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Bellflower girl succumbs to rare genetic disease

By Kelly Puente, Staff Writer

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Valerie Montgomery's 23-month-old daughter Caileen Harrison was born with a rare genetic disorder called Niemann-Pick Disease. Most children with the disease do not live past the age of 5; Caileen died earlier this month. (Brittany Murray/Press-Telegram)



Caileen Harrison and her mother, Valerie Montgomery, leave Kaiser Permanente after a physical therapy session last month. (Brittany Murray/Press-Telegram)

BELLFLOWER - Two weeks ago Valerie Montgomery was planning her daughter's second birthday party. Then, suddenly, she was planning a memorial service.

Montgomery knew her baby's lifespan would be short. The Bellflower mom was just hoping for at least a few more years, or months. But on the morning of April 19, Caileen Harrison quietly slipped away from complications from a rare terminal disease called Niemann-Pick Type A/B. She was 23 months old.

"I know people are telling me she's in a better place, but I wish I'd had more time with her," Montgomery said. "She was a little angel on this Earth."

Niemann-Pick is one of a group of genetic disorders that affect the body's metabolism. Only about 1,700 people in world are diagnosed with a type of the disease, according to statistics from the National Niemann-Pick Disease Foundation.

Caileen's form of the disease, Type A/B, is so rare there are no statistics for it.

"It's a disease that can be called 'ultra rare' because we have so few diagnosed cases," said Cate Walsh Vockley, an educational coordinator with the foundation.

In people with Niemann-Pick Type A/B, the body has an enzyme deficiency

and is unable to break down fats. Fatty substances build up in the liver, spleen and brain, causing brain damage and muscle weakness. There is no cure or treatment.

"The whole body basically begins to shut down," Walsh Vockley said.

Signs came early

A common symptom in children is a distended abdomen due to an enlarged liver and spleen, a sign Montgomery said she noticed minutes after Caileen was born on May 7, 2008.

"Her belly was round and hard like a beach ball, and I kept thinking something is not right with this belly," she said.

Still concerned about the size of her daughter's stomach, Montgomery brought Caileen to a doctor at Kaiser Bellflower Medical Center a few weeks later.

"The doctor told me I was feeding her too much," she said.

Over the next several months, Caileen's belly grew larger and her condition deteriorated. Next to other 1-year-olds she seemed small and less developed. Doctors couldn't figure out what was wrong, Montgomery said.

The baby, then 13 months old, was referred to Dr. Rebecca Mardach-Verdon, a genetics specialist at Kaiser Southern California Metabolic Center in Los Angeles.

Mardach-Verdon noticed the baby's round belly and performed a test for Niemann-Pick. Caileen tested positive for Type A/B, Montgomery said.

The prognosis was grim.

"They didn't expect her to make it past 5 years old," Montgomery said. "I lost my breath. All this time I'd been telling them something's not right."

Walsh Vockley says it's common for patients with Niemann-Pick to be misdiagnosed early on because the disease is so rare. The symptoms can be mistaken for other problems, such as mild retardation.

With the knowledge that her daughter might have only a few years left, Montgomery remained constantly at her baby's side. As her muscles grew

weak, Caileen required round-the-clock care.

"Every day is a struggle," Montgomery said last month in her Bellflower home. "I see her getting weaker."

But despite her ailments, Caileen was a happy baby who was always smiling and laughing, her mother said. She especially loved to eat. Her favorite food was liver and onions.

Already a mother of two adult daughters and a grandmother, Montgomery said she was surprised to learn she was pregnant with Caileen at age 41.

A former Metro bus driver, Montgomery met Caileen's father, a Metro supervisor, at work. The two didn't stay together after Caileen's birth, but they remained close friends.

Collis Harrison, a father of eight, doted on his tiny daughter, Montgomery said. And Caileen would light up whenever her father walked in the room.

"Her favorite thing in the world is her daddy," Montgomery said.

Parents show no signs

Niemann-Pick is autosomal recessive, meaning that children with the disease have two copies of the abnormal gene. Each parent carries one copy of the gene without having any signs of the disease themselves.

When both parents are carriers, there is one in four chance that a child will have the disease.

Niemann-Pick affects all segments of the population, but higher incidences have been found in certain populations, including the Ashkenazi Jewish population, the French Canadian population of Nova Scotia, the Maghreb region of North Africa and the Spanish-American population of southern New Mexico and Colorado.

Because it is so rare, research has been historically underfunded. It's a problem with many of the more than 7,000 rare diseases recognized by the National Institutes of Health. A disease is considered rare if fewer than 200,000 people in the United States have it.

Walsh Vockley said one of the biggest hurdles is finding enough patients for clinical trials. And in severe forms of the disease like Type A and Type AB,

patients may live only a few years.

However, research has progressed for the mildest form of the disease, Type B, with many of those patients living into adulthood. Mount Sinai School of Medicine in New York is conducting research on bone marrow transplants, enzyme replacement therapy and gene therapy.

DNA testing is available for some forms of the disease and has proved effective for cutting back on the numbers, especially in the Ashkenazi Jewish population, Walsh Vockley said.

Mom urges testing

Montgomery said she wanted to tell her story to raise awareness of the disease and urge parents to get tested.

"I would tell every parent to get their DNA checked," she said. "We don't want any more parents hurting from losing their little baby."

For now, Montgomery says she is working to stay strong and trying to move forward. She's staying with her mother in Los Angeles and is in the process of getting a job with the U.S. Census Bureau.

Although the past two years have been difficult, she says each moment with Caileen was precious.

"If I had to do it all over again I would," she said. "I wouldn't have sent her back for the world."

kelly.puente@presstelegram.com, 562-499-1305