



Combined II, VII, IX and X deficiency

This factsheet is about a bleeding disorder caused by a lack of a protein that the body needs for several blood clotting factors to work properly. It is known as inherited vitamin K deficiency or VKDCFD. It is written to go with our **Rare bleeding disorders booklet**, where you will find much more information on living with one of these conditions.

What is inherited vitamin K deficiency?

Inherited combined deficiency of the vitamin K-dependent clotting factors (VKDCFD) is very rare. Doctors have diagnosed it in fewer than 30 families worldwide. VKDCFD is an autosomal recessive disorder and is found more frequently in areas of the world where marriage between close relatives is common.

VKDCFD is a bleeding disorder caused by the body producing less of one of the enzymes required to activate factors II, VII, IX and X. These are referred to as vitamin K-dependent proteins as that is also necessary to make them fully active. It causes problems because the clotting reaction that would normally control any bleeding is blocked too early, so your body doesn't make the blood clots it needs to stop bleeding.

What causes VKDCFD?

This is an inherited genetic disorder. It is what is known as recessive, meaning you have to inherit the gene defect from both parents. It affects men and women.

If you carry one copy of the gene fault for VKDCFD, you are known as a carrier. You can only pass the condition on to your children if your partner also carries the gene fault. You will not have the condition yourself, but any children that inherit the gene fault from you will also be carriers of the condition.

Our **Rare bleeding disorders booklet** has more information about how bleeding disorders are inherited.

Symptoms of inherited vitamin K deficiency

Most people with inherited vitamin K deficiency are diagnosed early in life, at birth or in infancy or early childhood. They may have:

- bleeding from the umbilical cord stump at birth
- bleeding in the gut (gastrointestinal tract)
- bleeding into muscles and joints
- bleeding into soft tissues, such as

For everyone affected by a genetic bleeding disorder

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- bleeding gums
- bleeding into the brain at birth
easy bruising.

Girls and women usually have heavy periods that last for longer than normal (menorrhagia). Less often, people are diagnosed in early adulthood because of bleeding or routine blood tests during investigations for another condition. They may have:

- bleeding after surgery
- bleeding after an accident.

VKDCFD can be made worse at times because of other conditions. For example, after taking antibiotics that kill off normal bacteria in the gut which help the body to make vitamin K2. This will get better once the antibiotic course is finished and the normal bacteria recover.

It is very important that you contact your doctor or haemophilia centre if you think you or your child are having a bleed. If you have frequent bleeds, such as nosebleeds, you can become anaemic. This means you have low haemoglobin levels because of the frequent loss of blood. Anaemia can make you feel tired and breathless.

There is more information in our **Rare bleeding disorders booklet** about how to spot the different types of bleeds

Diagnosing Bernard-Soulier syndrome

Inherited vitamin K deficiency is diagnosed with a series of blood tests. These are specialised tests and have to be done at a haemophilia treatment centre.

Most people are diagnosed fairly young in life, either at birth after bleeding from the umbilical cord stump, or because of a bleed in childhood. Or your baby may be tested at birth because VKDCFD runs in your family.

Treatment for VKDCFD

VKDCFD is relatively easy to treat in most cases. Babies and children are started on a daily dose of vitamin K and that usually controls bleeding. If it doesn't work as well as it should, your doctor may suggest a weekly injection of vitamin K instead.

While vitamin K tablets or injections control bleeding well from day to day, you need extra treatment to prevent bleeding if you are at higher risk for any reason. If you have minor bleeding, or are going to have minor surgery, your doctor may suggest that you take tablets called tranexamic acid (Cyklokapron) for a few days. These help to stop blood clots breaking down.

If you have a serious bleed or are going to have major surgery, your doctor may also want you to have treatment called prothrombin complex concentrate (PCC). This contains clotting factors II, VII, IX and X. If this isn't available, you may have fresh frozen plasma (FFP), which also contains these clotting factors. Both these treatments are made from donated human blood – from plasma, the straw-coloured fluid that the blood cells are carried in. You have them through a drip into a vein (intravenously).

All these blood products are now treated during manufacture to kill off any known viral infections such as hepatitis and HIV.

You may also need treatment with PCC if you are having a baby. You start the treatment once you've gone into labour and continue with it for three days after you've had your baby.

You should not use Non-Steroidal Anti-Inflammatory Drugs (NSAIDs such as ibuprofen) as this increases the risk of bleeding. Other methods of pain relief should be used instead. Speak to your doctor if you are unsure.

You should have immunisations or other injections subcutaneously (under the skin) rather than intramuscularly (into a muscle) to reduce the risk of a painful bruised swelling (haematoma) developing.

Coping with your condition

Finding out that you or your child has a bleeding disorder can be upsetting and bring on a range of different emotions. Of course, this will take time to accept. Finding out as much as you can about your condition can help you learn to cope with it.

In general, VKDCFD will not affect your daily life too much, as long as you take your vitamin K as often as you should. You will only need extra treatment to prevent bleeding if you are due to have surgery or are having a baby. But you still have a higher risk of bleeding than normal and need to know the signs of a bleed so you can recognise when you need to seek medical help.

Do find out as much as you can about how to prevent bleeding and when it is likely to cause a problem. Our **Rare bleeding disorders booklet** has a lot of information about what to look out for and precautions you can take to keep yourself healthy. There is information on:

- carrying medical information with you
- dental care
- how to spot the early signs of a bleed
- information for girls and women about problems with periods and pregnancy
- ways to make bleeding less likely

A new diagnosis can feel scary or overwhelming but there's lots of great support available.