# Association of blood homocysteine levels with C677T and A1298C MTHFR gene polymorphisms in patients with NASH

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**Background and Aims:** It has been reported that increasing the level of homocysteine may affect the metabolism of lipids in the liver cells and provoke the development of fatty infiltration of hepatocytes. Frequent genetic mutations in enzymes that take part in folate metabolism can lead to increased levels of homocysteine in plasma.
The purpose of our work was to investigate the connection between the C677T and A1298C polymorphisms of the methylenetetrahydrofolate reductase (MTHFR) gene containing homocysteine plasma in patients with non-alcoholic steatohepatitis (NASH).

**Method:** We examined 86 patients with NASH. The control group consisted of 40 practically healthy persons. The determination of homocysteine was carried out by ELISA, the determination of allelic polymorphism was carried out by PCR with the detection of results by hybridization-fluorescence method in real-time.

**Results:** The frequency of the C677C, C677T and T677T genotypes of the MTHFR gene among patients with NASH and the control group was 53,5% 31,4%, 15,1% and 50%, 35%, 15% respectively. The frequency of A1298A, A1298C, C1298C genotypes of patients with NASH and the control group was 55,8%, 30,2%, 14% and 50%, 42,5%, 7,5% respectively. The plasma serum homocysteine level was higher in NASH patients as compared to control subjects (18.4±3.72 μmol/l, and 9.7±0.47 μmol/l ($p < 0.05$). The concentration of homocysteine in blood plasma in patients with C677C, C677T and T677T genotypes of the MTHFR gene was 14.8±2.27, 19.7±2.67 and 25.9±2.98 μmol/l, respectively ($p < 0.05$). The concentration of homocysteine in blood plasma in patients with A1298A, A1298C, C1298C genotypes was 18.8±2.39, 16.7±1.99 and 17.3±2.55 μmol/l, respectively ($p > 0.05$). We detected a reliable association between the frequency of genotypes for the C677T polymorphism of the MTHFR gene depending on the content of homocysteine. The T677T genotype carriers had a significantly higher homocysteine concentration compared to carriers of the C677T and C677C genotypes. We did not reveal a reliable association of plasma homocysteine with A1298C polymorphism of the MTHFR gene.

**Conclusion:** Patients with NASH, which were homozygous for the T677T genotype of the MTHFR gene, had a significantly higher homocysteine plasma level. The MTHFR C677T polymorphisms may be genetic risk factors for the development of NASH.

**Key words: homocysteine, non-alcoholic steatohepatitis, methylenetetrahydrofolate reductase**

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