

FACTOR V DEFICIENCY (PARAHEMOPHILIA)

AN INHERITED BLEEDING DISORDER

AN INFORMATION BOOKLET



Canadian Hemophilia Society
Help Stop the Bleeding



Canadian Association of
Nurses in Hemophilia Care
Association Canadienne des
Infirmières et Infirmiers en Hémophilie

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This booklet provides general information on factor V deficiency only. The Canadian Hemophilia Society does NOT practice medicine and does not suggest specific treatments. In all cases, we suggest that you speak with a doctor before you begin any treatment.



PREFACE

We are pleased to present this first edition of the information booklet *Factor V Deficiency: An Inherited Bleeding Disorder*.

This booklet has been written in order to inform people with factor V deficiency and their families about the disorder, and to educate the general public.

The information presented in this document was accurate at the time of its publication. The authors and editors do not assume responsibility for any problems that may arise related to its practical clinical application.

Be careful not to confuse factor V deficiency with factor V Leiden nor with the combined deficiency of factor V and factor VIII.

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Introduction

Factor V deficiency, also called *parahemophilia* or *Owren's disease*, is a very rare coagulation disorder. About one person in a million may be affected by this deficiency. Only 150 cases have been identified worldwide to date.

There is very little written information about this disorder. The purpose of this booklet is to describe the disorder and its treatment in order to help those affected by it deal with this health problem. The booklet explains the disorder and currently available treatments.

This booklet will not answer all your questions. The best source of information is still the comprehensive care team at the hemophilia treatment centre. However, it can provide a helpful reference for those affected, as well as their families.

Factor V deficiency

Factor V deficiency is a rare inherited bleeding disorder. It was first identified in 1943 in Norway by Owren.

Factor V is a blood protein that plays a role in the coagulation cascade, the chain reaction that is triggered when a blood vessel is damaged. Blood is carried throughout the body in a network of blood vessels. When we are injured, the blood vessels may break at the surface, as in the case of a cut. Or they can break deep inside the body, causing a bruise or internal hemorrhage. Clotting, or coagulation, is a complex process that makes it possible to stop injured blood vessels from bleeding. As soon as a blood vessel wall breaks, the components responsible for coagulation come together to form a plug at the break. Coagulation factors are required to hold the plug (or homeostatic plug) in place and form the permanent clot.

There are several steps involved in forming a clot:

- Step 1** The blood vessel is damaged.
- Step 2** The blood vessels contract to restrict the blood flow to the damaged area.
- Step 3** The platelets stick to the walls of damaged vessels and spread out. This is called *platelet adhesion*. These platelets then emit chemical signals that activate other nearby platelets so that they clump together at the site of the damage in order to form a plug, the *homeostatic plug*. This is called *platelet aggregation*.
- Step 4** The surface of these activated platelets forms a base on which blood coagulation can take place. The coagulation proteins circulating in the blood (including factor V) are activated at the surface of the platelets to form a fibrin clot which looks something like a mesh. This permanent clot is what finally stops the bleeding. (See **Figure 1**)

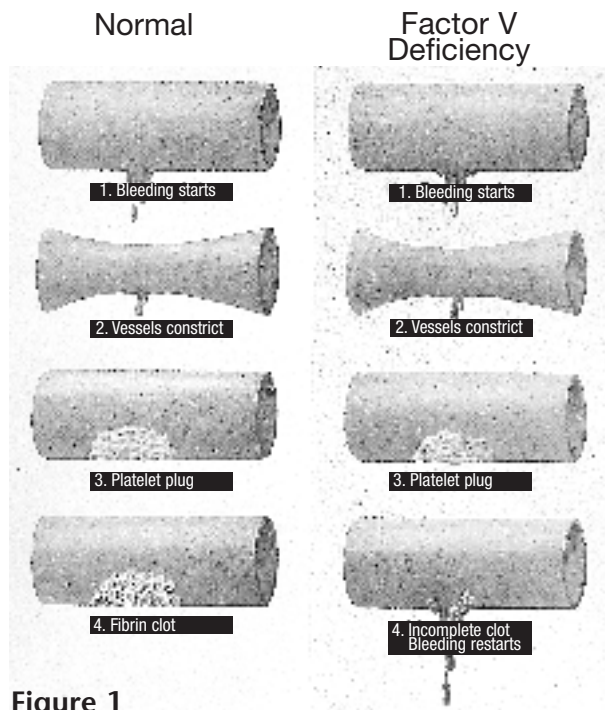


Figure 1

These proteins (factors I, II, V, VIII, IX, X, XI, XII and XIII, as well as von Willebrand factor) are triggered in a kind of domino effect, a chain reaction that is called the coagulation cascade.

