WISCONSIN STATE JOURNAL

Key Joe says no

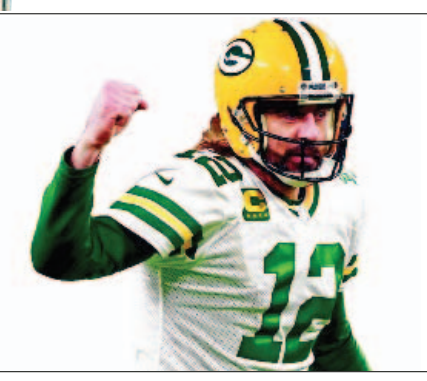
Manchin's rejection of $2 trillion social and environment bill may be fatal blow to Biden's top initiative

NATION. A9

MAJOR VICTORY

Rodgers, Packers clinch NFC North after denying Ravens' late 2-point attempt

SPORTS. B1



Diversity remains at issue

Monona City Council looks at a second contract for equity and inclusion consulting

LOCAL&STATE. A3

Students hone their pitches

Edgewood economics students' final project inspired by television's 'Shark Tank'

LOCAL&STATE. A3

UW VOLLEYBALL | 'THAT WAS PRETTY COOL'

Welcome home, champions

Enthusiastic fans are ecstatic about NCAA national title

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A line of people starting at the doors of the UW Field House and wrapping toward Camp Randall Stadium isn't rare.

It occurs before each University of Wisconsin volleyball match at its raucous home gym. The crowd awaiting entry Sunday was just starting to form lines at the Field House, but it was ready to greet a Badgers squad that made history a night earlier. Fourth-seeded UW captured the first NCAA national championship in program history after a 22-25, 31-29, 25-23, 23-25, 15-12 win against Big Ten Conference rival Nebraska at Nationwide Arena in Columbus, Ohio.

The Badgers defeated previously unbeaten and top-seeded Louisville in the semifinal match Thursday.

Fans were lining up outside the Field House gates more than an hour before they were slated to open for the welcoming party for the newly minted champions,

Please see VOLLEYBALL, Page A8

AMBER ARNOLD, STATE JOURNAL

Fans wait outside the Field House on Sunday as they prepare to celebrate the UW women's volleyball team, which defeated Nebraska in five sets Saturday to win the program's first NCAA title.

NO TIME TO LOSE | FINDING RARE DISEASES IN INFANTS

Prompt diagnosis vital

Treatment improves prognosis in cases of muscle disorder

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Mateo Medina has never walked or talked. The 10-year-old breathes and feeds through tubes.

His brother, Javier, 5, marches around their house in Fond du Lac and talks up a storm, but he can't run or jump and his speech is garbled. He can eat soft foods.

Their sister, Amelia, 3, sprints, chats and eats like most kids her age. An occasional morning tremor is the only sign that she, too, was born with the most serious form of spinal muscular atrophy, or SMA. The genetic disorder, which progressively weakens muscles, typically has been fatal by age 2.

New treatments are changing the face of SMA, and the Medinas are Exhibit A. Mateo was born five years before approval of the first treatment, which Javier received in a clinical trial. Amelia got a newer gene therapy — considered most effective if given within a few weeks after birth, before symptoms appear — when she was 11 days old.

For families without a history of the disease, the main way to catch it early is newborn screening. While Wisconsin and most states have added SMA to their infant testing programs, 12 states haven't, including Alabama, after a federal committee recommended it in 2018.

"A large part of me feels like the state of Alabama failed my son," said Lauren Hendrix, of Troy, Alabama, whose son Graham died in January. He was diagnosed with SMA nearly six weeks after birth after developing symptoms and received the gene therapy, called Zolgensma, two weeks later.

Hendrix

"Had he gotten Zolgensma a month sooner, because (SMA) had been tested for in the newborn screening, I fully believe my son would still be alive today," Hendrix said.

