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Research article

# Association of cystathionine beta synthase gene polymorphism with cognitive disorders in autistic children

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**Key words:** Autism; CBS; Homocystiene; G573A (rs73906420), CBS C699T (rs234706).

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**Abstract**

Folate, methionine and trans-sulfuration pathways and enzymes` are playing an important role in the pathophysiology of autism. Cystathionine beta synthetase (CBS) is a key enzyme of these pathways that associated with a lot of diseases such as brain atrophy and worsening neurological impairment in various central nervous system (CNS) disorders. CBS gene polymorphisms have been reported as a risk factor for neurodevelopment disorders and psychiatric disease. **Aim:** Hence the present study was designed to investigate the relationship between CBS gene polymorphisms from one side, and autism and the autistic behavior from another side. **Methods:** we sequenced the DNA fragment between exon 8 and exon 10 in CBS gene by using the polymerase chain reaction followed by direct sequencing methods in 40 autistic and 40 control children. **Results:** We found two polymorphisms CBS C699T (rs234706) and G573A (rs73906420). The frequency distribution of mutant and compound genotypes allele (T/T and C/T+T/T) of CBS C699T (rs234706) were (27.5%) and (52.5%) in the autism patients, respectively with a significantly higher association in autistic children; compared to controls (p=0.003 and 0.043). Also C/T showed significantly least frequency associated with sleep disorders and GIT disorders (p=0.016 and 0.001). No significant association was found between CBS genotypes and severity of the autism disorders. G573A (rs73906420) polymorphism was observed only in two autistic patients. **Conclusion:** This study demonstrates a role for CBS (C699T) polymorphism in sleep and GIT disorders and provides further support to the idea that CBS (C699T) gene polymorphism increased risk for autism spectrum disorders (ASD).