thefemalefactors

Haemophilia testing in women and girls

Your questions answered

Genetic testing • Factor level testing

HAEMOPHILIA FOUNDATION AUSTRALIA

Haemophilia testing in women and girls

If you are a woman or girl who may be affected by haemophilia, there are tests to help with your diagnosis. These tests can tell whether you have haemophilia or carry a genetic alteration that causes haemophilia.

This information has been developed for women, girls and their parents to explain what is involved in the tests and answer some common questions.

Table of contents

Why test for haemophilia?

There are important reasons for testing for haemophilia in women and girls:

- To know whether a woman or girl is affected by haemophilia
- If a woman or girl has a bleeding tendency and/or haemophilia, she will need a treatment plan to prevent bleeding complications
- Information for pregnancy or family planning
- Preparing a safe childbirth for mother and baby
- To help with diagnosing haemophilia in other family members.

Haemophilia is an inherited genetic bleeding disorder. It is caused by a **mutation** or alteration in the DNA code for **the factor VIII/F8 or factor IX/F9 gene**. As a shorthand, this F8 or F9 gene alteration is often referred to as 'the gene'. Some people in the community call it the 'haemophilia gene'.

- Females can carry the gene alteration for haemophilia without symptoms and pass it on to their children.
- Some women and girls with the gene alteration may also have a bleeding tendency and have haemophilia themselves.

If you are female, why might your doctor suggest you be tested for haemophilia?

- There is a history of haemophilia in your family
- Or a close relative like your brother, sister or mother has recently been diagnosed
- Or your child has haemophilia or is a carrier
- Or you have bleeding symptoms that suggest you may have haemophilia.

Types of haemophilia testing:

- **Genetic testing:** to identify if a woman or girl has an alteration in her F8 or F9 gene associated with haemophilia
- **Factor level testing:** to see if a woman or girl has low levels of factor VIII or IX and may have a bleeding tendency or haemophilia.

You may need to have both genetic testing and factor level testing. For example, a woman or girl can have normal factor levels and still carry the gene alteration that causes haemophilia.

How do you know if you have the gene alteration that causes haemophilia?

There are a number of steps to identify if you have an alteration in your F8 or F9 gene that causes haemophilia. The steps are outlined here and then explained in greater detail in the rest of this education resource.

Obligate carriers

In some cases a family history of haemophilia will identify whether you have the gene alteration. In genetics this is described as being an ***obligate carrier***, because the pattern of inheritance means you **must** have the altered gene.

Obligate carriers include:

- **ALL** daughters of a man with haemophilia
- Mothers of one child with haemophilia, and who have at least one other family member with haemophilia
- Mothers of one child with haemophilia, and who have a family member who is a known haemophilia carrier
- Mothers of two or more children with haemophilia.

Gene changes

Everyone has many gene changes throughout their DNA. These gene changes are called **variants or mutations** or **alterations**. Some are common and some are rare. Some gene changes have no effect on your health, while others can cause genetic conditions, like the alterations to the F8 and F9 gene that can result in haemophilia.

What is a *haemophilia carrier*?

In genetics females who have the gene alteration for haemophilia are described as 'carriers'. Most carry the gene alteration without symptoms. Some with the gene alteration have a bleeding tendency and have the medical condition haemophilia. A female with the gene alteration can pass it onto her children, whether she has bleeding symptoms or not.

Clotting factor level tests

- Clotting factor level tests do not take the place of genetic testing. However, if factor VIII (8) or factor IX (9) tests show that you have lower than normal factor levels, this suggests that you are likely to have the gene alteration and be a carrier.
- Normal or borderline clotting factor level tests will not tell you whether you carry the gene alteration. Many women and girls have normal factor levels and no symptoms but still have the gene alteration that causes haemophilia.

Genetic carrier testing

You may need to have genetic testing to confirm if you have the altered copy of the F8 or F9 gene.

There are a number of gene alterations causing haemophilia and families will share a specific alteration.

Identifying the gene alteration in your family can be part of the process in genetic testing. If your family gene alteration is not already known, this will involve some steps before you have genetic testing:

- Usually genetic testing of a male with haemophilia in your family - or less commonly, an affected female
- Once the genetic alteration in your family is known, you can have genetic testing for the same known or familial alteration to see if you are a carrier.

How is haemophilia diagnosed?

Haemophilia is a medical condition where a person has low levels of clotting factor VIII or factor IX. These low factor levels or deficiencies are caused by gene variants or mutations in the clotting factor VIII or IX gene.

In women and girls haemophilia is usually diagnosed through:

- The physical signs that you have unusual bleeding problems

And

- Checking the family history for bleeding problems

And

- Laboratory tests on a blood sample for your clotting factor levels

And

- Genetic testing that shows you have the gene alteration for haemophilia.

Diagnostic categories for females

In the past women and girls with bleeding symptoms were generally called 'symptomatic carriers'. In Australia, diagnostic categories have now changed.[1]

These categories can be helpful for many women and girls when their results are clear but if a woman or girl has borderline factor levels, singling out which category is appropriate can be challenging.

Factor level	Diagnostic category
Females with • an F8 or F9 gene alteration • and clotting factor levels in the range for haemophilia (0-40% of normal clotting factor levels)	**Haemophilia**
• Some females who carry the gene alteration and have factor levels at the lower end of normal (40-50%) may also experience abnormal bleeding. • If further investigation indicates this bleeding is related to a factor VIII or factor IX deficiency, they will be treated as having haemophilia.	**Symptomatic haemophilia carrier**
• Most females who carry the gene alteration will have normal factor VIII or factor IX levels and will not have bleeding problems.	**Asymptomatic haemophilia carrier**

The journey to diagnosis

Haemophilia is rare and some women have not been diagnosed until later in life, although they have bleeding symptoms. There is ongoing work worldwide to increase awareness about haemophilia in females, so that women and girls can be diagnosed earlier and the pathway to diagnosis can be a smoother experience.

> *"When I was diagnosed in my 40s, I was the first person I had ever known with haemophilia. Little did I know I had had it all my life. I was unsure how I was never tested before with all the bleeding and bruising I experienced throughout my life."*

Understanding haemophilia

What is haemophilia?

Haemophilia is a genetic bleeding disorder where blood doesn't clot properly. It is caused when blood does not have enough clotting factor. A clotting factor is a protein in blood that controls bleeding.

There are two types of haemophilia. Both have the same symptoms.

Haemophilia A is the most common form and is caused by having low levels of **clotting factor VIII (8)**.

Haemophilia B is caused by having low levels of **clotting factor IX (9)**.

Haemophilia is not contagious. It is a genetic condition and a person with haemophilia is born with it.

What causes haemophilia?

Everyone has the genes responsible for making factor VIII (the F8 gene) and factor IX (the F9 gene). These factors are necessary for blood to clot.

Haemophilia is caused by a mutation or alteration in the F8 or F9 gene. This alteration interferes with the way the gene functions, so that it doesn't work as well as it should. As a result, the body does not produce enough factor VIII or IX for blood to clot properly.

