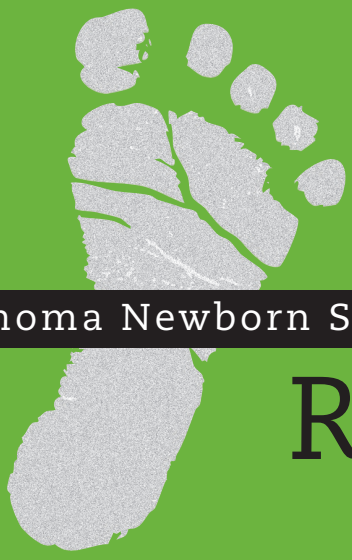


Oklahoma Newborn Screening Program · 2004

Regulations

Contents

Purpose

Under 63 O.S., Sections 1-533 and 1-534 the following rules and regulations are established concerning the screening of all infants born in Oklahoma for the disorders of phenylketonuria, congenital hypothyroidism, galactosemia, sickle cell diseases and after June 30, 2004, upon completion of validation studies and establishment of short-term follow-up services, infants shall be screened for cystic fibrosis, congenital adrenal hyperplasia, and medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD).

Definitions

The following words or terms, when used in this Chapter, shall have the following meaning, unless the context clearly indicates otherwise:

Certified Laboratory refers to the Oklahoma State Public Health Laboratory and/or a laboratory approved by the Oklahoma State Department of Health to conduct newborn screening.

CLIA '88 means the Clinical Laboratory Improvement Amendments of 1988, Public Law 100-578. This amendment applies to the Federal Law that governs laboratories who examine human specimens for the diagnosis, prevention, or treatment of any disease or impairment, or the assessment of the health of human beings.

Confirmatory Testing means definitive laboratory testing needed to confirm a diagnosis.

Congenital Adrenal Hyperplasia or ***CAH*** will refer to the most common form of CAH 21-hydroxylase deficiency. This genetic disorder is caused by the lack of an enzyme that the adrenal gland uses to process hormones. Serious loss of body salt and water can result in death. In girls the genitalia may appear as a male's, and can result in incorrect sex assignment. Hormone treatment is required for life.

Congenital Hypothyroidism means a disease caused by a deficiency of thyroid hormone (thyroxine) production, which if not treated leads to mental and physical retardation.

Cystic Fibrosis means a multisystem genetic disorder in which defective chloride transport across membranes causes dehydration of secretions. The result is a production of a thick, viscous mucus that clogs the lungs. This leads to chronic lung infections, fatal lung disease, and also interferes with digestion. Early detection and treatment can prevent malnutrition, and enhance surveillance and treatment of lung infections.

Days of Age means the age of a newborn in 24-hour periods so that a newborn is one day of age 24 hours following the hour of birth.

Department refers to the Oklahoma State Department of Health.

Discharge means release of the newborn from care and custody of a perinatal licensed health facility to the parents or into the community.

Disorder means any condition detectable by newborn screening that allows opportunities, not available without screening, for early treatment and management to prevent mental retardation and/or reduce infant morbidity and mortality.

Form Kit or Newborn Screening Form Kit is an FDA approved (or licensed) filter paper kit bearing a stamped lot number that has been approved by the Commissioner of Health. For an example of an FDA approved kit, see Appendix A, Oklahoma Health Department (OHD) Form Kit #450.

Galactosemia means an inherited disease caused by the body's failure to break down galactose due to a defective enzyme function, which if not treated early in life may cause mental retardation or death.

Hemoglobinopathy means an inherited hemoglobin disorder.

Infant means a child 6 months of age and under.

Infant's Physician means the licensed medical or osteopathic physician responsible for the care of the newborn.

Initial Specimen means the first blood specimen collected subsequent to birth, pursuant to these procedures.

Long-term Follow-up means follow-up services that begin with diagnosis and treatment and continues throughout the lifespan, including parent education, networking, referral, and case coordination.

Medium-chain Acyl Coenzyme A Dehydrogenase Deficiency or ***MCAD*** means a genetic disorder of fatty acid metabolism. This disorder can cause metabolic crisis when an infant/child fasts. This crisis can lead to seizures, failure to breath, cardiac arrest and death. Treatment is effective by preventing fasting.

Newborn means an infant 30 days of age and under.

Newborn Screening or ***Newborn Screening Tests*** means screening infants for the disorders of phenylketonuria, congenital hypothyroidism, galactosemia, sickle cell diseases, and after June 30, 2004, upon completion of validation studies and establishment of short-term follow-up services, screening infants for cystic fibrosis, congenital adrenal hyperplasia, and medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD).

Newborn Screening Laboratory means a laboratory operated by the Department or a laboratory certified by the Department to conduct the tests and carry out the follow-up required by these procedures.

Newborn Screening Program refers to the Public Health Laboratory and Family Health Services Short-term Follow-up Program at the Oklahoma State Department of Health.

Newborn Screening Program Coordinator refers to the coordinator of the Family Health Services Short-term Follow-up Program at the Oklahoma State Department of Health.

Pediatric Sub-Specialist means a physician licensed in Oklahoma, board certified in pediatrics and board certified in a pediatric subspecialty of pediatric endocrinology, pediatric pulmonology, or pediatric hematology; or a physician licensed in Oklahoma, board certified in pediatrics whose primary area of practice is pediatric endocrinology, pediatric hematology, pediatric pulmonology, or metabolic specialist.

Phenylketonuria or ***PKU*** means an inherited disease caused by the body's failure to convert the amino acid phenylalanine to tyrosine due to defective enzyme function, which if not treated early in life, causes mental retardation.

Planned Health Care Provider or ***Medical Home*** means the health care provider who will be providing health care for the infant after discharge from the hospital.

Premature Infant means an infant weighing less than 2500 grams or any live birth before the thirty-seventh week of gestation.

