Factor X deficiency

Description

Factor X deficiency is a rare bleeding disorder that varies in severity among affected individuals. The signs and symptoms of this condition can begin at any age, although the most severe cases are apparent in childhood. Factor X deficiency commonly causes nosebleeds, easy bruising, bleeding under the skin, bleeding of the gums, blood in the urine (hematuria), and prolonged or excessive bleeding following surgery or trauma. Women with factor X deficiency can have heavy or prolonged menstrual bleeding (menorrhagia) or excessive bleeding in childbirth, and may be at increased risk of pregnancy loss (miscarriage). Bleeding into joint spaces (hemarthrosis) occasionally occurs. Severely affected individuals have an increased risk of bleeding inside the skull (intracranial hemorrhage), in the lungs (pulmonary hemorrhage), or in the gastrointestinal tract, which can be life-threatening.

Frequency

Factor X deficiency occurs in approximately 1 per million individuals worldwide.

Causes

The inherited form of factor X deficiency, known as congenital factor X deficiency, is caused by mutations in the *F10* gene, which provides instructions for making a protein called coagulation factor X. 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. Some *F10* gene mutations that cause factor X deficiency reduce the amount of coagulation factor X in the bloodstream, resulting in a form of the disorder called type I. Other *F10* gene mutations result in the production of a coagulation factor X protein with impaired function, leading to type II factor X deficiency. Reduced quantity or function of coagulation factor X prevents blood from clotting normally, causing episodes of abnormal bleeding that can be severe.

A non-inherited form of the disorder, called acquired factor X deficiency, is more common than the congenital form. Acquired factor X deficiency can be caused by other disorders such as severe liver disease or systemic amyloidosis, a condition involving the accumulation of abnormal proteins called amyloids. Acquired factor X deficiency can also be caused by certain drugs such as medicines that prevent clotting, or by a deficiency of vitamin K.

Learn more about the gene associated with Factor X deficiency

- F10

Inheritance

When this condition is caused by mutations in the *F10* gene, it 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.

Acquired factor X deficiency is not inherited, and generally occurs in individuals with no history of the disorder in their family.

Other Names for This Condition

- Congenital Stuart factor deficiency
- F10 deficiency
- Stuart-Prower factor deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Factor X deficiency (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0015519/)

Genetic and Rare Diseases Information Center

- Congenital factor X deficiency (https://rarediseases.info.nih.gov/diseases/6404/index)

Patient Support and Advocacy Resources

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

Clinical Trials

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

Catalog of Genes and Diseases from OMIM

- FACTOR X DEFICIENCY (https://omim.org/entry/227600)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28factor+X+deficiency%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

References

- Brown DL, Kouides PA. Diagnosis and treatment of inherited factor Xdeficiency. Haemophilia. 2008 Nov;14(6):1176-82. doi:10.1111/j.1365-2516.2008.01856.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19141158)
- Girolami A, Scarparo P, Scandellari R, Allemand E. Congenital factor Xdeficiencies with a defect only or predominantly in the extrinsic or in theintrinsic system: a critical evaluation. Am J Hematol. 2008 Aug;83(8):668-71.doi: 10.1002/ajh.21207. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18506702)
- Girolami A, Vettore S, Scarparo P, Lombardi AM. Persistent validity of aclassification of congenital factor X defects based on clotting, chromogenic andimmunological assays even in the molecular biology era. Haemophilia. 2011Jan; 17(1):17-20. doi: 10.1111/j.1365-2516.2010.02328.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20546029)
- Herrmann FH, Auerswald G, Ruiz-Saez A, Navarrete M, Pollmann H, Lopaciuk S, Batorova A, Wulff K; Greifswald Factor X Deficiency Study Group. Factor Xdeficiency: clinical manifestation of 102 subjects from Europe and Latin Americawith mutations in the factor 10 gene. Haemophilia. 2006 Sep;12(5):479-89. doi:10.1111/j.1365-2516.2006.01303.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16919077)
- Karimi M, Menegatti M, Afrasiabi A, Sarikhani S, Peyvandi F. Phenotype andgenotype report on homozygous and heterozygous patients with congenital factor Xdeficiency. Haematologica. 2008 Jun;93(6):934-8. doi: 10.3324/haematol. 12211.Epub 2008 Apr 9. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18403394)
- Menegatti M, Peyvandi F. Factor X deficiency. Semin Thromb Hemost. 2009Jun;35(4):407-15. doi: 10.1055/s-0029-1225763. Epub 2009 Jul 13. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/19598069)
- Uprichard J, Perry DJ. Factor X deficiency. Blood Rev. 2002 Jun;16(2):97-110.doi: 10.1054/blre.2002.0191. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12127953)

Last updated January 1, 2015