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Common Thrombophilic Mutations among Sickle Cell Disease Patients in the Western Province of Saudi Arabia

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ABSTRACT Sickle cell disease (SCD), linked to vascular thrombosis, is an autosomal recessive disorder. Among the various thrombophilic mutations, Factor V Leiden (FVL) G1691A, prothrombin (PRT) G20210A, and methylenetetrahydrofolate reductase (MTHFR) C677T stand out as genetic alterations linked to thrombotic consequences. Thus, the study aims to examine the prevalence of these mutations in SCD patients, utilizing the polymerase chain reaction-based technique. The study design is an observational study. The study results revealed that there are significant levels of the heterozygous form of FVL G1691A and PRT G20210A in the SCD patient population compared to control subjects. While, MTHFR C677T showed no statistical significance. None of the homozygous forms of the three mutations was statistically significant. However, the incidence of FVL, PRT, and MTHFR mutations was higher among female participants than male participants in the study. The high prevalence of these mutations suggests that they may be significant risk factors for the vascular complications in this population. However, further studies are required to validate the current outcomes.