thefemalefactors

A snapshot of bleeding disorders in females

Information for women

Haemophilia - carrying the gene

What does it mean to "carry the gene"?

Everyone has the genes responsible for making factor VIII (8) and factor IX (9). These factors are necessary for blood to clot.

Haemophilia is caused by a mutation in the factor VIII (haemophilia A) or IX gene (haemophilia B). This altered gene is commonly called the "haemophilia gene".

If you are a female who "carries the gene" or a male with haemophilia, you will have a genetic alteration in your factor VIII or IX gene and can pass this altered gene on to your children.

Haemophilia is inherited and occurs in families, where the altered gene is passed down from parent to child. However, about one third of all cases appear in families with no previous history of the disorder. This happens when a new genetic mutation occurs during reproduction. The child who is conceived will carry the altered gene or will have haemophilia.

If someone is diagnosed with haemophilia or as carrying the gene, it is likely that other members of their family also have haemophilia or carry the gene. Diagnosis will also include checking the family history for bleeding problems. Other family members, including females, may also need to be tested for haemophilia.

Bleeding symptoms

Many girls or women who carry the gene do not have symptoms of a bleeding disorder. If at least one of their X chromosomes has a factor VIII or IX gene that works, their body can usually produce normal or near normal levels of factor and they do not have bleeding problems.

However, some girls and women who carry this gene may have a bleeding tendency. They used to be described as "symptomatic carriers". If their factor levels fall in the range for mild haemophilia (5 – 40% of normal clotting factor), they are now recognised as having mild haemophilia. In very rare cases, some girls or women have particularly low factor levels causing them to have moderate or severe haemophilia. Some women with factor levels between 40% and 60% of normal also experience abnormal bleeding.

Examples of having a bleeding tendency or symptoms may include:

- Bruising easily
- Heavy menstrual bleeding. This may lead to low iron levels or anaemia
- Excessive bleeding after dental surgery or extractions, other surgery and medical procedures, injuries or accidents
- Prolonged bleeding after childbirth (particularly with delayed or late postpartum haemorrhage)
- Females with very low clotting factor levels may also have joint or muscle bleeds.

Factor level testing

All females who carry or potentially carry the gene should have testing for their clotting factor levels. Ideally this should be done early in life so that females with low factor levels are identified and managed appropriately before they have problems with bleeding.

Unlike males with haemophilia, where the factor level is nearly always the same within the same family, the factor level in females who carry the gene is unpredictable and varies between family members.

Women and girls with lower levels should have theirs checked periodically, as their factor levels may change with age, pregnancy and hormonal medications. If their factor level is low, they will need a treatment plan to prevent bleeding problems and manage any situations that occur.

Factor levels and severity

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

Mild haemophilia	**5 – 40%** of normal clotting factor
Moderate haemophilia	**1 – 5%** of normal clotting factor
Severe haemophilia	**Less than 1%** of normal clotting factor

Genetic testing

A normal factor VIII or factor IX level test will not tell women or girls whether they carry the altered gene causing haemophilia. Some women or girls may have normal factor levels, but still carry the gene.

A common time for testing whether a girl or woman carries the gene is when she reaches childbearing age and can understand the process and implications fully and make the decision for herself. Finding out whether she carries the gene is a process which will take time, sometimes many months.

The Haemophilia Centre can help with information and advice about genetic testing and can provide a referral to a genetic counsellor, if needed.

Von Willebrand disorder

Von Willebrand disorder (VWD) is an inherited bleeding disorder. People with VWD have a problem with a protein in their blood called von Willebrand factor (VWF) that helps control bleeding. They do not have enough of the protein or it does not work the way it should. It takes longer for blood to clot and for bleeding to stop.

VWD is the most common inherited bleeding disorder worldwide. It affects both females and males equally.

Symptoms

Usually VWD is less severe than other bleeding disorders, such as haemophilia. Most people with VWD have few or no symptoms. The form causing moderate bleeding problems is uncommon, and the severe form of VWD is rare. However, with all forms of VWD there can be bleeding problems.