(See **Figure 2**)

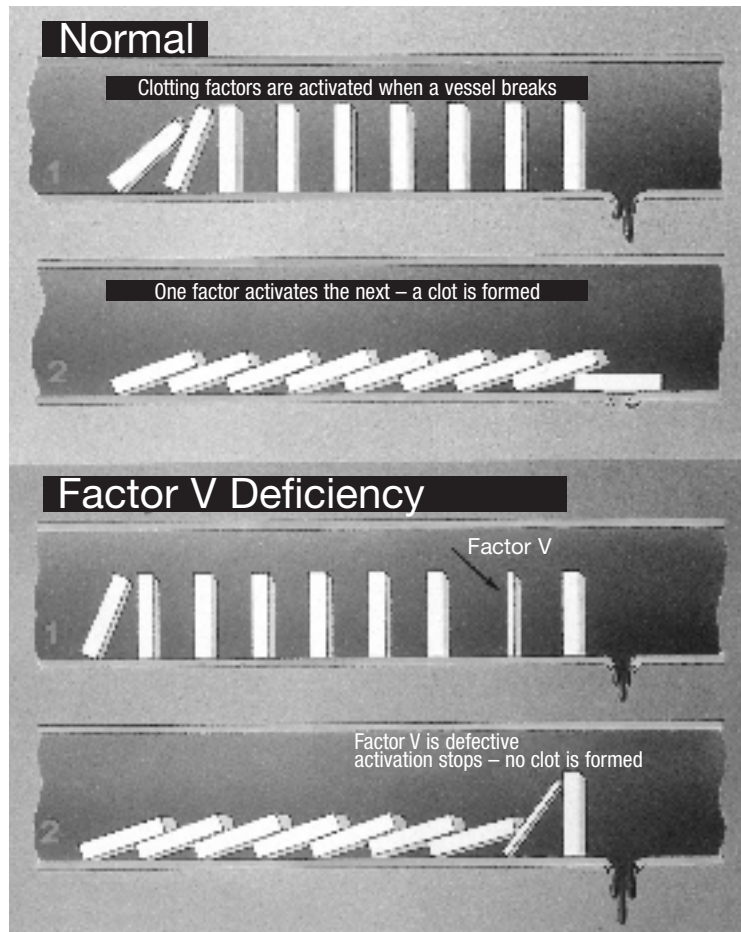
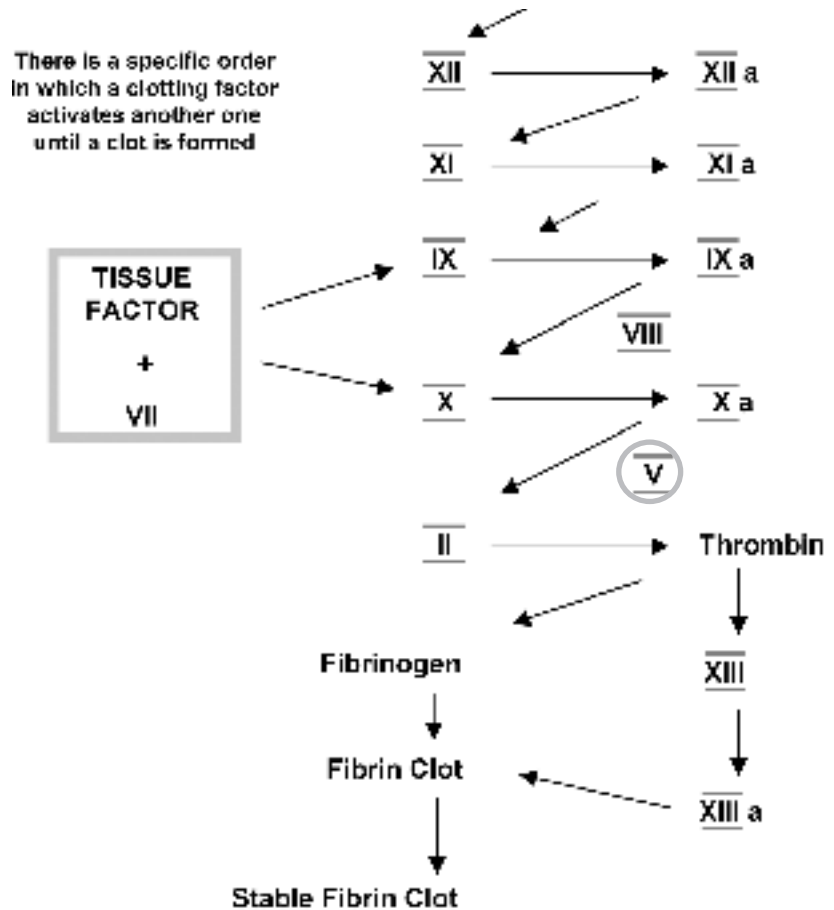


