

Factor V, factor VIII combined deficiency

This factsheet is about a bleeding disorder that is related to problems with two blood factors (factor V and factor VIII (pronounced factor 8)). 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 factor V, factor VIII combined deficiency?

This is a bleeding disorder caused by the body producing less of two particular factors than it should. This 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.

Factor V, factor VIII deficiency is very rare. Doctors estimate that it affects between one in a million and one in two million people overall. In some populations it is more common. In Ashkenazi Jewish and Iranian populations, it may be as common as one in 100,000. But it may also be under-diagnosed because the symptoms can be very mild.

What causes factor V, factor VIII combined deficiency?

This is an inherited genetic disorder. It affects both males and females. The condition is recessive, meaning you must inherit the gene defect from both parents in order to develop it.

There is only actually one gene fault involved, as the affected gene is involved in the production of both factor V and factor VIII.

If you carry one copy of the gene fault for factor V, factor VIII deficiency, you are known as a carrier. You can only pass the condition onto your children if your partner also carries the gene fault. Any children that inherit the gene fault from you will also be carriers of the condition but will not have it themselves.

Symptoms of factor V, factor VIII combined

The symptoms of factor V, factor VIII combined deficiency are generally quite mild.

The commonest symptoms are:

- bleeding after circumcision
- bleeding gums
- easy bruising
- heavy periods or periods that last longer than normal (menorrhagia)
- nosebleeds
- skin bleeding.

You may also have bleeding:

- after dental treatment
- after giving birth
- after surgery
- into the soft tissues.

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More serious bleeds, such as into the gut (gastrointestinal system) or brain and spinal cord (central nervous system) have been known to happen but are very rare.

Serious bleeds are rare with factor V, factor VIII combined deficiency. 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 factor V, factor VIII combined deficiency

Factor V, factor VIII combined deficiency is diagnosed with blood tests. These include a series of tests to measure how quickly your blood clots. Blood tests measure the levels at between 5% and 30% of normal. These are specialised tests, so you need to have them done at a haemophilia treatment centre.

Your doctor may suggest these blood tests at birth, because your baby had bleeding after the umbilical cord was cut, or when a circumcision was done.

The bleeding disorder may also show up later in life because of other bleeding symptoms, for example after dental work or other surgery.

Treatment for factor V, factor VIII combined deficiency

Generally, people with factor V, factor VIII combined deficiency only have treatment if they have a bleed or before planned surgery.

Replacement factor V concentrate is not yet available. So treatment for a bleed is usually with fresh frozen plasma (FFP), which contains all the clotting factors including factor V. It only has low levels of factor VIII, so you have factor VIII concentrate along with the FFP if you need treatment.

FFP is made from plasma, the straw-coloured fluid that the blood cells are carried in and factor VIII is made in a laboratory. You have all these treatments through a drip into a vein (intravenously).

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

Your doctor may also suggest treatment with a drug called desmopressin if you have a bleed or before surgery. This drug makes small blood vessels release stored factor VIII. You may also have treatment with a drug called tranexamic acid (Cyklokapron). This helps to stop bleeding by preventing existing clots from breaking down.

In some cases, the contraceptive pill might be prescribed to girls and women to make heavy periods lighter.

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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.

How much your bleeding disorder affects your daily life will depend on how severe it is. For many people, it won't have much affect at all. It may only be an issue if you are having dental work, major surgery, having a baby or have an accident. Others may need treatment to treat minor or more serious bleeds.

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.