

## Collection and Handling of Blood Specimens

Specimens shall be collected by a heel puncture using a disposable blood collection device. **Cord blood is not acceptable for newborn screening in SC and should never be applied on the filter paper collection form.** The blood should be allowed to begin flowing before attempting to fill the filter paper circles. It is essential that one drop of blood is applied to each circle and that it saturates the filter paper to the reverse side, giving an adequate homogeneous sample. Successive drops of blood should never be layered on the filter paper. The filter paper should not be saturated from both sides. Capillary tubes should not be routinely used for specimen collection. If capillary tubes are used to drop the blood, care must be taken to ensure that the end of the tube does not touch the filter paper since this will abrade the filter paper.

The specimen should be allowed to air dry in a horizontal position for at least 4 hours before mailing. Neither side of the blood spots should touch a surface. Specimens should be kept away from direct sunlight and heat sources during the drying process. Heat must not be used to facilitate drying. All specimens must be sent to the laboratory within 24 hours of collection. Staff should NOT hold specimens for large mailings. **SPECIMENS SHOULD NOT BE PLACED IN PLASTIC BAGS PRIOR TO MAILING!**

Delay in sending the specimen can cause deterioration of the sample, may affect the test result and delays testing. The phone number and address for the Newborn Screening Laboratory is on the top left hand corner of the collection form.

# Neonatal Screening

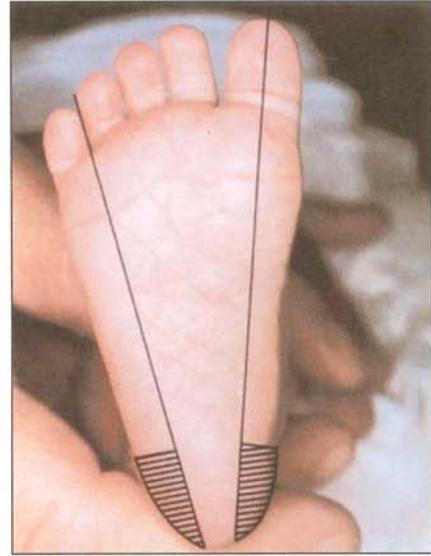
## Blood Specimen Collection and Handling Procedure



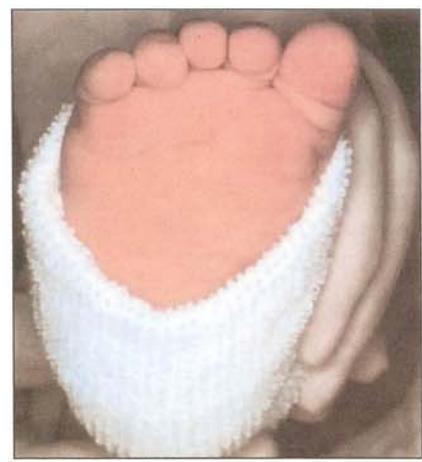
**1** Equipment: sterile lancet with tip approximately 2.0 mm – sterile alcohol prep, sterile gauze pads, soft cloth, blood collection form, gloves.



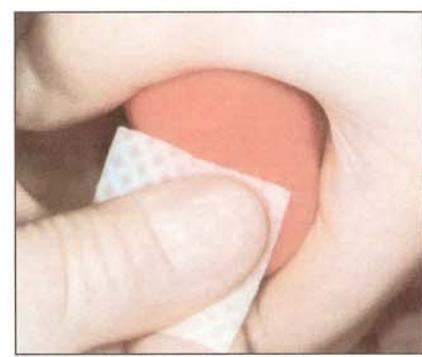
**2** Complete ALL information. Do not contaminate filter paper circles by allowing the circles to come into contact with spillage or by touching before or after blood collection. Keep "SUBMITTER COPY" if applicable.



