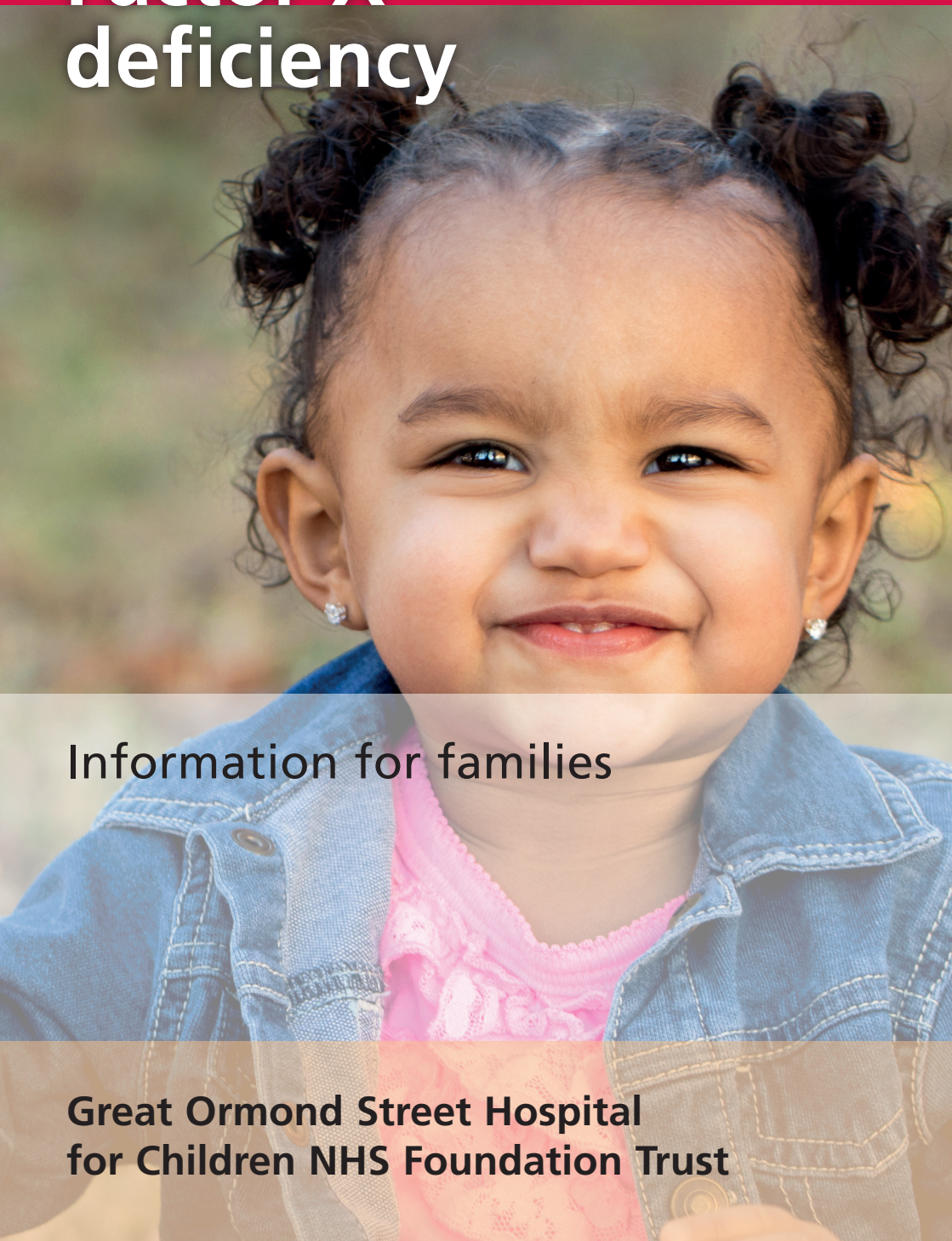
# Factor X deficiency



Information for families

Great Ormond Street Hospital for Children NHS Foundation Trust

**Factor X (previously known as the Stuart-Prower factor) deficiency is a type of clotting disorder. A specific protein is missing from the blood so that injured blood vessels cannot heal in the usual way. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Factor X deficiency and where to get help.**

