Genetic diagnosis may aid management of pediatric epilepsy

December 29 2021

For individuals with unexplained infantile or childhood-onset epilepsy,

genetic testing to establish a genetic diagnosis may impact medical care and prognosis, according to a study presented at the annual meeting of the American Epilepsy Society, held from Dec. 3 to 7 in Chicago.

Isabel Haviland, M.D., from Boston Children's Hospital, and colleagues assessed the individualized medical impact of a genetic diagnosis in a cohort of patients with infantile or childhood-onset epilepsy. The analysis included 152 individuals (46 percent female; median age of onset, 6 months) with a clinical diagnosis of genetic epilepsy, who underwent next-generation sequencing between 2012 and 2019.

The researchers found that a genetic diagnosis had a direct impact on medical management in at least one category for 72 percent of patients and in more than one category for 34 percent. In just under half of individuals (45 percent), treatment was impacted, including an impact on antiseizure medication choice (36 percent), use of disease-specific vitamin or metabolic treatments (7 percent), off-label use of medications (3 percent), and discussion of gene-specific clinical trials (10 percent). Additionally, care coordination was impacted in nearly half of individuals (48 percent), including surveillance for disease-associated features, disease-specific diagnostic testing, and referrals to specialists or disease-specific multidisciplinary clinics.

"A genetic diagnosis impacted medical management for nearly three out of four children in our study," Haviland said in a statement. "About two-thirds of pediatric epilepsy is unexplained, and genetic testing should be offered in these cases because of the significant potential impact on management, including on clinical treatment and eligibility for clinical studies."

More information: More information may be found here.