FAMILY AND INHERITANCE

Haemophilia is an inherited condition that occurs in families. The haemophilia gene is passed down from parent to child through generations. Mainly males have haemophilia. Men with haemophilia will pass the gene on to their daughters.

A small number of women also have haemophilia, but usually women who inherit the gene carry it without having the condition, although some have bleeding symptoms. Women who carry the haemophilia gene can pass the gene on to sons and daughters. Sons who inherit the gene will have haemophilia. Daughters who carry the gene can pass it on to their children.

Family members may need to be tested if there is someone in the family who has haemophilia.

When haemophilia is passed down within a family, the males in that family will always inherit the same type and severity of haemophilia, that is, a grandson with mild haemophilia A will have a maternal grandfather with mild haemophilia A. However, the level of severity for females is not related to the form of haemophilia gene they have inherited. For example, a girl whose father has severe haemophilia may have the same factor levels and symptoms as a girl whose father has mild haemophilia – but if she passes the gene onto any of her sons, they will have severe haemophilia.

Cells and chromosomes

The human body is made up of millions of cells. At the centre of each cell there are 46 chromosomes arranged in pairs. The person's genetic information or 'genes' are contained in their chromosomes. These determine the person's individual characteristics, such as the colour of their hair or their eyes.

Sex determination

Everyone has a pair of 'sex' chromosomes, which dictate what sex they are. Each parent contributes one of these chromosomes to their children. Females have two X chromosomes, and receive one from each parent. Males have one X chromosome, received from their mother, and one Y chromosome, received from their father. There are four possible combinations of sex chromosomes that children can receive from their parents.

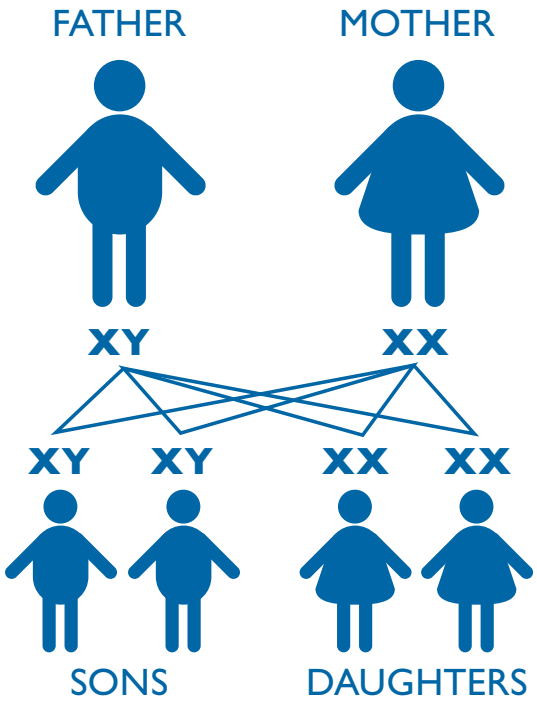
Inheritance

The genes for making factor VIII and IX are located on the X chromosome. Males with an altered factor VIII or IX gene on their X chromosome will have haemophilia. If a male with haemophilia has children, all his daughters will carry the haemophilia gene because he will pass his altered factor VIII or IX gene on to them. His sons will not have haemophilia as the 'normal' Y gene is passed on to them.

If a female who carries the haemophilia gene has children, her sons will have a 50% chance of having haemophilia and her daughters will have a 50% chance of carrying the gene. These percentages are the same with every pregnancy.

No family history of haemophilia

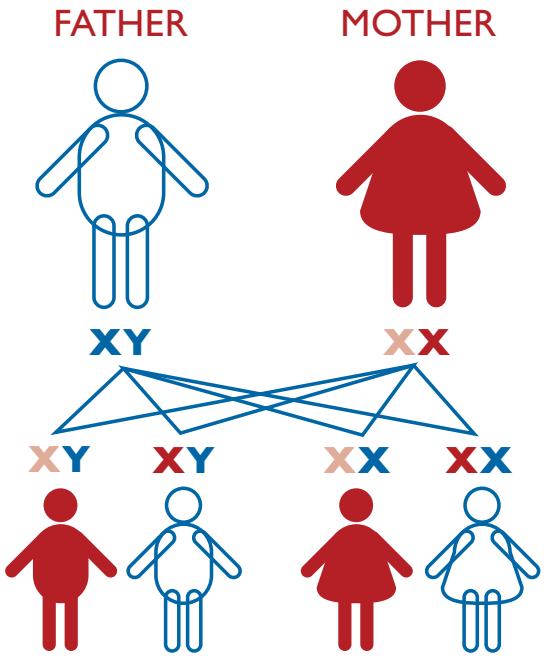
In about one third of people born with haemophilia, there is no history of the disorder in the family. This happens when a person has a genetic mutation in the factor VIII or IX gene on their X chromosome. It is often called a new or spontaneous mutation. Once haemophilia appears in a family the altered gene is then passed on from parents to children following the usual pattern for haemophilia. It is recommended that family members seek genetic counselling and testing if there is someone in the family who has haemophilia.



POSSIBLE CHILDREN

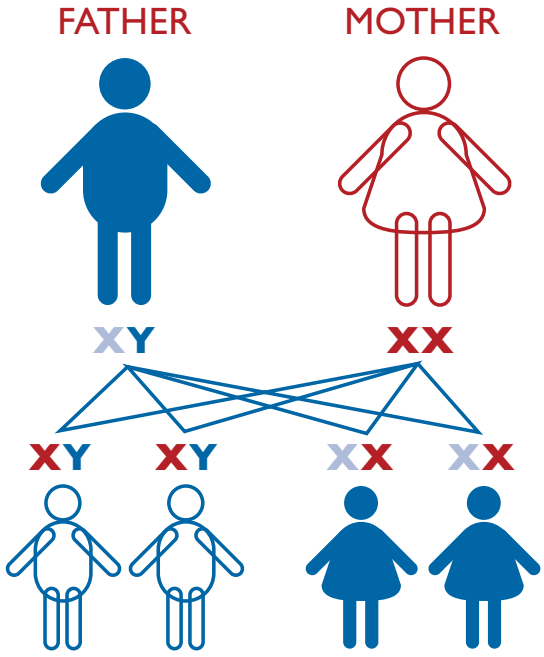
(The sex of the child is determined by the father)

When the mother carries the haemophilia gene and the father is unaffected



There is a 50% chance at each birth that a son will have haemophilia. There is a 50% chance at each birth that a daughter will carry the haemophilia gene

When the father has haemophilia and the mother is unaffected



None of the sons will have haemophilia
All of the daughters will carry the haemophilia gene

X or X = chromosome with haemophilia gene