Spinal Muscular Atrophy: Expansion of the California Newborn Screening Panel

Richard S. Olney,[1] MD, MPH, FAAP

Myles B. Abbott,[2] MD, FAAP

In July 2020, the California Newborn Screening Program of the California Department of Public Health (CDPH) added spinal muscular atrophy (SMA) to California's state-wide newborn screening (NBS) panel. The NBS program assays blood collected at 12-48 hours of age from nearly all infants born in the state for more than 80 serious but treatable diseases.

SMA is an autosomal recessive neuromuscular disorder caused by deletions of the Survival Motor Neuron 1 (*SMN1*) gene. SMA is a leading genetic cause of death in children and occurs in approximately 1 in 10,000 infants at birth. With California's birth rate of some half million per year, it is estimated that 50 infants will be diagnosed each year.

Screening will identify four types of SMA caused by homozygous deletions of exon 7 in the *SMN1* gene that are found in 90-95% of newborns with SMA. For SMA screen-positive infants, CDPH will:

* notify the ordering physician;
* provide educational information for both providers and families; and
* facilitate referrals to California Children's Services-approved Neuromuscular Centers to initiate definitive diagnostic testing and evaluation for treatment.

In addition to supportive care, current treatments include intravenous onasemnogene abeparvovec or intrathecal nusinersen.

For additional information, please visit the California Newborn Screening Program website or e-mail nbs@cdph.ca.gov.

[1]Division Chief, Genetic Disease Screening Program, California Department of Public Health
[2]Practicing Pediatrician in Berkeley and Orinda, California