SUNY Downstate Medical Center -University Hospital of Brooklyn Network
Department of Pathology Policy and Procedure

Subject: NEWBORN SCREENING FOR METABOLIC DEFICIENCIES

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Supporting Documents:

Approval Workgroup: Laboratory Administration Approval Group

Revision: 3

I. PURPOSE

To provide newborn screening, required by the New York State Department of Health, for detecting metabolic deficiencies.

II. POLICY

1. Newborns admitted to the NICU will have blood samples collected to screen for metabolic disorders: Phenylketonuria, Branched chain ketonuria, Hemocystinuria, Galactosemia, sickle cell disease/trait, Medium chain acyl-CoA – congenital Hypothyroidim, Cystic fibrosis, Congenital adrenal hyperplasia, Biotinidase deficiency and HIV.

2. In the event the parent or guardian of the newborn declines newborn screening because they are a member of a recognized religious organization whose teachings are contrary to the testing requirement, the following must be done:

    - The attending neonatologist along with the hospital administrator will inform the parents of the consequences of refusal.
    - Submit a completed and signed Refusal to Consent form to the Newborn Screening Coordinator and maintain original in chart.
    - The Refusal to Consent form must include a statement that the parent or guardian is a member of a recognized religious organization, has been fully informed of the possible consequences of not having their newborn tested, and is aware of and understands the consequences of not having their newborn tested, and is aware of and understands the possible consequences of refusal.

III. DEFINITION(s)

Phenylketonuria (also referred to as “PKU”)- is a condition associated with an elevation of phenylalanine in the blood. Newborn screening collection is often dubbed “PKU” collection TPN- Total Parenteral Nutrition

IV. RESPONSIBILITIES:

The following departments/responsible parties all share responsibility in maintaining compliance with the law on behalf of UHB’s Chief Executive Officer

- Lab Services
- Attending Physicians
- Physician extenders
- Registered Nurses
- Nursing Station Clerks
- Health Information Management
- Epidemiology
- Newborn Screening coordinator

V. PROCEDURE/GUIDELINES

1. <u>Specimen Collection Timing</u>

Infant Status	**Time of Collection**
Normal, healthy	Day 1 – If to be discharged from hospital or birthing center; and repeated on day 3 – 5. Day 2 – Acceptable

	Day 3 – Optimum
Transfused or TPN Treatment	Prior to transfusion or TPN treatment; If no collection took place prior to transfusion or TPN treatment, collect three days after most recent activity status; and with repeat three months after final transfusion or TPN treatment.
Premature, sick, or extended stay	Prior to transfusion or TPN treatment – any age; and on day 3 – 5 or three days after most recent transfusion or TPN; and at discharge or at one month of age; whichever comes first.
Transferred between hospitals	Transferring hospital provides written notification of specimen collections to receiving hospital. Following transfer, receiving hospital assumes responsibility for collection.
Need repeat specimens	Requests from the screening laboratory for repeats due to abnormal results will supersede these guidelines.

2. The clerk will:
    - Enter order for metabolic screening in the CERNER Laboratory Information System on admission of the newborn to the nursery or NICU and when a follow-up specimen is required.
    - Ensure that pertinent information is entered on the metabolic screening form including hepatitis B surface antigen (HBsAG) test results and HIV test required information.
    - Alert the head nurse/designee if (HBsAG) test results and HIV test information is not available.
    - Record name of the newborn, medical record number and metabolic screening number in the Metabolic Screening Log on the date screening is due.
    - Complete a metabolic screening form and place it in designated section of the newborn's chart.
    - Check Metabolic Screening Log daily to ensure that metabolic screening is ordered on all patients requiring screening (2^{nd} – 5^{th} day of life, after blood transfusion or TPN treatment if no specimen was previously collected, 30^{th} day of life or at discharge).

3. The nurse will:
    - Ensure that parents are informed of the reason for metabolic screening, and receive the New York State Department of Health education handout.
    - Ensure that metabolic screening is performed per New York State Department of Health Law (2^{nd} – 5^{th} day of life, before blood transfusion or TPN treatment, after blood transfusion if no specimen was previously collected. 30^{th} day of life or at discharge).
    - Ensure parents receive the pink newborn metabolic screening slip at discharge.
    - Record date newborn metabolic screening is performed on the NICU Discharge Checklist.
    - Perform metabolic screening prior to transfusion as required.
    - Document specimen collection in the patient's medical record.