**3** Hatched area ( // ) indicates safe areas for puncture site.

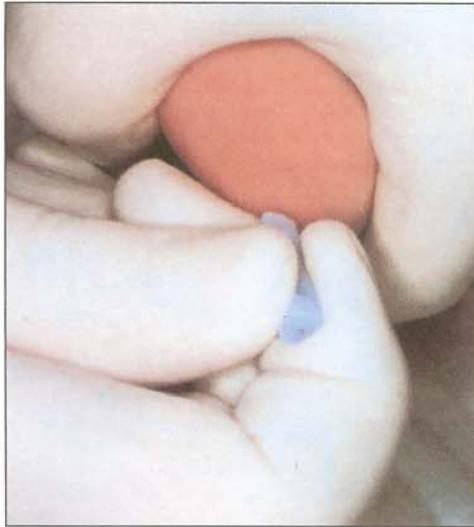


**4** Warm site with soft cloth, moistened with warmwater up to 41°C, for three to five minutes.

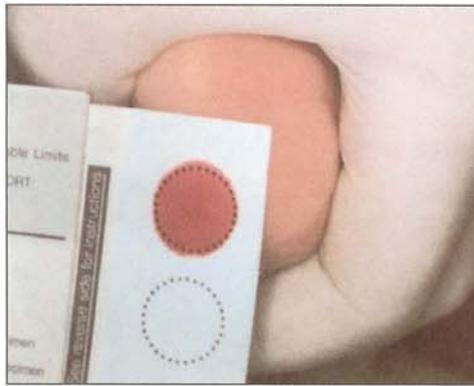


**5** Cleanse site with alcohol prep. Wipe DRY with sterile gauze pad.





**6** Puncture heel. Wipe away first blood drop with sterile gauze pad. Allow another LARGE blood drop to form.



**7** Lightly touch filter paper to LARGE blood drop. Allow blood to soak through and completely fill circle with SINGLE application of LARGE blood drop. (To enhance blood flow, VERY GENTLE intermittent pressure may be applied to the area surrounding the puncture site). Apply blood to one side of filter paper only.



**8** Fill remaining circles in the same manner as step 7, with successive blood drops. If blood flow is diminished, repeat steps 5 through 7. Care of skin puncture site should be consistent with your institution's procedures.



**9** Dry blood spots on a dry, clean, flat, non-absorbent surface for a minimum of four hours.



**10** Mail completed form to testing laboratory within 24 hours of collection.

Information provided by The New York State Department of Health.

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 Japan – Whatman Japan KK • Tel: +81 (0) 3 5215 1242 • Fax: +81 (0) 3 5215 1246 • E-mail: japaninfo@whatman.com  
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# Simple Spot Check

Whatman®

## Valid specimen:



Allow a sufficient quantity of blood to soak through to completely fill the preprinted circle on the filter paper. Fill all required circles with blood. Do not layer successive drops of blood or apply blood more than once in the same collection circle. Avoid touching or smearing spots.

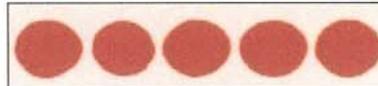
## Invalid specimen:



1. Specimen quantity insufficient for testing.



2. Specimen appears scratched or abraded.



3. Specimen not dry before mailing.



4. Specimen appears supersaturated.



5. Specimen appears diluted, discolored or contaminated.



6. Specimen exhibits serum rings.



7. Specimen appears clotted or layered.



8. No blood.

## Possible causes:

- Removing filter paper before blood has completely filled circle or before blood has soaked through to second side.
- Applying blood to filter paper with a capillary tube.
- Touching filter paper before or after blood specimen collection with gloved or ungloved hands, hand lotion, etc.
- Allowing filter paper to come in contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood specimen collection.
- Applying blood with a capillary tube or other device.
- Mailing specimen before drying for a minimum of four hours.
- Applying excess blood to filter paper, usually with a device.
- Applying blood to both sides of filter paper.
- Squeezing or "milking" of area surrounding the puncture site.
- Allowing filter paper to come in contact with gloved or ungloved hands or substances such as alcohol, formula, antiseptic solutions, water, hand lotion or powder, etc., either before or after blood specimen collection.
- Exposing blood spots to direct heat.
- Not wiping alcohol from puncture site before making skin puncture.
- Allowing filter paper to come in contact with alcohol, hand lotion, etc.
- Squeezing area surrounding puncture site excessively.
- Drying specimen improperly.
- Applying blood to filter paper with a capillary tube.
- Touching the same circle on filter paper to blood drop several times.
- Filling circle on both sides of filter paper.
- Failure to obtain blood specimen.

