

I NEED TO KNOW ABOUT HAEMOPHILIA

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What is haemophilia?

Patients with haemophilia have low levels of a protein essential for blood clotting.

Which protein is low?

There are two main types of haemophilia:

- **Haemophilia A** is most common type and is due to a deficiency of Factor VIII (8).
- **Haemophilia B** or 'Christmas Disease' is due to the deficiency of Factor IX (9).

Both types of haemophilia can range from mild to severe depending on how much Factor VIII or IX is missing.

What causes haemophilia?

Patients with haemophilia have a defect in the gene that controls the production of Factor VIII or IX.

Why is haemophilia a guy thing?

Both the Factor VIII and Factor IX genes are on the X chromosome. Males have one X and one Y chromosome. Any faulty Factor gene on the X chromosome will lead to low Factor levels.

Females have two X chromosomes (and no Y). For a female to be affected, both X chromosomes need to be abnormal. This is very rare. More commonly, a female will have one X with a faulty Factor gene and one with a normal Factor gene. Her body can make enough Factor proteins but she can pass the faulty gene on to a son. In this situation, she is called a 'carrier'.

How is someone with haemophilia affected?

Patients with haemophilia cannot make blood clots properly. This places them at risk of bleeding.

What type of bleeding?

The most debilitating bleeds occur in muscles and joints. These are very painful at the time and can lead to arthritis in the long term. Internal bleeding in the chest or head is rare but life-threatening.

Busting a couple of haemophilia myths

Myth #1: A person with haemophilia will bleed to death from a cut

NO. He will bleed for longer, but pressure on a cut will control the bleeding.



Myth #2: A person with haemophilia cannot play sport

NO. Sport will strengthen the muscles and joints hence making bleeding less likely. For some patients, Factor treatment is needed before playing some sports.

Myth #3: Haemophilia is contagious

NO. It is hereditary and cannot be 'caught' like a virus.

How is haemophilia treated?

Haemophilia is treated by replacing the missing Factor protein. Both Factor VIII and IX are available in 'recombinant' and 'plasma' forms.

- **Recombinant Factor** is made through genetic engineering technology.
- **Plasma-derived Factor** is made from the plasma in human blood. In Australia, plasma from the Blood Service goes to CSL for fractionation to make these products.

Factor is given by intravenous injection to treat bleeds but can also be given on a regular basis to prevent bleeds from happening.

BLOOD FACT

Queen Victoria was a carrier of Haemophilia A, hence the title 'the Royal Disease'.