all Newborn Screening Follow-up staff.....803-898-0337

Laboratory Services

If you have any questions about newborn screening laboratory services, please contact the staff listed below:

Retrieval of Newborn Screening Patient Lab Reports Online:

https://apps.dhec.sc.gov/Health/eReports/weblogin.aspx?MsgNum=0&Locale=1033

Request access to NBS Results Portal Email: nbslab@dph.sc.gov
Contact Lab Information Management Systems (LIMS fax)….803-896-3862

Testing/Technical questions:

Newborn Screening Director
Beth Bair, MS……..803-896-0991
Email: bairea@dph.sc.gov

NBS Laboratory Manager
Sandi Hall, BS, MLS(ASCP)…….....803-896-0891
Email: hallss@dph.sc.gov

NBS Lab Testing Supervisor
Graham McCaskill……803-896-0878
Email: mccasksg@dph.sc.gov

NBS Lab Testing Supervisor
D. Rodger Givens……...803-896-5938
Email: givensdr@dph.sc.gov

NBS Quality Improvement Specialist
Ashley Marchese....……..... 803-896-1140
Email: marchea@dph.sc.gov

NBS Outreach Coordinator
Christine Harrelson, MS, MLS(ASCP).....803-896-1140
Email: harrelcl@dph.sc.gov

Fax number for all NBS Laboratory staff.....803-896-0298

Office of Lab Quality Assurance (QA):

QA Program Manager
Patricia Myers, BS, MLS(ASCP).....803-896-3897
Email: myerspa@dph.sc.gov

Lab QA Technologist
Lynn Gleaton, BS, MLS(ASCP)……803-896-0899
Email: gleatoll@dph.sc.gov

Laboratory Information Management Systems (LIMS):

LIMS Administrator
Linda Conway, MLT(ASCP)……803-896-4777
Email: nbslab@dph.sc.gov

LIMS Specimen Gate Lead
Caroline Owen……..…….803-896-0945
Email: owencc@dph.sc.gov

Fax number for LIMS Department……...803-896-3862

To order Newborn Screening Collection Forms and Mailing Envelopes:

Lab Supply Manager
Todd Dufries.....803-896-0913
Email: dufriets@dph.sc.gov

Specimen Management, Lab Media, and Lab Billing:

Support Manager
VACANT.....803-896-2331
Email:

Mailing address for Newborn Screening Lab:

(NAME)
SC DPH
Newborn Screening
8231 Parklane Road
Columbia, SC 29223

Educational Materials Library (EML) and Forms

Contact:

LaMar O'Neil….....803-898-3539
Fax for all ERC staff……...803-898-3476

https://dph.sc.gov/professionals/health-professionals/educational-materials

To order:

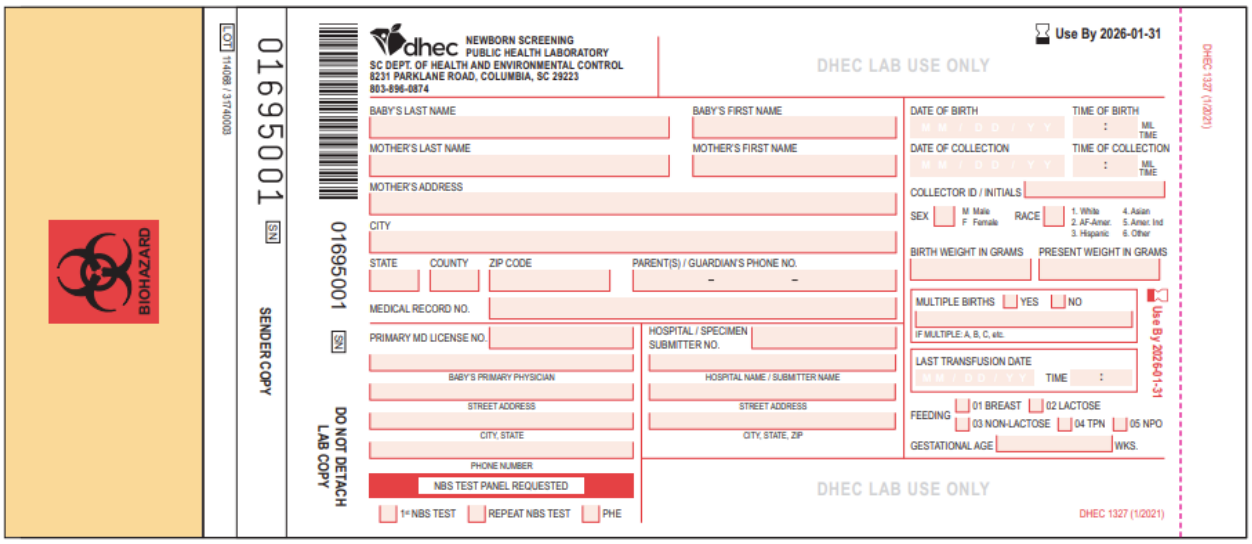
Newborn Screening Brochures "*For Your Baby's Health*" (ML-000032 in English)
Newborn Screening Brochures "*Para la salud de su bebe*" (ML-025096 in Spanish)

Parental Statement of Religious Objection Form (DPH 1804 in English)

Parental Statement of Religious Objection Form "*Declaracion de objection religiosa por parte de los padres*" (DPH 1804S in Spanish)

Mailing address for Education Materials Library staff:

SC DPH/EML
2600 Bull Street
Columbia, SC 29201



SOUTH CAROLINA DEPARTMENT OF PUBLIC HEALTH
PUBLIC HEALTH LABORATORY

Newborn Screening (Instructions for Completing DHEC-1327) Revised 1/2021

PURPOSE

Per the South Carolina Code of Laws, Sections 44-37-30, 44-37-35, and SC Regulation 61–80 for Neonatal Screening, the SC Newborn Screening (NBS) Program tests our youngest residents for numerous congenital disorders shortly after birth.

