EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding spinal muscular atrophy (SMA) to the Recommended Uniform Screening Panel (RUSP) in 2018.

About the disorder

SMA is a rare genetic disorder. Studies of patients with symptoms suggest that about 1 out of every 11,000 people has SMA. People with SMA have a change in the *SMN1* gene that prevents it from making enough of the protein that nerve cells need to survive. Some people make enough of this protein with a related gene called *SMN2*. There are different types of SMA. Most children have SMA Type 1, which causes weakness and, without treatment, can worsen quickly and lead to death.

Treatment for SMA

There is no cure for SMA yet, but early diagnosis allows early monitoring and treatment. Nusinersen is a recently approved medicine that can stop SMA problems from getting worse. When used early in the disease process, it can sometimes prevent damage to nerve cells. Other treatments can also help with certain symptoms, at least for a while. The timing and type of treatment for SMA depends on the disease type.

Detecting SMA in newborns

Newborn screening for SMA can be included with routine newborn screening for other disorders during the first few days of life. Newborn screening for SMA looks for problems with the *SMN1* gene. This process uses the same dried blood spots already collected for screening of other disorders. Newborns missing key parts of the *SMN1* gene are at high risk for SMA. They need more testing to know whether they have the disorder and to identify the right treatment.

Public health impact

Based on what is known about screening and the risk of being born with SMA, experts think that screening all newborns in the United States for SMA would find about 364 babies with the disorder each year. Each year, screening could prevent about 50 infants from needing a ventilator (breathing machine) and about 30 deaths due to SMA Type 1.

Committee decision

The Committee voted in 2018 to recommend adding SMA to the RUSP. As of 2018, the RUSP recommends that state newborn screening programs include SMA.