



NATIONAL HEMOPHILIA FOUNDATION
for all bleeding disorders

FACTOR X (F10)

DEFICIENCY

YOU ARE NOT ALONE

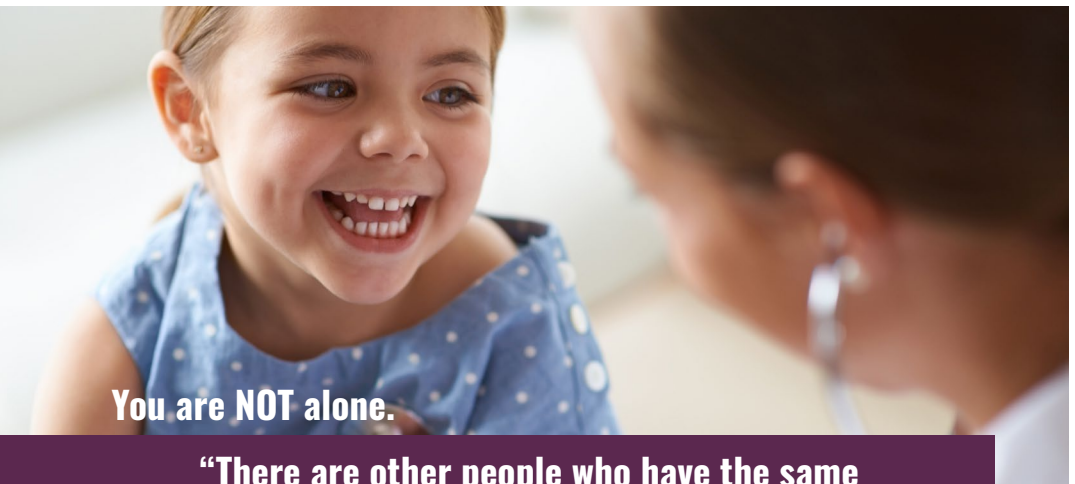


Factor X Deficiency

WHAT YOU SHOULD KNOW

Whether you are newly diagnosed with **Factor X Deficiency (FX Deficiency)** or have been diagnosed for some time, it is common to go through many ups and downs. We want to share information that you should know and some resources about this condition that can help you on your path to improved health and quality of life. **When you are first given a new diagnosis, one as rare as FX Deficiency, it is understandable to have many emotions or feel overwhelmed.** The good thing is that you're

not alone. There are other people who have the same condition and lead full lives. They have had their lives enriched by becoming closer to other people who are traveling on a similar path. With the care of an experienced team, you will find support from your health-care providers as well as from the vibrant and caring bleeding disorders community. You can be involved in this community and build meaningful relationships with your new extended family.



You are NOT alone.

“There are other people who have the same condition and lead full lives. They have had their lives enriched by becoming closer to other people who are traveling on a similar path.”

The **National Hemophilia Foundation (NHF)** is here to provide you with education and support as you manage your bleeding disorder whether you are a child, a teen, or an adult. In this spirit, NHF and the bleeding disorders community have selected some of the most frequently asked questions and answers below to empower you.



What Is Factor X Deficiency?

FX Deficiency is an ultra-rare bleeding disorder. Bleeding disorders are a group of medical conditions marked by an inability or decreased ability to form a stable blood clot. When the body is injured and an area bleeds, a clot is formed to stop the bleeding. Clot formation is a multistep process called coagulation. When the blood clots properly, the blood clot is held together firmly at the site of the injury. People with a bleeding disorder are unable to make strong clots or none at all. Improper

clotting can be caused by abnormalities in blood components such as platelets and/or blood clotting proteins, also called clotting factors. Platelets are small blood cells that help blood clot. Platelets act like first responders and stop bleeding by clumping and forming plugs in blood vessel injuries. This initial plug is strengthened by the formation of a mesh-like network termed fibrin that is generated by a series of clotting proteins.






“Bleeding disorders are a group of medical conditions that share an inability or decreased ability to form a stable blood clot.”

