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.

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.

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 genetic 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 Information

• What is genetic testing? /primer/testing/genetictesting
• Genetic Testing Registry: Factor X deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0015519/

Research Studies from ClinicalTrials.gov

• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22factor%20X%20deficiency%22

Other Diagnosis and Management Resources

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

Additional Information & Resources

Health Information from 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
• Encyclopedia: Factor X Assay
  https://medlineplus.gov/ency/article/003680.htm
• Encyclopedia: Factor X Deficiency
  https://medlineplus.gov/ency/article/000553.htm
• Encyclopedia: Gastrointestinal Bleeding
  https://medlineplus.gov/ency/article/003133.htm
• Encyclopedia: Nosebleed
  https://medlineplus.gov/ency/article/003106.htm

• Health Topic: Bleeding Disorders
  https://medlineplus.gov/bleedingdisorders.html

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
  http://www.cags.org.ae/pdf/227600.pdf
• MalaCards: factor x deficiency
  https://www.malacards.org/card/factor_x_deficiency
• Merck Manual Consumer Version: How Blood Clots
  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: Rare Clotting Factor Deficiencies
  http://elearning.wfh.org/elearning-centres/rare-clotting-factor-deficiencies/

Scientific Articles on PubMed
• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28factor+X+deficiency%5BTIAB
  %5D%29+AND+english%5Bl%5D+AND+human%5Bm%5D+AND+%22last
  +1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM
• FACTOR X DEFICIENCY
  http://omim.org/entry/227600
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Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/condition/factor-x-deficiency

Reviewed: January 2015
Published: October 29, 2019

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services