## What is a clotting disorder?

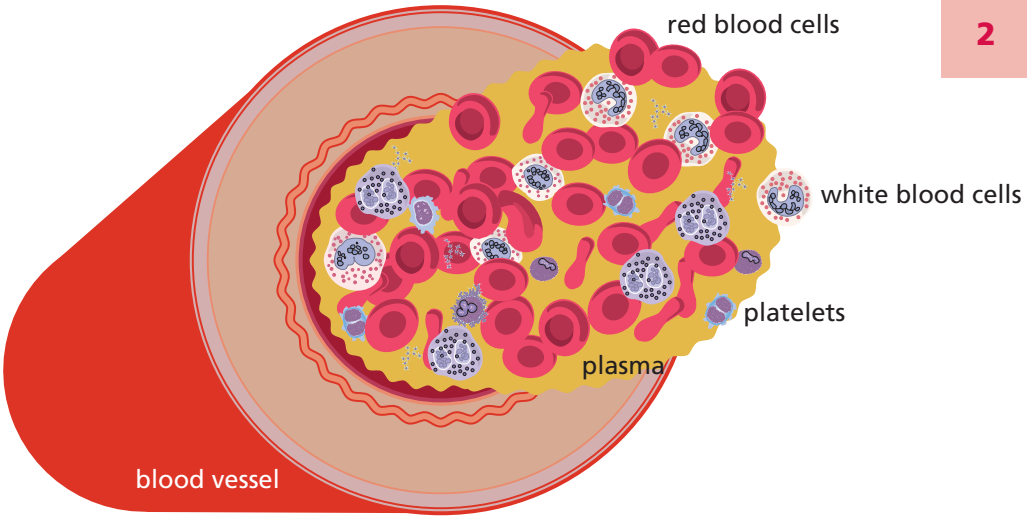
A clotting (or coagulation) disorder is a medical condition where a specific protein is missing from the blood.

Blood is made up of different types of cells (red blood cells, white blood cells and platelets) all suspended in a straw-coloured liquid called plasma. Platelets are the cells responsible for making blood clot. When a blood vessel is injured, platelets clump together to block the injury site. They also start off a complicated chemical reaction to form a mesh made of a substance called fibrin. This complicated chemical reaction always follows a strict pattern – with each clotting protein (known as a coagulation factor) turned on in order. When all of the factors are turned on, the blood forms a clot which stops the injury site bleeding any further.

There are a number of coagulation factors circulating in the blood, lying in wait to be turned on when an injury occurs. If any one of the factors is missing from the body, the complicated chemical reaction described above will not happen as it should. This can lead to blood loss, which can be severe and life-threatening. Each coagulation factor is given a number from I to XIII – they are always written as Roman numerals – and the effects of the missing factor will vary.

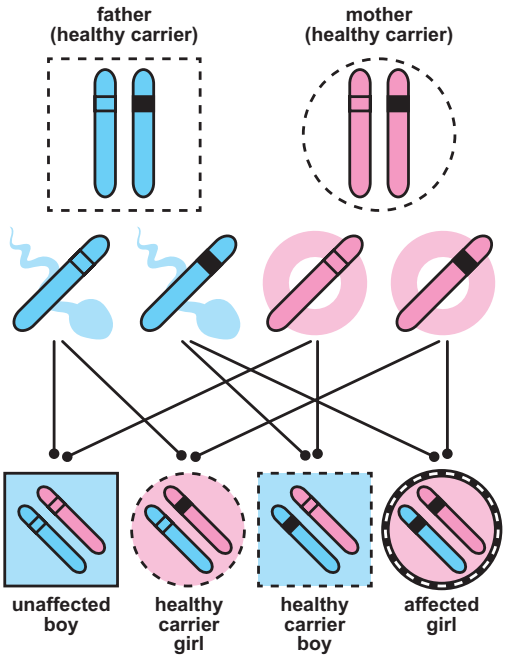
## What is Factor X deficiency?

Factor X deficiency is a very rare clotting disorder, affecting between 1 in 500,000 to a million people worldwide. The specific coagulation factor that is missing or reduced is Factor X which needs vitamin K from the liver to be produced.




## What causes Factor X deficiency?

Human beings have about 30,000 to 40,000 different genes, each of which has a function in making an individual person.




The genes are arranged in pairs (one of the pair from each parent) on 23 chromosomes – inevitably some of these genes are faulty. Factor X deficiency is caused by a mutation (change) on the F10 gene, which is inherited in an autosomal recessive manner. This means that a person has to inherit the faulty gene from both parents to develop the severe form of the disease. Autosomal recessive disorders are more common in areas of the world where marriage between close relatives is common.

Each pregnancy carries a:

- 75 per cent chance of having an unaffected child
- 25 per cent chance of having an affected child

People who carry one copy of the faulty gene are said to be a 'carrier'. The majority of carriers are healthy but occasionally, 'affected carriers' may show mild symptoms of Factor X deficiency, which may or may not need treatment.

