## HAEMOPHILIA GENETICS

PWH

**0%** The possibility of a man with haemophilia having a son with haemophilia. **100%** That the daughter of a man with haemophilia will be a carrier.

Haemophilia is an inherited bleeding disorder. It is a sex-linked recessive inheritence condition which mainly affects males, but is carried by women. The defect in the clotting factor is carried on the X chromosome.

Everyone has two chromosomes that are related to sex - men have one X and one Y chromosome whereas women have two X chromosomes.

A man with haemophilia has a defective X chromosome. Any daughters of a man with haemophilia will inherit the defected X chromosome and will be carriers of haemophilia. A man with haemophilia cannot have a son with haemophilia since his son must inherit his Y chromosome.

All female relatives of a man with haemophilia should be tested for their carrier status, but also have their own factor levels checked. Daughters can be tested at a young age to determine their factor levels. If her levels are normal, this does not mean she is not a carrier. Carrier testing can only be carried out on girls aged 16 years and over.

arrie

**50%** Chance of a carrier having a a daughter who is a carrier. **50%** Chance of a carrier having a son with haemophilia.

A carrier of haemophilia will have one normal and one defective X chromosome. She will have a 50 / 50 chance of having a son with haemophilia or a daughter who is a carrier as she has two X chromosomes which she can pass on.

The level of severity does not change within a family. For example if the daughter of a man with severe haemophilia has a son with haemophilia, he will have severe haemophilia also.



## The Irish Haemophilia Society

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