Information provided by The New York State Department of Health.

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 Whatman GmbH • Tel: +49 (0) 5561 791 0 • Fax: +49 (0) 5561 791 536 • E-mail: information@whatman.com  
 Japan – Whatman Japan KK • Tel: +81 (0) 3 5215 1242 • Fax: +81 (0) 3 5215 1246 • E-mail: japaninfo@whatman.com  
 Asia Pacific – Whatman Asia Pacific Pte Ltd • Tel: +65 6534 0138 • Fax: +65 6534 2166 • E-mail: wap@whatman.com

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## Special Circumstances

### Transfusion

If the infant is to undergo a transfusion of blood products, a blood specimen should be obtained prior to the transfusion. If this is not possible, collect the specimen and mark the form “Transfused Yes”, indicating the date of the most recent or last transfusion. Infants who receive transfusions should have a repeat hemoglobinopathy screening 2 months after the date of the last transfusion.

If the date of the last transfusion is unknown, put the date of hospital discharge on the collection form next to the “Transfused Yes” box. **DO NOT MARK THE “TRANSFUSED YES” BOX IF THE TRANSFUSION TOOK PLACE 2 MONTHS BEFORE THE SPECIMEN COLLECTION DATE.**

### Premature Infants

In premature infants, the results can be falsely abnormal due to system immaturity or the stress of prematurity. To assure the best specimen:

If the premature infant is receiving **only IV fluids or total parenteral nutrition/hyperalimentation**, the blood specimen may be collected as long as the infant is at least **24 hours of age and the lab slip is clearly marked “TPN or NPO.”**

**All premature infants shall receive their initial screening by 7 days of age regardless of their health status.**

Another specimen should be collected only if a report is abnormal, if a specimen is unacceptable or if the infant is initially screened before 24 hours of age. For example, if the screening result for congenital hypothyroidism is abnormal, the collection form will be marked "T4 /TSH" in the section labeled “Repeat Individual Test Only”. Routine repeat testing for premature infants is not necessary if the screening test results are normal and the infant is at least 24 hours of age at the time of specimen collection.

### Infants Being Placed for Adoption

The blood specimen should be collected from the infant as previously instructed. The adoption agency/lawyer's name, address and phone number should be used in place of the mother's information if the adoption agency/lawyer is considered the legal guardian. If the infant's legal name is not known, “Girl Adoption” or “Boy Adoption” should be used.

### Infants Born Outside of a Hospital

It is the responsibility of the person in attendance at a birth outside of a hospital to obtain a specimen for newborn screening. If the specimen has not been collected by the third day of life the person in attendance at the birth should contact Women and Children's Services for assistance.

### Testing Older Infants and Children

Analyte ranges for newborn screening are based upon expected results for infants tested within the first several weeks of life. **For that reason, the Laboratory will not process specimens for newborn screening of infants and children older than six months of age.** Health care providers

can contact staff in Women and Children's Services for assistance in locating another laboratory for that service. The Laboratory will process specimens from older infants, children and adults for monitoring of certain metabolic disorders (such as PHE and TYR for PKU) and for hemoglobinopathies.

