Working with your Haemophilia Treatment Centre

If you have a bleeding disorder, or carry the gene, it is important to see a haematologist who specialises in bleeding disorders. In Australia these haematologists can be found at Haemophilia Treatment Centres. There is at least one specialist Haemophilia Treatment Centre in every state and territory in Australia, located in major public hospitals.

Going to a Haemophilia Treatment Centre

If you have bleeding symptoms that are causing problems for you, your doctor may suspect that you have a bleeding disorder and refer you to a Haemophilia Treatment Centre. Or you may have a family history of males and females in your family with a bleeding disorder. If you have bleeding symptoms as well, it is recommended that you or your parents talk to your GP (family doctor) about a referral to a Haemophilia Treatment Centre.

Diagnosis and treatment of haemophilia, von Willebrand disease (VWD) and rare clotting factor deficiencies are a specialised area of medicine.

It is important to have a referral to a Haemophilia Treatment Centre where a team of health professionals who are experts in providing treatment and care to people with bleeding disorders can make sure you're getting the best care and information. In Australia nearly all of these Centres are part of public hospitals, where attendance is free for Australians with bleeding disorders and their close relatives.

What is comprehensive care?

Haemophilia Treatment Centres have a comprehensive care approach.

This means:

- They have a team of haematologists (doctor qualified to manage blood diseases), nurses, social workers, psychologists and physiotherapists who have expertise in caring for people with bleeding disorders and their partners and families.
- The Centre has access to specialised laboratory and diagnostic testing and can give referrals to other services such as genetic testing and counselling.
- The team can work with other services you might need such as paediatricians (children's health doctors), gynaecologists (women's health specialists) and obstetricians (for pregnancy and childbirth).
- **Comprehensive care is important for anyone with a bleeding disorder.**

If you are 16 years or older you can attend the Haemophilia Treatment Centre on your own but often it is useful to bring along a support person such as a parent, or other family member. This decision will depend on factors such as your ability to understand your condition, and whether you feel confident to ask questions and make decisions about your own health care. You might simply like some support during your appointment. You can always call the Haemophilia Treatment Centre first to discuss whether to come in on your own or with support.

It's often helpful to write down any questions before your appointment and bring them with you. That will help you remember to ask about things that are important to you.

As well as the care from your Haemophilia Treatment Centre, it is important to have specialist gynaecological care over your lifetime to manage any women's health issues that occur. These may or may not be related to your bleeding disorder, but in some cases having a bleeding disorder (or bleeding symptoms) may make a gynaecological condition worse.

Your medical team

Ideally all your doctors should work together on planning your health care. The team could include:

- A haematologist (doctor qualified to manage blood diseases) and who specialises in bleeding disorders.
- A gynaecologist (women's health specialist)
- A general practitioner (your GP or family doctor)
- A paediatrician (children's health specialist)
- An obstetrician (pregnancy and childbirth specialist) when this is relevant to you or your family situation.

Your regular GP is important for looking after aspects of your health that may not be associated with your bleeding symptoms and to recognise when it's time to involve the Haemophilia Treatment Centre.

"I would highly recommend anyone who thinks their bleeding isn't normal to seek further help. Speak to the team, get whatever help they can offer."

"Listen to your body. YOU are the expert when it comes to YOUR bleeding symptoms. If you think something isn't right, speak up and see someone."

Diagnosing a bleeding disorder

Bleeding disorders can be difficult to diagnose. 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 bleeding disorders.

Diagnosis can be tricky if you have heavy bleeding with your periods and a bleeding disorder. It can be hard to know if:

- Your bleeding disorder is causing the heavy periods

  Or
- You have a women's health problem that is unrelated to your bleeding disorder.

  Or
- Your bleeding disorder is making another problem worse.

The timing of the testing can also be important. Some tests can be affected by the natural rise and fall of your hormones in your menstrual cycle or taking the contraceptive pill and other factors such as stress, exercise or pregnancy.

Your GP or your Haemophilia Treatment Centre might refer you to a gynaecologist to help understand what is happening with you. This is an important step to rule out any other problems or issues that may not be related to your bleeding problems. It also allows your GP, Haemophilia Team and gynaecologist to work as a team. They will communicate with you and each other about the diagnosis and a treatment plan tailored to you and your situation. Sometimes it can take a while to unravel your diagnosis and to develop a treatment plan that works best for you. This might require more tests and trying out different medications, doses and even combinations of medications.

Diagnosing VWD

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 specialists with experience in VWD.

Many people's symptoms are mild and they may not be diagnosed until they have a major bleeding problem such as surgery or an injury. However if they have a severe form of VWD, they will usually have major bleeding problems as a baby or small child and will often be diagnosed within their first year of life.

