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[Clin Neurol Neurosurg](#). 2013 Sep;115(9):1693-6. doi: 10.1016/j.clineuro.2013.03.015.

Epub 2013 May 6.

# Association of methylenetetrahydrofolate reductase polymorphisms with susceptibility to Alzheimer's disease

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PMID: 23659764 DOI: [10.1016/j.clineuro.2013.03.015](https://doi.org/10.1016/j.clineuro.2013.03.015)

## Abstract

**Background:** Genetic risk factors play an important role in the pathogenesis of Alzheimer's disease (AD). In this case-control study, we examined the C677T and A1298C polymorphisms in the methylenetetrahydrofolate reductase (MTHFR) gene and their correlation with this pathology.

**Objective:** To verify the association between MTHFR C677T and A1298C polymorphisms and Alzheimer's disease.

**Method:** This work was conducted as a case-control study. Cases consisted of thirty-eight patients and 100 individuals without dementia constituted the control group. Genotyping of MTHFR polymorphisms was performed on patients and controls.

**Result:** Genetic analyses did not indicate a significant association between the MTHFR C677T mutation and AD (C/T: 63.15% versus 39%,  $p=0.087$ ). However, the genotype prevalence of the missense variant MTHFR A1298C was significantly different between patients and controls (A/C: 55% versus 7%,  $p<10^{-3}$ ). Our data suggest an association between the MTHFR A1298C mutation and AD; however, the MTHFR C677T mutation did not contribute to susceptibility for AD.

**Conclusion:** The MTHFR A1298C polymorphism is a possible risk factor for Alzheimer's disease.

**Keywords:** Alzheimer's disease; Human; MTHFR protein; Risk factor.

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