Factor XIII deficiency

Description

Factor XIII deficiency is a rare bleeding disorder. Researchers have identified an inherited form and a less severe form that is acquired during a person's lifetime.

Signs and symptoms of inherited factor XIII deficiency begin soon after birth, usually with abnormal bleeding from the umbilical cord stump. If the condition is not treated, affected individuals may have episodes of excessive and prolonged bleeding that can be life-threatening. Abnormal bleeding can occur after surgery or minor trauma. The condition can also cause spontaneous bleeding into the joints or muscles, leading to pain and disability. Women with inherited factor XIII deficiency tend to have heavy or prolonged menstrual bleeding (menorrhagia) and may experience recurrent pregnancy losses (miscarriages). Other signs and symptoms of inherited factor XIII deficiency include nosebleeds, bleeding of the gums, easy bruising, problems with wound healing, bleeding after surgery, and abnormal scar formation. Inherited factor XIII deficiency also increases the risk of spontaneous bleeding inside the skull (intracranial hemorrhage), which is the leading cause of death in people with this condition.

Acquired factor XIII deficiency becomes apparent later in life. People with the acquired form are less likely to have severe or life-threatening episodes of abnormal bleeding than those with the inherited form.

Frequency

Inherited factor XIII deficiency affects 1 to 3 per million people worldwide. Researchers suspect that mild factor XIII deficiency, including the acquired form of the disorder, is underdiagnosed because many affected people never have a major episode of abnormal bleeding that would lead to a diagnosis.

Causes

Inherited factor XIII deficiency results from mutations in the *F13A1* gene or, less commonly, the *F13B* gene. These genes provide instructions for making the two parts (subunits) of a protein called factor XIII. This protein plays a critical role in the coagulation cascade, which is a series of chemical reactions that forms blood clots in response to injury. After an injury, clots seal off blood vessels to stop bleeding and trigger blood vessel repair. Factor XIII acts at the end of the cascade to strengthen and

stabilize newly formed clots, preventing further blood loss.

Mutations in the *F13A1* or *F13B* gene significantly reduce the amount of functional factor XIII available to participate in blood clotting. In most people with the inherited form of the condition, factor XIII levels in the bloodstream are less than 5 percent of normal. A loss of this protein's activity weakens blood clots, preventing the clots from stopping blood loss effectively.

The acquired form of factor XIII deficiency results when the production of factor XIII is reduced or when the body uses factor XIII faster than cells can replace it. Acquired factor XIII deficiency is generally mild because levels of factor XIII in the bloodstream are 20 to 70 percent of normal; levels above 10 percent of normal are usually adequate to prevent spontaneous bleeding episodes.

Acquired factor XIII deficiency can be caused by disorders including an inflammatory disease of the liver called hepatitis, scarring of the liver (cirrhosis), inflammatory bowel disease, overwhelming bacterial infections (sepsis), and several types of cancer. Acquired factor XIII deficiency can also be caused by abnormal activation of the immune system, which produces specialized proteins called autoantibodies that attack and disable the factor XIII protein. The production of autoantibodies against factor XIII is sometimes associated with immune system diseases such as systemic lupus erythematosus and rheumatoid arthritis. In other cases, the trigger for autoantibody production is unknown.

Learn more about the genes associated with Factor XIII deficiency

- F13A1
- F13B

Inheritance

Inherited factor XIII deficiency is considered to have an autosomal recessive pattern of inheritance, which means that it results when both copies of either the *F13A1* gene or the *F13B* gene in each cell have mutations.

Some people, including parents of individuals with factor XIII deficiency, carry a single mutated copy of the *F13A1* or *F13B* gene in each cell. These mutation carriers have a reduced amount of factor XIII in their bloodstream (20 to 60 percent of normal), and they may experience abnormal bleeding after surgery, dental work, or major trauma. However, most people who carry one mutated copy of the *F13A1* or *F13B* gene do not have abnormal bleeding episodes under normal circumstances, and so they never come to medical attention.

The acquired form of factor XIII deficiency is not inherited and does not run in families.

Other Names for This Condition

- Deficiency of factor XIII
- Deficiency, Laki-Lorand factor
- Fibrin stabilizing factor deficiency

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Factor XIII, b subunit, deficiency of (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750481/)
- Genetic Testing Registry: Factor XIII, A subunit, deficiency of (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750514/)

Genetic and Rare Diseases Information Center

- Congenital factor XIII deficiency (https://rarediseases.info.nih.gov/diseases/10766/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 XIII deficiency%22)

Catalog of Genes and Diseases from OMIM

- FACTOR XIII, A SUBUNIT, DEFICIENCY OF (https://omim.org/entry/613225)
- FACTOR XIII, B SUBUNIT, DEFICIENCY OF (https://omim.org/entry/613235)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Factor+XIII+Deficiency%5BMAJR%5D%29+AND+%28factor+XIII*%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

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