Prothrombin deficiency

Description

Prothrombin deficiency is a bleeding disorder that slows the blood clotting process. People with this condition often experience prolonged bleeding following an injury, surgery, or having a tooth pulled. In severe cases of prothrombin deficiency, heavy bleeding occurs after minor trauma or even in the absence of injury (spontaneous bleeding). Women with prothrombin deficiency can have prolonged and sometimes abnormally heavy menstrual bleeding. Serious complications can result from bleeding into the joints, muscles, brain, or other internal organs. Milder forms of prothrombin deficiency do not involve spontaneous bleeding, and the condition may only become apparent following surgery or a serious injury.

Frequency

Prothrombin deficiency is very rare; it is estimated to affect 1 in 2 million people in the general population.

Causes

Mutations in the *F2* gene cause prothrombin deficiency. The *F2* gene provides instructions for making the prothrombin protein (also called coagulation factor II), which plays a critical role in the formation of blood clots in response to injury. Prothrombin is the precursor to thrombin, a protein that initiates a series of chemical reactions to form a blood clot. After an injury, clots protect the body by sealing off damaged blood vessels and preventing further blood loss.

F2 gene mutations reduce the production of prothrombin in cells, which prevents clots from forming properly in response to injury. Problems with blood clotting can lead to excessive bleeding. Some mutations drastically reduce the activity of prothrombin and can lead to severe bleeding episodes. Other *F2* gene mutations allow for a moderate amount of prothrombin activity, typically resulting in mild bleeding episodes.

Learn more about the gene associated with Prothrombin deficiency

- F2

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the 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.

Other Names for This Condition

- Dysprothrombinemia
- Factor II deficiency
- Hypoprothrombinemia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Congenital prothrombin deficiency (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0272317/)

Genetic and Rare Diseases Information Center

- Congenital factor II deficiency (https://rarediseases.info.nih.gov/diseases/2926/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Prothrombin deficiency%22)

Catalog of Genes and Diseases from OMIM

- PROTHROMBIN DEFICIENCY, CONGENITAL (https://omim.org/entry/613679)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28prothrombin+deficiency%5BTIAB%5D%29+OR+%28factor+II+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

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