### **Unsatisfactory Specimens**

The Laboratory receives some specimens that are considered unsatisfactory for officially reporting test results. While the Laboratory does test unsatisfactory specimens for extreme values when possible, improper collection and handling may compromise the accuracy of the test results. This delays the screening and diagnosis of a potentially affected infant and requires that a repeat specimen be submitted as soon as possible. If the testing of an unsatisfactory specimen yields an abnormal test result, the physician of record will be notified by Women and Children's Services. Depending upon the severity of the abnormal test result, the attending physician will receive either phone notification or mail notification. **The official laboratory result will still indicate that the specimen was unsatisfactory and that repeat specimen collection is necessary.**

### **Infants who Exhibit Clinical Signs of a Newborn Screening Disorder**

If signs and symptoms of one of the newborn screening disorders are evident clinically, the physician should proceed to diagnostic testing, pending the results of the screening or in spite of the results of the screening.

#### **▶▶ If the results of the newborn screening are pending:**

For any of the disorders, but especially those in which the metabolite accumulation is dangerous, treat the infant as if he or she has the disorder. Consultation with the appropriate medical specialist is indicated. For other disorders, contact the appropriate medical specialist for assistance with rapid diagnosis and institution of treatment as indicated.

#### **▶▶ If the results of the newborn screening are "normal":**

If clinical symptoms suggest one of the newborn screening disorders, the physician should treat the infant as if he or she has the disorder. Another specimen for newborn screening should be collected and the appropriate medical specialist should be contacted for assistance with further evaluation of the infant.

### **Infants with an Affected Close Relative**

Most of the disorders on the newborn screening test panel are genetic disorders. Prenatal diagnosis is possible for many of these disorders. If prenatal testing determines that the fetus is affected, arrangements should be made for delivery at the appropriate level hospital and treatment should be initiated as soon as possible. A specimen for newborn screening must still be collected to test for the other disorders on the panel. If prenatal testing predicts an unaffected fetus, a specimen must still be collected to confirm the results of the prenatal testing and to provide screening for the other disorders on the panel.

### **Report Related Issues**

Reports are mailed to the physician who is documented on the collection form and to the facility where the specimen is collected. **If a report is not received by the time of the infant's first visit, practices can call Linda Baker at (803) 898-0593 or Kathy Tomashitis at (803) 898-0619 for a verbal or faxed report.**

**Physicians and other providers should request a sender number from the Laboratory in order to ensure timely receipt of newborn screening reports. Requests should be made to Earleen Wilson at (803) 896-0966.** Practices may choose to receive reports by individual physician. To do so, the physician's sender number and name should be documented in the space under the "baby's doctor" section of the collection form. The physician's license number preceded by the letter "M" is used as the sender number.

They may also opt to obtain a group sender number from the Laboratory. Group sender numbers are assigned by the Laboratory and include the letter "G" before the number. However, only one address can be associated with a sender number, so if a practice has multiple locations, more than one sender number will be needed to route reports to the correct location.

If there is no sender number associated with the name of the physician as documented on the collection form, then the Laboratory will look up the physician's name in the on-line directory of the Department of Labor, Licensing and Regulation and create a sender number for the physician. **This process cannot be done if there is more than one physician with the same name or if the first and last name of the physician is not clearly documented on the collection form.**

**To keep the database as accurate as possible, all address changes must be sent in writing to the Laboratory.** It is acceptable to fax this information to the Laboratory at (803) 896-0983 or to email this information to Earleen Wilson at [wilsoney@dhec.sc.gov](mailto:wilsoney@dhec.sc.gov).

# Samples of Typical Newborn Screening Reports

## Report with Normal Screening Results:

	<b>S. C. DEPARTMENT OF HEALTH AND ENVIRONMENTAL CONTROL</b> <b>BUREAU OF LABORATORIES</b>	Submitter # <hr/> Physician #
TELEPHONE: (803) 896-0800	8231 PARKLANE ROAD COLUMBIA, S.C. 29223	CLIA #42-D0658606
<b>CONFIDENTIAL LABORATORY REPORT</b>		
SUBMITTED BY: PHYSICIAN: ADDRESS:		SPECIMEN #: TYPE: <b>Blood Card Specimen</b>
INFANT NAME: BIRTH:		COLLECTED: RECEIVED:
MED RECORD #:	RACE:	SEX:
PARENT NAME: ADDRESS: PHONE:	FEED TYPE:  PRINT DATE: 08/23/2007	

