Factor VII deficiency

Information for families

Great Ormond Street Hospital for Children NHS Foundation Trust
Factor V deficiency (also occasionally known as Owren’s disease or parahaemophilia) is a clotting disorder. A specific protein is missing from the blood so that injured blood vessels cannot heal in the usual way. It is completely different to the condition Factor V Leiden deficiency which increases the risk of blood clotting. The combination of Factor V and Factor VIII deficiency is also a separate condition and not covered here. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Factor V 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 V deficiency?

Factor V deficiency is a very rare clotting disorder affecting only around 150 people worldwide. The specific coagulation factor that is missing or reduced is Factor V. The severity of symptoms ranges from mild to severe depending on the amount of Factor V present in the blood and its activity.

What causes Factor V 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 V deficiency is caused by a mutation (change) on the F5 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 V deficiency, which may or may not need treatment.

What are the signs and symptoms of Factor V deficiency?

The symptoms of Factor V deficiency vary in severity, even between members of the same family. In many cases, the symptoms of Factor V deficiency are so mild that they cause no problems.

However, if the level of Factor V is low or absent or it does not work properly, symptoms may be present, often early in life. These can include bleeding in the brain, bleeding from the umbilical cord around birth, nose bleeds and bruising. Mouth bleeds following dental work can also occur, as can blood loss during childbirth or heavy menstrual periods. Occasionally bleeding can occur inside the body. This is because the level of Factor V in the blood is too low to complete the clotting process as described earlier.
How is Factor V deficiency diagnosed?

Often the suspicion of a clotting disorder might arise because of bruising or bleeding or during routine checks before an operation or following an injury.

Factor V 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 V in the blood will be checked as well as the level of Factor VIII as these sometimes occur in combination resulting in different and more serious bleeding. Doctors will try to identify the gene mutation as well, as this can be helpful for planning future brothers and sisters and for testing other family members.

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

Factor V 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.

How is Factor V deficiency treated?

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. Unlike other factors, Factor V is not available in replacement form so an infusion of fresh frozen plasma (FFP) is usually given instead. Alternatively, a medicine called tranexamic acid can be given to temporarily boost the proteins that stabilise blood clots. This is given by mouth, into a vein or topically (applied to the skin).

Females may have to take additional measures to make their monthly periods manageable. Options can include taking tranexamic acid before and during her periods, the contraceptive pill or having an intra-uterine device (IUD) inserted.

It is important that people with Factor V 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 V 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 V 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 V deficiency – details are available via The Haemophilia Society 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 V 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 V deficiency 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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