- Men and boys with the gene alteration **always** have haemophilia
- Most women and girls with the gene alteration do not have bleeding problems
- Some women and girls with the gene alteration have bleeding symptoms and may have haemophilia.

How common is haemophilia?

Haemophilia is rare. It occurs in all races and socio-economic groups.

Haemophilia in males

- Approximately 1 in 6,000 males has haemophilia A
- Approximately 1 in 25,000-30,000 males has haemophilia B

Severity in males

In Australia around **53%** of men and boys with haemophilia have **mild haemophilia**.

Around **47%** of men and boys with haemophilia have **moderate or severe haemophilia**.[2]

Haemophilia in females

- Research is being conducted worldwide to understand how haemophilia affects females and how common it is.
- One study of Haemophilia Treatment Centre patients found 1.6 female carriers to 1 male with haemophilia – but noted there would also be other females who did not know they were carriers.[3]

Severity in females

In Australia **most** women or girls who carry the gene alteration for haemophilia have normal factor VIII or factor IX levels and **do not have bleeding symptoms**.

Around **20-30%** of girls or women with the gene alteration causing haemophilia have reduced factor levels and may have **a bleeding tendency**.

If a girl or woman's factor levels are in the range for mild haemophilia (5-40% of normal clotting factor), she is now recognised as having **mild haemophilia**.

Some females with factor levels at the lower end of normal (40-50%) have abnormal bleeding. If investigation shows this is related to haemophilia, they will be treated as having mild haemophilia.

Very rarely, girls or women have particularly low factor levels (below 5% of normal clotting factor) causing them to have **moderate or severe haemophilia**.[1, 4]

What happens when you have haemophilia?

A key characteristic of haemophilia is to have a bleeding tendency.

Haemophilia is a lifelong condition. It can't yet be cured, but with current treatments it can be managed effectively.

There are different levels of severity in haemophilia related to the amount of clotting factor in the blood: mild, moderate and severe.

A person with haemophilia does not bleed any faster than anyone else, but bleeding can continue for longer if it is not treated, causing poor healing. Minor bruising or scratches on the skin are not usually a problem.

For people with haemophilia, situations become more serious when there is internal or prolonged bleeding. If normal first aid does not stop the bleeding, without other treatment the bleeding can continue for days. Specialised treatment will be needed so blood can clot normally.

A problem for many people with haemophilia is internal bleeding episodes or 'bleeds' into muscles, or organs or joints, especially knees, ankles or elbows. They cause painful swelling and bruising. They can often happen as a result of injury. Some bleeds don't seem to have an obvious cause (sometimes called 'spontaneous bleeds') - this is more common in people with severe haemophilia.

Treatment

Current haemophilia treatment helps the blood to clot normally.

Treatment may be used to prevent bleeding episodes or control a bleeding episode once it starts.

For more information on treatment, see the **Treatment Plan** section on page 29.

Providing a bleeding history

One of the first steps in diagnosis will be to provide a bleeding history.

You will need to be seen in the clinic by the haemophilia team. Your doctor or haemophilia nurse will ask you a comprehensive range of questions about your medical history and your medications, including over-the-counter medications.

They may also ask you to complete questionnaires, known as Bleeding Assessment Tools.

The purpose of these questions is to identify if you have bleeding problems that suggest a bleeding disorder and what kind of bleeding pattern you have.

What are signs you might have a bleeding tendency?

- Bruising easily
- Having more painful swelling and bruising than you would expect after an injury, eg falling off a bike, car accident, sporting injury
- Bleeding or oozing for a long time after dental surgery or extractions, other surgery and medical procedures, injuries or accidents
- Internal bleeding into joints, muscles, organs and soft tissues (more common in severe haemophilia)

Women and girls may also have:

- Heavy and/or long menstrual periods. This is called **heavy menstrual bleeding** or sometimes **abnormal uterine bleeding** or **menorrhagia** and may lead to low iron levels or anaemia
- Heavy bleeding for an extended time after childbirth (particularly with delayed or late postpartum haemorrhage)

Checking the family history

Haemophilia is inherited and another important step in diagnosis is to look back over your family history to see whether there are any other family members with haemophilia or who may have had a bleeding tendency.

Haemophilia and inheritance

A male or female who has the 'haemophilia' genetic alteration in their F8 (factor VIII) or F9 (factor IX) gene can pass this altered gene on to their children.

Because haemophilia is inherited, it occurs in families, and the altered gene is passed down the generations from parent to child.

No family history?

About one third of all cases appear in families with no previous history of the disorder. This happens when a new alteration in the genetic code of the F8 or F9 gene occurs by chance in an egg cell or sperm cell. The child who is conceived will have haemophilia or be a carrier and can pass the gene alteration on to their children.

Sometimes this genetic alteration has occurred a generation or two earlier and the family has been unaware until tested. For example, the alteration might have occurred originally at the time of a mother's conception so that when the mother of a child with haemophilia is tested, she finds she carries the gene. It may be that the grandmother was the original carrier - she passed the gene alteration onto her daughter who then passed the gene alteration onto her child who is diagnosed with haemophilia.

Testing the family

If someone is diagnosed with haemophilia or as being a haemophilia carrier, it is likely that other members of their family also have haemophilia or be a carrier. Diagnosis will also include checking the family history for bleeding problems. Other family members, both males and females, may also need to be tested for haemophilia.

Clotting factor genes and families

If you are thinking about genetic testing, it can be helpful to understand how haemophilia is passed on in families.

Haemophilia occurs when you have a mutation or alteration in the gene that makes clotting factor VIII (8) or factor IX (9). This gene alteration may have taken place for the first time in your generation or many generations ago. Once the gene alteration occurs, it is passed down from parent to child through the generations, creating a family history of haemophilia. The pattern of inheritance depends on whether a person is male or female. This is because the clotting factor gene is located on a sex chromosome.

Sex chromosomes and haemophilia

We all have millions of cells that make up our body. Each cell has 23 pairs of chromosomes, which contain our genetic information or 'genes'. The genetic information determines our individual characteristics, such as the colour of our hair or our eyes. It also determines how our body functions, for example, how blood clotting works in our body.

Twenty-two of these pairs of chromosomes look the same in both males and females and are called **autosomes**. The 23rd pair differ between males and females and are called the **sex chromosomes**.

Each parent contributes one of these sex chromosomes to their children:

- Females have two copies of the **X** chromosome, and receive one from each parent
- Males have one **X** chromosome, which they receive from their mother, and one **Y** chromosome, which they receive from their father.

Haemophilia

The genes for making factor VIII (8) and IX (9) are located on the X chromosome.

When there is a genetic alteration in the F8 or F9 gene causing haemophilia, this gene alteration will be found on the X chromosome.

Haemophilia and X chromosomes

Females	Males
• Because females have 2 **X** chromosomes, they have 2 copies of both the F8 and F9 genes, 1 inherited from each parent. • As a result, women and girls with an F8 or F9 gene alteration usually have another copy that functions normally. This helps factor VIII and IX to work properly in the blood clotting process and is often enough to control bleeding. • However, 20-30% of females have problems with blood clotting. This is because of a process called lyonization or skewed **X** inactivation – see below.	• Males have 1 **X** chromosome and therefore only have 1 copy of the F8 and F9 gene. • Males with an alteration on their F8 or F9 gene on their **X** chromosome do not have another F8 or F9 gene to help with blood clotting. They will all have haemophilia.