If any clotting protein is defective, deficient, or absent, the clot formation will be inadequate, and bleeding will happen. FX is one of the clotting factors that is an important part of this process and works by making the clot strong or stable. **FX Deficiency is caused by an abnormal function or decreased amount of the protein. FX Deficiency affects approximately 1 in 500,000-1,000,000.¹** It is also known as Stuart-Prower Factor Deficiency, named after the first two patients diagnosed with this condition.¹

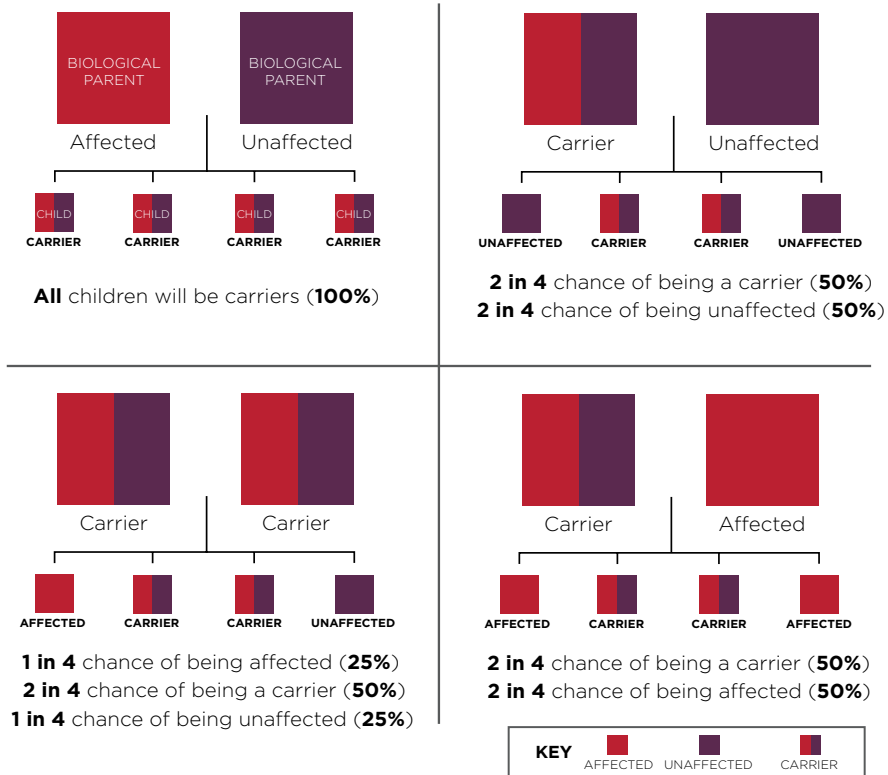


How does someone get Factor X Deficiency?

FX Deficiency is an inherited bleeding disorder, meaning that it is passed on from biological parents to child at the time of conception.

-  It affects both males and females equally.
-  The affected person has two abnormal copies of a defective gene.
-  A carrier has only one copy of a defective gene. Typically, carriers do not have bleeding symptoms.
-  For someone to inherit FX Deficiency they must inherit a copy of the defective gene from both biological parents. That means both parents must be affected or are carriers of FX Deficiency.
-  This pattern of inheritance is called Autosomal Recessive. It is different from what is called X-linked inheritance, as in the case of some other bleeding disorders like hemophilia. The different forms of autosomal recessive inheritance are illustrated on the next page.

Family Inheritance of an Autosomal Recessive Disorder



SUMMARY

If you are **AFFECTED** with Factor X Deficiency, you probably inherited one defective gene from each of your biological parents.

If you are a **CARRIER** of Factor X Deficiency, you inherited only one copy of a defective gene from a biological parent.

You can use the same logic to figure out the likelihood of your children being impacted by the deficiency, depending on your and your partner's genes. Genetic testing and counseling are available if you are interested in figuring out where the deficiency came from or who else in the family might be at risk of having the same condition.

Sometimes a person might develop FX Deficiency, rather than being born with it, as noted in patients with other medical conditions including some types of cancer.² Such an "acquired deficiency" is still quite rare. It can be caused by an antibody, which is a protein produced by the body's immune system. The antibody interferes with the way FX Deficiency works in the body. Acquired FX Deficiency can also be caused by an unrelated condition called amyloidosis, in which an abnormal protein aggregate (amyloid) binds FX and reduces its level in circulation. If you have acquired FX Deficiency, you may have variable bleeding symptoms that can range from mild to severe.

