Abstract

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Ataxia and myoclonic epilepsy due to a heterozygous new mutation in KCNA2: proposal for a new channelopathy.

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Abstract

We have recently performed exome analysis in a 7 year boy who presented in infancy with an encephalopathy characterized by ataxia and myoclonic epilepsy. Parents were not consanguineous and there was no family history of the disease. Exome analysis did not show any pathogenic variants in genes known to be associated with seizures and/or ataxia in children, including all known human channelopathies. However, we have identified a mutation in KCNA2 that we believe to be responsible for the disease in our patient. This gene, which encodes a member of the potassium channel, voltage-gated, shaker-related subfamily, has not been previously described as a cause of disease in humans, but mutations of the orthologous gene in mice (Kcna2) are known to cause both ataxia and convulsions. The mutation is c.890C>A, leading to the amino acid substitution p.Arg297Gln, which involves the second of the critical arginines in the S4 voltage sensor. This mutation is characterized as pathogenic by five different prediction programs. RFLP analysis and Sanger sequencing confirmed the presence of the mutation in the patient, but not in his parents, characterizing it as de novo. We believe that this discovery characterizes a new channelopathy.

KEYWORDS: KCNA2; ataxia; epilepsy; exome; mutation; potassium channel

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