Repeat Specimen means an additional newborn screening specimen to be collected after the initial specimen.

Satisfactory Specimen means a specimen collected using a single form kit which is suitable in both blood quantity and quality to perform screening for phenylketonuria, congenital hypothyroidism, galactosemia, sickle cell disease, cystic fibrosis, congenital adrenal hyperplasia, and medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD). All requested demographic information on the form kit must be completed. Federal CLIA '88 regulations require that the form kit's laboratory requisition contain sufficient patient data that must include patient's name, date of birth, sex, date of collection, test(s) to be performed, and complete name and address of person requesting the test.

Screened means a specimen that has been collected and tested on an infant less than 6 months of age.

Screening means a test to sort out apparently well persons who probably have a disease from those who probably do not. A screening test is not intended to be diagnostic.

Short-term Follow-up includes services provided by the Department and the health care provider that begins when the laboratory reports an abnormal or unsatisfactory screen result and ends with a diagnosis of normal, lost (repeat testing not achieved), or affected with appropriate treatment and referral has been initiated.

Sick Infant means an infant with any condition or episode marked by pronounced deviation from the normal healthy state; illness.

Sickle Cell Disease means an inherited disease caused by abnormal hemoglobin(s) which if not treated early in life may result in severe illness, mental retardation or death (one variation is commonly referred to as sickle cell anemia).

Specimen means blood collected on the filter paper Newborn Screening Form Kit.

Submitter means a hospital, other facility, or physician submitting a Newborn Screening specimen.

Transfer means release of the newborn from care and custody from one licensed health facility to another.

Unsatisfactory Specimen means a specimen which is not collected on a form kit and/or is not suitable in blood quantity and quality to perform screening for phenylketonuria, congenital hypothyroidism, galactosemia, sickle cell disease, cystic fibrosis, congenital adrenal hyperplasia, and medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD) and/or Federal CLIA '88 regulations are not followed and the form kit's laboratory requisition does not include patient's name, date of birth, sex, date of collection, test(s) to be performed, and complete name and address of person requesting test.

Testing of Newborns

All newborns in Oklahoma shall be tested by a Certified Newborn Screening Laboratory for phenylketonuria, congenital hypothyroidism, galactosemia, sickle cell diseases, and after June 30, 2004, upon completion of validation studies and establishment of Short-term Follow-up services, infants shall be screened for cystic fibrosis, congenital adrenal hyperplasia, and medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD); a parent or guardian may refuse screening of their newborn on the grounds that such examination conflicts with their religious tenets and practices.

A parent or guardian who refuses the newborn screening blood test of their newborn on the grounds that such examination conflicts with their religious tenets and practices shall also indicate in writing this refusal utilizing the Newborn Screening Program Parent Refusal Form as illustrated in Appendix C of this Chapter. This signed refusal form shall be placed in the newborn's medical record with a copy sent to the Newborn Screening Program Coordinator.

Specimen Collection

Hospital Births

For all live hospital births, the physician, licensed or certified birth attendant shall order the collection of a newborn screening specimen on all newborns prior to transfusion, **as early as possible after 24 hours of age** or immediately prior to discharge, whichever comes first. Due to the need to identify infants at risk for the disorders quickly, the specimen should be collected **as early as possible after 24 hours of age.** Specimens shall be collected on a single Newborn Screening Form Kit using capillary or venous blood. Cord blood is unacceptable. The hospital is responsible for collecting specimens on all infants.

- If the initial specimen for any infant is collected prior to 24 hours of age, the hospital and the physician are responsible for notifying the infant's parents verbally and in writing, utilizing the parent educational form on the Newborn Screening Form Kit, that a repeat specimen is necessary at three to five days of age. The infant's physician is responsible for ensuring that the repeat specimen is collected.
- The hospital is responsible for submitting a Satisfactory Specimen and for documenting all requested information on the form kit including the parent/guardian's name, address, phone or contact phone number and the planned health care provider who will be providing well care for the infant after discharge or if the infant is to be hospitalized for an extended period of time the name of the infant's physician.
- The hospital is responsible for documenting specimen collection and results in the infant's hospital record.
- Infants who are transferred from one hospital to another during the newborn period shall have specimen collection documented in the infant's hospital record. It is the responsibility of the physician and the receiving hospital to ensure the specimen is collected.
- It is the responsibility of the hospital and physician to insure that all infants are screened prior to discharge. If an infant is discharged prior to specimen collection, the Newborn Screening Program Coordinator shall be notified. The physician is responsible for ensuring the specimen is collected as required.

Premature/Sick Infants

For all premature/sick infants, the physician shall order the collection of a newborn screening specimen prior to red blood cell transfusion, at three to seven days of age or immediately prior to discharge, whichever comes first. Due to the need to identify infants at risk for the disorders quickly, the specimen should be collected **as early as possible after 24 hours of age.** It is recommended that a repeat newborn screening specimen be collected at 14 days of age. Specimens shall be collected on the Newborn Screening Form Kit using capillary or venous blood. The hospital and the physician are responsible for ensuring that specimens are collected on all premature/sick infants.

- Premature/sick infants screened prior to 24 hours of age must be re-screened between 7-14 days of age.
- Premature/sick infants who could not be screened prior to a red blood cell transfusion should be screened by the 7th day of life, with a repeat specimen collected when plasma and/or red cells will again reflect the infant's own metabolic processes and hemoglobin type (the accepted time period to determine hemoglobin type is 90 to 120 days after transfusion).
- The recommended follow-up study for an abnormal thyroid screen in a premature infant is a serum free T4 (measured by direct dialysis or an equivalent method) at 7-14 days of age.

