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Title	An elderly man with traumatic subdural hematoma and newly diagnosed inherited factor VII deficiency
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Abstract

Inherited deficiency in factors VII, XIII, XI, X, V, II are collectively known as rare inherited coagulation disorders (RICDs). Among them, the prevalence of factor VII deficiency ranked 4th with an incidence of 1 in 500,000[1]. However, local data and management guideline on RICDs were lacking. Our team would like to present a case with newly diagnosed inherited factor VII deficiency who suffered from internal haemorrhage.

A 86-year-old man was admitted in January 2024 for accidental fall with head injury. Computed tomography of brain (CTB) showed acute subdural hematoma over right temporoparietal region (0.4cm) and right frontal region (trace amount). There was also incidental finding of old lacunar infarcts at bilateral external capsules. He had known history of essential hypertension, hypercholesterolemia, lower urinary tract symptoms and chronic kidney disease. He was not on any antiplatelet or anticoagulant. He also denied use of over-the-counter medications or medications from family members. He did not have any history of bleeding. His Glasgow Coma Scale (GCS) remained 15/15 and limb power remained full during the in-patient stay.

The international normalised ratio (INR) on admission was prolonged to 2.3. Whereas the activated thromboplastin time (APTT) was normal. He received intravenous vitamin K1 injection and repeated plasma transfusions. However, the INR remained prolonged ranging from 1.8-2.3.

CTB was repeated 2 days later which revealed a new left subdural effusion of 0.4cm. 1:1 mixing study was arranged. The prolonged INR was corrected with the 1:1 mixing study, suggestive of factor deficiency. Subsequent factor level assessment revealed isolated factor VII deficiency of 12% (reference interval 50-150%).

Upon further enquiry, patient denied any parental consanguinity. He had 6 siblings all without major bleeding but all suffered from ischemic stroke. He received two doses of intravenous Factor VIIa injection at the dosage of 35microgram/kg, given 12 hours apart. No major side effect was encountered. Follow-up CTB showed resolution of the subdural effusion and hematoma. Patient was discharged uneventfully.

The human factor VII gene is located on chromosome 13 and the inheritance is autosomal recessive[2]. The correlation between factor VII level and bleeding tendency is poor[3, 4]. Common presentations of factor VII deficiency include excessive haemorrhage after invasive procedures, menorrhagia, mucosal or intramuscular bleeding[5]. Experts recommend a target of greater than 20% if a patient with known inherited factor VII deficiency with bleeding or before undergoing invasive intervention[1]. Recombinant factor VIIa, plasma transfusion and prothrombin complex concentrate (PCC) are all suitable choices.

Although the factor VII levels of the siblings of the patient could not be assessed, it was interesting to observe a high frequency of thrombosis among the family. Paradoxical arterial or venous thromboses in inherited factor VII deficiency patients have been reported (3-4%)[6]. Therefore, in addition to bleeding, physicians should also pay attention to any signs and symptoms suggestive of thromboembolism.

Our team would like to use this case to illustrate the diagnosis and management of factor VII deficiency. It is hoped that with the collaboration and sharing between local haematologists, we could manage RICDs better in the future.

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