



Category: Clinical Genomics

Genetic effect of monoamine oxidase B (MAOB) gene on ASD associated behavior phenotypes

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Abstract

Autism spectrum disorder (ASD) is a male predominance, behaviorally defined neurodevelopmental disorder which is characterized by impairment in social communication and restricted and repetitive activities. Abnormalities in serotonergic function play a major role in ASD pathophysiology. Monoamine oxidases, encoded by two X-chromosomal genes MAOA and MAOB regulate the serotonergic function by the degradation of serotonin and other biological amines. Therefore, the objective of present study is to investigate genetic correlation of MAOB markers with the severity of specific behavioral traits as scored by Childhood Autism Rating Scale (CARS) has been examined as quantitative trait (QT) analysis using IBM-SPSS program. A total of 225 ASD patients (190 male and 35 female) were recruited after psychometric evaluation done by DSM-IV-TR/DSM-5 criteria and assessment by CARS. Genotyping carried by PCR/RFLP/sequencing methods, and population were found in Hardy-Weinberg equilibrium. The outcome of the QT analysis indicating the increased score in overall CARS were associated with G and C allele of MAOB marker rs3027449 (p-value: 0.03) and rs1040399 (p-value: 0.01), respectively in male ASD children. In addition to this, major alleles of studied polymorphisms of gene were found to be statistically associated with the higher impairment in social communication domain only in male ASD children. Overall outcome of the study suggests likely involvement of MAOB with ASD in a gender-specific manner with the severity in behavior phenotypes. Considering the cumulative impact of these markers in regulating the severity of the behavioral symptoms of ASD, it is likely that MAOB gene is associated with the disorder.

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