

NEWBORN SCREENING PROGRAM HANDBOOK



Utah
Department
of Health

STATE OF UTAH
DIVISION OF COMMUNITY AND FAMILY SERVICES
BUREAU OF CHILDREN WITH SPECIAL HEALTH CARE NEEDS

December 2007

UTAH DEPARTMENT OF HEALTH
NEWBORN SCREENING PROGRAM

KIT ORDERING

Newborn Screening Kits

Fees Effective
July 1, 2007

TWO-PART KIT (First and Second Screen forms)

\$67.00

The two-part kit is the initial form used by the institutions of birth. It is issued to the parent at the hospital/birthplace.

Order two-part kits from: Utah Department of Health Laboratory
ATTN: Chris Peper
46 N Medical Dr
Salt Lake City UT 84113
Phone: (801) 584-8400
Fax: (801) 584-8421

MISCELLANEOUS FORMS

No Charge

Miscellaneous kits are supplied as replacements for inadequate specimens, recall specimens, or for use when the original kit has been lost.

Order miscellaneous forms from: Newborn Screening Program
ATTN: Shelley Morrill
PO Box 144710
Salt Lake City UT 84114-4710
Phone: (801) 584-8256
Fax: (801) 536-0962

UTAH NEWBORN SCREENING

INTRODUCTION

The Utah Newborn Screening handbook is designed to educate local hospital personnel such as nursery and laboratory staff, physicians, midwives, and other health care providers about the requirements for newborn screening in Utah. This handbook includes background information on the importance of newborn screening with specific instructions on completing the forms and submitting blood spot specimens.

The purpose of this handbook is to promote a better understanding of the newborn screening forms and the information entered on them. The quality of the newborn screening data and the ability to identify and locate families and medical providers quickly depends heavily on the correct completion of the forms. Forms sent in with missing or incorrect data or un-testable specimens cannot be processed, thus putting infants in jeopardy. The institution of birth or the midwife/practitioner providing assistance to the mother at the birth is responsible for initiating the newborn screening process.

The Newborn Screening Program uses the information on the form to efficiently locate and identify newborns with abnormal test results, as well as to notify health care providers when blood spot specimens are not adequate for testing.

IMPORTANCE OF NEWBORN SCREENING

Utah State Law UCA 26-10-6 [Appendix A] requires all infants born in Utah be tested.

The scientific, political and social advancements in the United States came together to foster the development of the newborn screening practices. In the 1960's, parent advocacy groups were instrumental in getting legislation passed for prevention of mental retardation. In 1965, Utah State legislators adopted mandatory testing of all newborns for phenylketonuria (PKU) and other metabolic diseases that might result in brain damage or death.

THE UTAH STATE DEPARTMENT OF HEALTH

The Utah Department of Health began managing the newborn screening process in 1979. At that time phenylketonuria (PKU), congenital hypothyroidism, and galactosemia were the diseases identified through screening. As of January 2006 the Newborn Screening Program has expanded to include screening babies for biotinidase, congenital hypothyroidism, congenital adrenal hyperplasia, hemoglobinopathies, galactosemia, disorders of amino acid metabolism (including PKU), disorders of organic acid metabolism, and disorders of fatty acid metabolism.

The Newborn Screening Program is administered under the laws [Statute 26-10-6/Appendix A] and rules [R398-1/Appendix B] of the State of Utah. The Newborn Screening Laboratory at the Utah Department of Health State Laboratory is in charge of the performance of all screening tests. The Newborn Screening Program, as part of the Bureau of Children with Special Health Care Needs, provides follow-up on abnormal specimens, education to the public and health care providers, and assists with identification of all public resources. The State Laboratory and the Newborn Screening Program work together to provide these valuable services to the community.

CONFIDENTIALITY OF NEWBORN SCREENING RECORDS

Newborn Screening personnel protect the information on the newborn screening forms and in the database from unwarranted or indiscriminate disclosure. Records are available only to persons who are authorized access by State Law and supporting rules. Legal safeguards for the confidentiality of records have been strengthened in recent years. Physicians, hospitals, and families are assured that extensive legal and administrative measures are used to protect individuals from unauthorized disclosure of personal information.

MEDICAL HOME



The Newborn Screening Program supports and encourages the “Medical Home” concept for all infants. The health care provider for a baby must be identified at birth. These providers will be contacted for any follow up needs.

What is a Medical Home?

A **medical home** is not a building, house or hospital, but rather a family-centered approach to providing health care in a high quality and cost effective manner. Primary care providers, families, and allied health care professionals act as *partners* to identify and access all medical and non-medical services needed by children and families to help them achieve their maximum potential. A medical home includes care that is accessible, family-centered, continuous, comprehensive, coordinated, compassionate, and culturally competent.

The ideal source of a child’s medical home is a primary care pediatrician or family doctor working in partnership with the parents. All children deserve a medical home to provide consistent and personalized care and this relationship may be even more important for children who may have special health care needs. Benefits of a medical home include: increased patient and family satisfaction, establishment of a forum for problem solving, improved coordination of care, efficient use of available resources, increased professional satisfaction, and increased child wellness due to comprehensive care.

(For more information contact Barbara Ward, RN, CSHCN Medical Home Project Coordinator at (801) 584-8584 or e-mail bward@utah.gov)

RESPONSIBILITIES REGARDING NEWBORN SCREENING SPECIMENS

FIRST SCREEN SPECIMEN

Hospital personnel, midwives, and birth attendants must complete the personal data required on the form and collect and submit a testable blood specimen (see Simple Spot Check on page --). Necessary procedures may cut across departmental lines, involving many different people. These procedures, when combined with the current emphasis on reducing the length of stay in hospitals, make it extremely important for one hospital staff member to be given the overall responsibility and authority to request and obtain the cooperation needed. Responsibilities regarding the first screening specimen:

- Develop efficient procedures for prompt assignment of a Newborn Screening Kit, preparation of data, and collection of filter paper blood spot specimen on every newborn. [R398-1]
- Collect and record all information requested on the data portion of the first screen form: infant name, sex, feeding, adoption, transfusion, date of birth, mother's name, address and phone number, mother's date of birth, and medical home/health care provider information. If the baby is being adopted, an identifying name and a contact person must be included. [R398-1-8 (e)]
- Prepare a legible form; make certain every item is complete and correct. Print in block, capital letters using black ink. Forms with missing information cannot be processed. *Completion of the second screen form at this time is discouraged as many items change between collection of the first and second specimens.*
- Collect an appropriate blood spot specimen using the heel-stick method. [R398-1-8] *The person drawing the specimen must complete the 'Specimen Collection Date' on the screening form, without which the specimen CANNOT be processed.* Use of capillary tubes to transfer blood to the filter paper is not recommended. Capillary tubes tend to roughen the filter paper and cause over absorption.
- Timing of collection: specimen should be drawn between 48 hours and five days of age, however there may be exceptions. [R398-1-5] *Results are based on the age of the infant at the time the specimen was drawn.*
- If original first screen card is unusable (contaminated, destroyed, blood specimen is inadequate, etc.), *it is not necessary to use a new 2-part kit and number.* Use a Miscellaneous Form. Remove bar code labels from the back of the original first and place them over the miscellaneous numbers, or cross out the miscellaneous numbers and write the original number in each place.
- If possible, collect the specimen prior to a transfusion. [R398-1-5 (2)] This may necessitate drawing it before 48 hours of age.
- Allow the specimen to dry horizontally at room temperature for 3 hours.
- Transport specimen *within 24 hours of collection* to the Newborn Screening Lab. Use of a courier is highly recommended to decrease delay in receipt and testing of the specimen. [R398-1-8 2)]
- To meet postal requirements, fold the cardboard flap over the dried blood spots before sending the specimen.
- Educate the family regarding the required screening, which disorders are screened, and how to obtain the second screen collection. [R398-1-6]
- Develop efficient procedures for prompt collection of the filter paper blood spot specimen on the newborn whose first specimen was determined to be abnormal [R398-1-9] or unsatisfactory for testing (could not test). [R398-1-10]

Instructions:

1. Place on the Delivery Record
2. Place on Newborn Hearing Record
3. Use for Newborn Screening Log.
4. Use for Medical Record Chart.
5. Use for Immunization record.