This form is used to provide identification and essential information and a means of submitting blood samples for newborn screening. Due to the makeup of this form and the information needed, it cannot be pre-addressed by the Public Health Laboratory. It must be filled out completely by the provider.

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.

HOSPITAL NO.: Enter hospital medical record number or Patient Number.

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

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

BILLING NUMBER: Used by Health Departments.

PROGRAM NUMBER: Used by Health Departments.

HOSPITAL/SPECIMEN SUBMITTER NO.: Enter the sender number. If a hospital, enter the number assigned by the Public Health Laboratory 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 or medical group/MD's city, state, and zip code.

NBS TEST PANEL REQUESTED: Check whether it is the 1st NBS TEST, REPEAT NBS TEST, or PHE (PKU monitoring sample) only.

NOTE: PKU is only one of the 60+ NBS screening tests available. Marking "PHE" when ALL tests are needed will mean a complete screening may NOT be performed.

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, 2024, would be 09/01/24.

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.

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

RACE: Insert appropriate number in block as outlined below:

1. White
2. African American
3. Hispanic
4. Asian
5. American Indian
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).

FEEDING: Check the appropriate box

DATE COLLECTED: Enter month, day, and year specimen was collected. Precede all numbers less than ten (10) with a zero (0). Example: September 2, 2023, would be 09/02/23.

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.

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 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 available to the sender(s) and the laboratory will retain the original paperwork.

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 (extracorporeal life support)	Hypotonia, seizures, developmental delay, abnormal movements, breathing problems, hair loss and hearing loss
Congenital Adrenal Hyperplasia	24-48 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, collection < 12 hours of age. False negative-meconium ileus; other GI	Failure to thrive, malnutrition, severe respiratory disease
Congenital Hypothyroidism	24-72 hours and 2-6 weeks	False positive-sick/stressed infant, preterm, topical iodine False negative-delayed TSH rise	Prolonged jaundice, lethargy, poor muscle tone, Intellectual disability (ID), abnormal movements, motor delays
Fatty Acid Oxidation Disorders	Birth - 48 hours	False positive-Carnitine, MCT supplementation or older babies False negative-Carnitine supplementation	Hypoketotic hypoglycemia, metabolic decompensation or crisis, seizures Can cause death in the newborn period
Galactosemia	Birth - 48 hours	False positive-liver disease False negative-red cell transfusion/ECLS extracorporeal life support	Hypoglycemia, jaundice, sepsis, failure to thrive, Intellectual disability (ID) Can cause death in the newborn period
Hemoglobin Disorders	Birth - 72 hours	False positives: rarely reported.	Chronic hemolysis, intermittent vaso-occlusive pain episodes, splenic

Condition	Best Age to Screen	Factors Affecting Tests	Consequences to Infant if Not Identified
		False negative-red cell transfusion/ECLS (extracorporeal life support)	dysfunction which can lead to life-threatening infection
Urea Cycle, Amino Acid, and Organic Acid Disorders	24-48 hours	False positive-PN, liver disease, immature liver enzymes False negative: collection < 24 hours of age	Seizures, lethargy, poor feeding, metabolic decompensation/crisis, coma, developmental delay, Intellectual disability (ID) Some disorders can cause death in the newborn period
Severe Combined Immunodeficiency (SCID)	24 -72 hours	Prematurity. SCID like diseases, such as DiGeorge syndrome, idiopathic T-cell lymphopenia, heparin anticoagulant interference, poor quality or low quantity of TREC-DNA extraction.	Excessive number of life-threatening infections and failure to thrive
Pompe	24 -72 hours	False positives – Pseudo-deficiency alleles and carriers. Prematurity with associated lower hematocrit, spike in newborn WBC count	Uniformly fatal if untreated, to more slowly progressive later onset forms
MPS I	24 -72 hours	False positives – Pseudo-deficiency alleles and carriers. Prematurity	May include coarse facies, hearing loss, umbilical hernia, hepatosplenomegaly, progressive dysostosis multiplex, cardiac valvular disease, corneal clouding, and developmental delay
Infantile Krabbe Disease		False positives – Pseudo-deficiency alleles and carriers. Older babies.	Newborns are asymptomatic and, if left untreated, survival beyond age 2 years is uncommon.
Late Onset Krabbe Disease		False positives – Pseudo-deficiency alleles and carriers.	All disease forms are associated with leukodystrophy; the age of onset and rate of progression vary widely.
Spinal Muscular Atrophy (SMA type 1)	24 -72 hours	Prematurity, LBW, and/or sick baby. Other types of SMA may be detected	If left untreated, death typically occurs by 2 years of age in the infantile form.

Adapted from *Newborn Screening for Preterm, Low Birth Weight, and Sick Newborns*. Clinical and Laboratory Standards Institute (CLSI) Approved Guideline NBS03, 2nd Edition, August 2019.