Out-of-Hospital Births

All infants who are not born in a hospital shall be tested **as early as possible after 24 hours of age.** The infant's physician, licensed or certified birth attendant is responsible for submitting a Satisfactory Newborn Screening Specimen. If there is not a physician, licensed or certified birth attendant involved in a non-hospital birth, the person attending the birth and the parents of the infant are responsible for submitting a Satisfactory Newborn Screening Specimen.

If a physician examines a child in the first three months of life who was not born in a hospital, or born out of state, the physician will verify that the child has been screened. If the child has not been screened or if results of screening are not available, the physician should submit a Satisfactory Newborn Screening Specimen.

Technique for Filter Paper Sample Collection

Specimens obtained with a Newborn Screening Form Kit should be collected in accordance with Appendix A of this Chapter.

Failure to follow these methods of blood collection may cause inaccurate results and require repeat specimens.

Submitters are responsible for submitting a Satisfactory Newborn Screening Specimen.

Hospital Recording

The hospital shall implement a procedure to assure that a newborn screening specimen has been collected on every newborn and transported to the Newborn Screening Laboratory within 24-48 hours of collection.

The hospital shall immediately notify the infant's physician, parents or guardians, and Newborn Screening Program Coordinator if an infant is discharged without a sample having been collected. This notification shall be documented in the infant's hospital record.

If no test results are received within fifteen (15) days after the date of collection, the hospital shall contact the Newborn Screening Laboratory to verify that a specimen had been received. If no specimen has been received, the hospital shall notify the physician.

Any hospital or any other laboratory which collects, handles or forwards newborn screening samples shall keep a log containing:

- name
- date of birth
- attending physician
- planned health care provider
- medical record number
- serial number of the Newborn Screening Form Kit
- date the specimen was drawn
- date the specimen was forwarded
- date the test results were received
- test results

Specimens should be transported in the manner designated by the Department.

Parent and Health Care Provider Education

The infant's physician or designee shall have the responsibility to assure that at least one of each newborn's parent or legal guardian is notified about newborn screening and is provided information about the disorders and instructed to obtain screen results from the planned health care provider or Newborn Screening Program.

The hospital will be responsible or designate a responsible party to distribute the Newborn Screening Program's written educational materials on newborn screening provided by the Department to at least one of each newborn's parent or legal guardian.

Hospitals shall provide ongoing training programs for their employees involved with newborn screening procedures. These training programs shall include methods of collecting a Satisfactory Newborn Screening Specimen.

The hospital is responsible for assuring that employees who collect, handle or perform newborn screening tests are informed of their responsibilities with respect to screening procedures.

Follow-Up for Physicians

If a physician examines a child in the first three months of life, the physician will verify that the child has been screened, and document results in the infant's medical record. If the child has not been screened or if results of screening are not available, the physician should submit a Satisfactory Newborn Screening Specimen within 48 hours or as soon as possible.

On written notification by the Newborn Screening Program of follow-up requirements for a newborn screen result of abnormal, unsatisfactory and less than 24 hours of age at time of collection; the infant's physician or designee will obtain required repeat screening, confirmatory testing, or diagnostic studies, in the time-frame specified so that therapy, when indicated, can be initiated expediently.

The infant's physician may selectively rescreen infants as clinically indicated.

Because patients may relocate without a forwarding address or contact information, where these rules place responsibility upon physicians and hospitals to follow-up or notify parents, then that shall be deemed to require only that a reasonable search be made and that if the parents are not contacted that the Newborn Screening Program Coordinator be notified of the non-follow-up or non-notification after efforts to contact the parents have been exhausted.

For appropriate comprehensive medical care, all confirmed cases of congenital hypothyroidism, galactosemia, phenylketonuria, sickle cell disease, cystic fibrosis, congenital adrenal hyperplasia, medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD) should have a referral to a pediatric sub-specialist, and the parent should be referred for enrollment in newborn screening Long-term Follow-up services as designated by the Newborn Screening Program. For referral information, please contact the Newborn Screening Short-term Follow-up Program at (405) 271-6617 or 1-800-766-2223, ext. 6617.

Physician Reporting

If confirmatory or follow-up testing is not performed by the Newborn Screening Laboratory or through a contract laboratory designated by the Newborn Screening Program, the infant's physician must report to the Newborn Screening Program Coordinator the results within 7 days after the completion of the medical evaluation, using the Department's Newborn Screening Report Form as illustrated in Appendix B of this Chapter.

A copy of the confirmatory test results must accompany the report form.

For all diagnosed cases of phenylketonuria, congenital hypothyroidism, galactosemia, sickle cell diseases, cystic fibrosis, congenital adrenal hyperplasia, and medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD), the infant's physician shall report treatment date if applicable, and referral information to the Newborn Screening Program Coordinator by completing the Department's Newborn Screening Report Form as illustrated in Appendix B of this Chapter.

These reports shall be confidential and may be utilized only for the purpose of assuring service delivery, program administration, data analysis, and evaluation.

Information

For information regarding laboratory procedures, or results of laboratory tests or to order form kits, contact:

Public Health Laboratory Service
Oklahoma State Dept of Health
P.O. Box 24106
Oklahoma City, OK 73124-0106
(405) 271-5070
FAX (405) 271-4850

For general information or information regarding follow-up, contact:

Newborn Screening Short-term Follow-up Program
Family Health Services
Oklahoma State Dept. of Health,
1000 NE Tenth St.
Oklahoma City, OK 73117-1299,
(405) 271-6617
FAX (405) 271-4892
1-800-766-2223

General information about the Newborn Screening Program is available on the OSDH Web site at **www.health.state.ok.us/program/gp**.

Standards, Procedures, and Follow-Up

The Commissioner of Health shall establish procedures for newborn screening laboratories which shall include laboratory methodology, proficiency testing, quality assurance, sample collection, reporting, follow-up, handling, use, retention, storage and disposition of form kits.

