

Shocked by Tay-Sachs, an Atlanta family embraces every day with their daughter

By Margie Fishman
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The Fiers can't recall exactly when their baby girl lost her smile.

It wasn't long ago that Rachaeli would flash a radiant grin whenever somebody new walked into the room, "like an angel visiting you for three seconds," her father would write later.

Then one day, without warning, the smile faded. In its place, a giggle-seizure episode, when Rachaeli would twist her mouth into a grimace and lift her eyebrows.

<http://atlanta.jewish.com/photos/2005/cover101405a.jpg>

Before her second birthday, she lost her sight. Rachaeli's world was dark, yet her father, Eric, was convinced that she could see right through him.

Today, he can no longer separate his daughter from Tay-Sachs, he wrote. They are one.

On Dec. 4, 2003, the Fiers learned that Rachaeli's stay with them would be brief. Her blood tests confirmed the absence of Hexosaminidase A, the enzyme missing in children with Tay-Sachs, a cruel, degenerative disease that is passed on by parents in the same way as eye color. Without Hex-A, a fatty substance, or lipid, accumulates abnormally in cells, especially in the nerve cells of the brain. Over time, that causes progressive damage to the cells.

<http://atlanta.jewish.com/photos/2005/cover101405b.jpg>

"In a strange way, I hate where I'm at. It's a station of false comfort, where I start to forget that things will get worse. For the past six to eight weeks, Rachaeli has been pretty stable. ... God hasn't dropped any new scary stuff on us in a while. ... And then I catch myself starting to think that maybe this is how things might stay. That's the part I hate — that moment of self-delusion when I let myself believe that she's chronically ill, but not terminally ill. 'She's leveled off,' I tell myself — 'this is as bad as it will get.' Then I wake up and remember that she's dying."

— From Eric Fier's online journal, "To Rachaeli With Love."

Inside a Genetic Death Sentence

What is Tay-Sachs disease?

Tay-Sachs is a rare hereditary disease caused by a genetic mutation that leaves the body unable to produce an enzyme (known as Hex-A) necessary for fat metabolizing in nerve cells. Without that enzyme, the central nervous system degenerates. The disease is named for a British ophthalmologist, Warren Tay, who first described the disease in 1881, and a New York neurologist, Bernard Sachs, who first described the cellular changes and the genetic nature of the disease in 1887.

In infants, Tay-Sachs is characterized by progressive mental deterioration, blindness, paralysis, epileptic seizures and death, usually between ages 2 and 5. Late-onset Tay-Sachs, which is far less common, occurs in people who have a genetic mutation that is similar but allows for some production of the missing enzyme.

There is no cure for Tay-Sachs disease.

How is the disease transmitted?

Only through heredity. A Tay-Sachs carrier has one normal gene for production of the Hex-A enzyme and one Tay-Sachs gene. The carrier does not have the illness and leads a normal, healthy life. When two carriers become parents, there is a one-in-four chance that any child they have will inherit a Tay-Sachs gene from each parent and have the disease. If only one parent is a carrier, none of their children can have the disease, but each child has a 50 percent chance of inheriting the Tay-Sachs gene and being a carrier. Carrier couples can explore in-vitro fertilization for family planning.

Who is at risk?

Tay-Sachs carriers are found most often among families of Eastern European Jewish descent (Ashkenazi Jews). In the United States today, approximately one in every 27 Jews is a Tay-Sachs carrier.

Among Jews of Mediterranean (Sephardic) origin and in the general, non-Jewish population, the carrier rate is about one in 250. There are some exceptions: French-Canadians and the Cajuns of Louisiana have the same carrier rate as Ashkenazi Jews, one in 27. Also, people with Irish ancestry are at increased risk for the Tay-Sachs gene.

Any person who can trace his or her lineage to a high-risk population should be tested. In addition, close relatives of carriers must be tested because they also could be carriers. The Tay-Sachs blood test identifies Tay-Sachs carriers by measuring the level of Hex-A in the blood. A second type of screening is a DNA-based carrier test, which looks for specific mutations in the gene that codes for Hex-A.

Since Tay-Sachs is more common in the Ashkenazi community, couples are urged to take the Tay-Sachs blood test before deciding to have children, along with being tested for Canavan disease, Niemann-Pick, cystic fibrosis and Gaucher. Screening has reduced the number of Tay-Sachs cases among Ashkenazi Jews in the United States and Canada by 90 percent.

Locally, Paul Fernhoff, the medical director of the division of medical genetics at Emory University, said that before the Fiers, the last four patients he had seen with Tay-Sachs were not Jewish.

The National Tay-Sachs & Allied Diseases Association is trying to raise awareness about the importance of testing in other communities. Today, there are 41 living children diagnosed as infants with Tay-Sachs in the United States and an additional 78 people with juvenile or late-onset Tay-Sachs. The association does not track numbers by state.

