

## CP 3.1 Phenylketonuria (PKU) - Follow-up of Positives

**POLICY:** All PKU confirmatory testing (via filter paper specimen) is to be conducted by the NAPS Laboratories.

### General Information

Total parenteral nutrition (TPN) solutions, frequently administered to babies in NICUs, usually include amino acids that can cause false positive results for PKU and other amino acid disorders. For this reason, when possible, the NBS specimen should be collected before TPN is initiated (as long as it occurs after 12 hours of age). However, screening should not be postponed until TPN is discontinued (unless the discontinuation will occur within 8 hours).

**Cutoffs:** For a newborn screen to be determined as positive for PKU, **both** the ratio of phenylalanine to tyrosine (phy/tyr) must be elevated ( $\geq 1.5$ ) **and** the phe level must be elevated ( $\geq 155 \mu\text{mol/L}$ ).

### Associated Forms and/or Documents:

- 3.1.1 Biopterin – Follow-up for Initial and Recall Results (Positive and Negative)
- 3.1.2 Instructions for Collecting and Shipping Bio-specimens for Biopterin Testing
- 3.1.3 Biopterin Specimen Collection Form

### PROTOCOL FOR INITIAL POSITIVE RESULTS:

Resp. Person	Action
NAPS Lab	<ul style="list-style-type: none"> <li>• <b>As soon as possible but no later than the end of the same day</b>, calls appropriate ASC when initial result is positive for PKU.</li> <li>• Enters Confirmation of Contact (C of C) and result into SIS.</li> </ul>
ASC NBS Coord. or Program Specialist	<ul style="list-style-type: none"> <li>• <b>As soon as possible, but no later than 48 hours after notification from the NAPS lab</b>, calls newborn's physician (and the hospital, if infant is still hospitalized) with results and to arrange for confirmatory filter paper specimen to be obtained using the NBS Test Request Form . Instructs physician/hospital staff to send specimen to NAPS Lab via routine courier (that delivers specimens within 24 hours) or overnight mail.</li> <li>• Enters appropriate tracking event into SIS.</li> <li>• Sends follow-up letter to physician confirming the results on the baby, including <i>Why Retest for PKU</i> brochure with letter.</li> <li>• Contacts the family directly as requested by the physician to arrange for collection of the confirmatory NBS specimen.</li> <li>• If baby has been discharged from the hospital, also sends letter to parent notifying them of need for retesting. Encloses brochure entitled "<i>Why Retest for PKU?</i>"</li> <li>• Notifies health facility where initial specimen was obtained (or if necessary, another facility more convenient to the family) about returning newborn and which test is needed. Provides instructions regarding collection, handling and transport.</li> <li>• Follows case closely until confirmatory testing is completed. After confirmatory specimen is processed, follows protocol for Follow-Up of Confirmatory Results (see page 2 below for protocols for negative and positive confirmatory results).</li> </ul>

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#### PROTOCOL FOR FOLLOW-UP OF CONFIRMATORY RESULTS:

Resp. Person	Action
NAPS Laboratory	<ul style="list-style-type: none"> <li>• Scans TRF or manually enters demographics into SIS.</li> <li>• <b>As soon as possible but no later than the end of the same day</b>, phones and faxes all <u>POSITIVE</u> results to the ASC. Enters a C of C into SIS.</li> </ul>
ASC NBS Coord.  or  Program Specialist	<p><b>POSITIVE CONFIRMATORY RESULT</b></p> <ul style="list-style-type: none"> <li>• <b>As soon as possible but no later than 48 hours after phone call from NAPS Lab reporting positive results</b>, calls the newborn’s physician with results and explains recommended follow-up (medical consultants are available through each ASC to provide additional information and consultation as needed).</li> <li>• Assists provider in referring baby to a CCS-approved metabolic center for diagnostic evaluation, and treatment if indicated.</li> <li>• Assures that metabolic center will obtain blood and urine specimens for biopterin studies (See 3.1.1 <i>Biopterin-Follow-Up for Initial &amp; Recall Results – Positive and Negative</i>) unless baby has siblings with PKU.</li> <li>• Sends or faxes a follow-up letter to PCP confirming results and verifying referral of baby to specific CCS Center. If the center has difficulty making contact with family, offers assistance in making a referral to public health nursing.</li> <li>• Documents all attempts at notification, interactions with physicians and parents using tracking events and case notes in SIS.</li> <li>• Reports unusual occurrences such as missed cases, anomalous/inconsistent results, lost to follow-up cases, delays in contacting family, delays in analysis or reporting of confirmatory results, etc., to the NBS Nurse Consultant/ASC Contract Liaison.</li> <li>• Refers case to Child Protective Services as appropriate, and with approval of NBS Nurse Consultant/Contract Liaison.</li> </ul> <p>Follows case until case is resolved on the Metabolic Service Report (MSR).</p> <ul style="list-style-type: none"> <li>• Resolves case on Case Resolution screen in SIS according to resolution entered on MSR.</li> </ul> <p><b>NEGATIVE CONFIRMATORY RESULT</b></p> <ul style="list-style-type: none"> <li>• <b>As soon as possible but no later than 48 hours after receiving negative confirmatory results</b>, phones physician to report results and closes case in SIS.</li> <li>• Enters appropriate TE’s, Case Resolution, and case notes into SIS.</li> </ul>