Hemophilia Inheritance Patterns

Hemophilia is a sex-linked recessive disorder. The abnormal gene responsible for hemophilia is carried on the X chromosome.

Males have one X chromosome and one Y chromosome. In a male, the presence of the abnormal gene results in the deficiency or absence of factor VIII or factor IX, as there is no protective X to make factor VIII or IX. Affected males cannot transmit the abnormal gene to their sons who inherit a Y chromosome from their father. However, all daughters of a man with hemophilia are considered obligate carriers of hemophilia because they must receive the affected X chromosome from their fathers to be female, and they can pass the gene to their sons.

A female with the hemophilia gene is called a hemophilia carrier. Females have two X chromosomes, so the presence of the hemophilia gene on one X does not result in hemophilia. The presence of the normal gene that codes for the production of factor compensates for the defective gene that does not. While usually not affected, females who have the gene can pass the gene to a son who will have hemophilia. In some cases, female carriers can have bleeding symptoms.

A woman is also considered an obligate carrier if she has more than one son with hemophilia, and they have at least one son and one other relative with hemophilia.