Sisters who share a progressive eye disease find hope in a gene therapy right here in Pittsburgh

Bethlehem's Tammy Krents recently became the city's first recipient of an FDA-approved therapy for retinitis pigmentosa

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Tammy Krents remembers thinking she would have to quit her job of 16 years as a preschool teacher due to progressive vision loss.

“I remember the day after I lost my [driver’s] license, I went in to my boss and told her I had to quit,” Krents, 55, of Bethlehem, Pa., recalled, “She said, ‘No, you won’t quit. We’ll find you a way back and forth to work.’ And that’s what they did.”

Some 10 years later, still in that job, Krents’ concerns about her vision remained, a result of an inherited retinal degeneration that until recent

years had no treatment. But again, she found that help wasn't far: Her sister, Kimberley Bell, 52, tracked down a solution, right here in Pittsburgh.

Krents and Bell were both diagnosed at a young age with the same genetic disorder, one that causes progressive blindness. Relying on each other for years to navigate a world slowly fading from view, they now have hope as the city's first recipients of an FDA-approved gene therapy. Bell's diligent research led the sisters to the treatment, with Krents the first to receive it, at the UPMC Vision Institute in the Hill District.

"Personally, I just don't want to go blind," said Bell of Johnstown, Pa. "A lot of people take things for granted, including your eyesight, which I did when I was younger. I just want to be able to experience life, and live as independently as I can and enjoy my family and be able to see everything."

Krents was approved first for the procedure, with surgeries on each eye — weeks apart, to ensure all went well — in June.

Bell faced an insurance roadblock, but, after winning an appeal unanimously, she now eagerly awaits the treatment that she discovered not long ago.

"A couple months ago, my disease started to progress, and my left eye, I realized I couldn't see well out of it at all," Bell said in June. Her local optometrist connected her with a retina specialist, she said, who mentioned that genetic testing was available.

UPMC Vision Institute patients Kimberley Bell, left, and her sister Tammy Krents are receiving a gene therapy to treat the inherited condition that has led to degeneration of their eyesight. (Sebastian Foltz/Post-Gazette)

While awaiting results, Bell conducted a quick Google search and noticed a treatment, Luxturna, available for a type of retinal dystrophy with a mutation in the RPE65 gene. After receiving the test results several weeks later, she learned she had a mutation of that very gene, indicating that she has retinitis pigmentosa.

Bell's Luxturna research led her to a solution less than 90 minutes from her home, at the UPMC Vision Institute, which opened in 2023. Once she made the initial call to the hospital system, things moved quickly. The sisters underwent hours of testing, with both genetically and clinically eligible for Luxturna.

"Out of over 220 mutations that can cause retinal dystrophy, there's only a treatment for one," Bell said, referring to retinitis pigmentosa (RP). "It felt like finding a needle in a haystack to be eligible."

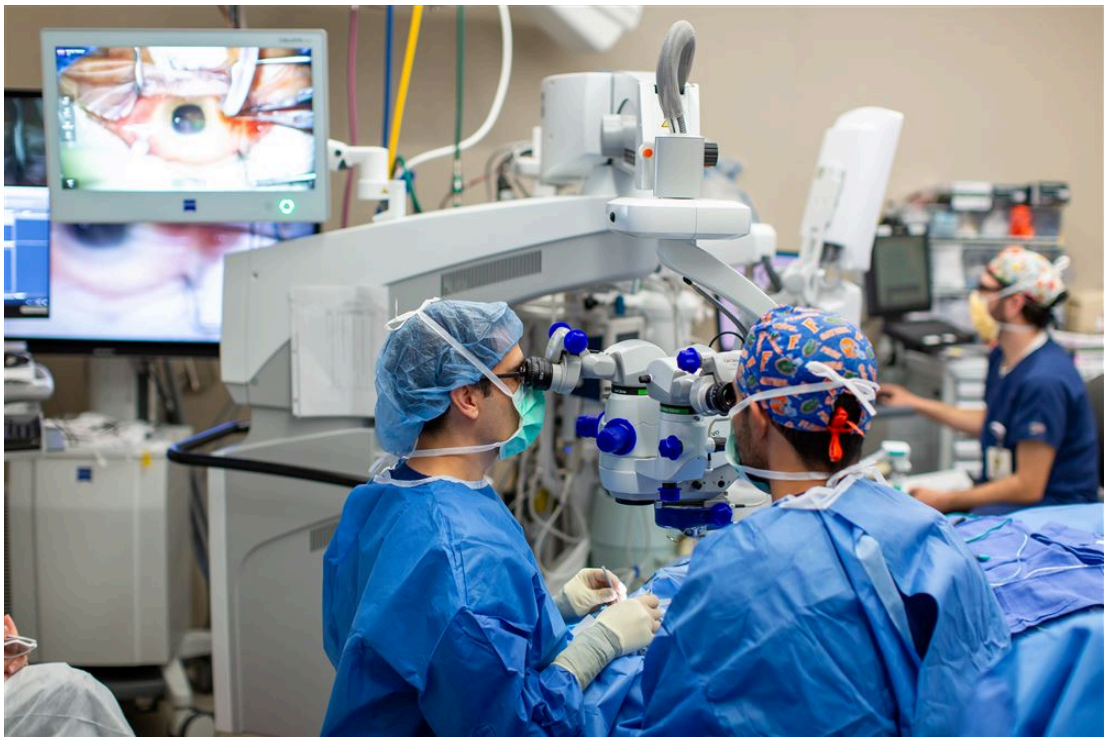
For many with RP, it can feel exceedingly rare. Constituting around 50% of cases of inherited retinal diseases, RP affects 1.5 million people worldwide, including an estimated 100,000 in the U.S.

