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Florida hopes to reduce ‘diagnostic odyssey’ for children with rare diseases

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Tiem Strouse seemed like a perfectly healthy baby boy when he was born in 2013 in St. Petersburg.

But at about 13 months his development stalled. The baby words that had come early stopped. He would only walk on the balls of his feet and at times would spin around for no reason, mom Liz Strouse said. He also appeared to be suffering from night terrors.

The first specialist that examined him wrongly concluded he had Down’s Syndrome. It was the start of a five-year struggle for his parents to know what was affecting their son’s development**.** It only ended when a genetic test revealed he had a neurodevelopmental genetic disorder caused by mutations in the *ADNP* gene.

Tiem’s path is common for children with extremely rare genetic diseases whose parents face years of heartache putting their child through a barrage of expensive medical tests and appointments with baffled specialists. That search for answers, dubbed the diagnostic odyssey, can average more than five years. Adding to the difficulty is that some genetic tests are not covered by some private insurance providers.

Florida has taken steps in the past two years to make that path easier for families. A 2023 bill provided funding for the state’s Medicaid program to pay for whole genome sequencing, which can cost more than $6,000. This year, the state also provided permanent funding for the Andrew John Anderson Pediatric Rare Disease Grant Program, which will fund research into obscure pediatric diseases.

State Rep. Adam Anderson, R-Palm Harbor, was one of those who sponsored the bill. The program is named after his son, who died aged 4 from Tay-Sachs disease, a rare genetic disorder that causes seizures, paralysis, vision and hearing loss. His son was misdiagnosed half a dozen

times and endured severe seizures before the cause was identified when the boy was 15 months, Anderson said.

“That lit the fire and led the passion to work on these things,” he said. “I really believe genetics in general, and testing and gene therapies are the future of medicine.”

Anderson also pushed for $1 million in state funds for the establishment of the Institute for Pediatric Rare Diseases at Florida State University and, longer term, hopes to file legislation to make genetic testing an option for all newborns.

Evidence backing that approach came from a pilot program run by Nicholas Children’s Hospital in Miami that provided genome testing to 50 children who required neonatal intensive care after birth, he said. A 2020 report on the project, known as Baby Manatee, found that 23 rare genetics conditions were identified among the infants tested, leading to changes in the care given to 19 of them.

The quick diagnosis and elimination of unnecessary medical tests and treatment saved an estimated $2.8 million, the report found.

Tiem, who now lives in Orlando, didn’t require whole genome sequencing but was diagnosed through whole exome sequencing, a subset of the genome.

The test was performed on saliva taken from mouth swabs of Tiem and his mother by GeneDx. The Connecticut company was able to reduce the cost of Tiem’s test to about $400. His father would have been tested too, but he had passed away a few months earlier.

It’s not just rare diseases that testing can identify, said Ashley Arthur, GeneDx’s vice president of market access. Cases of autism and cerebral palsy among other conditions can also be linked to genetic markers.

GeneDx was among those pushing Florida lawmakers to cover genome testing through Medicaid.

“We’re trying to get to a place where we show the value of testing newborns at birth,” Arthur said. “That’s the future vision.”

Tiem’s diagnosis of Helsmoortel-Van Der Aa syndrome, also known as ADNP syndrome, was difficult to accept at first, his mother said. But it gave her a roadmap to give her son as normal of a life as possible.

She knew that her son likely had dwarfism in his hands and feet, meaning they wouldn't grow at the same rate as the rest of his body. With the help of an orthopedic specialist she has been able to identify physical therapy that should delay corrective surgery until he reaches adulthood. It also highlighted slight cardiac abnormalities in his heart that had never been detected.

The night terrors turned out to be seizures. The exome testing provided additional insight into his seizure activity that his neurologist could use to identify the best prevention medication.

Tiem, now 11, has been seizure free for more than two years.

With targeted care, Tiem has been able to attend regular school. He also leads an active life of dance classes, painting, karate and is learning to play a ukulele.

"I'm sad my son has this but now I have the checklist and we can work on this together," she said. "It gave me hope that I can change my son's future so it doesn't have to be as dark as I thought it was."

As of 2019, only 205 affected children had been identified worldwide in medical literature or reported by the ADNP Kids Research Foundation. Tiem's diagnosis meant Liz Stouse found an online network of parents who share the same anxieties.

"I don't have to do this alone," she said.