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Novel Investigational Treatment Options for Rett Syndrome

Announcer:

Welcome to CME on ReachMD. This episode is part of our MinuteCME curriculum.

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Dr. Neul:

Hello, and thank you for joining us. I'm Jeff Neul. I'm a Child Neurologist at Vanderbilt University Medical Center. And today Joining me is Dr. Alan Percy, who's also a Child Neurologist at the University of Alabama in Birmingham. We're going to be talking a little bit about Novel Investigational Treatment Options for Rett Syndrome.

You know, Alan, at the beginning of the year, you know, we, as we have been since we've been treating people with Rett syndrome, we had no approved - FDA approved drugs for Rett syndrome. So, you know, maybe you could tell us a little bit about how you would manage symptoms of Rett syndrome, when we had no approved drugs.

Dr. Percy:

Yes, so the spectrum that is really quite broad. So first, there is an issue with eating or in bowel movements. So one has to look at whether the swallowing is happening normally, whether there's gastroesophageal reflux, or whether there's constipation. In addition, we have difficulties with sleep, difficulties with development of scoliosis. Some girls have difficulties with interaction with other children and/or with the family, and they may be quite aggressive, or maybe very anxious, can't go out in public because they just go ballistic. So there are a wide variety of therapies that one had to consider across time.

Dr. Neul:

Yeah, I mean, and you think about seizures. And of course, you know, we were - we've always just been using, you know, symptomatic treatments that we'd use for any other thing like constipation, as you mentioned, or seizures. And, you know, I think we all have just clinical experience and anecdotes about things working, but never have had formal tests of it. So we have guidelines on how to take care of people. But you know, they're really practice guidelines based on people's clinical experience.

You know, how do you think, you know, now that we have a first FDA approved drug for Rett syndrome, how do you think this is going to start changing the landscape of management of people with Rett syndrome?

Dr. Percy:

Yeah, certainly the families are quite interested in this agent and are eager to begin the treatment. The question is, one of the principal side effects of the agent is diarrhea, which is diametrically opposed to the problems of the Rett syndrome, which usually is constipation. So it will be important to address the management and whether it's a reduction in dose, a pausing in dose to modify and still maintain the effects of this agent, the positive effects.

Dr. Neul:

Yeah, I mean, obviously, I mean, since nearly everybody with Rett syndrome has constipation that can be quite severe, you know, we have a lot of people, almost everybody it seems like is on some sort of - form of laxative or modification of diet to decrease the

consistency, the hardness of the stool so they have to work on constipation. So I mean, thinking about putting an agent that probably causes diarrhea, we're probably going to have to adjust the medicines that we're giving them to decrease constipation even before starting the trofinetide, which will probably cause this diarrhea, you know, that has the likelihood of increasing diarrhea, which again, the opposite of what we normally see in Rett syndrome, so.

Dr. Percy:

Yeah, this will be a definite challenge, because it's quite opposite to what we have had experienced in the past. And in addition to that challenge is the issue of access for the various of individuals, whether there is private insurance or whether there is Medicaid or even Medicare issues, those will be - will have to be dealt with in the future. Fortunately, there is a single pharmacy which is responsible for providing the medication, and we hope that we gain experience with this rapidly over the next several weeks and months.

Dr. Neul:

Absolutely. Well, it's exciting times to have an actual FDA approved drug for Rett syndrome, and I expect we will see more clinical trials moving forward.

I just want to say thank you, Alan, for joining us, and thank you all for listening to this today.

Dr. Percy:

It was my pleasure.

Announcer:

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