thefemalefactors

Haemophilia testing in women and girls

Genetic testing | Factor level testing

A guide

HAEMOPHILIA FOUNDATION AUSTRALIA

Haemophilia testing in women and girls

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

Key points

- There are two types of haemophilia testing for women and girls
- **Genetic testing** shows if you have a gene change that is linked to haemophilia
- **Factor level testing** shows your blood clotting factor levels. If you have low levels, you may have bleeding symptoms/haemophilia.
- You may need to have both genetic testing and factor level testing.

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Why test for haemophilia in women and girls?

- To investigate whether a woman or girl is affected by haemophilia
- If a woman or girl is affected by haemophilia, she will need a treatment plan
- Preparing for puberty and possible issues with periods
- Information for pregnancy or family planning
- Preparing a safe childbirth for mother and baby
- In case she needs surgery or dentistry in the future
- To help with diagnosing haemophilia in other family members.

Meera's brother has haemophilia, but Meera doesn't have any bleeding problems. When her clotting factor levels were tested, they were normal. However, when she had genetic testing, she found out that she had a gene change for haemophilia. This was important to know because she might pass the gene change on to any children she has in the future.

Sarah has had heavy periods ever since she was a teenager, bleeds for a long time whenever she has dental work and bruises easily. Her doctor sent her to a Haemophilia Treatment Centre where they tested her clotting factor levels and found that they were lower than normal. She had genetic testing which confirmed that she had a gene change for haemophilia. Sarah was diagnosed with mild haemophilia.

The gene change she has can be passed onto her children. Her particular gene change causes severe haemophilia in males.

Now Sarah's specialist doctors have prepared an ongoing treatment plan for her so that she doesn't have heavy periods and won't have unnecessary bleeding when she visits the dentist or has surgery in the future. She has also had specialist genetic counselling on her options for planning and managing future pregnancies.

Understanding haemophilia

What is haemophilia?

Haemophilia is a bleeding disorder where blood doesn't clot properly. It occurs 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)**. It is also called *factor VIII deficiency*.

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

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

Haemophilia is not contagious. It is a genetic condition and a person with haemophilia is born with it. It can be inherited and passed on from parent to child.

What causes haemophilia?

Everyone has the gene that makes clotting factor VIII (the **factor 8 or *F8* gene**) and the gene that makes clotting factor IX (the **factor 9 or *F9* gene**). These factors are needed for blood to clot.

Haemophilia is caused by a change (mutation or alteration) in the *F8* or *F9* gene. The gene does not work as well as it should. As a result, the body does not make enough factor VIII (8) or factor IX (9) for blood to clot properly.

How common is haemophilia?

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

Haemophilia in males

Men and boys with the *F8* or *F9* gene alteration **always** have haemophilia.

- 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 males with haemophilia have **mild haemophilia**.

Around **47%** of males with haemophilia have **moderate** or **severe haemophilia**.[2]

Haemophilia in females

Researchers are currently investigating worldwide to understand how haemophilia affects females and how common it is.[3]

Severity in females in Australia

Severity in females affected by haemophilia

(*'carriers': have the F8 or F9 gene alteration causing haemophilia*)

Most have **normal factor levels** and **do not have bleeding symptoms**.

Around **20-30%** have **reduced factor levels** and **bleeding symptoms**.
They may have **haemophilia, usually mild haemophilia**.
It is very rare for females to have severe haemophilia.[1,4]

What happens when you have haemophilia?

If you have haemophilia, you will have what is called a *bleeding tendency*. This means you are likely to bleed more often than other people.

A person with haemophilia does not bleed any faster than anyone else, but bleeding can continue for longer if it is not treated. This results in 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.

Women and girls may have heavy and/or long menstrual periods. This is called **heavy menstrual bleeding** or sometimes **abnormal uterine bleeding** or **menorrhagia**. It can lead to anaemia (low levels of red blood cells or iron in the body).

Some women may also have heavy bleeding for a long time after childbirth.

Bleeding can occur internally in any part of the body, including muscles and joints, particularly after an injury or surgery. Bleeding can also occur into organs such as the brain. People with haemophilia should always seek medical advice after a blow to the head to make sure it has not caused internal bleeding.

It is important for people with haemophilia to involve their haemophilia treating team with any medical or dental procedures or surgery – anything larger than a blood test - to make sure it is managed safely.

Treatment

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

Haemophilia treatment helps the blood to clot and promotes healing.

