Von Willebrand disease

Summary

- Von Willebrand disease (VWD) is an inherited condition that affects males and females.
- A person with VWD is missing one essential blood clotting factor.
- This means it takes longer for their blood to clot and for bleeding to stop.

Von Willebrand disease (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. For people with VWD it takes longer for blood to clot and for bleeding to stop.

VWD is the most common inherited bleeding disorder worldwide. It affects both males and females from all racial backgrounds.

It is thought that up to 1 in 100 people have VWD, but most people have few symptoms. Many people with VWD may not know they have the disorder because their bleeding symptoms are very mild.

Most people with VWD are born with the disorder. Sometimes it is detected in childhood, but other people don't find out they have VWD until they have a bleeding problem as an adult, such as heavy bleeding after a serious accident or a dental or surgical procedure. Some people don't discover they have VWD until a relative is diagnosed and it is suggested that they are tested as well.

The severity of VWD someone experiences depends on the type of VWD and the level of von Willebrand factor in their blood. Usually VWD is less severe than other bleeding disorders, such as haemophilia.

With all forms of VWD there can be bleeding problems. Some people with VWD bleed quite often, for example with nosebleeds, bruising and heavy periods. A small number of people have a severe form of VWD and may experience joint and muscle bleeds, similar to haemophilia.

VWD is a lifelong condition and there is no cure. There are safe, effective treatments for all types of VWD.

What are the symptoms of VWD?

Most people with VWD have few or no symptoms. 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.

Less common symptoms that older people might experience are:

- blood in faeces (poo) from bleeding in the intestines or stomach
- blood in urine from bleeding in the kidneys or bladder.

People with severe forms of VWD, particularly type 3 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.

The types of symptoms a person with VWD experiences can change over their lifetime. For example, they may have nosebleeds and easy bruising as a child and find this occurs less often as they grow older. However, their type of VWD will not change.

Women are more likely to show symptoms of VWD than men. Without treatment, women with VWD often bleed more or for longer than normal with menstruation (their period).

Some women with VWD also have heavy bleeding a few days or weeks after giving birth and some have a lot of period pain or irregular periods. However, these symptoms are not always related to VWD and may have other causes. An assessment by a gynaecologist is an important part of understanding and treating these symptoms effectively.

What are the types of VWD?

There are three main types of VWD (bleeding symptoms can vary from person to person within each type):

- Type 1 VWD is the most common form. Around 80 per cent of all people with VWD have this form. In type 1 VWD, the von Willebrand Factor (VWF) works normally, but there is not enough of it. Symptoms are usually mild, depending on the level of VWF in the blood.
- In type 2 VWD, the amount of VWF in people's blood is often normal but the VWF doesn't work properly. Type 2 VWD is divided into subtypes 2A, 2B, 2M and 2N. Certain subtypes may be treated differently, which makes knowing the exact type of VWD you have very important.
- Type 3 VWD is very rare. People with type 3 VWD have very little or no VWF in their blood. Symptoms are more severe and can also include joint and muscle bleeding. Bleeding can occur more often.

How do you get VWD?

Von Willebrand disorder (VWD) is usually **inherited**, although it can also occur spontaneously (where there is no family history).

Inheritance of VWD

Changes in the VWF gene can disrupt the formation of blood clots and can cause VWD.

You inherit two copies of the VWF gene, one from your mother and one from your father. Most cases of type 1 and type 2 VWD are inherited in an 'autosomal dominant pattern', which means one copy of the changed gene is enough to cause the disease.

Type 3, some cases of type 2 and a small number of type 1 cases are inherited in an 'autosomal recessive pattern', which means that if someone has this type of VWD, both of their copies of the VWF gene will have changes. In these cases, each parent carries one copy of the gene variation (gene change) but they don't have symptoms themselves.

VWD affects males and females in equal numbers – both sons and daughters can inherit it. Their symptoms can be different to their parent's or to each other.

If one member of a family is diagnosed with VWD, the doctor may recommend testing other members to see whether they have VWD as well.

