BioNanoScience 2017 vol.7 N2, pages 265-268

Testing Genes Implicated in the Novel Case of Familial Hemiplegic Migraine

Khaiboullina S., Mendelevich E., Martynova E., Davidyuk Y., Giniatullin R., Rizvanov A.
Kazan Federal University, 420008, Kremlevskaya 18, Kazan, Russia

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

References

[1] van den Maagdenberg, A. M., et al. (2007). Migraine: gene mutations and functional consequences. Current Opinion in Neurology, 20(3), 299–305.

[2] Russell, M. B., & Ducros, A. (2011). Sporadic and familial hemiplegic migraine: pathophysiological mechanisms, clinical characteristics, diagnosis, and management. Lancet Neurology, 10(5), 457–470.

[3] Montagna, P. (2000). Molecular genetics of migraine headaches: a review. Cephalalgia, 20(1), 3–14.

[4] Stam, A. H., et al. (2008). CACNA1A R1347Q: a frequent recurrent mutation in hemiplegic migraine. Clinical Genetics, 74(5), 481–485.

[5] Vanmolkot, K. R., et al. (2003). Novel mutations in the Na+, K + −ATPase pump gene ATP1A2 associated with familial hemiplegic migraine and benign familial infantile convulsions. Annals of Neurology, 54(3), 360–366.

[6] Castro, M. J., et al. (2007). Recurrent ATP1A2 mutations in Portuguese families with familial hemiplegic migraine. Journal of Human Genetics, 52(12), 990–998.

[7] Bassi, M. T., et al. (2004). A novel mutation in the ATP1A2 gene causes alternating hemiplegia of childhood. Journal of Medical Genetics, 41(8), 621–628.

[8] Swoboda, K. J., et al. (2004). Alternating hemiplegia of childhood or familial hemiplegic migraine? A novel ATP1A2 mutation. Annals of Neurology, 55(6), 884–887.

[9] Fernandez, D. M., et al. (2008). A novel ATP1A2 gene mutation in an Irish familial hemiplegic migraine kindred. Headache, 48(1), 101–108.

[10] Lossin, C. (2009). A catalog of SCN1A variants. Brain and Development, 31(2), 114–130.

[11] Castro, M. J., et al. (2009). First mutation in the voltage-gated Nav1.1 subunit gene SCN1A with co-occurring familial hemiplegic migraine and epilepsy. Cephalalgia, 29(3), 308–313.

[12] Cestele, S., et al. (2013). Divergent effects of the T1174S SCN1A mutation associated with seizures and hemiplegic migraine. Epilepsia, 54(5), 927–935.

[13] Gargus, J. J., & Tournay, A. (2007). Novel mutation confirms seizure locus SCN1A is also familial hemiplegic migraine locus FHM3. Pediatric Neurology, 37(6), 407–410.

[14] Dichgans, M., et al. (2005). Mutation in the neuronal voltage-gated sodium channel SCN1A in familial hemiplegic migraine. Lancet, 366(9483), 371–377.

[15] Celebisoy, N., et al. (2008). Migrainous vertigo: clinical, oculographic and posturographic findings. Cephalalgia, 28(1), 72–77.

[16] Ophoff, R. A., et al. (1996). Familial hemiplegic migraine and episodic ataxia type-2 are caused by mutations in the Ca2+ channel gene CACNL1A4. Cell, 87(3), 543–552.

[17] Wada, T., et al. (2002). Wide clinical variability in a family with a CACNA1A T666m mutation: hemiplegic migraine, coma, and progressive ataxia. Pediatric Neurology, 26(1), 47–50.

[18] Jen, J. C., et al. (2007). Prolonged hemiplegic episodes in children due to mutations in ATP1A2. Journal of Neurology Neurosurgery and Psychiatry, 78(5), 523–526.

[19] Christensen, A. F., et al. (2016). The influence of genetic constitution on migraine drug responses. Cephalalgia, 36(7), 624–639.

[20] LaBianca, S., et al. (2015). Familial hemiplegic migraine and recurrent episodes of psychosis: a case report. Headache, 55(7), 1004–1007.