This can be a bit hard to follow! The inheritance diagram on page 11 might help to understand how it works.

Inheritance

If you are a female who has haemophilia or are an *asymptomatic* or *symptomatic haemophilia* carrier or you are a male with haemophilia, you will have an alteration in your F8 or F9 gene. This gene alteration may be passed on to your children.

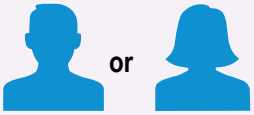
In genetics, females who have the gene alteration causing haemophilia are referred to as *carriers*.

Inheritance pattern in haemophilia

Haemophilia genetic inheritance

or

has an **X** chromosome with the "haemophilia" genetic alteration.



or

has an unaltered **X** chromosome.

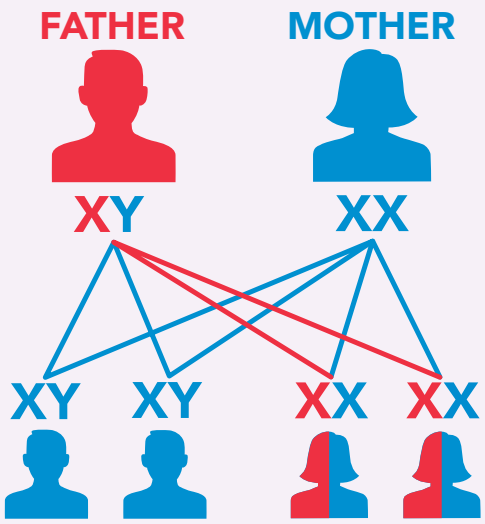
If you are a **female who is a haemophilia carrier**, there is a **50% chance with each of your pregnancies** that you will pass the gene alteration onto your baby:

- If you have a son who inherits the gene alteration, he will have haemophilia.
- If you have a daughter who inherits the gene alteration, she will be a carrier too and may have haemophilia.

If you are a **male with haemophilia**:

- **All (100%) of your daughters** will inherit the gene alteration from you. They will be carriers and some may have haemophilia
- **None of your sons** will inherit the gene alteration from you. They will not have haemophilia.

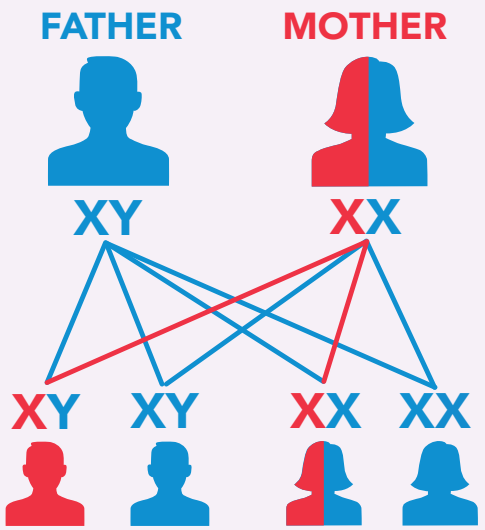
When the father has haemophilia and the mother is unaffected.



None of the sons will have haemophilia.

All of the daughters will carry the gene alteration. Some might have symptoms or have haemophilia.

When the mother carries the gene alteration causing haemophilia 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 gene alteration. Some might have symptoms or have haemophilia.

Female bleeding patterns

Women and girls may wonder why their bleeding pattern is different to the males in their family.

In families with haemophilia, males who are affected will nearly always have the same severity – for example, a grandfather and a grandson will both have severe haemophilia.

However, factor levels in females who carry the gene alteration are unpredictable and can vary between family members. For example, if two sisters have the gene alteration, one might have low factor levels and have mild haemophilia, while the other has normal factor levels and no symptoms. A father might have severe haemophilia and his daughter might have mild haemophilia

X-inactivation

This is because of a process called **X-inactivation** or **lyonization**.

X-inactivation occurs during the development of a female embryo. All females have two copies of the X chromosome. In X-inactivation each cell in the female embryo randomly turns off (inactivates or silences) most of the genes on one of the X chromosomes, including the F8 or F9 gene. Through this process only one copy of the F8 or F9 gene is switched on in each cell at any time.

What happens in haemophilia?

Because the X-inactivation process is random, usually it is an approximately 50:50 chance as to which X chromosome is inactivated. But sometimes the ratio may be skewed (for example, 70:30). This means:

- If the normal X chromosome is silenced more often than the X chromosome with the F8 or F9 gene alteration, a female's clotting factor level can be low.
- When the X chromosome with the haemophilia gene alteration is turned off more often than the normal X chromosome, a female's clotting factor level can even be at the higher end of the normal range.

This is a random process and it is different in each female. This is why two sisters who both carry the gene for haemophilia can have very different clotting factor levels. X-inactivation also helps to explain why 20-30% of women and girls who have an F8 or F9 gene alteration have reduced clotting factor levels.

When females have low factor levels, particularly when their factor levels are very low or equivalent to their male relatives with haemophilia, this may be due to other rare genetic conditions, for example, having two bleeding disorders or a different genetic disorder. This is a very complex area and the girl or woman would need to have specialised advice and genetic testing in liaison with a Haemophilia Treatment Centre.

Genetic testing and counselling

A genetic test looks for the F8 (factor VIII) or F9 (factor IX) gene alteration that causes haemophilia. This may be following a normal clotting factor level result. In almost all cases the genetic test result will give a definite answer.

Who can have a genetic test?

Current international haemophilia guidelines recommend that genetic testing should be available to:

- Females who are **obligate carriers** (see p 3)
- Female relatives of people with haemophilia or females who are potential carriers and where inheritance patterns suggest they may be at risk of having the gene alteration
- Females with bleeding symptoms that suggest they may be affected by haemophilia.[5]

Because having a genetic test has implications that last a lifetime, testing girls under the age where they can give informed consent will involve discussion with the specialist ordering the test and may need to be delayed until she can make the decision herself. The law in some Australian states does not permit a genetic test for a girl until she can give informed consent unless it is required medically. (see ***When to test*** on page 22).

Jane's story

One of the main reasons women choose to have genetic testing for the altered gene for haemophilia is to find out if they can pass it on to their children. For many women, including Jane, this is a complex issue that can stir up a lot of different feelings.

'I had been thinking about having a child, but was overwhelmed by the issues arising from my family history of haemophilia and the likelihood that I was a carrier. These issues were buried deep, and I felt that subjecting them to the light of day might unleash a tsunami of emotion and distress.'

Jane has one brother with haemophilia and a brother and a sister who are not affected. For Jane, discussing haemophilia with others would necessarily mean speaking about her brother.

'I was talking about my brother's life, and along with this perhaps my mother's greatest achievement – my brother's very survival and the maintaining of his health – and surely these were matters far too sacred to be adequately addressed in a mere "conversation".'

Then a friend suggested Jane see a genetic counsellor to help her think through some of the issues and to learn about her options around testing and pregnancy. 'Prior to that I had not even known such an avenue existed for someone like me.'