4. The Venipuncture Service will:
    - Check that required information is completed on the metabolic screening form prior to specimen collection.
    - Inform the head nurse/designee if required information is not completed on the metabolic screening form.
    - Collect specimen upon completion of form # MCH-3. Once the specimen is collected, the Venipuncture Service staff will initial the Newborn Screening Logbook, newborn's chart and the MCH-3 form.
    - Document specimen collection in the patient's medical record. (CERNER LIS)

5. The pediatrician/physical extender will:

- Ensure newborn metabolic screening is performed per New York State Department of Health Law.
- Follow-up and document HIV results of expedited testing and (HBsAG) test results to ensure timely metabolic screening.
- Notify the Neonatology Fellow if newborn screening specimen is not collected.

6. Newborn Screening Coordinator will:
    - Forward to Wadsworth Laboratories specimen cards (MCH-3) individually enclosed in a transparent, polyethylene, sealed plastic bag. All specimen bags are to be clearly labeled BIOHAZARD on the outside of the bag. Specimen cards must be thoroughly dry before being placed into the polyethylene bags.
    - Forward all MCH-3 specimens, within 24 hours after collection by priority mail to:
      New York State Department of Health
      Newborn Screening Program
      Wadsworth Laboratories
      P.O. Box 509
      Albany, NY 12201 – 0509
    - Receive reports from Wadsworth Laboratories indicating results that are within normal limits or unsatisfactory collections. Abnormal results will be mailed to the staff physician designee.
    - Forward original of reports and any accompanying letters to the UHB Health Information Management Department. Forward copies of reports to Epidemiology Department and to the Metabolic Screening Office.
    - Schedule for re-testing in collaboration with the listed responsible parties, under the supervision of the Laboratory Administrator.
    - Collect and submit all repeat specimens requested by the testing laboratory and maintain all tracking records and documentation.

VI. ATTACHMENTS

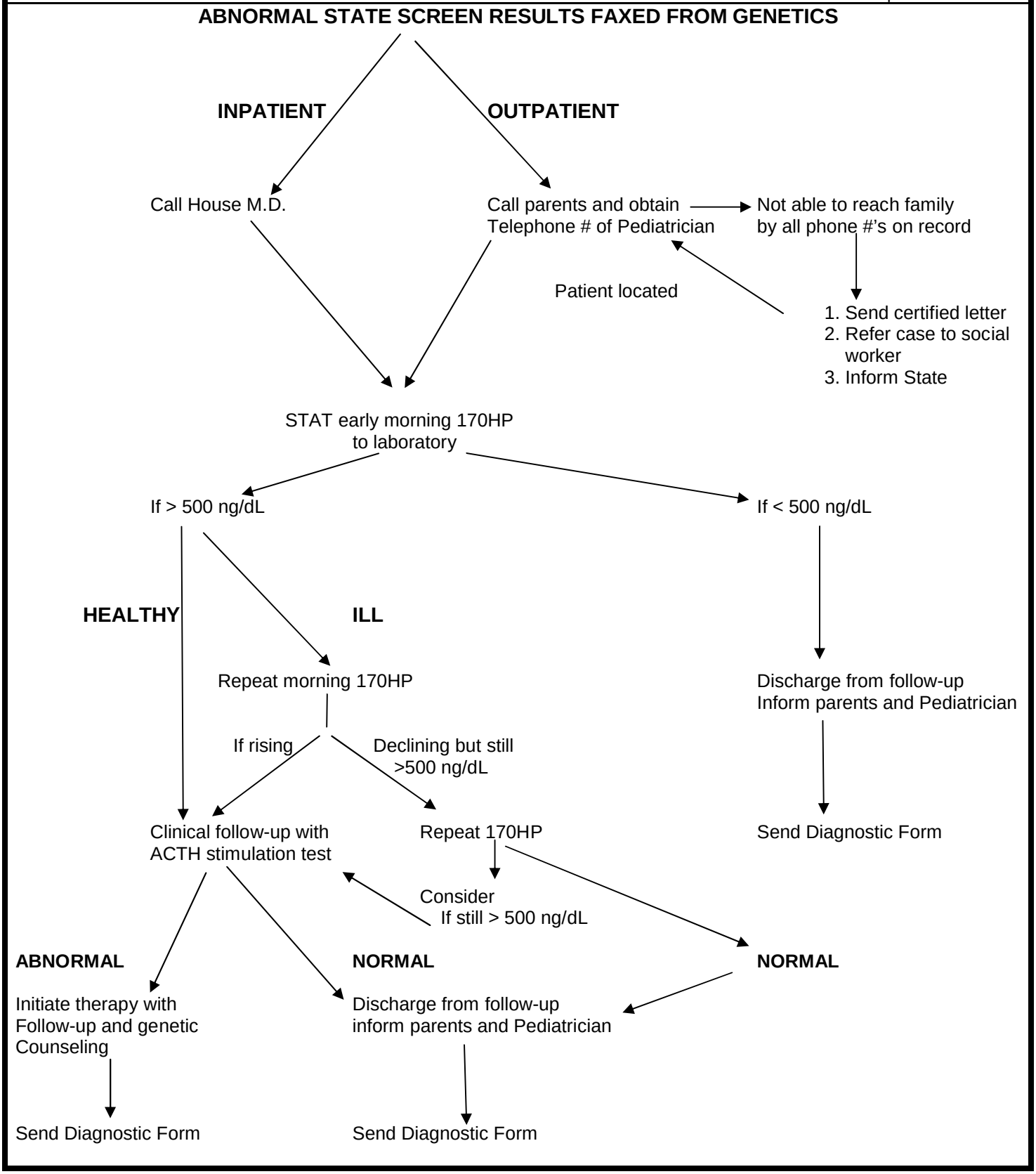
Newborn Screening Instructions for Handling and Mailing Biohazardous Specimens

