Stories of Hope: Batten Disease

Tony and Katie Ferrandino were out of hope when they left the hospital with their 4-year old son, Drew, that January day in 2007.

“Our plans for hospice and 24-hour nursing were in place. We were basically going home to watch our son die,” Tony Ferrandino said. Then Drew giggled. Loud.

It was like a call to arms. The couple knew they could not give up the battle they had waged since their son’s third birthday, when a seizure alerted them that something was going very wrong with their little boy.

Gradually, Drew lost his ability to walk, talk, and even eat. Their son had Batten disease, an always-fatal failure of the cells’ ability to dispose of its waste products.

StemCells Inc. of Palo Alto was conducting a very small trial — six patients — designed to determine the safety of injecting neuronal stem cells into the brains of children with Batten disease. Drew had little chance of being one of the six, the parents were told. But on July 31, 2007, he became the fourth child to undergo a stem cell transplant at Doernbecher Children’s Hospital in Portland, Ore.

“We consider ourselves lucky to be part of the hope of stem cells,” Ferrandino said.

- Watch the Spotlight on Batten Disease talks

Curing Batten Disease

What if no one ever took out the garbage? That’s essentially what happens in the cells of children with neuronal ceroid lipofuscinosis, sometimes called Batten disease. Every cell in our bodies contains tiny waste disposers called lysosomes. Within their acidic environment, a gang of various enzymes chop up the waste produced in the cell’s normal operation. But in Batten disease, a genetic defect interrupts the formation of a single enzyme. In time, the cells of these children swell with accumulating waste products and die. The brain shrinks. The children who once developed normally lose ground, forgetting how to walk and how to talk, losing their vision and finally, their lives.

StemCells Inc. of Palo Alto hopes to correct this genetic error by injecting neuronal stem cells into the brains of children born with this defect. Between 2006 and January of 2008, six children were injected with neuronal stem cells in a Phase 1 study designed primarily to evaluate the safety of the procedure. A surgeon drilled four dime-size holes called trephinations on each side of a patient’s head and injected stem cells just beneath the brain’s thin ribbon of cortex, said Stephen Huhn, MD, vice president of StemCells Inc., who directs its central nervous system program.
The children will be monitored for five years. Huhn, a pediatric neurosurgeon, took a leave from Stanford University to help direct the clinical trial conducted at the Oregon Health Sciences University.

"The opportunity to develop, even in a small way, a field that might offer potential for restoration and repair of the damaged brain is very exciting," he said.

In addition to monitoring transplant safety, researchers are looking for signs of some positive effects. In animal models, transplanted human neural stem cells successfully migrated to the correct portions of the brain, integrated with surrounding neurons and began producing the missing enzyme, said Ann Tsukamoto, PhD, StemCells’ executive vice president of research and development.

"Whether we’ll be able to show measures of efficacy in a safety study when these patients are very severe, that remains to be seen," Tsukamoto said.

Although the number of children with Batten disease is relatively small, there are more than 40 known forms of lysosomal storage disorders, about half of which affect the brain. Any of those may prove future targets for stem-cell therapies, Tsukamoto said.

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