Criteria for Notification of Abnormal Results (on Weekdays)

The following table outlines the methods by which the physician of record is notified of an abnormal screening result or an unacceptable specimen, as of 2/5/2025:

Condition	**CRITICAL results: Phone call with fax and mailed confirmation**	**Abnormal results: Mailed confirmation only**
CH	TSH >= 40 µIU/mL – *CRITICAL* TSH >= 100 µIU/mL - *PANIC*	TSH between 20 and 39 µIU/ml (for infants <= 7 days old) and TSH > 10.50 µIU/ml (for infants > 7 days old)
Galactosemia	GALT < 3.00 U/dL, regardless of GAO result	GAO >= 12.0 mg/dL with normal GALT
		GALT from 3.00 to 3.75 U/dL, regardless of GAO result
CAH	17-OHP >= 48 ng/mL for infants with birth weights >= 2500 grams	17-OHP from 30 to 47 ng/mL for infants with birth weights >= 2500 grams
	17-OHP >= 130 g/mL for infants with birth weights < 2500 grams	17-OHP from 76 to 129 g/mL for infants with birth weights < 2500 grams
Amino Acid disorders		
H-PHE or PKU	PHE > 120 and PHE/TYR ratio > 2 or PHE >= 300	PHE >= 120 only
TYR I or TYR II/III	TYR >= 800 µM regardless of SUAC result	TYR from 300 to 800 µM with normal SUAC result
TYR I	Elevated SUAC, regardless of TYR result	
HCY	MET >= 75, if indicated by CLIR	
MSUD	LEU+ILE >= 325 and Leu+Ile/Phe ratio > 4.00 and/or Leu/Ala ratio > 1.00	
Valinemia or MSUD	VAL >= 250 and Val/Phe > 4.50	
CIT I/II	CIT >= 60 and	CIT >= 60 only

Condition	**CRITICAL results: Phone call with fax and mailed confirmation**	**Abnormal results: Mailed confirmation only**
	CIT/ARG ratio > 15.00, and/or ORN/CIT ratio > 1.00, or CIT >=100	
ASA	CIT >= 60, ASA >= 1.00, and/or ASA/ARG ratio > 0.30	CIT >= 60 only
ARG	Elevated ARG and ARG/ORN ratio	Elevated ARG only
Fatty acid disorders	Most abnormalities (likelihood of disease status in some abnormal acyl carnitines can be determined using the CLIR validated tools)	Results from selected abnormal acyl carnitines that score likely normal using the CLIR validated tools
CUD	Low C0 <= 9.00 and C3+C16 ratio < 2 on infants < 7 days old	
CPT I	Normal to High C0 with Elevated C0/C16+C18 ratio	
SCHAD	C3DC+C4OH >= 0.40, if indicated by CLIR	
MCAD	C8 >= 0.50 C8 >= 1.5 *CRTICAL*	
VLCAD	C14:1 >= 0.58 with elevated C14:1/C2 ratio	
VLCAD	C14:1 >= 0.70 and C14:1/C2 ratio > 0.06 or C14:1 >= 1.5 *CRITICA*L regardless of ratio	
LCHAD/TFP	C16-OH >= 0.12 and C18OH/C18 ratio > 0.08	
CPT II/CACT	High C16 and C3/C16 ratio and/or High C18:1	
Organic acid disorders	Acyl carnitines other than C3: Most abnormalities (likelihood of disease status in some abnormal Acyl carnitines can be determined using the CLIR validated tools)	Results from selected abnormal acyl carnitines that score likely normal using the CLIR validated tools.
PROP, Cobalamin C, D	C3 >= 10.00 μM with elevated C3/C2 > 0.27 - *URGENT*	C3 from 7.00 to 10.0 μM

Condition	**CRITICAL results: Phone call with fax and mailed confirmation**	**Abnormal results: Mailed confirmation only**
MUT, Cobalamin A, B	C3 >= 10.00 μM with elevated C3/C16 > 3.00 - *CRITICAL*	C3 from 7.00 to 10.0 μM
PROP, MUT, MCD, Cobalamin A, B	C3 >= 15 μM with normal C3/C2 *PANIC*	C3 from 7.00 to 10.0 μM
MAL	C3DC+C4OH >= 0.40, if indicated by CLIR	
3-MCC, HMG, βKT	C4DC+C5OH >= 2.00 *CRITICAL*	C4DC+C5OH >= 0.70
GA-I	C5DC+C6OH >= 0.75 *CRITICAL*	C5DC+C6OH >= 0.30
IVA or SBCAD	C5 >= 0.50, if indicated by CLIR C5 >= 2.5 *CRITICAL*	
GA II	C4 >= 1.20 and C5 >= 0.50 with Multiple elevated acyl carnitines	
Hemoglobin Disorders		
Sickling Hemoglobin Diseases	Any sickling disease result: FS, FSA, FSB, FSC, FSD, FSE, FSV, FSO, or FSG (*Start Penicillin*)	Email notification to Regional Sickle Cell Program Coordinators in (CYSHCN) Sickle Cell Program
Non-sickling Hemoglobin Diseases	FC, FD, FE, FF, F+O (Other)	
Hemoglobin traits and carriers* (ex. FAS and FAC)	None	Lab notification to PCP and regional Sickle Cell community-based organizations (CBO's) for follow up, genetic education and counseling.
FA +Fast HGB Variant (aka Hemoglobin Bart's FAB)	FA + Fast HGB Variant >= 15% FA + Fast HGB Variant >= 25% - *CRITICAL*	FA +Fast HGB Variant < 15%
Other Disorders		
SCID	TREC < 100 copies *CRITICAL*	TREC < 250 copies
Cystic Fibrosis - IRT	IRT > 170	Any IRT < 170
Cystic Fibrosis – 2nd tier testing*	1 or more mutations found	No mutations found
Biotinidase Deficiency	BIO < 35 U/dL	BIO < 70 U/dL