Diagnosing VWD involves:

- **A personal history of abnormal bleeding or bruising from mucous membranes** (such as the nose, mouth, uterus, vagina, stomach and intestines) or from the skin after injury, trauma or surgery *and*
- **A family history of bleeding problems** *and*
- **Specialised laboratory test results for VWD**

Pregnancy and testing

Most women with VWD do not have a problem with delivering a healthy baby. With the most common form of VWD (mild type 1), pregnancy will very often cause blood levels of VWF (von Willebrand factor) to increase into the normal range by the last three months of pregnancy, so that bleeding complications during pregnancy and childbirth are far less likely. Women with rarer or more severe forms of VWD are much more likely to need treatment to reduce the risk of bleeding problems during delivery.

It is a good idea to talk to your HTC and your obstetrician before you get pregnant, and to ask them to liaise with each other. You might like to speak to a genetic counsellor. Ask your haematologist if there needs to be any special care with having tests like amniocentesis. If you have VWD and are pregnant, you would need to be monitored and have blood tests for your VWF levels during your last three months of pregnancy. This is to plan for childbirth, a safe delivery for you and your baby and for any treatments you might need to prevent possible heavy bleeding in the weeks after delivering the baby when your factor levels return to their usual level.

How were you diagnosed?

"I was diagnosed as a baby. I had fallen and cut my lip and it bled all night. My mum woke to me in my blood-soaked cot and I was taken to hospital by ambulance. I was diagnosed by the time we left the hospital."

Diagnosing haemophilia

If you know, or your family history suggests, that you might carry the gene causing haemophilia, it is important to have at least one visit to the Haemophilia Treatment Centre. This is to discuss your individual health situation and can include:

- **Clotting factor level testing to see whether you need a treatment plan.**

  Factor level testing is a good idea before you start menstruating (getting your period), or if you are having heavy periods. The initial treatment options for heavy periods are generally similar whether or not you have a bleeding disorder, but there may be other treatment options that are appropriate if your clotting factor level is low. Factor level testing is also really important before medical procedures such as dental work or surgery. Just keep in mind that having a test for clotting factor levels does not tell you if you carry the gene for haemophilia.

  You can have normal levels but still carry the altered gene.

- **Genetic testing to see if you have the gene**
  This includes discussing the pros or cons about having a genetic test to know for sure before you have testing (sometimes called 'genetic counselling'). Then if you go on with testing, it will identify whether you have the gene or not.

- **Planning a pregnancy and things to think about.**
  You can find out options to reduce the chance of passing the gene onto your children, and how to plan for a safe pregnancy and delivery. It's really important that you find out about these options **before** you become pregnant.

How were you diagnosed?

> "My grandfather was a haemophiliac, so I had clotting factor tests done at 5 years and genetic testing in my 20s."

> "We received additional genetic testing when I was 20 roughly which determined we were in fact carriers."