Dr. Mary Schroth, chief medical officer of Cure SMA, based in Elk Grove Village, Illinois, has been

Please see TREATMENT, Page A6

AMBER ARNOLD PHOTOS, STATE JOURNAL

Amy and Adan Medina of Fond du Lac have three children with the most serious form of spinal muscular atrophy, or SMA, a rare disorder that weakens muscles. Treatments that have become available in recent years are most effective if the disease is caught early, usually through newborn screening. From right, Amelia, 3, is barely affected by the condition; Javier, 5, is somewhat affected; and Mateo, 10, is significantly affected.

About this series

Sunday: Wisconsin doesn't screen newborns for some disorders, which can lead to disability and death.

Today: Testing and treatment have greatly improved the outlook for people with a rare muscle disease.

Tuesday: Doctors are increasingly turning to DNA sequencing to explain conditions missed by screening.

Wisconsin first state to test for immune disease

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Less than a week after Amanda and Brandon Hood's daughter Claire was born, they got a call from their doctor. Claire, who seemed healthy, had severe combined immunodeficiency disorder, or SCID, sometimes called "bubble boy disease."

Amanda Hood, a nurse, was familiar with immune deficiencies but not SCID, which affects about 1 in 45,000 babies. "We were very surprised," she said. "I thought maybe it was a lab error."

Further tests confirmed SCID, marked by having few or no T cells to fight infections. Without treatment, the condition is usually fatal by age 2.

Doctors at UW Health's American Family Children's Hospital introduced T cells in Claire's body by giving her a stem cell transplant when she was 4 months old, before showing any

Please see TEST, Page A7

Claire Hood, 8, reading in a swing in her backyard in West Bend. Born with a severe immunodeficiency, she received a stem cell transplant at 4 months old.

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Treatment

From A1

urging all states to add SMA to newborn screening. Other states that haven't are Alaska, Arizona, Hawaii, Idaho, Louisiana, Nevada, New Jersey, New Mexico, Oregon, South Carolina and Virginia. Some plan to start next year.

"An infant being diagnosed with SMA used to be essentially a death sentence, and it's not anymore," said Schroth, who previously was a pediatric lung specialist at UW Health. But, "the sooner a patient with SMA is identified and treated, the better the outcomes."

New treatments

Like most disorders detected in newborn screening through a few drops of blood collected from a baby's heel a day or two after birth, SMA is rare, occurring in about 1 in 11,000 births. If both parents are carriers, each of their children has a 25% chance of having SMA.

The condition is caused by a mutation in a gene that normally makes a protein needed by nerve cells that control muscles. Without the protein, the nerve cells die and people lose the ability to move.

A drug called Spinraza, approved in 2016 for SMA patients of all ages, helps a related gene make more of the protein. Given by spinal injection every four months for life, the drug costs $750,000 the first year and $375,000 each subsequent year.

