

# **F**inding out

## **about carrying the haemophilia gene**

Haemophilia is mainly an inherited disorder, which means it runs in families. If one or more of your family members, who may be your dad, brother, uncle or cousin, has haemophilia, this could mean that you 'carry' the haemophilia gene.

## this means...

When or if you decide it is time to have a baby, if you have a boy there is a 50% chance that he will have haemophilia. If a girl, there is a 50% chance that she will carry the haemophilia gene.



HAEMOPHILIA FOUNDATION AUSTRALIA

# More on haemophilia

Humans have 13 proteins labelled in roman numerals from I to XIII (13). The majority are clotting factors; many are made in the liver.

People with haemophilia don't have all their clotting factors working properly together. Those with haemophilia A (Classical Haemophilia) have a problem with clotting factor VIII (8). This is the most common form of haemophilia. For people with haemophilia B (Christmas Disease), their problem clotting factor is factor IX (9).

About 30% of cases of haemophilia occur in families where there is no family history of the disorder.

It is a myth (untrue) that people with haemophilia are likely to bleed to death from a cut finger. It is **internal** bleeding that causes the problems. Bleeding episodes may start after injury or they may just happen. The bleeding can be stopped by one or more infusions (injections) of the missing clotting factor.

## What exactly makes someone carry the haemophilia gene?

Everyone has two sex chromosomes and they determine whether we are male or female. A female has two X chromosomes while a male has one X and one Y chromosome.

The genes which make clotting factor VIII and factor IX are located on the X chromosome. A fault in the factor VIII gene causes haemophilia A and a fault in the factor IX gene causes haemophilia B.

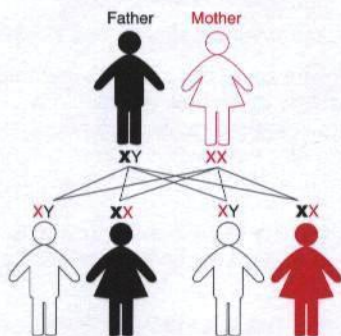


A female with a 'faulty' factor VIII or IX gene will not develop symptoms of haemophilia because usually her other X chromosome has a 'normal' gene which makes clotting factor. Females with this condition will *carry* the haemophilia gene, which they have inherited either from their father if he has haemophilia or from their mother if she carries this gene.

A small number of females who carry the haemophilia gene do have factor VIII or IX levels that are low enough to cause a bleeding tendency. Some of these females need factor replacement for surgery, teeth extractions and sometimes to treat heavy period bleeding.

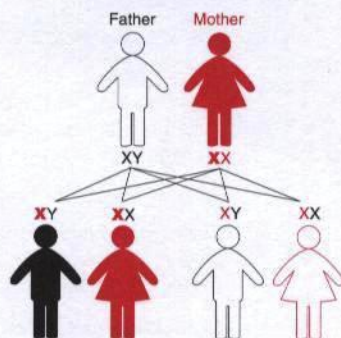
A male who has a faulty factor VIII or IX gene on his only X chromosome will have haemophilia as his X chromosome cannot make any clotting factor and his Y chromosome does not have a gene for making factors VIII or IX. If the male with haemophilia has children, all his daughters will carry the haemophilia gene because he will pass on his faulty factor VIII or IX gene to them. None of his sons will have haemophilia as the 'normal' Y gene is passed on to them. (see Fig 1)

**Figure 1**  
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.

**Figure 2**  
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.

A female who carries the gene can pass on to her children either the X chromosome with the normal gene or the X chromosome with the haemophilia gene. Each of her sons will have a 50% chance of inheriting the disorder from her and each of her daughters will have a 50% chance of carrying the haemophilia gene. (see Fig 2)

### Forget the baby for a minute. What about me?

You should be okay. Females who carry the haemophilia gene have one good X chromosome and one 'problem' X. Usually the good X is enough to make near normal levels of factor VIII or IX. But if the level of your clotting factor activity is low (normal levels are about 50-150 u/dl) and you have bleeding symptoms, then you are classed what is known as *symptomatic*.

When *symptomatic*, symptoms may include bruising, nose-bleeds, heavy periods (menorrhagia) and, after any surgery, you may have problems with bleeding. A severe accident may, in rare cases, cause joint bleeds as well as organ bleeding. The degree of bleeding depends on your level of clotting activity.

### What can I do about this?

For small bleeding problems, such as bruising, you may not need any treatment.

You can usually stop a nose-bleed by pinching the nostrils together for 10-15 minutes and repeating this procedure if you need to. Some bleeds in the mouth or tongue may be treated with medicine, such as, tranexamic acid tablets. This medication can only be prescribed by a doctor.

If you get very heavy periods your doctor may prescribe birth control pills to regulate the bleeding. You may also need iron tablets if you are low on iron. Tranexamic acid tablets may also be useful to control heavy periods.

Clotting factor concentrate will usually be infused (injected) if necessary before/after surgery or if you are involved in an accident.

For any problem or query regarding carrying the haemophilia gene it is best to go to your closest Haemophilia Centre.

