Provider action sheet for galactosemia

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

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. The SCC specialist may recommend the immediate cessation of galactose-containing feeds, including breast milk, pending results of confirmatory testing. Further evaluation may be required to make a final diagnosis.

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. **Review** the accompanying, "Family action sheet for galactosemia" with the family and ensure they understand.

4. **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.

5. **Advise** parents to (1) follow the plan for confirmatory testing right away, (2) keep their infant's appointments with the SCC specialist, and (3) maintain the diet. The family should receive services from a multidisciplinary team of specialists, including genetic counseling services.

Clinical information

Galactosemia is an autosomal recessive disorder caused by pathogenic variants in the *GALT* gene resulting in deficiency or absence of the GALT enzyme. There is a spectrum of symptoms associated with galactosemia, which correlate to GALT enzyme activity. Classic galactosemia presents with hypoglycemia, vomiting, lethargy, poor suck reflex, jaundice, and sepsis. Newborns with classical galactosemia are at risk for acute decompensation. It may be fatal if not treated.

With early identification and treatment, some classic signs and symptoms of galactosemia may never appear. Galactosemia can be best managed though a low-galactose diet.

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

- **Gene Reviews** (https://www.ncbi.nlm.nih.gov/books/NBK1518/)
- **Genetics Home Reference** (https://ghr.nlm.nih.gov/condition/galactosemia)
- **Baby's First Test** (https://www.babysfirsttest.org/newborn-screening/conditions/classic-galactosemia)

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 Public Health
Newborn Screening Program (www.cdph.ca.gov/NBS)