

NEWBORN SCREENING SERVICES

INTRODUCTION

Newborn screening is performed as mandated by Georgia Law. Effective January 1, 2007, Georgia law (OCGA 31-12-6 & 31-12-7) and Rules and Regulations (Chapter 290-5-24) require that every live born infant have an adequate blood test for 28 disorders. These disorders include: Phenylketonuria (PKU), Congenital Hypothyroidism, Maple Syrup Urine Disease (MSUD), Galactosemia, Tyrosinemia, Homocystinuria, Congenital Adrenal Hyperplasia (CAH), Biotinidase Deficiency, Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD), Sickle Cell Disorders (SS, SC, S-beta thalassemia), Isovaleric acidemia (IVA), Glutaric acidemia type I, 3-hydroxy 3-methyl glutaric aciduria (HMG), Multiple carboxylase deficiency, Methylmalonic acidemia, 3-Methylcrotonyl-CoA carboxylase deficiency (3MCC), Propionic acidemia, Beta-ketothiolase deficiency, Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD), Long-chain 3-hydroxy acyl CoA dehydrogenase deficiency (LCHAD), Trifunctional protein deficiency, Carnitine uptake defect, Citrullinemia, Argininosuccinic acidemia, and Cystic Fibrosis.

Newborn screening analyses are only performed on dried blood spot (DBS) specimens. Specific instructions for specimen collection, preparation of the requisition form, and guidelines for proper packaging and transport of specimens are described below. Details regarding procurement of collection kits, specimen acceptance policies, turn-around time, result reporting, and result interpretation are also given.

Reports are printed and mailed the day following completion of all testing. Results for presumptive positive specimens are faxed and/or called to the follow-up centers. For Sickle Cell Disorders, follow-up is performed by the Georgia Health Science University and the Newborn Screening Program in the Department of Public Health. For all other disorders, follow-up is performed by the Department of Human Genetics at the Emory University School of Medicine.

Questions concerning the collection and submission of newborn screening specimens or the reporting of test results should be directed to:

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SPECIMEN COLLECTION/LABELLING/REQUISITION FORM

See "Blood Collection on Filter Paper for Newborn Screening Programs", CLSI Document LA4-A5, Vol. 27 No. 21

Materials Needed

1. Form 3491 is a FDA-licensed medical collection device. It consists of a form and an attached filter paper strip for collection of the specimen. A pre-addressed UPS envelope is available for hospitals. Form 3491 and the pre-addressed envelope can be ordered from: Laboratory Services and Supply, 1749 Clairmont Road, Decatur, GA 30033, (phone: 404-327-7921). Clients should order amounts that can be used within six months. Store forms upright in a cool dry area. When submitting metabolic screens to the GPHL, use only the material supplied. The filter paper collection devices must be used prior to the expiration date, which is two years from the printing date. The expiration date is printed on the filter paper portion of the form. Destroy all forms after the expiration date. The forms currently in use include a protective wraparound cover for the filter paper, and do not require an individual envelope for each specimen card.
2. 75% isopropanol
3. Sterile lancets with a point of 2.5 mm in length. A longer point should not be used, because it may pierce the heel bone. Sterile prepackaged lancets designed for pediatric use are available through private vendors.
4. Sterile gauze
5. Gloves

Recommendations for Blood Collection

- a. Infants - The infant should preferably be on a protein diet 24 hours prior to collecting the blood sample. The sample should be collected at least 24 hours after birth.
- b. Early Discharge of an Infant - If the infant is discharged earlier than 24 hours after birth, a specimen should still be collected. The parents should be informed in writing that the child must be retested by one week of age.
- c. Preterm, Low Birth Weight, and/or Sick Infants – Infants who weigh less than 5 1/2 pounds (2500 grams) at birth should be retested when the child is four weeks of age. If the infant is transferred to a Special Care Baby Unit (i.e., NICU) prior to collection of a newborn screening specimen, a specimen should be collected

upon admission and prior to initiation of any treatment (e.g., transfusion, parenteral nutrition, or antibiotics). Collect a second specimen between 48 – 72 hours after birth on infants initially tested at less than 24 hours of age or with a birth weight less than 2000 grams. A third specimen should be collected 28 days after birth or at discharge, whichever occurs first, primarily for infants with a birth weight of less than 2000 grams. For more information, see “Newborn Screening for Preterm, Low Birth Weight, and Sick Newborns”, CLSI Document I/LA31-A, Vol. 28, No. 34.