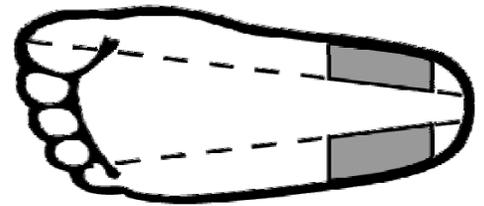


SECOND SCREEN SPECIMEN

Utah law requires that all newborns have a second specimen drawn between 7 and 28 days of age. These specimens are usually collected during the first visit to the medical home/health care provider. Office personnel, clinic personnel and midwives must assemble and record the personal data to be entered onto the forms and must collect and submit a testable blood specimen (see Simple Spot Check on page -). Responsibilities regarding the second screening specimen:

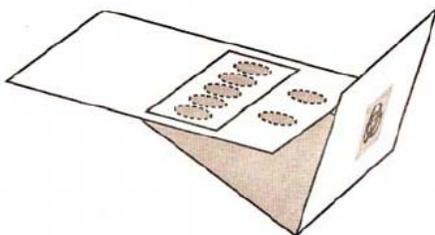
- Develop efficient procedures for preparation of data and collection of filter paper blood spot specimen on every newborn. [R398-1]
- Collect and record all information requested on the data portion of the second screen form: infant name, sex, feeding, adoption, transfusion (if applicable), date of birth, mother's name, address, and phone number, mother's date of birth, and medical home/health care provider information. If the baby was adopted, identifying information needs to be included to link the first and second specimens. [R398-1-8 (e)]
- Prepare a legible form; making certain that every item is completed and correct. Print in block, capital letters using black ink. Forms with missing information cannot be processed.

- Collect an appropriate blood spot specimen using the heel-stick method. *The person who is drawing the specimen must complete the 'Specimen Collection Date' on the form, without which the specimen CANNOT be processed.* Use of capillary tubes to transfer blood to the filter paper is not recommended. Capillary tubes tend to roughen the filter paper and cause over absorption. [R398-1-8]



Draw specimen from shaded area using heel-stick method.

- Timing of collection: specimen should be collected between 7 and 28 days of age, however there may be exceptions. [R398-1-7] *Results are based on the age of the infant at the time the specimen was drawn.*
- If possible, collect the specimen prior to a transfusion. [R398-1-5 (2)] If transfusion has been given, collect the specimen 7 - 10 days after the transfusion.
- Allow specimen to dry horizontally at room temperature for 3 hours, using designated drying rack.



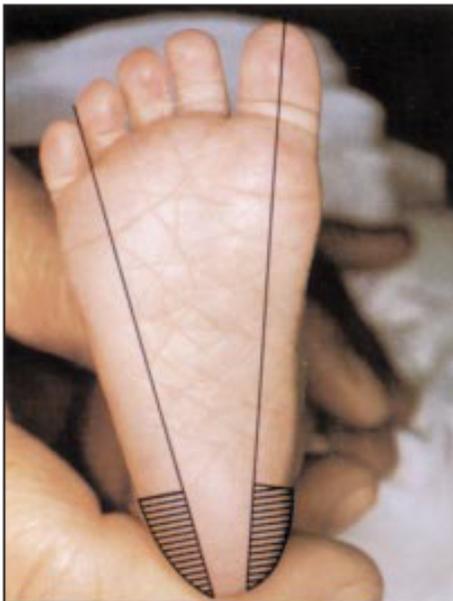
- Transport specimen *within 24 hours of collection* to the Newborn Screening Lab. Using a courier is highly recommended to decrease delay in receipt and testing of specimen. [R398-1-8 (2)]
- To meet postal requirements, fold the cardboard flap over the dried blood spots before mailing the specimen.
- Develop efficient procedures for prompt collection of filter paper blood spot specimen on the newborn whose specimen was determined to be abnormal [R398-1-9] or unsatisfactory for testing (could not test). [R398-1-10]



1 Equipment: sterile lancet with tip approximately 2.0 mm, sterile alcohol prep, sterile gauze pads, soft cloth (or disposable heat pack), blood collection form,



2 Complete ALL information. Do not contaminate filter paper circles by allowing the circles to come in contact with spillage or by touching before or after blood collection. Keep front copy "retain for your records."



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

Neonatal Screening

Blood Specimen Collection and Handling Procedure



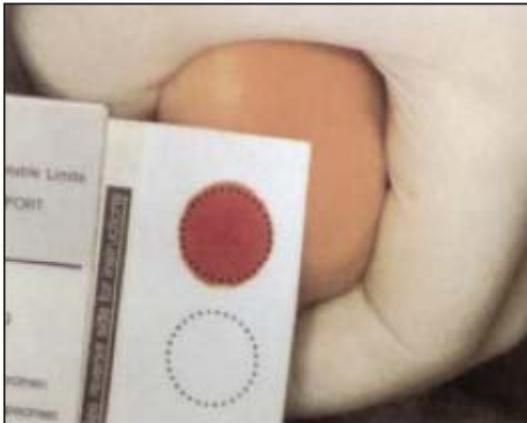
4 Warm site with a soft cloth moistened with warm water (up to 41° C), or a disposable heat pack, for three to five minutes.



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



6 Puncture heel with lancet. Wipe away the first drop of blood with a sterile gauze pad. Allow another LARGE drop of blood to form.



7 Lightly touch filter paper to LARGE blood drop. Allow blood to soak through and completely fill circle with a SINGLE application to LARGE blood drop. To enhance blood flow, VERY GENTLE intermittent pressure may be applied to area surrounding puncture site. Do NOT milk the site. Apply blood to one side of filter paper



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 institutions procedures.



9 Dry blood spots horizontally on drying rack provided by the Newborn Screening Program for at least 3 hours



10 Send completed form to testing laboratory within 24 hours of collection. Use of a courier is recommended. To meet postal requirements, fold cardboard flap over blood specimen before mailing.

Information provided by The New York State Department of Health.