The Commissioner of Health shall establish procedures for the Department's newborn screening short-term follow-up program which shall include quality assurance, notification of providers and parents, follow-up guidelines, and parent and provider education.

Hospitals, physicians, and laboratories shall comply with procedures for the Newborn Screening Program established by the Commissioner of Health.

Certified Newborn Screening Laboratories

(Applies to Laboratories that Perform the Newborn Screening Test)

Any laboratory performing newborn screening tests shall be certified by the Department as a Newborn Screening Laboratory. In order to be certified as a Newborn Screening Laboratory, a laboratory shall maintain technical proficiency and ensure that test reagents and equipment are properly standardized.

A Certified Laboratory refers to the Oklahoma State Public Health Laboratory or a laboratory approved by the Oklahoma State Department of Health to conduct newborn screening tests. A laboratory desiring certification as a Newborn Screening Laboratory shall make written application to the Public Health Laboratory Service of the Department. A certified laboratory shall meet the following minimum standards:

Eligibility for Approval

A laboratory in Oklahoma that meets the requirements of Section 353 of the Public Health Service Act (42 U.S.C. 263a) as revised by the Clinical Laboratory Improvement Amendments of 1988 (CLIA '88), Public Law 100-578. The Laboratory must have a CLIA certificate for tests of High Complexity and meet the criteria for those tests as specified in CLIA '88 and amendments. The lab must have the capacity to provide testing for the mandated newborn screening panel on a single satisfactory filter paper specimen submitted by the birth hospital or provider.

Minimum Tests

A laboratory shall perform a sufficient number of tests each week, a minimum of 300 blood samples from different Oklahoma infants a week for each disorder, to maintain technical proficiency and ensure that test reagents and equipment are properly standardized.

Record Keeping

The laboratory shall log in each sample received with an identifier. All patient information and test results shall be linked to the identifier and maintained as a permanent record for a period of 21 years.

The laboratory shall maintain quality control and proficiency test records and shall be available for inspection by the Department.

Standard Laboratory Screening Assay Methods

All assay methods must be approved by the Commissioner of Health.

Follow-up for Certified Laboratories

Within fifteen (15) days after specimen collection, the Certified Laboratory shall send a written report of the test results with repeat testing requirements, if indicated, to the submitter and physician listed on the filter paper requisition.

The Certified Laboratory will reject any unsatisfactory samples for testing.

The Certified Laboratory must maintain a database with the capacity to report abnormal results to the Department's Newborn Screening Program Coordinator or designee.

The Certified Laboratory must report abnormal results that are possible disease conditions within eight to twenty-four hours to the Department's Newborn Screening Program Coordinator or designee.

Reporting

Certified Laboratories shall compile quarterly and annual reports for the Newborn Screening Program of total screening tests, abnormal tests by disorder, unsatisfactory tests, and less than 24 hours of age at time of collection test.

Certification of Laboratories

Upon satisfying the requirements of these standards and demonstrating proficiency in the presence of an authorized representative from the Department the following is issued. A certificate of approval, which will specify:

- Name of laboratory
- Test of certification must be approved for all mandated tests.
- Date of issue and expiration: certificate issued for one (1) year and renewable annually.

Revocation of Certification

The laboratory shall be in compliance with all applicable Federal or State Laws, or regulations. The compliance with the requirements thereof shall be the responsibility of the laboratory, without reliance on or direction by the Oklahoma State Department of Health (OSDH). Following notice by OSDH of its intent to revoke the laboratory's certification and completion of an individual proceeding pursuant to Article II of the Oklahoma Administrative Procedures Act (APA), the certification of a laboratory may be revoked, based upon proof by a preponderance of the evidence for any of the following reasons:

- Failure to meet any requirements in these regulations.
- Failure to use a standard laboratory assay approved by the Commissioner of Health.
- Failure to participate in a recognized proficiency program and/or maintain proficiency.
- Failure to keep adequate records of test results and quality control.
- Failure to give prompt notice of changes in personnel performing the tests.

Upon notice of revocation the laboratory shall cease to perform newborn screening and return their certificate of approval.

Reinstatement of laboratory certification shall be contingent upon the following:

- A laboratory shall not apply for reinstatement until a minimum of three months has elapsed from date of revocation.
- All factors which can be identified must be corrected.
- A laboratory applying for reinstatement must meet the same requirements as for initial application.

Revocation of certified laboratory status by OSDH may be appealed pursuant to Article II of the Oklahoma APA.

Advisory Committee

A standing committee of the Oklahoma Genetics Advisory Council (OGAC) shall advise the Department on newborn screening issues.

Appendix A · Instructions for Filter Paper Sample Collection

Preliminary Steps

Ensure that the expiration date of the filter paper form kit has not passed. Complete the required information on the filter paper form kit. A ballpoint pen should be used; soft-tip pins will not copy through to the other sheets of paper. Address imprint devices (or adhesive labels) should never be used unless the handling process ensures that the patient information is not obscured and the blood collection area is not compromised. Do not use typewriters or printers that might compress the paper. Avoid touching the area within the circles on the filter paper section before, during and after collection (blood spots) of the specimen. Do not allow water, feeding formulas, antiseptic solutions, glove powder, hand lotion, or other materials to come into contact with the specimen card before or after use.

Precautions

Confirm the identity of the infant and ensure accuracy of the demographic data on the card. Wash hands vigorously before proceeding. All appropriate precautions, including wearing powder-free gloves (changing gloves between infants), should be taken for handling blood and disposing of used lancets in a biohazard container for sharp objects.

Site Preparation

Warm the newborn's heel, since warming the skin-puncture site can help increase blood flow. A warm, moist towel or diaper at a temperature no higher than 42° C may be used to cover the site for 3 minutes. This technique increases the blood flow sufficiently and will not burn the skin. In addition, positioning the infant's leg lower than the heart will increase venous pressure.

