Provider action sheet for biotinidase deficiency (BD)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for biotinidase deficiency (BD):

What steps do I need to take?

1. **Consult** with a California Children's Services Special Care Center (SCC) specialist. They will ask for your assessment of the infant's current health status and if the infant has any signs of the disorder. The SCC specialist or the NBS Program Area Service Center (ASC) staff will order confirmatory tests. Further evaluation may be required to make a final diagnosis. Once a definitive diagnosis is made, the goal of treatment is to manage the disease.

2. **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")

3. **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 specialist, will guide them through the next steps of confirmatory testing and follow-up services.

4. **Review** the accompanying, "Family action sheet for biotinidase deficiency (BD)" with the family and ensure they understand.

5. **Advise** parents to (1) follow the plan for confirmatory testing right away, (2) keep their infant's appointments with the SCC specialist, if needed, and (3) start treatment if indicated. Infants with BD must take a special biotin supplement every day.

The family should receive services from a multidisciplinary team of specialists, including genetic counseling services.

Clinical information

BD is an autosomal recessive metabolic disorder in which the biotinidase enzyme is absent or deficient. Without effective therapy, symptoms may include seizures, hypotonia, skin rash, alopecia, developmental delay, visual and hearing issues, ketolactic acidosis, and organic acidemias.

With early identification and proper treatment, BD can be managed successfully.

There may be a profound deficiency or partial deficiency.

Please visit these sites. Search for the site name and "biotinidase deficiency" if needed.

- **MedlinePlus** (https://medlineplus.gov/genetics/condition/biotinidase-deficiency)
- **HRSA Newborn Screening** (https://newbornscreening.hrsa.gov/conditions/biotinidase-deficiency)
- **Baby's First Test** (https://www.babysfirsttest.org/newborn-screening/conditions/biotinidase-deficiency)

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