Condition	CRITICAL results: Phone call with fax and mailed confirmation	Abnormal results: Mailed confirmation only
Spinal Muscular Atrophy (SMA)	Low to absent *SMN1* level	
Lysosomal Storage Disorders		
GAA enzyme (screen for Pompe Disease)	GAA (acid-a-glucosidase) Low to absent enzyme level	GAA (acid-a-glucosidase) < 20% of Daily Median
Pompe - 2nd tier testing	Mutations indicative of disease found	No mutations or pseudo deficiency alleles found (normal findings)
IDUA enzyme (screen for Mucopolysaccharidosis Type I - MPS I)	IDUA (a-L-iduronidase) Low to absent enzyme level	IDUA (a-L-iduronidase) < 10% of Daily Median
MPS I - 2nd tier testing	Mutations indicative of disease found	No mutations or pseudo deficiency alleles found (normal findings)
GALC enzyme **and** Psychosine (screen for Krabbe Disease)	Any elevated Psychosine level	Decreased GALC enzyme only
Unacceptable specimens	Hospital notification (for inpatient/NICU babies only)	Parent notification for any discharged baby whose initial specimen was unacceptable for testing.

***Note: Carrier Status**

Everyone has two copies of each gene, one from their biological mother and one from their biological father. Being a **carrier** means that one copy of a gene was altered (changed) and passed on to a person. However, the other copy is normal and functioning fine. Since carriers still have one working copy of the gene, most typically do not have health problems associated with carrying the altered gene.

Many people are carriers of a disease-causing gene alteration without even knowing it. Being a carrier, however, means there is an increased chance that a child of the carrier could be born with a genetic disease. Anyone can be a carrier of a genetic disease, although no one in the family is affected.

Critical and Panic Values

The SC Newborn Screening Program defines critical values (also known as panic values) as laboratory test results that exceed established limits (high or low) as defined by the laboratory for certain analytes as listed in the Critical (Panic) Limits.

Critical results are considered life-threatening and require immediate notification of the physician, the physician's representative, the ordering entity, or other clinical personnel responsible for the patient's care.

Note: Results that are higher or lower than the lab's established reference ranges are considered "*outside of acceptable limits*" or "*abnormal*". However, not all abnormal results are considered "*critical"* values. So, the terms "abnormal" and "critical" should not be used interchangeably.

Criteria for Notification of Abnormal Results (on Saturdays and Holidays)

The following table outlines the methods by which the physician of record is notified, when a result is indicative of immediate morbidity/mortality, as of 2/5/2025.

Abnormal Analyte(s)	Possible Condition(s)	Action
PHE	H-PHE or PKU	Wait until Monday
LEU+ILE and/or VAL	Valinemia or MSUD	Contact MD
MET	TPN or HCY	Wait until Monday
CIT and/or ASA	Citrullinemia I/II, ASA	Contact MD
SUAC	TYR I	Contact MD
TYR	TYR II, III	Wait until Monday
Low GALT and High GAO	Classic Galactosemia	Contact MD if GALT <= 3.75 and GAO >= 12.0
Very Low GALT	Classic or Duarte Galactosemia	Contact MD if GALT <= Lower limit and normal GAO (< 12.0)
High GAO	Other Galactosemia	Contact MD if GAO >= 24.0
Repeat GALT normal and high GAO >= 12.0	Other Galactosemia	Wait until Monday
Low C0 and ratio	CUD	Contact MD if C0 < 9.00 and C3+C16 < 2 and baby is < 7 days old
High C0	CPT I	Contact MD if indicated by CLIR tools
High C0/(C16+C18)	CPT I	Contact MD if Ratio >= 45 and infant <= 7 days old
C3 < 10	PROP, MMA, MUT, Cobalamin A, B	Wait until Monday
C3 > 10	PROP, MMA	Contact MD if C3 >10 with one or more corresponding high ratios
C5	IVA or SBCAD	Contact MD
C4 and C5 (or C8)	GA II	Contact MD Use CLIR dual scatter plot tool to differentiate MCAD from GA II
Multiple short and medium chain AC's C4, C5 (C8, C10, C12, C14, C14:1, C16OH)	GA II	Contact MD Use CLIR dual scatter plot tool to differentiate MCAD from GA II
C3DC+C4OH	MAL, SCHAD	Contact MD if indicated by CLIR
C4DC+C5OH	3-MCC, HMG, MCD, βKT, 3MGA, 2M3HBA	Contact MD if C4DC+C5OH > 2.00