Figure 2

Activation by contact



Source: Modified from Ks, P. M. and John, G. (eds) (eds) Hematology, 2nd Edition, St. Louis, C.V. Mosby Co., Chicago, 1987

Figure 3

Factor V is a protein involved in the complex cascade process that takes place when a blood vessel is damaged. (See **Figure 3**)

How Factor V deficiency is inherited

Factor V deficiency is an inherited bleeding disorder. This means that it is passed on from parent to child at the time of conception. It is an *autosomal recessive* disorder, which means that each parent must pass on a defective gene in order for the child to manifest the disorder. When only one of the two parents is a carrier of the gene responsible for the Factor V deficiency and it is passed on to a child, the child will not be affected.

A carrier is someone who carries the defective gene without being affected by the disorder. In order for a person to be affected by Factor V deficiency, he or she must have inherited two defective genes, one from the mother and one from the father. Both parents must therefore be carriers.

If a person inherits the defective gene from only one of the parents, he/she will be a carrier. His/her factor V level will be lower than normal. Symptoms of the disease may be absent or only slightly present.

Each cell of the body contains structures called *chromosomes*. A chromosome is a long chain of chemicals known as DNA. This DNA is arranged into about 30,000 units called *genes*. These genes determine such things as the colour of a person's eyes. In the case of factor V deficiency, one of these genes has a defect. The defective gene in factor V deficiency is on a chromosome that does not decide the sex of the child. This means that factor V deficiency can affect females as well as males.

The five illustrations below show how FV deficiency can be passed on for each pregnancy.

Figure 4 shows what can happen when both parents are carriers. There is one chance that the child will be normal, one chance that it will have the disorder, and there are two possibilities that the child will be a carrier.

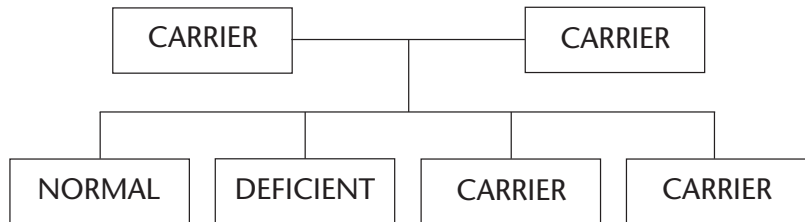


Figure 4

Figure 5 shows what can happen when both parents have FV deficiency. All their children will also have FV deficiency.

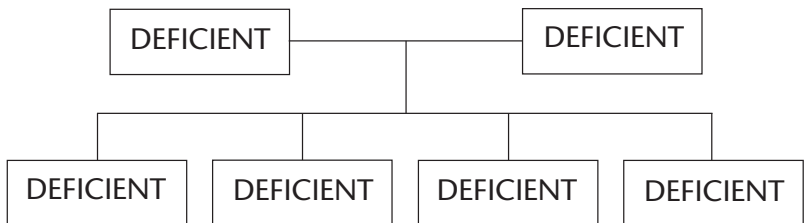


Figure 5

Figure 6 shows what can happen when one of the parents has the disorder and the other is normal. All their children will be carriers of FV deficiency.

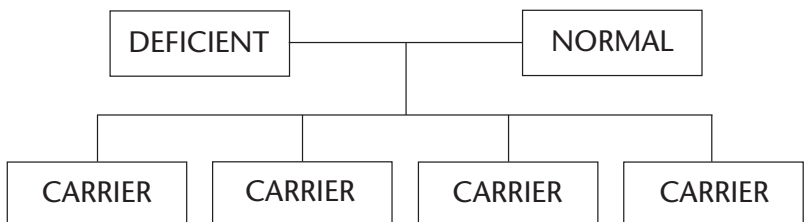


Figure 6

Figure 7 shows what can happen when one of the parents is a carrier and the other is normal. There are two possibilities that the child will be a carrier and two possibilities that it will be normal.

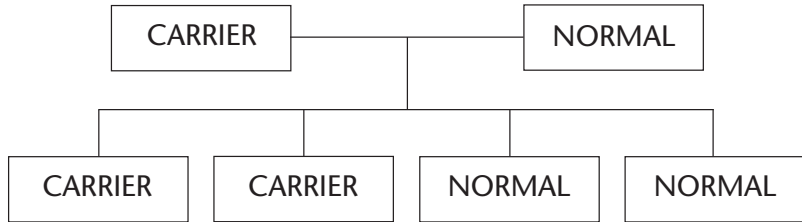


Figure 7

Figure 8 shows what can happen when one of the parents has the disorder and the other is a carrier. There are 2 possibilities that the child will have the disorder and 2 possibilities that it will be a carrier.