Schleicher & Schuell Inc. • 10 Optical Avenue • Keene N.H. 03431 USA • Tel. (603) 352-3810 • Fax (603) 355-6524 • Internet: <http://www.s-and-s.com> • e-mail: solutions@s-and-s.com
 Schleicher & Schuell GmbH • P.O. Box 4, D-37582 Dassel • Germany • Tel. 49-5561-791-0 • Fax 49-5561-791536 • Internet: <http://www.s-und-s.de> • e-mail: saleadtagcomp@s-und-s.de

GENERAL INSTRUCTIONS FOR SUBMITTING THE NEWBORN SCREENING FORM

The testing for the Newborn Screen is considered to be “moderate or high complexity testing, or both” by the Clinical Laboratory Improvement Amendments of 1988 (CLIA). The Newborn Screening Laboratory must comply with the CLIA regulations (specifically Subpart J which specifies facility standards). This includes CLIA’s minimum required data that must be collected to be in compliance.

The requested data is required in order to identify an infant, family, and health care provider, in compliance with federal regulations. Although the screened disorders are rare, their impact on an individual and family can be tremendous. Correct identification of the infant and rapid notification the health care provider is critical when there are abnormal results. Time is vital in getting the initial evaluation done and treatment started. Nationally, it is the standard of care to identify the infant, complete confirmatory testing, and begin treatment prior to 21 days of age.

- Follow the instructions on the form that comes with the newborn screening kit.
- Print legibly, in block letters, using black ink.
- Avoid abbreviations, except the standard abbreviations used in addresses (use those acceptable from the US Postal Service).
- Verify with the mother (or informant) the spelling of names, especially those that have different spellings for the same sounding name (Smith or Smyth, Gail or Gayle, and Wolf, Wolfe, or Woolf, etc.).
- Use the current form designated by the State of Utah. The filter paper is a Food and Drug Administration (FDA) regulated form. The filter paper has a shelf life of three years, after printing. *All forms are identified with the expiration date.*
- Avoid touching the filter paper at all times. Moisture, body oil, hand lotion, powder from gloves, and even compression of the filter paper fibers can interfere with the absorption of the blood and test results.
- Complete *all* requested data fields. Incomplete forms cannot be processed and the delay may result in poor infant outcomes
- The instruction part of the form is to be removed prior to collecting the specimen. It may be kept for your records.
- Collect a blood specimen by the heel-stick method. Use of capillary tubes to transfer blood to the filter paper is not recommended. Capillary tubes tend to roughen the filter paper and cause over absorption, invalidating results.
- Dry specimen thoroughly, 3-4 hours at room temperature, before mailing. Specimen should be dried in a horizontal position. A rack designed for this purpose is available, free of charge, through the program.
- Submit the specimen to the Newborn Screening Laboratory within 24 hours of the

specimen collection. Use of a courier is recommended to reduce delay in transportation and testing times. If using the US Postal system, the regulations/standards for mailing clinical specimens apply.

- Use the envelope supplied for sending the second screen specimen to the lab.

Utah Newborn Screening

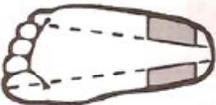
SEE BACK FOR BLOOD SPOT COVER

SECOND SCREEN:
General Instructions

Collect specimen after 7 days of life.

COLLECTION INSTRUCTIONS

1. Legibly print ALL information in spaces provided using block capital letters.
2. Collect specimen with heel stick. See newborn screening handbook for detailed instructions.
3. Fill all 7 circles.
4. Dry 3-4 hours before mailing.



COLLECT SAMPLE FROM SHADED AREA.

Mailing Instructions:

1. When blood is dry, fold stock card (from back of form) over blood spots. The flap should enclose the blood spots and reveal a biohazard symbol.
2. If using the postal service, place form with blood spots covered into envelope.

Mail to: Newborn Screening Laboratory
Utah Department of Health
46 N Medical Dr
Salt Lake City UT 84113-9903
Phone: (801) 584-8256

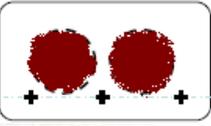
For more information, call, refer to your handbook or visit our website:
http://www.health.utah.gov/newbornscreening/HCP_Instructions.htm

Retain this sheet for your records.

XXXXXXX I.D. Number

PEEL AWAY THIS PART HERE

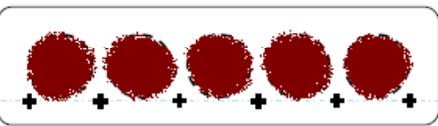
2



FOR UDOH LAB ONLY - DO NOT MARK



XXXXXXX



FOR UDOH LAB ONLY - DO NOT MARK

XXXXXXX

XXXXXXX

FOR UDOH LAB ONLY - DO NOT MARK

UTAH DEPARTMENT OF HEALTH
SECOND NEWBORN SCREENING FORM

BLOCK PRINT ALL CAPITALS - COMPLETE ENTIRE FORM
FORM EXPIRES DECEMBER 2007

Sample collection date MM/DD/YYYY 01 | 02 | 2007

Medical Record Number: SMITH Baby's first name: JOHN Sex: M F

YOUR HOSP Baby's Birthdate MM/DD/YYYY: 01 | 01 | 2007

Breast Bottle Adopted Transfusion date: 3 | 5 | 20
 Premature/lock

SMITH JANE
Mother's legal last name Mother's legal first name

DOE
Mother's maiden name

44 N MEDICAL DR
Mother's mailing address

SLC UT 84114
City State Zip

01 | 02 | 1986 801 | 584 | 8256
Mother's Birthdate MM/DD/YYYY Mother's Area Code & phone

JIM JONES
Baby's Medical Home: Doctor's Name / Clinic's Name

1234 S MEDICAL DR
Baby's Medical Home: Doctor's / Clinic's Address

SLC UT 84104
City State Zip

801 | 584 | 8256
Baby's Medical Home: Doctor's / Clinic's Area Code & phone

RECALL SCREEN MARK ONLY IF INSTRUCTED

Unacceptable ^{1st} Positive ^{2nd}

BELOW FOR UDOH LAB ONLY - DO NOT MARK

Sample Unacceptable

Fill all 7 circles with blood. The top two blood spots are sent off-site and MUST be filled in addition to the five below.

FOR UDOH LAB ONLY: Do not mark or place labels in any of these areas.

Sample Collection Date: Specimen cannot be processed without this date.

Birthdate: Several results are based on the baby's age and cannot be processed without the birthdate.

Birth Weight: Several results are based on birth weight and cannot be processed without it.

Medical Home Information: Who is called in case of an abnormal screen and where to send results.

Recall Screen Box: Mark only if instructed by program staff. This is used if another specimen is needed because of specimen being unacceptable or abnormal.

Used by UDOH lab to mark a specimen unacceptable.

* If baby is adopted, write *adoptive family information*, or adoption agency and contact person. We must be able to identify and connect the information from the second screen with the first screen.

Item-by-item instructions for completing the demographic

information on the newborn screening form

FOR UDOH LAB ONLY-DO NOT MARK: Leave this box blank. The Newborn Screening Lab uses it for the accession number and bar code.

Sample Collection Date (Month, Day, Year): Enter the sample collection date as eight digits: the first two digits for month, the next two for day, and the last four for year. Leading zeros are required. For example: 04-01-2001 for April 1, 2001.

This date establishes the parameter determining whether or not the specimen has been received within the acceptable time frame for testing. Enzymes and metabolites begin to break down as soon as the specimen is drawn. The older the specimen is when received for testing, the less likely the level of enzymes and metabolites will be accurate. The lab cannot guarantee results on a specimen that has been received outside of the acceptable time frame.