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.*

How is haemophilia diagnosed?

Haemophilia is a medical condition where a person has low levels of clotting factor VIII (8) or factor IX (9) AND these low factor levels are caused by an alteration in the gene for clotting factor VIII or factor IX.

In women and girls haemophilia is usually diagnosed through:

- The physical signs that you have a bleeding problem

  And
- Checking the family history for bleeding disorders

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

  And
- Genetic testing that finds you have a gene alteration for haemophilia.

Providing a bleeding history

One of the first steps in diagnosis will be to look at your experiences of bleeding over your lifetime.

This may happen when you are being seen by your general practitioner (GP) or by a specialist, eg a haematologist (doctor specialising in blood diseases) or the treating team at a Haemophilia Treatment Centre (HTC).

Your doctor or the HTC nurse will ask you questions about your medical history, including your periods and any other bleeding problems you have experienced. They will also ask you about your medications, including over-the-counter medications, herbal medicines and supplements.

These questions are to see if you have bleeding problems that suggest you have a bleeding disorder and what kind of bleeding pattern you have.

What are signs you might have a bleeding tendency?

- Bruising easily
- Having more pain, swelling and/or 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 (eg, brain, stomach, uterus, etc) and other soft tissues (body tissue under the skin that supports and connects the body parts). This is more common in severe haemophilia.

Women and girls may also have:

- Heavy and/or long menstrual periods.
- Heavy bleeding for a long time after childbirth.

Heavy periods can lead to low red blood cells or iron levels in the blood (anaemia), which can cause tiredness, shortness of breath, pale skin colour and poor concentration.

Genes and inheritance

What is a carrier?

In genetics **all females who have the gene alteration for haemophilia** are described as **haemophilia carriers**, because they 'carry' the gene alteration.

Another important step in diagnosis is to draw a family tree to identify any other family members with haemophilia or who are haemophilia carriers.

Haemophilia and inheritance

Haemophilia occurs when you have a mutation or alteration in the gene that makes clotting factor VIII (8) or factor IX (9). The gene alteration is inherited and passed down the generations from parent to child.

This means that haemophilia 'runs' in families.

No family history?

About one third of all cases of haemophilia appear in families with no previous history of the disorder. This happens when a new alteration in 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 gene alteration has occurred a generation or two earlier and the family has not known about it until they are tested.

Testing the family

If someone is diagnosed with haemophilia or as being a haemophilia carrier, it is likely that other members of their family may also have haemophilia or may be a carrier. To find out other male and female family members 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.

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

Chromosomes are packages in our cells that contain our genes. They decide our individual characteristics, such as the colour of our hair or our eyes. Sex chromosomes are different in males and females. They determine which biological sex we are. They also determine how blood clotting works in our body.

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.

Inheritance

Any male or female with an alteration in their *F8* or *F9* gene can pass it on to their children.

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:

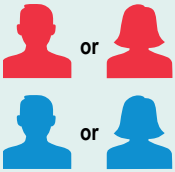
- If you have a son who inherits the gene alteration, he will have haemophilia.
- If you have a daughter who inherits the gene alteration, she will be a haemophilia carrier too and may have haemophilia.

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

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

Inheritance pattern in haemophilia

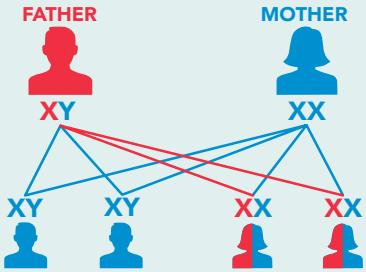
Haemophilia genetic inheritence



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

has an unaltered **X** chromosome.

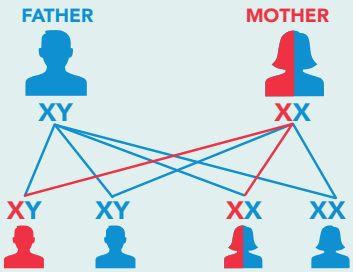
When the father has haemophilia and the mother is unaffected.



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.

Obligate carriers

Sometimes your inheritance pattern means you must have the gene alteration. In genetics this is called being an obligate carrier.

Obligate haemophilia 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 haemophilia carrier
- Mothers of two or more children with haemophilia.

Why are female bleeding patterns different?