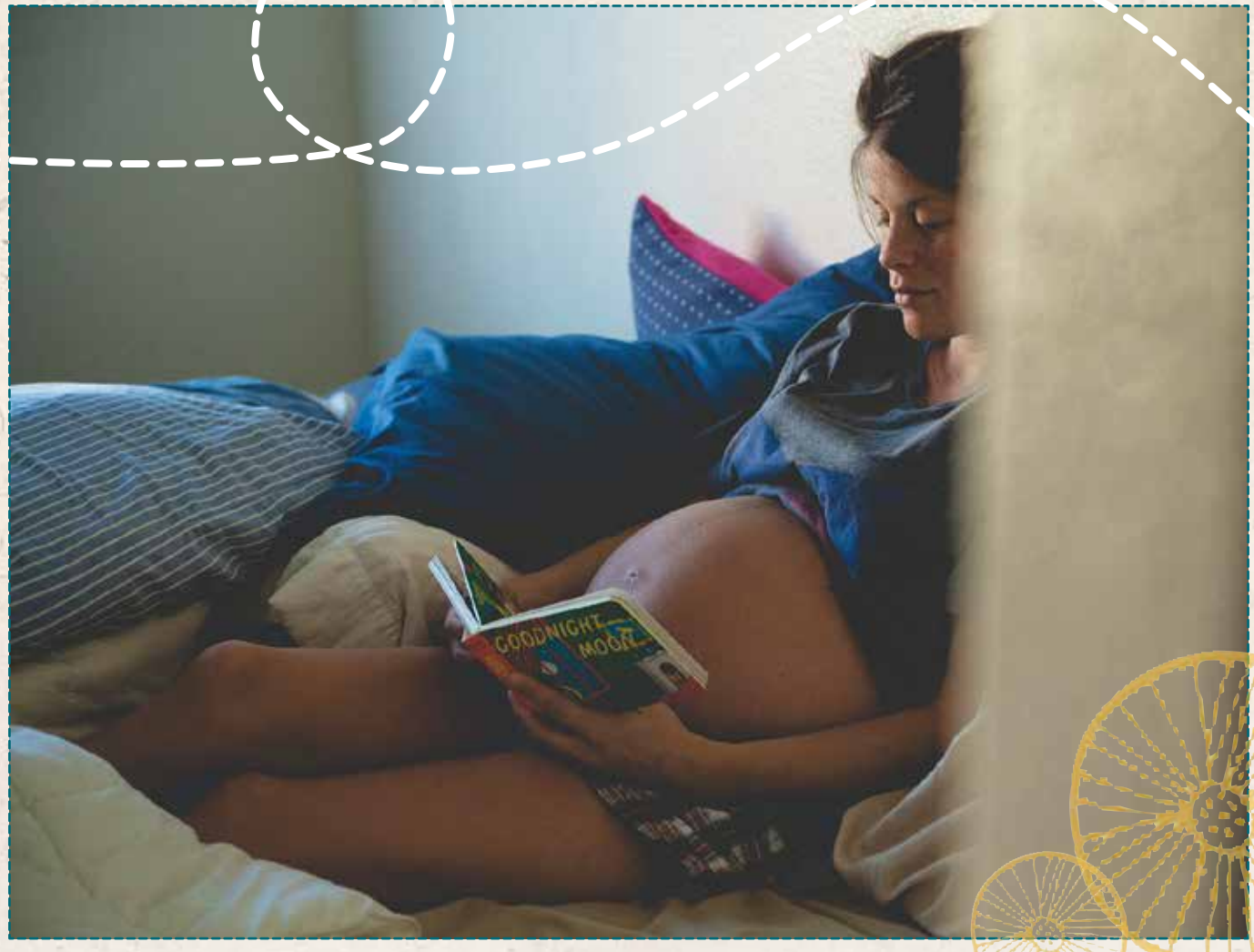
When to have clotting factor level testing

Clotting factor level testing is recommended for:

- All girls and women who have the gene for haemophilia
- Girls and women who are very likely to have the gene because of their family's history of haemophilia.

Ideally factor level testing should be done early in life, but the exact timing of the blood test is an individual decision.

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



Surgery or dentistry
If you are having planned surgery (such as removal of tonsils and adenoids) or dental procedures you **must** have your clotting factor levels tested. If your level is low your Haemophilia Treatment Centre will put a management plan in place to make sure the procedure can proceed safely.

Before periods start
You and your parents may also prefer to have your factor levels checked before your periods start, to be prepared and well-informed about the possibility of heavy periods.

Pregnancy
It is particularly important to have had clotting factor level testing before you become pregnant. If your factor VIII level is low and you become pregnant, you will also need more factor level testing. Usually factor VIII levels go up during pregnancy, so another test might need to be done to see if the level is high enough for a safe birth. If not, clotting factor concentrates or other treatment might be needed. Factor IX does not change during pregnancy so testing does not need to be repeated.

Other times for testing
Talk to your haemophilia team about other times you might need to have your clotting factor level done again. This would depend on your individual situation, and could relate, for example, to treatment, pregnancy or surgery.

It is important to remember that **normal factor VIII or factor IX levels will not tell you whether you have the altered gene causing haemophilia**. Many females who carry the gene have normal factor levels. Finding out your clotting factor level helps you and your Haemophilia Treatment Centre decide if you need to develop a treatment plan specifically for you. If your levels are normal a separate genetic test is needed to find out if you have the gene.

Genetic testing for haemophilia

Genetic testing shows if a girl or woman has the altered gene for haemophilia.

Deciding to have genetic testing is a personal decision and needs to be thought about carefully. The Haemophilia Treatment Centre can help with this.

A common time for genetic testing is when a young woman reaches childbearing age. It is important you understand the process and the meaning of genetic testing. This includes understanding how knowing your status might affect you, particularly in relation to current or future relationships, and for possible pregnancies.

Finding out whether you have the gene is a process which takes time, sometimes many months. Knowing before you get pregnant gives you the chance to think about family planning options that can lower the chance of passing on the altered gene. Many of these options aren't available if you don't know whether you have the gene before becoming pregnant.

