PP 32 - Identification of genetic variants associated with deep vein thrombosis (DVT) in a cohort of patients affected with DVT in Sri Lanka.

K. L. J. Hasaranga[1*], N. Noordeen[1], T. K. Wetthasinghe[1], V. H. W. Dissanayake[1]

[1]Human Genetics Unit, Faculty of Medicine, University of Colombo, Colombo 08, Sri Lanka

Introduction: Genetic thrombophilias are known to be inherited and led to an enhanced tendency of vascular blood-clot formation. Deep Vein Thrombosis (DVT) is a similar thromboembolic manifestation that occurs due to various genetic & non-genetic etiologies. The aim of this study was to design and implement an assay to identify genetic variants associated with DVT in a cohort of patients in Sri Lanka.

Methods: A comprehensive literature review was conducted to identify genetic variants associated with Deep Vein Thrombosis. A Tetra-Amplification-Refractory-Mutation-System Allele Specific Polymerase Chain Reaction (T-ARMS-AS-PCR) was designed and optimized to genotype the extracted DNA samples of the study population who had given informed consent for such studies. The genotype results were validated by Sanger sequencing for the specificity.

Results: Total of 110 individuals were genotyped using the designed assay. Out of which, 14 (12.7%) were normal (CC) for the variant, while 39 (35.5%) were heterozygotes (CA) and 57 (51.8%) were homozygotes (AA) for CYP4V2 c.775C>A (rs13146272) variant. The allele frequency for the variant allele at Hardy Weinberg's equilibrium was 0.6955. Out of the 110 individuals, 37 (33.6%) were normal (AA) for F5 c.2573A>G (rs4524) variant with 73 (66.4%) heterozygotes (AG). No homozygotes (GG) were found for the F5 c.2573A>G variant. The allele frequencies were 0.6651 and 0.3349 for both ancestral allele and variant allele respectively.

Conclusions: Allele frequencies of both CYP4V2 and F5 variants were consistent with South Asian population allele frequencies, suggesting the reliability of our finding. This is a simple genotypic assay which can be used as an efficient, sensitive and specific molecular diagnostic screening test for thrombophilic genetic variants in Patients affected with DVT in the Sri Lankan population.

Keywords: DVT, T-ARMS PCR, CYP4V2, F5

*Email: hasaranga@hotmail.com