

Finding out you carry the gene What does this mean to you?

Haemophilia - Information for women



Feelings following diagnosis

Women and girls respond in various ways when they find out they carry the altered gene that causes haemophilia. There is no right or wrong way to feel. How you react will depend on your own situation and what this news means to you. For some women this news confirms what they have thought for many years, perhaps because they have family members

with haemophilia. For others this news can come as a shock. Whatever your situation, getting support and current information can make a huge difference to how you feel. Treatment and support for people with haemophilia has come a long way in recent years. It's important to be up to date.

The following information, based on the experiences of other women and girls, describes some common reactions and where to go for information and support. The quotes are from Australian women speaking about their experiences of carrying the gene.

How do you know if you carry the gene?

Obligate carriers

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

Obligate carriers include:

- All daughters of a man with haemophilia
- Mothers of one child with haemophilia and who have another blood relative with haemophilia/who carries the gene
- Mothers of two or more children with haemophilia.

Genetic testing

In other cases, you need to have genetic testing to confirm if you carry the affected gene. If you do carry the gene, these tests also identify the specific genetic mutation associated with haemophilia in your particular family. If an affected male in your family has already been tested and the haemophilia gene mutation has been identified, testing is easier but may take some weeks. However, if the genetic alteration in your family is not known, it can take months to identify the mutation and complete the tests, as other family members may need to be tested. This is important to think about ahead of time if you are planning to have children. Genetic counselling is helpful before, during and after genetic testing to help you, your partner and your family understand what all this means.

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 may carry the gene
- Normal or borderline clotting factor level tests will not tell you whether you carry the gene. Many women and girls have normal factor levels but still carry the gene
- Genetic testing will be required to confirm whether you carry the gene or not.

For more information, see the HFA The Female Factors Carrying the Gene – Diagnosis booklet





Initial feelings

Reactions to finding out you carry the gene often depend on whether this news was expected or not. If you had reason to believe you carried the gene, perhaps because of family history, the results might not come as any surprise or you might not have any concerns. However, if this news has come 'out of the blue' you are likely to have at least some questions and concerns initially.

Sometimes a diagnosis can be upsetting, even if you suspected that you carry the gene. You might be experiencing a sense of confusion or worry about how this will affect you and your future. You could be worried that having the gene will affect relationships or decisions around having a family, including how it will affect your partner. Your concerns might be about being the parent of a child with haemophilia. Or they may be more about your own health and wellbeing.

It's really normal to feel uncertain at this early time. It's also normal for things to start feeling okay again with time and the right information and support.

Haemophilia is no-one's fault but many women and girls talk about feeling guilty. Mothers can feel guilty for passing the gene on to their child. And some females feel guilty because they believe the male members of their family who struggle with haemophilia seem more deserving of care and support.

Perhaps you feel angry this has happened to you, or just numb, not knowing where to go to from here. Or you might not have any of these concerns. Everyone is different.

“ It wasn't any great surprise - this just became a bit more information to help with decisions. ”

“ When I was diagnosed I felt gutted. ”

“ I found out that I carried the gene when I was 12 weeks pregnant. The stress was enormous as I had to make some important decisions fast. ”



Finding out when your baby is diagnosed

If you were tested because your baby was diagnosed with haemophilia, the news can feel overwhelming at first. This can be an emotional time as you work through what this means for your child, and for you and your family. It can be hard to take in a lot of information at this time but try to remember that life will settle back down again and you will be able to manage.

“ There was no history of haemophilia in my family so I was shocked when my son was diagnosed. ”

“ As time went on I felt more confident in being able to deal with what was on my plate, both for myself and for my boys. ”

“ ...at the beginning I felt guilty when family members became upset when hearing of my son’s diagnosis. I also wondered what my son’s future would be like. ”

“ It was a big shock even though my brother has haemophilia. It took me weeks to come to terms with it. ”

“ I felt frightened, and I was angry that I didn’t have a choice in being a carrier. ”

“ I was 12 when I found out and I didn’t understand the meaning of being a carrier. ”

Finding out when you were younger

Perhaps you were diagnosed as a girl or teenager. Maybe knowing since you were young has meant that it’s just part of who you are, and not something you think a lot about. Or this may have been a difficult time for you, especially if it made you feel different in some way. Everyone’s situation is different.

Heather's Story

Haemophilia runs in families, and yet sometimes it can go unrecognised for generations. This was the case for Heather who only realised after her son was born that haemophilia had been in the family for some time. Heather unexpectedly learnt she carried the gene after her baby boy was diagnosed with mild haemophilia at six months of age. "At that time I felt confused, shocked, bewildered and afraid. I had not known I was a carrier. There was no family history. I knew nothing about haemophilia. I didn't know what to expect or how to look after him. I thought my baby was 'perfect'."

