
Uncovering Rett: The Patient Journey to Diagnosis

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Dr. Tu:
Hello. I'm Dr. Jennifer Martelle Tu, and today I have the pleasure of speaking with the caregiver for our patient Zoe. Welcome, Angela. Please introduce yourself.

Angela:
Hi. Yes, I am Angela. I am the mother of Zoe, who's 9 years old with Rett syndrome.

Dr. Tu:
Angela, could you take just a few seconds to tell us a little bit more about Zoe?

Angela:
Zoe is a rambunctious 9-year-old who was diagnosed with Rett syndrome in 2019, so she was about 2 years old. She loves food, she loves to dance, and loves her Peppa Pig.

Dr. Tu:
All right, so today I want to focus a little bit on sort of the diagnostic journeys that some of our families go on. And so I'd like to ask you what were some of the first things that you noticed that were a little different about Zoe's development?

Angela:
I noticed that Zoe kind of was just existing in the spaces with us. She didn't really engage. She was just kind of there and would look past us, or if we spoke to her, she was not like she was really kind of getting it. She wasn't hitting many milestones, and she didn't want to use her legs at all. She would just like to be sat up or propped up by like pillows or a Boppy. Definitely wasn't hitting the regular milestones that I had experienced with my older daughter.

Dr. Tu:
And after you brought some of these concerns to medical attention, were there any specific challenges that you guys faced along the way?

Angela:
Yes. So we got told quite often that there was nothing wrong with Zoe. She was a 35-week-old preemie, and so they kept chalking it up to she was a preemie. “She's just going to be a little delayed. You need to stop looking for there being something wrong with your daughter. Leave well enough alone. You're chasing an empty rabbit hole. There's nothing we can do for you.”

Dr. Tu:
That sounds really frustrating, and it sounds like you had this just gut feeling that something was not right. How long did it take to

actually obtain a diagnosis for Zoe?

Angela:
So it took us 2 years to finally get a diagnosis. And it was by just luck that we happened to be in an emergency room, that a neurologist was on call and took one look at our daughter and was like, “Something's not right. I think maybe you guys need to be seen by genetics.” And that was the first time somebody had actually listened to our concerns and actually looked at our daughter like she was the patient; she just wasn't this long list of unknown question marks.

Dr. Tu:
It sounds like it was quite a challenge to get to the right place. After you got into the genetics clinic, how was it as far as getting the testing done? Was it easy to get the approvals done? Did those things happen relatively quickly?

Angela:
It wasn't as easy as I thought it would be. I just assumed that insurance would be like, yes, sure, let's pay for a testing. And it wasn't. There was a little bit of pushback and some red tape to get through. But once it was approved and we were able to do the lab testing, it was about a 7- to 8-week turnaround time from the date that they drew Zoe's labs, and then we had the phone call with the genetics clinic to give us the results. But it was about, yeah, I'd say 8 weeks.

Dr. Tu:
Angela, I really appreciate you sharing with us that whole process. It really was quite a journey for you and your family to kind of get Zoe where she needed to be. And hopefully the more we can talk with families and share these experiences, hopefully with time it will become less of an obstacle for patients in future diagnoses. So I really appreciate you sharing with us.

I have one final question, if that's okay. I know that Zoe has had some experience with trofinetide, which is the one FDA-approved medication for Rett syndrome. Would you be willing to share some of your experiences with using trofinetide?

Angela:
For Zoe, she's had a lot of GI issues, so that is always kind of where we go with medication is its effect on her, for her gastrointestinal tract. And so having a good team and just making sure that we are doing the best we can to make sure that they're just small little obstacles, and they're not anything that's long term or that's going to deter her. It's been pretty good with her.

Dr. Tu:
Are there any benefits that you feel like you're seeing since starting the medication?

Angela:
Not yet. I haven't seen anything traumatic yet or significant yet, but we're hopeful.

Dr. Tu:
So still early in the process and making sure she tolerates it and doesn't have significant side effects first?

Angela:
Yes. Yeah. We're kind of in that phase of just working out that part of it before we can say this is beneficial or not beneficial.

Dr. Tu:
Well, Angela, I really appreciate you sharing your time with us. Thank you, Angela.

Angela:
Thank you.

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