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Silent Angels

Rett syndrome is a neurological disorder affecting mainly girls in the critical early developmental stages of life.

By Cindy S. Krivoshik, BSN, RN, CCRN

My daughter Laura was diagnosed with Rett syndrome when she turned 8 years old. Many people have never heard of Rett syndrome, or Silent Angels, which children with the disorder are sometimes called.

Rett syndrome is a rare neurological disorder on the autistic spectrum that occurs almost exclusively in girls. It causes severe developmental delays, seizures and cognitive and physical disabilities. Seizures occur more frequently when the disease stabilizes and when children with the disorder enter puberty.

Laura's seizures were controlled by anticonvulsant medication for 5 years, until this past May when she had five tonic-clonic (grand mal) seizures, numerous complex partial seizures and absence (petit mal) seizures. We called 911 when she became unresponsive with very shallow, impaired respirations. After 14 hours in the emergency department of Hunterdon Medical Center, Flemington, NJ, where she continued to have seizure episodes, Laura was transferred to the neurological unit at Children's Hospital of Philadelphia (CHOP), where she eventually recovered well enough to come home.

Sometimes, loving a child with special needs is like that.

Stages of Decline

According to the National Institute for Neurological Disorders and Stroke (NINDS), Rett syndrome has four stages of deterioration: early onset, rapid destructive, plateau or pseudo-stationary and late motor deterioration.

Most girls with Rett syndrome seem to be healthy and normal at birth. They smile, sit up and may begin to walk and talk. After a normal birth and development stage during the first 6-18 months of life, a short period of developmental stagnation is followed by rapid regression in language and motor skills. Within the next few years, they begin to lose their motor and communication skills and are no longer able to do what they once could.

Twenty-five percent of girls with Rett syndrome never walk, and about half of those with the disease lose the ability to do so at some point, according to NINDS. Purposeful hand use is lost, replaced with repetitive hand movements. They often have sensory integration dysfunction and may easily burn, cut or hurt themselves without feeling it. The vast majority of girls with Rett syndrome never learn to speak or, worse, lose their ability to speak as the disease progresses.

Laura is rather unique to have developed and maintained her speech. While she has a host of problems that affect her in many different ways, one of the biggest is apraxia, which prevents her body from doing what her brain tries to tell it to do even moving and talking. Try to imagine how hard it would be to know what you want to do and not to be able to do it. Or to be able to understand what people say, but not be able to tell them you understand them.

Screaming fits and inconsolable crying are common by 18-24 months of age. Additional characteristics of Rett syndrome cited by NINDS include autistic features, panic-like attacks, bruxism, episodic apnea and/or hyperpnea, gait ataxia and apraxia, tremors and acquired microcephaly. After this stage of rapid deterioration, the disease becomes relatively stable, but patients frequently develop dystonia and foot and hand deformities as they grow older.

A Matter of Genetics

Andreas Rett, an Austrian physician, first described this syndrome in an Austrian journal article in 1966. But it was not until after a second article was published in 1983, this time in English, that the disorder was generally recognized by the medical community. In 1999, Rett syndrome was found to be caused by a spontaneous mutation in a specific gene (MECP2) that regulates the expression of other genes. Laura has tested positive for this genetic mutation.

While Rett syndrome is a genetic disorder, almost all cases occur spontaneously, which means the mutation occurs randomly and it is not inherited. Rett syndrome affects only one in every 10,000-15,000 girls, according to NINDS. Some cases of Rett syndrome are more severe than others because girls have two copies of the X chromosome and need only one working copy for genetic information. Their bodies turn off the extra X chromosome in a process called "X inactivation." This process occurs randomly so each cell is left with one active X chromosome.

In part, the severity of Rett syndrome in girls is a function of the percentage of cells with a normal copy of the MECP2 gene after X inactivation takes place. Because boys have only one X chromosome, they have no protection from the harmful effects of the disorder, and most die before or shortly after birth.

Frustration & Coping

There is no cure for Rett syndrome. The treatment is only symptomatic and supportive. Despite many assorted difficulties, most girls with Rett syndrome continue to live well into middle age, but with a significantly higher incidence of sudden, unexplained death.

Laura was classified as "multiple handicapped" when she was in preschool, although we did not know why until many years later. She is registered with the Department of Developmental Disabilities (DDD) in New Jersey. We receive some monthly respite care funds from the state for her and have a handicapped permit for parking.

Fortunately, Laura attends a top-notch special education school, The Midland School, 6 hours a day, although she must travel almost an hour each way. She will go there until she is 21. She participates in an extended school year program. Laura also attends the ARC of Hunterdon County's Saturday drop-off respite care program twice a month, and on Monday evenings goes to the Heads-Up Special Riders program at Hasty Acres Farm in Kingston, NJ, where her favorite horse is named Squirt.

Laura's memory is phenomenal, but her intelligence cannot be accurately measured because of her apraxia and short attention span. She struggles every day with the very basic activities of daily living. She is not toilet trained and needs help bathing, dressing and eating. She will never be able to read or write. Apraxia and tremors impair even her simplest everyday tasks. She has little physical endurance and walks very slowly. She needs oral stimulation to concentrate so she constantly chews on her plastic toys, even her dolls' extremities, and has increased oral secretions.

Laura's World

When not in school, Laura's small world revolves around taking care of her baby dolls, most of whom are called Honey Bunch, clutching toys and magazines about babies that she can't read, and watching movies in her room. She tends to repeat questions over and over and can get quite fixated on a subject.

We worry about her future and wonder what will happen when we can no longer take care of her. This is a chronic concern for my husband and me. We are grateful for her doctors at CHOP and Hunterdon Pediatric Associates and are thankful for the ARC of Hunterdon County, The Midland School, Hasty Acres Farm and all of the people who care for Laura and other disabled people like our daughter, as they are the true angels on earth.

We raise money for research, belong to the International Rett Syndrome Association (with which we are trying to be more active), and pray for a cure. We try to educate others by talking with them and writing articles. We also created a Web site, www.UnicornMeadows.org, about Rett syndrome.

As parents, caring for special needs children can be heartbreaking and exhausting. Some days I feel I simply lack the strength to continue on for another minute. Then Laura comes home from school with a huge smile and asks me if I'm happy. She looks at me with her gorgeous brown eyes and says, "I love you," and I am inspired and rejuvenated, if only for just another day. My days are busy keeping Laura safe — and crying when no one is looking. Somehow, I have to try to keep up my spirits and not dwell on Laura's disabilities.

As I said, sometimes that's what it's like loving a child with special needs.

Resources

International Rett Syndrome Association: http://www.rettsyndrome.org

National Institute of Neurological Disorders and Stroke. (2006, January). Rett Syndrome Fact Sheet. Retrieved Dec. 4, 2006 from the World Wide Web: http://www.ninds.nih.gov/disorders/rett/detail_rett.htm#55513277

Unicorn Meadows: http://www.ignatz.net/unicorn.htm

Cindy S. Krivoshik works in the ICU at Hunterdon Medical Center, Flemington, NJ.