

# RETT SYNDROME



**Britt Van Giesen shares a tender moment with daughter, Erika.**

families face life-changing challenges

by Tinka Davi

**M**ost people have never heard of Rett Syndrome. Have you?" That's the message Britt Van Giesen wears on his T-shirt and a question he'll readily answer when people stop him in the supermarket or elsewhere. Not many people or physicians know about Rett Syndrome, but Van Giesen is an expert at explaining it.

Van Giesen, 48, and his wife, Sharon, are the parents of Erika, age 6, who was diag-

nosed with Rett Syndrome on April 15, 2004. It changed their lives forever.

Rett Syndrome is a debilitating and devastating neurological disorder diagnosed almost exclusively in females. It occurs in one of every 10,000 female births in the US. Children with Rett Syndrome appear to develop normally until 6 to 18 months of age when they enter a period of regression, losing speech and motor skills. Most girls develop repetitive hand movements, irregular breathing patterns, seizures

and extreme motor control problems. Rett leaves its victims profoundly disabled, requiring maximum assistance with every aspect of daily living. There is no known cure.

Erika was born on Sept. 18, 2002, a normal birth, with no complications. "She was a perfect baby and never cried," says her dad. However, Van Giesen, a doctor's son, noticed that Erika missed some six- and 12-month "baby milestones." Instead of crawling, she bunny-hopped on her rear and began to have

## RETT SYNDROME



Fun in the tub – Erika, 6, and sister, Brianna, 9.

stereotypical hand movements and delayed developments of Rett.

Doctors urged the Van Giesens not to worry until Erika was 18 months old, because the local medical community had not seen any cases of Rett. Van Giesen started digging online and discovered Erika had 11 Rett Syndrome symptoms. He contacted Rett medical professionals and other parents. Erika underwent an MRI and CAT scan. A blood sample was sent to Greenwood Genetic Center in North Carolina, one of a half dozen Rett testing centers in the US. When a phone call relaying the results of tests confirmed that Erika had Rett, Van Giesen sat at his desk at work and cried.

Telling Sharon was devastatingly difficult as was explaining Rett Syndrome to their family and friends. He put together packets of information and a video and invited people to their home along with their pediatrician to answer

questions.

In the next year, Erika's medical condition worsened. She developed gastrointestinal problems and had trouble swallowing and sleeping.

Erika has a dazzling smile, thick curly hair and blue eyes that sparkle behind her tiny bifocals. Like other Rett girls, she is beautiful with perfect features. Notable is her constant hand wringing, her lack of verbal communication, her inability to walk. She rides around in a special wheelchair, has dramatic seizures and sometimes stays awake three nights in a row. The Van Giesens grind her food, feed her by spoon and use a syringe for liquids and six different medications. She watches the same Wiggles and Elmo videos repeatedly and has occupational therapy several times a week. Before and after school, she goes to the home of a nanny while the Van Giesens are at work.

“Erika can't do anything for herself and she has no ability to communicate,” says Van Giesen. “Changing a DVD or offering juice is just a guess, but we are working on her communication skills and hope to get a My Tobii device to help her.” It costs \$14,000. Erika's equipment is expensive: \$8,000 for a positioning chair so scoliosis won't set in, \$5,000 for a stander; \$1,200 for braces (ankle foot orthoses - AFOs) for her legs; multiple sets of bifocals at \$100 a pair; \$900 for a car seat; \$6,000 for her wheelchair.

Their home is completely handicapped accessible. A local builder, Shook and Waller Construction, took on the project after hearing about Erika through a mutual friend at their church. “They did not just build a house; any builder can do that. Their care and understanding built us a home,” says Van Giesen. The home has a handicapped-ready backyard, ramps throughout, solid-core doors and



rigid-board insulation to maintain the temperature at 78 degrees, which Erika needs. It's close to their former home so that the Van Giesen's older girls, Megan, 15, and Brianna, 9, did not have to change schools or move away from neighborhood friends.

Erika is a "poster child" for the International Rett Syndrome Foundation and a tender photo of her and Britt are on the cover of "The Rett Syndrome Handbook," second edition ([rettsyndrome.org/index](http://rettsyndrome.org/index)).

In 2005, Van Giesen, director of hospitality for Clos Pegase Winery, and his wife Sharon, and Katie Hamilton of Napa launched a fundraiser, "Erika's Dream," which last year brought in \$90,000 to benefit Katie's Clinic for Rett Syndrome at Children's Hospital in Oakland. The next event – an auction and dinner – is set for Feb. 6, 2010 at Artesa Winery in Napa.

Katie's Clinic ([childrenshospitaloakland.org/depts](http://childrenshospitaloakland.org/depts)) serves the Western states and Hawaii, and, in March, assessed its first international Rett family from the Philippines.

The clinic provides initial evaluation and ongoing coordination of care for Rett patients. Each evaluation is a multidisciplinary assessment, explains Pat George, program coordinator for Katie's Clinic, a former medical office manager and grandmother of Katie Nues for whom the clinic is named. (George took on the position about four months ago.)

Patients see a neurologist, communications specialist, behavioral pediatrician, occupational therapist, physical therapist, nutritionist, gastrointestinal specialist and orthopedic specialist. "Katie's Clinic is the only one (of six in the U.S.) where patients and parents see all specialists in one visit," says George.

"We are a gold standard for Rett clinics."

Patients range from babies to adults. "Girls can live well into adulthood," says George. The most frequent causes of death are complications from pneumonia or seizures, or a SIDS-like passing in sleep.