<u>Assay</u>	<u>Result</u>	<u>Value</u>	<u>Units</u>	<u>E x p e c t e d</u>
17-OHP	Normal		ng/mL	<75, Present Wgt >= 2500 g <125, Present Wgt <2500 g
Amino Acid	Normal Profile		µM	Normal Profile
Acylcarnitine	Normal Profile		µM	Normal Profile
Biotinidase	Normal		ERU	> 10
GALT	Normal		µM	> 60
GAO	Normal		mg/dL	< 10
HB	No Variant Detected			No Variant Detected
IRT	Normal		ng/mL blood	<100, Initial; <70 Repeat
SUAC	Normal		uM	<3
Test performed at Mayo Clinic, Rochester, MN				
T4	Normal		µg/dL	>7, <=7 days old; >4, >=8 days old
TSH	Normal		µIU/mL	< 23

**17-OHP (17-Hydroxyprogesterone):** marker for Congenital Adrenal Hyperplasia

**Amino Acid:** profile of several essential amino acids to detect Amino Acid metabolic disorders, including Phenylketonuria (PKU)

**Acylcarnitine:** profile of short to very long chain compounds to detect Fatty Acid Oxidation & certain Organic Acid Metabolism disorders.

**Biotinidase:** marker for biotinidase deficiency

**GALT (Galactose Uridyl Transferase) & GAO (Total Galactose) :** markers for Galactosemia

**HB (Hemoglobin) · No Variant Detected:** only known normal hemoglobin(s) were detected on screening

**IRT (Immunoreactive Trypsinogen):** marker for Cystic Fibrosis

**SUAC (Succinyl Acetone):** marker for Tyrosinemia Type I (test performed when Tyrosine >180 uM)

**T4 (Thyroxine) & TSH (Thyroid Stimulating Hormone):** markers for Congenital Hypothyroidism

**NOTE:** The purpose of newborn screening is to identify infants at risk & in need of more definite testing. As with any lab test, both false positive & false negative results are possible. Initial screening test results are insufficient information upon which to base definitive diagnosis or treatment.

**Report Sent When Amino Acid Profile is Abnormal**



S. C. DEPARTMENT OF HEALTH AND ENVIRONMENTAL CONTROL  
 BUREAU OF LABORATORIES  
 8231 PARKLANE ROAD  
 COLUMBIA, S.C. 29223  
 TELEPHONE: (803) 896-0800

LABORATORY REPORT

CLIA #42-D0658606

**SENDER:** **PHYSICIAN:**  
**NAME:** **DATE OF BIRTH:**  
**MOTHER'S NAME:** **SEX:**  
**SPECIMEN NUMBER:** **RACE:**  
**SPECIMEN TYPE:** Blood Spot **DATE COLLECTED:**  
**DATE RECEIVED:** **DATE OF REPORT:**

Amino Acid	Result (µM)	Expected (µM)
Citrulline		< 76
Leucine + Isoleucine		< 343
Methionine		< 75
Phenylalanine		< 181
Tyrosine		< 481
Valine		< 325
Leucine + Isoleucine /Phenylalanine		< 4.00
Methionine/Phenylalanine		< 1.50
Phenylalanine/Tyrosine		< 3.00
Valine/ Phenylalanine		< 3.00

THE NEWBORN SCREENING TESTING IDENTIFIED ONE OR MORE ABNORMAL AMINO ACID LEVELS THAT ARE LISTED ABOVE IN ***Bold/Italics***  
 THE RESULTS MAY BE INDICATIVE OF AN AMINO ACID DISORDER.

IF YOU HAVE ANY QUESTIONS, PLEASE CONTACT DR. BRYANT FORTNER (NBS MEDICAL CONSULTANT, 803-898-0362 OR E-MAIL HIM AT [fortnebr@dhec.sc.gov](mailto:fortnebr@dhec.sc.gov)).

