Crusading for a cure

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Ilyce Randell could have followed the doctors' advice to put 5-month-old Max in a nursing home and allow the rare genetic disorder known as Caravan disease to slowly claim his life.

Max couldn't hold up his head, roll over or reach for a toy. He seemed unaware of the world around him. He barely slept, waking every 30 to 40 minutes during the night and screaming for hours on end.

But instead of giving up on Max, Randell fought back. She established Canavan Research Illinois. She sought out treatments to prolong and improve Max's life. She created Web sites and met with public officials to raise awareness for Canavan disease. And she launched major fund-raising efforts to cover the cost of research and treatments that hold the only hope for the tiny victims of this fatal brain disease.

In the six years since Max was diagnosed, Randell has raised about half a million dollars for Canavan research - $30,000 with a charity ball on Make A Difference Day 2003 alone. For those fund-raising efforts, and for her work to help families touched by Canavan, Randell was chosen by the Daily Herald as this year's local Make A Difference Day winner and the recipient of a $500 charitable donation.

"She has been a major motivator, and an energetic person who brought a lot of attention to the disease," says Dr. Paola Leone, a leading Canavan researcher and director of the Cell and Gene Therapy Center at the Robert Wood Johnson Medical School in New Jersey. "Truly, this makes her a hero."

Randell, who lives in Buffalo Grove with her husband and two sons, never set out to be a hero. She was just a mom with a baby who wasn't developing the way he should have.

But rather than blindly accepting what doctors spelled out as the inevitable, Randell immersed herself in the world of genetic diseases. Nowadays, she talks about gene therapy and viral vectors with the ease of a med student.

It's all about hope. For the first time, there are treatments that offer a dose of that precious commodity to children born with Canavan - treatment that might someday help against Parkinson's, Alzheimer's and a host of other far more common diseases.

Devastating diagnosis

Max, now 6¨, watches his little brother dump a box of books out on their family-room floor. With a bit of coaxing from mom, Max smiles.

Max can't join his brother, but he can appreciate the toddler's antics. He can't talk, but he communicates a little with a blink of his blue eyes. He can't walk, but he can move his hands enough to maneuver a small electric wheel chair.

With special software, he can spell his name. He even attends afternoon classes at the Gillet Educational Center in Arlington Heights. His mother hopes he will one day learn to read.

A few years ago, all this seemed impossible.

When he was 5 months old, Max failed to meet those developmental milestones that pediatricians watch for in babies. He also suffered from tremors.

"He wasn't tracking anything," Randell says. "He seemed very disconnected from the world."

Doctors diagnosed Canavan, a rare genetic disorder seen mostly in Jews of Central and Eastern European descent. His parents, Ilyce and Michael, had never heard of it. Soon, they knew all there was to know.

Canavan is a neurodegenerative disorder that affects the formation of myelin, the protective sheath around the nerves in the brain. Because of mutations in the gene for the enzyme aspartoacylase, Canavan patients cannot break down an acid known as NAA, leading to a dangerous buildup in the brain and interfering with the formation of myelin.

The disease causes the brain to atrophy. Children remain trapped in immobile bodies, often unable to hold up their heads or sit without help. Most never speak. And as the disease progresses, it can lead to seizures, blindness, the inability to swallow and, eventually, death.

The disease affects about 500 kids in the United States and 4,000 worldwide. There is no cure, and most die before they turn 10.

Doctors held out little hope for Max and told the Randells he likely would die before he turned 2.

"They said, 'Take your baby home. Try not to get attached to him. And you might want to look at pediatric nursing homes,'" Randell recalls.

Michael and Ilyce refused to listen.

A viral weapon

In the battle for federal research dollars, a disease that affects just a few hundred kids is a hard sell. So Canavan families have had to raise much of the money on their own. Their efforts, however, are finally paying off.

In the mid-1990s, Dr. Leone and others worked on the world's first gene therapy for a genetic brain disease, and two Canavan patients received the preliminary treatment in 1996 in New Zealand.

The same therapy, with some modifications, was approved by the Food and Drug Administration in 1998 for a trial at Yale University in New Haven, Conn., and Jefferson Medical College in Philadelphia. A handful of children received the treatment, which was administered into the fluid of the brain. A few months after his diagnosis, Max was one of them.

At only 11 months old, he was the youngest child in the world to receive a gene transplant.

In 2001, Leone and her research team at Jefferson started using a more potent viral delivery system for the gene therapy. Basically, researchers alter the genes of a virus "to trick the virus to deliver what we want," Leone says.

Neurosurgeons drill six small holes into the patients' skulls. Leone then operates a pump that injects the virus, carrying the aspartoacylase gene, into the children's brains.

Ten children - including, once again, Max - have received the therapy since 2001 and six more patients are scheduled for this year, Leone says.

All but one have shown improvements in their quality of life. Max, for example, has regained some of his vision and his arm movements. Even his head and neck seem stronger.

"His head control has never been this good in his life," Randell says.

Still, the effectiveness of the treatment is limited by the damage Canavan has already done to the brains of these children.

By age 2, 60 percent of the brain has atrophied, Leone says. So for gene therapy to work well, it must be administered when the children are just a few months old.

"If a child has minimal brain damage, the treatment could do much more," Leone says.

Yet, she emphasizes, the treatment is still "very experimental," and there's no indication yet that it provides a cure.

That's why many families simply accept Canavan's slow death sentence. They don't see the point in fighting back.

Randell doesn't understand that approach. Even if current advances aren't enough to save Max, what doctors learn now might one day help her younger son Alex's children. Alex, though healthy, carries the Canavan gene and could pass it along to another generation.

Hope for the future

It is not yet known how much improvement Canavan families can expect if children are treated early enough, but already the system of delivering altered genes directly to the brain through viral means - known, in this case, as an adeno-associated virus or AAV - is being used to treat other diseases.

Last year, New York-Presbyterian Hospital started a clinical trial using virus-based gene therapy in severe Parkinson's patients. And soon, a trial using the therapy for Batten Disease, another progressive genetic disease that kills victims by their 20s, is slated to begin at Cornell University.

In addition, researchers are interested in looking at how stem cells could help children with Canavan. Leone says that research is about $800,000 away from a clinical study, again emphasizing the importance of fund raising.

Leone calls fund raising efforts, like those done by Randell, "crucial."

"Federal funds are difficult to come by ... and even more so for rare diseases," Leone says.

So, the Randells keep plugging away at fund raising and lobbying efforts. Her next event, a "Shop for the Cure" gift exhibition in Arlington Heights, is planned for May 2.

All of this happens against a backdrop of Max's daily needs. Ilyce and Mike Randell must carry him, bath him, reposition his limbs, even help him take a sip of water.

"It's such an uphill battle, trying to raise money, taking care of Max, advocating for him with the schools," Randell says.

Over the years, friends and family have pitched in. And even strangers who've read about Max have donated to Canavan research or attended a fund-raiser to help.

Randell wants them to know they've helped in so many ways.

"We really reached out to the community," Randell says. "We said, 'This is our situation, and we need help.' And people responded