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From the Heart! Maggie's Experience with Rett Syndrome

by Kathleen McMillan

In honor of National Rett Syndrome Awareness Month this October, I would like to share my daughter Maggie's story. Maggie is one of the earliest girls to be diagnosed with Rett Syndrome, a genetically-based developmental disorder that is now known to affect one in every 10,000 to 15,000 girls worldwide.

Maggie's Story

In 1974, God blessed our family with a beautiful baby girl, Maggie M. McMillan. I remember it as if it were yesterday. Her deep blue newborn eyes staring at the pin stripes of my old brown robe. Her lips pursed tightly around my nipple, as she took to breast feeding immediately. I beamed with pride. Only four hours old and already she demonstrated she was a fast learner! She was a happy baby.

For the first twelve months every milestone was met. Then it happened: she started regressing for no apparent reason. The doctors had no answers, just a lot of possible labels including autism, aphasia, mental retardation, schizophrenia, and epilepsy, to name just a few. To put it mildly, she was a mystery to the medical profession. Finally, after almost ten years of searching, we discovered that Maggie had Rett Syndrome (RTT), a syndrome that had just been recognized in the United States.

Rett Syndrome

At that time, Maggie was one of only 250 documented cases of children with Rett Syndrome in the United States. Rett Syndrome mostly targets females and develops in four stages:

- First Stage: mostly normal development up to the age of six to 18 months
- Second Stage: rapid destructive phase that occurs between ages one and four; characterized by regression and loss of acquired skills
- Third Stage: typically a plateau or period of improvement that occurs between ages two and ten
- Fourth Stage: late motor deterioration stage, characterized by loss of the ability to move that may last for years

During the Second Stage, girls start grasping their hands in a stereotypical hand-washing motion. Speech, fine and gross motor skills and other skills slow down at a rapid rate and are soon lost. Girls may have gait abnormalities and may stop walking, stop talking, have autistic-like behaviors, or have problems with breathing and growth. This is a very frustrating time for both parent and child.

When Maggie was diagnosed, she was already in the Third Stage of Rett Syndrome. The rapid destructive period was over, but what followed were motor problems, scoliosis, and seizures. She would have up to 22 seizures a day, and she also became extremely violent. The doctor told us that we basically had to learn to live with her behavior and seizures. My husband and I were devastated. After great frustration, I took Maggie to the doctor and carried her into the office. The doctor took one look at her and said she was having a reaction to toxic levels of her seizure medication. She was immediately admitted to the hospital. Once put on new medication, Maggie was a different person.

Maggie's temperament had changed considerably by the time she was in her late teens. As she entered the Fourth Stage of Rett Syndrome, she still had some self-abusive behaviors and would strike out when upset, but most of the time she was easy going and a joy to be around. She was experiencing fewer seizures, and had gained back some of her lost skills. She still was totally dependent on a caregiver for her basic needs, and like a small child, she required a lot of care. Even though she ate a lot she still looked frail, except for her swollen stomach, a side effect from continuous air swallowing.

Living with Rett Syndrome

Over the years Maggie has had to endure more than any person should in a lifetime. She had to go through scoliosis surgery, gall bladder surgery, pneumonia, and had a gastric feeding tube placed. She has repeatedly broken her collarbones, lost teeth, and broken her hip due to seizures. Yet in spite of all the pain and discomfort, she handles it better than anyone I know.

So much has happened in the last 25 years with Rett Syndrome. It took until 1983 before it was even recognized in the United States as a syndrome. More recently, a genetic marker (MECP2) was found, and is diagnostic in the majority of girls, though as many as 15% may lack this marker or be diagnosed with Atypical Rett Syndrome. Research is now in the beginning stages to develop trial treatments. This is promising not only for the newly diagnosed, but also for the older girls as well. We hope a cure is just around the corner!

Maggie and I have a special bond and have become inseparable. I have always been her primary care provider. When at home she depends on me for many things, and I depend on her for unconditional love. I always tell her she is my guardian angel. She nurtures my heart! Some people may look at our experience with Rett Syndrome and feel sorry for us. I view it as a unique adventure. God has led me down a much different road in life. But because of this detour, I have grown to appreciate and love the most amazing and incredible young woman! Because of her courage and day-to-day bravery, she gives me strength!

For more information:

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

Kathleen is a mother of three daughters and has three grandchildren. She resides in Jacksonville, North Carolina with her husband of 39 years. She graduated from the University of North Carolina at Wilmington with a Masters Degree in Education. She is a retired Onslow County teacher and currently works in the Onslow County Public Library as a Technical Assistant in Youth Services. She has also coauthored chapters about Rett Syndrome in The Handbook of Neurodevelomental and Genetic Disorders in Adults *(2005) and* The Handbook of Neurodevelomental and Genetic Disorders in Children*, second edition (2011).*