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Rare illness strikes one family three times

■ A couple's life turns upside down as each of their children are diagnosed with ataxia telangiectasia, a disease that usually kills by age 20.

By KIM GILMORE
Times Staff Writer

SPRING HILL — They were beautiful babies, creations of a man and woman who carefully planned parenthood after embarking on successful careers.

In seven years, Amy and David Madison brought Braun, Jamie and Andy into their world. Life was good.

Then it turned upside down as the children, one by one, were determined as having a disease so rare that most people have never even heard of it.

"We are living a nightmare," said Mrs. Madison at their home in River Country Estates in western Hernando County. "When I start to think how I will possibly never see my children get married or have grandchildren, it becomes more than I can bear."

Tune in

ABC's newsmagazine *Turning Point*, hosted by Barbara Walters, is scheduled to air a special on ataxia telangiectasia at 9 p.m. Monday. Afterward, local affiliate WFTS-Ch. 28 plans to broadcast a feature story about the Madison family on its 11 p.m. newscast.

Braun, now 15, Jamie, 12, and Andy, 8, have ataxia telangiectasia. Most children who contract the disease are in wheelchairs by age 10 and are dead by age 20.

"If you can imagine a disease that combines muscular dystrophy, cystic fibrosis and cancer all at once, that's what A-T is," said Brad Margus, president of the A-T Children's Project in Boca Raton, a non-profit foundation that promotes research of the genetic disorder.

Margus and his wife, Vicki, have two children with ataxia telangiectasia, a 7-year-old son, Jarrett, who first showed signs of the disease when he was 2, and Quinn, 5.

In August 1994, Dr. Stella Legarda, a pediatric neurologist and assistant professor at the University of Florida in Gainesville, recognized the broken blood

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vessels in the Madison children's eyes and on the tops of their ears as signs of A-T.

It was an amazing find.

"This is a disease so rare that many physicians will probably never see a case in their entire career," said Patrick Concannon, a member of the Virginia Mason Research Center and associate professor at the University of Washington School of Medicine in Seattle.

There are only about 500 diagnosed cases of A-T in the country. A-T attacks the immune system and destroys muscle control in the eyes, mouth, arms and legs.

A-T children usually develop respiratory problems, such as pneumonia and bronchitis. They are prone to lymphoma, leukemia and other cancers.

Braun first showed symptoms of A-T when he was 18 months old. "He took running steps instead of walking steps," Mrs. Madison said.

At 5, Jamie began to lose her balance and confidence in gymnastics. At 14 months, Andy also showed signs. "He was drooling. He was falling. He was wobbling. It was the same thing all over again," Mrs. Madison said.

The family drove hundreds of miles and saw dozens of specialists. Each poked, prodded and experimented on the children. Each gave a different diagnosis.

It took 11 years and more than 20 doctors to find the answer.

"It was a flood of relief we experienced. Fear comes from the unknown," Mrs. Madison said, her eyes tearing up. "It's kind of hard knowing a train wreck is coming, but it's kind of good, too, because you can prepare for it."

With three death sentences looming, the Madisons are on a desperate search for a cure.

Mrs. Madison, 40, has a master's degree from Oklahoma University and a background in computer software. Since her children were diagnosed, she has become an A-T evangelist. She travels the world speaking to other afflicted families, working with doctors and promoting the research of the A-T Children's Project in Boca Raton.

Mr. Madison, 41, earned a bachelor's degree at Oklahoma State University and was promoted three years ago by Wal-Mart in San Antonio, Texas, to be operations manager at the Wal-Mart Distribution Center in eastern Hernando County. Mrs. Madison calls him her Rock of Gibraltar, motivator and provider.

"David and I make a great team," she said. Their own health always has been good. Mrs. Madison was one of five children growing up with no indications of trouble. Mr. Madison was orphaned at birth but checked three generations into his family history and found no alarming health disorders.

The Madisons struggle to maintain a normal life. The children ride the school bus and attend public schools. They do household chores and earn an allowance.

At night the family gathers at the dinner table to share their frustrations. Like the time a schoolteacher told Mrs. Madison her daughter "simply had no bells in the tower."

"I started to stand up and strangle the woman," Mrs. Madison said. Instead she went to the bathroom and cried.

"Some people don't understand so they start making fun of me and stuff," said Jamie, a sixth-grade honor roll pupil at West Hernando Middle School. "They ask, 'What's wrong, are you tired or something?' "

Braun, a ninth-grader at Central High School, struggles to keep up with his classmates while dodging insults.

"Because of some of the abuse they've had to deal with, they've developed thick skin," Mrs. Madison said.

Unique to the Madisons is how they measure success. Many parents take for granted the natural ability of their children, such as catching a ball.

"When (Braun) does it, you ought to see his face," Mr. Madison said.

Andy, who is least affected by the disease so far, is learning to ride a bike. Jamie recently sang in an all-county choral ensemble.

"I was proud of her for just trying," Mr. Madison said, beaming. "We take everything one day at a time. We really appreciate what we've got . . . whereas other people might let that slip by or take that for granted, we treasure the moments."

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For the Madisons, 1995 was a momentous year.

In December, Braun received a $4,000 multimedia computer system from the Make A Wish Foundation in Tampa, a non-profit organization dedicated to fulfilling the wishes of children with life-threatening diseases.

The system is equipped with Dragondictate, a speech-recognition software package that allows Braun to dictate into the computer instead of typing.

As the A-T accelerates, so will the symptoms that rob Braun of his ability to read, write and speak clearly. The computer may eventually be his only means of communication.

"I'm going to dictate so I need silence," Braun announced, politely. "You . . . have . . . to . . . pause . . . between . . . each . . . word . . . you . . . say . . . so . . . the . . . computer . . . can . . . pick . . . it . . . up."

Volunteer Helen McQuade helped coordinate Braun's gift through Make A Wish. "The thing is, you get to make them smile, and that's what makes a difference," she said.

But the Madisons were smiling even before the arrival of the computer.

In June, after a decade of research, scientists in Israel discovered the defective gene that causes A-T, called ATM, for ataxia telangiectasia mutated.

Researchers now have the means to seek a treatment or cure. The gene also may provide vital information about cancer and the immune system.

"We now have a target for studying the biology of the gene that we didn't have before. We can now go in and ask, what's different about this gene," said Concannon of the Virginia Mason Research Center.

Concannon said researchers may now be able to determine which section of the ATM gene is damaged, how it is damaged and ultimately repair it.

It will not be easy.

So far, scientists have found 150 mutations of the ATM gene, which rules out, at least temporarily, a single solution.

"This is the same problem with the breast cancer genes and the colon cancer genes," said Dr. Richard Gatti of the UCLA School of Medicine in Los Angeles.

Gatti, who helped pioneer A-T research, said discovery of the ATM gene advanced research and opened the gates for future experiments. Already, scientists are trying to model the disease in mice to test drugs.

"It's already speeding the process of A-T research about tenfold. We're learning more in each month than we've learned in a year in the past," he said.

Gatti is optimistic that doctors soon may be able to diagnose the disease in children and determine whether someone is a carrier of the ATM gene.

"Getting the gene, of course, is the beginning of the end," Gatti said.

The question remains, will it happen in time for Braun, Jamie and Andy?

"We're going to find a cure for this, and we're going to get an answer," Mrs. Madison said. "If all this doesn't happen, I just want to live my life so I have no regrets, so if they are gone I'm not wishing I had done something that I had not done. I don't have any regrets today, and if I keep living my life this way I won't have any."

Times researcher Carolyn Hardnett contributed to this report.