Provider action sheet for sickle cell disease

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

What steps do you need to take?

1. **Consult** with a California Children's Services Sickle Cell Disease/ Hemoglobinopathies Special Care Center (SCC) specialist. The screening methodology results in few false positives, but confirmatory testing is needed. The SCC specialist or the NBS Program Area Service Center (ASC) staff will order tests. The SCC specialist should contact the family to educate them and schedule a clinic visit. When the confirmatory testing is completed, the SCC specialist will determine the type of SCD and share it with the family.
2. **Contact** the family to explain the positive newborn screening test result and tell them to expect the SCC specialist to contact them. 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 sickle cell disease" with the family and ensure they understand. Remind the family that their infant was positive on a screen and more testing is needed. 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. **Consider** prescribing penicillin (125 mg PO BID) if the infant's appointment with the SCC specialist gets delayed; it should be started by 56 days (8 weeks), before the fetal Hemoglobin begins to wane.
5. **Advise** parents to (1) follow the plan for confirmatory testing, (2) keep their infant's appointments with the SCC specialist, (3) give the infant penicillin when it is prescribed, and (4) seek immediate medical evaluation for a fever. The family should receive services from a multidisciplinary team of specialists, including genetic counseling.

Clinical information

SCD encompasses a group of disorders characterized by the presence of at least one hemoglobin S allele. The clinical manifestations of SCD result from intermittent episodes of microvascular occlusion leading to tissue ischemia and chronic hemolysis, both of which contribute to multiorgan dysfunction. Without prompt diagnosis and the initiation of prophylactic antibiotics and pneumococcal conjugate vaccination by 56 days of age (8 weeks), infants with SCD are vulnerable to life-threatening pneumococcal infections. Anyone can have SCD, not just African-American and Latinx populations.

Please visit these sites.

- **Gene Reviews** (https://www.ncbi.nlm.nih.gov/books/NBK1377/)
- **Centers for Disease Prevention and Control** (https://www.cdc.gov/ncbddd/sicklecell/facts.html)

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

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

CALIFORNIA GDSP Genetic Disease Screening Program