

Guide for Women Carriers & Women with Hemophilia



A publication by the Indiana Hemophilia & Thrombosis Center, Inc. 8326 Naab Road • Indianapolis, IN 46260
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Dear families with hemophilia,

The Indiana Hemophilia and Thrombosis Center, Inc. (IHTC), cares about your health. One of the initiatives at the IHTC is to focus on women with bleeding disorders.

Over the next several months, you will receive three educational newsletters designed specifically for **women with hemophilia and hemophilia carriers**. These newsletters highlight important issues related to your healthcare; topics covered include how to stay connected with the IHTC, what to tell your family, current treatments for females with bleeding symptoms, inheritance information, and more!

Share the information with your family. If you have family members who are experiencing bleeding symptoms, please put them in contact with us so that they can receive the very best care.

Please feel free to reach out to the IHTC if you have any questions or concerns. Call Meadow Heiman or Kristen Dieter, IHTC's Genetic Counselors, at 317.871.0000.

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I'm a carrier—what should I tell my family?

Now that you know you are a carrier of hemophilia, you should consider talking to family members

How do I get started?

Genetic counselors at the Indiana Hemophilia and Thrombosis Center (IHTC) will review your family history to help identify any family member who could be a carrier—generally sisters, mothers and daughters first, then consider aunts and female cousins. Individuals who need to be tested for hemophilia will then be determined.

Important facts to review with your family

You should discuss with your family members the type of hemophilia identified in your family: factor VIII deficiency, also called hemophilia A, and factor IX deficiency, also called hemophilia B or Christmas disease. You should also discuss the availability of testing, which can determine whether they are a carrier. Also consider:

- If a woman is *not* a carrier, her child will likely not have hemophilia
- If a woman *is* a carrier, there is a 25% chance of having a child with hemophilia with each pregnancy
 - Each son has a 50% chance of having hemophilia
 - Each daughter has a 50% chance of being a carrier
- If a woman *is* a carrier, she should:
 - Have her factor level checked, because carriers may have an increased risk of abnormal bleeding, especially with dental, surgical, or invasive medical procedures if their factor level is low
 - If pregnant, contact the IHTC to:
 - Help determine if there is a risk for bleeding during delivery



- Ask that recommendations for delivery be sent to your obstetrician
- Send cord blood testing - if the baby is a boy (need special kit and instructions which will be provided)
- If your family has a history of mild hemophilia, male family members of any age might not know they have hemophilia and need to be tested.
- There are multiple reproductive options available to carriers. These options can be discussed with a genetic counselor at the IHTC.

How can I get help with this?

Discussing your family history of hemophilia can be difficult. We're here to help any way we can. If you want to discuss who to talk to, what to say, or ways to approach family members, please contact one of the IHTC genetic counselors. Let your family know the IHTC is always available for any questions or concerns. You're always welcome to refer family members to us if they need assistance you cannot provide.

How do I know if I'm a Carrier of Hemophilia?

Females who may be carriers of hemophilia can be divided into two groups based on their family histories: obligate carriers and possible carriers. Genetic testing is sometimes necessary to determine whether a female is a carrier.

How is genetic carrier testing performed?

- Individuals with hemophilia have a change within their factor VIII or factor IX gene that has caused it to make lower-than-normal amounts of clotting factor VIII or clotting factor IX
- In order to perform carrier testing, it is preferred that a male family member with hemophilia be tested first to identify the genetic alteration in the factor VIII or factor IX gene causing hemophilia in the family
- Once the familial genetic alteration has been identified, female family members who are possible carriers can have a blood test to look for the presence or absence of this specific alteration within her factor VIII or factor IX genes, determining her carrier status

None of the males with hemophilia in my family have had genetic testing. Can I still be tested?

- Yes, but we would suggest you talk to your family members who have hemophilia to see if they would be willing to have genetic testing first.
- If a male family member with hemophilia is unavailable or unwilling to be tested, the second choice would be to perform genetic testing for an obligate carrier in your family first.
- If neither a male with hemophilia nor an obligate carrier in your family are available, a possible carrier can still have genetic testing performed. This, however, does not provide the most accurate results. There is a small chance of receiving a false negative result. This can be discussed further with a genetic counselor.

How can I get more information?

For additional information, to discuss your specific family history (including your chance to be a carrier), obtain more details about genetic testing, or to pursue genetic testing, please contact one of the IHTC's genetic counselors.

OBLIGATE CARRIER

Obligate carriers are females (any age) known to be carriers based on their family history alone and one of the following is true:

- Her father has hemophilia
- She has two or more sons with hemophilia
- She has a son with hemophilia and at least one other relative with hemophilia (such as a grandfather, brother, uncle, grandson, nephew, or cousin)

