ISSN 0972-3757
International Journal of
HUMAN GENETICS

 Int J Hum Genet, 16(1,2): 48-52 (2016)

Evaluation of Thrombophilic Genes in Recurrent Pregnancy Loss: A Case-control Study in Iranian Women

Mahmoud Shekari Khaniani[1,2], Fateme Afkhami[2], Fatemeh Abbasalizadeh[3] and Sima Mansoori Derakhshan[2]

[1]Department of Medical Genetics, Faculty of Medicine, Tabriz University of Medical Sciences, Tabriz, Iran
[2]Ebne Sina Medical Genetics Laboratory,
[3]Women Reproductive Health Research Center, Tabriz University of Medical Sciences, Tabriz, Iran

KEYWORDS FII G20210A. FVL G1691A. Miscarriage. MTHFR A1298C. MTHFR C677T. PCR–RFLP

ABSTRACT Although thrombophilia is associated with the etiology of recurrent miscarriage, the evaluation of specific inherited thrombophilic genes in women experiencing recurrent pregnancy loss remains controversial. This study compared the prevalence of four thrombophilic gene mutations among recurrent miscarriage and fertile control women. PCR–RFLP was performed to genotype four single nucleotide polymorphisms, *MTHFR* C677T, *MTHFR* A1298C, *FV* G1691A, and *FII* G20210A (causes of inherited thrombophilias) in both, case and control groups. About genotypes, there were statistically significant differences for mutant homozygote *MTHFR* A1298C genotype (P=0.01) between two cases and control groups, but other genotypes did not show any difference. About total allelic mutations, women experiencing recurrent pregnancy loss did not demonstrate more total mutations than control women (P=0.644). In conclusion, no association was observed between homozygous or heterozygous *MTHFR* C677T, *FV* G1691A, and *FII* G20210A mutants and pregnancy loss in Iranian women. However, the role of the homozygous *MTHFR* A1298C polymorphism should be investigated further.