

Starting Treatment for SMA Identified by Newborn Screening: Days Matter

Newborn screening for spinal muscular atrophy (SMA) is now routine in all 50 states. Clinicians are developing best practices and new processes for treating SMA identified through newborn screening.

“It has been an exciting period of time where we have learned from our experiences with our first years of newborn screening,” says Randal Richardson, MD, a pediatric neurologist at Gillette Children’s in St. Paul, Minnesota. “Now, we have the ability to treat these kids with three different medications in that first month of life.”



Early studies of the approved SMA therapies, particularly Spinraza and Zolgensma, show a clear benefit when treatment is started when a child is presymptomatic, compared to starting when they are symptomatic.

“We’ve learned over the years to become very aggressive with the timing between newborn screening and treatment,” Dr. Richardson says. “In my state, I describe the newborn screening alert as a ‘bat signal.’ The providers are informed that a child is positive, the primary care provider is informed, and then we do everything as quickly as possible.” Ideally, the family is alerted the day the screening result comes in, and they begin confirmatory testing and have an in-person meeting with a neuromuscular specialist the next day.

Dr. Richardson’s team has also gained experience working with insurance companies to get prompt treatment for SMA. “When I contact a family about a child with a newborn screen abnormality, I recommend that they call their insurance and make sure that the child is enrolled in their pharmacy benefits because, in some circumstances, it might take a month or two before a child gets a card and is fully enrolled.”

Starting treatment as quickly as possible is crucial, but choosing which treatment or treatments is also important.

According to Dr. Richardson, Evrysdi reaches a steady state after 10 to 11 days of dosing, while Spinraza takes about two months. “Some of the more advanced centers have turned to using an approach where they get Evrysdi to these children as quickly as possible, even if the parental choice is to get Zolgensma,” he says. “It's called the hyperacute Evrysdi protocol. It's basically treating children with a bridging dose until they can receive Zolgensma.”

In cases where SMA type 1 is a risk, Dr. Richardson will begin trying to secure Evrysdi before confirmatory genetic testing is done, so if the results show two or three SMN2 copies, they will have the drug available for immediate use. “I have seen cases where those extra days actually make a difference,” he says.

Clinicians are even beginning to explore management of prenatally diagnosed SMA. Dr. Richardson recommends prenatal amniocentesis for parents of a child with SMA type 1 who are pregnant again. “[This] shaves off a week of time (most state NBS programs do not send results until 5-7 days of life in general),” he says. “Although not a guarantee, some insurances will allow prenatal genetic reports, saving another half-week of wait. If parents elect for early induced delivery, this also may afford even more days, if not weeks.”

Behind each new practice, the guiding principle is that starting a disease-modifying therapy as early as possible leads to the best outcomes for babies with SMA.

Resources

- Watch MDA’s Medical Education webinar [What’s New in Spinal Muscular Atrophy \(SMA\)](#). It is CME-eligible and available on-demand.
- Learn about the [expansion of newborn screening for SMA](#) to all 50 states.
- To help patients learn about newborn screening and find resources, refer them to MDA’s [Newborn Screening for Neuromuscular Diseases](#) webpage.
- Cure SMA’s [Newborn Screening for SMA](#) also answers frequently asked questions and lists resources.