Results of pediatric genomic epilepsy tests often reclassified

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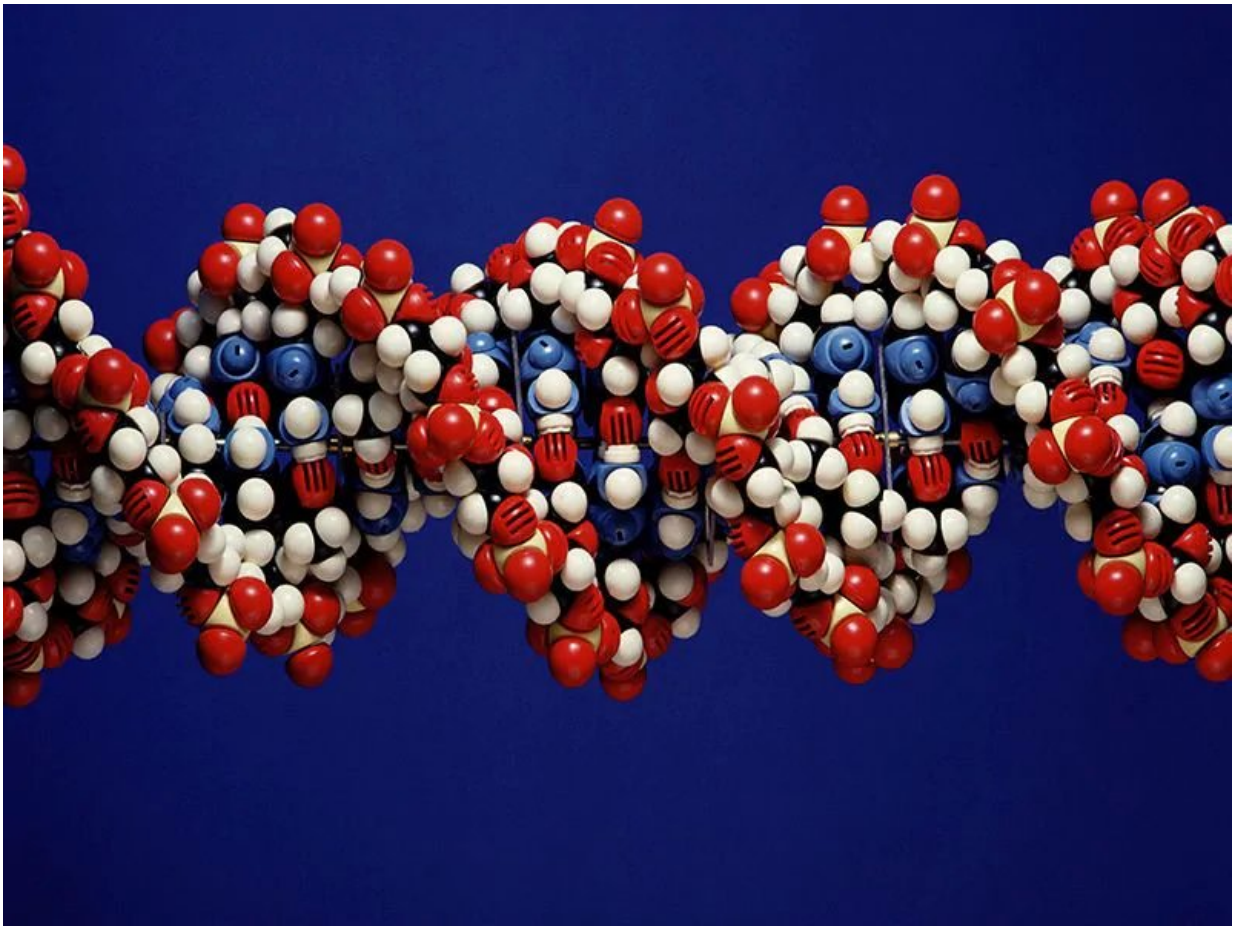

(HealthDay)—The interpretation of genomic epilepsy tests has evolved

rapidly in the last five years, and genetic variants identified in pediatric patients are often reclassified, according to research published online Nov. 5 in *JAMA Pediatrics*.

Jeffrey A. SoRelle, M.D., from the University of Texas Southwestern Medical Center in Dallas, and colleagues conducted a retrospective review and reinterpretation of genomic test results from July 1, 2012, to Aug. 31, 2015, for pediatric patients who previously underwent genomic epilepsy testing at a single facility. Patient reports from clinical genomic epilepsy tests were reviewed (309 patients), and all reported genetic variants were reinterpreted (185 patients) using 2015 consensus standards and guidelines.

The researchers found that based on the reported variants, there were 61 patients with and 124 patients without a genetic diagnosis (variant of uncertain significance [VUS] variants only). On reinterpretation of all 185 reported variants, 36.2 percent of patients had a change in variant classification; 31.3 percent of these experienced a change in diagnosis. During the five years of the study, results were reclassified for 31.1 percent of 61 patients with a genetic diagnosis and for 38.7 percent of 124 with undiagnosed conditions (VUS only). Upon review of genomic reports issued during the final two years of the study, 25 percent of 16 patients with a pathogenic or likely pathogenic variant and 26.8 percent of 41 patients with a VUS were reclassified.

"Patients with previous epilepsy genomic test results should have their test results reinterpreted at least every two years and before further genetic testing," the authors write.

One author disclosed having ties to genetic testing and interpretation services and having patent and pending patent applications for cancer diagnostics and therapeutics.

More information: Abstract/Full Text (subscription or payment may be required)