What are the symptoms of Factor X Deficiency?

The symptoms of FX Deficiency correspond well with how much FX you have in your blood. People who do not have FX Deficiency have a level above 60% of the normal amount of FX.³

Those with a FX level less than 10% of the normal amount are considered to have a severe deficiency; those with levels between 10%~40% are moderate; and those with levels greater than 40%-60% are considered to have a mild case of FX Deficiency.

If you have severe FX Deficiency, you may experience symptoms similar to persons with hemophilia, including joint bleeds (hemarthrosis), bleeding in the brain (intracranial hemorrhage), and

bleeding into the stomach or intestines (gastrointestinal bleeding). Those with levels greater than 10% typically have only minor bleeding without knowing why, or bleeding caused by trauma or surgery. Typically, these bleeds include easy bruising, nose bleeds (epistaxis), and gum bleeding. Up to 75% of women who are FX deficient have heavy menstrual bleeding and can experience heavy bleeding after childbirth.²

If you are a carrier and experience bleeding symptoms, it is important to seek medical care from a hematologist (a doctor specializing in the study of blood).

Classification of FX Deficiency^{1,2}



less than 10%

Higher risk of severe bleeding:

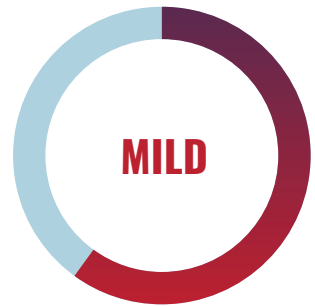
- Joint bleeds (hemarthrosis)
- Bleeding in the brain (intracranial hemorrhage)
- Bleeding into your stomach or intestines (Gastrointestinal or GI bleeds)



10%~40%

Low risk of severe bleeding:

- Minor bleeding without knowing why
 - Minor bleeding caused by trauma or surgery
- Typically these bleeds include:
- easy bruising
 - nose bleeds (epistaxis)
 - gum bleeding

