Thrombophilic mutations among women with spontaneous abortions

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INTRODUCTION

Spontaneous abortion is the most common complication of pregnancy, affecting approximately 10-15% of clinically recognized pregnancies. Inherited thrombophilia as a genetic risk factor for thrombosis has been implicated in etiology of spontaneous abortions and may contribute to adverse pregnancy outcomes, such as intrauterine growth restriction, intrauterine fetal death, placental abruption and pre-eclampsia. Maternal thrombophilia augment the prothrombotic tendency of pregnancy leading to impaired implantation and placentation of the embryo, and to inadequate fetomaternal circulation later in pregnancy.

AIM OF THE STUDY

The aim of this study was to determine the association of 10 mutations in 8 thrombophilic genes: Factor V Leiden G1691A, Factor V A1299G, Factor II G20210A, Factor XIII G34C, PAI-I -675 4G/5G, FG-ß -455G/A, MTHFR C677T, MTHFR A1298C, MTR A2756G and MTRR A66G with spontaneous abortions.

MATERIALS AND METHODS

Simultaneous genotyping of the 10 studied thrombophilic mutations was done by multiplex SNaPshot analysis, followed by separation of the fragments by capillary electrophoresis on ABI 3130 Genetic Analyzer (Figure 1).

The prevalence of these thrombophilic mutations was studied in 95 women with history of fetal loss (40 of Macedonian and 55 of Albanian ethnicity) and 72 matched fertile controls (37 of Macedonian and 35 of Albanian ethnicity) .

RESULTS

Genotype and allele frequencies of the studied mutations were not statistically different between patients and controls and between the two ethnic groups.

However, we found higher percentage of a combination of three and more thrombophilic mutations in patients than in controls although statistically not significant (≥3 mutations: 92.6% vs. 84.7%, ≥4 mutations: 74.7% vs 69.4%, ≥5 mutations: 52.6% vs 43.1%, ≥6 mutations: 25.3% vs 18.1%, ≥7 mutations: 13.7 vs 8.3%, respectively) (Figure 2).

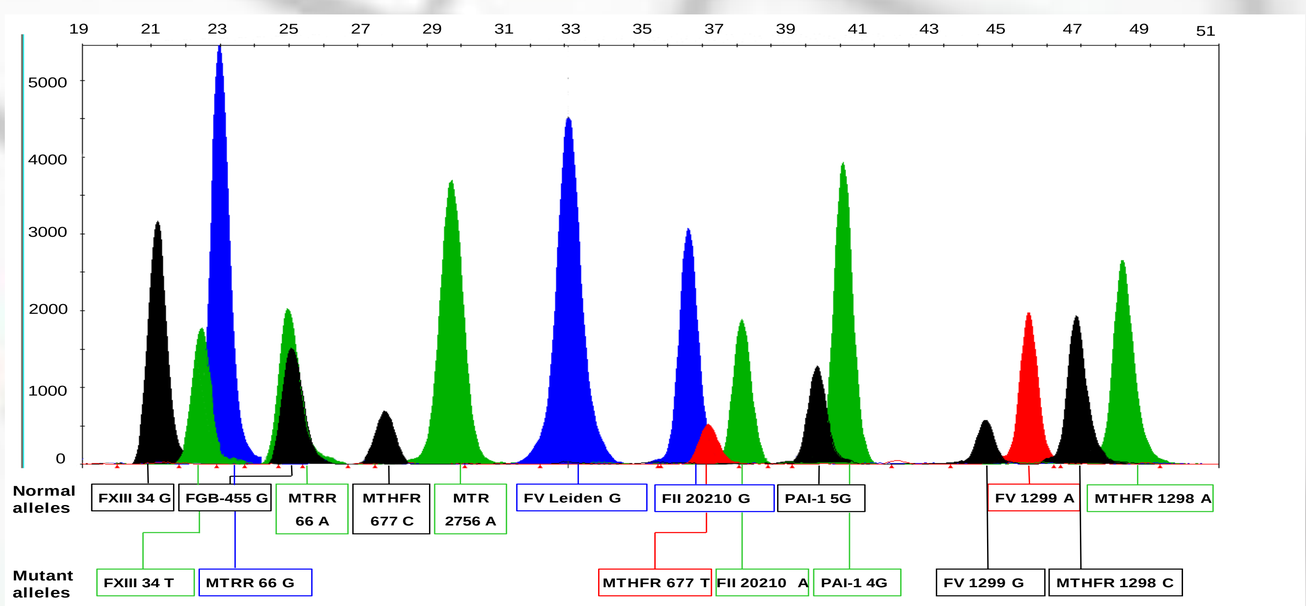

Figure 1. Electophoreogram from genemapper analysis of 10 mutations in 8 thrombofilia related genes.