Medical Record Number: Alpha or numeric digits may be entered.

This space is supplied at the request of the providers to assist in filing the results in the chart. This information may be the hospital medical record number or the health care provider medical record number (2nd screen specimens).

Baby's Last Name: Enter the baby's last name(s). The baby's last name does not need to be the same as the mother's last name. Hyphens and apostrophes are acceptable. *This field will accept up to 12 characters.*

Baby's First Name: Enter the baby's first name. If a first name has not been selected, leave this field blank. **Do not enter 'baby girl,' 'girl,' 'bg,' 'baby boy,' 'boy,' 'bb,' or sex and mother's first name.** You may enter 'twin A,' 'triplet C,' etc. *This field will accept up to 6 characters.*

Sex: Check M (male) or F (female) box.

Birthplace/Hospital: Enter place where baby was born. You may enter a hospital name, birth institution name, 'home birth,' 'out of state' [if born in another state], 'other' [usually precipitous birth occurring in a moving conveyance or at a non-facility], or 'unknown.'

This information is used to determine where the first screening results are to be sent. This information helps identify a baby from another born on the same day with the same last name and sex. The place of birth is a reference source for information. It is used in statistical reports to determine the number and/or percent of Utah babies screened.

Baby's Birth Date: Enter the baby's birth date as eight digits: the first two digits for month, the next two for day, and the last four for year. Leading zeros are required. For example: 04-01-2006 for April 1, 2006.

This information is used for identification, quality assurance issues, and diagnostic purposes.

Individual Items: Check any and all boxes that apply to the newborn:

Breast (feeding)
Bottle (feeding)

Adopted: Adoption issues may cause some confusion. The Newborn Screening Program maintains patient and record confidentiality. The first screen form must be completed with information to identify the baby and health care provider. If there is concern about entering the birth mother's information, the adoptive agency or adoptive mother's information may be entered. A contact person must be entered. [R398-1-8 (e)] The second screen form and educational information should be given to the adoptive agent or adoptive parents with instructions for collection and submission of the second screen specimen. Do not fill out the information on the second screen card. The card should be completed at the health care provider's office with the adoptive names entered.

Premature/sick: Mark if appropriate

Transfusion, enter date of transfusion: Transfusions prior to first screen specimens invalidate results for galactosemia and hemoglobinopathy. Transfusions given 7-10 days prior to second screen specimens could interfere with the results for phenylketonuria and congenital hypothyroidism. *This information is used for interpretation of the screening results, as well as diagnosis and treatment. Babies who have received transfusions may have results that are not valid and will need another specimen drawn at a later date.*

Birth Weight (grams): Enter the baby's birth weight in grams. Birth weight is necessary to determine testing cutoff values. *This field will accept up to 4 characters.* Leading zeros are required.

Mother's Last Name: Enter the mother's legal last name(s). *This field will accept up to 12 characters.* If the mother is unmarried, the last name and maiden name can be the same.

Mother's First Name: Enter the mother's first name. *This field will accept up to 8 characters.*

Mother's Maiden Name: Enter mother's maiden name, even if it is the same as the legal last name(s). *This field will accept up to 12 characters.* The maiden name is used for identification purposes.

Mother's Mailing Address: Enter the mother's mailing address. A P.O. Box or drawer is acceptable. Include apartment number, lot number, etc. *This field will accept up to 20 characters.*

City: Enter the mother's city. *This field will accept up to 11 characters.*

State: Enter mother's state, using the 2-digit abbreviation. *This field will accept up to 2 characters.*

Zip: Enter mother's zip code. *This field will accept up to 5 characters.*

Mother's Birth Date: Enter mother's birth date as 8 digits: the first two digits for the month, the next two digits for the day, and four digits for the year. Leading zeros are required. For example: 04-01-2006 for April 1, 2006.

Mother's Area Code & Phone: Enter the mother's phone number, including the area code. *This field will accept up to 10 characters.*

Medical Home Doctor/Clinic Name: Enter the name, last and first, of the health care provider or clinic that will provide health care to the newborn. *This field will accept up to 21 characters.* The information distinguishes providers with the same name.

Medical Home Doctor/Clinic Address: Enter the address of the health care provider or clinic that will provide health care to the newborn. *This field will accept up to 21 characters.* This information identifies health care providers that have the same last name by identifying the clinic where the physician is located (some providers work at two or more clinics).

City: Enter the city of the health care provider or clinic that will provide health care to the newborn. *This field will accept up to 12 characters.*

State: Enter the state of the health care provider or clinic that will provide health care to the newborn. Use the 2-digit abbreviation. *This field will accept up to 2 characters.*

Zip: Enter the zip of the health care provider or clinic that will provide health care to the newborn. *This field will accept up to 5 digits.*

Medical Home Doctor/Clinic Area Code & Phone: Enter the phone number of the health care provider or clinic that will provide health care to the newborn, including the area code. *This field will accept up to 10 characters.*

Recall Screen Mark Only If Instructed: Mark this area only when instructed by the program personnel, or in a recall notification letter sent to you from the program. When this area is marked, the lab staff members are able to distinguish the recall specimen from a routine specimen.

SPECIAL INSTRUCTIONS

Abnormal Results

Program personnel will call the doctor/clinic noted on the demographic form for all abnormal results. Instructions will be given for follow-up. You may be asked for the collection of the routine second specimen, a third specimen or for additional confirmation testing at any time. If there are any questions, call the program for clarification (801) 584-8256.



**Newborn
Screening Program**

Utah Public Health Laboratories
46 North Medical Drive
Salt Lake City, UT 84113-1105
Telephone: (801) 584-8400
FAX: (801) 584-8501

YOUR HOSPITAL
ATTN: MEDICAL RECORDS
100 N SOMEROAD
ANYTOWN UT 84000-0000

BABY
Infant's Name : SMITH
Sex : MALE
Birth Date : 01/01/2007
Birth Record # : 000A100
Hospital MR # : 99999999
Mother's Name : SMITH, JANE
SPECIMEN INFORMATION
Type : FIRST
AscN Number : F0010100200702
Date Collected : 01/02/2007
Date Received : 01/04/2007
Date Reported : 01/06/2007
Date Printed : 01/06/2007

NEWBORN SCREENING RESULTS

DISORDER/TEST	DATE TESTED	RESULTS	DETERMINATION	NORMAL RANGE
Biotinidase Deficiency <i>Enzyme activity</i>	10/26/07	Normal	Normal	Full enzyme activity
Congenital Adrenal Hyperplasia <i>17-OHP ELISA</i>	10/26/07	00.0 ng/dL	Normal	Based on baby's birth weight
Galactosemia <i>G-1-P uridylyltransferase activity</i>	10/26/07	0.0 U/gHb	Normal	> 4.0 U/gHb
Hemoglobinopathies <i>Isoelectric Focusing</i>	10/26/07	Normal - FA	Normal	FA
Congenital Hypothyroidism <i>T4</i>	10/26/07	00.0 ug/dL	Normal	> 4.0 ug/dL
Acylcarnitine Disorders <i>MS/MS screening</i>	10/26/07	Abnormal	ABNORMAL	Based on baby's birth weight
Amino Acid Disorders <i>MS/MS screening (Including PKU)</i>	10/26/07	Normal	Normal	Based on baby's birth weight

***Footnote:** This is where any specifics about results, actions needed and notes from the lab will be entered; not all disorders will have a footnote.

A newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider.

If you have questions regarding these results, please contact the Newborn Screening Staff at the Utah Public Health Laboratories or Visit our website <http://health.utah.gov/newbornscreening>

Example of Newborn Screening Results Mailer

Miscellaneous Forms

Miscellaneous forms are supplied without cost as replacement forms for inadequate specimens, recall specimens, or for use when the original kit form has been lost. (The original kit was issued to the parent at the hospital/birthplace.)

- Write the original Kit ID Number in each place on the miscellaneous form. This original kit number was issued by the hospital of birth and can usually be obtained from the Kit ID log kept in the hospital nursery.
- Mark the 'Test Requested' box for the appropriate screen needed. *Please mark if you are requesting a first or a second screen.*
- The 'Recall Screen' box is to be marked only if you have been instructed to do so by the program. It is used to notify the lab of the need for the recall specimen procedure and testing.

Instructions For Filling Out A Miscellaneous Newborn Screening Form

1 Read all instructions on cover sheet before proceeding. Remove cover sheet.

2 Check expiration date. Do not use if expired.

3 Fill out form completely. Specimen will not be processed until information is complete.

4 Replace miscellaneous number with a stick-on label from the original kit if available (or draw a single line through it and write the number). To obtain the original number call the nursery at the hospital of birth.

The image displays the 'Utah Newborn Screening Miscellaneous Screen' form. The cover sheet (left) provides 'COLLECTION INSTRUCTIONS' and 'MAILING INSTRUCTIONS'. The main form (right) features a barcode with the number 055M057, a 'FOR UDOH LAB ONLY - DO NOT MARK' label, and a detailed data entry section. The data entry section includes fields for 'Mother's last name', 'Mother's first name', 'Mother's mailing address', 'City', 'State', 'Zip', 'Mother's birthdate', 'Mother's Area Code & Phone', 'Baby's Medical Home, Doctor's Name / Clinic Name', 'Baby's Medical Home, Doctor's / Clinic's Address', 'City', 'State', 'Zip', 'Baby's Medical Home, Doctor's / Clinic's Area Code & Phone', and 'Sample collection date'. There are also checkboxes for 'TEST REQUESTED' (First Screen, Second Screen) and 'RECALL SCREEN MARK ONLY IF RESTRICTED' (Unacceptable 1st, 2nd, Positive, POST-TRANSFUSION). A 'Sample Unacceptable' label is at the bottom.

Replace miscellaneous number with a stick-on label from the original kit if available (or draw a single line through it and write the number). To obtain the original number call the nursery at the hospital of birth.

Transfusions

Transfused blood adds foreign red blood cells (adult hemoglobin) to the infant's circulation thereby altering the level of fetal hemoglobin and enzymes found in the blood. Infants who have received transfusions containing red blood cells may not have an accurate screen. In addition, dialysis and plasma exchange transfusions may temporarily reduce the concentration of circulating metabolites and hormones for phenylketonuria (PKU) or hypothyroidism. This change may result in a false negative screen for PKU or a false positive screen for hypothyroidism.

- When possible, collect the first screen specimen prior to a transfusion.
- If the first screen specimen is collected after a transfusion, another specimen will be needed when the foreign red blood cells are no longer in circulation (approximately 120 days after the last transfusion given).
- Collect the second screen specimen 7-10 days after a transfusion is given.
- Program personnel will follow up on all specimens with the 'Transfusion' box marked and/or if the hemoglobinopathy results are indicative of a transfusion (newborn's results will show a predominance of 'A' [adult] hemoglobin).

The Newborn Screening Program will need to review medical records and transfusion history. If a transfusion(s) is documented, instructions will be given for further action.

000A111

FOR UDOH LAB ONLY - DO NOT MARK

UTAH DEPARTMENT OF HEALTH
FIRST NEWBORN SCREENING FORM

BLOCK PRINT ALL CAPITALS - COMPLETE ENTIRE FORM

FORM EXPIRES December 2007

01-02-2007
Sample collection date MM/DD/YYYY

Medical Record Number

SMITH
Baby's last name

TWIN 1
Baby's first name

YOUR HOISP
Birthplace/Hospital

01-01-2007
Birthdate MM/DD/YYYY

Breast Adopted Transfusion date: 01/01/2007 Bottle Premature/sick

1236
BIRTHWEIGHT (gms)

Note that this specimen was drawn after the baby received a transfusion.

Unsatisfactory Specimens

The Utah State Lab frequently receives blood spot specimens in conditions that are unsatisfactory for testing. Unsatisfactory specimens are known to give invalid results. Submitting unsatisfactory specimens can result in delays, placing the newborn at risk, or even result in death. *Trained staff members review each specimen to determine if it is acceptable for testing or not.*

If the specimen is determined to be unsatisfactory, the Newborn Screening tests are not done. The newborn's status - normal or abnormal - is unknown.

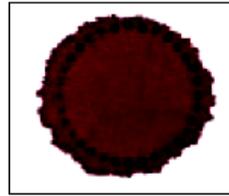
- Develop efficient procedures for recalling newborns, preparing data, and collecting filter paper blood spot specimens to replace the unsatisfactory specimens.
- Notification letter, screening result mailer (including the reason the sample was unsatisfactory), and a replacement miscellaneous newborn screening form are sent to the hospital/birth institution for unsatisfactory **first** screen specimens [R398-1-4], and to the medical home on unsatisfactory **second** screen specimens [R398-1-7]. The hospital/birth institution is responsible for the collection of a repeat first screen in the event of an unsatisfactory specimen, *even if the baby is no longer in the facility.*
- Collect the repeat specimen for an unsatisfactory first screen and the specimen for the routine second screen on different days, at least seven days apart. The Newborn Screening Program sometimes gets a repeat first specimen *after* having received the routine second specimen from the medical home. If the lab has not received the repeat first specimen, *do not draw the second specimen.* Call and find out when the repeat first specimen was drawn.

The image shows a portion of a newborn screening form. At the top, there is a green box labeled "TEST REQUESTED" with a "Mark One" instruction. Below this are two radio button options: "First Screen" and "Second Screen". A second green box contains the instruction "RECALL SCREEN MARK ONLY IF INSTRUCTED". Underneath, there are three radio button options: "Unacceptable", "Positive", and "POST-TRANSFUSION". The "Unacceptable" option is marked with an 'X' and has a small circle around the word "1st" above it and "2nd" below it. Below these options is a black box with the text "BELOW FOR UDOH LAB ONLY - DO NOT MARK". At the bottom, there is a label "Sample Unacceptable" followed by a radio button.

When sending in a recall specimen, you will be instructed to mark the recall screen box.

Simple Spot Check

Valid Specimen



Allow a sufficient quantity of blood to soak through to completely fill the pre-printed 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 Specimens:



1. Specimen quantity insufficient for testing



2. Specimen appears scratched or abraded.



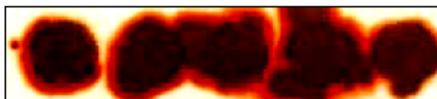
3. Specimen not dry before mailing.



