Factor VII deficiency

Patient brochure

changing haemophilia®

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Dear patient, dear parents,

You or your child have been found to have a factor VII deficiency (factor 7, VII is the Roman number for 7). This result may have heightened your level of concern and raised many questions. The purpose of this brochure is to help you understand factor VII deficiency and to help you answer your most important questions. In addition, your doctor will be happy to answer any questions you may have.

Description of factor VII deficiency

What is factor VII deficiency?
Factor VII deficiency is a rare congenital coagulation disorder in which a coagulation factor in the blood is reduced. Factor VII (FVII) deficiency is the most common of the so-called rare congenital coagulation disorders. The severe form of the disease occurs in around 1 in 300,000–500,000 people.[1–5] FVII deficiency was first described in 1951 by the American doctor Benjamin Alexander from Boston.[6]

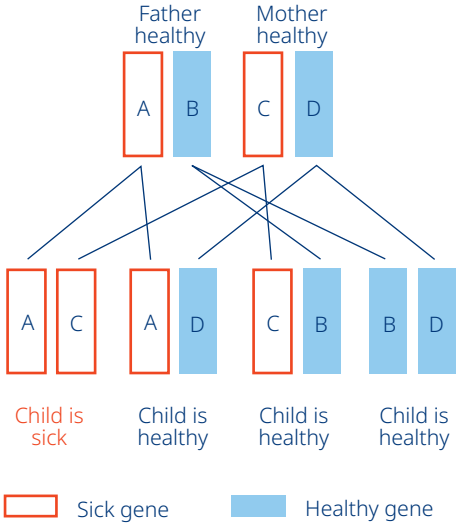
Is an FVII deficiency dangerous?
Many of those affected have few problems with it throughout their lives. Congenital FVII deficiency occurs in different forms: no signs through to a tendency towards severe bleeding. Because increased bleeding may occur in some situations, e.g. in the case of accidents or surgical procedures, it is important that you yourself are well informed about your illness and inform your doctors immediately about your FVII deficiency. This is also the reason why you will receive an emergency ID card, which you should always carry with you.

What is a coagulation factor?
In the event of a bleeding injury in humans, the coagulation system starts working in the blood to close the site of bleeding and prevent further blood loss. There are various substances (protein) in the human blood for this purpose, the so-called coagulation factors. The coagulation factors can have a coagulation-promoting or anticoagulant function.

Severity and inheritance

Are there different forms or severities related to FVII deficiency?
Yes. Depending on the amount of FVII present in the blood and its functional capability, the so-called FVII residual activity, a distinction is made between a mild, moderate or severe FVII deficiency. In mild and moderate cases, it is quite possible that those affected never have any problems with bleeding.

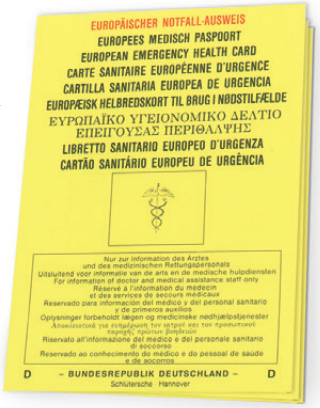
How do people develop FVII deficiency?
It is a congenital disease that is inherited from the parents to the children. FVII deficiency affects both men and women.[7] In order for a child to be born who has a FVII deficiency, both parents must have the affected gene. As the figure shows, in these cases, the risk for a child to inherit the FVII deficiency is 25%.[8]



Emergency situation

Does each patient with FVII deficiency receive an emergency ID card?
Yes. Even if you have a slight FVII deficiency, you will receive an emergency ID card. Often the tendency to bleed is not clearly predictable. It depends on the type and severity of the planned procedure, the residual FVII activity, but also on personal characteristics such as the type of FVII gene change that has been inherited and the previous history as to whether or not increased bleeding is to be expected. It is therefore important that you carry your emergency ID card and inform your doctor about your FVII deficiency.

European emergency ID card ▶



Tendency to bleed

When do I have an increased tendency for bleeding?
The tendency towards bleeding depends on the "coagulant activity", i.e. how effectively blood coagulation works. If "coagulant activity" is reduced, there is a tendency for blue spots (hematoma) without a discernible cause and an increased risk of bleeding with high blood loss. There is still a strong tendency to bleed in the case of accidents and surgery, as well as when giving birth (for the mother) and during menstrual periods.[1,9]

Is there a greater tendency for women to bleed?
In young girls with FVII deficiency, the first period may be longer and heavier than normal. This also applies to menstrual periods in general. It should be noted that regular and severe blood loss can slowly lead to iron deficiency, which can result in anaemia (iron deficiency anaemia). Therefore, sometimes attention must be paid to iron intake and/or iron substitution.[1]

What bleeding occurs frequently?
In addition to the increased periods and bleeding during surgery, bleeding of the mucous membranes (nose bleeding, bleeding of the gums), bleeding in muscles and

joints, bleeding in the gastrointestinal tract, blood in the urine and bleeding in the central nervous system occur in people with severe FVII deficiency.[1,9]

What should I consider when pregnant?
Pregnancies often go smoothly in the case of women with FVII deficiency. However, severe FVII deficiency during the birth process can lead to increased bleeding, which can be dangerous for the child and mother. Depending on the severity of the disease, it may be necessary to administer a preparation containing FVII before and/or during birth. In the case of bleeding during pregnancy, it is also possible to reduce the tendency to bleed by preventive administration of the clotting factor.