Due to its rarity, Sarah Zambotti, an Allegheny Health Network optometrist, said she may only see two to three cases per year. If she

suspects a patient has RP, she refers them to a retinal specialist for further testing.

In Pittsburgh, UPMC is the only health system that currently offers Luxturna. Zambotti said that because AHN doesn't have a retinal specialist, they cannot surgically treat patients with RP.

"I would definitely love for [Luxturna] to become something we can offer here, just not currently, but hopefully in the future we're able to," she said. "Of course, we can still see patients and diagnose RP and get you referred to the right place to have that done, if it's something you're eligible for."



UPMC Vision Institute surgeons, including Joseph Martel, perform the Luxturna subretinal injection for Tammy Krents on June 20.
(UPMC)

The Luxturna treatment Krents received, which was approved by the FDA in December 2017, is administered by a subretinal injection and works by replacing the defective RPE65 gene with a functional copy. To be eligible, individuals must have a confirmed mutation in both copies of the RPE65 gene and adequate retinal cells.

Joseph Martel, a physician at the UPMC Vision Institute, said that Luxturna will "halt the progression of the accelerated degeneration of the retinal cells." While the treatment is not intended to reverse vision loss, some improvement is possible.

Although it will take three months to see results, Krents said that she has noticed small yet meaningful changes, such as being able to put toothpaste on her toothbrush for the first time since her surgery.

“For the first 10 days to two weeks, my vision was blurry, like when you get your eyes dilated. I couldn’t even see the TV remote,” she said. “Now, I can see better, but it’s still foggy outside and dark inside. It’s just going to take time.”

The sisters are hopeful that certain symptoms don’t progress: blockages in their central vision, a common symptom of RP, as well as resulting night blindness and light sensitivity.

“We're very sensitive to light, and it's always been that way,” Bell said. "Sometimes if the sun would be rising in the morning, I'd be having tears coming down my face because it's so bright.”

Krents said that when it’s bright outside, she can't see well for a few minutes, which has occurred for “as long as I can remember.” Prior to receiving Luxturna, she recalled having to use a flashlight to read menus at restaurants.

While both sisters knew that one day they would lose their vision, changes over the years confirmed that reality.

Krents had long known about her eye disease — she was diagnosed at age 3 — but it “really hit me,” she said, that day she no longer had her driver’s license. “[A]t that point I didn’t see a reason to lose my license because I still had my peripheral vision, but now, there’s no way I could drive.”

Bell’s vision has remained overall more stable than her sister’s, though she recently lost her driver's license too.

The sisters’ message: Cherish your vision, advocate for yourself and your loved ones and never lose hope.

“Just get out and see everything you can,” said Krents. “Don’t take your eyesight for granted because one day, you might not have it, and if there’s a treatment available, pursue it because it can change your quality of life.”

The two have started doing a “sisters weekend” annually in the summertime, Krents said. “Our kids joke about ‘the blind leading the

blind,' but it's important for us."

Outwardly showing support for one another, the sisters are often seen walking arm-in-arm. And Krents and Bell are hopeful both for themselves and others who might benefit from such medical advancements.

"The older we get, the more we support each other because we're the only ones who truly understand what we're going through," said Bell.

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