



Billy Therriault, 3, of Fairfield, suffers from spinal muscular atrophy, a disease that is not widely known but kills more babies than any other genetic disease. Billy's mother Katy, top right, and friend Lee Baldwin Hafner are working to bring more awareness of SMA.

Courageous fighters

Fairfield family, group battles against devastating disease

This is part one of a two-part feature on Spinal Muscular Atrophy, a degenerative neuromuscular disease that affects the very young. Part two will appear in Wednesday's Health/Science section and will take a deeper look at the disease and advances in research. Today's story is about one Fairfield family's experience and Fighter Mom, an organization that helps parents advocate for their special needs children.

By **EILEEN FISCHER**
Staff Writer

Lying on his bed in his playroom, Billy Therriault, 3, quietly watches his mom, Katy, with big round eyes. One small arm and one leg are slightly raised by a wooden pulley system hanging over his bed that mom had specially made "so he'll always have the capability to move," she said.

Movement is important, Therriault

explained, for children like Billy who have spinal muscular atrophy, a genetic disorder that affects the motor neurons of the spinal cord and brain stem. This neuromuscular disease weakens the muscles of swallowing, breathing and limbs, according to the Web site, Fight SMA, an organization dedicated to finding a cure.

There are three types of SMA and Billy has Type 1, the most severe, said his mom.

"He's a paralytic. He can't swallow, he can't walk or talk and can't do anything by himself," she said. "He uses a breathing machine when he sleeps."

Billy is most comfortable, she said, when he's in water — taking his daily bath or, in summer, playing in their backyard pool with his sisters, Alexandra, 7, and Ella, 5 — because in the water he can sit up.

"He enjoys that," she said, stopping every few minutes to suction out saliva from his mouth because he can't swallow.

Billy is fed 18 to 20 hours a day on a drip feed, she said, which goes directly into his small intestine.

"Cognitively, he's completely normal," she said, as are most SMA children. "He greets you; he smiles; he'll wave 'hi' if I hold up his hand.

"He's a very happy child."

Arranging three toy dinosaurs in front of his smiling eyes, Therriault said they are her son's favorite playthings. "What does a dinosaur say?" she asks him, lovingly.

In this spacious room, Billy has a computer, a TV, a mirror on one wall that reflects everything and everyone around him — his nurse, his special education teacher, the family dog, Maxine, and his friend Sloane, 3, here for a visit with her mom, Lee Baldwin Hafner. In this swirl of activity, mom is at the center, explaining the daily drill of his care, watching over every detail.

"I take care of him. No one else 'coughs' him," said Therriault, explaining the machine that loosens her son's mucus. Colds are particu-

larly dangerous to these youngsters because they can't blow their noses or cough to clear their lungs and airways. Even though Billy can't speak, she said, "I can just read him. If he's not doing well, I know what he needs or I pray about it."

Therriault and her husband, Scott, who live in the Southport section of Fairfield, are fighters and optimistic about their son's future. With a protocol that's followed religiously 24 hours a day, seven days a week — there is no treatment for SMA — Billy is doing very well, said Therriault.

"I take one day at a time [and] stay completely optimistic," she said.

It's a daily grind with a special needs child, she said, and family, friends and other SMA moms she's met on the Internet have been helpful as has FightSMA and its latest program, Fighter Mom.

FightSMA was started by Martha and Joe Slay, of Richmond, Virginia, about 17 years ago, the Slays' oldest child, Andrew, now 22 and attending the University of Virginia, was diagnosed at 14 months with Type 2 SMA, a more benign form of the disease, said Martha Slay.

With promising drugs in the pipeline and proposed legislation for funding research, "There's tremendous reason to be hopeful" about SMA, said Slay.

In North America, 12.8 percent of children struggle with chronic health problems, according to the group. Over the years, she's had many, many requests from other parents of chronically ill children, who were not SMA patients, for information on how to advocate for their youngsters, too, she said.

Through Fighter Mom, the Slays share their expertise on the Internet (www.fightermom.org) and have compiled a manual that explains how to go about organizing a fundraiser, how to lobby for legislation and how to partner with researchers.

On Friday, Fighter Mom will have its first workshop with a panel discussion in Manhattan to help folks become proactive, said Slay.

Both Hafner and Therriault plan to attend the workshop. Hafner, also a Southport resident and the mother of three young daughters, has devoted a lot of her time to help her close friend and increase awareness about SMA.



Before Billy's diagnosis, Hafner said she had never heard of SMA, which surprised her since it is more common than some other genetic diseases that are more well-known, such as cystic fibrous, she said.

"I reached out to the medical community and when I did, nobody had heard of this disease," said Hafner. "This disease is the No. 1 genetic killer of babies under age 2. I couldn't believe that it was so unknown."

Pediatric neurologist Azimuddin Kazi, of Neurological Specialists in Fairfield, Stratford and Shelton, and at Bridgeport Hospital, said SMA affects one child in every 6,000 to 10,000 live births. In his practice, he sees one to two cases a year, he said.



Scott Therriault, left, talks to his son.

Tracy Deer-Mirek/
Staff photographer

Even though SMA is rare, more awareness about it is important, he said.

When Billy was born, he was a normal, healthy baby, said Therriault. There were no signs that anything was wrong until he was three months old and mom began to notice some weakness and that he wasn't moving his legs enough. When she breast-fed him, his tongue seemed to vibrate in a strange way, she said.

"I thought he wasn't strong or it was 'floppy baby' or that he had a form of cerebral palsy," she said. "There's a bunch of diseases that come up."

Kazi said decreased movement is one of the first signs of the disease, especially in Type 1. Tongue fasciculations, the vibration that Therriault felt, is also typical.

"Nerve fibers are degenerating in the tongue muscles," said Kazi. SMA "involves those muscles early on, but we don't know why."

At four months old, a neurologist diagnosed Billy with SMA Type 1.

"It was only his 15th [SMA] patient the doctor diagnosed," said Therriault. About one in every 40 people are carriers of the mutant gene, or about 7.5 million people, which is recessive, but if both parents are carriers, they have a 25 percent chance of having a child with SMA. Most of these children die by 9 months, generally of respiratory failure.

Last year, Hafner produced a documentary on the Therriaults and their battle with New York filmmaker Adam Goldstein titled, "Invisible Heroes: The Fight Against SMA."

"I decided that this disease needed more attention," said Hafner, who had no filmmaking experience before this endeavor. She is planning a benefit for SMA research and hopes the film will help with fundraising. Slay said she thought "Invisible Heroes" was very moving and should be a big help in increasing SMA awareness.

To the Therriaults, Hafner's friendship and support is immeasurable.

"Words can't say enough," said Scott Therriault. "If you have a child, you know how much you love the child and to have someone who is there to take such a big role in helping us find a cure, the gratitude is unexplainable."

The first Fighter Mom workshop to support mothers and others fighting children's diseases will take place Friday from 8 a.m. to 5 p.m. at the New York Palace Hotel in New York City. The event is free.

There will be several panel discussions during the day. Journalist and writer Catherine Crier, who started her career on CNN and hosted "Catherine Crier Live" on Court TV, will speak on leadership and hope for life's tough battles.

The event is sponsored by the Toy Industry Foundation.

For more information, visit: www.fightsma.org, www.fightermom.org, or www.toyindustryfoundation.org or call FightSMA at (804) 515-0800.