Abnormal Analyte(s)	Possible Condition(s)	Action
C5DC+C6OH	GA I	Contact MD if C5DC+C6OH >0.75
C8 (with C6 and C10)	MCAD	Contact MD
C10:2	Dienoyl reductase (DE RED)	Wait until Monday
C14:1	VLCAD	Contact MD if C14:1>=0.70 in isolation or C14:1 >=0.58 with elevated ratio. Use CLIR dual scatter plot tool to differentiate VLCAD vs. VLCAD (het)
High C16OH, and C18OH/C18 ratio	LCHAD, TFP	Contact MD, if indicated by CLIR tools
High C16 and C3/C16	CPT II/CACT	Contact MD if indicated by CLIR tools
Biotinidase < 70.0 U/dL	Biotinidase deficiency	Wait until Monday
Biotinidase < 35.0 U/dL	Biotinidase deficiency	Contact MD
TSH < 39	CH	Wait until Monday
TSH >= 40	CH	Contact MD
17OHP >= 48 in NBW or >= 130 in LBW baby	CAH	Contact MD
17-OHP abnormal result not indicated above	CAH	Wait until Monday
IRT > 170	Sick baby, CF, or CF carrier	Contact MD
One CF mutation found	CF carrier or CF	Wait until Monday
Two CF mutations found	CF	Contact MD
Abnormal Hgb	Sickling Hgb Disease	Contact MD
Abnormal Hgb trait	FA +Bart's >= 25%	Contact MD
Abnormal Hgb	Non-sickling Hgb disease	Wait until Monday
TREC < 250 copies	Prematurity, LBW, sick baby, or possible other disorder	Contact MD
TREC < 100 copies	SCID or other possible immune disorder	Contact MD
Borderline GAA or IDUA	Pompe or MPS I pseudo-deficiency or carrier	Wait until Monday
Low to absent GAA or IDUA	Pompe or MPS I disease	Contact MD
SMN1 low to absent	Possible type of SMA	Contact MD
Elevated Psychosine	Early (or Late) onset Krabbe Disease	Contact MD

Resources for Genetic and Metabolic Disorders in South Carolina

Call **866-262-3070** to access a clinical geneticist/metabolic specialist (on call) statewide.

You may also reach a clinical geneticist/metabolic specialist through their direct home offices as designated below:

Richard Schroer, **MD**
Sr. Clinical/Biochemical Geneticist
Greenwood Genetic Center: Charleston
3520 W Montague Ave, Ste 103
N. Charleston, SC 29418

LaToya 843-735-5093
Office Number: 877-679-0927
Main Fax: 866-676-9881
Alternate Fax: 843-735-5097
Cell: 843-206-9037

Emily Black, MD
Clinical Geneticist
Greenwood Genetic Center: Greenville
14 Edgewood Drive
Greenville, SC 29605

Tiffany 864-388-1065
Office (864) 250-7944
Fax: 864-250-9582

Neena Champaigne, MD
Clinical/Biochemical Geneticist
MUSC Department of Pediatrics
Division of Genetics
Rutledge Tower, Third Floor
135 Rutledge Avenue, MSC 567
Charleston, SC 29425

Office: 843-792-6735
Fax: 843-876-1518
Cell: 864-980-1295

Kristen Ann Lancaster, MD, FAAP
Pediatric Geneticist
MUSC Children's Health Genetics
Summey Medical Pavilion
2250 Mall Drive
North Charleston, SC 29406

Office: 843-876-1417
Fax: 843-792-1435

Laura Pollard, PhD, FACMG
Associate Director
Biochemical Genetics Laboratory
Greenwood Genetic Center
106 Gregor Mendel Circle
Greenwood, SC 29646

Phone: (864) 388-1070
Fax: 864-941-8133

Lauren Thompson, DO, FACMG
Clinical and Medical Biochemical Geneticist
Medical University of South Carolina
135 Rutledge Avenue
Charleston, SC 29425

Phone: 843-876-1229
Fax: 843-985-9744

Note: This list is not inclusive of all clinical geneticists and/or metabolic centers in South Carolina.

6/7/2024

Referral Sources for Pediatric Cystic Fibrosis in North & South Carolina

Low Country/Pee Dee: Allendale, Bamberg, Beaufort, Berkeley, Calhoun, Charleston, Colleton, Dorchester, Florence, Georgetown, Hampton, Horry, Jasper, Orangeburg, and Williamsburg counties

Sylvia E. Szentpetery MD, MPH
MUSC Comprehensive Cystic Fibrosis Center
Department of Pediatrics
135 Rutledge Ave, Ste 279
Charleston, SC 29425
Office: 843-876-1555
Fax: 843-876-1583
szentpet@musc.edu

Heather M. Staples, MD, Director
Prisma Health: Pediatric Pulmonology
Cystic Fibrosis Center
101 William H Johnson Street, Suite 400
Florence, SC 29506
Office: 803-434-2505
Fax: 803-758-0141
heather.staples@prismahealth.org

MUSC CF nurse coordinator – Jeralyn Wunderley (wunder@musc.edu) | Pediatric Cystic Fibrosis Center, 843-876-8518

For MUSC referrals use MEDULINE: 800-922-5250

MUSC Genetic counselor – Kimberly Brown Foil, MS, CGC | foilk@musc.edu

Midlands/Pee Dee: Aiken, Barnwell, Chester, Chesterfield, Clarendon, Darlington, Dillon, Edgefield, Fairfield, Kershaw, Lancaster, Lee, Lexington, Marion, Marlboro, Newberry, Richland, Saluda, and Sumter counties

Daniel Craig Brown, MD
Prisma Health: Pediatric Pulmonology
9 Medical Park, Ste 505
Columbia, SC 29203
Office: 803-434-2505
Fax: 803-758-0141
daniel.brown2@prismahealth.org

Heather M. Staples, MD, Director
Pediatric Cystic Fibrosis Center
9 Medical Park, Ste 505
Columbia, SC 29203
Office: 803-434-2505
Fax: 803-758-0141
heather.staples@prismahealth.org

PRISMA Health Pediatric CF coordinator – Sara Edwards (sara.edwards4@prismahealth.org) | 803-434-3293

