

Gene therapy eyedrops restored a boy's sight. Similar treatments could help millions

July 24 2023, by LAURA UNGAR and FREIDA FRISARO



Dr. Alfonso Sabater, checks Antonio Vento Carvajal's eye under a blue light after applying a stain to check to see if more ulcers had developed, before a gene therapy treatment, Thursday, July 6, 2023, at University of Miami Health System's Bascom Palmer Eye Institute in Miami. Antonio was born with dystrophic epidermolysis bullosa, a rare genetic condition that causes blisters all over his body and in his eyes. He was blind for much of his life but can see again after getting gene therapy eyedrops. Credit: AP Photo/Wilfredo Lee

Dr. Alfonso Sabater pulled up two photos of Antonio Vento Carvajal's eyes. One showed cloudy scars covering both eyeballs. The other, taken after months of gene therapy given through eyedrops, revealed no scarring on either eye.

Antonio, who's been legally blind for much of his 14 years, can see again.

The teen was born with dystrophic epidermolysis bullosa, a [rare genetic condition](#) that causes blisters all over his body and in his eyes. But his skin improved when he joined a clinical trial to test the world's first topical gene therapy. That gave Sabater an idea: What if it could be adapted for Antonio's eyes?

This insight not only helped Antonio, it also opened the door to similar therapies that could potentially treat millions of people with other eye diseases, including common ones.

Antonio's mom, Yunielkys "Yuni" Carvajal, teared up thinking about what Sabater did for her son.

"He's been there through everything," she said in Spanish during a visit to the University of Miami Health System's Bascom Palmer Eye Institute. "He's not only a good doctor but such a good human being and provided us with hope. He never gave up."

The family came to the U.S. from Cuba in 2012 on a special visa allowing Antonio to get treatment for his condition, which affects around 3,000 people worldwide. He had surgeries to remove scar tissue from his eyes, but it grew back. Antonio's vision kept getting worse, eventually deteriorating so much that he didn't feel safe walking around.



Dr. Alfonso Sabater, left, examines Antonio Vento Carvajal's eyes accompanied by his mother, Yunielkys Carvajal, right, Thursday, July 6, 2023, at University of Miami Health System's Bascom Palmer Eye Institute in Miami. Antonio, who spent much of his life legally blind, can see again after months of gene therapy delivered with eyedrops. Credit: AP Photo/Wilfredo Lee

Sabater had no answers then, and tried to reassure the boy: "I'll find a solution. I just need some time. I'm working on it."

"Yeah, I know you're going to do it," Sabater recalled Antonio saying. "That gave me the energy to continue."

At one point, Carvajal told Sabater about the experimental gene therapy gel for Antonio's skin lesions. He contacted drugmaker Krystal Biotech

to see if it could be reformulated for the boy's eyes.

Suma Krishnan, co-founder and president of research and development for the Pittsburgh-based company, said the idea made sense and "it didn't hurt to try it."

Antonio's condition is caused by mutations in a gene that helps produce a protein called collagen 7, which holds together both skin and corneas. The treatment, called Vyjuvek, uses an inactivated herpes simplex virus to deliver working copies of that gene. The eyedrops use the same liquid as the skin version, just without the added gel.



Dr. Alfonso Sabater, uses a syringe to apply gene therapy eyedrops to Antonio Vento Carvajal's eyes, Thursday, July 6, 2023, at University of Miami Health System's Bascom Palmer Eye Institute in Miami. Antonio was born with

dystrophic epidermolysis bullosa, a rare genetic condition that causes blisters all over his body and in his eyes. He was blind for much of his life but can see again after getting gene therapy eyedrops. Credit: AP Photo/Wilfredo Lee

