

**Rye Patch (<http://rye.patch.com>)** By [Satta Sarmah](#) | August 30, 2010

## Rye Brook Teen One of 350 People in the World with Rare Genetic Disease

**Rebecca Newman suffers from familial dysautonomia, a rare Jewish genetic disease. Her family will participate in the 7th annual Tour de Foliage bike race to raise money for research to fight the disease.**



Rebecca Newman, a student at Blind Brook Middle School, is, as she says, "13 and a half," an age that is accompanied by teenage angst involving school, friends and boys.

Rebecca is an ace card player (Poker, Black Jack 21 and Gin Rummy are her specialties) and she loves Nick Jonas—a member of the music group, the Jonas Brothers—and a male heartthrob and icon among the Disney-set.

She is, as her dad Jeff recently said, a "glass half full kind of kid."

Among all the similarities to children her age, Rebecca also has a distinction that no parent could ever wish for—she is one of 350 people in the world living with familial dysautonomia, a rare genetic disorder affecting the sensory and nervous systems attributed to a recessive gene only carried by those of Jewish heritage.

The disease, sometimes called FD or Riley-Day syndrome, affects normal bodily functions such as chewing, swallowing, blood pressure control, digestion and respiration. Only 640 people have ever been diagnosed with it, and about one-third of them live in the New York area, while another third live in Israel. As recently as 40 or 50 years ago, the mortality rate was high and the life expectancy of patients was limited, with most familial dysautonomia sufferers only living until age 5.

To raise awareness about the disease and money for research, the Newman family will co-chair the 7<sup>th</sup> annual Tour de Foliage, a bicycle tour that will benefit the Dysautonomia Foundation, a New York-based organization founded in 1951, not too long after FD was first identified in the United States.

More than 100 people are scheduled to participate in the event, which will take place on September 12 at 8 a.m. at Pace University's Pleasantville campus, including many of the Newmans' Rye Brook neighbors and friends.

"Jeff and I did not raise Rebecca by ourselves," Lisa Newman, Rebecca's mother, said. "A village of people raised this child. What our friends have done to help us—they participate in this bike tour; they ride in this bike tour, they donate money every year to this bike tour. Rebecca's life has touched so many people."

### **Searching for an Answer**

Rebecca was diagnosed with FD when she was a year old, after a maze of doctors' visits and misdiagnoses.

At only 13, Rebecca has the distinction of being the 513<sup>th</sup> person in the world diagnosed with the disease.

"How did we end up number 513 in the world of anything?" Lisa said she and her husband often ask themselves.

When Rebecca was born her mother had difficulty breastfeeding and she spent five days in the neo-natal intensive care unit. She didn't easily gain weight, as most babies do, and at 10 weeks old, she had her first respiratory infection. By the time she was a year old, Rebecca had almost 12 bouts with pneumonia.

Jeff and Lisa took their daughter to various hospitals around New York, and some of the doctors only diagnosed Rebecca with asthma or acid reflux. The Newmans didn't discover what was wrong with Rebecca until Jeff's mother told them that it could be familial dysautonomia, after she learned of the disease while talking about Rebecca with a man at a bar mitzvah. The man told her that his cousin suffered from the disorder and described symptoms similar to what Rebecca was experiencing. After talking to Jeff's mother, the Newmans took Rebecca to NYU Medical Center, where Dr. Felicia Axelrod, an expert in the field, diagnosed her with the disease.

Though FD has rendered Rebecca unable to perform even the most normal and basic of bodily functions, her case is considered mild.

Like many others afflicted with the disease, Rebecca can't cry tears and has to use eye drops every day. She also has to take multiple medications and gets 50 ounces of liquid every day from a feeding tube, since FD causes a swallowing dysfunction that makes it difficult for Rebecca to consume thin liquids.

"It's literally a safety valve for her body," Lisa said of the feeding tube.

Since Rebecca relies on a feeding tube, a nurse must accompany her to school every day.

The nurse, Joan, who is paid for by the school district, will likely be with her until she graduates high school. However, she is a welcomed presence in the 13-year old's world.

"She lets me be independent," Rebecca said.

In between school and homework, Rebecca's time is filled with singing lessons to help her breathing and regular doctors' visits to New York University Medical Center. NYU has been a leader in studying FD, establishing the Dysautonomia Center in 1969, the country's only medical facility solely devoted to research and treatment of the disease.

