Neuropsychiatric Disease and Treatment

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LETTER

Response to the paper titled "Identification of a novel *CACNA1A* mutation in a Chinese family with autosomal recessive progressive myoclonic epilepsy"

This article was published in the following Dove Press journal:
Neuropsychiatric Disease and Treatment

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Dear editor

I have just read this article.[1] I have issues with the authors denoting the insertion of CAG as a new mutation. This is located within a polyglutamine tract, which is known to be polymorphic in this gene. The normal number of CAG repeats in exon 47 of the *CACNA1A* gene ranges from 4 to 18; over 21 CAG repeats lead to the neurological condition SCA6.[2] Indeed, in this paper, Lv et al[1] call the mutation insertion c.6975_6976insCAG, that is, the insertion of one CAG codon; however, on the sequencing figure, the two affected members of the pedigree have 14 CAG repeats, whereas the control and unaffected descendant have 11 repeats. So, if they were going to call this a mutation, it would be an insertion of three CAG repeats. They also seem somewhat confused about the nature of this mutation, as in a later figure they suggest that the insertion of glutamine also causes deletion of alanine, which is not backed up by the data given on their sequencing figure, showing an in-frame insertion of CAG, which would not delete the following alanine residue.[1]

This is a known polymorphism and, therefore, cannot be attributed to be the cause of disease in these patients.

Disclosure

The author reports no conflicts of interest in this communication.

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http://dx.doi.org/10.2147/NDT.S179068