Talk to your Haemophilia Treatment Centre about when testing is right for you. The Haemophilia Treatment Centre can help with information and advice about genetic testing and can provide a referral to a genetic counsellor, if needed. Remember: genetic testing is not the same as clotting factor testing which only tells you whether your clotting factor levels are within normal range. Clotting factor level tests do not test for the gene.

For **more information about genetic testing and counselling**, visit the **Women with bleeding disorders** section on the HFA website - www.haemophilia.org.au/women.

Diagnosing rare clotting factor deficiencies

Most rare clotting factor deficiencies are diagnosed through a variety of blood tests, including tests to measure the amount of particular clotting factors in the blood. It is best if these are ordered by a specialist doctor (haematologist) at a Haemophilia Treatment Centre, who will use a specialist coagulation laboratory to do this kind of testing. The haematologist may need to request very specific tests, for example, a test for factor XIII (13) deficiency because factor XIII deficiency will not show up in routine clotting factor tests. The tests are complex and the specialist doctor will have to look closely at the results and may need to rule out other bleeding disorders.

How were you diagnosed?

> "I had a heel prick as a baby and it wouldn't stop bleeding."

Diagnosing inherited platelet disorders

To have an inherited platelet disorder diagnosed, you will need to see a specialist doctor at a Haemophilia Treatment Centre. The specialist will talk to you, and your parents if they are present, about your health and history of bleeding.

You will also need to have a series of blood tests, including tests that measure how well the platelets "aggregate" (stick to each other) in response to various stimuli (things that cause a reaction). Quite a few blood tubes need to be collected for these tests and the results can be affected by many factors in the process of collection, transport to the laboratory and testing. This may mean that repeated testing is necessary before your doctor is satisfied that a diagnosis of a platelet function disorder is appropriate.

Treatment plans

Most women and girls with bleeding disorders don't need regular treatment, but it is important that they have a treatment plan ("action plan") in case urgent treatment is needed (e.g., for urgent surgery or following an injury).

There are a number of situations where treatment may be required:

- To reduce or manage bleeding problems with your periods
- To reduce or manage other bleeding problems (eg. recurrent nosebleeds)
- In preparation for surgery, medical procedures, dental treatment or giving birth
- After an injury or accident

There are several different types of treatment for bleeding disorders.

The type of treatment that will be most useful may vary at different times and will depend on your individual diagnosis and situation. Your haematologist will look at all of this and will involve other doctors (for example a gynaecologist) and members of the Haemophilia Team (for example a physiotherapist). Together they will consider your thoughts and wishes before recommending the best treatment option for you. They may also need to reassess the treatment plan from time to time.

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

How does your treatment plan help you?

"Having a treatment plan meant I was no longer missing out on events, days at work and life in general because of bleeds. I was able to start living a "normal" life, and have treatment when needed."

"Recently my treatment plan changed to include prophylaxis after I developed joint damage. Starting prophylaxis has helped me recognise the minor bleeding issues I had been ignoring now that they are absent."

"Try to be as open as possible about your symptoms with your HTC team. They cannot provide appropriate treatment for symptoms that you keep to yourself."

What are the types of treatments used?

- **Protection, Rest, Ice, Compression and Elevation (PRICE):** These steps are important to minimise swelling or bruising. PRICE is a simple treatment for minor soft tissue injuries that may lead to bruising or haematoma (large bruises with a lump).
- **Tranexamic acid** is an oral medication (anti-fibrinolytic) that can be useful in managing heavy periods, nosebleeds and after minor injuries or procedures in the mouth, eg dental work.
- **Hormonal therapies** (for example, the contraceptive pill or the Mirena IUD/ intrauterine device) for problematic menstrual bleeding are prescribed with input from a gynaecologist.
- **DDAVP (Desmopressin)** is a laboratory-made version of a hormone that raises the levels of factor VIII and von Willebrand factor. It may be useful in managing bleeding or before surgery in girls and women who have a reduced factor VIII level or who have certain subtypes of VWD. It sometimes works well in treating platelet storage pool deficiencies. Not all young women and girls respond to this medication, so your Haemophilia Treatment Centre may arrange for you to have a test dose first with some blood tests afterwards to check your response.
- **Clotting factor concentrates** may be required to treat or prevent bleeds, or before surgery in girls and women with haemophilia and with some rare clotting factor deficiencies. It's also given to young women and girls with VWD who do not respond adequately to DDAVP. This is infused (injected) into a vein. Multiple infusions may be needed over a number of days depending on your clotting factor level and the procedure being done. Some girls and women with severe bleeding disorders have 'prophylaxis', which is regular treatment to prevent bleeds.

