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Managing Rett Syndrome: Building a Multidisciplinary Care Approach

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Dr. Lieberman:
This is CE on ReachMD. I'm Dr. David Lieberman. Here with me today is Dr. Payal Gu. As patients with Rett syndrome get older, their clinical needs continue to evolve. While many core features remain, new challenges can emerge that require ongoing, proactive management. Let's look at how this plays out in an older patient.

Our case here is an 8-year-old male with classic Rett syndrome diagnosed in early childhood with refractory epilepsy. This individual already has severe scoliosis, so they're preparing for spinal fusion. They have had prior hip surgery. They are G-tube dependent, and they have limited mobility.

Dr. Gu, what are the key management considerations in a patient at this stage?

Dr. Gu:
Thanks. So it's really important to recognize for a moment that we do see Rett syndrome in boys, about 3% to 5% of cases. And so they're also an important population to think about. Long term, in a case like this, it's really important to identify at baseline what providers are aware of in terms of a patient's mobility. This is something that the caregiver knows all about, but sometimes it's not shared or noted as easily in the clinic where the child is already sitting in a wheelchair. So it's helpful to know at home, at rest, is the child able to roll over in bed? Do they maintain capacity to sit up from lying down on their own? When they're put into a standing position, can they even bear weight through their legs with assistance or with minimal assistance? That helps to then guide the types of equipment considerations that we make in the clinic and the other complications we think about in terms of relative risk.

So some equipment considerations might be a gait trainer in someone who is still able to bear weight and initiate steps to maintain that mobility. A stander is another type of equipment where someone may not be weight-bearing or able to stand up on their own, but they can assume a standing position with assistance and then help optimize their respiratory health, because they're not sitting hunched over all day. Additionally, considerations made for things like AFOs, which help to maintain ankle positioning and range of movement, which otherwise could lead to contractures. So these are considerations that are important to make leading up to scoliosis surgery.

And also things to prepare for with surgical consideration is, right after surgery, we want to make sure we optimize goals of care and get children working on physical and occupational therapy to make sure that they're able to maintain the skills that they had leading up to surgery, which often could be lost if they're then subsequently in a more compromised state of mobility than they previously were.

And then finally, that leads us to also then think about bone health. So all of these measures even are really guided at preserving mobility and weight-bearing status so that we can think about important bone health considerations for this very special population.

Dr. Lieberman:
Thank you for your response. Some other things I was just thinking about is in this, especially in this boy with Rett syndrome, is seizures and seizure monitoring and seizure control. These kids can be very refractory in terms of their epilepsy. Right now, we don't have any particular anti-seizure medications that are more effective in Rett syndrome than in other disorders, but it's not uncommon for our kids, especially boys, to be on 2 or 3 medications. Ketogenic diet and VNS are also options for seizure control, especially in the boys.

And as these individuals are getting older, especially with all these different surgeries like posterior spinal fusion or hip surgery, having a G-tube in place for adequate nutrition is really critical. Healing is going to take a good amount of nutrition, and so that would really help with the healing process. But the G-tube is also important both for their nutritional status and to prevent aspiration risk.

So when we are following our patients over time, it's really important to do swallow studies periodically to reassess swallow, especially as silent aspiration could occur, and by the nature of it being silent, you wouldn't know.

We have to let families know, though, that the placement of a G-tube doesn't mean that their child no longer can eat by mouth. So they have to first, using the swallow study, figure out what consistencies of food are safest for that child to eat. And then we can just use supplemental nutrition via the G-tube to make sure they're meeting their nutritional goals.

Dr. Gu:
How do you approach behavioral and emotional symptoms in patients with Rett syndrome?

Dr. Lieberman:
So the first thing is it's really hard to assess some of their mental health status when the children are nonverbal or minimally verbal. We know anxiety is a very prevalent feature among our patients with Rett syndrome, and we have medications like SSRIs, like sertraline, or medications like buspirone that could help with anxiety.

Sometimes we just have situational anxiety before they come into a medical visit, maybe giving them clonazepam or other medication like lorazepam might be helpful. We also can have patients who have agitation—intermittent agitation—and sometimes we might want to use medications if behavioral interventions are not effective. Some of those medications could be hydroxyzine or, like I said, clonazepam or lorazepam.

For those who engage in a lot of self-injurious behaviors like biting their hands, biting the wrists, biting others, sometimes we have to use stronger medications like neuroleptic medications, and of course we want to use those at the lowest dose and for the shortest period of time possible.

But we also use other medications like alpha adrenergic agents like guanfacine to help reduce irritability, and they're of variable success.

Ultimately, caring for these patients means staying one step ahead. We need to recognize evolving needs and adapt management to support long-term outcomes. We hope this case review will be helpful in your practice. Thanks for tuning in.

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