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## Heartbroken, but not beaten

### Parents of terminally ill boys push for law to help catch, treat similar diseases early

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Even using walkers, their twin boys could no longer make it to the park at the end of the block. Bob and Sonya Evanosky knew something was seriously wrong, something that couldn't be explained by the cerebral palsy diagnosis the previous year.

They were right: In December 2004, they learned that John and Christopher, then just 3, did not have cerebral palsy but suffered from a genetic disease that strikes just one in 40,000 people.

There is no cure for metachromatic leukodystrophy, although there are signs that an early stem-cell transplant can stay some of its devastating effects.

But by the time the symptoms are apparent, as they were with John and Christopher, it is too late. An early death -- most children don't live past 5 years -- is certain.

"The doctors told us, 'take your kids home, make them comfortable, there's nothing we can do,'" said Sonya Evanosky, 37, of Aurora.

But after doctors found that their third child, Jack, then just 1 year old, also had the disorder, the Evanoskys decided they could do something to keep other parents from suffering the same fate.

Their efforts resulted in a bill, passed unanimously and now awaiting the governor's signature, that makes Illinois the first state in the nation to mandate blood samples drawn from newborns be tested for five diseases similar to metachromatic leukodystrophy, collectively known as lysosomal storage disorders. There are 40 of these disorders, which affect about 1 in 5,000 people, but tests for early detection have only been developed for the five included in the legislation.

The legislation, Bob Evanosky said Friday, "better justifies our tragedy."

"There's a part of me that is glad we did it, but there also is a part of me that wishes we never had to," he said.

The Illinois Department of Public Health now tests for 36 diseases and in July will add cystic fibrosis to the list. At

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that time, the cost for all the tests, which typically is paid by insurance carriers or Medicaid, will be \$56. Testing for the five lysosomal storage disorders would add another \$12.

"You are talking about saving lives and improving lives," said Sen. Dale Righter (R-Mattoon), who introduced the legislation after meeting with the Evanoskys. "That's a real tangible benefit. In fact, it's the most important one you can have."

Screening newborns for diseases began in the 1960s when Dr. Robert Guthrie developed a test for phenylketonuria and a system for collection and transportation of blood samples on filter paper. Today, newborn testing is regulated by individual states, concentrating on diseases that can be cured or effectively treated if diagnosed early.

Bob Evanosky, 39, and his wife both carried a recessive gene for metachromatic leukodystrophy, giving each child a 25 percent chance of having the disease and a 50 percent chance of being a carrier. But they didn't know that when they settled into a spacious home in Aurora, just west of Illinois Highway 59, a few years ago.

The couple had married as undergraduates in college and waited a few years to establish their careers before starting a family. After Sonya Evanosky became pregnant, they learned she was carrying identical twins.

"We were jumping up and down when we found out," said Bob Evanosky, who decided to leave his career as a commercial airline pilot to care for the boys. Sonya Evanosky works as an Information Technology director for a local financial services company.

John and Christopher Evanosky were born five weeks' premature but seemed initially to thrive.

By 3 months, they were sleeping through the night. By 5 1/2 months they were eating solid food and rolling over, according to a log kept by the Evanoskys.

But as they aged, their development slowed.

When neither boy could walk by 16 1/2 months, Bob and Sonya were concerned.

Initially, the boys were diagnosed with cerebral palsy, which can accompany premature birth. Months of intense physical therapy followed. John and Christopher learned to toddle with the help of reverse walkers. But even that progress ebbed away by the fall of 2004. It was then that the boys could no longer make the short trip to the park at the end of the block.

An MRI taken two days after Christmas showed significant degradation of the nerves in the boys' brains, a telltale sign of metachromatic leukodystrophy.

The disorder is caused by an enzyme deficiency. Waste builds up in the myelin sheath of nerve cells. There is no cure for the disease, but there is data to suggest that a transplant of stem cells harvested from a healthy umbilical cord performed before the symptoms appear can stay the cell deterioration.

But it was too late to try that treatment on John and Christopher, who have been devastated by the disorder.

Each is on nine medications to minimize pain and swelling and to keep unused muscles from constricting. There are medicines to minimize the saliva their mouths produce to keep them from choking, medicines to keep airways open and the digestive system working.

The boys can no longer talk, no longer walk. Their hips are displaced.

"For all intents and purposes, their bodies don't work anymore," Bob Evanosky said. "Cognitively they are there. They are trapped inside a broken body."

Dr. Joanne Kurtzberg, director of the pediatric stem-cell transplantation program at the Duke University Medical Center, said stem-cell transplants must be done early.

"In order to prove maximum neurological function, they need to be done in the first couple months of life," she said. "Without screening ... it is often too late."

Kurtzberg has performed 160 transplants on children with lysosomal storage disorders over 12 years. The risks of transplantation are high; 20 percent of patients do not survive. But rewards can be great, too.

"There's no question that if a child doesn't die early of a complication, [transplantation] is beneficial. It extends life and it improves the quality of life," Kurtzberg said, although the treatment is still too new to know long-term benefits.

Illinois would become the first state to test for five lysosomal storage disorders because the testing technology is so new. If the bill becomes law, the Health Department would begin testing the blood of newborns for Krabbe, Pompe, Guacher, Fabry and Niemann-Pick diseases in late 2009 or early 2010.

Genzyme Corp., a Cambridge, Mass.-based pharmaceutical company, developed the technology and is offering to give it away. The company sells enzyme replacement therapies for Gaucher, Fabry, and Pompe.

New York has tested for one of the diseases, Krabbe, for about a year. The state began testing thanks largely to the lobbying efforts of Hall of Fame quarterback Jim Kelly, whose son Hunter died of the disease in 2005 at age 8.

Metachromatic leukodystrophy was not included in the Illinois legislation because Genzyme still is developing a test for it. However, the public health director has the authority to add the test when it is available, and the Evanoskys hope that will happen soon.

Kurtzberg performed a stem-cell transplant on the Evanoskys' third son, Jack, in April 2005.

Today, Jack has a feeding tube in his stomach and a pump delivers nutrients and the 10 medicines that he must take every day.

But none of the medicines are for metachromatic leukodystrophy. Although the disease degraded Jack's nervous system for about a year while the stem cells grew new bone marrow, the degradation appears to have stopped.

"When you go to pick him up, you can feel him push off the floor," Bob Evanosky said. "What we see today is tenfold better than what we saw six months ago."

At one time, Bob Evanosky dreamed he would teach his boys to fly, teach them to work with wood so that they could decorate their own houses with millwork the way he had accented the family's home.

Today, the Evanoskys dream of helping other people's children.

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