4. Specimen appears supersaturated.



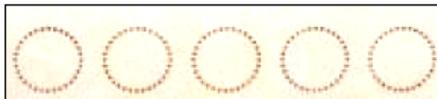
5. Specimen appears diluted, discolored or contaminated.



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.

DISORDERS COVERED BY THE PROGRAM

Effective January 1, 2006, Utah newborns are screened for the following disorders:

METABOLIC DISORDERS:

- Biotinidase deficiency: a recessive disorder of biotin metabolism.
- Galactosemia: a recessively inherited genetic disorder in which the individual is completely or partially incapable of normal metabolism of galactose due to a deficiency of the galactose-1-phosphate uridyltransferase enzyme.

Fatty Acid Oxidation Disorders: recessive disorders resulting from an enzyme deficiency needed for the break down of fatty acids.

- Carnitine uptake/transport defects
- Multiple acyl-CoA dehydrogenase deficiency (MADD)
- Short chain acyl-CoA dehydrogenase deficiency (SCAD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
- Long chain 3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Carnitine-Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase-1 Deficiency

Amino acid disorders: recessive disorders resulting from an enzyme deficiency needed for amino acid metabolism or transport.

- Arginase Deficiency
- Argininosuccinate lyase deficiency (ASA)
- Citrullinemia
- Homocystinuria
- Hyperphenylalanemia, including phenylketonuria
- Tyrosinemia

Organic Acid Disorders: recessive disorders resulting from an enzyme deficiency in the intermediary metabolism of amino acids or fatty acids.

- Beta-ketothiolase deficiency
- Glutaric acidemia, Type 1
- Isobutyryl CoA dehydrogenase deficiency
- Isovaleric acidemia

- Malonic aciduria
- Maple syrup urine disease
- Methylmalonic acidemias
- Propionic acidemia
- 3-Hydroxy-3-methylglutaryl (HMG) CoA lyase deficiency
- 2-Methyl-3-hydroxybutyryl CoA dehydrogenase deficiency
- 2-Methylbutyryl CoA dehydrogenase deficiency
- Multiple carboxylase deficiency

ENDOCRINE DISORDERS:

- Congenital adrenal hyperplasia (CAH): a genetic disorder in which there are defects in the enzymes of the adrenal cortex required for the biosynthesis of adrenal corticosteroids.
- Congenital hypothyroidism: a disorder in which the newborn is unable to secrete or produce thyroxine normally.

HEMOGLOBIN DISORDERS:

- Sickle cell disease and other hemoglobinopathies: recessively inherited genetic defects of the structure of hemoglobin found in red blood cells.

These are disorders that may have significant mortality and morbidity when not diagnosed pre-symptomatically and may not be consistently identified clinically in the neonatal period. Early detection and treatment may improve the health and development of newborns identified with these disorders.



Newborn
Screening Program

Utah Department of Health
Children with Special Health Care Needs
Newborn Screening Follow Up Program
44 N Medical Drive
PO Box 144710
Salt Lake City, UT 84114-4710
Phone: 801-584-8256
Fax: 801-536-0966
health.utah.gov/newbornscreening

Appendix A

NEWBORN SCREENING STATUTE

26-10-6. Testing of newborn infants.

(1) Except in the case where parents object on the grounds that they are members of a specified, well-recognized religious organization whose teachings are contrary to the tests required by this section, each newborn infant shall be tested for:

(a) phenylketonuria (PKU);

(b) other metabolic diseases which may result in mental retardation or brain damage and for which:

(i) a preventive measure or treatment is available; and

(ii) there exists a reliable laboratory diagnostic test method; and

(c) (i) beginning July 1, 1998, for an infant born in a hospital with 100 or more live births annually, hearing loss; and

(ii) beginning July 1, 1999, for an infant born in a setting other than a hospital with 100 or more live births annually, hearing loss.

(2) In accordance with Section **26-1-6**, the department may charge fees for:

(a) materials supplied by the department to conduct tests required under Subsection (1);

(b) tests required under Subsection (1) conducted by the department;

(c) laboratory analyses by the department of tests conducted under Subsection (1); and

(d) the administrative cost of follow-up contacts with the parents or guardians of tested infants.

(3) Tests for hearing loss under Subsection (1) shall be based on one or more methods approved by the Newborn Hearing Screening Committee, including:

(a) auditory brainstem response;

(b) automated auditory brainstem response; and

(c) evoked otoacoustic emissions.

(4) Results of tests for hearing loss under Subsection (1) shall be reported to:

(a) parents when results of tests for hearing loss under Subsection (1) suggest that additional diagnostic procedures or medical interventions are necessary; and

(b) the department.

(5) (a) There is established the Newborn Hearing Screening Committee.

(b) The committee shall advise the department on:

(i) the validity and cost of newborn infant hearing loss testing procedures; and

(ii) rules promulgated by the department to implement this section.

(c) The committee shall be composed of at least 11 members appointed by the executive director, including:

(i) one representative of the health insurance industry;

(ii) one pediatrician;

(iii) one family practitioner;

(iv) one ear, nose, and throat specialist nominated by the Utah Medical Association;

(v) two audiologists nominated by the Utah Speech-Language-Hearing Association;

(vi) one representative of hospital neonatal nurseries;

(vii) one representative of the Early Intervention Baby Watch Program administered by the department;

(viii) one public health nurse;

(ix) one consumer; and

(x) the executive director or his designee.

(d) Of the initial members of the committee, the executive director shall appoint as nearly as possible half to two-year terms and half to four-year terms. Thereafter, appointments shall be for four-year terms except:

(i) for those members who have been appointed to complete an unexpired term; and

(ii) as necessary to ensure that as nearly as possible the terms of half the appointments expire every two years.

(e) A majority of the members constitute a quorum and a vote of the majority of the members present constitutes an action of the committee.

(f) The committee shall appoint a chairman from its membership.

(g) The committee shall meet at least quarterly.

(h) (i) (A) Members who are not government employees shall receive no compensation or benefits for their services, but may receive per diem and expenses incurred in the performance of the member's official duties at the rates established by the Division of Finance under Sections **63A-3-106** and **63A-3-107**.

(B) Members may decline to receive per diem and expenses for their service.

(ii) (A) State government officer and employee members who do not receive salary, per diem, or expenses from their agency for their service may receive per diem and expenses incurred in the performance of their official duties from the committee at the rates established by the Division of Finance under Sections **63A-3-106** and **63A-3-107**.

(B) State government officer and employee members may decline to receive per diem and expenses for their service.

(i) The department shall provide staff for the committee.

Amended by Chapter 162, 1998 General Session

Last revised: Tuesday, October 03, 2006

RELIGIOUS OBJECTION TO NEWBORN SCREENING

I/We, _____ and _____,
Print, parent full name Print, parent full name

are the parents of _____, who was born on ____/____/____.
Last name and gender, if no first name

I/We understand that Utah law (§ 26-10-6(1)) requires that each newborn infant be tested for phenylketonuria, galactosemia, congenital hypothyroidism, and hemoglobinopathy.

I/We understand that failure to detect and treat any of these conditions within the first two weeks of life can be life threatening or cause significant handicaps, including mental retardation.

I/We have received a copy of the informational brochure and have read it. My health care provider has informed me of the seriousness of these conditions.

