

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. **Consult** today with a California Children's Services Endocrine Special Care Center (SCC) specialist. They will ask for your assessment of the infant's current health status and if the infant has any symptoms or signs of the disorder. Note that affected babies may be asymptomatic. Further evaluation and confirmatory tests 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 newborn screening test result. For information on how best to communicate this, consult the [Health Resources & Services Administration Newborn Communication Guide](https://bit.ly/HRSAGuide) (<https://bit.ly/HRSAGuide> or search online for "hrsa" "heritable" "communication")
3. **Review** the accompanying "Family Action Sheet for Primary Congenital Hypothyroidism (PCH)" with the family and ensure they understand it.
4. **Inform** the family that this result requires more testing and evaluation. Emphasize that you, along with NBS Program Area Service Center (ASC) staff and the SCC Endocrine specialist, will guide them through the next steps of confirmatory testing and follow-up services.

5. **Advise** parents to (1) follow the plan for confirmatory testing right away, and (2) keep their infant's appointments with an SCC 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, which can affect 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 refer to the site below for additional clinical information.

ACMG ACT Sheets and Algorithms

(www.ncbi.nlm.nih.gov/books/NBK55827/)

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](http://www.cdph.ca.gov/NBS) (www.cdph.ca.gov/NBS).