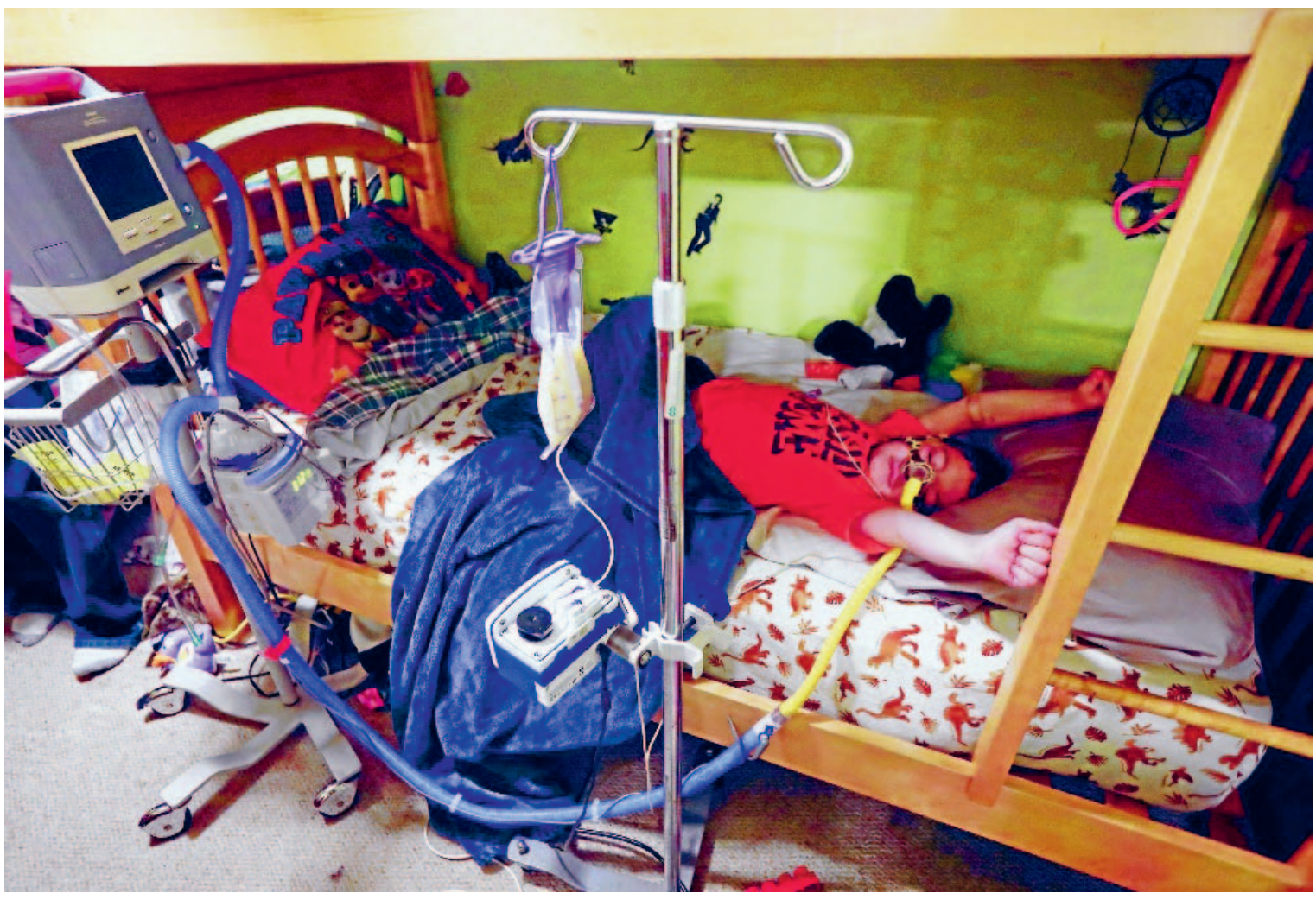
Zolgensma, approved in 2019 for SMA patients younger than 2 years, is a one-time infusion. It uses a harmless virus to replace the mutated gene with a normal one, restoring regular protein production. The gene therapy costs $2.1 million, believed to be the most expensive one-time treatment on the market.

A third treatment, Evrysdi, was approved last year for SMA patients 2 months and older. It's a daily oral liquid that helps make and maintain more of the protein, costing up to $340,000 a year.

Spinraza has helped most patients treated early maintain the ability to swallow and eat by mouth, its maker, Biogen, reported in June. With Zolgensma, most children with severe SMA sat, stood and walked alone at age-appropriate times, according to clinical trial data by Novartis. Most infants who started Evrysdi after developing symptoms were free of ventilators at their first birthday, Genentech said in July.

