Gene therapy helps boys with 'Lorenzo's Oil' disease

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This undated photo provided by Boston Children's Hospital shows Brian and Brandon Rojas, of Dover Plains, New York, before the pair were diagnosed with adrenoleukodystrophy. Study results published Wednesday, Oct. 4, 2017, by the

New England Journal of Medicine showed an experimental gene therapy treatment seemed to help boys with the inherited nerve disease featured in the movie "Lorenzo's Oil." Although the news is bittersweet for the Rojas family, as Brian was able to get the treatment, while Brandon, whose disabilities from the disease had progressed further, was not. (Courtesy of the Rojas Family/Boston Children's Hospital)

Called adrenoleukodystrophy (ah-dree-no-lewk-oh-DIS-tro-fee), it almost exclusively strikes boys, who have only one copy of the X chromosome, where the gene that causes the disease lies. It affects about 1 in every 20,000 boys worldwide.

They are unable to make a protein that helps break down certain fatty acids, causing the acids to build up and damage nerves, muscles and the brain.

Their plight became better known after the 1992 movie about a couple who, desperate for a cure for their son, developed a treatment from olive and rapeseed oils that seemed to help him. The real life Lorenzo died in 2008 at age 30, having lived two decades longer than doctors predicted.

Lorenzo's oil did not turn out to be a cure. The only effective treatment is a stem cell transplant from a matched sibling, but only about 1 in 5 boys with the disease have such a donor.

The gene therapy tested a way for boys to serve as their own cell donors.

In this Aug. 29, 2017 photo provided by Boston Children's Hospital, Paul and Liliana Rojas speak to their sons, Brian, second from left, and Brandon, at Boston Children's Hospital. The young boys suffer from adrenoleukodystrophy, an inherited nerve disease featured in the movie "Lorenzo's Oil." Brian was able to benefit from an experimental treatment while the disease had progressed too far in Brandon to qualify for the gene therapy study. Study results were published Wednesday, Oct. 4 by the *New England Journal of Medicine* and discussed at the Child Neurology Society conference in Kansas City. (Katherine C. Cohen/Boston Children's Hospital via AP)

The U.S. Food and Drug Administration requires gene therapy participants to be monitored for 15 years, so these patients will continue to be studied, Williams said. Eight more patients are being added to the study as part of Bluebird's plans to seek approval of the therapy in the U.S. and Europe.

Brandon Rojas now needs a wheelchair and feeding tube. He had been a

healthy, normal child until suddenly starting to drool and have learning problems in school about the time he turned 7. He was diagnosed three years ago and doctors advised the couple to have Brian tested too. They were on a Make-A-Wish Foundation trip when the results came.

"We were driving to one of the Disney parks and had to pull over to get the call," Paul Rojas said. "Once we got off the phone, we both looked at each other and said we need to move forward now and save our younger son any way we can."

The couple has joined an effort to increase newborn screening for the disease, especially now that a therapy may soon be available to help—if it's tried soon enough.