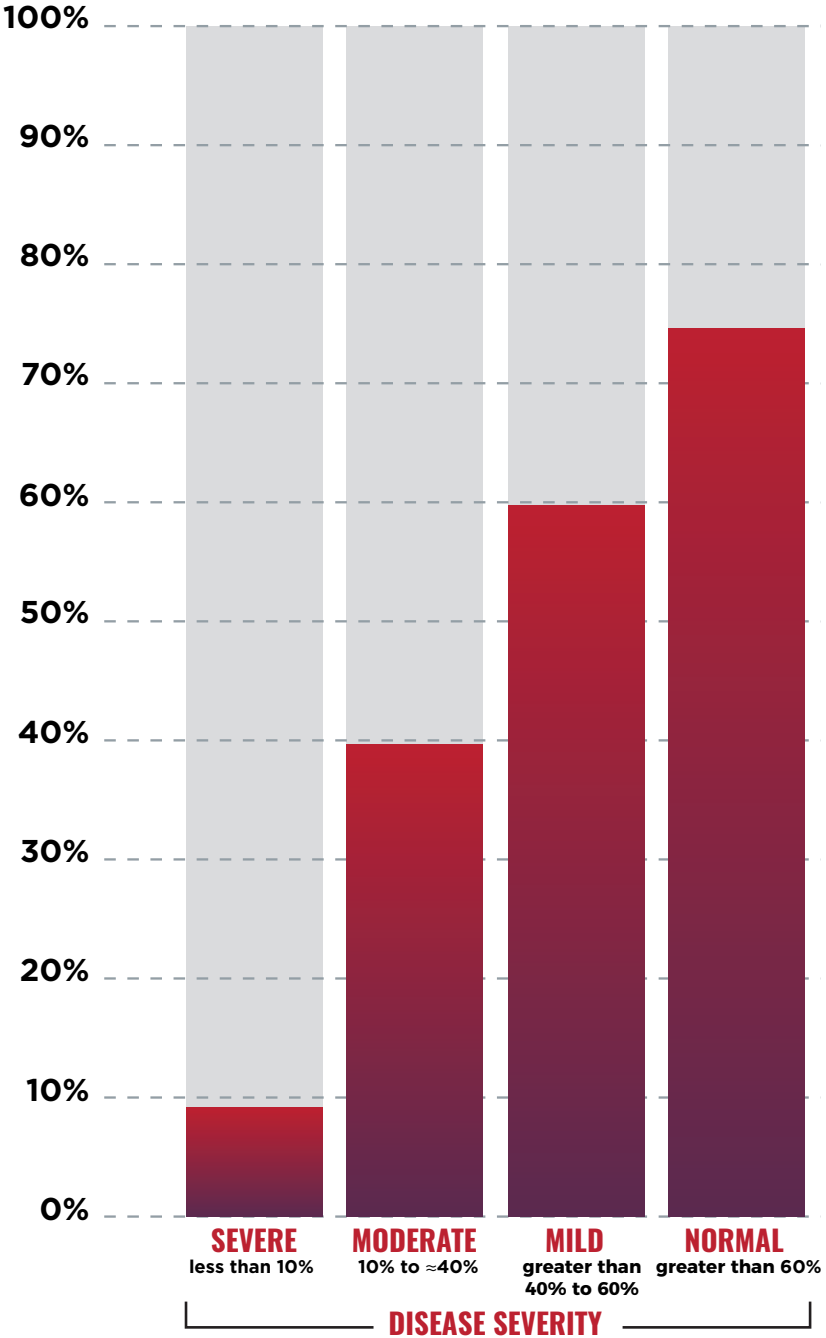


greater than 40%-60%

Mostly don't show symptoms, but might have problems with bleeding during:

- Trauma
- Surgical procedure
- Pregnancy / delivery

FX Activity Level





How is Factor X Deficiency diagnosed?

The diagnosis of FX Deficiency is challenging because it is rare and the common lab tests to look for this bleeding disorder may show results that are normal.

Persons with the severe form of FX Deficiency are often diagnosed with:

- 🩸 excessive bleeding during infancy, like bleeding following circumcision in boys
- 🩸 bleeding when the umbilical cord falls off, around 7-14 days after birth
- 🩸 bleeding into the brain (intracranial bleeding)
- 🩸 bleeding into the stomach or intestines (gastrointestinal hemorrhage)⁴


FX levels are normally low in all infants, even those without FX Deficiency and therefore the measurement may need to be repeated before a diagnosis of FX Deficiency is made. Persons with a mild or moderate deficiency may be diagnosed after some type of challenge, such as a surgery or trauma. The milder forms of the disorder may be diagnosed during routine screening or based on family history.² When a bleeding disorder is suspected, your healthcare provider will draw your blood and send your blood to be tested in a lab. The tests will measure how long it takes your blood to clot. If the results of these screening tests are suspicious then a FX Deficiency can be confirmed with a Factor X assay, to measure the level of FX in the blood. **The FX assay is a sensitive test that is best done through a Hemophilia Treatment Center (HTC), a comprehensive clinic that specializes in the care of individuals with coagulation disorders.** There are two types of FX Deficiency. Your physician can tell you what type you have.⁴

How is Factor X Deficiency treated?

For optimal care, it is recommended that you find a specialized doctor, or hematologist, who is experienced in the treatment of FX Deficiency. These doctors often work at a Hemophilia Treatment Center (HTC).

There is one FDA-approved FX concentrate that comes from human blood. In addition to the FX concentrate, fresh frozen plasma (FFP), a part of blood, and other types of factors have been used to treat bleeding in FX deficient patients.¹ FX concentrate may not be readily available, and another concentrate with FX (Profilnine) could be given.

For minor symptoms, topical therapies (applied to the skin) and/or antifibrinolytics (such as tranexamic acid pills that help the clot you make last longer) may be enough to control bleeding without needing to use replacement factor. These can also be used with the FX concentrate.¹ Patients with frequent bleeding, like repeated joint bleeds or severe nose bleeds, may benefit from “prophylactic” treatment 1-2 times per week. Heavy menstrual bleeding may often respond to standard hormonal therapy used for heavy periods in general. Your healthcare provider (HCP) will work with you to develop an ideal treatment plan based on your bleeding history.

