J Cell Mol Med. 2009 Oct;13(10):4229-38.

One carbon metabolism disturbances and the C677T MTHFR gene polymorphism in children with autism spectrum disorders.

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Source

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Abstract

Autism spectrum disorders (ASDs), which include the prototypic autistic disorder (AD), Asperger's syndrome (AS) and pervasive developmental disorders not otherwise specified (PDD-NOS), are complex neurodevelopmental conditions of unknown aetiology. The current study investigated the metabolites in the methionine cycle, the transsulphuration pathway, folate, vitamin B(12) and the C677T polymorphism of the MTHFR gene in three groups of children diagnosed with AD (n= 15), AS (n= 5) and PDD-NOS (n= 19) and their age- and sex-matched controls (n= 25). No metabolic disturbances were seen in the AS patients, while in the AD and PDD-NOS groups, lower plasma levels of methionine (P= 0.01 and P= 0.03, respectively) and alpha-aminobutyrate were observed (P= 0.01 and P= 0.001, respectively). Only in the AD group, plasma cysteine (P= 0.02) and total blood glutathione (P= 0.02) were found to be reduced. Although there was a trend towards lower levels of serine, glycine, N, N-dimethylglycine in AD patients, the plasma levels of these metabolites as well as the levels of homocysteine and cystathionine were not statistically different in any of the ASDs groups. The serum levels of vitamin B(12) and folate were in the normal range. The results of the MTHFR gene analysis showed a normal distribution of the C677T polymorphism in children with ASDs, but the frequency of the 677T allele was slightly more prevalent in AD patients. Our study indicates a possible role for the alterations in one carbon metabolism in the pathophysiology of ASDs and provides, for the first time, preliminary evidence for metabolic and genetic differences between clinical subtypes of ASDs.

PMID:

19267885

[PubMed - indexed for MEDLINE]