Family Copes With Rare Genetic Disorder

By David Ruppe
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Hunter Moff is a 6-year-old baseball fanatic who likes to rollerblade and swim in the neighborhood creek. His sister Sydney, age 11, takes horseback riding lessons, likes arts and crafts and keeps a gerbil.

Last month, both were diagnosed with Sanfilippo syndrome, a little-known, fatal genetic disorder affecting an estimated one in 24,000 people, or around 12,000 Americans.

The diagnosis, of course, was a terrible blow for the family. But Steve and Theresa Moff, married 16 years, are now, like other parents of children with rare diseases, strongly motivated to promote awareness of a disorder so unusual that few doctors have ever encountered it.

Lacking an Essential Enzyme

Named for one of the first doctors in the United States to describe the disorder, Sanfilippo syndrome is caused by a rare inherited defect. It usually leads to a deterioration of the body and mind. Patients usually die between the ages of 10 to 15 years, although some do live longer.

So far, science has produced nothing that can slow down the deterioration or cure the disorder.

As with six other, similar disorders, Sanfilippo children are missing an essential enzyme that breaks down a complex sugar produced in the body.

Over time, the sugar builds up in the brain, stopping normal development and usually resulting in hyperactivity, sleeping disorders, loss of speech, coarse features, loss of physical ability, mental retardation, dementia and finally death. Depending on the severity of the case, affected children can lead active, nearly normal lives to a point — until they plateau and begin to deteriorate.

For a child to have Sanfilippo syndrome, both parents must have the recessive trait. Even then, there is only a one-in-four chance the child will have it. The Moffs also have an 8-year-old boy, Stefan, who is perfectly healthy.

Puzzling Symptoms

The fact that it took 11 years before the Moffs found out they had children with a fatal disorder illustrates just how little is known about it.

Beginning in infancy, Hunter and Sydney both showed a variety of symptoms that puzzled doctors but did not appear life-threatening.

Hunter was diagnosed with attention deficit hyperactivity disorder, pervasive developmental disorder and autism. His everyday behavior was most striking — a constant lack of focus, speech trouble, and a relentless drive for getting into things.

Hunter looks and acts, in many ways, like a normal kid, though one a couple of years younger than his actual age. But his tantrums and periods of aggressive activity are more intense than for most youngsters, his parents say.

Sydney, who doctors say has a milder case, was diagnosed with three different hearing disorders and with Retinitis pigmentosa, a group of eye diseases that eventually cause loss of vision.

The parents were frustrated because doctors could find no explanation for all of these ailments.

“We went through nine years of searching for an answer,” says Steve Moff.

Mixed Signals

Sydney was examined by one of the country’s top pediatric specialists. Both children were tested for standard genetic disorders, but nothing turned up.

“There was no reason to think about this with them,” says Theresa Moff, a pediatric nurse practitioner who has spent a great deal of time with children and families coping with catastrophic illness.

Because the siblings showed different symptoms, doctors did not necessarily think the two had the same illness. Both have learning disabilities, speech difficulty and behavioral problems. Sydney, though, unlike Hunter, has unusual patches of light skin, and doesn't show some of the central identifying features of the disorder, such as hyperactivity.

"When I had two kids with problems, I said there's got to be some relationship here. I always knew in the back of my mind there's got to be something," says Steve, a professor of marketing at the Pennsylvania College of Technology in Williamsport.

"What they've found with improved testing and improved recognition is there is obviously a whole range of variability in this disease," says Theresa.

Scott Myers, a specialist in neuro-developmental pediatrics at the Geisinger Medical Center in Danville, Pa., finally diagnosed Sanfilippo syndrome in July.

"I think the key was putting together the two kids," says Myers. "That's a lot of bad luck for two kids in one family to have such significant problems and there's probably a way to tie those together.

"I was looking for things that could sort of present cognitive problems, this type of behavioral issue, the short stature, the facial features, and the fact there's this sibling who has Retinitis pigmentosa and sensory neural hearing loss," he says. "It was those things that made me think about Sanfilippo syndrome and check for it."

