Understanding Rett Syndrome

The signs of the rare, life-altering Rett syndrome are often missed by doctors. For the mostly girls affected by Rett, intense therapy is required to manage the disorder.

By Amber Greviskes

Rett's symptoms can seem like those of autism, cerebral palsy, Parkinson's, epilepsy, and anxiety disorders—all at once, all in one little girl.

Zoe Rothschild of Stamford, Connecticut, was a happy child. She never crawled, but scooted around on her bottom. Then, her vocabulary stalled, and daycare teachers encouraged Rachel, her mother, to visit her pediatrician. But it wasn't until genetic tests were ordered (after a period of continuous therapy with physical, occupational, and speech therapists that did not positively impact her symptoms) that it became apparent that Zoe had Rett Syndrome.

Of the genetic testing experience, "there's a list of everything they're testing the child for, and you know there are certain ones—like Angelman's and Rett syndrome—that are considered the 'bad' ones," Rachel Rothschild recalls. "Our doctor said they'd start with the worst ones and work their way backward. Rett was the worse-case scenario."

Rett is a neurodevelopmental disease that begins in infancy or early childhood and is seen almost exclusively in girls. According to the National Institute of Neurological Disorders and Stroke, Rett affects about one in every 10,000 to 15,000 girls born each year. These children are usually born healthy and show normal development until they're six to 18-months old. At this point, new skills stop developing and a period of regression begins that includes losing communication skills, hand use and, at times, walking ability. The child's head stops growing at a normal rate.

Although each child experiences Rett differently, the child may wring her hands, have breathing problems, seize, or cry inconsolably. The condition can mirror autism, cerebral palsy, or non-specific developmental delay. Rett's wide-ranging symptoms can influence every aspect of the child's—and family's—life.

"For the parent, the most frustrating part of the condition is the fact that there was normal development initially," says Christine J. Milhaila, Ph.D., a neuropsychologist at The Brain Specialists in Nassau County. "Additionally, there is the occasion of misdiagnosis, which only adds to frustration."

Varying Symptoms

Rett, which is not inherited, can be diagnosed by the presence of an X-chromosome mutation, called an MECP2 mutation. The severity of the disease is determined by the location and type of mutation, which causes the variation in symptoms among children with Rett. The disease can be diagnosed through a blood test or a clinical diagnosis based on signs and symptoms.

Although there are four stages of Rett, children will spend most of their time in Stage III, the plateau stage, or Stage IV, when motor development is lost. Children who never develop motor skills move immediately from Stage II, the rapid destructive stage, to Stage IV. Based on the stage the child reaches, treatment plans are created that may include regular visits with more than a dozen doctors and specialists.

"Treatments geared at improving quality of life and maximizing levels of independence are key," Dr. Milhaila says. "Occupational, physical, feeding, and speech or language therapies are all integrated into a child's medical management of the disorder as well."

Research, Confusion, and Hope

The main reason that treatment focus is on improving the quality of life is because there is presently no cure for the disease. However, the identification of the gene mutation that causes Rett in 1999 has led to new research. Two years ago, Rett was reversed in animals. Scientists are now determining ways to reverse the mutation in humans.

After the initial diagnosis, Rothschild sought information from numerous sources, including websites, which she now regrets. Many only reinforced the family's fears, made the Rothschilds feel more alone, or confused them. It wasn't until Zoe Rothschild was referred to the Rett Syndrome Center at The Children's Hospital at Montefiore in the Bronx that her family began to receive answers from an 18-specialist team that focuses on children with Rett.

Aleksandra Djukic, M.D., the director of the Rett Syndrome Center, encourages her team to improve the patient's quality of life, increase her communication ability, and discover ways to assist in learning. Research has shown that the girls have a normal brain foundation. When they are given access to special communication tools, it's possible for them to express feelings, identify characters from books, or even make television or music clips play by following simple instructions.

"It's obvious that they understand you," Dr. Djukic says. "They just can't communicate."

Making Connections

Pioneers at Montefiore communicate in new ways with patients.

Adi Talwar

Dr. Aleksandra Djukic uses eye-gaze technology with five-year-old patient Maeve Curry at the Rett Syndrome Center at the Children's Hospital at Montefiore. Using the device, children can even get a song or video to begin playing just by staring at the screen.

One of the most devastating parts of Rett Syndrome is its ability to affect every aspect of the young patient's life.

Although Rett is a neurological disorder that affects each child differently, it can be best described as having symptoms of Parkinson's disease, cerebral palsy, autism and epilepsy in one. Until recently, doctors were unaware of how much children with Rett understood. However, it has become apparent that those with Rett have brains with a normal foundation. The neurons in their brain do not work as quickly as those in others' brains, but they are still intact.

For parents of those with Rett, this means that their daughters are capable of understanding much of the world around them. However, without voices or the ability to point at preferences, they have no way to communicate.

"These girls are imprisoned in their own bodies," says Aleksandra Djukic, M.D., director of the Rett Syndrome Center at The Children's Hospital at Montefiore in the Bronx. "They cannot communicate with the world in normal ways."

New eye-gaze technologies that are housed at Montefiore, however, are making communication possible. The eye-gaze system, which costs between $10,000 and $15,000, tracks eye movements using cameras and reflected infrared light when the patient is seated in front of the eye-gaze monitor, which is very similar to a computer screen.

Children may, for example, be asked to identify a specific cartoon character from among numerous others; the children who cannot gesture are able to point out the character by staring at it. Other times children may be able to activate music or play videos by staring at certain images.

For parents who have spent years trying to communicate with their daughters, finally knowing that their daughter understands language can be overwhelming.

In fact, when the Rett Syndrome Center opened, many researchers expected that the team's first focus would be on feeding, which is a physical need. However, after surveying parents of children with Rett, it became apparent that communication should be a main focus. With the ability to communicate through eye-gaze technologies, children can make choices (like what to have for lunch), express pain, or show that they are capable of learning.

"The eye-gaze test shows that there is a cognitive process going on," Dr. Djukic says. "To communicate through language is a very human need—it's what makes us human. The eye-gaze technology opens up the world of communication for these little girls."

For Zoe and Rachel Rothschild, being able to communicate would be a huge step in their relationship. Rachel realizes that Zoe has more complex thoughts than those that express basic physical needs; but, like many children with Rett, she can only make sounds.

"She knows she's loved, but she also knows she's being left out," Rachel Rothschild says. "I hope she continues to be happy and (relatively) healthy, and I hope for a cure. When you deal with the day-to-day, you have to believe that there's a cure. That's what keeps me going."

According to Rett Syndrome Research Trust, science tells us that we have every reason to be hopeful for finding an eventual cure to this debilitating brain disorder. Visit www.rsrt.org to learn more or support their research efforts.