Continued on page 14 >

'First I met with a clinical geneticist. I remember how carefully she listened to me and how comforted I was by her specialised support. Then I was referred to a genetic counsellor. I was able to express my grief for my brother and my parents and the years of worry and struggle to help and treat my brother. The counsellor was incredibly helpful and supportive. He talked to me about the choices I had regarding finding out my status, including whether I even wanted to undergo testing. We also talked about what I might do with or without test results.'

Jane decided to go ahead with testing and her brother agreed to provide his genetic information. This was used to look for the genetic alteration specific to her family. The same alteration was confirmed in Jane. Looking back, she has no regrets about getting tested.

'It was the only way forward. It is so much better to live with the reality than to not know. It was an essential step in arming myself with information so that I was ready to go ahead and have a child.'

Jane and her partner met with the genetic counsellor to discuss how the gene is passed on through inheritance and the choices they had, for example, with prenatal diagnosis. They followed this with a discussion with the haematologist and haemophilia nurse at the children's hospital, to understand what the realities are for a child born with haemophilia today.

Jane now has a daughter. When she learned she was carrying a girl at the first ultrasound, 'it was almost a shock, having done so much work to prepare for the possibility of a boy with haemophilia.'

Reflecting on her experience, Jane felt that the process of genetic counselling had a very positive effect on her life.

'There was no pushing, nor judgement of me or my relationship, the time it had taken me or the age that I was, only the calmest and most helpful support that I could ever have wished for. It left me feeling ready to embrace the future which was not something I could ever remember feeling previously. I believe that my daughter was literally born out of this new hope for life.'

Where can you have a genetic test?

Testing to see if you have the gene alteration for haemophilia is complex and is only done at specialist centres. The testing is usually undertaken by your state or territory clinical genetics service and they will use a specialist laboratory associated with their service to analyse the results.

It is preferable that you have a referral for genetic testing through a Haemophilia Treatment Centre so that they can support you with current information about haemophilia and help you to navigate the process of being diagnosed. This may involve genetic counselling through a specialist clinical genetics service, but this should be done in conjunction with the Haemophilia Treatment Centre. If you are speaking to your general practitioner (GP) about genetic testing, ask them to refer you to a Haemophilia Treatment Centre for diagnosis. If your GP is reluctant to refer you for genetic testing, you can request a referral to a clinical genetics service to discuss your options.

The genetic test for haemophilia is done by looking for gene mutations or alterations in either the F8 or F9 gene. There are thousands of different changes to these genes that can cause haemophilia.

What is a proband?

A **proband** is the first person in a family to have a genetic test which identifies a family gene alteration. In haemophilia the proband is often a male with haemophilia.

How long does genetic testing take?

Finding out whether a woman or girl has the gene alteration is a process which will take time, varying from weeks to many months. It may take longer if the family alteration has not yet been identified, as the family alteration will need to be identified first.

It is important to think about genetic testing ahead of time if you are planning to have children.

How much do these tests cost?

There may be some costs involved in genetic testing, but this can vary. Speak to your HTC or the genetic counsellor about any costs involved.

What does a genetic test involve?

Genetic testing is a process which involves several steps.

The test itself is a simple blood test. However, before you have the test, it is essential that you have the opportunity for education and genetic counselling so that you can give informed consent. If the testing is for your daughter, it may need to be delayed until she is old enough to give informed medical consent herself.

1a. Information, discussion and counselling

Discussion with a haemophilia specialist and/or genetic counsellor before giving permission to have the test.

- Information about haemophilia, treatment and inheritance
- Understanding the testing procedure, the benefits and limitations of the test, and the possible consequences of test results
- Weighing up the pros and cons of genetic testing with advice and support from specialists, counsellors and other experts

1b. Mapping the family tree

- Looking at the family tree to identify other family members who may have the gene alteration

2. Giving consent to having the test

Written consent is required for a genetic test. This can be provided by:

- A woman or a teenage girl who is legally able to give medical consent.
- The parents or legal guardians of an underage girl if the test is recommended by the medical specialist and it is permitted under state/territory law.

You can choose not to go ahead with genetic testing or withdraw your consent at any stage before the result is issued by the laboratory.

3. Having a blood test

This is usually a simple blood test.

Your doctor or nurse practitioner will request the test for you and will refer you to a clinical blood taking service for the test. This may be at the same hospital or you may be referred to a local pathology service, particularly if you are in a regional area.

The amount of blood taken is not large and will vary depending on whether you are having other blood tests at the same time, eg factor level testing.

4. Genetic analysis in a laboratory

If the family mutation is known

If other members with the altered gene (for example, a brother with haemophilia) have already had genetic testing and the haemophilia gene alteration has been identified, testing is easier but may take some weeks.

Knowing the family gene alteration tells the laboratory where to look, increases the accuracy of testing and assists with interpreting your genetic test results.

If the family mutation is not known

If the specific genetic alteration in your family is not known, it can often take months to identify your family alteration and complete the tests.

Where possible, a family member who has been clinically diagnosed with haemophilia should tested first to identify the family alteration. This is usually a male family member with haemophilia.

5. Receiving the results

If you are tested through a genetic testing service, the way your genetic test results are given will be discussed and agreed on during the genetic counselling.

In all cases, you will usually receive your results in a face-to-face or formal telehealth consultation with your doctor, nurse practitioner or genetic counsellor. They will explain what the results mean and provide other education and genetic counselling, as required. If you are not already receiving genetic counselling, they can also refer you to a psychologist or genetic counsellor for further discussion.

The impact of receiving the results is very individual and will vary from person to person.

"When I had genetic testing, I went in for the blood test and got the results only a few weeks later as I was due for surgery. I had genetic counselling through the Haemophilia Treatment Centre Director. It helped to make me aware of who else in my family needed to be tested. I was the very first member of my family to have the genetic testing done and to be diagnosed."

Who do you need to tell about the test results

Family and partners

Being diagnosed with carrying the gene alteration for haemophilia can be an emotional time. You may feel you need some time and support to come to terms with it, while you decide who you wish to tell and how to tell them. Family and partners can be a great source of support, but their reactions may also be challenging. You may feel unsure how to talk about your diagnosis with them.

There is no legal obligation to tell your family or partner that you have the gene for haemophilia. However, it might be very important to the health of some relationships, particularly with the people who are directly affected by your diagnosis and where trust is involved:

- Your partner
- Family members who may also need to consider testing
- Your immediate family and other people you are close to.

If you are the first person in your family to be diagnosed with the gene alteration for haemophilia, you may be asked to tell your other family members so that they can have genetic counselling and consider testing as well. Letting family members know gives them the opportunity to find out if they have haemophilia or carry the gene alteration and whether that will affect their or their children's health. But this may be a challenging conversation for you.

Your Haemophilia Treatment Centre and genetic counselling services can assist with this:

- Information about who might be affected in your family
- Helping you to plan how to tell them
- Support for all affected family members considering genetic or factor level testing

Haemophilia is a health condition that often affects a partner and a whole family, even when they do not have haemophilia or carry the gene alteration themselves. They all feel the impact of living with haemophilia and have their own relationship with the condition. Telling them might be quite straightforward or it may not feel easy for you.