Factor concentrates for haemophilia A and B are 'recombinant', which means they are synthetic, made by genetic engineering. The factor concentrate that is currently available for VWD in Australia is made from the plasma (pale yellow fluid part) in human blood, but is then highly purified and treated.

In some rare bleeding disorders a specific clotting factor concentrate has not yet been developed or is not suitable for treatment. Other blood products may be used:

- **Fresh frozen plasma** is made from the plasma (pale yellow fluid part) in human blood. It contains the range of proteins (factors) needed to help blood to clot. It is stored frozen and thawed for treatment, when it is infused (injected) into a vein. In Australia it is sometimes used to treat rare clotting factor deficiencies, such as factor V (5) deficiency.
- **Cryoprecipitate** is also made from the plasma (pale yellow fluid part) in human blood and is infused (injected) into a vein. It contains specific blood clotting proteins (factors) including fibrinogen, factor VIII (8), factor XIII (13) and von Willebrand factor. In Australia it would only be used in an emergency when other suitable products are not available.
- **Platelet transfusion** may be required for more severe inherited platelet function disorders. To avoid reactions to platelets, the transfusions are often carefully matched to the individual person. This requires specialised testing which can take some weeks.

Other forms of treatment

- Treatment can also involve **exercise guided by a haemophilia physiotherapist**, especially if you have had an injury or have pain in your joints or muscles. This can help to prevent bleeds and protect joints, and keep muscles strong. If you have a joint or muscle bleed, the physiotherapist can also give advice to help reduce pain and a program of advice and exercises to rehabilitate the joint or muscles around where you had the bleed back to full function. General exercise is a common recommendation as another approach to dealing with pain during your period.
- **Pain management:** talk to your specialist doctor (haematologist) before you take any medications containing ibuprofen (e.g., Nurofen™) or aspirin – these medications may worsen your bleeding tendency. Doctors usually recommend paracetamol (e.g, Panadol™) for initially managing pain with a bleeding disorder, but if you feel that something else is needed it is important to discuss this with your haematologist or pain specialist. Heat packs and rest may also help with period pain. If you are having problems with joint pain or other pain that is not period pain, talk to your HTC team or your local GP – there might be a few different issues to take into account.

Registering with your Haemophilia Treatment Centre

If you have bleeding symptoms, registering and staying in contact with your local Haemophilia Treatment Centre is an important part of managing your bleeding disorder.

If you move interstate, touch base with your new Haemophilia Treatment Centre to give them your details and to see how the system works locally. Let your Haemophilia Treatment Centre know if you change any of your contact details so they can keep you up to date with the latest news, workshops or events. It is important they have your latest contact details so that information about appointments (or any other information you request) are sent to the correct address. You can find contact details of HTCs in Australia on the HFA website at www.haemophilia.org.au.

If you move overseas for a long period, eg with study or work or permanently, make sure you research your new local Treatment Centre and get in touch before you leave home. Talk to your Haemophilia Treatment Centre about what you will need to do when you move and how to register with your new HTC. For details of HTCs in other countries, click on the FIND A TREATMENT CENTRE button on the World Federation of Hemophilia website – www.wfh.org.

ABDR and an ABDR patient card

The **Australian Bleeding Disorders Registry (ABDR)** is the online system used by Haemophilia Treatment Centres to manage and record the treatment and care of their patients.

The **ABDR patient card** explains your diagnosis (for example, mild haemophilia A), what treatment should be given and who should be contacted for further advice. It is the size of a credit card and fits in a wallet or purse. Keep the ABDR patient card on you for quick reference.

Previously all females who carried the gene for haemophilia and had reduced factor levels were registered in the ABDR as 'symptomatic carriers', but this has recently changed. Girls and women who carry the gene and have factor levels of less than 40% will now be registered as having haemophilia. Some girls and women who carry the gene for haemophilia have bleeding symptoms but their factor levels are between 40% and 60% and are higher than a person with haemophilia. These girls and women are registered as 'symptomatic carriers' in the ABDR.

MyABDR is the app and website that people with bleeding disorders who are on home treatment use to record their treatments and bleeding episodes. If you use MyABDR, your MyABDR entries are added directly to your patient record in the ABDR every time you sync. The main purpose of MyABDR is for people with bleeding disorders to share information about their use of clotting factor concentrates at home or bleeding episodes with their Haemophilia Treatment Centre.

> "If I experience a serious bleed I will note it in myABDR"

For more information visit 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 Treatment Centre, contact:

Haemophilia Foundation Australia

P: **1800 807 173**

E: **hfaust@haemophilia.org.au**

W: **www.haemophilia.org.au**