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

- Males with haemophilia in the same family will nearly always have the same severity – for example, a grandfather and a grandson will both have severe haemophilia.
- Factor levels in females who are carriers are unpredictable and can vary even within the same family.

Nur and Fatimah are sisters who are both haemophilia carriers.

Nur has low factor levels and mild haemophilia.

Fatimah has normal factor levels and no symptoms.

Their father has severe haemophilia.

Nur and Fatimah have inherited the same 'family' gene alteration that their father has, but this gene alteration causes severe haemophilia in males.

Both Nur and Fatimah can pass on the gene alteration to their children.

- **Their male children who inherit the gene alteration will have severe haemophilia.**
- **Their female children who inherit the gene alteration may have normal or low factor levels.**

Why are factor levels unpredictable in females?

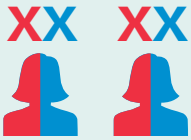
To understand factor levels in females who have a gene alteration for haemophilia, we need to look more closely at X chromosomes. Interested? Read on!

Haemophilia is due to a problem in one of the genes on the X chromosome – either the factor VIII (*F8*) gene or the factor IX (*F9*) gene.

AFFECTED MALES

A male with haemophilia (like Nur and Fatimah's father) has an **X chromosome with an *F8* or *F9* gene problem**.

Because he is male, he only has one X chromosome and so he has haemophilia.



AFFECTED FEMALES

Affected females (like Nur and Fatimah) have inherited **the X chromosome with the factor gene problem** from one parent (eg, their father).

However, females inherit two X chromosomes, one from each parent.

Usually their other parent will have **a normal X** (like Nur and Fatimah's mother).

So, although they have a **problem with a factor gene on one X**, affected females (like Nur and Fatimah) still have **a second normal factor gene on their other X**.

What happens to X chromosomes in females?

This is where it gets a bit more complicated.

In a female's body, only one of her two X chromosomes is needed in each of her cells.

The other X chromosome is permanently 'switched off' or inactivated.

This process is called **X-inactivation**.

X-inactivation is a random natural process that **happens in all females before birth**.

How does X-inactivation affect factor levels?

If a female is affected by haemophilia, she usually has **one normal X chromosome** and **one problem X** in each cell.

In some of her cells **the normal X** will be switched off

In the rest of her cells **the problem X** will be switched off.

This switching off process is random, so it is not possible to predict how many of her cells will have the normal X inactivated.

How much factor each cell makes depends on which X chromosome is switched off.

If more than half of a haemophilia carrier's normal X chromosomes are switched off, most of her cells might not be able to produce clotting factor properly. She could have low clotting factor levels and bleeding problems.

This is why two sisters who both carry the gene alteration for haemophilia can have different clotting factor levels.

50% of Fatimah's cells have an *F8* or *F9* gene that works properly.

This is enough for her blood to **clot normally**.

Only 30% of Nur's cells have an *F8* or *F9* gene that works properly.

This is why her blood doesn't clot well and she has **mild haemophilia**.

Why do some females have very low factor levels?

When females have very low factor levels, like the levels in moderate or severe haemophilia, this may sometimes be caused by other rare genetic conditions, for example, having two bleeding disorders or a different genetic disorder. Or in very rare cases, it could be that she has had the majority of her normal X chromosomes inactivated or inherited factor gene problems from both her parents.

This can be very complex and the girl or woman would need to have specialised advice, genetic counselling and genetic testing in liaison with a Haemophilia Treatment Centre and/or a Clinical Geneticist.

Genetic testing and counselling

When you have a genetic test, this is to look for the factor VIII (*F8*) or factor IX (*F9*) gene alteration that causes haemophilia.

You may already have had a clotting factor test with a normal result and need to find out if you have the gene alteration and are a carrier. The genetic test result will nearly always give you a definite answer.

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

Who can have a genetic test?

It is recommended that genetic testing should be available to females:

- who are **obligate carriers** (see page 11)
- who are relatives of people with haemophilia or of females who may be haemophilia carriers - if inheritance patterns suggest these females may also be haemophilia carriers
- who have bleeding symptoms that suggest they may be affected by haemophilia.[5]

Where can you have a genetic test?

Usually your state or territory clinical genetics service or your Haemophilia Treatment Centre will undertake your genetic testing. They can also provide genetic counselling.

They will use a specialist laboratory to analyse the results.

You will need a referral.

If you are speaking to your GP about genetic testing, ask them to refer you to a Haemophilia Treatment Centre who will advise you or arrange the appropriate testing.

