BioNanoScience 2017 vol.7 N2, pages 265-268

Testing Genes Implicated in the Novel Case of Familial Hemiplegic Migraine

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Abstract

© 2016, Springer Science+Business Media New York.Familial hemiplegic migraine (FHM) is a rare monogenetic form of migraine associated with aura and motor dysfunctions. Molecular mechanisms of FHM still remain unknown; however, genetic predisposition to the disease has been suggested. We report several cases of FHM diagnosed in three generations of ethnic Tatars, Russian Federation. Apart from clear family character, patients presented with typical features of FHM including aura and ataxia. In order to determine disease genetic markers in these cases, we sequenced several genes (CACNA1A, ATP1A2, and SCN1A) previously reported in FHM. Sequence analysis demonstrated absence of the previously identified mutations in genes studied. This is the first genetic study of FHM in the Tatar population. The lack of known FHM associated mutation in these patients suggests presence of yet unidentified mutations in some other genes. Further studies including full genome sequencing are needed to clarify the genetic background of FHM cases in the Tatar population.

http://dx.doi.org/10.1007/s12668-016-0314-x

Keywords

CACNA1A, ATP1A2, SCN1A genes, Familial hemiplegic migraine (FHM), Mutations

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