What is Rett Syndrome?

Rett Syndrome is a neurological disorder seen primarily in females; although it also rarely occurs in males. It is usually caused by mutations of the MECP2 gene on the X chromosome.

Early developmental milestones appear normal, but between 6-18 months of age, there is a delay or regression in development, particularly affecting speech, hand skills and coordination. Intellectual development appears to be severely delayed.

A hallmark of Rett Syndrome is repetitive hand movements that may become almost constant while awake. Other features include seizures, irregular breathing, and curvature of the spine.

The most severe handicap in Rett Syndrome is Apraxia which means that the will to move is present, but the ability to carry through with movements is not.

Most girls and women need assistance for all activities of daily living: feeding, bathing, dressing, and toileting.

25% of girls and women may never walk at all, and about half of those who do walk will lose the ability at some time.

Other symptoms include gastro esophageal reflux disease (GERD)

Natalie Hong's Story

Our first daughter, Natalie, was born on February 1, 1999. She was born healthy and we were so happy to finally be a family.

Early on, she was so gentle and good. There were no foretelling signs that she was going to be any different from any other baby. This all changed right before her first birthday when she became very ill and was hospitalized. The doctor diagnosed her illness as a simple case of dehydration and said that she was going to be fine. Needless to say, she has not recovered and our lives have not been the same since then.

I can't help to think about how she was before this incident. The last memorable moment before her regression accelerated was in the hospital when she was about to get a shot from the nurse. I can vividly recall her

saying, "no, no, no" in such a pleading yet sweet manner. That was the last time she ever spoke to us in a coherent and meaningful way.

From that moment on, instead of making a full recovery from her dehydration, she regressed daily. Instead of reaching developmental milestones, she reached regression milestones. Instead of starting to walk, she stopped crawling. Instead of being able to pick up the smallest of sesame seeds, she lost purposeful use of her hands and repeatedly put her hand in her mouth. I could not believe what was happening to our little Natalie.

We desperately searched for answers. We went from one doctor to another doctor getting a different diagnosis each time. One doctor said she's just developmentally delayed and told us not to worry. Another doctor diagnosed her with Pervasive Developmental Disorder. Each diagnosis we got was worse than the prior one and the prognosis looked bleaker and bleaker.

After getting a diagnosis of Rett Syndrome around her second birthday and researching into what it is, I was in shock. Actually, from that moment up until her third birthday, I was in denial and thought the doctors had it wrong or she would snap out of it and go on to live a normal childhood. I have now accepted her condition but at the same time hope and pray for a cure. But in the meantime, we look to providing her with treatments and therapies that will improve her quality of life.

The Future

Although Natalie's illness has made our life difficult to say the least, she is still our daughter and we will treat her as such. So as we hope and pray for a cure, our family will attempt to live as normal as a life as possible partaking in everyday family activities.