Factor X deficiency

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.

Genetic Changes

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.

Inheritance Pattern

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

Diagnosis & Management

Genetic Testing

• Genetic Testing Registry: Factor X deficiency

Other Diagnosis and Management Resources

• MedlinePlus Encyclopedia: Factor X Assay
  https://medlineplus.gov/ency/article/003680.htm

General Information from MedlinePlus

• Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
• Drug Therapy
  https://medlineplus.gov/drugtheraphy.html
• Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
• Palliative Care
  https://medlineplus.gov/palliativecare.html
• Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

• Encyclopedia: Bleeding into the Skin
  https://medlineplus.gov/ency/article/003235.htm
• Encyclopedia: Blood Clot Formation (image)
  https://medlineplus.gov/ency/imagepages/19462.htm
Genetic and Rare Diseases Information Center

- Factor X deficiency
  https://rarediseases.info.nih.gov/diseases/6404/factor-x-deficiency

Educational Resources

- Centre for Arab Genomic Studies

- Disease InfoSearch: Factor X Deficiency
  http://www.diseaseinfosearch.org/Factor+X+Deficiency/2712

- MalaCards: factor x deficiency
  http://www.malacards.org/card/factor_x_deficiency

  https://www.merckmanuals.com/home/blood-disorders/blood-clotting-process/how-blood-clots

- Orphanet: Congenital factor X deficiency
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=328

- WomensHealth.gov: Bleeding Disorders Fact Sheet
  https://www.womenshealth.gov/a-z-topics/bleeding-disorders

Patient Support and Advocacy Resources

- Foundation for Women and Girls with Blood Disorders
  http://www.fwgbd.org/

- National Hemophilia Foundation
  https://www.hemophilia.org/Bleeding-Disorders/Types-of-Bleeding-Disorders/Other-Factor-Deficiencies/Factor-X

- World Federation of Hemophilia
ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22factor+X+deficiency%22

Scientific Articles on PubMed

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

OMIM

- FACTOR X DEFICIENCY
  http://omim.org/entry/227600

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19141158

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18506702

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20546029

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16919077

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18403394

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19598069

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