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The Gene Editor-in-Chief: Dr. Kiran Musunuru's Race to Save a Baby With a Rare Genetic Disorder Affecting One in a Million

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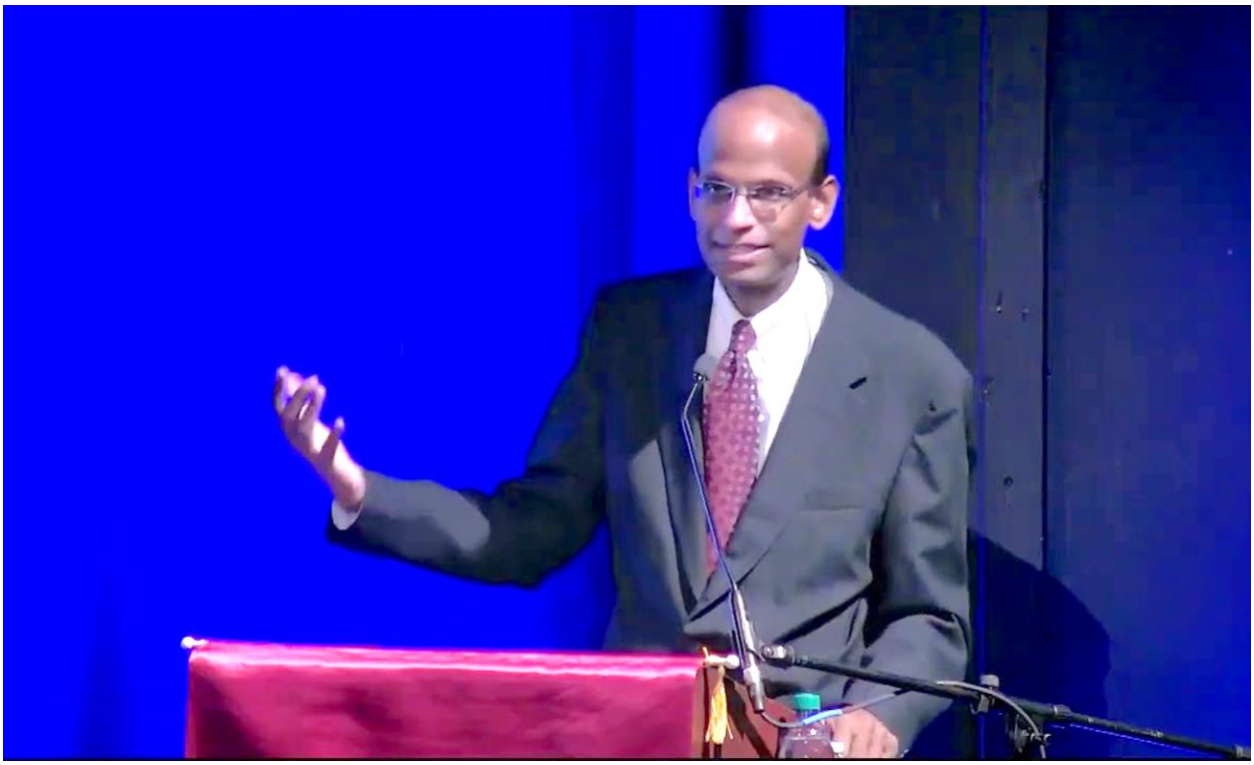
The Indian American researcher's race against time is said to stand as a testament to what becomes possible when scientific innovation meets human compassion.



In the predawn hours of a February morning in 2025, Dr. Kiran Musunuru stood anxiously in a hospital room at Children's Hospital of Philadelphia. Before him lay six-month-old KJ Muldoon, sleeping peacefully in the same crib that had been his home since birth. As a clear liquid flowed through an IV into the infant's tiny veins, Dr. Musunuru felt a conflicting surge of emotions.

“I was both excited and terrified,” recalls Dr. Musunuru, the Barry J. Gertz Professor for Translational Research at the University of Pennsylvania’s Perelman School of Medicine.

What KJ received that morning was unlike any treatment given to a patient before — a personalized gene-editing therapy created specifically to correct the single DNA letter in his genetic code that had caused a rare, life-threatening condition. That infusion would make medical history and potentially open the door to treating thousands of other genetic diseases that have long been considered untreatable.

The Midnight Email

The story began on August 8, 2024, when Dr. Musunuru received an urgent email from Dr. Rebecca Ahrens-Nicklas at Children’s Hospital of Philadelphia. A newborn boy had been diagnosed with CPS1 deficiency, a rare genetic disorder affecting just one in 1.3 million babies.

CPS1 deficiency prevents the body from properly processing ammonia, a toxic byproduct of protein metabolism. If left untreated, the condition can cause severe brain damage or death. Half of all babies with the disorder die within their first week of life. In her email, Dr. Ahrens-Nicklas’s asked if Dr. Musunuru can save the baby’s life considering that most babies don’t survive unless they get liver transplant at 3 years of age.

“At this point, the clock starts in my mind,” Dr. Musunuru said, as reported by The New York Times. “This is real life. This is not hypothetical.”