"Rett is random and unexpected and life changing for the whole family, relatives and friends," she adds. "(The parents) will have the child with them forever. The girls won't go to proms and will never marry. It's heart breaking, but there's a special thing about them. They are so loving and understanding."

Some Rett girls are able to communicate via "Eye Gaze," a camera with lasers affixed to the top of a computer and trained on the girl's eyes. When asked what she would like for lunch from among four foods on the computer screen, the camera captures what she stares at the longest. "The girls want to communicate so badly; but the effort can be so subtle that sometimes just raising an eyebrow means 'yes,'" explains George.

The clinic offers consultations to schools. "The girls can do academic work. They can learn and do have memory, but they can't write or stack blocks," says Paige Nues, 39, founding parent of Katie's Clinic. The clinic is named for Paige and Jesse Nues' daughter, now 6½, and older sister to Melissa, 2½, and Abigail, 14 months.

"When Katie got her diagnosis, it turned us into a world of disability and children's health issues," says Nues. She and her husband flew to Baylor College of Medicine in Houston, Texas, to visit the Rett Syndrome Clinic there. As they talked to other Rett families, they saw a need for a local clinic. Children's Hospital

of Oakland was open to the idea and Katie's Clinic was founded with funds from Erika's Dream and other donors.

Katie has thick brown hair and expressive brown eyes. "She can't walk, can't sit up unassisted and has no functional use of her hands. Speech is absent, but she communicates with her eyes," says Nues. "Rett girls are incredibly social, and Katie's a really happy kid. If she's upset, I know there's a physical need but she can't tell me if it's a stomach ache, headache, leg cramp or rock in her shoe." Katie has frequent seizures, difficulty chewing and swallowing and takes liquids through a G-(gastrostomy) tube.

Rett's most severe disability is apraxia, described as having the will to move, but the body can't coordinate the physical motion. Girls frequently are diagnosed as having autism or cerebral palsy. "During their regression period they are withdrawn, but after regression, they become very social again. That's very different from autism," says Nues.

Katie attends first grade, mainstreams language arts, science, music, recess and gardening and has one-on-one communication and fine motor skill sessions in her special education classroom. She reads in a group via a small computer box with the words programmed by her aide. It's one of her happiest times in the school day, says her mom.

Word about Rett Syndrome is getting out to the public thanks to Donald Trump's "Celebrity Apprentice" on NBC. One of the celebrities, country singer Clint Black, has named the International Rett Syndrome Foundation as his charity.

That's encouraging to the Van Giesens

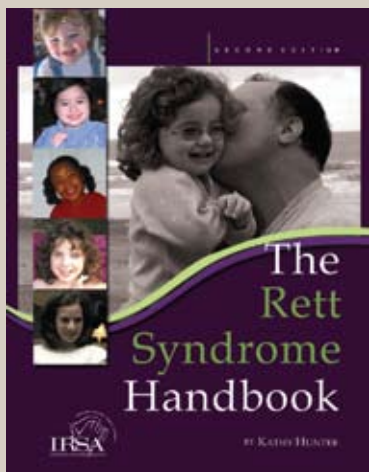
## RETT SYNDROME

who want more people to be aware of Rett Syndrome. They willingly talk about Rett, but admit that it's a tough life. "Some days we win the battle and some days we don't, and we spend a lot of time behind closed doors in tears. But we love Erika so much. She will always need us and always have a special place in our life."

**For more information, Rett syndrome, visit the International Rett Syndrome Foundation (IRSF) at [rettsyndrome.org/index](http://rettsyndrome.org/index); [childrenshospitaloakland.org/healthcare/depts./rett](http://childrenshospitaloakland.org/healthcare/depts./rett); (Katie's Clinic); and [erikavangiesenfoundation.org](http://erikavangiesenfoundation.org).** •



**Below: Erika and Britt Van Giesen are featured on the cover of a guide written for everyone affected by Rett Syndrome.**



## FACTS ABOUT RETT SYNDROME

- Rett syndrome is the leading genetic cause of impairment in girls
- Rett syndrome is caused by mutations in the gene MECP2. 95 percent of girls diagnosed with Rett syndrome in the United States have this mutation
- Rett syndrome is the only autism spectrum disorder with a known genetic cause
- Girls and women with Rett syndrome have unusually bright eyes and seem to understand more than they can express
- A girl is born every five hours with Rett syndrome
- A recent, amazing breakthrough in science is the ability to reverse Rett symptoms in an animal model
- Thousands of girls and women worldwide are undiagnosed or misdiagnosed with cerebral palsy or autism
- Rett syndrome affects all ethnic, racial and socio-economic groups; every parent is at risk for having a child with Rett syndrome
- Rett syndrome often is characterized as a "Rosetta Stone" disorder that can give insight into a host of other late onset neurological disorders such as autism

### Medical Facts

- Rett syndrome is one of four diseases that primarily affect females
- The American medical community did not recognize Rett syndrome until 1983
- 20 percent of all girls and women may never walk at all and about one-fourth of those who do walk will lose the ability
- 80 percent of those affected by Rett syndrome will experience at least one seizure in their life
- Approximately 90 percent are at risk for some degree of curvature of the spine
- The most severe handicap in Rett syndrome is apraxia, which means the will to move is present but there is an inability to carry through with movement
- Most girls and women also live with nearly constant repetitive hand movements while awake, irregular breathing and sleep patterns as well as gastrointestinal issues

— **Paige Nues**,  
Director, Family Support,  
International Rett Syndrome  
Foundation  
1.800.818.RETT (7388),  
[rettsyndrome.org](http://rettsyndrome.org)