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. Before you have the blood test for genetic testing, you will be offered education and genetic counselling so that you can give informed consent.

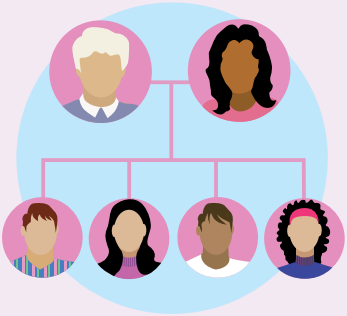
The steps in genetic testing

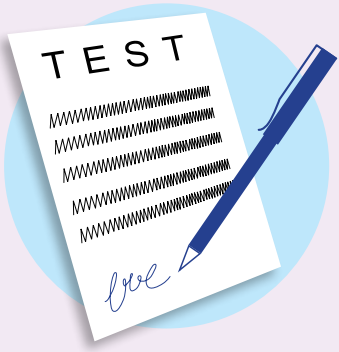
1a. Information, discussion and counselling

You will have the opportunity for discussion with a haemophilia specialist and/or genetic counsellor before giving permission to have the test.

1b. Mapping the family tree

They will look at your family tree with you to identify other family members who may have the gene alteration.



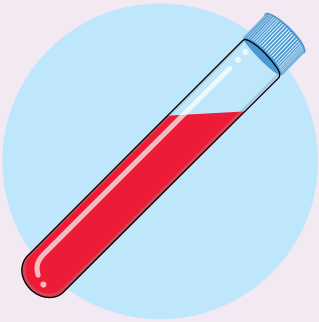


2. Giving consent to having the test

You will be asked to sign a consent form for the genetic test. 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.

A genetic test for an obligate carrier is also a paternity test and this needs to be part of the discussion.

If the testing is for your daughter, it may need to be delayed until she is old enough to give informed medical consent herself. You may choose to only have factor level testing in a child.



3. Having a blood test

Genetic testing is a blood test.

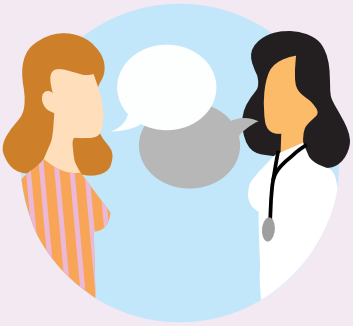
Your doctor or nurse practitioner will request the test for you and explain where you can have the test done locally.

About a teaspoon of blood is taken in the test. A little more blood may be taken if you are having other blood tests at the same time, eg factor level testing.

4. Genetic analysis in a laboratory

Genetic testing results may take weeks to months, depending on whether the family gene alteration is already known.

Where possible, a family member who has been clinically diagnosed with haemophilia should be tested first to identify the family alteration. This is usually a male family member with haemophilia. Knowing the family gene alteration makes genetic testing easier and quicker.



5. Receiving the results

If you are tested through a genetic testing service, you will discuss and agree on the way to receive your genetic test results 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 to suit your needs.

They can also refer you to a psychologist or genetic counsellor.

The impact of receiving the results is very individual and will vary from person to person. You may want to have a support person with you when you receive your results.

> 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.

To test or not to test?

Many people find that genetic testing gives them a lot to think about. For women, girls and parents of girls there are many issues to consider.

What to consider with genetic testing

- What are the benefits of genetic testing at this stage?
- Will genetic testing help to understand a woman or girl's bleeding disorder? If she is a child, will it provide helpful information now, or would it be better to wait until she is older? Would factor level testing provide enough information about her bleeding disorder at this time?
- Some women or girls with the gene alteration may not have bleeding symptoms. Will genetic testing be important to confirm whether she is a haemophilia carrier?
- 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? If she has a partner, would it be valuable to involve them?
- What is her understanding of what it's like to have haemophilia with current treatments?
- What will be the impact on her children and other family members? Could there be unexpected news from genetic testing (eg, non-paternity/different biological father) that may impact on relationships?
- 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 in her medical record for her lifetime. She may have to answer questions about genetic testing in the future if she is applying for certain types of insurance and sometimes 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?

Emma is expecting to be an obligate carrier of haemophilia as her father Steve has haemophilia. She is considering IVF treatment and has genetic testing to confirm her gene alteration.

Testing shows she does not carry the gene alteration as she expects. When she brings this up with her mother, she discovers that her biological father is not Steve but someone else.