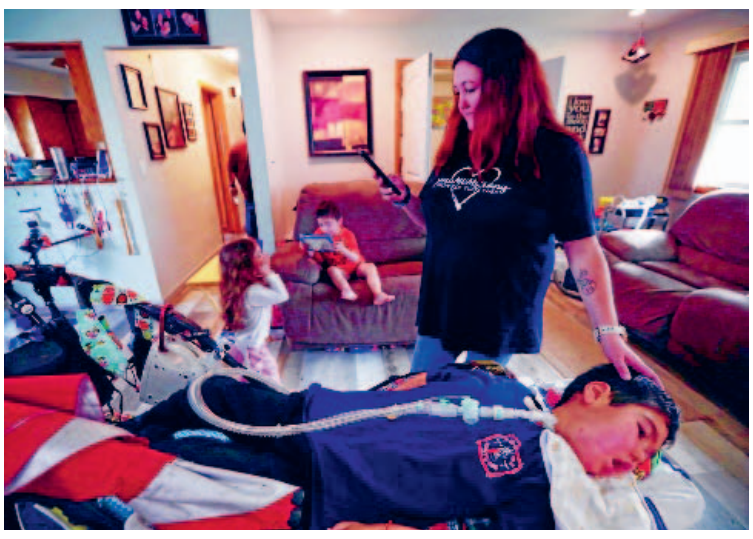
With its hefty, ongoing price tag, Spinraza "exceeds common thresholds for cost-effectiveness," according to the Boston-based Institute for Clinical and Economic Review, or ICER, which studies the cost and benefits of medical treatments.

Zolgensma's price is "right at the upper bounds of what could be considered cost-effective," said David Whitrap, a spokesperson for ICER, which hasn't reviewed Evrysdi.



AMBER ARNOLD PHOTOS, STATE JOURNAL

Javier Medina, 5, wakes up at his home in Fond du Lac. He started treatment for spinal muscular atrophy less than two weeks after he was born, but he still has some symptoms of the muscle-weakening disease. At night, he receives supplemental oxygen and nourishment through a feeding tube.



Amy Medina, with children Mateo, 10, foreground, Amelia, 3, and Javier, 5, said she and husband Adan worked full-time jobs, with opposite shifts, until their daughter was born. "We didn't see much of each other," she said. Adan started staying home with the kids until recently, when he went back to work and she started staying home.

Javier Medina gets breathing treatments to remove secretions from his airway and mucus from his lungs twice a day. His brother, Mateo, gets the treatments three times a day.

Javier Medina, left, plays on his tablet as his brother Mateo lies on his bed at their home in Fond du Lac. Sometimes Javier or his sister Amelia will grab Mateo's hand and help him play a game, their father said. "They include him as much as they know how," Adan Medina said.

Among about 80 patients with SMA at UW Health, including adults with a later-onset form, 17 take Spinraza, a dozen have received Zolgensma and about 50 are on Evrysdi, said Dr. Jennifer Kwon, a UW pediatric neurologist. Insurance coverage is sometimes a challenge for Spinraza and Evrysdi, but most plans promptly cover Zolgensma, especially for infants, Kwon said.

Since Wisconsin added SMA to newborn screening in October 2019, nine babies have been found to have the condition through the testing. Eight have received Zolgensma and one got Spinraza.

One Wisconsin baby who had symptoms of SMA before getting Zolgensma shortly after birth has had physical delays, but the others who got the gene therapy "are doing remarkably well," Kwon said.

What their lives might be like years from now "is a question we can't answer," she said. "While it seems like it's continuing to be effective, we just don't have longer-term data."

Series online

Point your smartphone's camera here to view this series online, including additional photos and video.

