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Clinical Pearls in Adult Rett Syndrome Care: Supporting Long-Term Needs

Announcer:

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Dr. Gu:

This is CE on ReachMD, and I'm Dr. Payal Gu. Here with me today is Dr. David Lieberman. Let's review some key clinical pearls to help guide management of older patients with Rett syndrome. With newer therapies now available, how are you thinking about incorporating disease-targeted treatments into the management of Rett syndrome?

Dr. Lieberman:

Thanks for that question. The new agent is trofinetide that was FDA-approved in 2023 for pediatric and adult patients 2 years of age and older. What I hear from my patients most consistently is that their kids who are on the medication are more alert and more engaged with the environment, and they use their communication device more readily. And for most of our patients they're either minimally verbal or nonverbal. They may be able to use an augmentative communication device, like an iPad screen where they could touch the icons, but many of them don't have that dexterity anymore because of Rett syndrome, and they need to use eye gaze technology. And so the families say that their kids are using the device more frequently.

Now, when we're proposing the use of trofinetide for the families, we have to let them know there is a high risk of diarrhea and vomiting, and sometimes weight loss can also be seen. But we can mitigate those problems by starting the medication at a lower dose, increasing the dose slowly over time, and adding fiber supplements to help bulk up the stool, and we get much less diarrhea with that. And even over time, our patients have such a strong tendency towards constipation that even when they're on a medication like trofinetide, they become constipated again in the longer time span.

So we have to utilize trofinetide as a tool along with other agents that we might be using in terms of medications for seizure, medications for muscle tone, medications for sleep or anxiety. So we're hoping that we're going to have other agents that we can add to the armamentarium to help improve the lives of our patients with Rett syndrome.

Dr. Gu, what are some of the most important long-term complications that require continued monitoring even after major interventions?

Dr. Gu:

So orthopedic issues are often persistent even after surgery, so it's really important to maintain the preexisting skillset and mobility that the patient had or to try to get them back into therapies to help maintain those skills despite recovery from surgeries. The things we're thinking about are how is the patient positioned during the day, are they developing contractures, are parents remembering to do range

of motion exercises to maintain lower risk for development of contractures?

Bone health is something we also are always worried about, given that this is a population where there is reduced weight-bearing status, so getting them into their equipment if they have it, like a stander or a gait trainer, helps to stave off risk for lower bone density as they're getting older and improve bone health long term.

In general, many of our patients with Rett syndrome have a high fracture risk. They are often in a malnourished state and also have less fat and therefore also are at high risk for fractures. So these are really important proactive surveillance techniques and prevention strategies that we try to employ and invite our primary docs to also think about.

There's really no clear endpoint to monitoring; this is a lifelong thing.

How do you approach symptom recognition and overall care in patients who are nonverbal while also supporting the caregivers needs over time?

Dr. Lieberman:

Good question. I think it's important to look both at the care of the patient and the care of their family over the longer term. So one thing to take in consideration is the behavior of the individual, especially as they get older, so anxiety can become worse or even depression can set in. Especially as these individuals get older, they may be removed from their normal social settings because maybe their parents are getting older and now they're in a group home and some of their social contacts are lost. And so depression is sometimes something that we have to identify.

The caregiver is really the focal point and helping us to interpret what their child is experiencing, especially if their child is nonverbal and can't let us know.

And we have to support the caregivers as well. Because our individuals who have Rett syndrome, 70% of them are living to their 50s and we have patients that probably are going into their 60s or 70s. So we have to look at the need that the family has to support these kids over time.

One thing we face in Rett clinic is when the child is turning 18, a guardianship has to be placed and that requires a team of physicians. We have a psychologist, a social worker who help determine their eligibility for guardianship, and that has to be processed through the state, through the court, and that's something that will fall on all individuals with Rett syndrome.

Dr. Gu:

Well, I think our time is up, and I think we did well. Thanks so much. We'll see you guys next time.

Announcer:

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