FDA panel mulls gene therapy for kids with rare eye disease (Update)

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(HealthDay)—A U.S. Food and Drug Administration advisory panel was

poised on Thursday to recommend approval for a gene therapy that could grant the gift of sight to young people with a rare type of inherited vision loss.

Replacing a nonworking gene with a new one is opening a new world for children and teens with the inherited retinal disease called Leber congenital amaurosis.

"This is a gene therapy that can restore some vision to people who have very limited vision or no vision due to the mutation in the RPE65 gene, and as such, it's a great breakthrough," said Stephen Rose, chief research officer at the Foundation Fighting Blindness.

For those who have already received the therapy, the treatment has been life-changing.

Eleven-year-old Cole Carper got the treatment when he was 8, according to the *Associated Press*. Afterwards, "I looked up and said, 'What are those light things?' And my mom said, 'Those are stars,'" he said.

His 13-year-old sister, Caroline, was treated when she was 10. "I saw snow falling and rain falling. I was completely surprised," she told the wire service. "I thought of water on the ground or snow on the ground. I never thought of it falling."

If approved by the FDA advisory panel, the treatment would be on its way to becoming the first gene therapy approved for an inherited disease, said Rose. His foundation helped fund the research that led to the treatment. The FDA typically follows the advice of its expert panels.

Only one other gene therapy has so far met FDA approval—a cancer treatment that the agency sanctioned in August.

This type of vision loss is rare and affects about 1,000 people in the United States, Rose said. "But for these individuals who are essentially blind, it's a huge impact," he said.

In total, about 200,000 Americans have some type of inherited disease that causes blindness involving some 250 different genes, Rose said.

"We are not restoring 20/20 vision," he said. "We are restoring functional vision."

This means that people can be mobile without the need for a guide dog or a cane, Rose said.

Rose said this therapy treats only this one type of vision loss. "There are 22 different genes that can cause Leber congenital amaurosis—RPE65 is only one of them," he said.

The treatment, voretigene neparvovec (Luxturna), was developed by Philadelphia-based Spark Therapeutics.

Clinical trials using gene therapy, drugs or cell therapy to treat other types of inherited visions are currently going on, Rose said.

This new treatment is proof that replacing a gene in the eye can restore some vision, he said, and along with other therapies, offers hope for people with previously untreatable inherited vision loss.

It has been tried on kids as young as 4, Rose said. "The earlier you treat the better," he said. "Ideally, you would treat people as early as possible and prevent any retinal degeneration."

Whether the treatment will last a lifetime isn't known, Rose said. But people who received the treatment in early trials more than 10 years ago

continue to have their sight, he noted.

Dr. Jean Bennett, a professor of ophthalmology at the University of Pennsylvania in Philadelphia, is one of the researchers who is actually giving the treatment.

Using a harmless virus to carry a new, working RPE65 gene, doctors perform microscopic surgery and, with a tube about the width of a human eyelash, implant the new gene into cells in the retina, she explained.

For maximum vision improvement, the procedure has to be done in each eye, Bennett said.

"We think this same procedure would be effective for other genes," she said. "It would just require that the gene we used be swapped out and replaced with a different gene."

Dr. Zenia Aguilersa, a pediatric ophthalmologist at Nicklaus Children's Hospital in Miami, said that the FDA is taking a big step in gene therapy for these genetic eye diseases.

"If we can treat the disease early, we may prevent blindness in all these kids," Aguilersa said.

It isn't known how much the treatment will cost or if it will be covered by insurance, Rose said. But the foundation believes that everyone who needs the treatment should get it.

"Our goal is that there will be treatments for individuals who need these treatments such that nobody will ever have to hear: 'You have retinal degeneration, get a guide dog, learn braille, get a cane,'" Rose said.

More information: Jean Bennett, M.D., Ph.D., professor, ophthalmology, University of Pennsylvania, Philadelphia; Stephen Rose, Ph.D., chief research officer, Foundation Fighting Blindness; Zenia Aguilersa, M.D., pediatric ophthalmologist, Nicklaus Children's Hospital, Miami

For more on inherited retinal disease, visit the American Academy of Ophthalmology.