Who can help?

- The team 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 or other counselling, if needed.
- You, your partner or family can talk to the Haemophilia Treatment Centre or the counsellor individually or together before testing. Many find this helpful.
- Your general practitioner (GP) or any other doctor can also refer you to a genetic counsellor or other counselling.

If you start exploring genetic testing but decide against it, you do not have to complete the process.

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

When to test?

There is a lot of discussion about the best age for females to have genetic testing for haemophilia. This is because the effects of having a genetic test last a lifetime.

In Australia a common time to have a genetic test for haemophilia is when a young woman reaches childbearing age and can understand what will happen with testing and what it means 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.

As a parent this can be a difficult discussion. It will be important to consider both genetic and factor level testing to understand the benefits of each test and the information they can provide. Would you choose to have both tests, or only factor level testing as a priority to manage bleeding problems?

You may find it valuable to talk through the issues with the team supporting you during genetic counselling.

This might include 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.

What are the issues for when to test?

Will there be more benefits than harm for your daughter to have genetic testing now?

- Is she mature enough to understand what it means?
- What are the health and psychological benefits for her vs the harm for her self-esteem and sense of identity?
- When is it best for her to know for her family planning – finding out the risks for her future children before she becomes pregnant?
- This will be making a decision to have a test that stays on her medical record permanently – should she be the one to choose whether she wants to have this information?
- What would be the impact on mental wellbeing for you and your daughter?

For more information, see the HFA booklet **Haemophilia testing in women and girls – your questions answered** on the HFA website, www.haemophilia.org.au.

Factor level testing

Factor level tests measure how much clotting factor VIII (8) or IX (9) the body is producing.

This will help to understand whether you or your daughter are likely to have abnormal bleeding and will need a treatment plan.

When to have factor level testing over a lifetime

It is recommended that girls and women have clotting factor level testing if they:

- Are haemophilia carriers
- Are likely to be carriers because of their family's history of haemophilia
- Prior to any surgery if they are a carrier or possible carrier
- Have bleeding symptoms that suggest low factor levels.[5]

Girls or women should have factor level testing at least once, preferably as soon as a girl shows bleeding symptoms or before she starts menstruating.

Testing may need to be repeated over her lifetime: her factor levels may change with pregnancy and hormonal medications such as the contraceptive pill and as she grows older.

Testing a fetus or newborn baby?

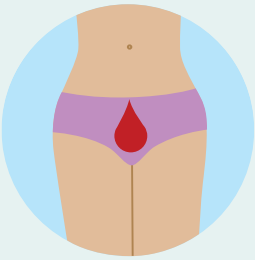
It is not recommended to do fetal blood sampling for factor levels. This is a complex procedure where blood from the fetus (unborn baby) is taken while it is still in the mother's uterus (womb) and has significant risks for the fetus.

It is generally not recommended to do routine umbilical cord blood testing or newborn tests on a female baby for factor levels. Newborn factor level testing can be challenging and in some situations factor levels can change in the first weeks or months before they settle to the level they will be in the long-term.

When symptoms appear

Testing factor levels is important if a female baby, girl or woman has abnormal bruising and bleeding that suggests haemophilia, including heavy periods, oozing or bleeding for a long time after cuts or losing teeth, or nose bleeding that lasts for 10 minutes or longer in spite of constant pressure.

Before starting periods (menstruation)



If haemophilia is a possibility, a girl should have factor level testing before she starts menstruating (getting her period).

Testing before periods start helps a girl and her parents to be prepared and well-informed.

A treatment plan can be developed to manage heavy periods.

Surgery, medical procedures or dentistry

Factor level testing is essential if you are having planned surgery or medical or dental procedures that pierce the skin, no matter how minor.

You may need a management plan from your Haemophilia Treatment Centre so that the procedure can proceed safely.

Vaccinations generally do not cause a problem when constant pressure is applied for a 2-5 minutes after the injection.

Pregnancy

Factor level testing is particularly important **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.

Other times?

Talk to your haemophilia team about other times you might need to have your clotting factor level done again.

Who arranges a factor level test?

Ask your haematologist (doctor specialising in blood disorders) or Haemophilia Treatment Centre (HTC) about factor level testing. They will advise you and can arrange testing.

If you or your daughter are not registered with a Haemophilia Treatment Centre, speak to your GP about a referral to an HTC. Your GP can also arrange factor level testing through a local pathology provider.