Please see TREATMENT, Page A7

ADULTS WITH SPINAL MUSCULAR ATROPHY

New treatment can have mixed results

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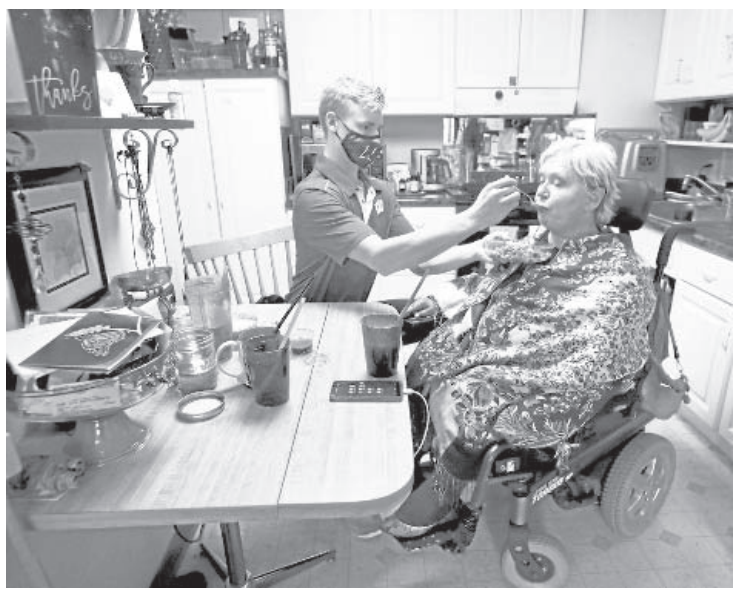
Karen Foxgrover relies on personal care workers to help her eat, go to the bathroom and get dressed. She hoped a new medication for spinal muscular atrophy would give her enough strength to live more independently.

But after swallowing the fruity liquid, called Evrysdi, daily for five months, she's not sure if it has helped with the muscle-weakening condition. She struggles to find enough workers to help her get by day to day.

"I don't feel like I'm getting any weaker," the 64-year-old from Madison said. "But I don't feel like I'm getting significantly stronger. … I don't think I've noticed any difference."



AMBER ARNOLD PHOTOS, STATE JOURNAL

Personal care worker Andrew Wolfram helps Karen Foxgrover eat lunch at her Downtown Madison apartment. Foxgrover, who has a later-onset form of spinal muscular atrophy, has been taking a new drug for the condition for about five months.

Evrysdi, approved last year for SMA patients 2 months and older, is one of two treatments available for adults with SMA. The other, Spinraza, a spinal injection given three times a year, was approved five years ago.

There are four main types of SMA. Type 1, the most common and most severe, is usually diagnosed within the first few months of life and is typically fatal by age 2 if not treated. People with type 2, diagnosed between 6 months and 2 years, and type 3, diagnosed as late as the teen years, generally can move or walk in childhood but eventually need wheelchairs. Type 4, or adult-onset SMA, is very rare.

Tyler Engel, 32, a Madison man who has type 2 SMA, has been taking Evrysdi for a year. His strength has remained about the same, but he doesn't get tired as quickly as he previously did doing activities like wheelchair soccer, he said.

Engel

He hopes the drug will allow him to maintain the strength he still has. "It's life-changing knowing this is the worst-case scenario that I think I'm going to be in," Engel said.

Foxgrover, who has type 3 SMA, moved to Madison in 1982 after graduating from UW-Whitewater with a business degree. She was born and raised in Appleton.

She walked with difficulty as a young child, on the sides of her feet, quickly growing tired from the exertion, she said. By fifth grade, she started using a wheelchair and attending a school for children with disabilities.

Her oldest brother had what was likely SMA and died in his teens. Their other six siblings don't have it, in line with the 25% chance that parents who are carriers will pass it on to each of their children.

When Foxgrover moved into her Capitol Centre apartment in Downtown Madison nearly 40 years ago, she was still strong enough to feed herself and transfer herself from her wheelchair to the toilet and the shower.

The disease eventually left her unable to use her arms and reliant on regular help.

"It's just so obvious how much I've lost," she said. With Evrysdi, "I was hoping I could get some muscles back … but I think I'm a little too old for it."

Test

From A1

symptoms. Now 8 and in third grade, she is doing well, playing with her 3-year-old brother on their backyard swing set in West Bend and writing a short story tentatively titled "Fire Mountain."

"What would have happened if we didn't have newborn screening then?" Amanda asked.