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Dr. Musunuru wasn’t a newcomer to the field of gene editing. As the Barry J. Gertz Professor for Translational Research in Penn’s Perelman School of Medicine, he had dedicated years to studying how gene editing could be used to treat genetic conditions. He is also a professor of Cardiology, Professor of Genetics and professor of Pediatrics at University of Pennsylvania school of medicine. Incidentally, Dr. Musunuru has designed gene therapy for the enzyme PCSK9 that decreases high cholesterol that is undergoing stage 2 clinical trials. If approved it will change the treatment of coronary heart disease. It’s like vaccine for heart attacks.

According to the Penn Medicine report, Dr. Musunuru and Dr. Ahrens-Nicklas had been collaborating since 2023 to study the feasibility of creating customized gene editing therapies for individual patients. Both are members of the NIH-funded Somatic Cell Genome Editing Consortium, which supports collaborative genome editing research.

Their focus had been on urea cycle disorders — precisely the category of disease affecting baby KJ. But developing a personalized treatment would typically take years, time the infant simply didn't have.

"Developing a gene editor to treat patients is a deliberate process that can take years," noted the New York Times report. "But KJ did not have years to wait — perhaps as few as six months before a mounting risk of severe brain damage or death."

The Race Against Time

What followed was an unprecedented scientific sprint. Dr. Musunuru assembled a team that included Dr. Fyodor Urnov at the University of California, Berkeley, and reached out to Danaher Corporation and other biotechnology companies to help produce the treatment.

Their approach used a refined CRISPR technique called "base editing," which precisely changes a single letter in the DNA sequence without cutting the DNA strand. This method, invented in David Liu's Harvard laboratory, reduces the risk of unintended genetic modifications.

The team worked around the clock. In Berkeley, as Dr. Urnov told The New York Times, "scientists burned a vat of midnight oil on this the size of San Francisco Bay." He added, "such speed to producing a clinic-grade CRISPR for a genetic disease has no precedent in our field. Not even close."

Dr. Liu himself described the timeline as "astounding."

"These steps traditionally take the better part of a decade, if not longer," he said.

Within just six months — lightning speed in the world of medical development — Musunuru's team had designed, tested, and manufactured a treatment ready for human use. The FDA expedited the regulatory approval process, recognizing the urgency of KJ's condition.

The Breakthrough

On February 25, 2025, KJ received his first infusion of the experimental therapy, followed by additional doses in March and April. The results were remarkable. Within two weeks of the first treatment, KJ was able to eat normal amounts of protein — something that would have been dangerous before. When he contracted typical childhood illnesses, which would normally have caused dangerous spikes in ammonia levels, he "sailed through them," according to Dr. Ahrens-Nicklas.

Now 9½ months old, KJ is growing well and meeting developmental milestones. His weight has increased from the 7th percentile to the 40th percentile for his age. While he still requires some medication, the dosage has been greatly reduced, and his doctors are preparing to discharge him from the hospital — allowing him to go home with his family for the first time in his life.

"We've been in the thick of this since KJ was born, and our whole world's been revolving around this little guy and his stay in the hospital," his father, Kyle Muldoon, told Penn Medicine News. "We're so excited to be able to finally be together at home so that KJ can be with his siblings, and we can finally take a deep breath."

Beyond One Patient

For Dr. Musunuru, saving KJ's life is just the beginning. The implications of this treatment extend far beyond one rare disorder.

"We want each and every patient to have the potential to experience the same results we saw in this first patient," Dr. Musunuru explained to Penn Medicine News. "And we hope that other academic investigators will replicate this method for many rare diseases and give many patients a fair shot at living a healthy life."

More than 30 million Americans suffer from one of over 7,000 rare genetic diseases. Many of these conditions are so uncommon that pharmaceutical companies have little financial incentive to develop treatments. But the approach used for KJ — essentially creating a molecular GPS that can be reprogrammed to target different genetic mutations — offers a template that could be adapted for countless other conditions.

The cost of development, while significant, was comparable to that of a liver transplant — the standard treatment for CPS1 deficiency in older children. As the process becomes more refined, Musunuru believes costs will decrease further.

"As we get better and better at making these therapies and shorten the time frame even more, economies of scale will kick in and I would expect the costs to come down," he told ABC News.

The Future of Personalized Medicine

The breakthrough represents what Dr. Peter Marks, who recently resigned from his position overseeing gene-therapy regulation at the FDA, called "one of the most potentially transformational technologies out there."

The methodology could eventually be applied to more common genetic disorders like sickle cell disease, cystic fibrosis, Huntington's disease, and muscular dystrophy.

"The promise of gene therapy that we've heard about for decades is coming to fruition," Dr. Musunuru said, "and it's going to utterly transform the way we approach medicine."

For the Muldoon family, the significance is measured in simpler terms — in milestones met and moments shared.

"Any time we see even the smallest milestone that he's meeting – like a little wave or rolling over – that's a big moment for us," Nicole Muldoon told ABC News.

As KJ prepares to leave the hospital and experience the world beyond his crib for the first time, Dr. Musunuru's race against time stands as a testament to what becomes possible when scientific innovation meets human compassion — and a reminder that behind every medical breakthrough are the very real lives hanging in the balance.