Provider action sheet for spinal muscular atrophy (SMA)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for spinal muscular atrophy (SMA).

What steps do I need to take?

1. **Consult** with a California Children's Services Neuromuscular 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. Further evaluation and confirmatory tests will be arranged by the SCC.
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 spinal muscular atrophy (SMA)" with the family and ensure they understand.
4. **Inform** the family that this result requires more testing and evaluation. Emphasize that you, with NBS Program Area Service Center (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) start treatment if indicated. The family should receive services from a team of specialists, including genetic counseling.

Clinical information

SMA is an autosomal recessive neuromuscular disorder caused by variants of the survival motor neuron 1 (*SMN1*) gene. SMN protein is needed to maintain the health and function of the motor neurons. Insufficient SMN protein results in progressive muscle weakness and atrophy, leading to difficulty breathing, eating, crawling, and walking.

Survival motor neuron 2 (*SMN2*) gene also plays a role in producing SMN protein and can estimate the severity and course of SMA. While the number of *SMN2* copies varies from person to person; infants with more *SMN2* copies usually have a less severe form of SMA.

There are several forms of SMA based on the age of onset and maximum motor milestone achieved. More severe forms of SMA are associated with an earlier age of onset and greater motor impairment.

There are disease modifying treatments for SMA that work by increasing the SMN protein. Early treatment is important for reduction of motor neuron and muscular atrophy and best outcomes.

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

- **HRSA** (https://newbornscreening.hrsa.gov/conditions/spinal-muscular-atrophy)
- **Cure SMA** (https://www.curema.org)

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)