The symptoms of VWD vary greatly from person to person. Even members of the same family may have different symptoms.

The more common symptoms are:

- Having nose bleeds often or that are difficult to stop
- Easy bruising
- Very heavy or long menstrual periods
- Bleeding for a long time with minor cuts
- Bleeding from the gums
- Bleeding after injury, surgery or dental work that continues for a long time.

Bleeding in people with VWD usually involves the mucous membranes, the delicate tissues that line body passages such as the nose, mouth, uterus, vagina, stomach and intestines.

People with severe forms of VWD may also have other bleeding problems similar to haemophilia, such as:

- Bleeding episodes that are spontaneous or happen for no obvious reason.
- Bleeding into joints and muscles which can cause swelling and pain.

Menstrual problems

Because they usually menstruate (have periods) regularly for many years of their life, females are more likely to show symptoms of VWD from an early age than males.

Heavy bleeding with menstrual periods (menorrhagia) is a common symptom of VWD for women and girls.

Menorrhagia may involve:

- Heavy menstrual periods (eg, soaking through a tampon and pad around two hourly, or needing to change during the night)
- Menstrual bleeding for longer than normal (eg, longer than 8 days)
- Bleeding with clots bigger than a 50 cent piece in size.

Heavy menstrual bleeding can lead to anaemia (low red blood cell count/low blood iron levels), with symptoms of fatigue, paleness, lack of energy and shortness of breath.

Some women and girls with VWD also experience:

- Pain during their menstrual periods (dysmenorrhoea)
- Abdominal pain and sometimes bleeding during ovulation (when an egg is released from the ovaries, around the middle of the menstrual cycle).

Although these can be symptoms of VWD, they can also be symptoms of a gynaecological disorder, so it is important to consult a gynaecologist.

If you are a woman or girl with VWD, a holistic or comprehensive care approach to your health care can help you to achieve better health and quality of life. Specialist gynaecological care over your lifetime is important to manage any gynaecological issues that occur. These may not be related to VWD, but in some cases VWD may make the bleeding problems worse.

Ideally your medical care team should work together on your health care and should include:

- A gynaecologist
- A haematologist specialising in bleeding disorders
- A GP
- A paediatrician or obstetrician, if relevant at the time.

Rare clotting factor deficiencies

A clotting factor is a protein in the blood that controls bleeding. There are a number of other bleeding disorders in females caused when their body does not produce enough of a specific clotting factor, or when the factor does not work properly. These disorders are known as rare clotting factor deficiencies.

These conditions are very rare and most are inherited in an autosomal recessive pattern. This means that the person with the rare factor deficiency has nearly always inherited a mutated or altered gene from both parents. They are often found in people whose parents are blood relatives, e.g. cousins. Both females and males can be affected by the disorders.

Factor deficiency	How rare? (estimates)	Severity of bleeding
Factor I (1): Afibrinogenemia Hypofibrinogenemia Dysfibrinogenemoa	5 in 10 million people Not available 1 in 1 million people	Usually mild, except in afibrinogenemia
Factor II (2)	1 in 2 million people	Usually mild. Moderate to severe when factor levels are low
Factor V (5)	1 in 1 million people 1 in 100,000 in some populations, including Israel, Iran and Italy	Usually mild. Moderate to severe when factor levels are low
Combined Factor V (5) and Factor VIII (8)	1 in 1 million people	Usually mild
Factor VII (7)	1 in 500,000 people	Severe when factor levels are low
Factor X (10)	1 in 1 million people	Moderate to severe when factor levels are low
Factor XI (11)	1 in 100,000 people	Mild to moderate when factor levels are low
Factor XIII (13)	1 in 3 million people	Moderate to severe when factor levels are low

Source: National Blood Authority. *Australian Bleeding Disorders Registry Annual Report 2013-14*. NBA: Canberra, 2015.

