عنوان مقاله:

Association Study of Sequence Variants in Voltage-gated Ca2+ Channel Subunit Alpha-1C and Autism Spectrum Disorders

محل انتشار:

مجله گزارش های بیوشیمی و زیست شناسی مولکولی, دوره 8, شماره 1 (سال: 1398)

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خلاصه مقاله:

Background: Autism spectrum disorders (ASDs) (MIM 209850) are a group of distinct neurodevelopmental disorders characterized by impaired social interactions and communication abilities and abnormal repetitive activities. Many genetic variants have been shown to be associated with ASD. Channelopathies are among putative culprits in the pathogenesis of many neurodevelopmental disorders, including autism. The calcium channel, voltage-dependent, L type, alpha 1C subunit gene (CACNA1C) encodes an alpha-1 subunit of a voltage-dependent calcium channel. Genetic variants within this gene have been associated with psychiatric disorders including Autism Spectrum Disorders (ASD). Our aim was to determine whether the SNPs rs1006737, rs4765905, and rs4765913 were associated with ASD in an Iranian population. Methods: In the present case-control study we investigated the associations of rs1006737, rs4765905, and rs4765913 polymorphisms within CACNA1C and the risk of ASD in a population of 529 Iranian ASD patients and 480 age, gender, and ethnicity-matched healthy subjects. Results: None of these SNPs were associated with ASD risk in the assessed population. Although previous studies have shown an association between these polymorphisms and psychiatric disorders and an association between rs4765905 and ASD, we did not replicate those results in our study. Conclusions: Our data indicate that these CACNA1C variants are

.not involved in the pathogenesis of ASD in the Iranian population

کلمات کلیدی:

.Autism Spectrum Disorder, Channelopathy, polymorphism