

# Taking notice OF AN obscure illness

Research into  
spinal muscular  
atrophy is put  
on a fast track



Tracy Deer-Mirek/Staff photographer

**Katy Therriault of Fairfield hugs her son, Billy, 3, who suffers from spinal muscular atrophy, a disease that kills more children than any other genetic disorder.**

*This is part two of a two-part feature on spinal muscular atrophy, a degenerative neuromuscular disease that affects the very young. Part one ran on Sunday.*

By **EILEEN FISCHER**  
Staff Writer

**A** little known disease is getting some major attention. Spinal muscular atrophy is a debilitating genetic disease that produces weakness in the muscles of swallowing, breathing and the limbs.

According to the Fight SMA Web site,

SMA occurs when a vital gene — the survivor motor neuron gene or SMN — is deleted or mutated. This prevents the creation of a protein necessary for muscle strength.

It is a relatively rare illness that affects one in every 6,000 to 10,000 births, but has a high infant mortality rate due to respiratory failure. Close to 70 percent of children who are diagnosed with SMA Type 1 (there are three types) don't live to see age 2, said Martha Slay, founder of Fight SMA, which

raises awareness and money for research. Her son, Andrew, now 22, is living with the SMA Type 2, a more benign form.

Scott and Katy Therriault's son, Billy, 3, has Type 1 SMA. The Fairfield family is the subject of a documentary on the disease, "Invisible Heroes: The Fight Against SMA," produced by a close friend, Lee Baldwin Hafner, also from Fairfield.

Katy Therriault said she had never heard of SMA until her son was diagnosed.

One out of every 40 people, or more than 7 million Americans, are carriers. SMA is a recessive gene, but if both parents are carriers, they have a one in four chance that their child will have SMA. Most people don't know if they are at risk.

"Since it is recessive, you have no idea until you have a child," said Slay. While there is genetic testing available, there is usually no reason to ask for testing unless another family member has already been known to have the gene.

At present, there is no cure or treatment for the disease, but what has made Slay, the Therriaults and others optimistic about finding a cure is the support from the National Institutes of Health and the National Institute of Neurological Disorders and Stroke that put SMA research on a fast-track.

"Some promising compounds [drugs] have almost come through that are ready to go into clinical trials," said Slay who was in Washington, D.C., recently with other SMA parents, visiting legislators to ask them to support the funding bill, the SMA Treatment Acceleration Act.

Research for a cure for SMA can conceivably help other neuromuscular diseases such as Parkinson's and Duchenne Muscular Dystrophy — the most common type of MD — say its supporters.

Dr. Kenneth Fischbeck, chief of neurogenetics at the NIND said, "SMA is an important problem."

It is "the most common severe heredity disease of infancy and early childhood and there is no treatment," he said.

The NIH got behind SMA research several years ago, said Fischbeck, not only because it is such a devastating disease, but because SMA is "a good target for developing treatment."

The discovery of the SMN1 gene on chromosome 5 in 1995 and its copy — SMN2 — is promising information for researchers to investigate, said Fischbeck. Studies with animals have also had some encouraging results.



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**Katy and Scott Therriault of Fairfield comfort their son, Billy. He has spinal muscular atrophy.**

With the NIH coordinating independent work and clinical trials on SMA, there is hope for faster progress towards a cure.

Dr. Alex MacKenzie, a molecular geneticist and pediatric neurologist at the Children's Hospital of Eastern Ontario, said it's an exciting time for SMA research.

"It's a perfect storm of a fairly poignant and tragic disorder caused by a unique mutation, which is something we can do something about," he said.

In Ottawa, he sees about 30 or so children a year with SMA. While there is no cure, progress has been made taking care of children who have it. Babies with the severe form are living longer than they were five years ago, he said, because "parents and clinicians are getting more aggressive with nutrition and respiratory [therapies]."

The role of exercise is being recognized as well, he said.

In Connecticut, about 20 children and six adults are living with SMA, according to Families of SMA spokeswoman Mary Ellen Barrelle. It's difficult to get statistics on the prevalence of the disease and FSMA reports

on what information it has been able to gather through its contacts with families.

The group raises funds for research and offers families support. They also have an equipment pool, such as wheelchairs and cough assistant machines, said Barrelle.

While the medical experts are cautiously optimistic about finding a treatment soon, Slay is very encouraged by the NIH support and the SMA Treatment Acceleration Act.

"There is tremendous reason to be hopeful," she said. "People really know how to take care of these children so they're ready for these therapies."