For more information on rare bleeding disorders, see the ***Rare clotting factor deficiencies*** section on the World Federation of Hemophilia website – www.wfh.org

Working with your haemophilia centre

If you or your daughter are thought to have a bleeding disorder, or to carry the gene, it is important to see a haematologist (doctor qualified to manage blood diseases) who specialises in bleeding disorders. In Australia these haematologists can be found at Haemophilia Centres. There is at least one specialist Haemophilia Centre in every state and territory in Australia, located in a major public hospital. Talk to your doctor about a referral.

Haemophilia Centres have a comprehensive care approach. This means they have a team of doctors, nurses, social workers, counsellors and physiotherapists with expertise in providing treatment and care to people with bleeding disorders. They have access to specialist laboratory and diagnostic testing and can give referrals to genetic testing and counselling services. They can also liaise with other specialities where needed, such as pain management, gynaecology and obstetrics.

Haemophilia: if you carry the gene

If you know or your family history suggests that you or your daughter might carry the gene causing haemophilia, it is important to have at least one visit to the Haemophilia Centre to discuss your individual issues, including:

- Genetic testing and its implications
- Future pregnancies and their management, if appropriate
- Factor level testing to see whether a treatment plan will be needed. This is particularly important to do before surgery or before a young girl starts menstruating, or if you are having heavy periods.

VWD and rare factor deficiencies

Diagnosis and treatment of VWD and rare factor deficiencies are a specialised area and it is important to attend a Haemophilia Centre to access a team of health professionals with expertise in providing treatment and care to people with bleeding disorders.

VWD can be difficult to diagnose and repeated testing may be needed to confirm the diagnosis. Understanding the laboratory test results is complex and needs to be done by a haematologist and laboratory with experience in VWD.

The Haemophilia Centre can work with you or your daughter to make an appropriate treatment plan and advise on ways to live well with the bleeding disorder.

Treatment plans

There are several different types of treatment for bleeding disorders. The type of treatment that will be useful may vary at different times and according to the situation. The haematologist will look at all of this when they work with you or your daughter to decide the best treatment option for each situation. They will also need to reassess the treatment at times.

Treatment may be given to prevent bleeding complications:

- To reduce or manage bleeding problems with menstruation
- In preparation for surgery, medical procedures, dental treatment or childbirth
- After an injury or accident
- If bleeding doesn't stop.

If you or your daughter have a lot of problems with bleeding symptoms, the Haemophilia Centre may ask you to visit regularly to develop and monitor a treatment plan, but this will depend on the individual situation.

Registering with the Haemophilia Centre

For women and girls who have bleeding symptoms, registering and staying in contact with your local Haemophilia Centre is an important part of managing your bleeding disorder.

If you move interstate, touch base with your new Haemophilia Centre to give them your details and see how the system works locally. **Let your Haemophilia Centre know if you change any of your contact details.**

If you or your daughter have bleeding symptoms, you may also be asked about joining the Australian Bleeding Disorders Registry (ABDR), which is used by Haemophilia Centres to manage the clinical care of their patients. With your consent, statistics that do not identify you can also be drawn from the ABDR. These statistics will be able to help improve care, for example, by assisting medical researchers to understand better how bleeding disorders affect females.

MyABDR is the app and website that people with bleeding disorders who are on home treatment use to record their treatments and bleeding episodes. Their MyABDR entries are added to their patient record in the ABDR.

ABDR patient card

If you or your daughter have bleeding symptoms and have registered for the ABDR, ask your Haemophilia Centre to organise you an ABDR patient card.

The ABDR patient card explains your diagnosis, what treatment should be given and who should be contacted for further advice. It is credit card sized to fit in a wallet. Keep the ABDR patient card on you for quick reference.

MyABDR

Jo's story*

Jo carries the gene causing haemophilia and has bleeding symptoms. With her childbearing years behind her, she is now dealing with a range of health issues very familiar to other women her age. But for Jo, managing these issues requires extra vigilance.