Blood Collection Procedure

In order to provide sufficient blood, if retesting is required, five (5) spots of blood should be submitted.

If the infant is to be transfused, collect a specimen before the transfusion regardless of the age. If the child is less than 24 hours old, collect a second sample 24 hours after the last transfusion and a third sample 4-6 months after the last transfusion.

The blood collector should wear gloves and take universal precautions for handling blood.

Collect the blood from the infant's heel, using the most medial or lateral portion of the plantar surface of the heel, where "medial" is defined as that closest to the midline of the body, and "plantar surface" as the walking surface of the foot. Do not use previous puncture sites or the curvature of the heel. Do not perform skin punctures on the central area of a newborn's or infant's foot (area of the arch) as this may result in injury to the nerves, tendons or cartilage. Do not perform skin punctures on the fingers of newborns or infants.

Warming the skin-puncture site can increase blood flow to the site. A warm, moist towel or diaper at a temperature no higher than 42C may be used to cover the site for three minutes.

Clean the skin with an alcohol swab (75% isopropanol). Wipe off the excess alcohol with dry sterile gauze, and allow the skin to air-dry. Alcohol residue remaining on the skin may dilute the specimen and adversely affect test results.

To obtain sufficient flow of blood, forcefully puncture the infant's heel with a sterile lancet with a tip no longer than 2.5 mm, or with an automated lancet device. Wipe away the first drop of blood with sterile gauze.

Hold the infant's heel loosely so not to impede the flow of blood. If bleeding does not immediately occur, massage the lower portion of the leg in a downward direction. Avoid milking or squeezing the puncture site, because this may cause hemolysis of the specimen and/or dilute the blood with tissue fluid. If this occurs, the specimen will be rejected as “unsatisfactory, contaminated”.

When a large drop of blood appears, fold back the protective wrap-around cover and gently touch the filter paper to the drop of blood. Allow a sufficient quantity of blood to soak through to completely fill a printed circle on the filter paper. Only apply blood to one side of the filter paper. Do not layer successive drops of blood in the printed circle. If blood flow diminishes so that the circles are not completely filled, repeat the sampling in a new circle. Examine the opposite side to be sure that blood has penetrated through to make a circle which is approximately the same size as the printed circle. Success lies in allowing the blood drop to grow to its full size, and touching with the filter paper when it is about to fall. Repeat this process until all circles are filled. Keep the cover folded away from the wet blood while it is being air-dried. Each circle should be filled with only one application of free-flowing blood. If not, the specimen will be rejected as "unsatisfactory, unevenly saturated".

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.

After the blood has been collected, keep the cover folded away from the wet blood. Allow the blood to dry at room temperature for a minimum of three hours by placing the form horizontally on a table top or rack with the wet blood spots extending over the edge to allow air drying from both sides. Do not place the form inside an envelope until completely dry. Do not put near a heat source, in direct sunlight, or on an absorbent surface. Do not touch the filter paper with your hand at any time.

Requisition Form

The test requisition (form 3491) is combined with the collection device (i.e., filter paper). All of the information requested on the form is important for test result interpretation or physician/parent notification. The infant's name, date and time of birth, submitter (e.g., hospital of birth or clinic), sex, date and time of specimen collection, the infant's healthcare provider (pediatrician), and the telephone number to report abnormal test results are mandatory. The birth weight, protein feeding source, and transfusion status are needed to interpret the results. Complete all fields with block print letters. To aid in researching missing reports, keep the submitter's copy of the requisition. This can be useful in tracking and identifying missing reports.

