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The association between thrombophilic gene mutations and recurrent pregnancy loss

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Abstract
Purpose To determine whether the Factor V (1691G/A), Factor V HR2 (4070A/G), Prothrombin (20210G/A), PAI-1 (-675 I/D, 5G/4G), ACE (intron 16 I/D), Factor VII (Gln353Arg), Factor XIII (Val34Leu), β-fibrinogen (-455G/A), Glycoprotein Ia (807C/T), tPA (intron 8 D/I) gene mutations could be risk factors for recurrent pregnancy loss (RPL).

Methods Genotyping of thrombophilic gene mutations were carried out by amplification Refractory Mutation System-PCR (ARMS-PCR) method after DNA extraction.
Results We found that the mutant allele frequencies of Factor V (1691G/A), Factor V HR2 (4070A/G), Prothrombin (20210G/A), PAI-1 (-675 I/D, 5G/4G), Factor XIII (Val34Leu) and β-fibrinogen (-455G/A) were more seen in the case group compared with the healthy control; However, the difference between the two group is not statistically significant ($p>0.05$). Whilst the mutant allele frequencies of other studied genes were lower in the case in comparison to the fertile control women ($p>0.05$).
Conclusion Taken together, our data has shown that the prevalence of thrombophilic gene mutations was similar in women with RPL and healthy controls. Therefore, it appears that further studies on large-scale population and other genetic variants will be needed to conclusively find candidate genes for RPL unknown etiology in the future.

Capsule It seems that our selected thrombophilic gene mutations is not significantly associated with recurrent pregnancy losses in North western Iranian women.

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Keywords Recurrent pregnancy loss · Thrombophilia · Thrombophilic gene mutations

Introduction

Recurrent Pregnancy Loss (RPL), defined as two or more consecutive pregnancy losses, is a serious reproductive problem, affecting 1–5 % of reproductive-age woman [29, 31]. There is a strong belief that RPL is a multifactorial condition that many factors affect such as chromosomal abnormalities, uterine anatomic malformation, endocrine dysfunction, immunologic factors, infections, and environmental factors [17, 18, 25]. However, the etiology of RPL remains unknown in ~50 % of cases [1].

In an attempt to find candidate genes for RPL, various genetic investigations have been performed on diverse genetic variants. The several studies focused on thrombophilic gene mutations that lead to maternally inherited thrombophilia and their association with RPL [12, 20, 40]. Of note, the possible

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