



Genetics

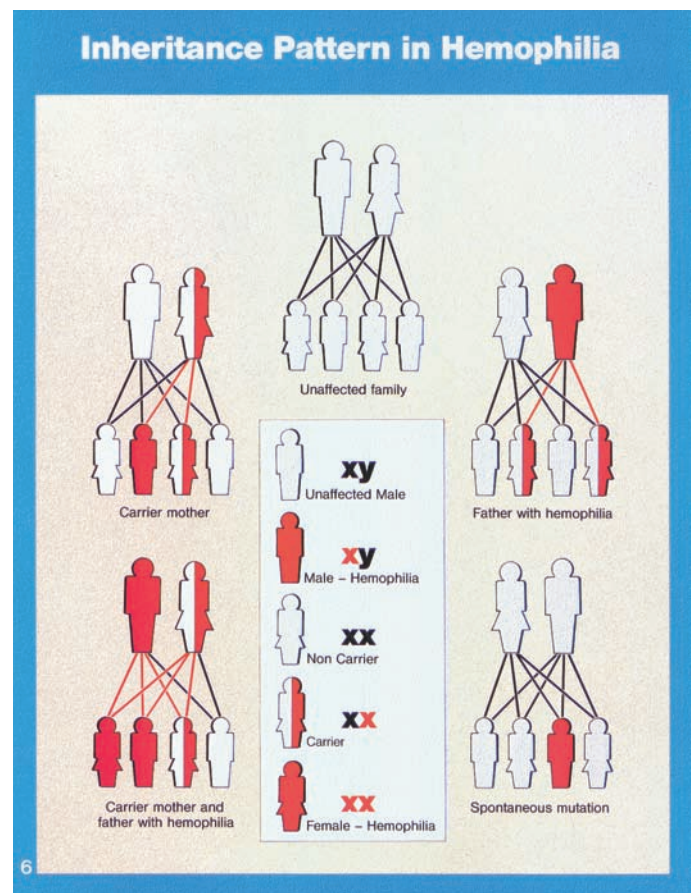
Hemophilia is caused by a mutation, or change, in the genetic material on the X chromosome (a threadlike structure that contains genetic information passed down through families)

General genetics

1. Females have two X chromosomes (XX) and males have one X and one Y chromosome (XY)
2. Everyone inherits two chromosomes from their parents
3. A female inherits an X chromosome from her mother and an X chromosome from her father
4. A male inherits an X chromosome from his mother and a Y chromosome from his father

Genetics and hemophilia

1. People with bleeding disorders have a defective blood clotting gene located on the X chromosome
2. Females have two X chromosomes—if the clotting factor gene on one X chromosome doesn't work, the gene on the other chromosome can do the job
3. Males have only one X chromosome, so if the clotting factor gene on the X chromosome doesn't work, they have hemophilia. This is why hemophilia affects mostly males.
4. Because the father contributes a Y chromosome to his son, none of the sons of a man with hemophilia will have hemophilia
5. Because the man's X chromosome determines that the baby will be a girl, all daughters of a man with hemophilia will be carriers
6. Up to 1/3 of people with hemophilia have no family history of the disease which means that a mutation of the clotting gene occurred
7. In rare instances, a female can have severe hemophilia from different mutations of the clotting gene



Hemophilia carriers

1. A carrier is a female who can pass hemophilia on to her children
2. Women who have the hemophilia gene on one X chromosome are carriers of hemophilia
3. There are 3 ways to determine if a woman is a hemophilia carrier:
 - Look at the family tree (if she has a son and another male relative with hemophilia, she is a carrier and no further testing is required)
 - Determine the clotting factor level (If the clotting factor level is less than 50%, she is probably a carrier and may have mild hemophilia. It is possible that a woman is a carrier even if the clotting level is above 50%)
 - Perform a DNA test to look for the hemophilia mutation
4. With each pregnancy, each son has a 50% risk of having hemophilia and each daughter has a 50% chance of being a carrier. The four possibilities for each pregnancy are:
 - A girl who is not a carrier (25% chance)
 - A girl who is a carrier (25% chance)
 - A boy without hemophilia (25% chance)
 - A boy with hemophilia (25% chance)
5. Women who are carriers can also have bleeding symptoms because of low factor VIII or factor IX levels
6. Women who are pregnant and may be carriers need to talk to their obstetrician about the potential for the baby to be born with hemophilia and to a hematologist so that plans can be made for a safe delivery
7. A diagnosis of hemophilia can be made soon after birth. Tests can be run on blood obtained from the umbilical cord or drawn from the newborn's vein

Genetic counseling

1. Genetic counseling is important for people with hemophilia
2. Genetic counseling includes a wide range of tests for diagnostic and carrier detection and individual counseling
3. Some families with a history of hemophilia request prenatal testing to see if a fetus has hemophilia
4. Testing can be done early in a pregnancy so that the family can make informed decisions and preparations
5. The HTC can give you further information about where to go for genetic counseling

[World Federation of Hemophilia. Guidelines for the management of hemophilia. 2005.]

[Medical Encyclopedia: Hemophilia A. Available at: www.nlm.nih.gov/medlineplus/print/ency/article/00538.htm.]

[National Hemophilia Foundation. Caring for your child with hemophilia.]