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Case Study: Diagnosis, Treatment, and Management of Rett Syndrome

Announcer:
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Dr. Percy:
Hello, I'm Alan Percy. I'm a Child Neurologist at the University of Alabama in Birmingham. Today, we're going to talk in Case Study, about the Diagnosis, Treatment, and Management of Rett Syndrome.

The recent history of this young girl is that she's a 2.5-year-old who had a recent diagnosis of Rett syndrome. She appeared normal at birth, and was a good baby who did not advance quickly and was slow to roll over or sit up on prompt. She lost fingers skills and her first few words at about 12 months of age, and developed hand stereotypies in the form of hand patting and hand mouthing. She was referred to a geneticist who suggested Rett syndrome and genetic test had identified a mutation in the gene which has been associated with this disorder.

The current problem is that she was seen by her pediatrician for sleep issues, both in going to sleep and maintaining sleep. She had experienced no seizures or breathing issues, and these were not issues of concern. Sleep is a common comorbidity related either indirectly or directly to Rett syndrome. This girl had no history of noisy breathing or snoring, suggesting any obstructive sleep apnea, but the mother had noticed occasional spit-up on the bed close.

So, potential issues therefore are altered feeding. Is she a slow or poor eater, with poor swallowing, frequent choking or coughing? Does she have GE reflux, delayed stomach emptying, or constipation? And it's important to review the history of breathing during sleep to be certain. And any history of sudden arousals after being asleep.

Diagnostic suggestions therefore include you need to check the growth pattern and type of diet in this young lady. Growth pattern in girls with Rett syndrome is slower than normal in most individuals, with weight increase dropping below the 2nd percentile by age 2, and in length by at least age 12. The responses to feeding or altered bowel movements also need to be followed. Does this girl have constipation? That is common in Rett syndrome, and is important to consider that. Also difficulties with feeding are very common. It's important to examine the abdomen for distension or stool pattern, and obtain an x-ray of the abdomen if necessary. You can consider a referral to a gastroenterologist as well.

If the issue is poor swallowing, the girl needs a swallow study, which is easy to perform to see if various thickening agents in food make it more easy to follow the normal feeding route. Failure to pass a swallow study indicates the child may need a gastrostomy tube. About a third of girls with Rett syndrome ended up with one of these. If it is GE reflux, H2 blocker or proton pump inhibitors should be included. If constipation, you need to suggest polyethylene glycol or MiraLAX.

If none of the above, consider sleep aids. For the younger individual, we use gabapentin at 10 to 30 milligrams per kilogram at bedtime. You also can use Klonopin. Antihistamines are a poor choice as a chronic medication because the individual can adapt to the medication, and it loses its efficacy.

With the new treatment now available for Rett syndrome, it is important particularly to pay attention to the growth and to constipation. Because one of the principal side effects of the new agent is diarrhea, it's important to stop the polyethylene glycol before the agent is begun, and to pay close attention to the frequency of bowel movements after the medication is begun. This may require either lowering the dose or a dose holiday to get things back in normal pattern.

Furthermore, weight loss was seen in slightly more than 10% of individuals. And as that goes along with Rett syndrome, one must be certain to differentiate the two. This isn't part of the typical pattern of Rett syndrome, but is in fact related to the new agent.

I hope this has been helpful. And thank you very much for your attention.

Announcer:
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