

Mother crusades for newborn screening

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Had Stephen Monaco been born a few hours down Interstate 95 in North Carolina, he would have avoided a life of wheelchairs, feeding pumps, medications, therapies and disability.

But after three years of a mostly normal, healthy life, including conquering "the big slide," helping his mom bake a cake for his grandmother's birthday and playing with his big brothers, Stephen Monaco succumbed to a disease lurking secretly in his body.

Woodbridge resident Jana Monaco gave birth to her third son, Stephen, seven years ago. Monaco, her husband Tom and area doctors never knew Stephen's picky eating habits, strange odor and slow recovery from Roto Virus at 18 months of age warned of his genetic condition.

"I wish I would have known then, I wish I would have picked up on it," Jana Monaco said. "He had all the signs and symptoms, but it's the basic knowledge that you don't know."

Although Tom and Jana Monaco's other two sons were born healthy, Stephen inherited his parents' recessive traits for a very rare genetic disease called Isovaleric Acidemia, or IVA.

Stephen's body lacked the enzyme to break down the amino acid leucine, a byproduct of protein.

On the night of May 29, 2001, Jana Monaco sent her youngest son to bed after making sure he had a Pedialyte ice pop to combat dehydration from a what she thought was a common stomach virus.

Stephen Monaco awoke around 2 a.m. to go to the bathroom. Jana Monaco gave her son another ice pop, and tucked him back into bed. Stephen Monaco told his mother he loved her for the last time.

The next morning she found her son in his crib breathing very fast and with glazed, unfocused eyes.

"I repeatedly called his name, but he did not respond. When I picked him up, my precious child felt like a floppy rag doll with his teeth clenched tightly," Jana Monaco wrote in a story recounting Stephen's ordeal.

She called 911 and rode with Stephen to the hospital, where he would stay for the next few months.

After a transfer to a pediatric intensive care unit, a variety of medical tests and odd questions repeated by many doctors, the Monaco family finally learned the truth.

This rare disease affecting one in 50,000 American babies each year could have been detected at birth. With a special diet and medication, Stephen could have avoided this metabolic crisis that damaged his brain and severely disabled him.

Each state regulates its own newborn screening for genetic diseases, and Stephen's disease was not included in the nine tested for by Virginia hospitals. North Carolina screens for 36, including IVA, according to Jana Monaco, who has become an expert on the subject through experience.

Stephen's family considered it a blessing that he merely made it out of the hospital alive. But the Monaco family and Stephen would never be the same.

They slowly adjusted, and made it through the first, most difficult months with help from friends, family and hospital staff. The Monacos also turned to their strong Roman Catholic faith and the St. Elizabeth Ann Seton Church in Lake Ridge.

A private medical fund allows him to receive therapy at home. Lifting his head up, smiling and laughing have been great strides for Stephen Monaco.

THERAPEUTIC ACTIVISM

Preventing other families from experiencing a similar fate through activism became the Monacos' mission.

"When Stephen was diagnosed, and we went through that whole experience, that was devastating," Jana Monaco said. "But when we learned it was preventable, that was even harder to swallow. I made a promise to myself a long time ago and to Stephen that if he could survive, we would put a hand in and make a difference so that no one else would have to go through this ordeal."

They became the poster family for newborn screening a year ago when Jana Monaco gave birth to Caroline, who also inherited IVA. Doctors immediately treated her, released her within three days, prescribed medication and dietary restrictions. They expect her to live a normal life.

"It became a very therapeutic thing to do," Jana Monaco said. "Preventing other families going through such a tragedy seems too important to us. I felt that Stephen's suffering won't be in vain. He's already made a difference with [Caroline]."

Jana and Tom Monaco have spoken in Washington, D.C., Richmond and elsewhere, advocating for expanded newborn screening. She presented a slide show to a group of pediatric doctors and nurses in Fairfax County. The Organic Acidemia Association asked Jana Monaco to join its board of directors.

"One of the problems is the lack of understanding, even within the medical community, with these metabolic disorders," Jana Monaco said.

TAKING ACTION

A federal Department of Health and Human Services committee is expected to release guidelines for each state to follow on newborn screening, although states can not be forced to comply.

The Virginia General Assembly's Joint Commission on Health Care is also working to expand Virginia's testing, and plans to use the federal recommendations to create a bill, according to Kim Sneed, executive staff director of the Joint Commission.

Dumfries Delegate Jeffrey M. Frederick, R-52nd District, also plans to submit a bill which would expand Virginia's newborn screening, he said.

"I decided it was a good idea. It makes a lot of sense," Frederick said. "I think it's something that can easily be implemented and have a huge impact."

Frederick decided this summer he would submit the bill, and doesn't want to wait for the Commission to act. If two legislators submit similar bills, they will just combine and work together, he said.

"I don't know what the Commission is going to do," Frederick said. "I am not accountable to any commission. I am accountable to the people who voted for me."

Frederick believes the bill has a good shot at passing, but every piece of legislation has opponents, he said.

He expects insurance companies might dislike the bill for cost reasons, but he will argue this change would actually save money.

Virginia has already purchased three new Tandem Mass Spectrometers used to test for a variety of diseases like IVA. Current estimates say extended screening would cost approximately \$20 to \$40 per baby.

"I understand any new cost is a new cost," Frederick said. "I don't discount their argument in the sense it will cost a little bit of money, but these screenings can avoid so many future medical costs. It's a preventative measure.

"If this prevents one additional doctor visit for a baby then you've paid for it."

Stephen's new wheelchair alone cost his parents \$825 out-of-pocket. Their insurance company paid the remaining \$3,675.

It's a no-brainer, according to Dr. Piero Rinaldo of the Mayo Clinic in Minnesota. He has spent over 20 years working with children and metabolic disorders and he helped diagnose Stephen Monaco.

He also sits on the federal advisory committee preparing screening guidelines.

Moral, ethical and financial reasons all justify increased screening in all states, Rinaldo said.

"People are getting tangled in a meaningless debate," he said.

There are 54 diseases of this type that current technology can test for, and new information emerges all the time, Rinaldo said

He hopes the federal government will provide funding as an incentive for states to increase screening efforts. Rinaldo stressed the importance of fairness, uniformity and universality in screening.

"This should have been done a long time ago in a uniform way across the country," he said. "Perfectly normal children within a matter of hours die. My job is to go and tell the parents this is why your child died. It's gut-wrenching when you tell them ... 'sorry your baby was born in the wrong state.' "