Where there is no family history of VWD

Sometimes there is no family history of VWD and the genetic change may arise randomly during pregnancy Although their parents and their brothers and sisters do not have the VWD gene, the child will be able to pass the VWD gene on to their own children in the future.

Some people develop a form of VWD later in life due to other medical problems that affect their von Willebrand factor. This is called Acquired von Willebrand Syndrome (AVWS) and is very rare.

How is VWD diagnosed?

Many people's VWD symptoms are mild and they may not be diagnosed until they have a major bleeding problem, for example from 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 bleeding or bruising more than normal from mucous membranes or skin after injury, trauma or surgery, and
- a family history of bleeding more than normal, and
- specialised laboratory blood tests for VWD, and
- genetic testing that identifies a change in the VWF gene that causes VWD.

Routine blood tests often give normal results, which is why the person's history of bleeding is so important. Testing is often repeated because a person's VWF and factor VIII levels can vary at different times.

Things that can cause the level of VWF to rise in the blood and appear to be normal are:

- stress
- exercise
- pregnancy
- normal hormonal changes during a woman's monthly menstrual cycle
- hormone treatment
- inflammation with other health conditions
- inflammation in combination with blood type.

If you think you have a bleeding problem, it is important to see a haematologist who specialises in bleeding disorders. In Australia, these haematologists can be found at **haemophilia treatment services** which are at some major hospitals. Talk to your general practitioner or your gynaecologist about a referral.

Other family members may also need to be tested for VWD.

How is VWD treated?

Specialist haemophilia treatment services have a team of health professionals with expertise in providing treatment and care to people with bleeding disorders including VWD. They can work with you to make a treatment plan and advise on ways to live well with VWD.

The team includes:

- haematologists: doctors who specialise in blood disorders
- haemophilia nurses
- social workers or counsellors
- physiotherapists
- other specialist health professionals.

The recommended treatment for VWD depends on the type of VWD you have and how severe it is. Usually people with mild VWD will only need treatment if they have surgery, dental work or an accident or injury. For some minor bleeding problems, like bruising, treatment may not be necessary.

There are several **treatment options** available:

- **desmopressin** – a synthetic hormone that releases the body's stored VWF and factor VIII into the bloodstream to help blood clot. It can be given as a slow injection into a vein; as an injection into the fatty tissue under the skin; or as a nasal spray. Desmopressin is not suitable for everyone
- **clotting factor concentrate made with von Willebrand factor (VWF) and factor VIII (FVIII)** – this replaces the missing VWF and FVIII in the blood and helps blood to clot. It is made from the plasma (pale yellow fluid part) in human blood and is produced from blood donations. It is used when desmopressin is not suitable, or when it is likely the person will need treatment for more than two to three days. It is injected into a vein in the

arm.

- **tranexamic acid** and **aminocaproic acid** are medicines that stop blood clots being dissolved once they have formed. They can be used to stop bleeding in the mouth or nosebleeds, gut bleeding, bleeding after dental work, minor surgery or an injury. Most commonly they are taken as tablets, syrup or as a mouthwash. They may be used by themselves or together with DDAVP or a clotting factor concentrate.
- **fibrin glue** is a medical gel made from fibrinogen and thrombin, which are proteins in the body that help blood to clot. It can be applied directly onto a wound to stop bleeding
- **hormone treatment**, such as **oral contraceptives** (birth control pills), can help women who have heavy menstrual bleeding. The hormones can increase VWF and factor VIII levels.

The treatment you need may vary depending on your medical needs, what works best for you and the situation. Your haematologist will consider all of this when they work with you to decide the best treatment option.

Living well with VWD

Von Willebrand disorder may be part of life, but it doesn't need to rule it. If you have VWD, you can still go to school, have adventures, play sports, raise a family, work, travel, and do the things that inspire you or enjoy the fun activities you love.

Physical activity and VWD

Exercising regularly and being physically active is a great way to keep muscles and joints strong and stay in good health. Exercise can help to boost VWF levels and may even reduce the number of bleeding problems and episodes you experience.