If you are concerned about telling your partner or your family about your diagnosis, consider getting some extra support. Stay in touch with the Haemophilia Treatment Centre – they can talk things through with you and support your partner and other family members too. They can also refer you to other counselling services if that would be helpful.

Telling others about bleeding disorders is a free HFA information booklet which explores some of the issues that arise for women, girls and their parents in a range of situations - with partners, family, friends, school, work and applying for insurance. It has suggestions on how to prepare to tell them, information that is useful to share with others, and tips and stories from other women and parents.

Available from

- The HFA website – www.haemophilia.org.au
- Haemophilia Treatment Centres
- Or ask HFA to post you a print copy – hfaust@haemophilia.org.au

Insurance and employers

In some circumstances, you may be asked if you have a **health condition** or have had **genetic testing**. This includes applying for insurance and to be employed by the Australian Defence Force (ADF) or the police force.

Questions about **genetic testing** often relate to the implications for health issues over your lifetime and are more likely to be part of life, income protection, disability or health insurance applications than other types of insurance.

When you are asked about having a **pre-existing health or medical condition**, this relates to having a clinical diagnosis of *haemophilia*, where you have bleeding symptoms and factor levels in the range for haemophilia.

If you carry the gene alteration and do not have bleeding symptoms (an *asymptomatic haemophilia carrier*), you do not have the medical condition haemophilia and you are not likely to develop haemophilia. In that case you do not have a medical condition that would need to be disclosed.

If you have been diagnosed a *symptomatic haemophilia carrier*, you may need to speak to an insurance company to find out if they classify this as a 'medical condition'. Full disclosure is always the best policy.

The ins and outs of disclosure

Employers

- If you have had genetic testing and are asked about this by an employer such as the ADF or police force, you are required to answer honestly.
- Employers cannot ask you to have genetic testing.
- You must provide the ADF or police force medical officer with information about any known health condition, including being a symptomatic haemophilia carrier, so they can make a decision about how relevant it is. If it is not disclosed and causes a problem, you can be discharged from the ADF or police force.

Insurance generally

- If you are asked about pre-existing health conditions and/or genetic testing when applying for insurance, you are required to answer this honestly, or your insurance policy may become void and you would not be covered for any claims.
- If you are a symptomatic carrier and the insurer refuses to cover you or charges a higher premium it would be advisable to obtain expert advice. Insurers can only apply exclusions or charge increased premiums where they can identify with statistical analysis known as 'actuarial data' that the relevant condition will lead to a greater risk of claims being lodged.

Life insurance products

(including cover for death, trauma, permanent disability and income protection)

- In Australia, life insurers can charge you higher premiums or apply exclusions because of pre-existing medical conditions.
- In certain circumstances, they may also rely on genetic testing where it indicates a likelihood of developing an illness than runs in the family.
- Life insurers can ask for or use results of genetic testing, but the guidelines recommend that they only make this request where the amount of cover being applied for exceeds certain financial limits and only where the result is relevant to the insurer's risk.

Private health insurance

- Private health insurers cannot refuse to cover you or charge higher premiums for pre-existing medical conditions (including results of genetic testing)
- They can apply waiting periods of up to 12 months for any claims relating to pre-existing medical conditions.

Travel insurance

- Affordable travel insurance for people with haemophilia can be a problem. You will need to disclose your haemophilia if asked and may need to pay a higher premium, but you may find it helpful to shop around for a travel insurer that provides the best value for you.

For more information on genetic testing and life insurance, visit the Human Genetics Society of Australasia website, www.hgsa.org.au

To test or not to test?

Many people find that undertaking genetic testing gives them a lot to think about.

There are many issues to consider for women, girls and parents of girls:

- What is the role of genetic testing in understanding a woman or girl's bleeding disorder? Will it provide helpful information now, or if she is a child, would it better to wait until she is older?
- What is the effect of genetic testing on a woman or girl's perception of herself and on her relationships, now and in the future? Does she have a partner? Would it be valuable to support her partner with information or include them in the counselling?
- What is her understanding of what it's like to have haemophilia? Has this been influenced by the experiences of family members in the past? Is she aware of current treatments and experiences of growing up and living with haemophilia?
- What will be the impact on her children and other family members? Will they need to be tested? What do they need to know? How will she tell them? Will it affect having children or family planning in the future?
- How important are her personal religious and cultural beliefs in decision-making? Would she like to speak to anyone else about it, eg a spiritual leader?
- If she has genetic testing, her results will be part of her medical record for her lifetime. She may be required to answer questions about genetic testing in the future if she is applying for certain types of insurance and occasionally in other circumstances. How will this impact on her in the future?
- What costs are involved?
- Are there any other issues relevant to her personal situation?

The haematologist (specialist doctor) or nurse practitioner at the Haemophilia Treatment Centre can help with information and advice about haemophilia, genetics and genetic testing and can provide a referral to a genetic counsellor, if needed. Women, their partner, parents or family can talk to the Haemophilia Treatment Centre or genetic counsellor individually or together prior to testing and many find it helpful. Your general practitioner (GP) or any other doctor can also refer you to a genetic counsellor.

If you start exploring genetic testing but decide against it, there is no obligation to complete the process.

For more information about genetic testing, see the Policies and Position Statements on the Human Genetics Society of Australasia web site – www.hgsa.org.au

When to test?

In Australia a common time to have a genetic test for haemophilia is when a young woman reaches childbearing age and can understand the process and implications fully and make the decision for herself.

However, some parents may wish to consider genetic testing of their daughter when she is a baby or a child.

In haemophilia there is ongoing debate about the best age for females to have genetic testing. Approaches to genetic testing of children can differ between Australian states and territories.

The legal age for informed medical consent can also vary from 16 to 18 years between Australian states and territories.

As a parent this can be a difficult discussion. It can be valuable to talk through the issues with the team supporting you during genetic counselling - your daughter's specialist doctor, the other members of the HTC team, the genetic counsellor and any others whose understanding and advice you find helpful.

The debate has centred around the benefits and harms of genetic testing for a child and concerns about delaying carrier testing. Some important issues to consider are outlined below.

Reason FOR genetic/carrier testing in childhood

- For the health benefits of the child and to help predict and manage symptoms
- So that a girl can come to terms with her diagnosis and get the information and support she needs as she is growing up
- So that a girl is aware of the risks of haemophilia for her children before she has relationships or becomes pregnant. Genetic testing takes time and can cause unnecessary stress and limit family planning options when she is already pregnant
- If she does not have the gene alteration, early testing will eliminate much stress and concern.

Reason AGAINST genetic/carrier testing in childhood

- The child may not be mature enough to understand what a carrier is or the risk to her children in the future
- The psychological harm and impact on a girl's self-esteem and sense of identity
- A genetic test is permanently part of the girl's medical record and must be disclosed in certain circumstances in the future if asked, eg applying for insurance, or employment with the Australian Defence Force or police, or to migrate to another country
- The difficulty for a parent to decide for their child whether testing their child is beneficial or harmful
- Parents may be distressed or anxious about informing their child about her carrier status
- Parents may be concerned about the impact on their daughter's prospects for marriage or having children
- Genetic testing in childhood can remove an individual's own right to choose whether they wish to obtain this information.

