

PRINT-Newborn Screening for Spinal Muscular Atrophy Begins This Month

OSF Lab Assistant's Testimony Supported New Law

A new law that took effect in Illinois at the start of this month will likely save lives and it'll make for a better life for infants who otherwise might not receive early treatment that can have a lifelong impact on their development.

The law adds [Spinal Muscular Atrophy](#) (SMA) to Illinois' infant screening program which means every baby born in the state will be tested for the neurodegenerative disease. This month marks the two-year anniversary of when the US Secretary of Health and Human Services recommended SMA be added to state newborn screening panels. Since then Illinois and Michigan along with 29 other states started SMA screening for newborns with permanent or pilot programs.

SMA is a group of inherited conditions that affect the motor neurons of the spinal cord that control muscles used for activities such as breathing, crawling, and walking. Early treatment can slow disease progression and improve quality of life.

Tracy Parlier and her five year-old daughter Lily were the inspiration behind the Illinois law. Parlier, a lab assistant at OSF St. Mary Medical Center in Galesburg, found herself questioning her then-infant daughter Lily's inability to sit up at five months. Lily was born prematurely and had other health complications, so it took 11 months before she was diagnosed with type II spinal muscular atrophy (SMA) in June of 2016.

Lily needs a wheelchair and has mild scoliosis. She has a machine to assist her coughing and also wears a shaker vest at least once a day to make her coughs more productive. The little girl also has trouble eating, so most of her nutrition is delivered via a port in her stomach, although she occasionally eats a little of her favorites – French fries and popcorn chicken.

Despite the devastating diagnosis, hope arrived with drug called Spinraza, that when administered early – even for babies with the most severe form of SMA – can greatly improve quality of life and chance for survival.

“With early treatment, they're walking, they're talking. They still have SMA but they have a childhood they would never have had otherwise. As a mom, I can't even explain to you what that would mean – the difference,” Parlier said choking back tears as a feisty Lily squirmed, giggled and smiled while sitting next to her mom on the couch.

Tracy and Randy Parlier enlisted help from state Rep. Dan Swanson (R-Alpha) to lobby for Illinois' testing law. The Parliers traveled twice to Springfield to testify. Swanson says their testimony put a face on the disease and helped lawmakers understand the impact of early screening.

“Lily had the whole room just in awe when she grabbed the microphone and started talking into the microphone. It was pretty amazing,” according to Swanson who joked Lily has a future in broadcasting.

University of Illinois College of Medicine at Peoria Genetic Counselor Jennifer Burton worked with the Parliers as they sought to confirm a diagnosis for Lily. Burton is also on the Illinois Department of Public Health's Newborn Screening Disorder Panel and says the legislation will make a significant difference.

“Our hope is to catch those babies at birth with newborn screening and start treatment younger, before they even have symptoms and hope that that will greatly improve their quality of life, reduce complications and of course extend survival.”

Swanson has asked the Illinois Medical Society to educate members about the disease which affects one in 10,000 children in the US. It's estimated one in 50 Americans are carriers.

Parlier said the support has been overwhelming, especially from what she calls her OSF HealthCare family.

“Every Mission Partner is praying for us and supporting us. It just means so much,” said Parlier.

Representative Swanson has also been impressed the support from OSF HealthCare.

“Speaking in front of committees, your employer has to support that. OSF has been there to do that. As she mentioned earlier, the support of the team that she's a part of – to be there to pick up when she can't be there – to pick up the workload is tremendous,” observed Swanson.

Mission Partners (employees) have attended trivia nights and hosted mini fundraisers. One Mission Partner designed and built a new bedframe for the Parlier's eight year-old son Evan to make it easier for Lily to hold and stand while the two play together. Another raised money to get glow-in-the-dark wheels for Lily's wheelchair.

Adam Peterson, a kindergarten teacher in Morris, IL, is the son of a Mission Partner who works with Tracy. His class adopted the Parliers for Christmas in 2018 and created a book called, "[All The Same Yet Totally Different](#)" which he began selling on Amazon.com. [In this Youtube video](#), Peterson explains the project and how proceeds will support Cure SMA, an organization supporting research and advocacy.

Peterson was there to support Lily at Quad City River Bandits baseball game a couple of years ago where Lily was allowed to throw out the first pitch. Former OSF HealthCare St. Mary Medical Center President Jennifer Junis agreed to spend some time in a dunk tank for the "Raise Awareness for SMA" promotion. (Junis is now senior vice president for OSF St. Gabriel Digital Health).

Junis agreed to subject herself to multiple soggy submersions because she says OSF HealthCare's culture promotes Mission Partners getting involved in their communities and supporting co-workers.

"Their stories are so powerful and when they tell their stories and really advocate for something, it's for the greater good. It's really what our Mission is about – serving others."

The larger Galesburg area has also rallied around Lily Parlier, with many posts on her [Longevity for Lily Facebook page](#). Mom Tracy says since she began fighting for the newborn screening law in 2018, Lily has suffered compression fractures from low bone density and her right hip is dislocated and she's not a good candidate for surgery. If the FDA approves a new drug, as expected next month, the family hopes to transition Lily to [Risdiplam](#) in December.

The drug is considered the next generation [orphan drug](#) for treatment of SMA. It is more easily administered; given daily in a liquid form – a big improvement from the spinal injections to deliver Spinraza – and the Parliers look forward to a change that could make life just a little bit easier.