For Rebecca, the one-hour car rides to the hospital are sometimes tiring, but she said she never really gets weary of going to the doctor, though she admits that "sometimes it might get a little annoying."

### **FD Takes a Toll on Families, Rallies the Community**

Familial Dysautonomia is a disease that affects not only those it afflicts, but also the people who love them.

Rebecca's parents, Lisa, 47, and Jeff, 50, have been married for 19 years. They also have an older daughter, Julia, who is 15 and a sophomore at Blind Brook High School.

The disease sometimes causes a difficult balancing act for the family. Jeff said one of he and his wife's concerns was making sure Julia did not feel slighted because Rebecca's medical needs often take priority. However, the girls have maintained a good bond, he said.

"What I can say is that Julia is remarkable in that she doesn't resent her sister. She takes everything pretty much in stride," Jeff said. "But I'm not sure what she shares and doesn't share outside of here."

FD has also reshaped Lisa's life. Lisa, who previously worked in advertising, is a board member of the Dysautonomia Foundation and earned a master's degree in special and elementary education after Rebecca was diagnosed with FD.

"I went into it thinking that Rebecca was going to need me to be an advocate for her for the rest of her life," Lisa said.

Jeff has advocated for his daughter in a different way. Several years ago, he began cycling with two of his neighbors to train for the Tour de Foliage. The group has since expanded to more than 10 people, now known as the Rye Brook Riders, who train together every weekend and ride through Rye Brook and Greenwich to build their endurance and speed for the fundraising event.

Ben Tauber is one of the Rye Brook Riders. Tauber learned about the Newman family after reading about them in a newspaper article two years ago. Tauber's 27-year-old sister, Allison, was diagnosed with FD when she was 2-years old and died of the disease a few months before Tauber read about Rebecca.

After reading the article, Tauber contacted the Newmans, who live in his neighborhood, and eventually began riding with Jeff and other neighbors during the weekend.

"That's what made it so random, not only someone in the same town, but literally in the same neighborhood," Tauber said.

"From the moment I saw Rebecca, I could see that exact same feeling that I saw in my sister," Tauber said. "Seeing Rebecca is a good thing, quite honestly. It's very therapeutic to stay involved with the disease."

## **A Disease Without a Cure**

Though there have been significant strides in treating the disease, doctors have not come closer to finding a cure.

The life expectancy of patients has increased—the oldest person living with FD recently died at the age of 61—but as patients have gotten older, they've experienced other ailments such as kidney problems, according to David Brenner, executive director of the Dysautonomia Foundation.

Brenner said awareness is key to diagnosing and treating FD. FD is a recessive disease, which typically only occurs in populations that have been dramatically reduced, such as is the case with Jewish survivors of the Holocaust. A small number of FD cases existed in the beginning, Brenner said, and likely spread through the population via marriage.

Because of this history, the foundation has tried to get the word out to Rabbis to counsel marrying couples to get tested for FD to determine whether they might carry the recessive gene.

Brenner, whose 23-year-old son Michael is also living with FD, said Rebecca has handled the disease with courage.

"She struggles against tremendous odds and she does it with grace and intensity to be as normal as possible and do the things that normal children do," Brenner said.

Still, parents like Lisa and Jeff Newman, have had to reshape their definition of normal.

FD is a regressive disease, which means one of the possible outcomes is that Rebecca's condition could worsen. For the Newmans, each day comes with its challenges—and fears—that Rebecca will be able to live her life as fully as possible.

"My hope is that she can live independently," Jeff said. "I think she will be able to."

"We try to keep the bad days to a minimum," Lisa said. "And we really try to savor the good days."

*The Tour de Foliage will be held on Sept. 12. Breakfast and lunch will be served to riders and all riders receive a free Tour tshirt and water bottle. Registration is \$25 with a minimum \$75 donation requested for riding the 25-mile route and \$100 for riding the 50 mile route. For more information on the Tour de Foliage event, visit [www.fdcycletour.org](http://www.fdcycletour.org).*

*To learn more about familial dysautonomia, visit the website for the Dysautonomia Foundation, [www.familialdysautonomia.org](http://www.familialdysautonomia.org).*