## What are the signs and symptoms of Factor X deficiency?

The age at which symptoms appear varies, depending on the amount of Factor X in the blood and how well it is working. Children with little or no Factor X may start to show symptoms soon after birth, whereas those with some functioning Factor X may not show symptoms until later, often following surgery or injury. The symptoms of Factor X deficiency vary in severity, even between members of the same family. In many cases, the symptoms of Factor X deficiency are so mild that they cause no problems.

Babies with severe Factor X deficiency may have prolonged bleeding from their umbilical cord following childbirth and are also at risk of bleeds inside the brain (intracranial haemorrhage). This is because the level of Factor X in the blood is too low to complete the clotting process as described earlier.

Other symptoms that can occur at any age include bleeding inside the joints and muscles and also mouth bleeds following dental work. Bruising is more common and blood may also be present in the urine (haematuria). Bleeding may be more severe or last longer than usual following surgery or injury.

Women with Factor X deficiency will often have heavy menstrual periods. Pregnancy should be carefully planned as there is an increased risk of early miscarriage or extended bleeding following childbirth.

## How is Factor X diagnosed?

Factor X deficiency can be diagnosed before birth (prenatally) if there is a family history. There are several options for this including chorionic villus sampling (CVS) early in pregnancy or amniocentesis around 15 to 20 weeks or so.

In babies, a clotting disorder may be suspected following a bleed inside the brain (intracranial haemorrhage) or if bleeding continues after the umbilical cord is cut or after surgery such as circumcision. In older children and adults the suspicion of a clotting disorder might arise during routine checks before an operation or following an injury. Mild Factor X deficiency is diagnosed more in females than males due to menstruation and child birth.

Factor X deficiency can be diagnosed using a sample of blood for testing in the laboratory. A test to measure how long a sample takes to clot may suggest a clotting disorder, which would then be investigated further. The level of Factor X in the blood will be checked and doctors will try to identify the gene mutation as well, as this can be helpful for planning future brothers and sisters.

Imaging scans, such as MRI, CT or ultrasound scans may be used to identify any internal bleeds, for instance, inside a joint.

## How is Factor X treated?

In mild cases, if symptoms are not problematic, treatment may not be needed. In many cases, what treatment is needed is on an on demand basis to treat symptoms as they occur rather than preventative (prophylaxis).

Treatment may be required as part of planning for an operation or treating an injury. An infusion of fresh frozen plasma (FFP) can be given during the operation and afterwards. Another blood product, prothrombin complex concentrate (PCC) can also be given. Children with a severe deficiency of FX need to have FX replacement every 48 to 72 hours which is usually given via a central venous access device so that the treatment can be given at home.

It is important that people with Factor X deficiency should not use Non-Steroidal Anti-Inflammatory Drugs (NSAIDs such as ibuprofen) as this greatly increases the risk of bleeding. Other methods of pain relief should be used instead. Caution is needed for injections as well – immunisations for instance, should be given subcutaneously (under the skin) rather than intramuscularly (into a muscle) to reduce the risk of a painful bruised swelling (haematoma) developing.

Children and young people with Factor X deficiency will need regular reviews at their specialist centre to check that they are responding to treatment and not experiencing any side effects. This review will usually involve a check-up from the physiotherapist and dentist as well.

## What happens next?

Children and young people with Factor X deficiency have a normal life span. Some activities may need to be avoided, such as contact sports that carry a high risk of head injury for instance, but most day to day activities will cause few problems. It is always advisable to wear a medical alert identity necklace or bracelet to alert health care professionals about having Factor X deficiency – details are available via the support organisation. Any surgery or future pregnancy will need careful planning in advance so it is important that all health care professionals involved are aware.

As Factor X deficiency is a genetic condition that can be passed on from parent to child, it is possible to have genetic counselling before planning a family, both for affected individuals and unaffected carriers. Talk to us about a referral to our Clinical Genetics service.

Children and young people with Factor X deficiency may need life-long monitoring and treatment, so as your child approaches their teenage years, we will start to talk to them about getting ready to move on to adult health services. This is a planned process so that they become more independent as they grow older and able to manage their own health.

## Further information and support

Call the **Haemophilia Comprehensive Care Centre** at GOSH on 020 7829 8837.

The **Haemophilia Society** offers support and advice to anyone affected by haemophilia or any other clotting disorder. Call their helpline on 020 7939 0780 or visit their website at www.haemophilia.org.uk.

A wide range of resources is also available on the World Federation of Hemophilia website at www.wfh.org and UK-specific information regarding haemophilia, bleeding disorders and the National Haemophilia Database can be found via the United Kingdom Haemophilia Centre Doctors' Organisation website at www.ukhcdo.org. Useful details of Haemophilia Centres when travelling in Europe can be found at www.euhanet.org/centrelocator.

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