Upstate: Abbeville, Anderson, Cherokee, Greenville, Greenwood, Laurens, McCormick, Oconee, Pickens, Spartanburg, Union, and York counties

Steven M. Snodgrass, MD
Prisma Health: Pediatric Pulmonology
200 Patewood Dr, Ste. A300
Greenville, SC 29615
Office: 864-454-5530
Fax: 864-241-9246
steve.snodgrass@prismahealth.org

Steven M. Snodgrass, MD
Prisma Health: Pediatric Pulmonology
249 N Grove Medical Park Dr, Ste. 200
Spartanburg, SC 29303
Office: 864-454-5530
Fax: 864-241-9246
steve.snodgrass@prismahealth.org

North Carolina/South Carolina border:

Levine Children's Specialty Center Pulmonology

Dr. Dennis E. Schellhase, Center Director
Atrium Health Cystic Fibrosis Program
1001 Blythe Blvd. Suite E
Charlotte, NC 28203
Pulmonary Clinic: 704-381-8840 or 4813
Fax: 704-381-8836
dennis.schellhase@atriumhealth.org

Atrium Health Nurse Coordinator/Navigator - Kendra Blevins, RN, Office: 704-355-2000 or email: kendra.blevins@atriumhealth.org

Heather M. Staples, MD, Director
Prisma Health: Pediatric Pulmonology
1003 West Meeting Street
Lancaster, SC 29720
Office: 803-434-2505
Fax: 803-758-0141
heather.staples@prismahealth.org

NOTE: This list is not inclusive of all pediatric pulmonologists in North & South Carolina.
Updated: 06/26/24

Resources for Pediatric Hemoglobin Disorders in SC

PRISMA Health Children's Hospital
Center for Cancer and Blood Disorders
7 Richland Medical Park
Columbia, SC 29203
(803) 434-3533
(803) 296-5437
https://prismahealthchildrens.org/locations/practices/pediatric-hematology-oncology-columbia

PRISMA Health Children's Hospital
Pediatric Hematology and Oncology
BI-LO Charities Children's Cancer Center
900 W. Faris Road, 2nd Floor
Greenville, SC 29605
(864) 455-8898

PRISMA Health Children's Hospital
Pediatric Hematology and Oncology
BI-LO Charities Children's Cancer Center
249 N. Grove Medical Park Drive, Suite 200
Spartanburg, SC 29303
(864) 573-8732

PRISMA Health Children's Hospital
Pediatric Hematology and Oncology
BI-LO Charities Children's Cancer Center
2000 E. Greenville Street, Ste 3500
Anderson, SC 29261
(864) 716-6490

https://prismahealthchildrens.org/locations/practices/pediatric-hematology-oncology-greenville

Medical University of South Carolina (MUSC)
Shawn Jenkins Children's Hospital
10 McClennan Banks Drive
Charleston, SC 29425
(843) 792-1414

Medical University of South Carolina (MUSC)
Rutledge Tower
135 Rutledge Ave
Charleston, SC 29425
(843) 876-0444

https://www.musckids.org/our-services/sickle-cell-center

Referral Sources for Pediatric Endocrine Disorders

Prisma Health Pediatric Endocrinology – Columbia

Alison Joan Lunsford, MD
9 Richland Medical Park Drive, Suite 230
Columbia, SC 29203
Phone: 803-434-7990
Fax: 803-758-0140

MUSC – Low Country

MUSC Children's Health R. Keith Summey Medical Pavilion
2250 Mall Drive
North Charleston, SC 29406
Phone: 843-876-0444
Fax: 843-985-0180

MUSC Children's Health Specialty Care - Mt Pleasant
2705 Highway 17, Suite 100
Mount Pleasant, SC 29466
Phone: 843-876-0444

MUSC Children's Health Specialty Care - Summerville
4330 Ladson Road
Summerville, SC 29485
Phone: 843-876-0444

Prisma Health Pediatric Endocrinology – Greenville

Elaine Apperson, MD
200 Patewood Dr, Ste 200
Greenville, SC 29615
Phone: 864-454-5100
Fax: 864-241-9238

NOTE: This list is not inclusive of all pediatric endocrinologists in South Carolina.

Updated: 10/18/2024

Resources for Pediatric Immune Disorders in South Carolina

Lower State (MUSC): Charleston, Berkeley, Dorchester, Colleton, Hampton, Jasper, Beaufort, Georgetown, Williamsburg, Horry, Marion, Dillon, Marlboro, Darlington, Florence

Kelli Williams, MD, MPH
West Ashley Medical Pavilion
2125 Charlie Hall Blvd
Charleston, SC 29414
Office: 843-876-3151
Fax: (843) 876-8933

Maria Streck, MD
Ben Sawyer Primary Care
1440 Ben Sawyer Blvd, Ste 1109
Mt Pleasant, SC 29464
Office: 843-876-8333
Fax: 843-876-8330

Upstate (PRISMA): York, Chester, Union, Laurens, Greenwood, McCormick, Abbeville, Anderson, Oconee, Pickens, Greenville, Spartanburg, Cherokee

Joshua Brownlee, MD
Pediatric Infectious Diseases
PRISMA Health Patewood Hospital
200 Patewood Dr. Ste A200
Greenville, SC 29615
Office: 864-454-5130
FAX: 864-454-5698

All Counties/Midstate: Clarendon, Chesterfield, Lee, Sumter, Calhoun, Orangeburg, Bamberg, Allendale, Barnwell, Aiken, Lexington, Richland, Kershaw, Lancaster, Fairfield, Newberry, Saluda, Edgefield, Aiken

