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Compassionate Communication: Keeping the Patient and Family Informed Throughout the Patient's Journey Through the Four Stages With Rett Syndrome

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

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Dr. Percy:
Hello, I'm Alan Percy. I'm a Child Neurologist at the University of Alabama at Birmingham. Today we're going to talk about Compassionate Communication: Families Who Have a Child with Rett Syndrome.

To review, Rett syndrome is a developmental encephalopathy which mainly affects females and is due mainly to a mutation in the gene MECP2. Because all girls with Rett syndrome do not have this mutation, the diagnosis is clinical. These girls are apparently normal at birth and through early development, although there are key features that may suggest that they are going to have difficulties. There are multiple comorbidities and a broad clinical spectrum.

Initially, there's an arrest of developmental progress with a regression followed that - following that, with reduced or absent speech and finger skills, absent or abnormal gait, enhanced stereotypies principally the of the hands, wringing and mouthing, and so on. Later on, there's a stabilization with better social contact and very good eye interaction. But over time, over the next 10 or 20 years, there's a gradual slowing of motor function. It's important to address these issues with each family.

So initially, during early infancy, there is an arrest of developmental progress. Now, I said development was apparently normal, but during the first 6 months, one may note the deceleration in the rate of head growth, which is greater than normal. And these girls may appear to be too good. They eat and sleep, but they don't fuss much. But between 6 and 18 months, there is a slowing of development. They may be slow to sit, slow to play with toys, or finger feed, and instead of developing a normal crawl, they may develop a combat crawl or scoot around or shuffle as they sit on their bottoms, and then hand stereotypies begin.

Later on, where there's regression, there is a partial or loss of skills, but they also may be less interactive or appear as autistic, in their own world. Seizures and abnormal breathing are rarely seen at this stage.

Following the regression period is a post-regression period, where eye contact is improved, there's better interaction, they emerge from this on uninterested or detached phase. However, seizures, abnormal breathing, slowing of growth may begin and may become more and more apparent. Walking, over time, which occurs and about half of the girls, may slow and become more difficult for them. Scoliosis may be seen as early as 4 years of age, but progresses usually after age 8, especially if they're not walking. There's also a poor feeding pattern, and many of them, up to a third may require alternative routes of feeding, such as gastrostomy tube, and premature puberty may emerge.

Finally, there is a period of late motor decline. During this period, which may be seen as early as 10 or 15, but more likely a little bit later, where there's an increase in muscle tone, which is different from what they - how they appear at early on, and the development of

rigidity which may mimic that of parkinsonism. There are other issues including dystonia and contractures. Seizures and breathing abnormalities may diminish as they age, and yet they remain alert, interactive, and happy.

So it is important to stress the positive and to make certain that the parents are devoted to looking for things that may occur.

We've made significant progress and understanding Rett syndrome over the past 16 years. And I won't go into all of these, but you can see that we have made a variety of comments and improvements in dealing with individuals with Rett syndrome.

I think that the key feature here is the development of qualitative rating scales, which have been used in the development of new drugs for therapies and were essential in the progress to prove that these drugs were effective.

The expectations of the parents need to be supported throughout. They may have multiple concerns, and one must be available to discuss these concerns, to find appropriate solutions or resolutions, and to make certain that the child is developing on a proper perspective from all points of view, but most importantly, from that of the parents.

Thank you again for your attention.

Announcer:
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