



Fibrinogen (factor I) deficiency

This factsheet is about bleeding disorders related to problems with a blood-clotting factor called fibrinogen or factor I (one). 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 are fibrinogen deficiencies?

Fibrinogen (pronounced fie-brin-oh-jen) abnormalities are bleeding disorders caused by the body producing less fibrinogen than it should, or because the fibrinogen your body makes doesn't work properly. They cause 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 I deficiency is an umbrella term for several related disorders known as congenital fibrinogen defects. There are several different types of factor I bleeding disorders. All of them affect males and females equally.

- Afibrinogenaemia is pronounced ay-fie-brin-oh-jen-ee-mee-ah. It means a complete lack of fibrinogen. It is very rare. Doctors

estimate that it affects about one in a million people.

- Dysfibrinogenaemia is pronounced diss-fie-brin-oh-jen-ee-mee-ah. It means that your body makes fibrinogen, but it doesn't work in the way it should. The inherited form is rare. But it can also develop as a result of other diseases and this is more common.
- Hypofibrinogenaemia is pronounced high-po-fie-brin-oh-jen-ee-mee-ah. It means having lower than normal levels of fibrinogen. This is also a rare condition.
- Hypodysfibrinogenaemia is pronounced high-po-diss-fie-brin-oh-jen-ee-mee-ah. It means that you have low levels of fibrinogen and that the fibrinogen you do have doesn't work as it should. It is very rare.

Because some people with hypofibrinogenaemia and hypodysfibrinogenaemia do not have any symptoms, we don't know exactly how rare these conditions are.

For everyone affected by a genetic bleeding disorder

To find out more, visit haemophilia.org.uk or contact us on 020 7939 0780 or at info@haemophilia.org.uk

What causes fibrinogen deficiencies?

Afibrinogenaemia is an inherited genetic disorder. It is what is known as recessive, meaning you have to inherit the gene defect from both parents.

Hypofibrinogenaemia, dysfibrinogenaemia, and hypodysfibrinogenaemia are usually inherited genetic conditions. They can be recessive, meaning that both parents must carry the gene defect in order to pass it on to a child. Or they can be dominant, meaning that only one parent needs to carry the gene fault in order to pass the condition on to a child.

People can also develop dysfibrinogenaemia later in life. This is called acquired dysfibrinogenaemia. It is much more common than the inherited form and is usually caused by liver disease.

Our Rare bleeding disorders booklet has more information about these conditions and how they are inherited.

Symptoms of fibrinogen deficiencies

The symptoms of fibrinogen bleeding disorders vary, depending on the type of condition that you have.

Symptoms of afibrinogenaemia and hypofibrinogenaemia are similar. But the symptoms you have with hypofibrinogenaemia will depend on how much fibrinogen you have in your blood. The less you have, the more symptoms you are likely to have and the more severe they will be.

Common symptoms include:

- abnormal bleeding after circumcision
- abnormal bleeding after injury or surgery
- bleeding after childbirth (post-partum

haemorrhage)

- bleeding from the umbilical cord stump at birth
- bleeding in the mouth, particularly after dental surgery
- bleeding into joints
- bleeding into muscles
- easy bruising
- heavy periods or periods that last longer than normal (menorrhagia)
- nosebleeds
- problems during pregnancy, including miscarriage.

You may also have these symptoms, but they are less likely:

- bleeding in the gut (gastrointestinal haemorrhage)
- bleeding into the brain or spinal cord (the central nervous system)
- blood clots (thrombosis)
- Problems with slow wound healing.

Diagnosing fibrinogen deficiencies

Fibrinogen bleeding disorders are diagnosed with blood tests. These include tests to measure how quickly your blood clots and a specific test that measures the amount of fibrinogen in your blood. These are specialised tests, so you need to have them done at a haemophilia centre. To get a definite diagnosis of an inherited factor I deficiency you need to have DNA testing. But this may not be necessary if there is a clear history of the disorder in your family.

Your doctor may suggest these blood tests at birth, because your baby had bleeding from the umbilical cord or when a circumcision was performed. Afibrinogenaemia is usually diagnosed in this way.

Or the bleeding disorder may show up later in life because of other bleeding symptoms – after dental work or other surgery, for example. This is more usual with hypofibrinogenaemia and

dysfibrinogenaemia, which can be diagnosed in adulthood.

Treatment for fibrinogen deficiencies

How often you need to have treatment will depend on how severe your condition is.

Many people with hypofibrinogenaemia and dysfibrinogenaemia do not need any treatment at all.

If you have very heavy periods, your doctor may suggest that you take the contraceptive pill to lighten them. Or you may have a tablet to take during your periods to stop the breakdown of blood clots, such as tranexamic acid (Cyklokapron).

Your doctor may also suggest you take tranexamic acid tablets for minor bleeding, such as nosebleeds or before minor surgery.

There are three main treatments for more serious bleeding in fibrinogen deficiency. All are made from donated human blood from plasma, the straw-coloured fluid that the blood cells are carried in. They are:

- cryoprecipitate
- fresh frozen plasma (FFP)
- fibrinogen concentrate.

Although it is not treated to destroy viruses, the likelihood of getting an infection from a transfusion of FFP or cryoprecipitate is very low. Fibrinogen concentrate is treated during manufacture to kill off any known viral infections such as hepatitis and HIV.

You have these treatments through a drip into a vein.

Depending on how severe your condition is, you may need regular treatment once or more a week, to keep your fibrinogen levels up. Your doctor may call this prophylaxis (pronounced proff-ill-ax-iss). Or you may just

need treatment if you have a serious bleed, before you have surgery or dental work. If you have very low fibrinogen levels and become pregnant, you may have fibrinogen concentrate twice or more a week until you deliver your baby.

You may also need treatment to prevent abnormal blood clots forming, as this can be a complication of these types of condition. If you have had problems with clots in the past, your doctor may suggest you take a low dose of an anti-clotting drug such as heparin (enoxaparin), particularly during pregnancy.

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.

Caution is needed for injections as well – immunisations should be given 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 effect 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 regular treatment to prevent bleeding or treat 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.