Then as Heather started putting the pieces of her family's puzzle together it all fell into place. Initially this brought great sadness. "I felt bereaved all over again when I realised that this was why my father, an undiagnosed haemophiliac, inexplicably bled to death after surgery at the age of 54."

Like her son, Heather's father would have had mild haemophilia, which can be undiagnosed until there is a bleeding problem requiring treatment, for example after a serious injury or other event such as surgery. Re-thinking the loss of her father also gave Heather hope for her son. "I was subsequently comforted to think that if my dad could survive 54 years with no treatment how much better must be the prognosis for my son, given improved awareness and treatment. The more I learnt the better I felt. The prognosis was good."

As time went on Heather gathered the information and support she knew she would need to take control. "We learnt what to do when my son was injured, we connected with a supportive group of people at the Haemophilia Treatment Centre and through the local Haemophilia Foundation and our Women's group."

One thing Heather hasn't changed her opinion of is her son who is now in his early twenties. "Turns out I was right - I still think he's perfect."





When you have bleeding symptoms

Women and girls with the gene and who have bleeding symptoms might feel a sense of relief to finally know the cause of their bleeding problems. Knowing what has caused problems such as heavy periods can be the first step towards understanding the condition and how to manage it. Sometimes females worry that having a bleeding tendency will affect their general health and wellbeing, especially if other family members have had a tough time with a bleeding disorder. There are ways females with a bleeding tendency can ensure their health is well looked after. Staying in touch with the Haemophilia Treatment Centre is key to this.

“ I had a feeling I was a carrier because of some bleeding symptoms. Everything fell into place with a sickening thud. ”

Normal feelings

All the reactions mentioned so far are common feelings that women and girls experience as they come to terms with this news. It's normal for feelings to come and go over time. Your reactions might be different to these, or they might change as your life changes. Maybe starting a new relationship, or thinking about having a family will bring your diagnosis into sharper focus. Most women find things get easier as time goes on even if there are difficult times along the way.

“ I just accepted that that was part of what made me who I am. ”

“ I don't ever remember a time when I didn't assume I also carried the gene. ”

Next steps

Dealing with these feelings can be hard but there are steps you can take to lighten the load. You are not alone – information and support is available.

Getting support

You may have already talked to a genetic counsellor about the tests, or with someone from a Haemophilia Treatment Centre. Located throughout Australia, Haemophilia Treatment Centres are staffed by doctors, nurses, social workers, psychologists, and other health workers who specialise in caring for people affected by haemophilia. They provide confidential information, counselling and support to women with the altered gene, and referral to a specialist genetic counselling service if required. Haemophilia Treatment Centres and genetic counsellors can also provide information and support for anyone else who may be affected by your diagnosis, such as partners and other family members.

There can be specific times when getting support is really useful, for example if you are thinking about having children or starting a new relationship. You can follow up with your Haemophilia Treatment Centre for support at that time. Partners and family members can speak to someone too. You can also contact your state or territory Haemophilia Foundation, or Haemophilia Foundation Australia.

“ The doctor gave me my results and I just put them away. I couldn't think about it without panicking. But after a couple of weeks I decided to talk to the Haemophilia Counsellor and it was like a weight lifted off. ”

Staying informed

One of the keys to adjusting to this news is having up to date information. Knowing about the condition helps you to manage it and to take back control.

“ We don't need to deal with this condition on our own – there is lots of help and advice available to us. ”



Many women are surprised to learn that the experience of living with haemophilia has changed a great deal in recent years. Changes in testing, treatment and services have made a very big difference to people's lives. Keeping up to date helps you to deal with situations and decisions as they come up. Keep in contact with your Haemophilia Treatment Centre. They will help you stay informed. It's normal to have lots of questions - don't be afraid to seek answers. This might mean asking the same questions more than once!

Talking to others

Many women find sharing their feelings with close friends or family helps them come to terms with the diagnosis. This is often best done once you are clear about the meaning of your diagnosis.

“ I have always been pretty open about it – which has helped me accept it as being ‘just the way it is’ for me. ”

“ The guilt comes and goes depending on what’s happening in our life but I find the support I get from my family, friends and the Haemophilia Treatment Centre helps me to overcome these feelings... ”

Family members who have experience of living with haemophilia can be a great source of information and support. However, some women may choose not to turn to family members, especially in the early days. This could be because family members are also affected by the diagnosis and might have their own

reactions to work through. Who you go to for support is a personal decision. Choose someone you trust and who cares about your wellbeing. If you sense friends or family are overwhelmed, or don’t understand, you can always come back to the Haemophilia Treatment Centre.