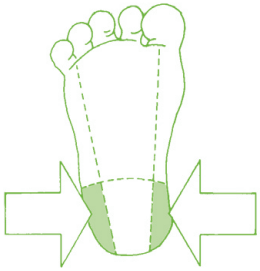
Cleaning the Site

The skin should be wiped with alcohol (isopropanol/water: 70/30 by volume, "70%"). Allow the skin to air dry.

Puncture

To obtain sufficient blood flow, puncture the infant's heel on the plantar surface of the heel with a sterile lancet or with a heel incision device. The incision device provides excellent blood flow by making a standardized incision 1.0 mm deep by 2.5 mm long. Any puncture device used should be selected so that the puncture does not exceed 2.0 mm in depth. For infant safety, scalpel blades or needles must not be used to puncture the skin for blood collection. Disposable skin puncture lancets of different designs are commercially available for performing the heel stick on infants. For worker safety, disposable skin puncture devices that protect the user from unintentional self-inflicted skin punctures should be used.

In small, premature infants, the heel bone (calcaneus) might be no more than 2.0 mm beneath the plantar heel skin surface and



half this depth at the posterior curvature of the heel. Studies indicate that for some infants (including full-term infants) a puncturing depth of 2.0 mm might be excessive and might cause bone damage. In this situation other collection methods should be considered.

Direct Application

After the heel has been punctured, wipe away the first drop of blood with a sterile gauze pad or cotton ball and allow a larger drop of blood to form. (Intermittently apply gentle pressure to the heel with the thumb, and ease this pressure as drops of blood form). Touch the filter paper gently against the large blood drop and, in one step, allow a sufficient quantity of blood to soak through and completely fill a preprinted circle on the filter paper. Do not press the filter paper against the puncture site on the heel. Blood should be applied only to one side of the filter paper. Both sides of the filter paper should be examined to assure that the blood uniformly penetrated and saturated the paper. During collection avoid milking or layering:

Milking · Excessive milking or squeezing the puncture might

cause hemolysis of the specimen or result in an admixture of tissue fluids with the specimen and might adversely affect the test result.

Layering · Do not apply layers of successive blood drops to the same printed circle. Applying successive drops of blood to already partially dried spots causes nonuniform analyte concentrations and invalidates the specimens.

After blood has been collected from the heel of the newborn, the foot should be elevated above the body, and a sterile gauze pad or cotton swab pressed against the puncture site until the bleeding stops. It is not advisable to apply adhesive bandages over skin puncture site on newborns.

Collection

The required blood spots should be collected so that there is one in each pre-printed circle of the filter paper. Failure to collect and fill each pre-printed circle might result in the specimen being rejected (unsatisfactory) for testing. If blood flow diminishes so that a circle is not completely filled, repeat the sampling technique using a new circle or, if necessary, a new blood collection card.

For alternative methods to specimen collection (e.g., capillary tube, dorsal hand vein, umbilical venous catheter or umbilical arterial catheter) refer to the *NCCLS Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard-Fourth Edition (LA4-A4, Vol. 23 No. 21)* or contact the Newborn Screening Program Coordinator.

Drying

Avoid touching or smearing the blood spots. Allow the blood specimen to air dry on a horizontally level, nonabsorbent, open surface for at least 3 hours at an ambient temperature of 15° C to 22° C. Keep the specimen away from direct sunlight (indirect room light is not usually detrimental unless accompanied by heat). Blood spots on the filter paper should not be heated, stacked, or allowed to touch other surfaces during the drying process.

The Filter Paper has a fold-over protective cover. This protective cover is used to protect the blood spots from contamination and can be used in the drying process. To use the protective cover in the drying process simply elevate the blood spots to gently rest on the edge of the protective cover. After drying, the protective cover should be placed over the spots to prevent contamination.

Stacking

Since leaching (cross-contamination) between specimens might occur, specimen-to-specimen contact is not appropriate. Before placing the specimens in a paper envelope for mailing, use the fold-over protective cover to cover each individual blood spot. When stacking of exposed blood spots cannot be avoided, the following procedure should be done:

Before placing the specimens in a paper envelope for mailing, the dried blood spots on the collection card should be rotated 180° from the blood spots on the cards in the stack immediately above and below.

If the physical barrier is used (fold-over protective cover), specimen rotation is not necessary.

Mailing

Specimens should be transported in the manner designated by the Department. The collection card should be transported or mailed to the Newborn Screening Program laboratory within 24 hours after collection. Mailing delays at collection sites should be avoided, and the postal or transport environment relative to possible delays should be considered. Never place the filter paper specimen in plastic bags. Use the form kit's protective overlay to cover the filter paper spots when mailing or transporting. If mailing the specimens use a U.S. Postal Service approved envelope.

Information

For information regarding specimen collection, Postal regulations, envelope and form kit purchasing, please contact the Newborn Screening Program Laboratory at (405) 271-5070.

Source: *NCCLS Blood Collection on Filter Paper for Newborn Screening Programs; Approved Standard-Fourth Edition (LA4-A4, Vol. 23 No. 21)*

OHD #450 Form Kit's Detachable Educational Slip

Front

Baby's Last Name

Baby's First Name

Oklahoma State Department of Health

Newborn Screening Program

OKLAHOMA NEWBORN SCREENING Dried-blood Spot PROGRAM

THE NEWBORN SCREENING BLOOD TEST

A special blood test has been done to protect your baby from hidden disease. The test screens for the disorders listed on the back of this form. These disorders are harmful if treatment is not started within the first month of life (each disorder is explained on the back of this sheet).

TIME OF TESTING

☐ Under 24 hours of age ☐ Over 24 hours of age

ATTENTION PROVIDER

DETACH AND GIVE TO PARENT OR GUARDIAN

WILL FURTHER TESTING BE REQUIRED?