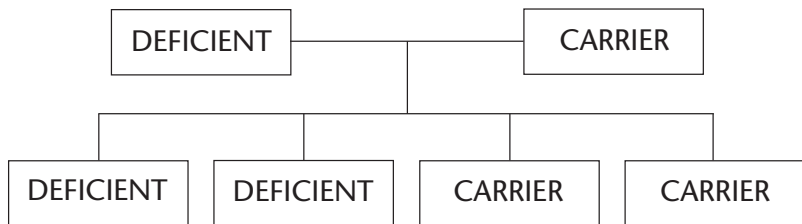


Figure 8

Incidence

Factor V deficiency is an extremely rare disorder. It is estimated that one person in a million has factor V deficiency. Only one hundred and fifty cases have been identified worldwide to date.

Diagnosis

Factor V deficiency is usually discovered in a patient by accident through a routine coagulation blood test done prior to surgery or following a family history of coagulation problems. The physician will order a blood test called PT (prothrombin time) and PTT (cephalin time). These tests measure the time it takes for a clot to form. In the case of factor V deficiency, the PT and PTT will be prolonged. The coagulation time will be longer than normal in a third of patients with severe deficiency. A specific dose of factor V level is needed to confirm the diagnosis. A factor V deficiency is diagnosed if the factor V level is below normal.

It is also important to check the blood level of factor VIII. This is because another inherited bleeding disorder combines factor V and factor VIII deficiencies. The two disorders must be distinguished before appropriate treatment can be administered.

Symptoms

By comparison with some other coagulation factor deficiencies, the bleeding that accompanies factor V deficiency is generally harmless. Factor levels are generally between 5% and 30%. These levels are not generally associated with a hemorrhagic tendency. A factor V blood level between 10% and 20% of normal is enough to prevent bleeding, even after surgery.

The most common symptoms of factor V deficiency are:

- nosebleeds (in patients with both light and moderate deficiency);
- internal hemorrhaging (bruises) at the slightest trauma;
- abundant or prolonged menstruations (menorrhagia);
- bleeding after a trauma (wound);
- bleeding after surgery;
- occasional post-partum hemorrhaging;
- hemarthrosis (bleeding into joints), rarely;
- a few cases of gastro-intestinal bleeding and central nervous system bleeding (brain) have been reported.

The severity of the symptoms may vary within a single family.

Treatment

It is very important to mention this deficiency to a dentist, physician, and/or other specialists in order to prevent a delay or cancellation of surgery.

Treatments are needed only in major bleeding or as part of pre-operation preparation.

Fresh frozen plasma is the usual treatment for factor V deficiency. DDAVP and antifibrinolytic agents (agents that prevent clots from dissolving) will be used sometimes, depending on the circumstances.

Most nosebleeds are controlled with local measures and antifibrinolytics.

Recommendation for the patient with factor V deficiency

It is important to wear a *MedicAlert* bracelet chain engraved with the bleeding disorder you have at all times. Even if the person has never had significant bleeding, this will inform medical professionals of the coagulation disorder. As a result, any questions about coagulation that may arise as a result of abnormal blood tests will be avoided, and the person can be cared for more easily.

The comprehensive care team

As the name suggests, a hemophilia comprehensive care team provides most of the medical services required by a child or adult with an inherited bleeding disorder.

The team is composed of several professionals, including:

- a medical director, usually a hematologist
- a nurse coordinator
- a physiotherapist
- a social worker
- a psychologist.

The team works closely with specialists – a surgeon, an orthopedist, a rheumatologist, a dentist, a geneticist and a psychiatrist, among others. The purpose of this multidisciplinary team is to ensure the well being of the patient and, in the case of a child, the parents as well.

Conclusion

Factor V deficiency is an extremely rare bleeding disorder. It affects both men and women and in some cases, can cause serious bleeding.

Factor V deficiency can also be combined with factor VIII deficiency. It is necessary to distinguish between these two deficiencies in order to be able to administer appropriate treatment.



For more information

You can obtain a list of Hemophilia Treatment Centres by contacting the National Office of the Canadian Hemophilia Society at the following address:

Canadian Hemophilia Society
625 President-Kennedy Avenue
Suite 505
Montréal, Québec H3A 1K2
Telephone: (514) 848-0503
Toll free: 1-800-668-2686
Email: chs@hemophilia.ca
Web site: www.hemophilia.ca

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