Abstract

Pulmonary thromboembolism (PE) is obstruction of pulmonary vessels with one or more thrombi. It carries a very high mortality. Hyperhomocysteinemia is a hereditary thrombophilic condition predisposes to venous thromboembolism (VTE) and PE. Mutations in methylene tetrahydrofolate reductase gene, which involved in homocysteine metabolism, cause hyperhomocysteinemia commonly in homozygous state. Heterozygous mutation with VTE is a rare entity.

We report a case of young Sri Lankan male presented with unprovoked right lower limb deep vein thrombosis complicated with moderate risk PE. He had hyperhomocyteinemia and vitamin B_{12} /folate deficiency. We observed the unique association of MTHFR 677 C>T mutation particularly in heterozygous state in this patient.

Hyperhomocysteinemia predisposes both arterial and venous thrombosis. Therefore, we would like to emphasize on the value of screening for hyperhomocysteinemia and related genetic mutations in young patients presenting with PE.