If your baby was tested before 24 hours of age, the test must be repeated at 3 to 5 days of age. If the blood test is abnormal or inadequate to test, a repeat test will be needed. If the time of testing is not indicated on this form, please contact your baby's physician to determine if your baby needs a repeat test.

ASK YOUR BABY'S DOCTOR FOR THE TEST RESULTS

Please take this form with you to your baby's first doctor visit and ask for test results. If your baby's doctor does not have the test results and you have not been notified by mail, please call the Oklahoma State Department of Health when your baby is two to three weeks of age at **405-271-6617 or 800-766-2223**.

Serial No. 000000

DETACH AND GIVE TO THE PARENT OR GUARDIAN
NEWBORN SCREENING PROGRAM

Back

Early Detection and Treatment Provide Oklahoma Infants a Healthy Start

Congenital Hypothyroidism – If not treated, this disorder results in mental retardation and poor growth. Congenital hypothyroidism is usually caused by abnormal development or absence of the thyroid gland. Treatment with daily thyroid medication is needed to **prevent** mental retardation and poor growth.

Classic Galactosemia – If not treated, this genetic disorder can harm the baby's eyes and liver and can result in mental retardation or even death. Galactosemia occurs when the baby cannot break down a special sugar in milk called galactose. A special diet without galactose is needed to **prevent** mental retardation or death.

Phenylketonuria (PKU) – If not treated, this genetic disorder can result in severe brain damage. PKU is caused by the body's inability to break down the protein in food called phenylalanine. Treatment with a special diet is needed to **prevent** mental retardation.

Sickle Cell Disease & other hemoglobin diseases – If not treated, this genetic disorder may result in severe illness or death. Sickle cell disease occurs when the hemoglobin in the red blood cells does not develop normally. Red blood cells have the important job of delivering oxygen to different parts of the body. Treatment with antibiotics helps **prevent** infections that could cause severe illness or death. Other hemoglobin diseases can be detected by screening and will require medical follow-up.

Cystic Fibrosis – If not treated, this genetic disorder results in malnutrition. Cystic Fibrosis is a disorder that causes thick mucus to collect in the lungs and other body organs, which can result in breathing problems, lung infections, and poor digestion of food. To **prevent** malnutrition and to address the infant's special health care needs, expert medical care from a Cystic Fibrosis Center is needed.

Congenital Adrenal Hyperplasia 21-hydroxylase Deficiency (CAH) – If not treated, this genetic disorder may result in severe illness or death. CAH is caused by the lack of an enzyme that the adrenal gland uses to process hormones. Serious loss of body salt and water can result in death. In girls the genitalia may appear like that of a male, and can result in incorrect sex assignment. Hormone treatment is needed to **prevent** serious illness and death.

A new test scheduled to start March 2005 - Medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD) [Ask your health care provider if testing for this disorder has started] – If not treated, this genetic disorder may result in severe illness or death. MCAD is a disorder of fatty acid metabolism. This disorder can cause metabolic crisis when an infant/child fasts. This crisis can lead to seizures, respiratory failure, cardiac arrest, and death. Frequent feedings and a special diet are needed to **prevent** serious illness and death.

Special Note: For sickle cell disease and cystic fibrosis screening, the blood test might find that your baby is a "carrier" of a disorder. Carriers do not need treatment or any special medical care. Genetic counseling is recommended.

Questions about the newborn screening blood test?
Call: 405-271-6617 or 800-766-2223
E-mail: newbornscreen@health.state.ok.us
Web site: www.health.state.ok.us/program/gp

OHD #450 Form Kit

Ser. No. 000000

Newborn Screening Form
Oklahoma State Department of Health-P.O. Box 24106,
Oklahoma City, OK 73124-0106 (405) 271-5070
ODH #450 Rev. **1/2005**

DO NOT WRITE IN THIS BOX

INFANT'S INFORMATION

1. Infant's Last Name ____ Infant's First Name ____
2. Sex: ☐ M ☐ F
3. Date of Birth: MM DD YY
4. Birth Time: : 24 Hour Clock
5. Birthweight in Grams
6. If Multiple Birth Indicate Birth Order: ☐ A-H
7. Infant's Medical Record or I.D.
8. Provider ID
9. Infant's Provider or Physician's Name
10. Mom's Medicaid Number
11. ( ) - Provider's Phone Number

SPECIMEN INFORMATION

1. Collection Date: M M D D Y Y — Collection Time: : 24 Hour Clock
2. Transfusion Date: MM DD YY — Time: : 24 Hour Clock
3. Has a previous metabolic blood test been done anywhere? ☐ Yes ☐ No
   Previous OSDH Lab Number
4. Age of Infant at time of specimen collection:
   ☐ Under 24 hours of age ☐ Over 24 hours of age
5. Test requested:
   ☐ All Tests ☐ HGB only ☐ Monitor for: _____

MOM'S INFORMATION

1. Mom's Last Name, First Name
2. Mom's Age
3. Mom's Address
4. Apt. #
5. Mom's City
6. State
7. Zip
8. ( ) - Mom's Telephone or Contact
9. - - Mom's Social Security #
10. Mom's Race/Ethnic: ☐ 1. White ☐ 2. Black ☐ 3. Hispanic ☐ 4. Asian ☐ 5. American Indian ☐ 6. Other

☐ **Adoption** *(check if baby is being adopted)*

SUBMITTING HEALTH PROVIDER :

1. ID #
2. Return to:

Hearing Screening Results:

Right Ear	Left Ear	Screen Method
☐ Pass	☐ Pass	☐ ABR ☐ Other (Specify)_____
☐ Refer	☐ Refer	☐ OAE

If not screened, reason:
☐ Technical problem ☐ No equipment ☐ Delayed
☐ Caregiver refused ☐ Baby discharged ☐ Other_____

Hearing risk status – Check all that apply:
☐ Blood relatives of the infant have a permanent hearing loss that began at birth or in early childhood.
☐ Infant is suspected of having a congenital infection (neonatal herpes, cmv, rubella, syphilis, toxoplasmosis).
☐ Infant has craniofacial anomalies (pinna/ear canal abnormality, cleft lip/palate, hydrocephalus).
☐ Infant had exchange transfusion.
☐ Infant has serum bilirubin level ≥ 15 mg/dL.
☐ Infant was placed in a Level II or III nursery for more than 24 hours.