With full knowledge of the possible consequences, I/we object to the testing on the grounds that

1. I/we am/are (a) member(s) of the _____
religion

or

2. I/we have conscious, personal objections to newborn screening.

_____/_____/_____
Mother's signature Date

Mother's address City State Zip
Area Code and Phone

_____/_____/_____
Father's signature Date

Father's address City State Zip
Area Code and Phone

_____/_____/_____
Witness's signature Date

Appendix B

RULE R398-1. NEWBORN SCREENING RULE.

R398-1-1. Purpose and Authority.

- (1) The purpose of this rule is to facilitate early detection, prompt referral, early treatment, and prevention of disability and mental retardation in infants with certain metabolic, endocrine, and hematological disorders.
- (2) Authority for the Newborn Screening program and promulgation of rules to implement the program are found in Sections 26-1-30(2)(a), (b), (c), (d), and (g) and 26-10-6.

R398-1-2. Definitions.

- (1) "Abnormal test result" means a result that is outside of the normal range for a given test.
- (2) "Appropriate specimen" means a blood specimen submitted on the Utah Newborn Screening Kit form that conforms with the criteria in R398-1-8.
- (3) "Blood spot" means a clinical specimen(s) collected by carefully applying a few drops of blood, freshly drawn by heel stick with a lancet from infants, onto the filter paper (specially manufactured absorbent specimen collection paper) of the Newborn Screening Kit.
- (4) "Department" means the Utah Department of Health.
- (5) "Follow up" means the tracking of all newborns with an abnormal result, inadequate or unsatisfactory specimen or a quantity not sufficient specimen through to a normal result or confirmed diagnosis and referral.
- (6) "Inadequate specimen" means a specimen determined by the Newborn Screening Laboratory to be unacceptable for testing.
- (7) "Institution" means a hospital, alternate birthing facility, or midwife service in Utah which provides maternity or nursery services or both.
- (8) "Medical home/practitioner" means a person licensed by the Department of Commerce, Division of Occupational and Professional Licensing to practice medicine, naturopathy, or chiropractic or to be a nurse practitioner, as well as the licensed or unlicensed midwife who takes responsibility for delivery or the on-going health care of a newborn.
- (9) "Metabolic diseases" means those diseases due to an inborn error of metabolism, for which the Department of Health shall screen all infants.
- (10) "Newborn Screening Kit" means the department's demographic form with attached Food and Drug Administration (FDA)-approved filter paper medical collection device.
- (11) "Quantity not sufficient (QNS specimen)" means a specimen that has been partially tested but requires more blood to complete the full testing.
- (12) "Unsatisfactory specimen" means an inadequate specimen.

R398-1-3. Implementation.

- (1) Each newborn in the state of Utah shall submit to the Newborn Screening testing, except as provided in Section R398-1-11.
- (2) The Department of Health, after consulting with the Genetic Advisory Committee, will determine the Newborn Screening battery of tests based on demonstrated effectiveness and available funding.

R398-1-4. Responsibility for Collection of the First Specimen.

- (1) If the newborn is born in an institution, the institution must collect and submit an appropriate specimen, unless transferred to another institution prior to 48 hours of age.
- (2) If the newborn is born outside of an institution, the practitioner or other person primarily responsible

for providing assistance to the mother at the birth must arrange for the collection and submission of an appropriate specimen.

(3) If there is no other person in attendance of the birth, the parent or legal guardian must arrange for the collection and submission of an appropriate specimen.

(4) If the newborn is transferred to another institution prior to 48 hours of age, the receiving health institution must collect and submit an appropriate specimen.

R398-1-5. Timing of Collection of First Specimen.

The first specimen shall be collected between 48 hours and five days of age. Except:

(1) If the newborn is discharged from an institution before 48 hours of age, an appropriate specimen must be collected within four hours of discharge.

(2) If the newborn is to receive a blood transfusion or dialysis, the appropriate specimen must be collected immediately before the procedure, except in emergency situations where time does not allow for collection of the specimen. If the newborn receives a blood transfusion or dialysis prior to collecting the appropriate specimen the following must be done:

(a) Repeat the collection and submission of an appropriate specimen 7-10 days after last transfusion or dialysis for a second screening specimen;

(b) Repeat the collection and submission of an appropriate specimen 120 days after last transfusion or dialysis for a first screening specimen.

R398-1-6. Parent Education.

The person who has responsibility under Section R398-1-4 shall inform the parent or legal guardian of the required collection and submission and the disorders screened. That person shall give the second half of the Newborn Screening Kit to the parent or legal guardian with instructions on how to arrange for collection and submission of the second specimen.

R398-1-7. The Second Specimen.

A second specimen shall be collected between 7 and 28 days of age.

(1) The parent or legal guardian shall arrange for the collection and submission of the appropriate specimen through an institution, medical home/practitioner, or local health department.

(2) If the newborn's first specimen was obtained prior to 48 hours of age, the second specimen shall be collected by fourteen days of age.

(3) If the newborn is hospitalized beyond the seventh day of life, the institution shall arrange for the collection and submission of the appropriate specimen.

R398-1-8. Criteria for Appropriate Specimen.

(1) The institution or medical home/practitioner collecting the appropriate specimen must:

(a) Use only a Newborn Screening Kit purchased from the Department. The fee for the kit is set by the Legislature in accordance with Section 26-1-6;

(b) Correctly store the Newborn Screening Kit;

(c) Not use the Newborn Screening Kit beyond the date of expiration;

(d) Not alter the Newborn Screening Kit in any way;

(e) Complete all information on the Newborn Screening Kit. If the infant is being adopted, the following may be omitted: infant's last name, birth mother's name, address, and telephone number. Infant must have an identifying name, and a contact person must be listed;

(f) Apply sufficient blood to the filter paper;

(g) Not contaminate the filter paper with any foreign substance;

(h) Not tear, perforate, scratch, or wrinkle the filter paper;

(i) Apply blood evenly to one side of the filter paper and be sure it soaks through to the other side;

- (j) Apply blood to the filter paper in a manner that does not cause caking;
 - (k) Collect the blood in such a way as to not cause serum or tissue fluids to separate from the blood;
 - (l) Dry the specimen properly;
 - (m) Not remove the filter paper from the Newborn Screening Kit.
- (2) Submit the completed Newborn Screening Kit to the Utah Department of Health, Newborn Screening Laboratory, 46 North Medical Drive, Salt Lake City, Utah 84113.
- (a) The Newborn Screening Kit shall be placed in an envelope large enough to accommodate it without folding the kit.
 - (b) If mailed, the Newborn Screening Kit shall be placed in the U.S. Postal system within 24 hours of the time the appropriate specimen was collected.
 - (c) If hand-delivered, the Newborn Screening Kit shall be delivered within 48 hours of the time the appropriate specimen was collected.

R398-1-9. Abnormal Result.

- (1) If the Department finds an abnormal result, the Department shall inform the medical home/practitioner noted on the screening specimen form.
- (2) The Department may require the medical home/practitioner to collect and submit additional specimens and conduct additional diagnostic tests.
- (3) The medical home/practitioner shall collect and submit specimens within the time frame and in the manner instructed by the Department for the particular diagnostic test.
- (4) As instructed by the Department or the medical home/practitioner, the parent or legal guardian of a newborn identified with an abnormal test result shall promptly take the newborn to the Department or medical home/practitioner to have an appropriate specimen collected.
- (5) The medical home/practitioner who makes the final diagnosis shall complete a diagnostic form and return it to the Department within 30 days of the notification letter from the Department.