The decision as to whether an FVII-containing product is used in a pregnancy of a patient suffering from FVII deficiency must always be made by an experienced physician (clotting specialist). The benefit of treatment for a decreased tendency to bleed must be carefully weighed against a possible increased tendency to clot (thrombosis) during pregnancy.

Diagnosis

How can FVII deficiency be detected?
Often, congenital FVII deficiency is a coincidence. This means that in the course of general check-ups or before an operation, an abnormal blood value will be found in the blood test without you having experienced bleeding. Some patients also first notice a tendency to bleed or bruise and then the FVII deficiency is detected in the doctor's surgery.[4]

What blood test can be used to determine the FVII deficiency?
The FVII deficiency is picked up by a decrease in a laboratory value (quick value). Following this, the activity of the coagulation factors such as factor VII can be determined by a further laboratory test. Using a genetic test, with which the genetic material for FVII is examined, it is possible to discover the genetic change that is the basis.[10,11]

What are the benefits of an examination of the FVII genetic material?

The tendency to bleed varies greatly among people with FVII deficiency. On the other hand, the examination of the genes serves to find the gene change responsible for the FVII deficiency. On the other hand, the genetic examination of many affected persons and the accompanying recording of the bleeding can contribute to a better understanding of the disease and ultimately, one day, to the predictability of a tendency towards bleeding due to certain genetic changes.[10–12]

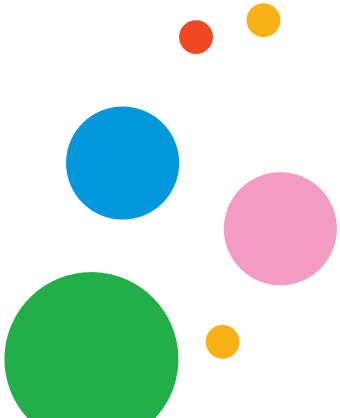
Treatment

Is there a treatment for my FVII deficiency?
Yes. The treatment consists of replacing the missing clotting factor, (so-called substitution treatment).[4] The clotting factors must be injected into a vein, as they cannot be administered as tablets. Depending on the residual level, it may also be necessary to administer a factor as a preventative measure in the event of expected bleeding (surgery). Because the tendency to bleed is very different in FVII deficiency, some people with FVII deficiency do not require the administration of the clotting factor.

What to do in the case of bleeding
In any case, you should visit a hospital.

What kind of substances can be used to treat FVII deficiency?
There are two ways to replace the missing FVII. One possibility is a genetically manufactured product (this is known as a recombinant). It is free of components from human donor blood and therefore safe from infection with foreign viruses or bacteria. The second option is made up of human blood donations; the product is plasmatic in terms of its origin.[4,13]

Are there any risks in the administration of coagulation factors?
Two important hazards must be considered: firstly, the risk of excessive coagulation resulting in an undesired blood clot (a so-called thrombosis) and secondly, the risk of antibodies being formed against the coagulation factor that is supplied (so-called inhibitors). The risk of thrombosis and the possibility of inhibitor formation is low in FVII deficiency.[2,4,13,14]



Day-to-day management

Can a child with a FVII deficiency participate in kindergarten and school activities?
Yes. FVII deficiency does not usually affect everyday life. However, teachers should be aware that your child has a FVII deficiency so that they can react correctly in an emergency. The child can also take part in normal school sports or exercise.

What needs to be observed when changing or pulling teeth?
If a severe tendency towards bleeding is known of in the case of injuries, preventive administration of a preparation containing FVII should be considered before dental interventions. If frequent major bleeding events occur during tooth changes in children with FVII deficiency, preventive treatment with the clotting factor should be discussed with a coagulation expert.

Can I take my medicine with me when I travel?
Ask your doctor's office for a customs certificate to help you take your medication with you on your travels.

CUSTOMS CERTIFICATE

for medication

GB

To whom it may concern,

The following patient has haemophilia and can only control his/her bleeding with the help of factor concentrates.

Mr/Ms

Patient name

who has

e.g. Haemophilia A or B

is travelling to

The above-named person cannot start his/her journey without taking factor concentrates. He/She is carrying sufficient medication for autoinfusion and additional quantities for emergencies.
Accidental confiscation/seizure of the factor concentrates could put the life of the above-named person at risk because this medication is essential for him/her.

Thank you for your co-operation.
Yours sincerely

Signature, name and title of the attending physician

MEDICAL CERTIFICATE

for presentation to the authorities

GB

Mr/Ms

Is suffering from the blood disease called:

i.e. a clotting factor deficit.

I know the patient personally. In case of bleeding he/she responds well to prompt autoinfusions of factor concentrates. Preferably of:

Name of the concentrate

You can safely follow the instructions given by Mr/Ms

Name of the patient

concerning when and what quantity of factor concentrates are to be administered.

Yours sincerely

Signature, name and title of the attending physician

Source: dhg.de/leben-mit-blutungserkrankungen/reisen.html ▲

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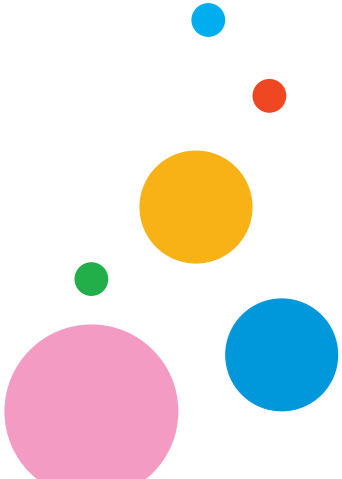

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haemcare.de is your portal for everything you need to know about blood coagulation disorders, sports, nutrition, travel … and what's happening right now.

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