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FROM GENE TO CLINIC: INSIGHTS INTO FACTOR VII DEFICIENCY

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ABSTRACT
FVII is synthesised in the liver in dependence on vitamin K. Therefore, the causes of acquired deficiency are vitamin K deficiency, hepatopathies, or the use of vitamin K antagonists. Rarely do other forms of acquired FVII deficits occur. Here we report a case of 22-year-old female with a history of menorrhagia and easy bruising on top of pain and swelling in her left shoulder. Laboratory tests showed decreased factor VII activity levels and an extended prothrombin time. She was treated with with factor VII replacement therapy, oral tranexamic acid during monthly menstrual cylces, amoxcillin, and hormonal OCPs. This case illustrates the variety of clinical signs and difficulties associated with diagnosing and treating a female patient's factor VII insufficiency.

Keywords: *Factor VII*

INTRODUCTION
FVII is synthesised in the liver in dependence on vitamin K. Therefore, the causes of acquired deficiency are vitamin K deficiency, hepatopathies, or the use of vitamin K antagonists. Rarely do other forms of acquired FVII deficits occur. Acquired bleeding problems are often the result of scarcity of many clotting factors. Acquired single defects of clotting factors are rare conditions. The most generally known is the factor X (FX) deficiency linked with amyloidosis (Choufani and Sanchorawala 2001). Bleeding or thrombosis are frequently linked to coagulation problems in neoplastic diseases. The most common alterations are thrombocytopenia (found in acute leukemias), thrombocytosis (found in myeloproliferative disease [MPD]), and thrombophilic state (found in lung and pancreatic tumors). Disseminated intravascular coagulation (found in promyelocytic leukemia and several solid tumors) is also common. These are well-known conditions, and the compassionate doctors are aware that they could develop. Nevertheless, there are additional disorders that are frequently overlooked or misdiagnosed that are linked to clotting abnormalities (Zili *et al.,* 2005). The reason for this is that certain defects, like FVII deficiency, may only be mild to moderate in nature and only manifest as a slight extension of the prothrombin time (PT). If the treating physician does not order specialized coagulation factor assays to look into the isolated prolongations of the PT, the proper diagnosis can go unnoticed.
Some of these illnesses are sometimes referred to as paraneoplastic syndromes. The definition, however, is incorrect because it is unclear if the action mediated by a known or unknown hormone-like substance—a need of a paraneoplastic syndrome—is always present.
Other than neoplastic diseases, sepsis or the development of antibodies are other conditions that can cause isolated FVII deficiency (Grish *et al.,* 1997).
In order to raise awareness of the issue, the current study aims to report a case of acquired FVII insufficiency. Given the finding that the occurrence of a single FVII deficiency may have significant clinical implications, this could have a significant clinical impact.

Case
A 22-year-old woman arrived with a history of menorrhagia and easy bruising on top of discomfort and swelling in her left shoulder. She also reported feeling exhausted and lightheaded. A physical

examination of the left shoulder revealed limited range of motion and discomfort. Laboratory tests showed decreased factor VII activity levels and an extended prothrombin time. Multiple subchondral cystic regions of erosions in the humeral head were found by imaging. The patient's symptoms were resolved after receiving conservative treatment with factor VII replacement therapy, oral tranexamic acid during monthly bleeding, amoxcillin, and hormonal OCPs. This case illustrates the variety of clinical signs and difficulties associated with diagnosing and treating a female patient's factor VII insufficiency. Her shoulder synovium was biopsied. Her histopathological examination showed villonodular synovitis.

DISCUSSION

Alexander et al. published the first description of factor VII deficiency (Alexander *et al.,* 1951). In nations where consanguineous marriage is more prevalent, frequency is higher. India has a dearth of case reports (Mahale *et al.,* 2010). Decreased biosynthesis or rapid clearance cause type 1 deficiencies; a malfunctioning molecule is the cause of type 2 abnormalities. over 100 mutations, the majority of which are missense (Cooper *et al.,* 1997). This missense mutations were located on chromosome 13. Liver disease, vitamin K antagonist medication, or vitamin K insufficiency can all result in acquired FVII deficiency. Lower levels of other vitamin K-dependent components are correlated with lower FVII levels under these circumstances. Compared to genetic deficit, acquired FVII deficiency is significantly more prevalent (Biron *et al.,* 1997).

The amount of FVII coagulant activity detected in plasma does not necessarily correspond with clinical bleeding and can vary greatly. Severe bleeding, which typically results from CNS hemorrhage as was the case in the index case, is associated with mortality. The majority of severe cases of FVII insufficiency are identified in children, frequently in the first six months of life. 60% to 70% of bleeding incidents in infants originate in the gastrointestinal system or central nervous system (Mariani *et al.,* 2004).

The mainstay of management for acute hemorrhage is the administration of FVII replacement therapy to address bleeding (Ingerslev and Kristensen, 1998). Generally speaking, levels above 10% are hemostatic; but, in the event of a serious bleeding episode, higher levels might be advised. With the exception of small bleeding episodes, repeat treatment may be required due to the short half-life of FVII (3–4 hours). Fresh frozen plasma is one of the treatment options, although it is the least efficient due to the volume needed to replace sufficient amounts of FVII. Factors II, IX, and X are present in prothrombin complex concentrates together with FVII.

There is a chance that these medications will cause thrombogenic side effects, especially if repeatedly administered. FVII concentrates are preferable over untreated plasma if they are available. The original purpose of recombinant activated FVII (rFVIIa) was to treat hemophiliac patients with factor inhibitors. Patients with congenital FVII deficiency may be treated with lower doses of this medication. For the majority of surgical procedures, maintaining FVII levels of at least 15–25% ensures sufficient hemostasis levels.

CONCLUSION

A rare abnormality known as isolated FVII deficiency appears to be linked to a number of morbid illnesses. The patients presenting with mild bleeding diathesis like easy bruisability should be investigated. Also, a mild prolongation of the prothrombin time should be thoroughly investigated, particularly in cases when the partial thromboplastin time and fibrinogen levels are normal.

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