Endocrine Abstracts (2011) **26** P110

MTHFR mutations in female patients with autoimmune thyroiditis

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Introduction: Methylenetetrahydrofolate reductase (MTHFR) is a key regulatory enzyme involved in folate and homocysteine metabolism. The enzyme is encoded by a gene located on chromosome 1p36.3. MTHFR mutations frequently met in the population are C667T and A1298C. The impairment of homocysteine metabolism due to MTHFR gene polymorphism influences the risk for diseases such as CVS diseases, certain types of cancer and is associated with certain complications of pregnancy including chromosomal abnormalities, congenital malformations, neural tube defects: spina bifida, anencephaly, recurrent abortions, diseases of the placenta and preeclampsia. Autoimmune thyroiditis is a relatively common disorder found in the female population, which can influence fertility in a negative sense.

Purpose: The study aims to assess the risk of thrombophilia in patients with autoimmune thyroiditis.

Subjects and methods: There were assessed 50 patients aged 22 years to 39 years diagnosed with autoimmune thyroid disease, diagnosis established by means of thyroid ultrasound and by detection of ATPO and antithyroglobulin antibodies titre. The reasons for presentation for examination were primary and secondary infertility. Because the patients presented in their personal history spontaneous abortions there were also performed further evaluation of coagulation profile: V Leiden factor, C protein, S protein, detection of MTHFR mutations.

Results: In this group of patients with autoimmune thyroiditis 15 patients had MTHFR mutations: 3 patients with C667T homozygous mutation, 5 patients with heterozygous C667T mutation, 2 patients with A1298C homozygous mutation, 4 patients with heterozygous A1298C mutation and 1 patient with compound heterozygous C667T/A1298C.

Conclusions: MTHFR mutations are relatively frequent in the population (11%). Additional studies are needed to explain the association of MTHFR mutations with autoimmune thyroiditis, an increased prevalence of these mutations among the patients with autoimmune thyroid disease being observed.

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