SHIPMENT

Due to the life threatening nature of several of these diseases, the law requires that the specimen collection, testing, follow-up of suspected cases, and specific diagnosis be performed before the infant is three weeks old. For this reason the blood specimen must be collected when the infant is no less than 24 hours old, but not later than one week of age. **In order to expedite testing and to insure the integrity of the sample, all specimens should be shipped to the laboratory on the day of collection.** Specimens must be completely dry before covering and inserting in the mailing envelope. The dried blood spot specimens **must not** be packaged in airtight, leak-proof bags. The lack of air exchange in the inner environment of a sealed plastic bag may

cause heat buildup, moisture accumulation and/or chemical leaks from the plastic that can damage the specimen integrity. Once collected, specimens can either be mailed to the Georgia Public Health Laboratory using the pre-addressed envelope that is part of the collection device, or they may be sent by courier. Precautions should be taken that mailed specimens are not put in mail boxes. **The specimen must reach the laboratory within seven days of the collection date or it will be rejected as "unsatisfactory, delayed".**

REPORTING/INTERPRETATION OF TEST RESULTS

Normal results for all tests other than hemoglobins are reported as **Within Normal Limits**. Abnormal results for Galactosemia are reported as **Positive** or **Inconclusive**. For all other diseases, abnormal results are reported as **Above Normal Limits** or **Below Normal Limits**, and the value of the analyte(s) is (are) given. For the CF Mutation test, the number of mutations identified (0, 1, or 2) is given, and any identified mutations are listed.

Normal hemoglobin results are reported as FA or AF. Heterozygote results (abnormal hemoglobin plus A) are reported as traits. Homozygote abnormal results (absence of hemoglobin A) are reported as a disease. Traits that cannot be identified by the methods used by the GPHL are reported as "FAV" (V= variant), and confirmation is recommended.

Initial tests for all diseases are performed (or, in the case of overnight assays, started) the day of receipt in the lab or the next working day. Reports are mailed to the hospital of birth and the healthcare provider listed on the NBS from within one day after completion of all testing. This is generally 2-3 working days after specimen receipt. All critical abnormal results are also reported by telephone and/or fax to the appropriate follow-up center (see above) for diagnosis, treatment, and counseling of presumptive positive results.

UNACCEPTABLE SPECIMENS

Specimens will be deemed unsatisfactory to test for the following reasons:

1. Inadequate blood collection/Quantity not sufficient (QNS) - A specimen will be considered QNS if the blood spot has an inadequate quantity of blood, or if the spots are not soaked completely through the filter paper.
2. Oversaturated – A specimen will be considered to be oversaturated if too much blood is allowed to soak into the filter paper, causing the paper to bend, fold or crumple. Oversaturation may also result in clots on the surface of the paper.
3. Delayed – A specimen will be considered to be delayed if it is received in the lab more than seven days after the date of collection.
4. Contaminated – A specimen will be considered to be contaminated if there is visible evidence of dilution of the specimen with a disinfectant, alcohol, water, tissue fluid, or other foreign substance.

5. Scratched/Abraded- Recognizable as unevenly soaked blood in a circular pattern with roughed up filter paper tracks caused by dragging the tip of the capillary across the surface of the filter paper.
6. Unevenly Saturated – A specimen will be considered to be Unevenly Saturated if there is visible evidence of multiple applications of blood causing a layering effect of blood on the filter paper.
7. Roughed Up - Collection of specimen on damaged filter paper so that fibers from paper are standing on end.
8. No Blood – Collection device submitted with no blood applied.
9. No Information - Specimens without a name (infant or mother).
10. Insufficient Information – Specimens missing critical information (i.e., date and time of birth, date and time of collection, birth or current weight).
11. Invalid/Illegible demographics – Dates in the future; date/time combinations that result in the calculation of a negative age for the infant; demographic information that is not interpretable.
12. Obsolete or wrong device – The use of an expired or recalled filter paper device.

** For more information on education and training material, call (404)-657-4143.