Is there any treatment for Tay-Sachs?

There is no cure for Tay-Sachs, and no treatment will prevent the disease from running its course. It appears that stem cell transplants can slow or stop the progression of the disease. There is no evidence that stem cells can reverse the damage.

In July, the pharmaceutical company Actelion approved a contract for the first clinical trial to involve infants suffering from Tay-Sachs using the drug Zavesca. Zavesca works by reducing the amount of substrate the cells produce so there is less for the Hex-A enzyme to break down. (A substrate is a molecule affected by an enzyme.) It has shown positive results in clinical trials of people with Type 1 Gaucher disease.

Scientists still believe that they are years away from the gene therapy technology to transport the genes into neurons, which would be necessary to cure Tay-Sachs patients.

Sources: The National Tay-Sachs & Allied Diseases Association; Paul Fernhoff, medical director of the division of medical genetics at Emory.

Excerpts from "To Rachaeli With Love"

June 2004:

"I sometimes look at my baby and wonder if she knows that she is sick. Can she see it in my eyes? Though her vision has faded, she seems to see right through me. Her eyes are open windows, bespeaking quiet sadness; then I realize that they are merely mirrors of mine."

September 2004:

"As she lies here in my arms, I miss her terribly. I wonder if this non-responsive breathing child in my arms resembles the non-responsive ashen child I will hold in the future. Is this what it will feel like when I hold her that last day? Maybe I should appreciate every breath, every hiccup, every twitch. Maybe every seizure is proof of life, each convulsion an announcement that she is still here with me."

March 2005:

"Rachaeli is teaching me to listen differently. I listen to her heavenly breathsounds, to her soft vocal sighs. She says more in one sweet sigh than I ever imagined. In her tremors, she dances; in her giggle-seizures, she sings. And when I let myself truly hear her silent words, she tells me that she is choosing to be here with us. We are her family; this is her home. She is fighting to stay, with her own silent determination. She is my little hero."

Rachaeli became only the second Tay-Sachs baby in Georgia in a decade, according to the Fiers, both psychiatrists who live in northeast Atlanta. Her doctors capped her life span at five years.

Eric and Nicole were stunned by the diagnosis. Years earlier, before the births of their three healthy boys, Eric, an Ashkenazi Jew, tested negative as a carrier of Tay-Sachs, even though his sister had tested positive. Tay-Sachs carriers are found most often among families of Eastern European Jewish descent; about one in every 27 Jews in the United States carries the gene.

Nicole, a convert to Judaism of mixed Philippine and Dutch descent, didn't bother getting screened because both parents must be carriers of a recessive disease gene for a child to be affected.

After Rachaeli's diagnosis, Eric was retested and learned that a University of Pittsburgh laboratory had misinterpreted his earlier results. Nicole was informed that she, too, was a carrier, even though the rate in the non-Jewish population is about one in 250.

Immediately, Eric began composing his thoughts. Last year, he launched a Web site, www.rachaeli.com, after noticing a dearth of Tay-Sachs sites from the father's perspective. Eric's site has links to information about the disease, lyrics to "Here Without You" by 3 Doors Down, and a video of Rachaeli's birth, with a man's voice barely audible in the background: "No penis, right? Oh, I love it."

There are dozens of gorgeous pictures of Rachaeli on the site, including a black-and-white portrait of Eric cradling his naked baby as she touches his chin, a feeding tube protruding from her stomach. His online diary spans the nearly two years since his daughter's diagnosis.

"We felt that she was meant to be here with us and she was given to us with a purpose," Eric

said in a recent interview. "There were too many elements of supposed coincidence for this just to be happenstance."

"I let M.B. [a sensitive and spiritual friend] know that my daughter has Tay-Sachs. I remind her that when she met my daughter many months ago, she described her as an 'old soul.' I asked her today what she meant. She says that you can look in the eyes of some people and simply see a depth; a depth that tells you that they have been here before."
From "To Rachaeli With Love."

She was born Eliana Rachael on Oct. 25, 2002; her older brothers nicknamed her Rachaeli. The obstetrician dubbed her a "designer baby" after a flawless delivery. But Nicole couldn't shake that eerie feeling. Her daughter's wide brown eyes stared so intensely, as if she had awakened in the wrong place.

Three months later, Eric, who has a background in developmental pediatrics, knew something was wrong. Rachaeli wasn't rolling over. At 6 months, her body flopped on the couch, and she didn't attempt to crawl. At 9 months, a pediatric neurologist diagnosed her with hypotonia, a relatively mild condition of low muscle tone that can be corrected through therapy.

