

# 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.<sup>5</sup>

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.'

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*'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 be 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](http://www.haemophilia.org.au)
- Haemophilia Treatment Centres
- Or ask HFA to post you a print copy – [hfaust@haemophilia.org.au](mailto: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](http://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](http://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	Reason AGAINST genetic/carrier testing in childhood
<ul style="list-style-type: none"><li>• For the health benefits of the child and to help predict and manage symptoms</li><li>• So that a girl can come to terms with her diagnosis and get the information and support she needs as she is growing up</li><li>• 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</li><li>• If she does not have the gene alteration, early testing will eliminate much stress and concern.</li></ul>	<ul style="list-style-type: none"><li>• The child may not be mature enough to understand what a carrier is or the risk to her children in the future</li><li>• The psychological harm and impact on a girl's self-esteem and sense of identity</li><li>• 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</li><li>• The difficulty for a parent to decide for their child whether testing their child is beneficial or harmful</li><li>• Parents may be distressed or anxious about informing their child about her carrier status</li><li>• Parents may be concerned about the impact on their daughter's prospects for marriage or having children</li><li>• Genetic testing in childhood can remove an individual's own right to choose whether they wish to obtain this information.</li></ul>

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NB: All photos in this booklet are stock images.

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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:

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## Important note

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