

Rett Syndrome and Rallying Around Tiana: Why Vega Family Participates in Clinical Trials



In December 2015, the Vega family's world was rocked when they received their two-year-old daughter Tiana's Rett Syndrome diagnosis.

The Mayo Clinic defines Rett Syndrome as “a rare genetic neurological and developmental disorder that affects the way the brain develops, causing a progressive loss of motor skills and speech. This disorder primarily affects girls. Over time, children with Rett Syndrome have increasing problems with the use of muscles that control movement, coordination, and communication. Rett Syndrome can also cause seizures and intellectual disability. Abnormal hand movements, such as repetitive rubbing or clapping, replace purposeful hand use.”

In the five years since learning this news and navigating this new trajectory, there has been no end to the depths this Madison family will go to make sure their daughter has the best of everything that is available to help not only Tiana live her life to the fullest, but also future children and families who may also face this diagnosis.

Victor and Jeannette Vega didn't know after Tiana was born that this diagnosis was looming, as most babies with Rett Syndrome seem to develop normally for the first six to 18 months of life.

Victor shared that as Tiana neared 18 months old, developed skills began disappearing. She also missed certain milestones, including not walking until her second birthday.

Their family began seeking therapy of all kinds for Tiana and wondered if maybe she was just delayed. Victor says that “we all knew she'd be okay and that everything would be fine.”

As Tiana's symptoms worsened, the Vega family sought a neurologist's help.

“Nobody really knew what was going on, but eventually they diagnosed her with partial complex seizures.”

Victor shared that Tiana was prescribed medicine that gave her various negative side effects with absolutely no positive side effects.

“She became super aggressive and very unhappy.”

The Vegas continued seeking answers, and things began clicking when Tiana's neurologist suggested they be a part of the CSER study, a project funded by the National Institutes of Health which provides the means for DNA sequencing to identify the genetic causes of undiagnosed conditions.

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Jeannette Vega | #NowIncluded Member

The CSER project's main goal uses whole-genome sequencing to identify the causes of the symptoms in children with unexplained intellectual and developmental disabilities. Tiana's previous brain diagnostic evaluations did not provide an answer, but the Vega family hoped that whole-genome sequencing would.

After participating in the project, the Vega family then waited six full months before they had results. Then at age 2, Tiana was officially diagnosed with Rett Syndrome.

Though there was a range of emotions after her diagnosis, Victor shared that having some concrete answers meant that they could finally see specialists and get their hands on medicine that actually works.



One aspect of this journey that Victor and Jeannette became passionate about is their participation in clinical trials when the opportunities become available. Victor says that he and his wife are committed to alleviating some of Tiana's day-to-day struggles by continuing to seek out the best treatment.

Jeannette shared that years ago they started doing a deeper dive into Rett Syndrome studies and understand its lifelong implication.

"We've lived through this for four years now of actually knowing what she has, and we want to be part of finding answers so that future families don't have to go through this."

She continued to say that "what you do today may not just benefit your own child but actually the next generation as well, and it's cool to think that we're a part of that."

When asked how Tiana feels about being a part of various studies, Victor shared that they always communicate with her what's going on and make sure that she's onboard.

"She excited about helping other people with Rett Syndrome."

One aspect of clinical trial participation that the Vega family deeply cares about is ensuring that trials of all kinds are diverse. They believe that all sorts of people need to be part of these studies because "our bodies are not the same or responsive to the same things."

The Vega family says that they had another eye-opening experience at a conference several years ago where they saw various research studies and learned that there was very little diversity among those being studied.

Victor also shared that the asthma medicine that he used for years never seemed to work, learning years later that it's actually been proven ineffective for 77% of Puerto Ricans.

"Now I just think back and wonder if that's the sole reason it never worked."

He says that once they learned more about the lack of diversity in clinical trials of all kinds, they both realized "Wow! This is a problem. Maybe this is why that asthma medicine never worked because it wasn't initially tested on someone like me."

After arriving home from their conference, the Vegas say that they started asking themselves how they could help other people and spread the word about encouraging different cultures to participate in clinical trials.

Jeannette, who is half Puerto Rican and half Costa Rican, says "I immediately contacted my family members and said 'Let them test your blood!'"

Victor shared that he doesn't believe clinical trials generally lack diversity on purpose but says that "the way the system goes about asking patients to participate,

where the studies are located, how the pamphlets are communicated, all of it. What happens then, in turn, is that you don't attract a diverse population for your study."

Victor says participating in various trials is never a wasted opportunity because one can get out of it at any time.

"Don't be scared of going into trials because there is always an exit," he says. "Any amount of collected data is helpful to researchers, but ultimately you are in charge of your medical treatment."

Regarding getting a daunting diagnosis, both Victor and Jeannette encourage the family to keep going until you find answers.

"Find those answers, find support groups, and treat properly. This provides a better quality of life for everyone."

"Also, just take the hits as they come," shared Victor. "If you like the rainbow, you have to put up with the rain."



Learn more about racially diversifying clinical trials via Acclinate

This article was inspired by the work that Bob Jones High School graduate Tiffany Whitlow does via Acclinate as the Chief Development Officer. Acclinate says that "There is a significant under-representation of racially and ethnically diverse people in clinical trials, leading to greater health disparity and inequality."

"We believe diversifying clinical trials to include more people of color increases knowledge about health issues and makes a difference in personalized healthcare for all."

To learn more about Acclinate, browse their website [here](https://www.acclinate.com). You can also follow along with their #NOWINCLUDED movement via Facebook, Instagram, or by signing up for our email newsletter.



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