Being active with VWD can be a balance between taking on physical challenges and preventing injuries or dealing with accidents quickly. Know your limits and be prepared.

People with VWD generally have to find out for themselves what physical activities they can and can't do. Many people with mild VWD take part in all kinds of sports, including active sports like soccer and moderate-risk sports like downhill skiing.

People with type 3 or severe VWD might find that contact sports or sports with a high risk of injury, such as football or boxing, lead to serious bleeding and may prefer less injury-prone sports or to modify their activities.

It is important to give children the chance to discover what activities they can safely do. As they grow up, they will want to take part in the same sports as their friends. Naturally parents may want to protect their children from harm, but they can do this by making sure their children follow the current safety guidelines for all children in sport and being prepared for accidents.

Travelling with VWD

If you are planning a trip overseas or interstate, contact your haemophilia treatment centre a few months before departure to plan ahead. They can:

- help with preparations tailored to your situation
- advise on documentation, medication and travel insurance – for example, documentation to carry medication and treatment equipment through security and customs may be required
- advise you on what to do if you need treatment while travelling.

In case you need them, find out the names, addresses, phone numbers and hours of operation of haemophilia treatment services along your route. Knowing where to go in the hospital may also be helpful.

Be sure to get travel insurance for overseas travel.

School and VWD

Having VWD shouldn't affect a child's ability to attend childcare, kinder or school. Most bleeding with Types 1 or 2 VWD is not serious enough to keep a child out of school. Occasionally, with type 3 or severe VWD, a serious bleed into a muscle or joint might mean that a child is absent from school for a short time – but with prompt treatment, this should only be for a day or two.

Tips include:

- Provide information on VWD to childcare, kinder or school staff, including how to manage your child's common bleeding problems, such as nosebleeds.
- You may find it useful to organise an information session. Haemophilia treatment service staff are experienced in this and can assist.
- Let staff know that it is important that they contact you at any time if there is an emergency.
- Provide the telephone number of your child's haemophilia treatment service, and let them know when to use it.
- When you think your child is old enough, teach them how to manage their common bleeding problems too.

VWD issues for women and girls

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

- heavy menstrual periods (such as soaking through a tampon and pad around two hourly, or needing to change a pad during the night)
- menstrual bleeding for longer than normal (for example, longer than eight 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, including during family planning, 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:

- gynaecologist
- haematologist specialising in bleeding disorders
- GP
- paediatrician (when relevant)
- obstetrician (when relevant).

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.

If you are planning to have a child, talk to your haemophilia treatment service and your obstetrician before you get pregnant, and to ask them to liaise with each other. You might also like to speak to a genetic counsellor. Prenatal diagnostic testing may be an option if the genetic variant in your family is known. Ask your haematologist if there needs to be any special care with having tests like amniocentesis.

If you have VWD and are pregnant, let your medical care team know. You will need to be monitored and have blood tests for your VWF levels during your last three months of pregnancy to plan a safe delivery for you and your

baby. This will also help in planning any treatments you might need to prevent possible heavy bleeding in the weeks after delivering your baby, when your factor levels return to their usual level.

Genetic counselling and VWD

If you, your child or another family member have been diagnosed with VWD, or if it runs in your family, it may be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand VWD and what causes it, how it is inherited, and what a diagnosis means for your or your child's health and development, and for your family. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

If VWD runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

Victorian Clinical Genetics Services (VCGS) provides genetic consultation, counselling, testing and diagnostic services for children, adults, families, and prospective parents.

Support for people and families with VWD

Haemophilia Foundation Victoria offers a variety of services and support for people and families living with VWD.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can also connect you with other individuals and families affected by VWD.

Where to get help

- Your **GP (doctor)**
- **Haemophilia Foundation Victoria** Tel. **(03) 9555 7595**
- **Haemophilia Foundation Australia** Tel. **1800 807 173**
- **Genetic Support Network of Victoria** Tel. **(03) 8341 6315**
- **Victorian Clinical Genetics Services** Tel. **1300 118 247**

This page has been produced in consultation with and approved by:

Haemophilia Foundation Victoria