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Clinical Pearls in Rett Syndrome Management: Practical Approaches to Multidisciplinary Care

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Dr. Lieberman:
This is CE on ReachMD. I'm Dr. David Lieberman. Here with me today is Dr. Jennifer Tu. In a previous episode, we discussed a patient with Rett syndrome and the complexity of managing multiple comorbidities. Our focus now is on practical clinical pearls, those real-world insights that can help clinicians deliver more effective and coordinated care every day.

Dr. Tu, when managing a patient with Rett syndrome, what are the most important priorities clinicians should focus on early?

Dr. Tu:
That's a great question, Dr. Lieberman. I think the things that we really want to focus on, the things that are important and kind of keep the case grounded for physicians when taking care of these patients, is to look at the big picture items. The most important things initially, I think, are going to be things like seizure control, nutritional stability, whether or not that means a patient requires a G-tube or other feeding supports. We really want to focus on respiratory safety, making sure that we're focusing on the highest-risk issues first before we dive into some of the other issues. But certainly there's a lot there. There's sleep and anxiety, there's movement disorders, and certainly there's lots of medications that can be considered throughout the course of patient care.

And one last thing we have to remember is that we now have a new medication called trofinetide, which is the first FDA-approved medication for Rett syndrome. And we can certainly look at data from their clinical trials, and we see improvements in behavior scores, both parent/caregiver reports as well as clinician scales.

So what are some of the "don't miss" aspects when you're caring for these patients that you don't want clinicians to overlook?

Dr. Lieberman:
Well, number one, these patients are nonverbal, and we need to support the ability for them to express their needs and wants. And so communication strategies, using alternative and augmentative forms of communication, such as eye gaze technology, is really helpful for the families and others to know what the desires of those children and adults are.

We want to make sure that we keep our patients as healthy as possible over the long term. So we try to survey those areas that need attention, and one of those is looking for cardiac arrhythmias. So we monitor with yearly EKGs, looking at QT intervals to watch if they get prolonged, because that is a risk factor for having a cardiac arrhythmia.

We want to make sure they have good dental care and have a dentist see them. We have our patients followed up by orthopedics to make sure that they are monitoring the spine to see if their scoliosis is developing or significant kyphosis. We want to monitor for hip stability to make sure their hips are well placed and they don't have any hip dislocation. They look at the ankle tone to see if they need orthotics like AFOs.

And importantly, for girls and women as well as boys with Rett syndrome, bone health is an issue, especially for those who are nonambulatory. And there's also thoughts that just the way the bone density is developed over time is impaired in Rett syndrome, so we want to monitor vitamin D levels and calcium intake over time.

Dr. Tu, what care coordination do you feel is critical for Rett syndrome management? What does that look like in practice?

Dr. Tu:
Right. So for these patients, we certainly have to work on coordinating their care. There's a lot there, a lot of specialties that are all involved, and so it's really important to get the full team involved, right? So that often includes social work. It includes working with rehab medicine or orthopedics, that they need medical equipment, all the devices that they might need for their mobility and support at home and in school.

And in school, we want to make sure that they've got the right supports there. So really engaging with the school system, sometimes having education navigators helping families to make sure they have the right education plans. And certainly these are challenging patients because they have a lot of comorbidities, and so we want to make sure they have the right supports for insurance, right? Can they get the right devices? Can they see the right providers? Can they see the right teams? So it's really complex. There's a lot of people involved, and so our job really is to be as supportive as possible, making sure they have all the pieces in place.

Dr. Lieberman:
These practical insights can really shift care toward a more proactive, coordinated approach for patients with Rett syndrome. Thanks for listening.

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