

The Monmouth Journal

The Monmouth Journal, LLC • 46 English Plaza • Suite D • Red Bank, NJ • 07701 • 732-747-7007 • Fax 732-747-5445

April 10, 2009

www.themonmouthjournal.com

I Know You're in There

By Diane Carlton
The Monmouth Journal

RUMSON — Sarah Harding is an 8-year-old girl who is trapped — and her mother, Ingrid Harding, has vowed to set her free.

Sarah cannot walk, speak and has no use of her hands due to the debilitating disease she is afflicted with. But, her mother says Sarah comprehends everything in the world around her and believes that one day soon, a cure will be found to unleash her daughter from the prison her body has created.

Rett syndrome is a unique developmental disorder that is first recognized in infancy and seen almost always in girls, but can, rarely, be seen in boys.

To Sarah and the approximately 500,000 girls living with Rett Syndrome, Harding says, "Hold on. We're coming to get you."

The 40-year-old mother of three can make such a bold statement because, since 2006, neuroscientists have made groundbreaking strides naming and locating the female gene which causes the sporadic onset of Rhett Syndrome — the most severe form of autism. But through two foundations that Harding has founded and co-founded, she maintains a strong optimism that Sarah will be singing and dancing at her Sweet 16 party.



Rett Syndrome is a debilitating neurological disorder that is classified as a pervasive developmental disorder. It is a genetic disorder — resulting from a faulty gene, or genes — of which less than 1 percent of recorded cases are inherited or passed from one generation to the next. In most cases the mutation occurs randomly and is not inherited. "Any little girl anywhere in the world has equal chance of getting it," said Harding.

Sarah was born in January 2001 while the family was living in Biloxi, Mississippi where Harding's husband, Peter, 41, was studying to become a physician in the Air Force. After enduring what she describes as a difficult pregnancy which required bed rest, Harding said, "We always say Sarah fought to be here — she won and she's still fighting."

Her mother details Sarah's first six months as witnessing nothing noticeable. Her daughter achieved every milestone and natural development for a baby her age.

Sarah's brother, Pierce, who at the time was 3 years old, was the first to notice something was not right, according to Harding.

"He said, 'Mommy, why won't she look at me?'" she recalled.

The family began to notice that Sarah's toes were continually curled and that her thumbs remained tucked in the palm of her hand. When family members and outsiders began to ask questions, the family called Early Intervention — a



state-run agency whose mission is to enhance the capacity of families to meet the developmental and health-related needs of children birth to age three. They specialize in children who have developmental delays or disabilities by providing services and support to families and their children.

Believing the family was dealing with a slowly developing child, the Hardings noticed that Sarah began to regress rapidly and within a month had completely lost every skill and milestone she had reached.

Harding lists the shocking discoveries.

"She couldn't hold her head up, couldn't focus, couldn't sit up, couldn't chew ... we thought she was dying." As their concern for Sarah turned to panic, Harding remembers thinking, "My little girl is dying. We have to find out what's wrong ... we have to save her."

Because the vast majority of the public and medical communities do not know about Rett Syndrome, Harding offers a sobering statistic: "Every 90 minutes another little girl is diagnosed and another family is going through this." It took three grueling years for Sarah to, eventually, receive a diagnosis of Rett Syndrome at the age of 4. Enduring a CAT scan, MRI, spinal tap, geneticists "poring over every inch of her body" and copious amounts of blood work, "looking for things I had never heard of," Harding is now the owner of volumes of lab tests that were performed on Sarah, all having been returned "normal."

Alarmingly, they received one misdiagnosis during the ordeal when Harding was called on her cell phone by a geneticist and told that Sarah's spinal tap showed her brain was leaking into her spinal cord and that the young girl did not have long to live.

Reflecting on the bad memory, Harding recalls, "It's good to have a doctor for a husband sometimes." Sarah was quickly retested and the diagnosis was found to be wrong.

In a relay of ups and downs, the Hardings were told by some doctors to stop looking for what the medical diagnosis was and were told to be at peace with who their daughter was.

Determined to find an answer, the Hardings traveled to Children's Hospital of Philadelphia, where, in a waiting room, Harding saw a poster of actress Julia Roberts advocating for Rett Syndrome awareness.

Tested for the neurological disorder, it too came back negative.

At home, Harding entered countless symptoms and keywords into her Internet browser search box. The common answer kept coming back Rett Syndrome.

Meeting two other girls in medical offices with Rett Syndrome, she noticed both were just like Sarah. Harding also watched videos of girls with Rett Syndrome on the Web site YouTube, and all acted just like Sarah, as well. Stating that mother's intuition goes a long way, Harding said many in the medical community saw her as a desperate mother who was looking to hang her hat on something. Of her uncanny instinct she laughs, saying, "Nobody was on that train with me."

Reaching out to the Scientific Director of the Rett Syndrome Research Foundation in Ohio, Harding sent video, pictures and lab test results figuring there might be other undiagnosed girls like Sarah in the world. Recommended for a study of 10 similar girls taking place in Israel, the Hardings sent vials of Sarah's blood, along with their documentation, and received a confirmed diagnosis that their daughter had Rett Syndrome.

Though Sarah had been tested before, Harding said, "They didn't tell us the test was not 100 percent accurate." Harding describes their experiences with Rett as an emotional roller coaster.

Though she said, in such a situation, you come to a place of resolve, Harding said she can still shed tears when it comes to Sarah.

"Not about me or what it has done to my family," she said. "I cry for Sarah — imagine not being able to speak ... she hurts and can't tell me about it."

In the years since Sarah's diagnosis, Harding has found a 23-year-old woman with Rett Syndrome on the Internet who has limited gross motor function and cannot speak, but uses a special computer to type with a "facilitator" who holds her hand for her so that her index finger stays straight. She uses that finger to type on a special keyboard.