The Importance of Knowing

The diagnosis, of course, has been devastating for the family — though neither child has been told the disorder is fatal. But knowing the cause of the children's problems has been important, the parents say.

Now specific therapies can be applied to treat known symptoms. Surgically inserting a shunt, or tube, designed to drain fluid from the cranium has been found to improve behavior in some Sanfilippo patients. And Hunter's hearing will be regularly tested so he can get hearing aids as soon as he needs them.

The family recently wrote a letter to their relatives, describing the illness and explaining how other members of the family could also be carriers of the recessive gene. With that knowledge, other family members can decide whether to be tested for the recessive gene. The test can involve analyzing cells from a blood sample.

“From a procreation point-of-view, you need to know if you’ve got these things going on because you could to pass it on to your kids,” says Theresa.

Having a diagnosis also has helped the family discover government resources available to the children. Some information has come from doctors, some from Theresa’s contacts in the medical community, and some from word-of-mouth — other parents who have children with disabilities.

“There’s a special kids network, a parents advocacy group. Usually if you go to the therapeutic preschools out there they can put you in touch with people who can help you know your legal rights,” says Theresa.

Increasing Awareness

The family has applied for state programs to have people regularly help with Hunter’s behavior at school and home. Other services provide respite-care aides who come to the home and allow the parents to go to work, shop or just to get away for a short time.

Hunter’s non-stop activity “has a cumulative effect on us. It’s every day. It’s constant. It’s no break,” says Steve.

The Moffs are planning to have key-operated deadbolt locks installed on doors leading outside, “So I can take a shower upstairs and not worry about Hunter running out into the street,” says Steve.

Next year, they hope to begin building a handicapped-accessible house in a more rural location.

Because the disorder is so rare, there isn’t a lot of data to allow doctors to predict when and how quickly Hunter and Sydney will begin to get worse. Some people with Sanfilippo syndrome have been found to live into their 20s and 30s.

There isn't enough information out there "to know who won't really decline for a long time, and who is going to really go downhill before 10 years of age," says Myers.

Funding for research is much less than might be found for more common childhood illnesses and disorders. So Steve and Theresa hope to help promote public and government awareness of Sanfilippo syndrome.

There are a handful of researchers around the globe studying the disorder, looking at possible enzyme replacement, cell replacement and gene therapies. A foundation set up in 1995 by another family has granted more than $1.2 million for research, which remains in the early stages.

The disorder recently got some national exposure when the April 6 episode of the NBC-TV series ER featured a story about a girl with Sanfilippo syndrome.

As Normal as Possible

Coming to terms with the medical consequences of Sanfilippo syndrome is toughest, says Steve. "The hardest part is knowing your two children are going to pass away in your lifetime."

But Sanfilippo syndrome, like other catastrophic illnesses and disorders, presents numerous other challenges for families, such as dealing day after day with the behavioral effects and physical circumstances of the disease; the teasing children with disabilities encounter; and finding out and accessing just what resources are available to families.

"You've got these tremendous battles to fight, just with your children and their health. Then these other things start compounding on top of it," says Theresa.

The Moffs hope to give all of their children as normal a life as possible, including their healthy child Stefan. "This is as much his reality as it is mine. He's going to grow up to lose his brother and sister," says Theresa.

Baseball is a help. Both boys love to watch and love to play. Hunter is in a league for kids with special needs. The family has also taken kayaking, hiking and skiing trips and spends several afternoons a week swimming.

The children are encouraged to strive and take risks, the parents say. Sydney recently knocked out a tooth on a skiing trip.

“I don’t want pity to be the reason people do things. I don’t like that as a driving force. In the long run that’s not going to do them any good,” says Theresa. “They still need to be disciplined. They still need to be held to the same expectations, to be pushed to their maximum potential. And when you bring in pity, it doesn’t happen.”

As for 8-year-old Stefan, “He’s learning a very valuable lesson in tolerance and patience and unconditional love,” his mother says. “I think he’s going to be a better kid for having to go through all of this.”