HEARING SCREENING INSTRUCTIONS

Hearing screening is to be completed with results recorded and forwarded to the Oklahoma State Department of Health at the same time as the blood specimen. Follow the instructions below:

1. Screen the infant's hearing using the available technology.
2. Record results in the **Hearing Screening Results** area on the front page of the form. Place a check mark in the appropriate Pass or Refer box for the right ear and the left ear.
3. Indicate the method used to screen hearing (ABR, OAE, Other). If "Other" is checked, specify the technology used.
4. If **hearing cannot be screened**, check the appropriate box for the reason; **if screening will be delayed**, follow instructions below.
5. Complete the **Hearing risk status** indicator section by placing a check mark in the box of any item that applies to this infant. The first question about familial hearing loss is to be asked of the birth mother. Information for the other indicators should be available in the infant's chart.
6. Detach and give the Newborn Hearing Screening parent form (pink sheet) to the infant's parent or guardian at discharge.

DO NOT DELAY SENDING THE BLOOD SPECIMEN. ALL BLOOD SPECIMENS <u>MUST</u> BE SENT WITHIN 24 HOURS OF COLLECTION.

For infants whose hearing screening cannot be completed by the time the blood specimen must be sent (including those transferred **<u>within</u>** the facility) **and it is anticipated hearing will be screened prior to discharge, do the following:**

1. On the original form in the **If not screened, reason:** area, mark the "**Delayed**" box.
2. Complete the Hearing risk status section. For infants placed in "special care" nursery, be sure to mark the **Infant was placed in a Level II or III nursery for more than 24 hours** box. ***Be certain there are no marks in the* Screen Method box**.
3. Detach and retain the parent's copy of the hearing screening form (pink sheet). It will be used to record hearing screening results.
4. Be sure that the infant's last and first names are legible on the detached document.
5. Mail blood specimen.
6. Perform the hearing screening prior to discharge.
7. Record the results as indicated above in the appropriate boxes on the pink parent copy.
8. Mark any appropriate boxes in the Hearing risk status area if this has not already been completed.
9. Photocopy the front of the completed form (pink sheet). **Be certain that infant's name and the form's serial number are legible on photocopy**.
10. Mail the photocopy to OSDH, Public Health Laboratory Service, PO Box 24106, Oklahoma City, OK 73124-0106.
11. Give the completed pink sheet to the infant's parent or guardian.

Ser. No. 785625
FORM ODH #450 REV 12/2003
FILL ALL CIRCLES WITH BLOOD
S&S 903® LOT# W-011
Expiration Date: 01/01/2006

Instructions on Specimen Collection and Mailing
(Complies with NCCLS Standard LA 4 – A4)

SUBMITTER RESPONSIBILITY

1. Completion of form.
2. Collection of an adequate specimen for testing.
3. Send specimens within 24 hours of collection.
4. The quality of the specimen received by the Public Health Laboratory Service.
5. Listing the planned health care provider who will be providing well care for the infant after discharge or infant's physician if the infant is to be hospitalized for extended period of time.

SCREENING REQUIREMENTS FOR ALL NEWBORNS

1. Prior to blood transfusion, as early as possible after 24 hours of age or immediately prior to discharge, whichever comes first.
2. If infant is screened at less than 24 hours of age, repeat screen at 3-5 days of age (if premature or a sick infant, repeat screen at 7-14 days of age).
3. All premature and sick infants should have a repeat screen at 14 days of age.

COMPLETION OF FORM

1. Legibly print and complete all information requested.
2. List submitter's return address and submitter's ID number. Submitter means the facility or provider who has collected the specimen.
3. List the provider or physician who will be following the baby for well care or the attending physician if the infant is hospitalized for an extended period of time.
4. List the parent's correct address and phone number for notification of abnormal results.

Note: All results are sent to the submitter and the provider listed on the form.

COLLECTION OF BLOOD SPECIMEN

1. To prevent specimen contamination, do not touch any of the filter paper circles before or after collection.
2. Select puncture site and cleanse with 70% isopropyl alcohol and allow heel to air dry. Usual puncture site is illustrated below.

3. Use a sterile, disposable lancet or heel incision device to perform a swift clean puncture.
4. Wipe away first drop of blood with a sterile gauze or cotton ball.
5. Gently touch the filter paper against a large drop of blood and allow a sufficient quantity of blood to soak through to completely fill the preprinted circle on the filter paper. Blood must be applied to only one side of the filter paper and circle area should be fully saturated.
6. Fill **all five circles** with blood.
7. Protect freshly collected specimens from contamination.
8. Allow blood specimen to air dry at room temperature for 2-6 hours in a horizontal position. **Do not stack wet specimens. Insufficient drying will adversely affect the test results. DO NOT PLACE FILTER PAPER SPECIMENS IN A PLASTIC BAG.**
9. Specimens may be "Unsatisfactory for Testing" for the following reasons:
   a. Circles not completely filled in or not thoroughly saturated.
   b. Uneven saturation of circles or multiple sample application.
   c. Specimen appears contaminated.
   d. Clotted or caked blood on filter paper, or damaged filter paper.
   e. Assay inhibition due to antibiotic or other substance.
   f. Incomplete elution of blood from filter paper.
   g. Laboratory requisition incomplete or improperly completed.
   h. Results inconsistent – possibly due to improper sample collection.
   i. Specimen submitted on incorrect form or expired form.
   j. No specimen received with form.
   k. Specimen placed in plastic bag while wet.
   l. Receipt of specimen was more than 14 days from date of collection.