Sharri's story

You often hear about mothers of children with haemophilia devoting huge amounts of time to getting their child's medical issues sorted out. Frequent appointments at haematology, physiotherapy, pathology and elsewhere take time in an already busy world for parents. This is one of the reasons Sharri, whose son has haemophilia, found it hard to get on top of her own medical situation – which turned out to be a little complicated.

'After he was diagnosed at a few days old and the dust settled (several years later) I took myself off to get blood levels checked for the very first time. I didn't know I was a carrier before I had my son. My factor levels were 38% which I was told was not great but if I required any surgery probably best to check in with them first. As I was fit and healthy, I was fairly sure any surgery was not in my plans for the next 20 or so years, so I just carried on not thinking about it much.'

'I continued my haematology checks and the last one had my factor levels at just 17% and I was also diagnosed as having mild von Willebrand disorder. I still wasn't too worried as it didn't really affect me day to day. I had a hormone-releasing IUD implanted, which managed the curse of my menstruation.'

Things took a bit of a twist when Sharri needed surgery.

'I told my surgeon about my bleeding issues and his clinic communicated with the Haemophilia Treatment Centre. Turns out I definitely needed treatment product and a pre- and post-surgery treatment plan was written up. I required factor replacement therapy over the five days I was in hospital and could not go home until the last blood test said my levels were good and everyone was happy.'

Sharri hopes others will learn from her experience by taking the time to look after themselves.

'I want to encourage women who are carriers to get their factor levels checked. It has been an interesting learning experience for me because for so long it has been all about my son and managing his care, but this time - for the first time - it has been all about me.'

Factor level testing

Clotting factor level tests measure how much factor VIII or IX the body is producing. This will help to understand the risk of abnormal bleeding.

Who should have factor level testing?

Clotting factor level testing is recommended for:

- All girls and women who have the gene alteration for haemophilia
- Girls and women who are very likely to have the gene alteration because of their family's history of haemophilia
- Girls and women who have bleeding symptoms that might suggest low factor levels.[5]

Checking factor levels will help to determine if you or your daughter need a treatment plan.

It is also often the first step in finding out if a female is a haemophilia carrier. Lower than normal factor levels usually indicate a female is a haemophilia carrier if there is already a family history of haemophilia.

However, if you have normal clotting factor levels, you should not take this result to mean you aren't a haemophilia carrier. It just means you are producing normal levels of clotting factor. You will need genetic testing to confirm whether you carry the gene alteration or not.

When to have factor level testing

Ideally factor level testing should be done early in life, in case a girl is likely to have bleeding problems that need to be managed. The exact timing of the blood test is an individual decision but is recommended before she has her first menstrual period (menarche).

Factor level testing is not recommended for an embryo or fetus as it is a complex procedure with risks.

For young children, a good time to collect the blood for a clotting factor level test can be at the same time as a blood test for another reason. If your daughter is already going to have a blood test, you can contact the Haemophilia Treatment Centre who can arrange the form to collect the extra sample of blood for clotting factor testing at the same time.

As an adult you may also have factor level testing at the same time as other blood tests such as genetic testing.

Factor level testing over a lifetime

When symptoms appear

Factor level testing may be recommended if a girl or woman has abnormal bruising and bleeding which suggests haemophilia.

Periods (menstruation)

Factor level testing is important before a girl starts menstruating (getting her period) or if you are having heavy periods.

This can give a girl and her parents the opportunity to be prepared and well-informed about the possibility of heavy periods before her periods start. It also helps with a managing heavy periods once they occur. The initial treatments for heavy periods are generally similar whether or not you have a bleeding disorder, but there may be other treatment options that are appropriate if your factor level is low. Managing heavy periods with a treatment plan can make a big difference not only to your health, but also to study, work and generally participating in and enjoying life.

Surgery, medical procedures or dentistry

If you are having planned surgery or medical or dental procedures that pierce the skin, no matter how minor, it is essential that you have your clotting factor levels tested. If your level is low your Haemophilia Treatment Centre will put a management plan in place to make sure the procedure can proceed safely.

Pregnancy

It is particularly important to have had clotting factor level testing **before** you become pregnant, or as soon as possible if you did not plan the pregnancy. This is to manage any potential bleeding throughout pregnancy and childbirth:

- with invasive procedures where there is a risk of bleeding, such as in procedures with IVF (in vitro fertilisation) and prenatal diagnostic testing of the fetus
- preparing for bleeding risks during childbirth and after delivery.

Factor VIII deficiency (haemophilia A): If your factor VIII level is low and you become pregnant, you will also need more factor level testing as your pregnancy progresses. Usually factor VIII levels increase during pregnancy, so another test might need to be done in your third trimester to see if the level is high enough for a safe birth and to prevent bleeding after delivery. If not, replacement factor therapy, tranexamic acid or other treatment might be needed.

Factor IX deficiency (haemophilia B): Factor IX does not change during pregnancy, so testing does not need to be repeated.

Other times?

Talk to your haemophilia team about other times you might need to have your clotting factor level done again. This would depend on your individual situation.

Factor levels and severity

The normal level of factor VIII or IX in a person's blood is between 50% and 150%

Severity and factor level	What to expect if you are female
All severity and factor levels	• Likely to bruise easily and have prolonged bleeding after minor cuts • Likely to have bleeding problems after a bad injury, tooth extractions, surgery or medical procedures that pierce the skin
Mild haemophilia 5 – 40% of normal clotting factor	• May have heavy menstrual bleeding (heavy periods) • Might have bleeding problems with childbirth • Other than periods, might only have bleeding problems requiring medical attention very occasionally
Moderate haemophilia 1 – 5% of normal clotting factor	• Likely to have heavy menstrual bleeding (heavy periods) • Sometimes have bleeding problems with childbirth • Might have bleeding problems with minor injuries, such as sporting injuries • Occasionally have a bleeding episode for no obvious reason ('spontaneous bleeds')
Severe haemophilia Less than 1% of normal clotting factor	• Likely to have heavy menstrual bleeding (heavy periods) • Likely to have bleeding problems with childbirth • Often have bleeding into joints, muscles and soft tissues • Can have bleeding episodes for no obvious reason ('spontaneous bleeds') as well as after surgery, dental work or injuries including minor bumps and knocks.

Women and girls can have very variable symptoms with haemophilia.

What does the factor level test involve?

"Factor level testing is just like a normal blood test – it's painless and quick. The process isn't scary. You're fully informed by your haematology doctor and nurse about the results and what to do if the test returns positive."

Factor level testing is a simple blood test.

Factor level testing can be done at the blood collecting service at the same hospital as your Haemophilia Treatment Centre or you may be referred to a local pathology service.

Only a small amount of blood is taken – 3.5mls or around a teaspoon or less.