VII. REFERENCES

PUBLIC HEALTH LAW 2500-a and 2500-f, Subpart NYCRR69-1
Policy #SAF-14

CAH SCREEN FLOW CHART
Division of Pediatric Endocrinology

Updated 2-17-2004



Congenital Adrenal Hyperplasia Protocol
Section of Pediatric Endocrinology

1. Phone call is received from the New York State Health Department Newborn Screening Program – Abnormal 17-OH Progesterone.

2. Contact is made with the Primary Care MD or NICU (most referrals for prematures in NICU setting) – depending on the initial results, either repeat serum 17-OHP is requested, or the baby comes in for visit day of referral or immediately the next morning. If clinically indicated, the baby may be admitted for IV hydrocortisone therapy.

3. If the baby is unable to be located by the end of the business day, referral is made to the Public Health Nursing Agency in that County or the NYS Troopers in an effort to locate the baby and family.

4. Baby is then seen frequently (every 2-4 weeks) for follow-up for laboratory evaluation and growth parameters. Visits are then extended out to every three months.

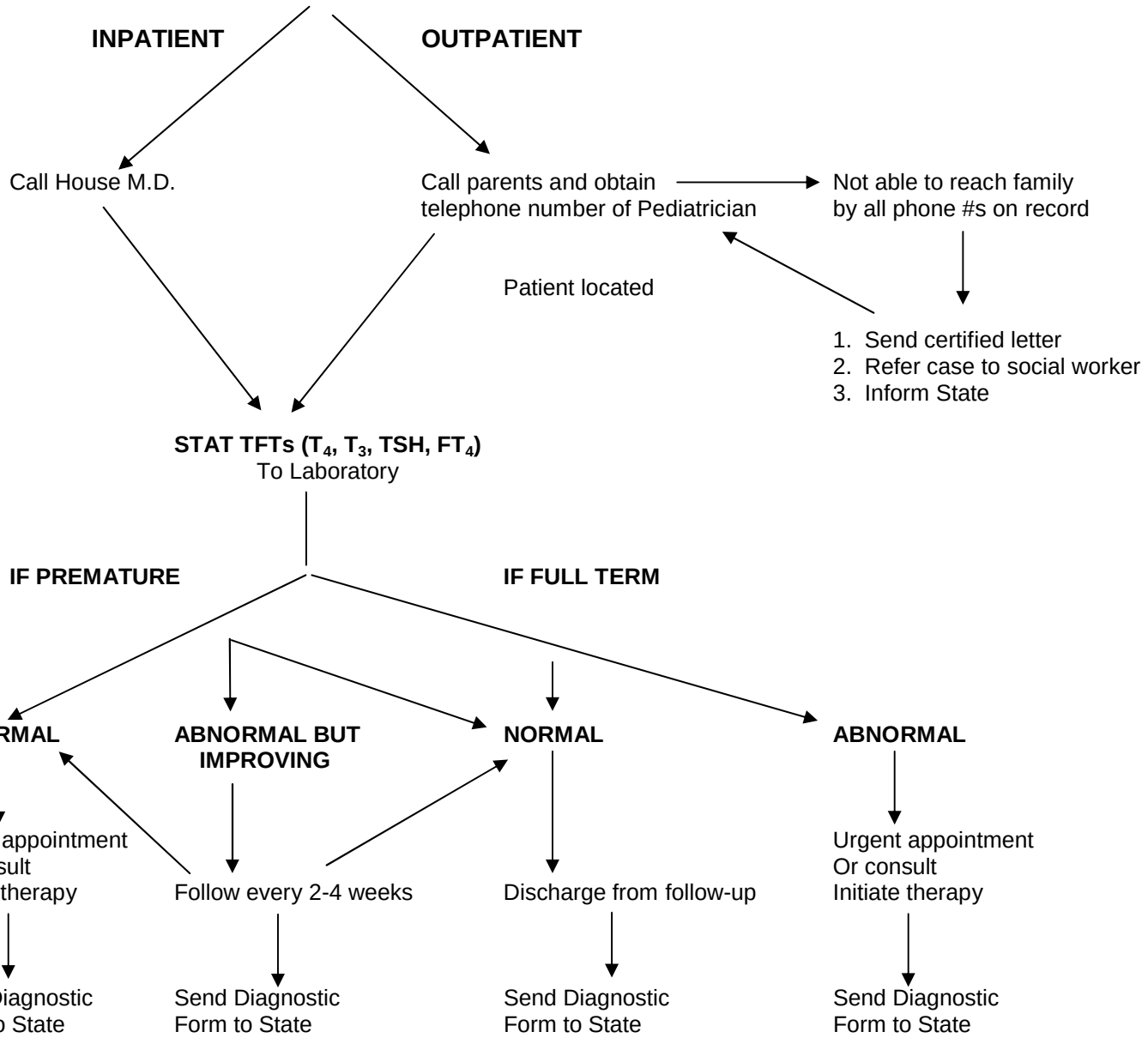
5. At the initial visit, whether the baby is outpatient or inpatient, the parents undergo extensive education, learning proper administration of tablets in infants (crushing and using a syringe type device to administer medication by mouth), giving an intramuscular injection of hydrocortisone in case of emergency, stress dosing of hydrocortisone for illness.

6. Older children are seen every 3-6 months, depending on clinical status.

THYROID SCREEN FLOW CHART
Division of Pediatric Endocrinology

Updated 2-17-04

