Sickle-cell trait as a risk factor for unprovoked venous thromboembolism

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Background: Sickle-cell trait (SCT), characterized by the presence of a single mutated allele for hemoglobin S, is typically considered asymptomatic and benign. However, emerging evidence suggests a potential link between SCT and increased venous thromboembolism (VTE) risk, encompassing deep vein thrombosis (DVT) and pulmonary embolism (PE).

Case Presentation: We present a case of a 44-year-old African American male with SCT and obesity, without other significant risk factors, who developed pulmonary embolism. This occurred despite low scores on conventional risk assessment tools such as the Wells and Geneva scores and meeting the pulmonary embolism rule-out criteria (PERC). His clinical presentation included shortness of breath, chest pain, and a normal physical examination. Labs were normal except for elevated D-dimer levels. Subsequent imaging revealed extensive pulmonary emboli, leading to immediate anticoagulation therapy and plans for long-term management.

Discussion: This case underscores the limitations of current risk assessment models in patients with SCT, echoing findings from studies like that of Little et al. and contributions from Naik and Noubiap. These models, while robust, often overlook inherited risks like SCT. Our case suggests the necessity for a more nuanced approach to VTE risk assessment and management in SCT carriers, potentially including the use of alternate tools like D-dimer measurement and DASH scores. It also raises the question of considering SCT as a persistent risk factor in management strategies, particularly in decisions regarding indefinite anticoagulation.

Conclusion: This case report highlights the need for heightened awareness and reevaluation of risk assessment strategies in patients with SCT, considering the emerging evidence of their increased risk for unprovoked VTE. It also provides insight into the potential adaptations in management strategies for these patients.