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 blood test.

It can be done at the blood collection service at the same hospital as your Haemophilia Treatment Centre or you may be referred to a local pathology service. In some cases the HTC may do the blood test.

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

Results

Your results will be given to you by the haematologist or the nurse practitioner at the Haemophilia Treatment Centre and they will explain what the results mean.

The way results are reported may vary depending on the laboratory and your Treatment Centre will need to interpret this for you.

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
Mild haemophilia 5 – 40% of normal clotting factor	• 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 • 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 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 • 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 bruise easily and have prolonged bleeding after minor cuts • 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 a range of symptoms with haemophilia.

> As long as I can remember, I was told that I was a carrier. My older brother didn't have haemophilia. We wrestled, rode our bikes, fought and were just kids that loved to play outside. And yes, very often I would end up covered in bruises from head to toe!!

> I mainly experience bleeding issues during dental surgery - excessive bleeding, swelling and bruising. I also had marginally heavier bleeding after childbirth than is considered normal. Occasionally I come up with bruises and I'm not sure why! But on a daily basis, haemophilia doesn't affect my life.

> I am a female with mild haemophilia A. I was only diagnosed following my son's diagnosis. If I hadn't been tested, I would never have known I was a carrier and had the condition. I get very heavy periods but I didn't think anything of it and once I had very minor surgery with an internal bleed afterwards. They were the only occasions where I have been affected.

Your diagnosis and treatment

What does your diagnosis mean?

In the past women and girls with bleeding symptoms were generally all called 'symptomatic carriers'.

In Australia, diagnosis is now more specific – but you may be wondering the terms mean.

Diagnosis	Meaning
genetic test negative	The genetic test did not find a gene alteration known to cause haemophilia. You are **not** a carrier and you **do not have** the medical condition *haemophilia*.
asymptomatic haemophilia carrier	You have the gene alteration for haemophilia but you do not have any haemophilia bleeding symptoms. You **do not have** the medical condition *haemophilia*. However, you can pass the gene alteration on to your children and some of your children may have haemophilia.
symptomatic haemophilia carrier (40-50% of normal clotting factor)	You have the gene alteration for haemophilia. Your clotting factor levels are on the borderline between normal and haemophilia, but you have haemophilia bleeding symptoms. You will be treated as having haemophilia. You can pass the gene alteration on to your children and some of your children may have haemophilia.
haemophilia and haemophilia carrier (less than 40% of normal clotting factor)	You **have** the medical condition *haemophilia*. Therefore you are also a *haemophilia carrier*. You can pass the gene alteration on to your children and some of your children may have haemophilia. Your male children may have more severe haemophilia than you do.

Treatment plan

If you have been diagnosed with haemophilia, you will have an individual treatment plan.

The plan will be worked out with your haemophilia treatment team and will cover the best option for each situation. You may be prescribed different types of treatment at different times.

You may also be referred to a gynaecologist (specialist women's health doctor) if heavy menstrual bleeding is a problem for you.

If you have anaemia (low red blood cells or low iron levels) as a result of heavy menstrual bleeding, your doctor may suggest treatment such as iron supplements.

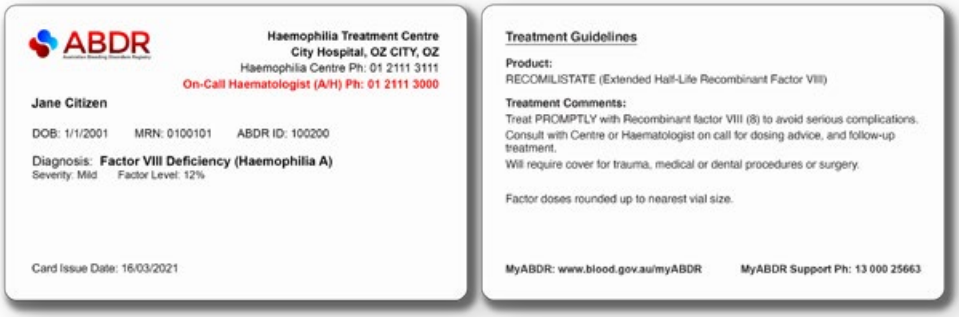
Australian Bleeding Disorders Registry (ABDR)

If you have bleeding symptoms, it is important to register and stay in contact with your local Haemophilia Treatment Centre (HTC), 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 a 'carrier', you may be asked by your HTC to register in the Australian Bleeding Disorders Registry (ABDR). This is the online system used by HTCs across Australia to manage and record the treatment and care of their patients.