A photograph of a man with a beard and short hair, wearing a light grey t-shirt, kissing a young boy on the cheek. The boy has dark, curly hair and is wearing a light blue shirt. They are outdoors, with a blurred green background of trees and grass. The man's hands are gently cupping the boy's face.

**“There is one
FDA-approved
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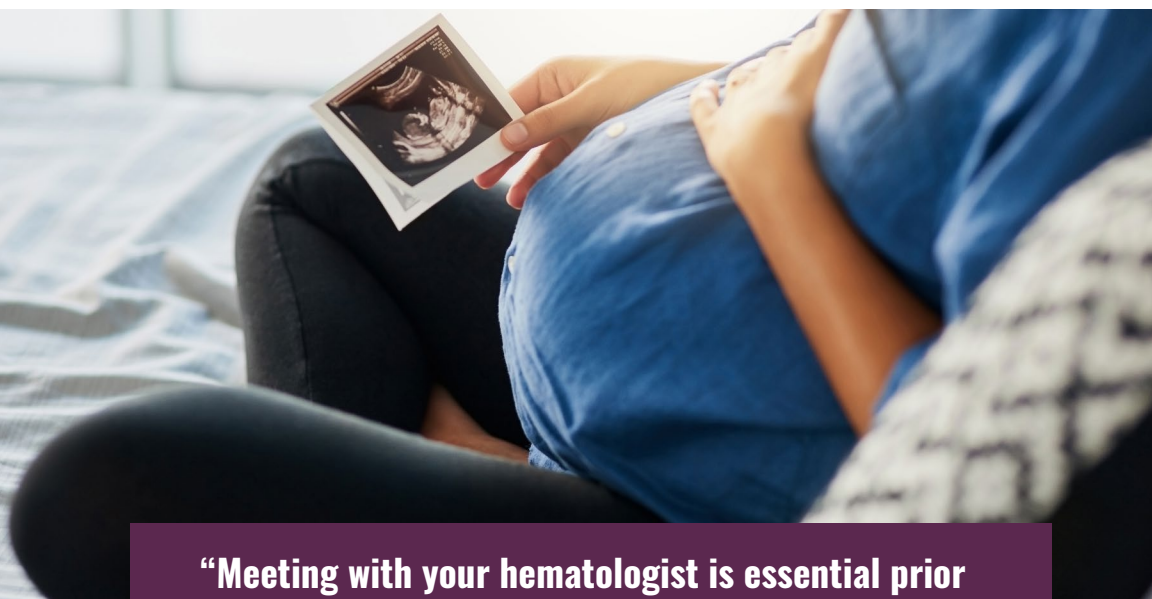
For the most current list of FDA-approved treatments for all bleeding disorders, including FX Deficiency, visit:

www.hemophilia.org/healthcare-professionals/guidelines-on-care/products-licensed-in-the-us

What special precautions need to be taken when considering pregnancy? _____

Women with FX Deficiency can experience successful pregnancy, but most will have some degree of bleeding. **Women with a severe FX Deficiency can experience several pregnancy-related complications and may require preventive treatment with Factor X concentrate (prophylaxis) during their pregnancy.**⁵

Meeting with your hematologist is essential prior to getting pregnant. It is important to work closely with a hematologist who is experienced in the treatment of FX Deficiency. Often these specialized doctors are found at a HTC. They can guide your women's health doctor (OB/GYN) to help develop a treatment plan during your pregnancy, labor and delivery, and up to 4-6 weeks after delivery (the postpartum period). HTC hematologists will also be able to provide care to your baby and provide testing as needed.



“Meeting with your hematologist is essential prior to getting pregnant.”

ADVICE FROM OTHER FX COMMUNITY MEMBERS



While we set out with the best intentions for our children, sometimes we encounter pitfalls we never could have envisioned. But often the most challenging journeys become opportunities for finding our best selves. If we seek out knowledge and steer with confidence and optimism, we can enjoy this extraordinary journey alongside those other rare gems sharing the same path.”

Parent of a child with FX Deficiency



Having NHF in your corner turns a FX diagnosis from the end of the world into, oddly enough, a unique life opportunity. It is difficult, but you are not alone; there’s a whole community waiting to meet you with open arms that you would never have known otherwise.”

