Provider action sheet for phenylketonuria (PKU)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for phenylketonuria (PKU), please see the accompanying screening test results.

What steps do I need to take?

1. **Consult with the California Children's Services Special Care Center (SCC) specialist.** They will ask for your assessment of the infant's current health status and whether the neonate has any signs of the disorder as they formulate a diagnostic follow-up plan. The SCC specialist or the NBS Program Area Service Center (ASC) staff will order confirmatory tests and, depending on the result, may schedule a clinic visit for the neonate. Further evaluation may be required to make a final diagnosis. If the infant has a diagnosed condition, the SCC will provide clinical follow-up care.

2. **Contact the family to explain the positive NBS test result.** For information on how best to communicate, consult the **Health Resources & Services Administration Newborn Communication Guide** (https://bit.ly/HRSAGuide or search online for "hrsa" "heritable" "communication").

3. **Remind the family that their infant was positive on a screen, and it is not diagnostic.** Emphasize that you, with the help of ASC staff and the SCC specialist, will guide them through the next steps of confirmatory testing and follow-up services. Please see the accompanying, "Family action sheet for phenylketonuria (PKU)." You should give this information to the family and review it with them to ensure they understand.

4. **Encourage parents** to (1) follow the plan for confirmatory testing right away, and (2) keep their infant's appointments with SCC specialists, if any are scheduled. The family should receive services from a multidisciplinary team of specialists, including genetic counseling services.

Clinical information

PKU is a metabolic disorder caused by variants in a specific gene. When untreated, classical PKU can cause intellectual disability, seizures, and behavioral or developmental problems. Early identification and dietary restriction of phenylalanine intake, beginning with a special PKU infant formula, can prevent long-term problems.

For more information, please visit these sites. Search for the site name and "phenylketonuria" if needed.

GeneReviews
(https://www.ncbi.nlm.nih.gov/books/NBK1504/

MedlinePlus
(https://medlineplus.gov/genetics/condition/phenylketonuria/)

Questions?

For follow-up questions, call the telephone number provided on the screening test result notification. This number can also be found on the center left of the initial patient result mailer.

For program questions, please email NBS Program staff at NBS@cdph.ca.gov or visit the NBS Program website (www.cdph.ca.gov/NBS).

California Department of PublicHealth

California Department of Public Health
Newborn Screening Program (http://www.cdph.ca.gov/NBS)

CALIFORNIA GDSP Genetic Disease Screening Program