Most states didn't test babies for SCID in 2013, when Claire was born. Only 16 did, including Wisconsin, which five years earlier became the first state to add the condition to newborn screening. SCID was the first disorder included in the federal Recommended Uniform Screening Panel in 2010. By 2018, all states were testing infants for it.

Many Americans are familiar with SCID from the 1976 film, "The Boy in the Plastic Bubble," starring John Travolta, who played a child with the disease based on two real patients.

On Jan. 1, 2008, Wisconsin started a pilot project to test babies for the disorder, using funding from Children's Wisconsin hospital and the New York-based Jeffrey Modell Foundation, named after a boy who died from a related condition at 15.

New molecular technology presented an opportunity for testing, and stem cell transplants were shown to be beneficial, said Dr. Mei Baker, co-director of newborn screening at the Wisconsin State Laboratory of Hygiene.

"With the … transplant, it's the first disorder where we ever can use the word 'cure,'" Baker said.

The state, which has more than 60,000 births a year, has found 19 cases of SCID in babies since 2008.

Claire Hood's transplant helped, but she had to live in isolation until she was nearly 3 because it took a long time for her transplanted T cells to grow. She didn't leave home except for doctor visits, and visitors were limited to her grandparents when they weren't sick.

Once she was able to socialize, Claire was shy during play dates because she was not used to interacting, her mother said. But that soon changed.

In 4K, Claire was exposed to chicken pox, for which she has not been vaccinated because the live vaccine can still be dangerous for her. She received preventive antibodies and didn't get sick.

In school today, she wears a face mask to help prevent COVID-19, as do a few classmates even though West Bend schools don't require them. Amanda stopped working as a nurse because she didn't want to bring infections home. Brandon is a sergeant with the Washington County Sheriff's Department.

Other than washing their hands frequently and trying to avoid sick people, the family lives like any other.

"She's like a normal kid, where she gets sick and she gets better," Amanda said.

Brandon Hood, a sergeant with the Washington County Sheriff's Department, plays with children Claire, 8, and Evan, 3, in their backyard in West Bend.

Treatment

From A6

Seated in her high chair, Piper Droessler bounced up and down, kicked up her feet and ate strawberry-banana yogurt with a spoon — until she decided to scoop it up with her fist and smear it across her face.

A few weeks before her second birthday last month, the toddler chased her older brother and sister around their home near Platteville, occasionally falling on the floor before springing back up and racing again.

Piper was the first child in Wisconsin to test positive for SMA through newborn screening and the first identified through screening to get Zolgensma, 23 days after she was born. Today, she shows no signs of the disease.

"You wouldn't even know," said her father, Ben Droessler.

Caiti Droessler, Piper's mother, interacts online with parents whose children with SMA have died or use feeding tubes. "I don't know their struggle at all," she said.

In Alabama, after Lauren Hendrix's son died, she started a petition to get SMA added to newborn screening. The state plans to start screening for SMA by late January, said Arrol Sheehan, spokesperson for the Alabama Department of Public Health.

Graham Hendrix looked normal at birth in October 2020, but a few weeks later he stopped moving his arms and his head, his mother said. After being diagnosed with SMA, he got Zolgensma at Children's of Alabama hospital in Birmingham. The treatment seemed to help, but he stopped breathing in January and died the next day.

"It blows my mind that we don't test for (SMA) in our state, but we have a team for it at the children's hospital," said Hendrix, who is expecting a daughter in April.

Haley Comer of Middleton, Idaho, said she and medics had to resuscitate her son, Ryder, a month after he was born. Tests showed he had SMA, and he got Zolgensma two weeks later.

Today, at 10 months, Ryder is moving his arms and sucking on his fingers again, but he doesn't sit or hold his head like most children his age, his mother said. Idaho plans to start screening for SMA in February, said Greg Stahl, spokesperson for the Idaho Department of Health and Welfare.

