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Living With Rett Syndrome: What to Expect at Each Stage

Announcer:

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

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Dr. Percy:

Good afternoon. This is Dr. Alan Percy. I'm a Child Neurologist at the University of Alabama at Birmingham. And joining me is Dr. Jeffery Neul, who is a Child Neurologist at the Vanderbilt Medical Center. And we're going to discuss Living with Rett Syndrome: What to Expect at Each Stage.

And to start off, this question is: How do we approach the family at the time of diagnosis? Jeff, do you want to comment on that?

Dr. Neul:

Yeah, I think first of all, it's important to listen to the family and their concerns. And I think that sometimes the concerns that the families might bring up, that might help make the diagnosis regarding some subtle developmental delay, and then regression of skills like hand skills and spoken language, which may not be a complete loss, then but the families may be really picking it up. So it's important to really hearing what the family is saying, to identify their concerns. I think this will help make a diagnosis earlier, but also to understand the issues that may be affecting the child with Rett syndrome, and really impacting the family.

Dr. Percy:

Yeah, so I think it's important this is an issue of the family. The age of this child as well as the difficulty itself makes a response directly from the child impossible.

The next question is: What therapeutic communication do we want to give them? And I think it's important to stress what we see as the possible future guides in this child, so we are concerned about growth and development, particular services, physical/occupational therapy, communication therapy, and perhaps even advanced skills with communication therapy. You have anything to add to that, Jeff?

Dr. Neul:

Yeah, clearly there's a need for an interdisciplinary team. I mean, you mentioned the therapists. But we know there's changes that happen over time that you might need to involve people like gastroenterologists or physical medicine doctors or orthopedic surgeons. So I mean, I think that's what really highlights the importance of bringing in together this multidisciplinary team and communicating with each other.

Dr. Percy:

Yes, and it's important to look at this across the progression of the disease. Initially, the child is perhaps a little bit difficult to engage. But as the regression ends, and there is an awakening, so to speak, and there are great improvement, but then begin to appear seizures, breathing aspects. And as time goes on muscle toning increases in their developed Parkinsonian-like features and other issues. But all the time, their interaction is better and better and better.

So the issue then becomes: How do we address the new medication that's now available? And I think that we should offer this wherever a family is willing to invest in this enterprise. What do you think, Jeff?

Dr. Neul:

Yeah, absolutely. And I think so, you know, it's going to be an open question, as we're using this in the real world, how this new FDA approved drug actually changes what might be happening over time. So I think, you know, we have an idea how things progress and what issues come up. But as we use trofinetide, this might change, and our experience may change.

Well, thank you, Alan, for being here and talking with me today. And thanks to everybody for listening.

Dr. Percy:

Yes, thank you. Indeed.

Announcer:

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