Results

When you are given your results by the haematologist or the nurse practitioner at the Haemophilia Treatment Centre, they will explain what the results mean. There may be some variation in the results, depending on the laboratory and your Treatment Centre will need to interpret this for you.

Michelle's story

Although Michelle had a family history of haemophilia, it took many years to connect the dots with her bleeding symptoms. But it was a journey that resulted in a great improvement to her quality of life.

'Haemophilia B is in my family. We often spoke at family gatherings about my grandfather who was diagnosed in the 1920s at around age 12 after falling out of a tree and spent many months in hospital. He was told by a doctor to "come back and find me if you live to see 21" – which he did.'

'I was studying Science at university, majoring in genetics, when I read in a textbook that haemophilia B is X-linked and as my grandfather and cousin have haemophilia B, there was a chance I carried the gene.

Over the years Michelle saw a few GPs about her heavy menstrual bleeding and anaemia and raised her family history of haemophilia. 'The GPs considered haemophilia too rare and unlikely to affect women to agree to a factor level test, let alone a genetic test.' Eventually in her 30s she sought help from a women's health clinic and was referred to the specialist Haemophilia Treatment Centre.

'I had counselling from the Haemophilia Treatment Centre clinical nurse consultant prior to having the genetic test.

'The clinical nurse consultant was very approachable, sensitive, knowledgeable and professional. She immediately put me at ease.'

After her diagnosis, Michelle had a hormone-releasing IUD implanted and no longer has bleeding symptoms.

Treatment plan

Each person affected by haemophilia will have their own treatment plan worked out with their haematologist (specialist doctor qualified to manage blood diseases) or haemophilia nurse practitioner that takes into account their individual type of haemophilia and bleeding pattern along with the best option for each situation. Treatment for women and girls may also involve a gynaecologist (specialist women's health doctor) if heavy menstrual bleeding is a problem.

Treatment can be given to:

Prevent bleeding episodes, for example:

- regular 'prophylaxis' (preventive) treatment
- or treatment before surgery, medical or dental procedures
- or treatment before and after childbirth
- Some women find hormonal treatments such as contraceptive pills, implants or IUDs helpful in preventing bleeding.

Control a bleeding episode **once it has started**
This is called **'on demand'** therapy.

Types of haemophilia treatment can include factor replacement therapy, non-factor therapies, hormonal treatments, and antifibrinolytic drugs such as tranexamic acid. You may be prescribed different types of treatment at different times.

Researchers are always looking for new ways to treat haemophilia and new treatments are now in development and becoming available. This includes clinical trials for gene therapy, which may in the future be able to cure haemophilia.

Australian Bleeding Disorders Registry (ABDR)

If you have bleeding symptoms, registering and staying in contact with your local Haemophilia Treatment Centre (HTC) is an important part of managing your bleeding disorder, even if you receive most of your care from another doctor, such as your general practitioner (GP).

If you are diagnosed with haemophilia or as carrying the gene, you may be asked by your HTC to register in the **Australian Bleeding Disorders Registry (ABDR)**. This is the online system used by HTCs nationally to manage and record the treatment and care of their patients.

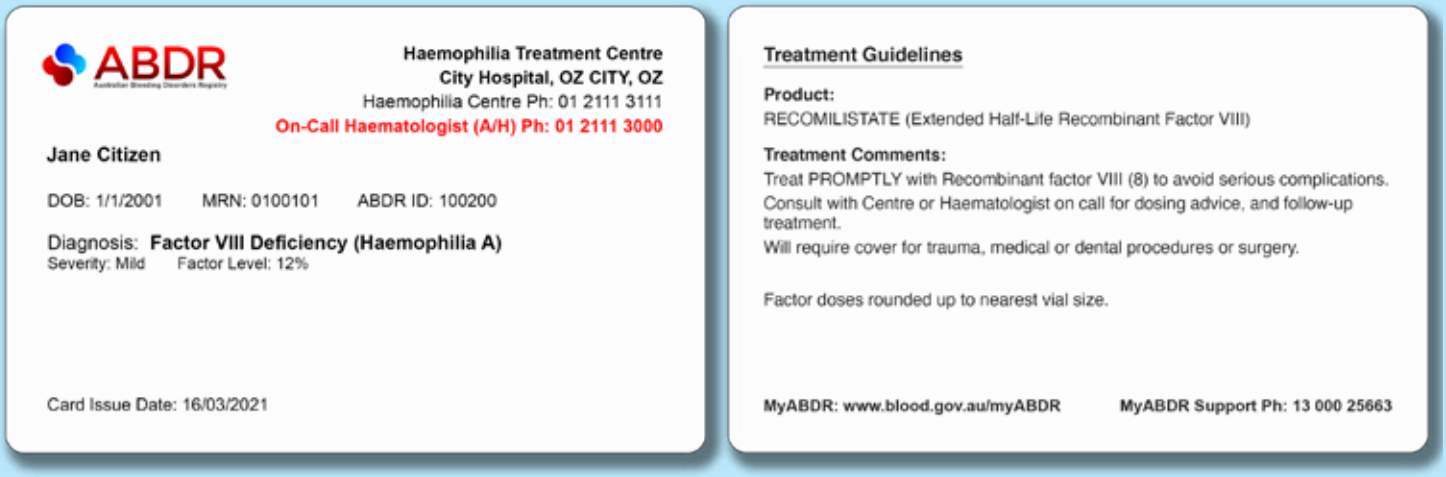
The ABDR is an important resource for people with bleeding disorders.

- Your diagnosis and treatment data is centralised and easily accessible by your treating team
- With your permission, your full data can be shared with another HTC if you move from a children's to an adult HTC, move interstate, or are travelling within Australia
- Statistics from the ABDR can support research into understanding and treating bleeding disorders, along with planning for treatment product supply nationally.