FX Patient



Don’t let your diagnosis define you, let it inspire you. Be a champion for yourself and others within the bleeding community.”

FX Patient



Even though you are one in a million you are not alone. The Factor X community is here for you. It’s great to be a 10. It’s an exclusive club.”

FX Patient

Where else can I obtain additional information?

You are now part of a family known as the bleeding disorders community. You are not alone and you can turn to other members of this community for support if needed:



Know how to navigate disclosing your or your child’s bleeding disorder to daycare, school, work, emergency rooms, and non-hematology specialists.



Learn more about advocating for appropriate treatment in an emergency room (ER) or with other healthcare providers who may not know much about FX Deficiency. Always carry your treatment plan letter provided by your Hemophilia Treatment Center when travelling or going to the emergency room.



Find out where to connect with others with bleeding disorders locally.

Resources:

The National Hemophilia Foundation

- **Factor 10 webpage:** <https://www.hemophilia.org/bleeding-disorders-a-z/types/other-factor-deficiencies/factor-x>
- **Educational Resources:** <https://stepsforliving.hemophilia.org/>
- **HemAware magazine:** <https://hemaware.org/>
- **HANDI toll-free hotline:** 1-800-42-HANDI

Foundation for Women & Girls with Blood Disorders

- **Website directory of specialty women's clinics:** <https://www.fwgbd.org/clinics>

Hemophilia Federation of America

- **The Learning Central:** <https://www.hemophiliafed.org/the-institute/>

Rare Coagulation Disorders

- **Factor VII Deficiency:** <http://www.rarecoagulationdisorders.org/diseases/factor-x-deficiency/disease-overview>

The National Institute of Health

- **Genetic and Rare Diseases Information Center:** <https://rarediseases.info.nih.gov/diseases/6404/factor-x-deficiency>

Mayo Clinic

- **Autosomal Recessive Inheritance Pattern:** <https://www.mayoclinic.org/autosomal-recessive-inheritance-pattern/img-20007457>

Comprehensive Health Education Services (CHES)

- **Website:** <https://www.ches.education/rare-bleeding-disorders>

National Organization for Rare Disorders (NORD)

- **Rare Disease Database:** <https://rarediseases.org/rare-diseases/factor-x-deficiency/>

References:

1. Rare Coagulation Disorders Resource Room. <http://www.rarecoagulationdisorders.org/diseases/factor-x-deficiency/medicationtreatment>. Accessed December 17, 2020.
2. Brown, D.L. and P.A. Kouides. Diagnosis and treatment of inherited factor X deficiency. *Haemophilia*. 2008, 1176-1182.
3. Pagana KD, Pagana TJ, Pagana TN. *Mosby's Diagnostic & Laboratory Test Reference*. 14th ed. Elsevier; 2019.
4. National Organization for Rare Disorders website. <https://rarediseases.org/rare-diseases/factor-x-deficiency/>. Accessed December 17, 2020.
5. Nance, D. et al. Factor X deficiency and pregnancy: preconception counselling and therapeutic options. *Haemophilia*. 2012, e277-e285

Acknowledgements:

The **National Hemophilia Foundation (NHF)** is dedicated to finding cures for inheritable blood disorders and to addressing and preventing the complications of these disorders through research, education, and advocacy enabling people and families to thrive. The NHF would like to express its appreciation to Sonia Nasr/Gloval LLC and Nikole Scappe for the content development, the rare working group members, Carlisa Magee, Natalia Winberry, MNLM, Lena Volland, PT, DPT, and Kate Nammacher, MPH, for their insights and review. A special thank you to Peter Kouides, MD, Leonard Valentino, MD, and all the individuals who reviewed drafts of this publication. This publication was developed through the support of NHF's 2021 Community Education Program sponsors: BioMarin, Genentech, Hemophilia Alliance, Sangamo, Sanofi Genzyme, and Takeda.

This booklet is intended for informational purposes only. It is not intended to be used to make healthcare coverage or treatment determinations. NHF's Medical and Scientific Advisory Council (MASAC) recommends that the product and corresponding treatment regimen used by an individual should remain a decision between patient and physician.

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