\*\*\*PLEASE REPEAT THE NEWBORN SCREENING.\*\*

IT IS IMPERATIVE THAT THIS REPEAT SCREENING BE COMPLETED AS SOON AS POSSIBLE AND THE DRIED BLOOD SPOTS BE SENT TO THE LABORATORY.

**Report Sent When Acyl Carnitine Profile is Abnormal:**



S. C. DEPARTMENT OF HEALTH AND ENVIRONMENTAL CONTROL  
 BUREAU OF LABORATORIES  
 8231 PARKLANE ROAD  
 COLUMBIA, S.C. 29223  
 TELEPHONE: (803) 896-0800

LABORATORY REPORT

CLIA #42-D0658606

**SENDER:** **PHYSICIAN:**  
**NAME:** **DATE OF BIRTH:**  
**MOTHER'S NAME:** **SEX:**  
**SPECIMEN NUMBER:** **RACE:**  
**SPECIMEN TYPE:** Blood Spot **DATE COLLECTED:**  
**DATE RECEIVED:** **DATE OF REPORT:**

Acylcarnitine	Result (µM)	Expected (µM)
Free carnitine		> 5.00
C2 (Acetyl carnitine)		> 2.00
C3 (Propionyl carnitine)		< 4.81
C4 (Butyryl carnitine)		< 1.86
C5:1 (Tiglyl carnitine)		< 0.33
C5 (Isovaleryl carnitine)		< 1.00
C4-OH (3-Hydroxy-butyryl carnitine)		< 0.75
C6 (Hexanoyl carnitine)		< 0.62
C5-OH (3-hydroxy-isovaleryl carnitine)		< 1.18
C8 (Octanoyl carnitine)		< 0.72
C3-DC (Malonyl carnitine)		< 0.35
C10:2 (Decadienoyl carnitine)		< 0.32
C10:1 (Decenoyl carnitine)		< 0.30
C10 (Decanoyl carnitine)		< 0.55
C5-DC (Glutaryl carnitine)		< 0.32
C12:1 (Dodecenoyl carnitine)		< 0.49
C12 (Dodecanoyl carnitine)		< 0.89
C6-DC (Adipyl carnitine)		< 0.55
C14:2 (Tetradecadienoyl carnitine)		< 0.15
C14:1 (Tetradecenoyl carnitine)		< 0.64
C14 (Myristoyl carnitine)		< 0.84
C16 (Palmitoyl carnitine)		< 9.73
C16-OH (3-hydroxyl Palmitoyl carnitine)		< 0.32
C18:2 (Linoleyl carnitine)		< 0.60
C18:1 (Olelyl carnitine)		< 3.99
C18:1-OH (3-hydroxyl Olelyl carnitine)		< 0.30
C3/C2		< 0.20
C3/C16		< 2.00
C8/C10		< 3.00
C14:1/C12:1		< 3.00
C14:1/C16		< 0.20

THE NEWBORN SCREENING TESTING IDENTIFIED ONE OR MORE ABNORMAL ACYLCARNITINE LEVELS THAT ARE LISTED ABOVE IN **Bold/Italics**. THE RESULTS MAY BE INDICATIVE OF A FATTY ACID OXIDATION OR ORGANIC ACID DISORDER. **IF YOU HAVE ANY QUESTIONS, PLEASE CONTACT DR. BRYANT FORTNER (NBS MEDICAL CONSULTANT, 803-898-0362 OR E-MAIL HIM AT [fortnebr@dhec.sc.gov](mailto:fortnebr@dhec.sc.gov))** \*\*PLEASE REPEAT THE NEWBORN SCREENING.\*\*IT IS IMPERATIVE THAT THIS REPEAT SCREENING BE COMPLETED AS SOON AS POSSIBLE AND THE DRIED BLOOD SPOTS BE SENT TO THE LABORATORY.