

## Identification of genetic variants associated with Deep Vein Thrombosis in the Sri Lanka population.

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**Background & Objectives:** Deep Vein Thrombosis (DVT) is a major preventable cause of morbidity and mortality worldwide. DVT is a multi-factorial disorder and occurs due to various acquired conditions, and inherited genetic risk factors. Genetic risk factors include mutations in the *Factor V*, *prothrombin* and *MTHFR* (5, 10- methylenetetrahydrofolate reductase) genes. Apart from these established genetic risk factors, studies have implicated various other candidate genes that might be involved in DVT pathogenesis. The aim of this study was to identify novel genetic variants associated with DVT in the Sri Lankan population.

**Method (s) and Results:** The study population comprised of 110 individuals diagnosed with DVT, who were referred to the Human Genetics Unit, University of Colombo, for genetic screening retrospectively following ethical clearance. Through literature search, globally studied genetic variants were identified, which were shown to be associated with DVT in various populations. Two gene variants were identified through literature review to be associated with DVT in South Asian populations, *CYP4V2* c.775C>A (rs13146272) and *Factor 5 (F5)* c.2573A>G (rs4524). A T-ARMS-PCR assay was designed and optimized to genotype extracted patient DNA. Genotyping results were validated by Sanger sequencing. For the *CYP4V2* variant, the ancestral (C) allele frequency was 0.3045 and variant (A) allele frequency was 0.6955 at Hardy Weinberg's equilibrium. For the *F5* variant, the allele frequency was 0.6651 and 0.3349 for ancestral (A) allele and variant (G) allele, respectively. The allele frequencies of both gene variants in the Sri Lankan population were consistent with other South Asian populations.

**Conclusions:** The optimized T-ARMS-PCR assay protocol that was developed in this study can be introduced to genotype the *CYP4V2* c.775C>A variant and *F5* c.2573A>G variant, in DVT patients. Our findings suggest that *CYP4V2* c.775C>A variant and *F5* c.2573A>G variant maybe new possible candidate genetic risk factors for DVT in the Sri Lankan population.

**Conflict of interest disclosure:** The authors declare no potential conflicts of interest, whether scientific, financial and personal.

**Keywords:** Deep Vein Thrombosis, Sri Lankan population, T-ARMS PCR, *CYP4V2*, *Factor 5*