Our patient presented at 3 years of age with delayed language development. He was noted to have poor socialization, poor eye contact, inflexibility in new situations, and repetitive behaviors. He was administered the Autism Diagnostic Observation Schedule—Module 2 by a developmental psychologist specializing in autism when he was age 10 and was found to meet DSM-IV criteria for autistic disorder. His physical examination was notable for obesity (body mass index 31 kg/m²), macrocephaly, and poor eye contact but normal tone.

His mother developed cryptogenic epilepsy at age 11. His father reported learning disabilities and social anxiety but no formal neurologic diagnoses. There was no consanguinity (figure, A).

At age 15, he developed focal epilepsy with secondary generalization. EEG revealed left frontotemporal epileptiform activity (figure, B). MRI was remarkable for a left anterior temporal lobe nonenhancing fluid-attenuated inversion recovery hyperintensity (figure, C). He has been seizure-free for more than 1 year on a regimen of zonisamide and levetiracetam.
phenotype, as has been identified in other individuals with developmental delay.\(^6\)

Of note, there is an increased prevalence of epilepsy in individuals with autism. The prevalence of epilepsy in those with high-functioning autism is 9% and increases to 24% in those with comorbid intellectual disability.\(^7\) The mechanism for this increased prevalence is currently unclear but may be due to similar molecular mechanisms, such as neuronal hyperexcitability. Together, our data and previously published reports suggest an important role for SLC9A9 in a small subset of individuals with autism and epilepsy.

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