Factor VII Deficiency: A Paradoxical Bleeding Disorder with Prothrombotic State

Introduction

Factor VII deficiency is a rare bleeding disorder which can vary in both its severity and presentation signs and symptoms. The common presentations are excessive bruising, hemarthrosis, or hematuria. Spontaneous thrombosis occurs in 3-4% of the factor VII deficiency patients. This case is an example of a 47 year old male who presented coronary artery thrombosis and progressive ischemic cardiomyopathy and was found to have factor VII deficiency.

Case Presentation

- A previously healthy 47 year old male with a past medical history of coronary artery disease status post a coronary artery stent placement after a myocardial infarction two years prior, hypertension, non-insulin dependent diabetes, and a transient ischemic attack. He has no history of smoking or alcohol use and presented to the hospital with progressive worsening shortness of breath.
- Echocardiogram showed a reduced ejection fraction of 25%. This ejection fraction is reduced from his prior admission two years ago when he was admitted for an acute myocardial infarction and underwent cardiac catheterization.
- It was noted during that admission that he was found to have an isolated elevated PT and INR of 1.7 and was not on any anticoagulation. During his current presentation, the patient’s INR was 2.1. With such an elevated INR, cardiac catheterization could not immediately be performed. Despite multiple trials of Vitamin K, his INR did not change.
- Further testing with coagulation mixing studies and factor activity testing revealed a factor VII activity level of 13% compatible with factor VII deficiency. In addition, no factor VII inhibitor was detected.
- The patient subsequently underwent cardiac catheterization after receiving recombinant factor VII and was found to have multi vessel disease as well as stent restenosis. No post procedural hemorrhagic events were noted.

Discussion

Factor VII is a vitamin K dependent factor produced by the liver. When it binds to tissue factor, its activation begins the intrinsic coagulation cascade leading to thrombin production. Deficiency results in excessive bleeding since thrombin production is impaired. Thrombosis associated with Factor VII is uncommon and the mechanism is not well understood. This case is a rare presentation of a young male with no personal or family history of atherosclerotic vascular disease or sudden cardiac death and no underlying liver disease who presents with recurrent thrombosis despite appearing hypocoagulable by in vitro coagulability testing. Therefore, having bleeding disorder does not necessarily rule out prothrombotic states and must still be considered. The management of such patients who are prothrombolic yet appear to be hypocoagulable is complicated as a balance needs to be maintained between risk of bleeding versus thrombus formation.

References