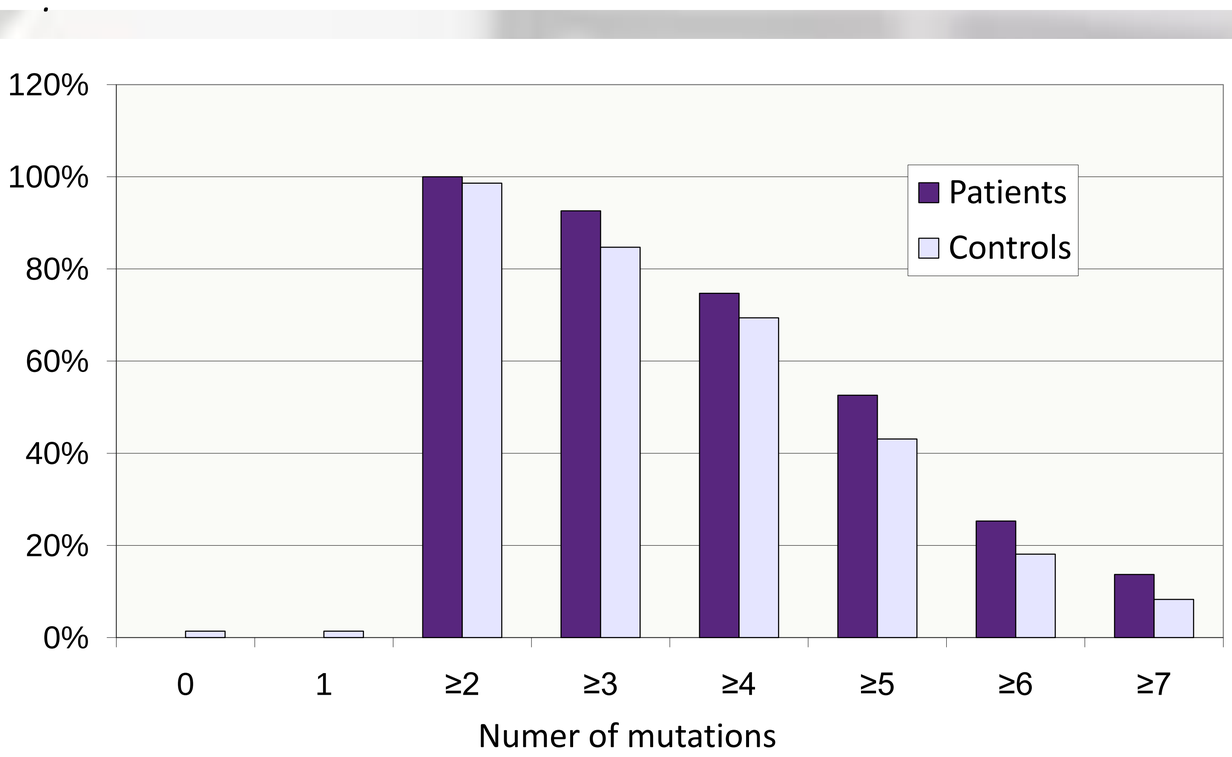

Figure 2. Distribution of co-occurence of the 10 studied thrombophilic mutations (Factor V Leiden G1691A, Factor V A1299G, Factor II G20210A, Factor XIII G34C, PAI-I -675 4G/5G, FG-ß-455G/A, MTHFRC677T, MTHFRA1298C, MTR A2756G and MTRR A66G) among patients with spontaneous abortions and fertile controls.

CONCLUSION

The results from this study showed higher percentage of a combination of three and more thrombophilic mutations among patients with spontaneous abortions indicating that thrombophilia might contribute to etiology of spontaneous abortions.

ACKNOWLEDGMENTS

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