

Reanalyzing gene tests prompt new diagnoses in kids

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Lori Fullen sought genetic testing for her son Lorenzo after his epilepsy medications didn't stop the frequent seizures. Credit: UT Southwestern

Lori Fullen recalls waking to screams from her little boy's room. She remembers rushing in and seeing the terror in his eyes, wondering if he was having recurring nightmares.

She later realized the severity of the situation when her 7-year-old started shaking uncontrollably during an afternoon nap on the couch. "It was heartbreaking to see him suffer and there's nothing I could do," said Mrs. Fullen, her voice cracking with emotion.

Doctors diagnosed Lorenzo with nocturnal epilepsy and prescribed medications, but the treatment only slightly reduced the frequency of his seizures. He was up to eight pills a day by age 13 when UT Southwestern experts conducted a genetic test that yielded a life-changing discovery: The boy's epilepsy stemmed from the nicotine receptors in the brain—targets that could be controlled by nicotine patches.

More than three years into his new treatment, Lorenzo remains seizure-free.

"Science is an incredible thing," Mrs. Fullen said. "A genetic test changed Lorenzo's life, and it's exciting how quickly we're learning about other genes that cause all kinds of diseases."

A new study from UT Southwestern quantifies for the first time how quickly these rapid advancements in genomics may benefit patients. Research published in *JAMA Pediatrics* includes a five-year review of more than 300 epilepsy cases that showed about a third of children had a change in diagnosis based on new data. In some cases, the review helped doctors prescribe a more effective treatment.



Lorenzo Fullen has been seizure-free for more than three years after a genetic test pinpointed the cause of his epilepsy and helped doctors prescribe an

effective treatment. Credit: UT Southwestern

Based on these data, scientists are calling for parents to review gene tests done in children with epilepsy at least every two years to ensure their diagnoses and treatments are based on the latest discoveries. This finding is significant because the leading genome societies have suggested periodic checks but have not recommended how frequently these should be made.

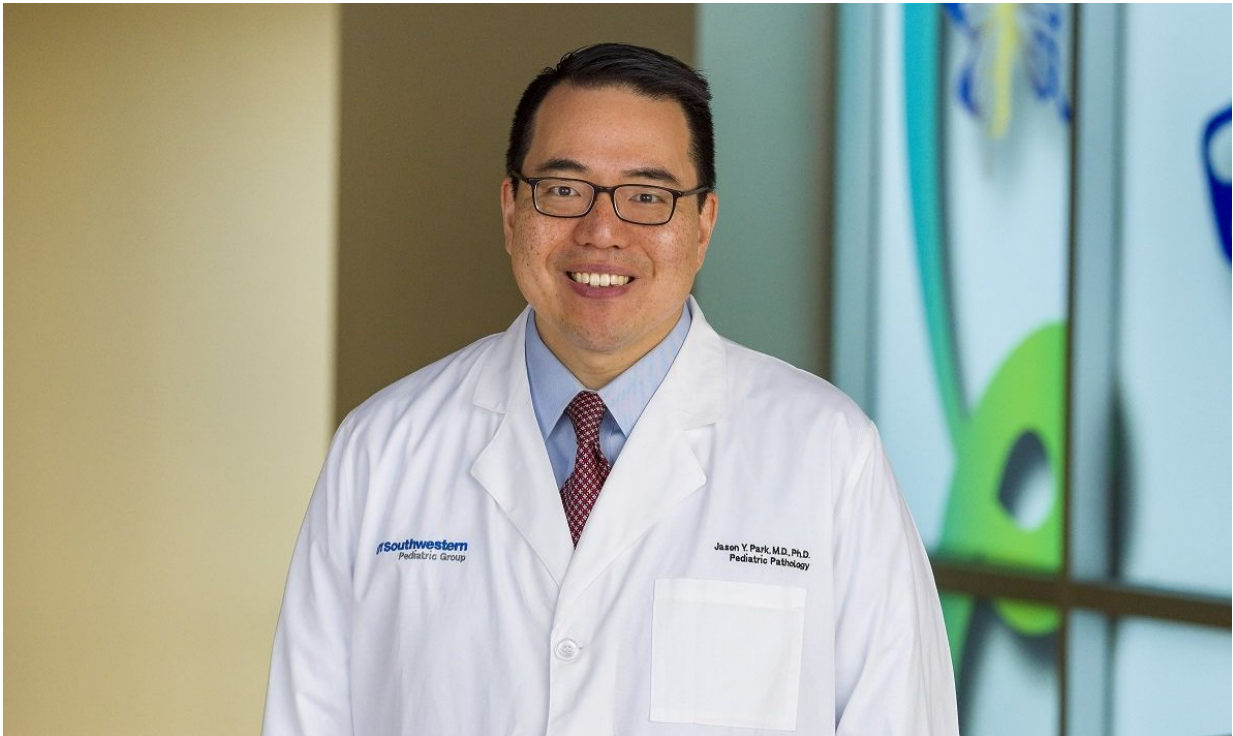
"This will start a national conversation for a number of diseases," said Dr. Jason Park, the study's senior author and a member of UT Southwestern's Eugene McDermott Center for Human Growth and Development. "Many of these [test results](#) come back inconclusive; we identify an unusual genetic change but we don't know what it means. That unknown status often changes within a few years, allowing these patients to be helped if the providers and families check the new data."

Much remains unknown about the human genome. One widely used database contains about 175,000 "variants of uncertain significance—approximately double the number of genetic variants believed to cause disease.

Still, the new research shows how quickly scientists are piecing the puzzle together. The study found that 32 percent of the patients received a diagnosis based on a new understanding of their genetic variant within the five-year window. In patients who received a genetic test result as recently as two years prior, researchers still found 25 percent had a disease-causing variant reclassified.

The study calls for further research to determine whether test reviews for other diseases and conditions have a similar limited shelf life.

"I was surprised how much change we saw," said Dr. Drew Thodeson, a child neurologist and epilepsy expert at UT Southwestern's Peter O'Donnell Jr. Brain Institute. "It's a testament to how quickly genetics is evolving and how much we are learning with new laboratory techniques and with more people getting their genes tested."



'This will start a national conversation for a number of diseases.' Dr. Jason Park, Associate Professor of Pathology. Credit: UT Southwestern

Dr. Thodeson recalls one of his patients who was initially placed on an ineffective medication after receiving test results that were inconclusive. He later prescribed a different treatment after [new data](#) clarified the nature of the patient's genetic variant.

"If we would have had that information before, then the patient never would have been given that initial prescription," Dr. Thodeson said. "It's up to both families and doctors to have [test](#) results periodically reviewed to make sure the best care is being given. Our knowledge of the [human genome](#) is growing so fast."

Mrs. Fullen's family is reaping the benefits of that knowledge every day as 16-year-old Lorenzo is sleeping restfully and performing better in school.

She still gets emotional recalling her son's screams in the middle of the night—an occasional waking nightmare she bares with the comfort of knowing Lorenzo can now live a normal life.

"I have my kid back," she said. "I don't have to watch him suffer anymore."

Provided by UT Southwestern Medical Center

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