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Caring for Adults With Rett: Evolving Needs and Ongoing Challenges

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Dr. Lieberman:
Hi, I'm David Lieberman. I'm the director of the Comprehensive Rett Syndrome Clinic at Boston Children's Hospital. Today I have the pleasure of speaking with the mother of my patient Sammy. Welcome, Jenny.

Jenny:
Thank you. It's a pleasure to be here.

Dr. Lieberman:
Thank you for joining us today. So there are some things we wanted to discuss about challenges and issues for our adult Rett patients and those challenges that have been faced both by the patient and their families, what kind of obstacles they've had to navigate.

And so maybe just tell me a little bit about Sammy’s first diagnosis in terms of the journey it took to get there.

Jenny:
Sure. So Sammy is now 18 years old. She was diagnosed with Rett syndrome when she was 16 months. She developed pretty typically until she was about 9 months old. She reached all of her developmental milestones. At about 9 months, her development started to plateau. We first noticed that she wasn't meeting her gross motor developmental milestones, so she never crawled and she never learned how to walk. At 12 months, we started receiving early intervention. And around 16 months we went to genetics at Boston Children's Hospital, and that was when we received the genetic diagnosis of Rett syndrome.

About that time, she also went through the regression phase of Rett syndrome. So she lost all of her purposeful hand movements. She lost her neat pincer grasp. She lost her ability to pick up any objects. She developed hand stereotypies, so her hands are constantly going to her mouth. And at that point, we started all of the therapies, so speech therapy, occupational therapy, physical therapy. And that's really where our journey with Rett syndrome started.

Dr. Lieberman:
Right, right. I know that's a very hard time to go through. And thank you for sharing.

Tell us about some of the medical challenges that Sammy has faced as she has gotten older with Rett syndrome.

Jenny:
Sure. So a lot of the challenges that Sammy has faced have to do with the fact that she is nonambulatory, so she doesn't walk. She is in a wheelchair, so she's had a lot of orthopedic challenges. She had severe scoliosis, so she's had a spinal fusion. She has hip dysplasia, so she's had multiple procedures on her hips. She used to eat by mouth, and then over time she slowly just lost interest in eating by

mouth, and so we did have a G-tube placed while she was still eating by mouth. However, she's now completely G-tube dependent.

So I think a lot of the challenges that we face now really have to do with her just getting a lot bigger, and it's just physically more difficult to take care of her since she doesn't walk, she can't assist with transfers, so it's challenging for me as her mom. Basically, I lift her to move her. We do have a lift at home, but I find it's just faster and easier for me just to pick her up.

So, yeah, I think a lot of the challenges are just around her getting bigger as she's gotten older.

Dr. Lieberman:
No, and it’s terrific that you can help her through all of this.

What was your experience in getting guardianship?

Jenny:
That's definitely been a challenge. So we started the guardianship process before her 18th birthday. I wish that there was more guidance and support around that process. We did end up having to hire a special needs attorney to help us through the guardianship process, and it took probably about 6 months for us to complete that process. So we didn't actually obtain full guardianship probably until about 4 months after she had turned 18. So that's definitely a challenging process and one where I feel like more resources could be available for our families going through that transition.

Dr. Lieberman:
Yeah, I wish we had more social workers to help our families do that.

Jenny:
Yes, yeah.

Dr. Lieberman:
What is your biggest fear kind of navigating Sammy’s progression into adulthood now? What’s your biggest fear?

Jenny:
Yeah, I think a couple things. I think one fear is the transition away from Boston Children's Hospital into the adult medical care. I know we're still a few years away from that, and I know that she'll still be seen in the Rett clinic, but I do worry about who she's going to see for medical providers outside of Boston Children's Hospital.

I worry about who will take care of her as we, her parents, age. And you feel like you're between a rock and a hard place when thinking about that, because her dad and I plan to take care of her at home for as long as we're physically able. We know that there are residential placements, but that's not the direction that we plan to go in. So I worry about if she outlives me. I can't imagine that, and I can't imagine me not being here to take care of her.

Dr. Lieberman:
To take care of her, yeah. Yeah.

Jenny:
Yeah. Yeah.

Sammy is just overall very happy and content, and she shows so much emotion, and she understands so much. And even though we can't measure her intelligence or her cognition, if you spend any time around her, you just see the love that is in her eyes. You know that there is a person in there. And I feel hopeful about the future with the therapies that are being developed now. And I think that’s it.

Dr. Lieberman:
That's great. No, thank you. To leave it on an optimistic note is terrific.

Jenny:

Yes.

Dr. Lieberman:
Thank you, Jenny, for being here and helping us to better understand Rett syndrome. We'll see you soon.

Jenny:
Thank you.

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