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Clinical Pearls in Recognizing Rett Syndrome: Early Signs and Diagnosis

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Dr. Gu:
This is CE on ReachMD, and I'm Dr. Payal Gu. Here with me today is Dr. Edward Dabrowski.

In real-world practice, patients don't always follow a textbook pattern, so recognizing when to pursue evaluation can significantly impact time to diagnosis and patient outcomes, and that diagnosis really matters. It's what opens the doors to appropriate specialist referrals, helps to connect families to the right support systems, and ensures that patients can access the targeted therapies appropriate for their diagnosis. It also provides opportunity for genetic counseling and increasingly determines eligibility for emerging treatments, including gene- or genetic-based therapies.

So our focus now is on the key insights that can help recognize Rett earlier and navigate the diagnostic process with greater confidence.

So, Dr. Dabrowski, in your experience, which symptoms or patterns of presentation are most suggestive of Rett syndrome? And how can clinicians avoid missing them in everyday practice?

Dr. Dabrowski:
I think really the biggest piece is the developmental regression, and I'm going to go back then to basically the 4 criteria that define Rett, and that includes partial or complete loss of purposeful hand skills, a substitution of those hand skills with nonsensical movements, handwashing, rubbing, etc.

You're also very concerned when you see that language that is beginning to emerge all of a sudden disappears. Kid no longer says mama or dada, or what have you, which is very, very, very upsetting to the parents and is actually one of the big things that parents will tell you in their histories first up. And then finally, if the child is ambulatory, their ability to walk is reduced and unstable, etc.

Dr. Gu:

Thank you for reminding us again of those key 4 features that characterize the major criteria for Rett syndrome.

So, Dr. Dabrowski, what are your key clinical pearls you would give to others when it comes to the diagnostic workup for suspected Rett syndrome?

Dr. Dabrowski:
Well, we most certainly have talked about the criteria, and then we would look for atypical criteria as well, where an individual has 2 of the 4 main criteria and at least 5 of the 11 supporting criteria. And these including breath issues—or breathing issues I should say—bruxism, abnormal muscle tone, and a series of other symptoms.

But on top of that, when we look at the child as a whole, we get an understanding of its phenotype. What does it really look like? And you'll have children that have a classic—not necessarily completely classic—but most certainly if you take a look you'll see a classic facies. You'll start seeing head circumference reducing. You'll see them protruding their upper lips, for example, or making very odd lip movements. They'll also make eye movements that are somewhat different. If they're older, they'll cover up, etc. Then you'll see their loss of their coordination, their tone, etc. So those are the things that you're looking for as a clinician.

So the most important thing is the regression, and that is lost many times in the history. But talking to the parent, they'll always come back to that, and that leads you down the correct path.

And if it comes to the actual diagnostic workup, some people might do an MRI or an EEG beforehand. But truthfully, it comes down to the genetics and looking for that MECP2 gene or variants.

Dr. Gu:
Yeah, these are some really great points. It's once you kind of look back at that genetic diagnosis and see an MECP2 mutation that maybe you'll go back to the family and say let's make sure we didn't miss a regression. Was there a change during this period of time in these skill sets? And sometimes families will then bring them up. You bring up a really good point.

And with that, thank you so much. This has been such a wonderful learning opportunity. We hope you all have found these perspectives useful, and thank you again for listening to us.

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