

Earlier Intervention, Better Outcomes: Pushing the Limits of Early SMA Diagnosis

Newborn screening has reinforced a critical lesson in spinal muscular atrophy (SMA) care: Earlier intervention leads to better outcomes.

Today, all 50 US states include SMA in their newborn screening programs, and Puerto Rico is in the process of implementing screening. Infants identified through newborn screening undergo additional testing to determine their SMN2 copy numbers, helping clinicians predict disease severity and make treatment decisions within the first weeks of life.

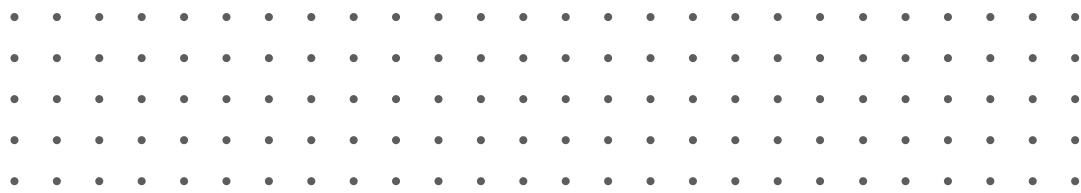
Infants treated with the gene therapy onasemnogene abeparvovec (Zolgensma), who historically would have developed the most severe form of SMA, are now reaching developmental milestones, walking independently, and avoiding the respiratory and nutritional complications that once defined the disease course. Other disease-modifying therapies — nusinersen (Spinraza) and risdiplam (Evrysdi) — may also be in the treatment plan for babies born with SMA.

But many infants with severe forms of SMA already have evidence of disease activity at birth, reflecting motor neuron loss that begins during fetal development. As a result, the focus is shifting from treating symptoms to preventing irreversible damage before it occurs.

Robert Lopez-Alberola, MD, associate professor of clinical neurology and pediatrics, chief of the Pediatric Neurology Division at the University of Miami, and director of the SMA Treatment Center for South Florida, has treated 25 infants diagnosed with SMA. The earliest gene therapy administration occurred at four days of life, while other SMN-targeted therapies have been initiated on the first day of life.

Even small delays can have meaningful consequences. “I have seen how a one-week delay, even in the newborn period, does have a significant clinical impact on the outcome,” he says.

This growing understanding of the neurodegenerative process associated with SMA has driven interest in prenatal treatment strategies.



In 2025, researchers at St. Jude Children’s Research Hospital reported the first known case of prenatal treatment for SMA. In that case, the mother received oral risdiplam during pregnancy, allowing the medication to cross the placenta and reach the fetus. Treatment of the infant continued after delivery using standard newborn dosing. The infant showed no signs of SMA at birth and continued to meet developmental milestones through the first year.

In the St. Jude case and subsequent cases now being studied, the goal is to preserve motor neurons before symptoms emerge and further improve long-term outcomes. “It’s amazing what we’re able to do today,” Dr. Lopez-Alberola says. “We know that the earlier the initiation of treatment, the better the outcome.”

Resources

- MDA Education resources:
 - [Spinal Muscular Atrophy Medical Management](#) webpage
 - [Spinal Muscular Atrophy](#) fact sheet
- Learn about [Cure SMA](#) and the research it’s driving for treatments and a cure.
- Read more about the [St. Jude’s case study](#).