

Factor VII Deficiency

Subjects: Genetics & Heredity

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Factor VII deficiency is a rare bleeding disorder that varies in severity among affected individuals.

Keywords: genetic conditions

1. Introduction

The signs and symptoms of this condition can begin at any age, although the most severe cases are apparent in infancy. However, up to one-third of people with factor VII deficiency never have any bleeding problems. Factor VII deficiency commonly causes nosebleeds (epistaxis), bleeding of the gums, easy bruising, and prolonged or excessive bleeding following surgery or physical injury. Bleeding into joint spaces (hemarthrosis) and blood in the urine (hematuria) occasionally occur. Many women with factor VII deficiency have heavy or prolonged menstrual bleeding (menorrhagia). Severely affected individuals have an increased risk of bleeding inside the skull (intracranial hemorrhage) or in the gastrointestinal tract, which can be life-threatening. Although factor VII deficiency is primarily associated with increased bleeding, some people with the condition have excessive blood clotting (thrombosis).

2. Frequency

Factor VII deficiency is estimated to affect 1 in 300,000 to 1 in 500,000 people. It is the most frequently occurring of a group of disorders classified as rare bleeding disorders.

3. Causes

The inherited form of factor VII deficiency, known as congenital factor VII deficiency, is caused by mutations in the *F7* gene, which provides instructions for making a protein called coagulation factor VII. This protein plays a critical role in the coagulation system, which is a series of chemical reactions that forms blood clots in response to injury. These mutations reduce the amount of coagulation factor VII in the bloodstream. Such a reduction prevents blood from clotting normally, causing episodes of excessive bleeding. It is not known why some people with this condition have problems with thrombosis. Researchers also do not know what determines the severity of the condition; it does not appear to be related to the amount of coagulation factor VII in the bloodstream.

The noninherited form of the disorder, called acquired factor VII deficiency, is less common than the congenital form. It can be caused by liver disease or by blood cell disorders such as myeloma or aplastic anemia. Acquired factor VII deficiency can also be caused by certain drugs such as medicines that prevent clotting, or by a deficiency of vitamin K.

3.1. The Gene Associated with Factor VII Deficiency

- *F7*

4. Inheritance

Congenital factor VII deficiency is inherited in an autosomal recessive pattern, which means both copies of the *F7* gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Acquired factor VII deficiency is not inherited.

5. Other Names for This Condition

- F7 deficiency

- hypoproconvertinemia
- proconvertin deficiency
- prothrombin conversion accelerator deficiency
- serum prothrombin conversion accelerator deficiency

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