



Make today a breakthrough.

Testimony of

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In support of

Newborn screening for spinal muscular atrophy in Nevada

My name is Stephanie Alcantar from North Las Vegas. I am sharing my family's experience with spinal muscular atrophy, a rare disease, to help inform the Nevada Legislature's Legislative Commission about SMA, as it is called, and to highlight the importance of approving the newborn screening rule under consideration.¹

My son, Aidan, was born on January 28, 2021, with SMA, a rare genetic disease that impacts the muscles used for activities such as walking, eating, and breathing. However, because Nevada does not screen for SMA, we would not receive the SMA diagnosis for another 23 days.

I suspected something during the final two months of my pregnancy. I noticed fewer kicks and other movements. When Aidan was born, he had limited arm movement and poor reflexes, which the doctors initially believed was related to the delivery, such as possible broken bones. We were sent home where he ate well and breathed fine. However, at our 5-day checkup, the doctors noticed further declines and grew concerned over his breathing.

We were sent to the emergency room where the ER doctor immediately suspected SMA, ordering bloodwork and other tests to confirm. That was February 3, 2021. Aidan was five days old. Given his significant decline since birth, the doctor told us Aidan would not make it past six months of age without medical intervention. But without a diagnosis, we couldn't start treatment.

We waited two long weeks for his SMA diagnosis.

It was a costly wait given the rapid degeneration of the disease. Time is everything with SMA. When he was officially diagnosed with SMA Type 1 on February 20, 2021, Aidan went from showing some arm and leg movement to absolutely nothing. He lost his ability

to swallow, and his breathing grew very labored, requiring surgery to insert breathing and feeding tubes.

On March 5, 2021, one month and five days after Aidan was born, he received his first SMA treatment. His SMA treatments are making a world of difference. He can sit unassisted now for about 5 minutes, he no longer requires permanent ventilation, and he no longer requires a G tube for feeding. He has decent upper body strength, can roll over, feed himself, and reach for objects. He especially loves playing with stackable toys, such as large Legos, and stuffed animals.

He has the greatest attitude toward everything and is growing more independent each day. My goal is for him to reach other key milestones, like crawling and walking, but I am also realistic. He lost a lot of strength between when he was born to when he received his diagnosis and treatment.

Time really is everything with SMA, which is why I ask the Legislative Commission to immediately approve the newborn screening regulation. No other Nevada family with babies born with SMA should have to experience the frustration of delayed diagnosis and missed opportunities for better outcomes. Through newborn screening, those families will receive a diagnosis at birth that they can use to make timely decisions about treatment and care.

Unfortunately, Nevada is one of only three states that does not screen for SMA, as recommended by the federal government in 2018. The Nevada Legislature took the first important step in approving legislation in 2019 to ensure that Nevada screens for conditions on the federal Recommended Uniform Screening Panel, or RUSP. The regulations being considered today simply implements the bipartisan 2019 law, while still maintaining an opt-out option for parents.

While approval of the newborn screening regulation will not directly benefit Aidan, it will help other Nevada families of children born with SMA. More than 60,000 Nevada residents are SMA carriers, according to Cure SMA, a national organization that represents people with SMA. If both parents are SMA carriers, every child they have together has a 25% chance of being diagnosed with SMA.

Your action will mean Nevada will finally join the other 47 other states that already screen for SMA. Newborn screening will give Nevada residents born with SMA their best shot at success. It is the right thing to do and I respectfully ask that you immediately approve the newborn screening regulation.

Thank you for considering my views.

ⁱ Newborn Screening Regulation;
https://www.leg.state.nv.us/Register/IndexesRegsReviewed/LCMtg_List_2022_Sept27.pdf