The woman writes in her blog, "Since it took 12 years of living with Rett before I was diagnosed, we saw so many different doctors. They each had an opinion about me that they shared with my mom in front of me as though I couldn't hear. Much of what they shared was something they never would have if they believed I was capable of understanding them."

The young woman, identified only as Karly, tells of how her mother and sister read to her and how she basically taught herself to read by looking at the letters and words in books while being read to as a child.

The first three years of Rett Syndrome are excruciating for the child, according to Harding. She recounts an episode where Sarah once went 52 hours without sleeping and screamed all night. With Peter going through his residency, she said, "There were nights all three of us were standing there crying at 4 a.m."

Sarah suffered from intense gastrointestinal problems, a common complication of Rett. At age 3- and one-half, Harding removed wheat and dairy from Sarah's diet, a frequent course of treatment pursued by parents in the Autism community.

Within two weeks, Sarah showed remarkable improvement.

Though the proteins in wheat and dairy are difficult for most people to digest, said Harding, in Autistic children — who often have gastrointestinal difficulties — the buildup of proteins can cause serious complications. “Casein, the dairy protein, builds up in the body and is as potent as cocaine to the central nervous system,” said Harding. “Gluten messes up the gut lining and allows yeast overgrowth that puts huge holes in the stomach lining and messes up the food processing and causes pain.” Describing the miraculous ability for Sarah to now make eye contact, put weight on her feet, and roll towards a desired object on the floor, Harding said, “I felt at that point we had stopped regression and we could progress. Her body wanted to move.”

In 2007, after Peter spent five months serving in Afghanistan, the Hardings moved back to the home in Rumson where Ingrid was raised. Living in a three-generational environment with Ingrid’s parents, Douglas and Peggy Love, Sarah is surrounded by her immediate and extended family.

Peter practices Internal Medicine as a hospitalist close to home at Riverview Medical Center in Red Bank while Sarah spends time in a pink-colored playroom with sister Gretchen, 6, and brother, Pierce, 11.

Grandmother Peggy Love said, “The house has come alive!”

Last year Sarah entered the first grade at Deane Porter School, here, where she has progressed to the second grade in a typical classroom setting with a one-to-one aid. On fair weather days, the family bicycles to school with Sarah in tow in a cart pulled by a bike.

But 2007 proved to be a banner year for advancements in Rett Syndrome, as well as for the Harding family. Harding said Dr. Adrian Bird, a molecular geneticist at the University of Edinburgh, Scotland who in the 1980s discovered the MeCP2 gene that, when mutated, causes Rett, performed tests on mice to see if there is a window of opportunity for a cure. The tests concluded that Rett could be reversed at any age without permanent damage to the mice.

“This sent shockwaves through the neuroscience community,” said Harding. “It is the first time any neurological disorder has been proven reversible.”

Knowing this fact, Harding said there is renewed interest in the research field and that the progress is getting closer to a cure. “There’s a half million girls who can be saved,” she said.

Through two separate foundations, Harding has been advocating awareness and raising funds towards the massive research needed to zero in on the cure.

In 2006, she founded GirlPower2Cure which is a grassroots effort targeted towards girls ages eight to 15 who do not have Rett.

“These girls have the possibility to not only witness the cure, but they can be a part of making it happen,” she said. With a self-designed, brightly colored flower as its logo, Harding hopes the pink and orange daisy can do for Rett what the pink ribbon achieved for breast cancer awareness and research. Designing one of the flower petals as slightly pointed she said, “Just because something is a little off, doesn’t mean it’s not still beautiful.”

GirlPower2Cure has sponsored Dress Up for a Cure on April 1 for the last three years and this year the event consisted of 23 teams across the country including local Rumson schools. For a \$1 donation — or more — students were able to raise close to \$100,000 to go toward research for a cure by dressing up in “silly” outfits or participating in other creative ways to dress up. Hoping to sweep the nation, and one day promote the event on the Oprah Winfrey Show, Harding said, “April Fool’s Day should be synonymous with Dress Up for a Cure.”

The funds raised from GirlPower2Cure are designated for the Rett Syndrome Research Trust (RSRT) that Harding co-founded with Monica Coenraads, Scientific Director of the Rett Syndrome Research Foundation. In 2008 RSRT committed \$2.4 million in its first year of operation towards funding a cure.

Describing RSRT as a streamlined organization where all involved are on the same path, focused and working toward the same goal, Harding said, "We have the top neuroscientists in the field on our scientific advisory board and we're setting the roadmap to the cure."

Harding said neuroscientists are calling Rett Syndrome the "Rosetta Stone" — or scientific key — of research. Its cure will open the door to all neurological disorders like Parkinson's disease, Huntington's disease, Alzheimer's disease, Schizophrenia and Autism, she believes.

"It's an important piece of the puzzle our girls hold," she said.

In telling Sarah's story and expressing optimism for its happy ending, Harding displays remarkable stoicism mixed with a natural sunny disposition. She is the first one, however, to let her guard down and express concerns that no mother should have to face. She counts irregular breathing, the possibility of uncontrollable seizures, and complications like pneumonia and the potential for sudden death while sleeping as some of the things that keep her up at night.

She said, "Their hearts simply stop beating. I pray Sarah will make it through to see her cure, but I give her a huge kiss and hug every night just in case it is my last chance to hold her."

For more information on Rett Syndrome or to help fund the cure, visit www.rsrt.org or visit www.girlpower2cure.org to find out about Sarah and how your daughter can help spread awareness. To read more of Karly's blog visit www.spiritdances.wordpress.com and click the "My Rett Body" tab.