R398-1-10. Inadequate or Unsatisfactory Specimen, or QNS Specimen.

- (1) If the Department finds an inadequate or unsatisfactory specimen, or QNS specimen, the Department shall inform the medical home/practitioner noted on the screening specimen form.
- (2) The medical home/practitioner shall submit an appropriate specimen in accordance with Section R398-1-8. The specimen shall be collected and submitted within two days of notice, and the form shall be labeled for testing as directed by the Department.
- (3) The parent or legal guardian of a newborn identified with an inadequate or unsatisfactory specimen or QNS specimen shall promptly take the newborn to the medical home/practitioner to have an appropriate specimen collected.

R398-1-11. Testing Refusal.

A parent or legal guardian may refuse to allow the required testing for religious reasons only. The medical home/practitioner or institution shall file in the newborn's record documentation of refusal, reason, education of family about the disorders, and signed waiver by both parents or legal guardian. The practitioner or institution shall submit a copy of the refusal to the Utah Department of Health, Family Health Services, Newborn Screening Program, P.O. Box 144710, Salt Lake City, UT 84114-4710.

R398-1-12. Access to Medical Records.

The Department shall have access to the medical records of a newborn in order to identify medical home/practitioner, reason appropriate specimen was not collected, or to collect missing demographic information.

R398-1-13. Noncompliance by Parent or Legal Guardian.

If the medical home/practitioner or institution has information that leads it to believe that the parent or legal guardian is not complying with this rule, the medical home/practitioner or institution shall report such noncompliance as medical neglect to the Department.

R398-1-14. Confidentiality and Related Information.

- (1) The Department releases test results to the institution of birth for first specimens and to the medical home/practitioner, as noted on the demographic form, for the second specimen.
- (2) The Department notifies the medical home/practitioner noted on the demographic form if the test results are abnormal, inconclusive or QNS.
- (3) The Department releases information to the medical home/practitioner noted on the demographic form for timely and effective referral for diagnostic services or to ensure appropriate management for individuals with confirmed diagnosis.
- (4) Upon request of the parent or guardian, the Department may release results as directed in the release.
- (5) All requests for test results or records are governed by Utah Code Title 26, Chapter 3.
- (6) The Department may release information in summary, statistical, or other forms that do not identify particular individuals.
- (7) A testing laboratory that analyzes newborn screening samples for the Department may not release information or samples without the Department's express written direction.

R398-1-15. Blood Spots.

- (1) Blood spots become the property of the Department.
- (2) The Department includes in parent education materials information about the Department's policy on the retention and use of residual newborn blood spots.
- (3) The Department may use residual blood spots for newborn screening quality assessment activities.
- (4) The Department may release blood spots for research upon the following:
 - (a) The person proposing to conduct the research applies in writing to the Department for approval to perform the research. The application shall include a written protocol for the proposed research, the person's professional qualifications to perform the proposed research, and other information if needed and requested by the Department. When appropriate, the proposal will then be submitted to the Department's IRB for approval.
 - (b) The Department shall de-identify blood spots it releases unless it obtains informed consent of a parent or guardian to release identifiable samples.
 - (c) All research must be first approved by the Department's IRB.

R398-1-16. Retention of Blood Spots.

- (1) The Department retains blood spots for a minimum of 90 days.
- (2) Prior to disposal, the Department shall de-identify and autoclave the blood spots.

R398-1-17. Reporting of Disorders.

If a diagnosis is made for one of the disorders screened by the Department that was not identified by the Department, the medical home/practitioner shall report it to the Department.

FREQUENTLY ASKED QUESTIONS

What do we do if the family brings their baby in for his/her second screen and they forgot/lost the second screen card?

- *Use a miscellaneous form. Miscellaneous forms are supplied without cost as replacement forms for inadequate specimens, for recall specimens, or for use when the original kit form has been lost. (The original kit was issued to the parent at the hospital/birthplace.)*
- *Write the original Kit ID Number in place of the miscellaneous number in all three places on the form. This original kit number was issued by the hospital of birth and can usually be obtained from the Kit ID log kept in the hospital nursery.*
- *Mark the 'Test Requested' box for the appropriate screen needed. Please mark if you are requesting a first or a second screen.*
- *The 'Recall Screen' box is to be marked only if you have been instructed to do so by the program. It is used to notify the lab of the need for the recall specimen procedure and testing.*

We have to use a miscellaneous form, how do we get the baby's original Kit ID number?

- *This original kit number was issued by the hospital of birth and can usually be obtained from the Kit ID log kept in the hospital nursery. If you are having trouble obtaining the number from the hospital, please call the Newborn Screening Program for assistance.*

We have a baby that was born at home and he/she never had a first screen, what do we do?

- *If the newborn is born outside of an institution, the practitioner or other person primarily responsible for providing assistance to the mother at the birth must arrange for the collection and submission of an appropriate specimen. If there is no other person in attendance of the birth, the parent or legal guardian must arrange for the collection and submission of an appropriate specimen. If the parents were not given a first or second newborn screening card, use a miscellaneous form. The Kit ID number on this form will become the baby's ID number.*

We accidentally put the blood for the first screen on the second screen card or vice versa?

- *Sometimes there is an error and the blood is placed on the wrong card. Rather than throw out the specimen, you may use that card and send it in. Make sure to write on the card what screen it is. If it is a first screen on a second card – write **FIRST SCREEN** on the card. If it is a second screen on a first card – write **SECOND SCREEN** on the card.*

What if the baby was not born in Utah?

- *Follow the protocol for the state the baby was born in. Each state's newborn screening laws and rules are different. We will do a courtesy screen if requested by the Medical Home.*

The baby was adopted, what demographic information should we put on the newborn screening card?

- *Adoption issues may cause some confusion. The Newborn Screening Program maintains patient and record confidentiality. The first screen form must be completed with information to identify the baby and health care provider. If there is concern about entering the birth mother's information, the adoptive agency or adoptive mother's information may be entered. A contact person **must** be entered. [R398-1-8 (e)/Appendix B] The second screen form and education should be given to the adoptive agent or adoptive parents with instructions for collection and submission of the second screen specimen. Do not fill out the information on the second screen card. The card should be completed at the health care provider's office with the adoptive names entered.*

We have a baby that needs a second screen but he/she has casts on both feet, what should we do?

- *If the baby has casts on both feet and the baby did not have any abnormal tests on his/her first screen, obtain second screen when/if casts are changed or when the casts are removed. We can screen babies up to a year of age. If the baby had abnormal test results please contact the Newborn Screening Program to determine plan of care.*

What if the parents refuse to have a newborn screen done?

- *A parent or legal guardian may refuse to allow the required testing for religious reasons only. The medical home/practitioner or institution shall file in the newborn's record documentation of refusal, reason, education of family about the disorders, and signed waiver by both parents or legal guardian. The practitioner or institution shall submit a copy of the refusal to the Utah Department of Health, Family Health Services, Newborn Screening Program, P.O. Box 144710, Salt Lake City, UT 84114-4710.*