“ I didn’t feel any different. I think my dad was more worried about me than I was. ”

Once you have talked to the Haemophilia Treatment Centre, and are clear about your own diagnosis you may find it helpful to talk to other women who have faced similar challenges. Other women who have the gene often understand the issues you face, and how you are feeling. They might have information and tips based on their own experience that you find helpful. The Haemophilia Treatment Centre, your local Haemophilia Foundation, or Haemophilia Foundation Australia may be able to put you in contact with other women who carry the gene.

“ There’s lots of support out there, and it’s great to get in touch with other women who are in a similar space to you. ”



Kirsty's Story



Kirsty has always known about haemophilia. Having other family members who were affected led her to believe she probably carried the altered gene. "As I had grown up with a brother who had haemophilia and a mum who had symptoms because she was a carrier, I was always pretty sure I carried the gene. My factor levels were first tested when I was quite young, and were found to be "borderline". You can have normal factor levels if you carry the gene so my results didn't give us a clear answer, but when DNA testing technology became available, my status was confirmed. I expected it – by that stage I had figured out that I too had symptoms that I needed to manage".

For Kirsty talking to others about her status has helped her come to terms with it. "I'm pretty upfront about it. I think that has been good because talking about it has made it feel normal for me. Talking to other women has been great too - knowing how others deal with it, how it affects their life, and what to look for is

invaluable. As a bonus, I became great friends with another woman as a result of sharing our haemophilia journeys!!!"

Dealing with health workers has sometimes had its challenges. Knowing her status has let Kirsty stay in control and to plan for a future. "It's a good thing to know about because you can take charge of your health and be informed. There have been times when I've had to stand firm with doctors and dentists who aren't as aware of the issues, and who have told me being a carrier won't affect my health. I've had to explain to them that women can have problems with bleeding too. Having all the information has also helped my partner and I make decisions going forward."

****Females with normal clotting factor levels can still be carriers. In Kirsty's case her levels were 'borderline' but she still needed genetic testing to find out if she carried the altered gene.***

Feeling hopeful about the future

Adjusting to the news you have the gene that causes haemophilia is a process. At first it can feel like your world has been rocked. Then, as you take steps towards becoming informed, things usually get easier. Finding out what you need to know, and talking to others can put you on the path to feeling in control.

If you have a child with haemophilia you will get better at managing their condition. This will come with experience, knowledge and support. You'll start to feel in control of everyday situations and you'll know where to go for additional help.

Give yourself time to adjust to your diagnosis. If you're struggling, keep in mind that life will start to feel normal again. Try not to judge how you feel, and seek information and support as you need it.

“ Knowing I carried the gene empowered me to take charge and push for the information, treatment and support I needed at the times I needed it – from friends, family, the Haemophilia Treatment Centre, and other medical practitioners, even when doctors, physios, and dentists have told me there was no issue. ”



Sharron's Story

Haemophilia has been a part of Sharron's life since childhood, but it hasn't always been easy. Diagnosed at the age of three, she often wondered why she couldn't do all the things other children were allowed to do. "My dad was known to have haemophilia. We think his mother had low factor levels due to her history, but was never formally diagnosed. I was diagnosed as a 'symptomatic carrier' as soon as I was able to have the test, when I was just three. I was very protected and resented my restrictions as a child at times. I think I felt that way because I felt different, although with my dad having haemophilia we grew up with it as a way of life. As I got older I understood my parents' concern."

Like so many women who carry the altered gene, in later years Sharron had to think about the options involved in having a family of her own. There were times when she thought it might not happen. "I wasn't sure if it would be something my future partner would want to risk. I wasn't sure with the low factor levels if there could be complications with child birth or labour. And what if I needed a caesarean? As it happened, I was very up front with my future husband, and he accepted it well and was happy to do whatever kept me out of danger".

Getting the right information and support helped Sharron and her partner make their decision. "As time went on I thought about having children. We also considered only having one child. But after some genetic counselling we decided to try naturally, and have three beautiful children, the eldest with haemophilia."



More information

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

Haemophilia Foundation Australia

T: 1800 807 173 Toll free

E: hfaust@haemophilia.org.au

W: www.haemophilia.org.au

Sources

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The Female Factors

This booklet is part of the suite of resources that will be published in *The Female Factors* project. Other resources will cover:

- A snapshot of bleeding disorders in females
- Diagnosis
- Symptoms, treatment and care
- Family planning, pregnancy and birth
- Telling others
- Information for teenage girls and young women

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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