Jennifer Joi Jaroscak, MD
Pediatric Hematology/Oncology
MUSC Shawn Jenkins Children's Hospital
10 McClennan Banks Drive
Charleston, SC 29425
Office: (843) 876-0444 or (843) 792-2957

Resources for Pediatric Neurologic Disorders

Rebecca K Lehman, MD
Prisma Health Pediatric Neurology
9 Richland Medical Park Drive, Suite 110
Columbia, SC 29203
Phone: 803-434-7961
Fax: 803-758-0134

Rebecca K Lehman, MD
Prisma Health Pediatric Neurology-Orangeburg
1724 Village Park Drive
Orangeburg, SC 29118
Phone: 803-434-7961
Fax: 803-758-0134

Addie Stark Hunnicutt, MD
Prisma Health Pediatric Neurology
200 Patewood Drive, Suite A350
Greenville, SC 29615
Phone: 864-454-5110
Fax: 864-241-9206

Addie Stark Hunnicutt, MD
Pediatric Specialties-Spartanburg
249 North Grove Medical Park Drive, Suite 200
Spartanburg, SC 29303
Phone: 864-454-5110
Fax: 864-241-9206

Neena Champaigne, MD, FACMG
Division Chief, Pediatric Genetics
Medical University of South Carolina
135 Rutledge Avenue
Charleston, SC 29425
Phone: 843-792-6735

Elizabeth Sekul MD
Augusta University Neuroscience
1446 Harper Street
Augusta, GA 30912
Phone: (706) 446-5455
Fax 706-721-3377

NOTE: This list is not inclusive of all pediatric providers in South Carolina and Georgia.

Krabbe Disease/ List of providers at GGC, MUSC, and Duke*

Richard Schroer, MD
Sr. Clinical/Biochemical Geneticist
Greenwood Genetic Center (GGC)
3520 W Montague Ave, Ste 103
N. Charleston, SC 29418

Office: 877-679-0927
Fax: 866-676-9881
Alternate Fax: 843-735-5097

Emily Black, MD,
Clinical Geneticist
Greenwood Genetic Center (GGC)
14 Edgewood Drive
Greenville, SC 29605

Office: 864-250-7944
Fax: 864-250-9582

Neena Champaigne, MD
Clinical/Biochemical Geneticist
MUSC Department of Pediatrics
Division of Genetics
Rutledge Tower, Third Floor
135 Rutledge Avenue, MSC 567
Charleston, SC 29425

Office: 843-792-6735
Fax: 843-876-1518

Kristen Ann Lancaster, MD, FAAP
Pediatric Geneticist
MUSC Children's Health Genetics
Summey Medical Pavilion
2250 Mall Drive
North Charleston, SC 29406

Office: 843-876-1417
Fax: 843-792-1435

Jennifer Joi Jaroscak, MD
MUSC Shawn Jenkins Children's Hospital
Pediatric Hematopoietic Stem Cell Transplantation
10 McClennan Banks Drive
Charleston, SC 29425
Phone: (843) 792-2957

Lauren Thompson, DO, FACMG
Clinical and Medical Biochemical Geneticist
Medical University of South Carolina
135 Rutledge Avenue
Charleston, SC 29425
Phone: 843-876-1229
Fax: 843-985-9744

***Joanne Kurtzberg, MD**
Duke University Medical Center
Director, Pediatric Blood and Marrow Transplant Program
Co-Director, Stem Cell Transplant Laboratory

Duke Blood Cancer Center
2400 Pratt Street, Room 9026
Durham, NC 27705
Office Phone: 919-668-1102 or
919-668-1119, Attention Tina Hayes

NOTE: This list is not inclusive of all prospective providers in North and/or South Carolina.

South Carolina Newborn Screening Laws and Regulations

Laws

Neonatal Screening for Inborn Metabolic Errors and Hemoglobinopathies, Sections 44-37-30 and 44-37-35 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 Public Health, 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.

(D)(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.

(E)(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 ($50,000) or imprisoned for not more than three years."

(H) The department shall establish the Newborn Screening Advisory Committee to review the feasibility and advisability of including additional metabolic, genetic, and congenital disorders in the neonatal testing conducted pursuant to this section. The committee must be multidisciplinary and composed of members deemed appropriate by the department.

HISTORY: 1962 Code Section 32-655.1; 1965 (54) 641; 1978 Act No. 514, Section 1; 1986 Act No. 484, Section 1; 1994 Act No. 418, Section 1; 2002 Act No. 225, Section 2, eff May 1, 2002; 2019 Act No. 55 (H.3036), Section 2, eff May 16, 2019.

Effect of Amendment:

2019 Act No. 55, Section 2, added (H), establishing the Newborn Screening Advisory Committee.

Proviso 34.37. (DPH: 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.

SECTION 44-37-35. Required neonatal genetic testing.

(A) Neonatal testing conducted pursuant to Section 44-37-30 must include testing for the following:

(1) Krabbe disease;

(2) Pompe disease; and

(3) Hurler syndrome (MPS I).

(B) The department shall require additional lysosomal storage disorders to be tested upon the recommendations of the Newborn Screening Advisory Committee and in accordance with Section 44-37-30 pursuant to a duly promulgated regulation as testing for such disorders becomes available.

HISTORY: 2019 Act No. 55 (H.3036), Section 1, eff May 16, 2019.