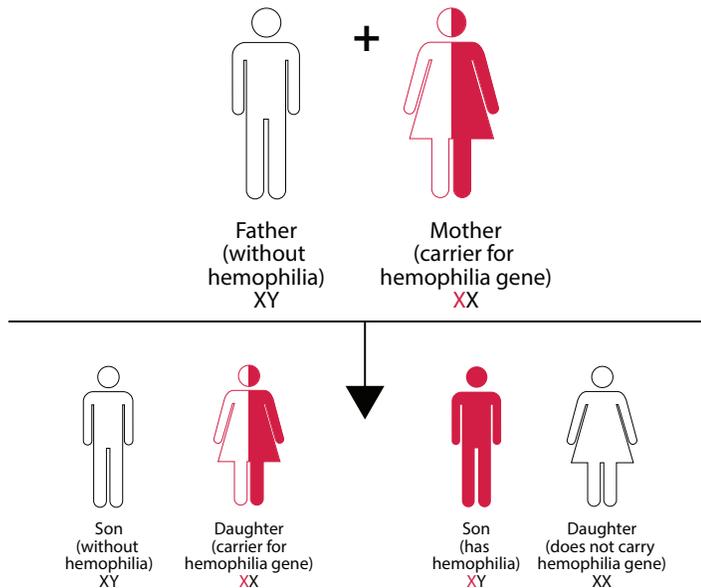
Obligate carriers *do not* require genetic carrier testing.

POSSIBLE CARRIER

Possible carriers are Females (any age) who have a chance of being carriers for hemophilia, but their carrier status cannot be determined by family history alone. Possible carriers *do need* genetic carrier testing to determine their carrier status.

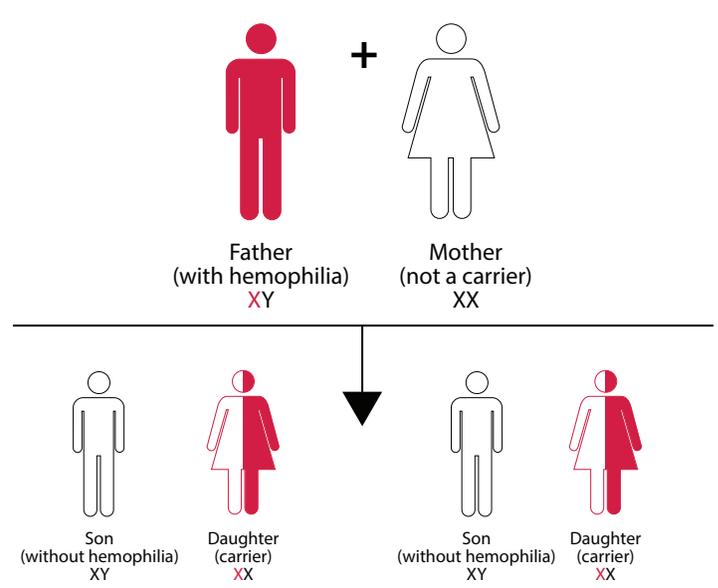
Inheritance of Hemophilia

"Carrier" Mother and Father without Hemophilia



Inheritance of Hemophilia

Father with Hemophilia and Mother Who is Not Carrier



Inheritance of Hemophilia A & B

X-linked Inheritance

- » Females have two "X" chromosomes (XX) and males have a single "X" and a single "Y" chromosome (XY)
- » X-linked disorders are associated with alterations, also known as mutations, on the X-chromosome. These disorders affect males more often than females because females have an additional X-chromosome that acts as a "back-up"
- » The factor VIII and factor IX genes, which cause hemophilia A and B respectively, are located on the X-chromosome
- » Because males only have one X-chromosome, any alteration in the factor VIII or IX gene will result in hemophilia
- » Females with an alteration on one X-chromosome are called "carriers"

A female carrier has a 50% chance of passing on the gene causing hemophilia to each of her children. Each son has a 50% chance of having hemophilia and each daughter has a 50% chance of being a carrier, like her mother. A man with hemophilia will pass the gene on to none (0%) of his sons and all (100%) of his daughters. Therefore, his sons will not typically have hemophilia, but all of his daughters will be carriers. See diagrams above.

Staying Connected.....

through life's stages.....

Every life stage brings new events and experiences which may require assistance from the IHTC

All Ages

Notify the IHTC about these events in case medication is required to prevent or manage bleeding:

- Surgeries
- Procedures
- Dental extractions or extensive dental work
- Accidents/Injuries
- Suspected bleeding episodes

It is important to stay connected and current with the IHTC to ensure the best care can be provided quickly in an emergency.

Adolescence and Young Adulthood

Menstrual Periods

Heavy or prolonged menstrual periods

Genetic Counseling

Information about the chance of having a child who has hemophilia or who carries the hemophilia gene

Pregnancy, Delivery, & Postpartum

Notify the IHTC about pregnancies
The IHTC will develop a delivery plan to prevent bleeding complications for both you and your baby. The IHTC will provide cord blood testing kit.



Mid-Life to Older Adulthood

New medications, herbs, or vitamins

There are certain medicines, supplements, and herbs that can increase your risk for bleeding

Example: Doctors might recommend starting an aspirin regimen as people age, depending on certain medical conditions

The IHTC can provide recommendations on medications or herbs that will not cause bleeding or discuss your risk of bleeding with medications or herbs you are using.

Routine screenings that may require pre-treatment to prevent bleeding:

- Colonoscopy
- Biopsy

Heavy menstrual bleeding during menopause

Questions? Concerns?

If you have any questions about your family history as it relates to your bleeding disorder, or if you have any family members who are experiencing bleeding symptoms, please feel free to reach out to one of IHTC's Genetic Counselors at 317.871.0000 or via the email addresses noted below.

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