Medical knowledge about women with bleeding disorders among health professionals in the community is still catching up and Jo's experiences have taught her about the need to be proactive. Even recently, although her carrier status was known to her cardiologist, Jo was prescribed a combination of a non-steroidal anti-inflammatory drug and aspirin to help her heal after a heart operation and needed intervention from the Haemophilia Centre to manage the bleeding complications she experienced.

Bleeding disorders are rare in the community and it is not surprising that many of the health professionals Jo has encountered have not been familiar with the specialist care required. However, Jo has found that their attitudes can make a big difference to complications with medical procedures.

Some years ago Jo had a number of teeth extracted and bled quite badly. *"My sister had tried to tell the dentist that there was haemophilia in the family and I could be a carrier,"* said Jo, *"but the dentist said that it doesn't matter, haemophilia only affects boys."*

In contrast, she has found that when doctors speak with the Haemophilia Centre and find out more, they are more likely to learn about the complications and short-circuit any potential problems. *"My GP is a brilliant doctor but he didn't know a lot about my bleeding disorder to start with,"* Jo commented, *"but I know that he has checked up a lot in between my appointments without him even saying so because every time I see him, he has so much more information."*

Jo's experiences have taught her the importance of taking more control of her care. *"It's easy to think it's not important, or that you haven't had problems before so you won't now,"* she said. *"Or to be too embarrassed to ask questions or change doctors. But if something goes wrong, you are not in any position to argue. You need to be prepared."*

Jo's tips for women:

- Learn about your bleeding disorder and be well-informed yourself. Be vigilant about the medications you are prescribed and don't be afraid to ask questions about everything
- Contact your Haemophilia Centre first before you have a medical procedure and discuss the procedure with them and what you need to know. Ask your doctor to speak with your Haemophilia Centre before undertaking the procedure – and check that they have
- Shop around until you find a doctor who works well with you and with the Haemophilia Centre. Listen to the answers they give to your questions – if they are a bit dismissive or are fixed on knowledge they learned at university and are not prepared to talk to the Haemophilia Centre and find out more about you and your bleeding disorder, or they do not want to treat someone with a complicated health problem, it might be time to find another doctor. Your Haemophilia Centre may also be able to tell you which specialists they have worked with before.

"It's worth taking the time to find the right doctor for you," said Jo. "When you have chronic health conditions, you need doctors who can provide your care over your lifetime. And when you have a problem, knowing what to do and having the right people around you can really make a difference to your life."

* Jo is not her real name.

What do I tell my other doctors or dentist?

Bleeding disorders are relatively rare conditions. Most doctors and dentists are not familiar with the treatment and will not be aware of your individualised treatment plan.

Understanding the impact of bleeding disorders on females is quite a new area and many doctors, nurses and other health professionals do not know that females as well as males can have bleeding symptoms from haemophilia. This can be an added challenge for women and girls who have haemophilia, or who carry the gene and have bleeding symptoms.

It is important to have some personal strategies to help manage your health care when you are using services outside of the Haemophilia Centre.

These tips and personal stories give examples of the strategies some Australian women with bleeding symptoms have used.

Tips

Make sure you know what type of bleeding disorder you have and ask the Haemophilia Centre to organise you an ABDR patient card. The ABDR patient card explains your diagnosis, what treatment should be given and who should be contacted for further advice. Keep the card on you for quick reference.

Show your other doctors, dentist, and other health care providers your ABDR patient card and ask them to liaise with your Haemophilia Centre. This will help with getting appropriate treatment. It will also make it easier to obtain treatment if you need it when you are away from your usual hospital or Haemophilia Centre, for example, if you are travelling or have moved interstate or overseas.

Always inform your doctor, dentist or surgeon that you have a bleeding disorder before having any medical, dental or surgical procedures, no matter how minor.

Before you have any procedures, contact your Haemophilia Centre and discuss the medical support you may need to prevent bleeding complications. Where possible, plan this well ahead of time. The Haemophilia Centre team may also need to liaise with your surgical or dental team or other health professionals involved in your care to discuss the best approach for you individually and any pre- or post-treatment care you may need.

