Provider action sheet for primary congenital hypothyroidism (PCH)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for primary congenital hypothyroidism (PCH):

What steps do I need to take?

1. **Review** the California state recommended guidelines for follow up on PCH provided by the Area Service Center (ASC) staff. **Confirmatory testing is needed immediately.**
2. **Consult** with a California Children's Services Special Care Center (SCC) endocrinology specialist. They will ask for your assessment of the infant's current health status and whether the infant has any signs of the disorder as they help formulate a diagnostic follow-up plan. 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.
3. **Contact** the family to explain the positive newborn screening 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")
4. **Review** the accompanying, "Family action sheet for primary congenital hypothyroidism (PCH)" with the family and ensure they understand.
5. **Remind** the family that their infant was positive on a screen, and it is not diagnostic. Emphasize that you, with ASC staff and the SCC endocrinology specialist, will guide them through the next steps of confirmatory testing and follow-up services.
6. **Advise** parents to (1) follow the plan for confirmatory testing right away, and (2) keep their infant's appointments with an SCC endocrinology specialist, if any are scheduled. The family should receive services from a multidisciplinary team of specialists, including genetic counseling services.

Clinical information

PCH is an endocrine disorder characterized by absent or decreased thyroid hormones that can affect normal metabolism, growth, and brain development. Treatment, usually thyroid hormone replacement, should be started within the first week of life to prevent development of intellectual disabilities and/or growth delays.

Please visit these sites. Search for the site name and "congenital hypothyroidism" if needed.

MedlinePlus
(https://medlineplus.gov/genetics/condition/congenital-hypothyroidism)

National Center for Biotechnology Information
(https://www.ncbi.nlm.nih.gov/books/NBK558913/)

Questions?

For follow-up questions, please call your NBS Program Area Service Center.

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 (www.cdph.ca.gov/NBS)

CALIFORNIA GDSP Genetic Disease Screening Program