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# HEALTH&FITNESS

## A family copes as a child battles a rare disease

# Niemann-Pick Type C

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(Mel Greer/Staff photo)
**The Marella family, from left, Andrew, 3, mother Andrea, Dana, 9, Julia, 11, Philip, 6, and father Philip, has had to learn how to deal with illness. Dana has Niemann-Pick Type C, a disease characterized by the inability of cells to properly metabolize cholesterol.**

From an early age, Dana Marella was considered clumsy. It was not unusual for her to spill her milk or walk into tables, say her parents, Philip and Andrea Marella.

She developed an unsteady gait and appeared to be pigeon-toed. She also started having difficulty moving her eyes up and down quickly.

But initially the Marellas, who live in Greenwich, were not alarmed by the behavior of their second oldest child, now 9.

"That's just Dana" was the pat explanation. "She is how she is, clumsy, and that's how she'll always be," her parents recall thinking. But by the time Dana was five, it was apparent her problems went far beyond coordination.

Over the next four years, as Dana's condition grew progressively worse, doctors told the Marellas about several possible causes. They told them Dana may have leukemia or Lyme disease, that she may possibly be suffering from Attention Deficit Disorder or retardation.

But none of those diagnoses could account for all of the symptoms that Dana was exhibiting. Worried about her lack of balance, her parents had her take ballet and karate lessons. When they discovered she had a bullseye rash on her body, possibly caused by a tick bite, Dana began therapy for Lyme disease.

But no matter the diagnosis or therapy program, Dana's condition did not improve. No one could tell Andrea and Philip what was wrong with their daughter, and they grew increasingly frantic.

"We went everywhere to find out what she had," says Andrea. Her husband adds, "I can't even tell you how many different doctors we saw."

Then, this spring, the Marellas finally met someone who could give them some answers.

Dr. Marc Patterson is a pediatric neurologist at Columbia University. He specializes in a rare genetic disorder called Niemann-Pick Type C, a disease characterized by the inability of cells to properly metabolize cholesterol. That means that patients with Niemann-Pick Type C accumulate excessive amounts of cholesterol in the liver, spleen and brain. This causes the cells to become smothered by the cholesterol. Then the cells die one by one.

The disease, according to Patterson, is often misdiagnosed as clumsiness, a learning disability or mild retardation, and has only been diagnosed in between 300 and 400 people in the United States.

"Unless you suspect the disease and know the correct test to use, you won't know the diagnosis," Patterson says.

The primary indicator of Niemann-Pick Type C is speed of movement of the eyes, Patterson says. Patients with the disease suffer from a condition called vertical supranuclear gaze palsy, which is the inability to voluntarily move the eyes up and down quickly.

"It's the tip-off to the diagnosis," Patterson says.

Even when properly diagnosed, there is no treatment or cure for the disease; it is always fatal.

When Patterson examined Dana Marella, he knew almost immediately she had Niemann-Pick Type C.

Dana is a fourth-grader at Parkway School, where she participates in a specialized curriculum. An effect of Niemann-Pick Type C is that Dana suffers from dementia, and has difficulty with speech and walking. At the school she receives speech and occupational therapy, and was recently fitted for a walker.

Yet while the effects of her condition are apparent to others, they are not so apparent to Dana, her parents say.

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"She has a dementia," Andrea says, adding, "I think she thinks she's normal."

Though the disease presents physical and mental challenges, Dana is a happy child, her parents say.

"She's a very happy kid, who loves her friends," says her father.

"People bond with her quite quickly and have always wanted to help her," says Andrea, noting Dana's very best friend, who she identifies only as Alix. "She's an angel from heaven. This little girl doesn't leave her side."

And the students and faculty at Parkway School have also been wonderful to their daughter, the Marellas say. For instance, administrators are discussing a way for the school to participate in a fund-raiser for Niemann-Pick Type C research, (Fund-raising has suffered because so few people have been diagnosed with the disease.)

"The moms I've gotten together so far are terrific, everybody just stepping up to the plate and contributing tremendously," says Andrea.