AMBER ARNOLD, STATE JOURNAL ARCHIVES

Dr. Jennifer Kwon, a pediatric neurologist at UW Health, tests Piper Droessler's strength in this photo from January 2020, as her parents Caiti and Ben Droessler look on.

Piper Droessler eats with her family at their home near Platteville a few weeks before turning 2 on Nov. 25. Piper was the first baby to test positive for spinal muscular atrophy through newborn screening in Wisconsin, which started the testing in October 2019. She got a new gene therapy for the condition shortly after birth and shows no signs of SMA.

"It's very frustrating" Idaho hasn't started the testing yet, Comer said. "Getting treatment sooner gives them such a better chance."

Audrey Wiley's daughter Adelynn was born in Storm Lake, Iowa, on June 25, 2020. Six days later, Iowa started screening newborns for SMA.

Adelynn, diagnosed with the disease at 3½ months, got Zolgensma a month later. She started moving more but stopped breathing in January and died.

"It's as bad of luck as it gets," Wiley said. "Obviously, she was not treated soon enough."

Two sons with SMA

Like most parents affected by SMA, Amy and Adan Medina had never heard of the disease before their son Mateo was diagnosed with it a month after he was born in 2011.

He has used a feeding tube since he was 3 months old and a breathing tube, attached through a hole in the front of his neck, since 7 months. He wears diapers. He can't move, except for his eyes, and, very subtly, his feet and left index finger and thumb. "You have to really be watching to see it," Amy Medina said.

Three times a day, his parents or a nurse use machines to clear secretions from his airway and mucus from his lungs.

Mateo spends his days in bed or on a wheelchair-stroller, extended upright to support his spine and prevent the buildup of secretions. He goes to public school, where he has been trying to use a communication device activated by eye movement to converse with fellow fourth-graders.

When the Medinas got pregnant again, they knew they had a 25% chance of having another child with SMA. But, "one in four is also a 75% chance you're not going to have an affected baby," Amy said.

More than five months into the pregnancy, amniocentesis confirmed the baby would have SMA. The news was no easier to accept the second time. "It was still heartbreaking," Adan said.

After Javier was born, the family drove 14 hours to Johns Hopkins University in Baltimore, where he got his first dose of Spinraza in a clinical trial at 12 days. They made 18 trips to Baltimore, most by airplane, before Javier started getting the injections last year in Chicago.

Mateo also got Spinraza, starting at age 6, before switching last year to Evrysdi.

Javier eats — mostly oatmeal, mashed potatoes, cookies and Cheetos — but gets much of his nourishment through a feeding tube at night. He also receives supplemental oxygen while sleeping, especially when he's sick.

In addition to getting treatments to remove secretions and mucus twice a day, the kindergartner receives physical therapy, occupational therapy and speech therapy.

Family photos show him grinning until he was 3, when his facial muscles weakened. "He lost his smile," his mother said. "He says it's broken."

A third child

When the Medinas got pregnant a third time, they figured the odds must be in their favor. But they learned, again through amniocentesis, that their third child would also have SMA.

"I cried, loud, uncontrollably and a lot for a period of time," Amy wrote on the family's CaringBridge page.

Amelia was born in March 2018, when another clinical trial was available, for Zolgensma. After getting the gene therapy at UW's American Family Children's Hospital, she is doing well today.

Outside the family's house, a big red van sits in the driveway, equipped to carry the five Medinas and all of their gear. On a recent morning, Amelia ran along the sidewalk. Javier swung his arms as if he was running, but his feet stayed on the ground. Mateo watched them from his horizontal perch.

Amelia pushed her scooter, adorned with a unicorn on the front. Javier rode his three-wheel, lime green adaptive bicycle, sporting a hotdog costume he likes to wear around the house.

"They're two peas in a pod, but they fight like crazy too," Amy said.

With three children who represent the spectrum of SMA today, the Medinas see the importance of newborn screening.

"Catching it early makes a world of difference," Amy said. "You shouldn't have to wait until your child is showing symptoms to figure out what's happening."