To Order Newborn Screening Collection Kits (ODH #450): Call (405) 271-5070

Specimens should be transported in the manner designated by the OSDH Public Health Laboratory Service. Send specimens within 24 hours of collection.

Courier Service address:
NEWBORN SCREENING SECTION
Public Health Laboratory Service
1000 NE 10th Street
Oklahoma City, OK 73117-1299

Mailing address: *(using United States Postal Service)*
NEWBORN SCREENING SECTION
Public Health Laboratory Service
P.O. Box 24106
Oklahoma City, OK 73124-0106

Adoption

If infant is being adopted, check the Adoption box on the front of the form. List the agency or lawyer that is handling the adoption in the "Mom's Information" section. Please note: for proper identification, the "Infant's Information" section must be completed accurately. Questions? Please call (405) 271-6617 or (800) 766-2223.

Appendix B Report Form

Newborn Screening Program Report Form

Infant's Name _____ Infant's Birth Date ___ ___/___ ___/___ ___

Newborn Screening Program Lab # _____ Mother's Name _____

[ ] Pending Diagnosis, Follow-up Plan:

_____

_____

Final Diagnosis (please attach confirmation lab results)

[ ] Normal

[ ] Trait Condition (specify carrier status):

_____

[ ] Classic Galactosemia (GG phenotype/genotype)

[ ] Duarte/Galactosemia Compound Heterozygote (DG phenotype/genotype)

[ ] Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency

[ ] Cystic Fibrosis

[ ] Classic Phenylketonuria (PKU)

[ ] Hyperphenylalaninemia (not clinically significant)

[ ] Hyperphenylalaninemia (clinically significant treatment required)

[ ] Congenital Hypothyroidism

[ ] Medium-chain Acyl Coenzyme A Dehydrogenase Deficiency (MCAD)

[ ] Sickle Cell Disease (specify type):

_____

[ ] Hemoglobin disease (specify type):

_____

[ ] Other (specify):

_____

Treatment Indicated? [ ] yes [ ] no

Date Treatment Started ___ ___/___ ___/___ ___

Referred to Pediatric Sub-specialist:

[ ] Endocrinologist (specify name) _____

[ ] Hematologist (specify name) _____

[ ] Metabolic Specialist (specify name) _____

[ ] Pulmonologist (specify name) _____

Family Referred for (check all that apply)

[ ] Genetic counseling (check provider): ___ Sickle Cell Association ___ Geneticist ___ Other _____

[ ] Enrollment in Newborn Screening Long-term Follow-up Program

[ ] Early Intervention Services

Print Physician's Name _____ Telephone _____

Physician Signature _____ Date ___ ___/___ ___/___ ___

Mail or Fax this follow-up form with complete diagnostic information and confirmation lab results to:

Oklahoma State Department of Health · Family Health Services · Newborn Screening Program Coordinator
1000 NE Tenth Street · Oklahoma City, OK 73117-1299 · Fax (405) 271-4892

For questions or referral information, please call the Newborn Screening Program Coordinator at (405) 271-6617 or 1-800-766-2223.

Appendix C **Refusal Form**

Newborn Screening Blood Test: Religious Tenets & Practices Refusal Form

Infant's Name _____ Medical Record Number _____

Infant's Birth Date ___ ___ /___ ___ /___ ___

Attending Physician or Provider (please print) _____

Place of Birth:

[ ] Hospital (print name) _____

[ ] Birthing Facility (print name) _____

[ ] Home Birth

- I have received and read the parent educational brochure printed by the Oklahoma Department of Health on the newborn screening blood test. I understand that these disorders are easily detected by testing a small blood sample from my baby's heel.
- I have been informed that all newborns are required by law (under 63 O.S. 2004, Sections 1-533 and 1-534) to have a newborn screening test collected.
- I have been informed and I understand that this screening is done to detect these disorders because symptoms sometimes do not appear for several weeks or months, and irreversible damage can occur before symptoms become apparent to a family or a physician.
- I have been informed and I understand that, if untreated, these conditions may cause permanent damage to my child, including mental retardation, growth failure, and even death. This permanent health damage can be prevented through early detection and treatment.
- I have discussed the newborn screening test with my physician or health care provider and I understand the risks to my child if the screening test is not completed.
- I understand that the law allows a parent or guardian to refuse newborn screening based on the grounds that such examination conflicts with a person's religious tenets and practices. I elect to refuse newborn screening on that such testing of my infant conflicts with my religious tenets and practices. My decision was made freely and I accept the legal responsibility for the consequences of this decision.

Parent or Legal Guardian Name (please print) _____

Parent or Legal Guardian Signature _____ Date ___ ___ /___ ___ /___ ___

Witness Name (please print) _____

Witness Signature _____ Date ___ ___ /___ ___ /___ ___

Original to infant's record, provide a copy to parent, and forward a copy by fax or mail to:
Oklahoma State Department of Health · Family Health Services · Newborn Screening Program Coordinator
1000 NE Tenth Street · Oklahoma City, OK 73117-1299 · Fax (405) 271-4892

For questions, please call the Newborn Screening Program Coordinator at (405) 271-6617 or 1-800-766-2223.

This publication, printed by B & S Quality Printing, was issued by the Oklahoma State Department of Health, as authorized by James M. Crutcher, MD, MPH, Commissioner of Health. 3,000 copies were printed in December 2004 at a cost of $2,047. Copies have been deposited with the Publications Clearinghouse of the Oklahoma Department of Libraries.