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. Some medicines, vitamins and supplements can interfere with blood clotting and healing, or can irritate your mucous membranes such as your nasal passages or stomach lining. This includes some herbal and homeopathic medicines and others available over-the-counter, such as aspirin and non-steroidal anti-inflammatory drugs like ibuprofen. Speak with your haematologist about medicines to be cautious with or to avoid.

Sharron's story

Sharron has mild haemophilia A. As a child her factor levels were considerably lower than they are now, but even though they have increased as she matured, they still remain in the mild haemophilia range.

"I have had the advantage of knowing from a young age that I had low factor levels and needed to advocate for myself if my parents weren't around, especially as many health professionals didn't understand that females could have haemophilia too," explained Sharron.

For Sharron, this has meant involving her Haemophilia Centre in her care.

"I had to have a tooth out about 18 months ago, and I told the dentist I would need to have it removed at the hospital where my Haemophilia Centre is. We organised a referral to the dental department there, and all went well. A recent gastroscopy was also organised through this hospital and the Haemophilia Centre was involved in pre- and post-procedure care, as with the dental procedure. My care in both those cases was tailored to my haemophilia."

What does Sharron think works best for her?

"I always ring my Haemophilia Centre and talk to one of the team before any procedure is booked. Sometimes in the past I have needed an update on my factor level before a procedure. I always let a new GP or specialist know up front about my haemophilia. I had plans in place with the obstetrician and HTC working together for each of my three children."

This also involves recognising the limitations of some health services.

"Sometimes I have made compromises to fit in with my care," she commented. *"For example, when living in a regional centre we decided I couldn't have a hysterectomy for heavy periods there, and I had an endometrial ablation instead (burning off the lining of the uterus) and at the same time I had coils put into my fallopian tubes for permanent contraception instead of having a laparoscopy (little cuts in my tummy) to tie my tubes . It was brilliant. I still had treatment but it was managed locally."*

Sharron's tips for women:

- Always know your factor level
- Always work with your doctor and the Haemophilia Centre. Liaise with your Haemophilia Centre to see where and how the surgery or procedure needs to be carried out. I usually say to my GP or surgeon/physician, "I will contact my Haemophilia Centre before we book that."
- Have a plan to manage your care, whether it is at a major centre or rural hospital
- Always look after your health so you are able to be there for your family. It teaches others to do the right thing for themselves too in relation to their bleeding disorder
- Contact your local Haemophilia Centre for advice. If you aren't sure how to contact the Haemophilia Centre, ring your local Haemophilia Foundation or, if you have a child with a bleeding disorder, ask your child's haematologist.

You are not alone

Although bleeding disorders may only affect women and girls at certain times in their life, they and their parents often comment that it is helpful to talk to others in a similar situation and know that they are not alone. Haemophilia Foundations are a great way to connect with others and share experiences.

State and Territory Haemophilia Foundations have:

- Newsletters and web sites to update people with bleeding disorders and their partners, families, friends and carers
- Social activities where you can meet, talk about common experiences and enjoy a meal or a day out, such as family camps, community picnics and BBQs, women's groups, grandparents' groups.

Haemophilia Foundation Australia also supports:

- A national website with information and personal stories for women with bleeding disorders
- Social media sites, such as Facebook and Twitter
- A youth program (***Youth Lead Connect***) and a website (***Factored In***) for young people affected by bleeding disorders.

For more information, visit

- ***HFA*** website – www.haemophilia.org.au
- ***Factored In*** youth website – www.factoredin.org.au

More information

For more information about bleeding disorders, or to find out how to get in touch with your local Haemophilia Foundation or a specialist Haemophilia Centre, contact:

Haemophilia Foundation Australia
T: 03 9885 7800 Toll free: 1800 807 173
E: hfaust@haemophilia.org.au
W: www.haemophilia.org.au

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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.