Editor's Note

2019 Act No. 55, Section 3, provides as follows:

"*SECTION 3. This act takes effect upon approval by the Governor. Implementation of the act is contingent upon available funding from public sources.*"

South Carolina Regulation 61-80

South Carolina Department of Public Health, Regulation 61-80
Neonatal Screening for Inborn Metabolic Errors and Hemoglobinopathies.

Contents:

Section A. Purpose and Scope
Section B. Definitions
Section C. Testing
Section D. Collection of Specimen
Section E. Assurance of Diagnosis and Follow-Up
Section F. Storage of Specimen
Section G. Use of Stored Specimen
Section H. Forms
Section I. Enforcement Provision

Appendix A. Religious Objection Form: DPH 1804, Newborn Screening Program, Parental Statement of Religious Objection

Appendix B. Information Release Form: DPH 1878, Consent to Release Information Relative to Newborn Screening for Inborn Metabolic Errors and Hemoglobinopathies

Appendix C. Blood Sample Storage Options Form: DPH 1812, Blood Sample Storage Options, Screening of Inborn Metabolic Errors and Hemoglobinopathies

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 Public Health 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 Public Health.

6. Laboratory--shall mean the South Carolina Department of Public Health Public Health Laboratory.

7. Bureau of Maternal and Child Health--shall mean an organizational unit of the South Carolina Department of Public Health.

8. Official Departmental Instructions--shall mean detailed instructions approved by the Commissioner of the South Carolina Department of Public Health 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 (CLIA) 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 Specimens

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: DPH 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 Public Health 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 Public Health, 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: DPH 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 Public Health 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.

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

APPENDIX C: Blood Sample Storage Options Form: DPH 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 Public Health 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 Public Health, 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 Public Health 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 Public Health, 2600 Bull Street, Columbia, SC, 29201, of address or name changes.

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

Name: _____ Date: _____

DPH can store your baby's blood sample for special study. Studies help DPH find out new information about diseases. If a study finds something in your child's blood sample that can help your child, DPH 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.

DPH 1804, Parental Statement of Religious Objection Instructions

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: SC DPH, Newborn Screening Program, 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.

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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.

Other Resources

HRSA's Maternal and Child Health Bureau (MCHB) **Newborn Screening Information Center** for Newborn Screening Programs, provides up-to-date information and resources about newborn screening. All newborn babies in the United States are screened for rare but serious health conditions. Screening helps identify babies with these conditions before they become sick.

https://newbornscreening.hrsa.gov/

https://mchb.hrsa.gov/programs/newborn-screening

HRSA's **Advisory Committee on Heritable Disorders in Newborns and Children** (ACHDNC) was established under the Public Health Service Act, Title XI, § 1109 (42 U.S.C. 300b-10), as amended by the Newborn Screening Saves Lives Reauthorization Act of 2021:

https://www.marchofdimes.org/sites/default/files/2022-11/Newborn-screening-2021-Fact-Sheet.pdf

The ACHDNC advises the Secretary of the U.S. Department of Health and Human Services (HHS) on the most appropriate application of universal newborn screening tests, technologies, policies, guidelines, and standards:

https://www.hrsa.gov/advisory-committees/heritable-disorders/index.html

American College of Medical Genetics and Genomics (ACMG) ACT sheets provide immediate steps for physicians to take upon receiving an abnormal screen for infants in their practice. https://www.acmg.net

Baby's First Test provides current educational and family support and services information, materials, and resources about newborn screening at the local, state, and national levels and serves as the Clearinghouse for newborn screening information. https://www.babysfirsttest.org

The March of Dimes leads the fight for the health of all mothers and babies and has helped millions of babies survive and thrive:

https://www.marchofdimes.org/baby/newborn-screening-tests-for-your-baby.aspx

The Centers for Disease Control (CDC) and Prevention, Newborn Screening Portal serves as the national resource for newborn screening activities and data to prevent death or disability and enable children to reach their full potential. https://www.cdc.gov/newbornscreening

The Clinical and Laboratory Standards Institute (CLSI) provides resources on specimen collection and newborn screening lab tests. https://clsi.org

Sickle Cell Disease (SCD) is a complex group of inherited blood disorders associated with debilitating pain and complications that can affect every organ in the body. Learn about the types of sickle cell disease, diagnosis, treatment, and more resources here:

https://www.cdc.gov/sickle-cell/about/index.html

The mission of the **Cystic Fibrosis Foundation** is to cure cystic fibrosis and to provide all people with the disease an opportunity to lead full, productive lives. https://www.cff.org

The Immune Deficiency Foundation (IDF) is a national nonprofit patient organization dedicated to improving the diagnosis, treatment, and quality of life of persons with Primary immunodeficiency diseases (PI) such as Severe Combined Immune Deficiency (SCID) and others, through advocacy, education, and research:

https://primaryimmune.org

National Organization for Rare Disorders (NORD) is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them:

https://rarediseases.org

Acknowledgements

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

ACMG ACT Sheets and Confirmatory Algorithms
https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets_and_Algorithms.aspx

Newborn Screening, Oregon Health Authority
https://www.oregon.gov/oha/PH/LaboratoryServices/NewbornScreening/Pages/index.aspx

Health Professionals Guide to Newborn Screening, Wisconsin Newborn Screening Laboratory
http://www.slh.wisc.edu/clinical/newborn/health-care-professionals-guide/

New England Consortium of Metabolic Programs at Children's Hospital of Boston
https://www.newenglandconsortium.org/