Pressed by her parents to talk about school and her best friend Alix, Dana, who has difficulty following conversation, manages to say, "We do everything together. She tickles me. She makes me laugh."

Knowing their daughter has Niemann-Pick Type C, the Marellas realize her condition will only worsen. Right now she can swallow without assistance, but many patients end up on a feeding tube.

"There's no doubt she has regressed physically, but she is far from those stages," says Philip, but he adds, "It's not hard to discern where we're going."

Patterson says the prognosis for someone in Dana's condition can vary. The disease usually affects children of school age, but it may strike at any time from early infancy to adulthood. The vast majority of children who have it die before age 20 (and some die before the age of 10). Late onset of symptoms can lead to longer life spans but it is extremely rare for any person to reach 40.

"It really is a progressive dementia, and in that way is similar to Alzheimer's disease," Patterson says. "I can't really predict how long someone will live. That depends on other considerations, such as nursing resources."

In addition to vertical supranuclear gaze palsy, other indicators of the disease include liver and spleen problems in the first few months after birth. (The presence of Niemann-Pick Type C is confirmed by taking a skin biopsy, growing the cells in the laboratory, and then studying their ability to transport and store cholesterol).

"It's a slow progressing disease," Patterson says. "A progressive and lethal disease," - one that cannot be cured or treated, though some of its symptoms, such as seizures or cataplexy, can be controlled or tempered by drugs.

While Niemann-Pick Type C is characterized by an inability to metabolize cholesterol, doctors find that cutting down on cholesterol does not curb the effects of the disease. Nothing has proven capable of slowing the progression of the disease, though a new human trial is underway.

The Marellas, meanwhile, retain hope.

While traditional medicine has thus far produced no solutions, the family has explored some unconventional avenues. Dana is on a homeopathic regimen of pills, which she takes seven times a day, and last spring she and her mother went on a pilgrimage to Lourdes, France. There, at the Roman Catholic shrine, Dana was bathed in waters that some feel hold miraculous healing powers.

"You do everything you can in the interim," says Andrea, adding, "It's very devastating for us as the parents to watch our child deteriorate and know there's nothing we can do."

While researching Niemann-Pick Type C, the Marellas met Mike and Cindy Parseghian. The Parseghians have had three children with the disease, two of whom died. Mike Parseghian is the son of former University of Notre Dame football coach Ara Parseghian, for whom a Niemann-Pick research foundation was named.

Working with the Ara Parseghian Medical Research Foundation, and the National Niemann-Pick Disease Foundation, the Marellas hope to raise money and coordinate research of the debilitating disease. They recently founded Dana's Angels Research Trust, and last month held a private fund raiser at a backcountry Greenwich home.

Motivated by their daughter's condition, and by the hope that other families will not suffer similar experiences, the Marellas are focusing their attention on funding research for Niemann-Pick Type C.

"When you're talking about success with a disorder like this, you're talking about slowing the effects of the disorder," Philip says.

Studies have shown that a drug called Zavesca can slow progression of the disease. Consequently, Patterson is using the drug in human trials at Columbia University. The trial is set to last one year, and is limited to patients 12 years of age and older.

"While people are working on the ultimate cure, we're trying to find other treatments that will improve length of life and chance of survival," says Patterson, who hopes to extend a trial to younger patients such as Dana by next year.

The Marellas three other children - Julia, 11, Philip, 6, and Andrew, 3 - have not exhibited symptoms of the genetic disorder that afflicts their sister.

"What I'd want people to understand is that we see hope in all the efforts that are being made, which will quite probably lead to treatment for an even wider range of these dementia disorders," says Philip. "If there is something in this for us, it would be wondering if it had an even greater good. When it comes down to challenges like this, there aren't a lot of easy answers. You have to persevere."

Donations for research into Niemann-Pick Type C can be sent to Dana's Angels Research Trust, 15 E. Putnam Ave., #117, Greenwich, Connecticut, 06830-5424.