At the time, Eric, who likes to be emotionally prepared, pressed for further testing. But Nicole resisted, especially after doctors agreed that Rachaeli was progressing, albeit at a slower rate.

Shortly after her first birthday, Rachaeli experienced what appeared to be two severe asthma attacks. The more serious episode came Thanksgiving night, when she began coughing while slurping mashed potatoes and didn't stop for 48 hours, the Fiers remembered. Doctors confirmed that food was entering Rachaeli's lungs rather than her digestive tract, triggering asthmalike symptoms.

Paul Fernhoff, a prominent Emory geneticist, felt Rachaeli's enlarged liver and noticed that she had an exaggerated response to noise. He discussed the possibility of a metabolic disorder and inquired about a family history of Tay-Sachs.

Fernhoff said an eye exam would yield more clues. A cherry-red spot on the retina would indicate that fatty waste products were accumulating in Rachaeli's brain.

Eric was distraught. He knew from his training and from scouring the Internet that every diagnosis with a cherry-red spot was likely fatal.

Nicole convinced herself that the ophthalmologist would confirm what she knew all along: that her daughter did not have one of those horrible diseases.

The next day welcomed the arrival of the consulting team and a swarm of medical students from Children's Healthcare of Atlanta, Nicole recalled. Within seconds of gazing into Rachaeli's eyes, the ophthalmologist was overwhelmed with academic excitement. He encouraged his colleagues to take a look at Rachaeli's bright spot surrounded by a grayish-white halo.

Nicole left the hospital room.

"This is where I have to go pray and just say, 'That didn't just happen,' " she said in a later interview. "Let's just go back to yesterday and start this whole day over again, and you know, I didn't hear the words. La la la la."

Nicole said she felt as if she were hovering above her body, disassociating, before the doctor prodded her to look into her daughter's eyes. "It's either Tay-Sachs or Sandhoff," a disease similar to Tay-Sachs with an even shorter life span, she recalled him telling her. Nicole pulled Rachaeli to her and held on tight.

At 3 p.m., Eric was sitting in his office, counseling a suicidal patient about appreciating the power to choose her own destiny, when his receptionist buzzed in and insisted that the session was over.

He called Nicole, and she told him the news, emotionless. Eric drove the 35 minutes to the hospital, screaming all the way, he recalled.

That night, after reviewing the list of possible outcomes, still uncertain which one to pray for, Eric pretended everything was normal as he corrected his older children's homework. Later, with a sense of urgency, he drove to the home of a woman whose son had died at age 5 of a genetic disease. Eric had never spoken to the woman before, but he had heard about her in the Orthodox community. He arrived on her doorstep unannounced, explained that his daughter was dying, and asked to hear about her son. An hour later, he left feeling comforted.

The next day, Rachaeli's blood tests confirmed Tay-Sachs, and Eric and Nicole were inducted into the "Parents of the Terminally Ill Children Club, an unofficial club to which no one wants or chooses to belong," Eric would write later.

Eric, 37, remembered what it was like growing up with the specter of Tay-Sachs, knowing it was one of the worst illnesses that could strike a Jewish child. Nicole, 39, whose parents had never heard of Tay-Sachs, would later joke bitterly that she was now an official Jew. Her tests revealed a previously undiscovered mutation of the Tay-Sachs gene.

The couple watched their images of life with Rachaeli dissolve, Eric would write later. Rachaeli holding their hands as the family walked home from synagogue. Falling in love. Kissing her own children. Saying "Abba" one last time.

Eric was the crier, and he would comment later that he felt as though medical science had robbed him of the capacity to view his daughter's existence as anything but pathology.

Nicole, by contrast, was the cheerleader, with no time to mourn. She thumbed through the stories of infants living with Tay-Sachs in the National Tay-Sachs & Allied Diseases Association newsletter. He went straight to the death announcements.

Their coping styles were "divergent," she would say later.

"Deadly," he corrected her.

In July 2004, the Fiers investigated a stem cell transplant, an endeavor that both say nearly destroyed their marriage.

Nicole found Joanne Kurtzberg, the director of the pediatric stem cell program at Duke University Medical Center in Durham, N.C., who had performed successful transplants on a handful of children with Tay-Sachs.

Eric was skeptical, worried that the procedure would merely stabilize Rachaeli and not lead to meaningful improvement. He began to wonder what it would be like to live with a child with Tay-

Sachs for 20 or 30 years.

Still, they drove to Durham to learn that Rachaeli's Tay-Sachs was too advanced for her to be a transplant candidate. There was a 90 percent likelihood that she wouldn't even survive the procedure, Kurtzberg told them.

Four months later, one year to the day after her first hospitalization, Rachaeli suffered a life-threatening gastrointestinal bleed that kept her in the hospital for 10 days. Eric later said he viewed the illness not as a linear progression, but as a stepwise decline punctuated by periods of relative calm.

