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Transitioning to Adult Care in Rett Syndrome: Navigating Challenges

Announcer:

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Dr. Gu

This is CE on ReachMD, and I'm Dr. Payal Gu. Here with me today is Dr. Jennifer Martelle Tu. As patients with Rett syndrome move into adulthood, the challenges often shift from diagnosis in early management to then navigating a much more complex and fragmented care landscape. Families are no longer supported by structured school-based services, and clinicians are often faced with coordinating care across multiple specialists that may be less familiar with Rett syndrome. So how do we ensure continuity, maintain quality of life, and support both patients and caregivers during adulthood? Let's explore a real-world case.

So we have a 24-year-old woman who has a history of classic Rett syndrome. That means there was a period of regression of purposeful hand use as well as speech. Now, this case involves a girl who is actually still ambulatory. She never lost any of her motor skills, but maybe her gait is a little bit more off balance or ataxic, and she overall has a milder clinical phenotype. There is no history of epilepsy or seizures. And recently, this family has relocated from Oregon to California, and they're seeking to reestablish care. As she has aged out of the school system, her family is navigating changes in therapy access, community support, and long-term care planning, including concerns around mood, sleep, and caregiver burden.

So when a patient like this enters adulthood, especially in a setting of a new healthcare system, how do you approach maintaining continuity while reestablishing care?

Dr. Tu:

Thank you, Dr. Gu. I really love this question because it highlights one of the major challenges that patients face as they age. So most often, these patients lose their clear pediatric multidisciplinary care, and then they head off into a fragmented adult system, right? So it's very challenging to figure out who do I see, where do I go, who takes care of which part of my medical care? So it's really important in these instances to have direct, clear communication.

So one of the things we really focus on is reaching out to their primary care doctor or their primary care team to make sure that they understand all of the comorbidities a patient might have and which specialists should be involved in their care. But it's really challenging, and many patients face this struggle because it can seem quite fragmented, so it's very important. Clear communication is really the biggest thing.

So Dr. Gu, in that frame of mind, we know that they've now aged out of most of their healthcare system, but what do you do for the

school services? How does that impact the patient? And how can clinicians help fill that gap?

Dr. Gu:

Thanks for that question. It's really important as we're transitioning from the school system to nonschool-based life to maintain activities in the daytime to help improve and maintain cognitive flexibility as well as maintaining the physical skillset of that individual. So that might look like various daytime activities like going out to a community center or a gym with adaptive physical activities to maintain physical mobility, also maintaining motivation and alertness in these individuals. Otherwise, we worry that in the home they're vulnerable to just being in one place, watching TV, and potentially falling asleep and taking naps multiple times a day. And that has a really significant influence on sleep at night, let alone mental health overall.

So coming to the outcomes then that we sometimes see in sleep as a result of lack of activity during the daytime, we worry about difficulty falling asleep, difficulty staying asleep, or waking up too early.

So overall, the exposure to activities outside of the home that benefit both cognitive flexibility as well as physical endurance and mobility are really optimal to the benefits of mental health as well as sleep and helping balance caregivers.

How do you approach reproductive and women's health in patients with Rett syndrome, particularly around menstrual management and its broader impact on care?

Dr. Tu:

Wow, this is a really big topic, and it's certainly something that a lot of caregivers come to us asking about. And I think it's important not to ignore it and really help families understand. And you can kind of break it down into 2 big categories.

The first one really is does something need to be done, right? These are women just like any other woman across the world that is going through their reproductive years, and so they're going to have their menses. And for many of our patients, you don't need to do anything, right? But then there's the bigger question of how is this impacting the family? How is it impacting the patient? Because just like every other woman out there, there can be instances of dysmenorrhea or challenges with menses.

And so in that case we have to think about not only the impact of reproductive management and hormonal management regulation, the side effects there, but also in the microcosm of Rett syndrome, right? And so we know that with Rett syndrome there can be issues with mobility; we're concerned about our bone health overall.

And then if we look at the bigger picture of reproductive management and adjusting hormones, we have to look at consequences and side effects there, things like blood clots or worsening of osteoporosis. And so for each individual patient we have to look at the combination.

And so for a patient that is ambulatory, I might worry a little bit less about the risk of blood clots versus a patient who's not ambulatory, is in a chair all day long, I really have to weigh those side effects and think about the best option.

But similar to every other woman out there, there are lots of choices. And so it's a really important conversation to have, and it really is nuanced. And so it's important to think about each individual patient and find the best thing for that patient and that family.

Dr. Gu:

Dr. Tu, this has been a wonderful discussion. Our time is up. Thanks for listening.

Announcer:

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