ABNORMAL STATE SCREEN RESULTS FAXED FROM GENETICS



Congenital Hypothyroidism Referral Protocol
Section of Pediatric Endocrinology

1. Phone call is received from the New York State Health Department Newborn Screening Program – Abnormal thyroid function.

2. Contact is made with Primary Care M.D. – depending on initial results, either repeat serum TFT's are requested, or baby is brought in for visit day of referral or immediately the next morning. If results indicate scan is necessary, an I-123 uptake and scan (2-day test as an outpatient) is scheduled the following day, Tuesday-Friday. Nuclear Medicine is unable to have the tracer dose of radioactive iodine on Mondays.

3. If baby requires thyroid replacement therapy, baby is then started on therapy after the TFT's are drawn, either by our office or the PMD if the child cannot come to Albany, i.e., lives a long distance or the potential for bad weather.

4. Baby with repeat serum TFT's is seen by the PMD, labs ordered ASAP and reported by phone/fax to our office to ascertain whether visit and scan are necessary as well as treatment.

5. If the baby is unable to be located within one business day, the Public Health Nurses and/or the NYS Troopers are contacted to find the baby and family.

6. After the initial visit and initiation of therapy, the baby has laboratory studies obtained within 10-14 days. A follow-up visit is scheduled within the next 4-6 weeks, with laboratory studies as well, and then every 2-3 months after that.

7. Older children with congenital hypothyroidism are seen for follow-up on a clinical judgment basis every 3-6 months.