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Case Study: Relatively Late Diagnosis & Management of Rett Syndrome

Announcer:
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Dr. Neul:
Hi, I'm Dr. Jeffrey Newell, the Director of the Vanderbilt Kennedy Center and Professor of Pediatrics at the Vanderbilt University Medical Center. Thank you for joining me today to talk about later diagnosis of Rett syndrome.

So to present this, I'm going to present a case of a child, a 7-year-old girl who comes to a clinic visit for concerns related to neurodevelopmental features. She was born after a normal birth and delivery, and her initial development was very normal in the first year of life. At 6 months old, she was sitting independently when placed, she was holding a bottle to feed, she was cooing with some occasional sounds, such as ‘ma.’ By her first birthday, she had bilateral pincer grasp with small objects, she was pulling to stand, walking with support, and using multiple single words with meanings such as ‘mama’ and ‘dada.’ She started walking independently at 14 months, and by 2 years old, she was using 2-word phrases, climbing stairs with assistance, and scribbling with a crayon. At 3 years old, though, there was a change. She started having decreased social engagement. She seemed withdrawn and not as interactive with her family. Her vocal output declined to only single simple words from the previous 2-word phrases. She was no longer able to draw, she could still use her hands to feed, but she started having trouble walking up and down the stairs. When she was overstimulated, she flapped or even clasped her hands. She was diagnosed with Autism at that time. At 4 years old, she started using 2-word phrases again. She was also starting to climb stairs independently again, but she was having difficulty initiating steps, and gait was unsteady at times, and seemed wandering and slow for her age. She had also started scribbling with the crayon again. By 6 years old, she was using occasional 3-word phrases and starting to copy a circle. She had exome sequencing, which found a pathogenic mutation in the X-linked gene, methyl-CpG binding protein, or MECP2.

So the case presented is representative of a later diagnosis of Rett syndrome. And I think it's important to review what the diagnostic criteria for Rett syndrome are, which is a regression of acquired skills, specifically, purposeful hand skills and spoken language. And then onset of characteristic features, gait abnormalities, stereotypic hand movements. And this regression is stabilized after the regression doesn't continue on. And the majority of people have mutations in MECP2.

Now this case is informative because the regression was somewhat more subtle than people sometimes think. She had a loss of words from multiple words to single words, it wasn't a complete loss, and then she regained it. And similarly, with her hand skills, she was scribbling and then no longer was, and then regained it. She has gait abnormalities and had a history of stereotypic hand movements. And this is very representative of the preserved speech variant, which is really maybe better called the better regained speech variant, where you have a loss and it may be more subtle, but then you might improve and regain some of those skills. She's still behind, obviously at 6 years old. So she does meet the clinical criteria for a diagnosis of Rett syndrome.

And so thinking about that, we can approach the clinical management as we would for anybody else with Rett syndrome. There are clinical care guidelines that have been published and we need to be evaluating her, considering a risk for seizures, motor tone problems,

scoliosis, behavioral issues, GI problems such as constipation. And it will require the multidisciplinary team management, including targeted therapies such as physical, occupational, communication to maximize ability.

Now there are targeted treatments now for Rett syndrome. Trofinetide was approved in March of 2023 for people - by the FDA for people with Rett syndrome over 2 years old, so this could be considered for her. And furthermore, there are ongoing clinical trials either initiated or planned using novel interventions such as gene therapy, which also could be considered for this child.

So in closing, Rett syndrome is a clinical diagnosis. Most people have MECP2 mutations are female, but not all. The regression may be subtle, and skills may be partially regained. Careful assessment and history are very important in making the diagnosis. And you can use the clinical care guidelines to guide your care. And these novel treatments should be considered to improve the lives of affected individuals.

Thank you for joining me today and I hope this was informative.

Announcer:
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