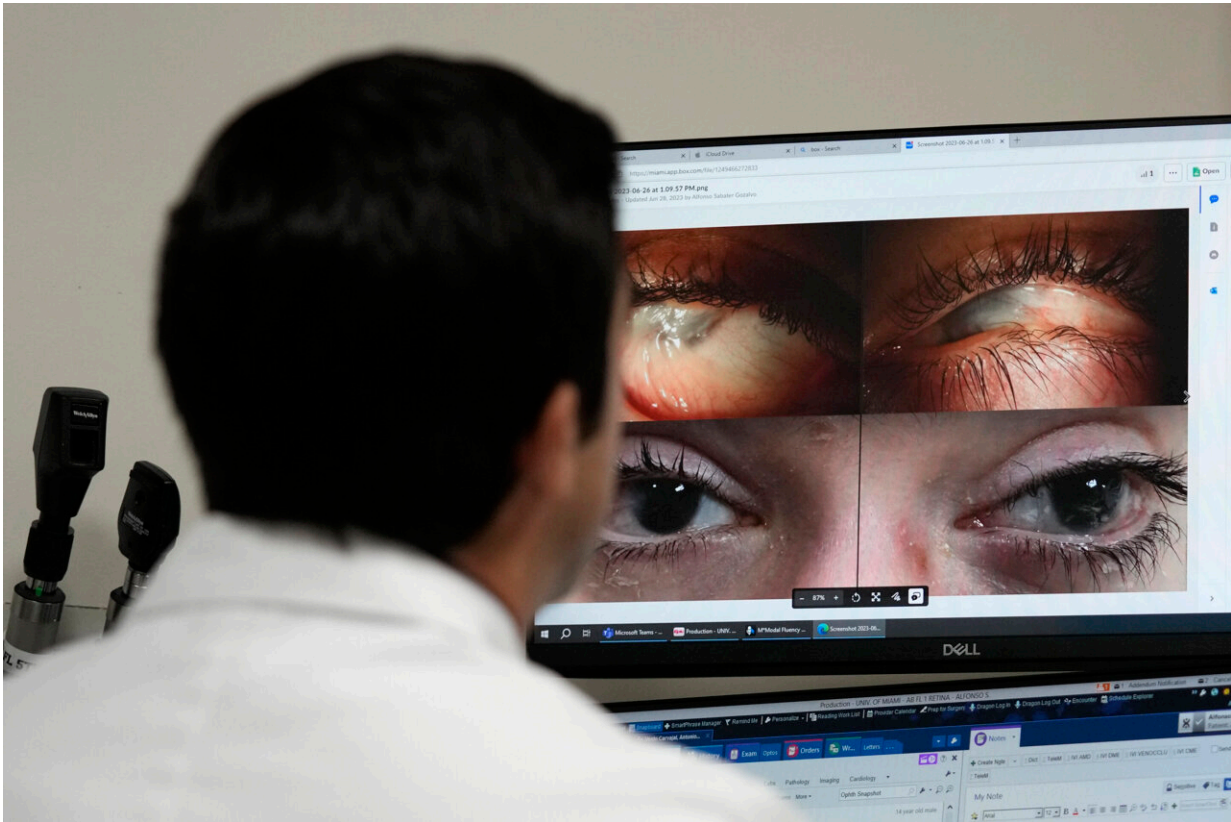
After two years, which included testing the drug in mice, the team got "compassionate use" approval from the U.S. Food and Drug Administration and permission from university and hospital review boards. Last August, Antonio had surgery on his right eye, after which Sabater started treating him with the eyedrops.

Krishnan said they were cautious, frequently watching to see that it was safe.

Antonio's eye recovered from the surgery, the scarring didn't return and there was significant improvement each month, Sabater said. Doctors recently measured the vision in Antoni's right eye at a near-perfect 20/25.

This year, Sabater began treating Antonio's left eye, which had even more scar tissue. That one is also steadily improving, measuring close to 20/50, which Sabater said "is pretty good vision."

Antonio comes to the eye institute for checkups almost weekly and gets the drops once a month. During visits, Antonio must wear [protective clothing](#) covering his arms, hands, legs and feet. Like other kids with the condition—who are sometimes called "butterfly children"—his skin is so fragile that even a touch can wound him.



Dr. Alfonso Sabater, left, pulls up photos of Antonio Vento Carvajal's eyes photographed before and after surgery and gene therapy, Thursday, July 6, 2023, at University of Miami Health System's Bascom Palmer Eye Institute in Miami. Antonio was born with dystrophic epidermolysis bullosa, a rare genetic condition that causes blisters all over his body and in his eyes. He was blind for much of his life but can see again after getting gene therapy eyedrops. Credit: AP Photo/Wilfredo Lee

Antonio still uses the skin gel, which was [approved by the FDA](#) in May and can also be used off-label on eyes. It doesn't modify DNA, so it's not a one-time treatment like many gene therapies.

Sabater, director of the Corneal Innovation Lab at the eye institute, said gene therapy eyedrops could potentially be used for other diseases by

changing the gene delivered by the virus. For example, a different gene could be used to treat Fuchs' dystrophy, which affects 18 million people in the U.S. and accounts for about half the nation's corneal transplants.



Dr. Alfonso Sabater, left, examines Antonio Vento Carvajal's eyes, Thursday, July 6, 2023, at University of Miami Health System's Bascom Palmer Eye Institute in Miami. Antonio, who spent much of his life legally blind, can see again after months of gene therapy delivered with eyedrops. Credit: AP Photo/Wilfredo Lee



Photos of Antonio Vento Carvajal's eyes, left, photographed before and after surgery and gene therapy, are displayed on a computer screen as his mother, Yunielkys Carvajal, center, looks into his eyes while they attend a doctor's appointment, Thursday, July 6, 2023, at University of Miami Health System's Bascom Palmer Eye Institute in Miami. Antonio was born with dystrophic epidermolysis bullosa, a rare genetic condition that causes blisters all over his body and in his eyes. He was blind for much of his life but can see again after getting gene therapy eyedrops. Credit: AP Photo/Wilfredo Lee



Yunielkys Carvajal, gives her son, Antonio Vento Carvaja, a hug and a kiss after a doctor's visit, Thursday, July 6, 2023, at University of Miami Health System's Bascom Palmer Eye Institute in Miami. Antonio, who spent much of his life legally blind, can see again after months of gene therapy delivered with eyedrops. Credit: AP Photo/Wilfredo Lee

The prospect of treating more conditions this way is "exciting," said Dr. Aimee Payne, a dermatology professor at the University of Pennsylvania who isn't involved in the research. The approach "delivers gene therapy that really addresses the root cause of disease."

With his vision restored, Antonio has enjoyed a typical teen pastime he's wanted to do for quite a while: playing video games with his friends. And he finally feels safe walking around.

Sabater said the two-year journey seeking government and hospital approvals "was worth it. Just for Antonio, it was worth it ... but also because it opens the space to treat other patients in the future."

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