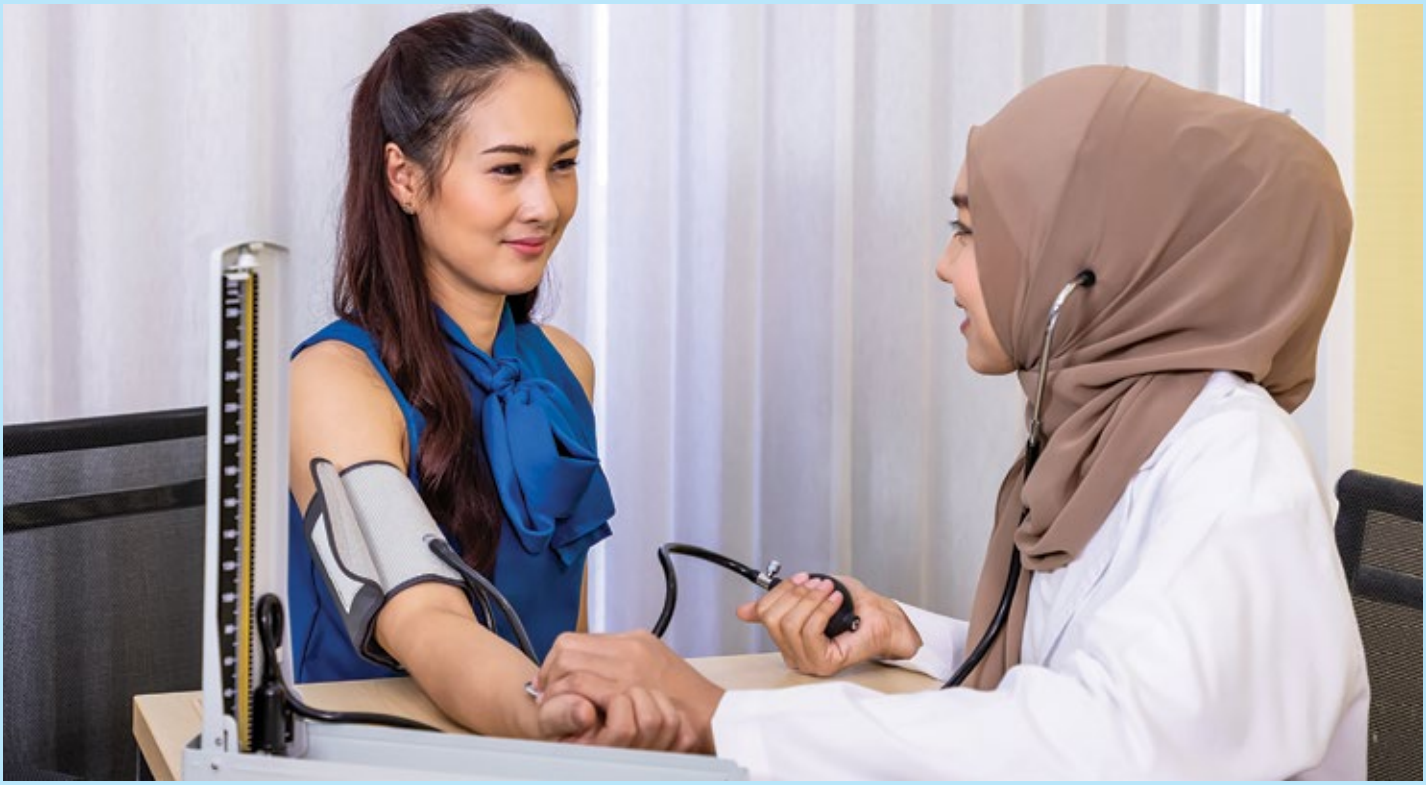
Ask your HTC about an **ABDR patient card**.
This is a wallet card which explains:

- Your diagnosis and severity
- Your treatment plan
- Who to contact for further medical advice.

Keep your ABDR patient card on you for quick reference and to show to other health professionals treating you.



Another important tool is the **MyABDR** app and website. If you are using haemophilia treatment products at home, you can use MyABDR to record your treatments and bleeding episodes and share the information with your HTC. Your MyABDR entries will be added directly to your record in the ABDR.



What can you tell your other doctors?

Bleeding disorders like haemophilia are relatively rare conditions. Most doctors and dentists may not have treated female patients with bleeding disorders before and will not be familiar with current treatment guidelines for haemophilia.

- Give your other doctors, dentist and health care providers a copy of any letters explaining your diagnosis and the contact details of your Haemophilia Treatment Centre. Ask them to liaise with your HTC.
- Show them your ABDR patient card, if you have one.
- Always inform your doctor, dentist or surgeon you have a bleeding disorder before having any medical, dental or surgical procedures, no matter how minor.
- Before you have any procedures, contact your HTC and discuss the medical support you may need to prevent bleeding complications.
- Your HTC might also have specific brochures you can take with you, eg on surgery or dentistry.
- Before you start taking anything prescribed by your doctor, naturopath or other health practitioner, check with them whether it is safe for someone with a bleeding disorder.

Where can your other doctors get information?

Your other doctors may wish to look at current evidence-based clinical care guidelines. In Australia these are produced by the Australian Haemophilia Centre Directors' Organisation (AHCDO) and can be downloaded from their website – www.ahcdo.org.au.

With your permission, your treating haematologist (specialist bleeding disorders doctor) or the team at your HTC would be happy to speak with your doctors and other health professionals about your diagnosis and treatment plan and liaise with them about your care.

How are you feeling?

Being diagnosed with haemophilia or carrying the gene can affect people in very different ways. There is no right or wrong way to feel. How you react will depend on your individual situation and what this news means to you.

You may have been tested because your baby or child has been diagnosed with haemophilia – or because haemophilia is in your family. Your testing may result from bleeding symptoms that suggest haemophilia. Or it may be your daughter who is being tested.

Feelings and reactions can be very complex. The diagnosis may have been something you expected and perhaps a relief to have your symptoms confirmed or it may have been a shock to you. You may feel guilt about your child having haemophilia or worry about the future. There can be a ripple effect to partners and family.

Support

Whatever your experience, you may need time to digest the information while you consider what this means to you or your daughter.

If you would like to talk to someone about your questions or feelings, don't hesitate to reach out for support.

Your Haemophilia Treatment Centre is there to provide information and support at any time along the way and also in the future, if issues come up for you later. They are also available for your partner and family and can talk to them separately if that works better.

The Haemophilia Treatment Centre can refer you to a genetic counsellor or other counsellors.

You can also talk to your GP and they can refer you to counselling and other services to support you.

Talking to other women – you are not alone

You may find it valuable to talk to other women with the gene alteration, who have faced similar challenges and understand how you are feeling. The Haemophilia Treatment Centre, your local Haemophilia Foundation or Haemophilia Foundation Australia may be able to put you in contact with other affected women.

Finding out you carry the gene – what does this mean to you? is a free HFA booklet which explores feelings after diagnosis, support and information, with tips and personal stories from Australian women.

Available from:

- The HFA website – www.haemophilia.org.au
- Haemophilia Treatment Centres
- Or ask HFA to post you a print copy – hfaust@haemophilia.org.au

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NB: All photos in this booklet are stock images.

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Reviewers

Australia and New Zealand Haemophilia Psychosocial Group: Nicoletta Crollini, Dr Moana Harlen, Jane Portnoy.

Australian Haemophilia Centre Directors' Organisation: A/Prof Chris Barnes, Dr Stephanie P'ng.

Australian Haemophilia Nurses' Group: Jaime Chase, Susan Dalkie, Janine Furmedge, Penny McCarthy, Joanna McCosker, Megan Walsh.

Genetics and genetic counselling: Clinical A/Prof Kristi Jones, Senior Staff Specialist in Clinical Genetics, and Lucy Kevin, genetic counsellor, The Children's Hospital at Westmead, Sydney.

Haemophilia Foundation Australia: Sharon Caris.

HFA Women's Consumer Review Group – individuals not named for privacy reasons.

Maurice Blackburn Lawyers: Katherine Bedford, Senior Associate.

More information

To find more information about haemophilia and carrying the gene alteration, or to find out how to get in touch with your local Haemophilia Foundation or a specialist Haemophilia Treatment Centre, contact:

Haemophilia Foundation Australia

7 Dene Ave Malvern East Victoria 3145

T: 03 9885 7800 Toll free: 1800 807 173

E: hfaust@haemophilia.org.au

Or visit the HFA website: www.haemophilia.org.au

Important note