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Prevalence of genetic thrombophilic polymorphisms in the Sri Lankan population - implications for association study design and clinical genetic testing services

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Abstract

We investigated the prevalence of genotypes/alleles of single nucleotide polymorphisms (SNP) and haplotypes defined by them in three genes in which variations are associated with venous thromboembolism in 80 Sinhalese, 80 Sri Lankan Tamils and 80 Moors in the Sri Lankan population and compared the SNP data with that of other populations in Southern India and haplotype data with that of HapMap populations. The genes and polymorphisms investigated were Methylenetetrahydrofolate reductase (MTHFR) - 677C>T (rs1801133), 1298A>C (rs1801131), 1317T>C, 1793G>A (rs2274976); Factor V (F5) - 1691G>A (rs6025) and 4070A>G (rs1800595); and prothrombin (F2) - 20210G>A (rs1799963). The polymorphisms were genotyped using PCR/RFLP methods. The prevalence of the variant alleles of each polymorphism in the Sinhalese, Tamils, and Moors was MTHFR 677T: Sinhalese - 13%, Tamils - 9%, Moors - 9%. 1317T>C: Sinhalese - 0%; Tamils - 0%; Moors - 0%. 1793A: Sinhalese - 19%, Tamils - 19%, Moors - 19%. F5 1691A: Sinhalese - 2%, Tamils - 3%, Moors - 2%. 4070G: Sinhalese - 6%, Tamils - 5%, Moors - 8%. F2 20210A: Sinhalese - 0%, Tamils - 0%, Moors - 0%. The frequencies observed were similar to data from other South Indian populations; the haplotype data showed haplotypes unique to the Sri Lankan population when compared to HapMap populations. rs9651118 was identified as a SNP that splits the haplotypes harbouring the functionally significant 677T allele in the MTHFR gene. This data would be useful in planning genetic association studies in the Sri Lankan population and in deciding on which genetic variants should be tested in a clinical genetic testing service.