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Frequency of thrombophilic genetic polymorphisms among Saudi subjects compared with other populations

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Thrombophilic mutations increase the tendency toward thromboembolic disease. The aim of this study was to estimate the prevalence of the genetic variants related to thrombophilia among Saudis compared with other populations. Real-time polymerase chain reaction (PCR) genotyping was carried out to determine the polymorphic variants of factor V Leiden 1695G/A, prothrombin 20210G/A, plasmin activator inhibitor 1 4G/5G, methylene tetrahydrofolate reductase (MTHFR) 677C/T, MTHFR 1298A/C, and angiotensin-converting enzyme (ACE) insertion/deletion (I/D) among a representative sample of healthy Saudi subjects. Carriage rate for each of the mutant variants of factor V Leiden (FVL) and FII genes constituted 2% of the surveyed subjects giving an allele frequency of 0.01, homozygous forms of plasminogen activator inhibitor-1 (PAI-1) gene 4G/4G, MTHFR 677TT, 1298CC, and ACE DD were present among 7.7, 2.55, 7, and 51.8% of subjects with a mutant allele frequency of 0.4, 0.19, 0.29, and 0.73, respectively. This study showed that the Saudi population has a peculiar pattern regarding thrombophilic mutations that might warrant additional considerations for prophylaxis.

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