Association of gene polymorphism MTHFR C677T and MTHFR A1298C with atrial fibrillation

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Introduction: MTHFR C677T and MTHFR A1298C polymorphisms are associated with hyperhomocysteinemia that results in prothrombogenic and atherogenic effect and could influence atrial fibrillation (AF) onset.[1,2] The goal of our study is to investigate relationship between MTHFR gene polymorphisms and AF.

Patients and Methods: We included 55 patients (31M, 23W) with AF. To all patients MTHFR C677T and MTHFR A1298C polymorphisms were determined, routine laboratory tests were done, transthoracic echocardiography was performed, body mass index was determined.

Results: In analysis of MTHFR C677T polymorphisms (**Figure 1**), there were 20 patients (36,4%) with healthy genotype (without present mutation) CC (cytosine–cytosine), 29 patients (52,7%) heterozygous CT (cytosine-thymine), and 6 patients (10,9%) with homozygous mutation TT (thymine-thymine). In analysis of MTHFR A1298C (**Figure 2**) there were 24 patients (43,6%) with healthy genotype AA (adenine-adenine), 23 patients (41,8%) heterozygous AC (adenine-cytosine), and 8 patients (14,5%) with homozygous mutation CC. Average weight of patients was 91.1±15.47kg, height 174.8±9.15 cm, and determined body mass index 29.54±3.68kg/m². There were no differences in left atrium diameter in different genotype groups of patients.

Conclusion: While incidence of MTHFR C677T homozygous mutation TT was similar in our group of patients with AF as in general population in our geographic region, the incidence of MTHFR C677T heterozygous mutation (52.7%) was significantly higher than the incidence of general population; approximately 20-40% of Caucasian.[3] MTHFR A1298C homozygous mutation CC incidence in our group of patients with AF (14.5%) was higher than in European population (7-12%). Pathological MTHFR C677T and MTHFR A1298C polymorphisms distribution in our group of patients with AF, that includes high incidence of heterozygous mutation and higher incidence of homozygous mutation than in general population, could indicate association of pathological MTHFR polymorphisms with AF onset.

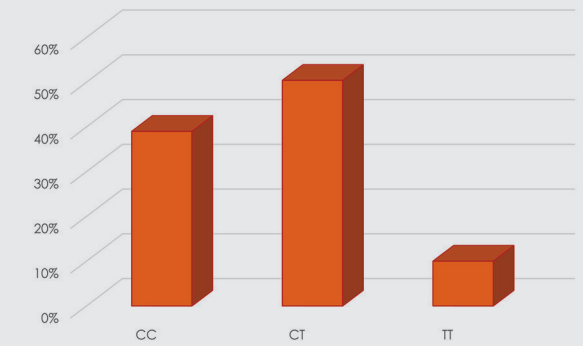

FIGURE 1. MTHFR C677T polymorphisms distribution.
CC = cytosine–cytosine, CT = cytosine-thymine, TT = thymine-thymine

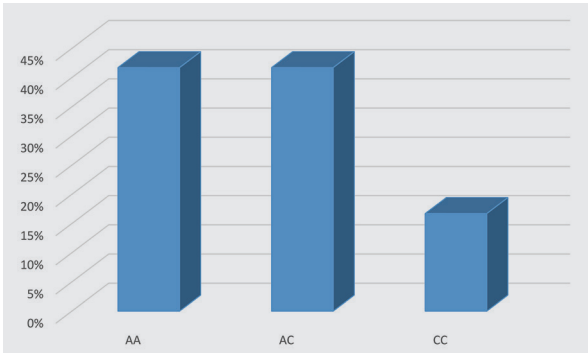

FIGURE 2. MTHFR A1298C polymorphisms distibution.
AA = adenine-adenine, AC = adenine-cytosine, CC = cytosine-cytosine

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