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.

What can you tell your other doctors?

Haemophilia is rare. Many health care providers like doctors and dentists may not have treated female patients with haemophilia before and may not be familiar with the current treatment.

Planning is important! If you are going to have a procedure that will need preventive treatment, such as surgery, your doctors and HTC will need to organise the treatment ahead of time.

- Show your other doctors, dentist and health care providers your **ABDR patient card** if you have one.
- Ask them to liaise with your HTC.
- Always inform your doctor, dentist or surgeon you have a bleeding disorder before having any medical, dental or surgical procedures, no matter how minor.
- If you are planning surgery, contact your HTC beforehand 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 or your pharmacy whether it is safe for someone with a bleeding disorder.

Pregnancy

If you are planning a pregnancy or are pregnant, speak to your HTC. You may wish to explore your reproductive options, such as testing for the sex of the baby, finding out if it has a gene alteration for haemophilia or options for having a child without haemophilia.

Ask your obstetrics team to liaise with your HTC.

Where can your other doctors get information?

If you consent, 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. HTC contact details are on the HFA website – www.haemophilia.org.au.

Your other doctors may find other useful medical information on the Australian Haemophilia Centre Directors' Organisation (AHCDO) website – www.ahcdo.org.au.

Telling others

Who do you need to tell about the test results?

Family and partners

Diagnosis 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 want 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.

Legally you do not have to tell your family or partner that you have the gene for haemophilia. However, telling others close to you may be very important to your relationship.

Are you the first person in your family to be diagnosed with the gene alteration for haemophilia? You may be asked to tell other family members so that they can have genetic counselling and think about testing for themselves and their children.

Telling others might be quite straight-forward or it may not be easy for you.

If you are concerned about telling your partner or your family about your diagnosis, consider getting some extra support.

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

- Information about who might be affected – your partner and in your family
- Helping you to plan how to tell them
- Support for your partner
- Support for all affected family members considering genetic or factor level testing
- Referral to other counselling services

Telling others about bleeding disorders is a free HFA information booklet for women, girls and their parents. It explores a range of situations - telling partners, family, friends, school, work and applying for insurance.

Available from HFA

- Download from the website – www.haemophilia.org.au
- Or ask HFA to post you a print copy – hfaust@haemophilia.org.au

Employers

Generally you do not have to tell an employer or a prospective employer (eg, if applying for a job) about your health condition unless it will have a big ('fundamental') impact on your ability to do your job or will impact on your ability to do your job safely.

Some specific examples of situations where you need to tell an employer include applying to be employed by:

- The Australian Defence Force (ADF)
- The police force.

What do you need to do in this situation?

- When asked about your health conditions or genetic testing, you are required to answer honestly.

Otherwise you may be discharged from the ADF or police force.

Insurance

Another common question is about telling insurance companies. When applying for insurance you may be asked if you have a **health condition** or have had **genetic testing**.

What do you need to do in this situation?

- If asked, you are required to answer honestly
- Let them know if you have been diagnosed as a *symptomatic haemophilia carrier*, even though this is not the medical condition *haemophilia*
- Otherwise your insurance may not be valid.

People with the medical condition *haemophilia* may need to pay a higher premium for **life insurance** (including income protection) and **travel insurance**. Shop around for an insurer who provides the best value for you.

More information

Haemophilia testing in women and girls: your questions answered – read the *Genetic testing and counselling* section of this resource on the HFA website – www.haemophilia.org.au

Talking to your employer about illness – under *Health Topics* on the HealthDirect website - www.healthdirect.gov.au

How are you feeling?

Being diagnosed with haemophilia or as a carrier 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, why you are seeking testing now and what this news means to you and others in your life.

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 in the future, if issues come up. They are also available for your partner and family and can talk to them separately, if preferred.

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 helpful to talk to other women or girls affected by haemophilia, 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 women or girls in your situation.

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 HFA

- Download from the website – www.haemophilia.org.au
- Or ask HFA to post you a copy – hfaust@haemophilia.org.au

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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
PO Box 1208 Darling 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

NB: All photos in this booklet are stock images.

Important Note

This booklet was developed by Haemophilia Foundation Australia for education and information purposes only and does not replace advice from a treating health professional. Always see your health care provider for assessment and advice about your individual health before taking action or relying on published information.

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