### **How can I find out if I carry the haemophilia gene?**

Your nearest Haemophilia Centre can carry out all the necessary tests (or refer you to a hospital that does). The tests (which involve blood samples) will involve you and other members of your family, including the person with haemophilia. The reason for this is that there is more than one haemophilia gene, so you can't be the only person in your family tested. These tests can give a definite answer about carrier status in a large number of cases.

### **Different types of 'carriers' of the haemophilia gene**

Females who carry the haemophilia gene will be in one of the following categories:

**Obligatory** carrier of the haemophilia gene. This category covers females who *must* carry the haemophilia gene. This includes all daughters of men who have haemophilia. This category also covers a female from a family where there is haemophilia, who has a son with haemophilia;

**Possible** carrier of the haemophilia gene. This is any female from a family where there is haemophilia. She may have inherited the haemophilia gene;

**New mutation.** This can be any female at all who has experienced a mutation on one of her X chromosomes. Once in the family, haemophilia will be passed down in the same way as in families where it is an inherited condition.

### **I don't plan to have children, so what's the point in finding out whether I carry the haemophilia gene?**

The number of women who don't have children is increasing. Changing our minds, though, is part of being human. Things change, even though this is hard to believe sometimes. Everyone at one time or another has ended up doing something they said they would never do.

The worst time to find out that you carry the haemophilia gene is **after** you become pregnant. Then all of a sudden you may want to know and waiting for the tests could stress you out. Once you do know, you may have to make some decisions fast (ie. whether to have the baby) and this too can be stressful.



Another problem is that carrier testing is not always reliable if undertaken when the female is pregnant as factor VIII levels increase during pregnancy.

Finding out whether you carry the haemophilia gene is also important for your own health. There is a small chance only, but testing may reveal you are *symptomatic* and may need treatment.

### **How "bad" is haemophilia?**

In the past, haemophilia was a life-threatening disorder. Treatment improved, but bleeds into muscles and joints usually left people who have haemophilia with severe arthritis. You may have relatives with haemophilia who have joint damage because of this condition.

Now, children with severe haemophilia are treated *prophylactically*. This means they have infusions of treatment product **before** they have a bleed. In the past, people were only treated after the bleed had started. Treating before a bleed occurs, means that some young children with haemophilia have had few or no bleeds.

Children growing up with haemophilia now do have as good a chance of a long life as the next person. However, you should be aware that problems can still occur. Apart from coping with the pain caused by bleeds, children with haemophilia (and their parents) have to deal with infusions of the treatment product. Some people manage this easier than others.

Children with haemophilia, especially if they have the severe form, do need to limit some of their activities. For example, usually any type of contact sport is not on as sports such as football and rugby can increase the chances of having a bleed.

About 10% of people with haemophilia, including children, develop inhibitors to the treatment product. This means their body rejects the infusions of factor. People in this situation and their families will tell you that living with an inhibitor can be very difficult.

### **If I get pregnant can I find out whether the baby I am carrying has haemophilia?**

Yes, there are several tests available but testing does take time.

### **How much would having a child with haemophilia affect my life? For example, can I still have a career?**

This is a difficult question to answer because every child is different. In many ways, life for people with haemophilia is easier now. In the past many people with haemophilia and their families will tell you that it was a drag having to go to hospital all the time for treatment. Now, as you probably know, *home therapy* is available for many people who have haemophilia. This means they can infuse their treatment product at home, school, work, most places really. Home therapy makes it much easier for everyone. Many mothers of children with haemophilia have careers.

However, it would be unrealistic to say that haemophilia won't affect your life. Most parents with children who have haemophilia will tell you that they have had their ups and downs, especially in worrying about the child, ie. - *Will this fall cause a bleed? Will there be another bloodborne virus?*

# I'm feeling...

On finding out you carry the haemophilia gene, even if you knew there was a very good chance you do, you may experience some, or all of the following emotions:

**Confusion:** You had held out hope that the tests would show you don't carry the gene. You find it hard to believe that your instincts were wrong in this case.

This can be a difficult time. When you begin to feel like it, talk to other females in your family who carry the gene and/or try to talk it through with people who you feel close to. The counsellors at the Haemophilia Centres have a lot of experience in this area too.

**Worry:** You keep thinking of the word **haemophilia**. It seems like such a serious disorder.

You are concerned about how carrying this gene will affect your future plans.

Try to turn your worry into positive thinking time - this can help sort through how you feel. You might want to try writing down how you feel and/or talk it through with someone. Carrying the haemophilia gene won't necessarily disrupt your future plans. You can still have a partner, children, career...

**Anger:** *Why me?* is a common feeling.

There's nothing wrong with feeling angry. Try to work through your anger in a positive way: write, walk, play sport... it may take some time but the anger does usually go away.

**Denial (like you want to forget about it):** This is a tricky one. By pushing it to the back of your mind, you think (hope) your haemophilia gene will go away.

Your haemophilia gene won't go away but usually time does make it a little easier to cope with this fact. So, if you need to, give yourself a break from thinking about your carrier status for a while and when you feel ready, come back to it. You can face up to it gradually.

Remember all these are common reactions. These feelings may come weeks, even months, after the test results. You may go up and down for some time. Fear may also creep in for a while. Usually at the end of this time though, slowly, **acceptance** sets in and you will find that you can cope, and that you're okay.

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