Newborn Critical Care Center (NCCC) Clinical Guidelines

Newborn Metabolic Screening

GENERAL INFORMATION

1. State policy recommends that a repeat metabolic screen be done on all transfers within 48 hours after admission, **regardless of whether or not a sample was obtained at the transferring institution.**
2. **Repeat newborn metabolic screen should be obtained at 30 days of life for ALL infants who are still admitted to the NCCC at that time.** This is to assess for thyroid anomalies that may not have been caught on the first newborn screen, especially for VLBW infants with birthweight <1500g and infants with congenital heart disease or Trisomy 21.

   *Parents/legal guardians have a right to refuse both the metabolic and hearing testing for their child but **must** sign refusal forms.*

METABOLIC SCREENING

- The optimal time for metabolic screening is 24 - 48 hours of age. The screen must be obtained after 24 hours unless patient is being transfused.
- A blood specimen should be obtained from every infant born at UNC Health Care System. If the patient is **discharged or transferred prior to 24 hours of age, a specimen should be obtained prior to discharge or transfer.**
- Infants discharged and screened prior to 24 hours of age should have a repeat screening test performed by one week of age. It is ultimately the responsibility of the infant's health care provider whose name is noted on the newborn screening form to obtain a repeat specimen in a timely manner.
- An infant should be screened regardless of the status of feedings between 24 - 48 hours of age. The infant may need to be screened again at a later time.
- The blood specimen should not be obtained from a central line if an amino acid solution is being infused.
- Make sure to thoroughly clean the heel with an alcohol swab to remove any soap residue from baths, as this can cause false positives
- Use of capillary tubes to transfer blood is **not** recommended because the tubes disable filter paper and cause over absorption.
- If an infant is less than 24 hours of age and needs a blood product transfusion, it is optimal to collect a specimen before the infant is transfused. Another screen should be sent between 24 - 48 hours of age. **If a transfusion is given prior to the initial screen**, a repeat screen will need to be drawn 4 months after the last transfusion. This is primarily used as a screen for sickle cell disease or trait given the time required for Hb S to increase following a transfusion.

RESULTS AND REPORTING

- Results can be accessed at the NC State Laboratory of Public Health website.
- The reporting of abnormal screening results by the state lab should be accompanied by recommendations for follow-up or additional testing. NCDHHS Newborn Screening Reporting
- If there are questions, discuss these issues with NCCC attending/fellow and consider consulting appropriate subspecialties for further recommendations.

FURTHER SCREENING

Congenital Adrenal Hyperplasia (CAH)

NCDHHS Newborn Screening Reporting

ACMG Congenital Adrenal Hyperplasia ACT Sheet

ACMG Elevated 17-OHP Algorithm

Cystic Fibrosis

- This is a two-tier screening process – first IRT then DNA testing.
- Specimens with an IRT value of < 100 ng/mL and no variants identified are reported as normal for CF
- Specimens are reported abnormal for CF if
  - IRT > 175 ng/mL OR
  - IRT ≥ 100 ng/mL AND one or two variants identified on DNA testing
- Infants with abnormal CF results on newborn screen should then undergo sweat chloride test once the patient is > 2.5 kg. A result of > 60mEq/L is positive for cystic fibrosis (CF).
- *Contact Pediatric Pulmonology to schedule sweat chloride test:*
  Tonya Stafford via Epic in-basket message
  Phone: (919) 966-1055
  Email: trstaff@email.unc.edu

Severe Combined Immunodeficiency (SCID)

Abnormal Result – Contact Immunology for additional testing.

- If the infant is < 28 weeks GA, an abnormal result does not necessarily trigger an immunology consult unless the TREC assay is undetectable. Otherwise, repeat at 28 weeks CGA.

Borderline Result

- Infants with BW < 2300 grams *AND* < 36 weeks adjusted gestational age
  - Repeat NBS every two weeks until one of the conditions below is met:
    1. Screen is normal – no further workup or additional NBS indicated
    2. Infant is 36 weeks CGA and screen remains borderline
       - Consult Immunology
    3. Screen becomes abnormal

        - Consult Immunology
    - Infants BW < 2300 grams *AND* ≥ 36 weeks adjusted gestational age
        - Repeat NBS:
            - Repeat screen normal
                - No further workup or additional NBS indicated
            - Repeat screen borderline or abnormal
                - Consult Immunology

    - Infants BW > 2300 grams
        - Repeat NBS within 48 hours
            - Repeat screen normal
                - No further workup or additional NBS indicated
            - Repeat screen borderline or abnormal
                - Consult Immunology
- If a UNC Allergy Immunology consult is indicated, please place an order for the consult in EPIC with the comment "increased risk for SCID on NBS." Please page the on-call Fellow to alert them of the consult or if you have additional questions.

Thyroid Screening

- See Thyroid Screening and Therapy Guidelines

FOR ADDITIONAL INFORMATION

- Contact UNC Newborn Screening Coordinators for specific disorders:
    - Abnormal amino acids/acylcarnitines or X-linked adrenoleukodystrophy
        - Jennifer Sullivan → jennifer_sullivan@med.unc.edu OR Epic Chat
        - Shauna Coleman → shauna_coleman@med.unc.edu OR Epic Chat
    - Mucopolysaccharidosis type 1 (MPS1)
        - Kristin Clinard → kclinard@email.unc.edu

- NC State Laboratory of Public Health:
    - Newborn Screening website
    - List of conditions tested
    - Contact Number: 919-733-3937 (customer service) or 919-807-8938 (newborn screen director – April Burrell)

- American College of Medical Genetics and Genomics
    - The ACMG ACT Sheets and their accompanying algorithms are a great resource for health care providers looking for information on genetic conditions (identified through newborn screening and beyond) to help inform clinical decision making.