"You just keep redefining normal, but each time something hits, it still catches me by surprise," he wrote. "With each major change I feel that we're one step closer to the end."

In April, Nicole quit her job to care for Rachaeli full time, every day negotiating the delicate dance of managing her daughter's phlegm. If it's too wet, she chokes; too dry and it becomes difficult to cough up.

During a recent interview, the couple alternated using a suction machine to clear Rachaeli's lungs five times in less than three hours. Rachaeli's glassy eyes stared at the ceiling, a purple barrette keeping her hair off her face, opening her mouth like a baby bird.

She now receives medical treatments every three hours, including an experimental drug, Zavesca, which slows the deposit of fatty deposits in the brain and has been tested on patients with Gaucher, another lysosomal storage disease. Rachaeli was too old to participate in a trial for infantile Tay-Sachs patients, so her parents contacted the drug company directly and bought a supply of pills at a cost of \$300 a day, which is covered by their insurance.

The Fiers agree that it may be too late for Rachaeli, but they wanted to try the treatment to help other children with Tay-Sachs. Since beginning the regimen, Eric said, he has noticed hints of emotion registering on his daughter's face.

Nicole said she has prepared for this role her whole life. When she converted to Judaism and became Orthodox before meeting Eric, the rabbi gave her the Hebrew name of Nechama, which means comfort or solace.

Rachaeli has not been admitted to the hospital in a year, and the couple are convinced that is a testament to the in-home care of two doctors. They essentially converted their daughter's pretty pink bedroom into a hospital room, installing a sink and a refrigerator, and purchased wheelchairs, feeding pumps and home nebulizers.

In the mornings, Nicole davens with her daughter. She feeds her through a tube every six hours. When Nicole leaves her to take a shower, she worries that Rachaeli will stop breathing.

Nicole's only free time is when she is sleeping and for two hours on Wednesday mornings, when the in-home nursing aide works an extended shift.

The couple lost a court hearing to have Medicaid pay for additional nursing hours after a lawyer for the state argued that Nicole just wanted time to go to Lenox Square Mall and get a manicure, Nicole said. After the judge denied the Fiers' appeal for another hearing, the couple decided to hire an attorney.

When Rachaeli is not vomiting all night or her tiny foot isn't twitching uncontrollably, her three brothers, ages 5, 8 and 10, adjust to life as normal. She attends their soccer games, cheering silently. They fall asleep hugging her. When their friends ask questions, they respond that "Tay-Sachs is kind of like dying but it's not" or "She can eat as much cotton candy as she wants," Eric said. The other day, the youngest, Ari, turned to a friend after a coughing spell and said, "Don't worry, Avi, it's not Tay-Sachs."

But there are also the serious moments when the boys struggle to understand why Rachaeli will go up to shamayim (heaven) when she is very young while they must wait until they're very old.

When asked what he will do when he meets Rachaeli in heaven, the middle child, Yoni, said: "I will be crying because it will be the first time that she understood me."

As Rachaeli weakens physically, she grows closer to the family spiritually, Eric said. "Every one of their kisses, every one of their caresses, her soul is feeling their soul."

This summer, the family took its first trip together to Israel. The boys prayed for Rachaeli at the Western Wall, stuffing messages into its cracks. "Rachaeli's health remained thankfully well throughout our journey," Eric wrote. "We felt that God, Himself, carried Rachaeli ever so gently through His beautiful home."

The couple left this year's National Tay-Sachs Conference with a new perspective. While the previous year's conference left them feeling drained and depressed, they now understood that despite the absence of one small enzyme, their child was a truly perfect being, Eric wrote.

Sometimes he tries to re-create the hugs he once had with his daughter, Rachaeli's flaccid body resting in his lap, her head nestled in the bend of his left arm. He places her right arm over his shoulder, strokes her hair, kisses her cheek and tells her he wishes they could do this forever.

"She knows that her abba is with her. That she is not alone," he wrote. "And neither am I."

As Rachaeli approaches her third birthday Oct. 25, Eric acknowledges feeling scared about what the next chapter will bring and how he will cope. Earlier this year he was overcome by the beauty of normalcy, he wrote, watching a child at synagogue climb up in a chair, holding her head up with ease and whispering in her father's ear.

"When I'm old, I don't think I'll be afraid of dying as much because I'll see her again," Eric said in a recent interview. Then he began to cry, explaining that sometimes when you open a window and peer in, you want to close it right away.

"It's important to open it every now and then," his wife said softly.

Both know in their hearts, after all, that if Rachaeli needs to be here, who better to take care of her?

God "chose us to care for her soul," Eric said. "I recognize that there are some gifts you keep forever and some gifts you need to return."