hämophilie von-willebrand zentrum köln

PATIENT BROCHURE

Specialized center in Cologne for the diagnosis and treatment of all bleeding disorders, providing comprehensive care and support.

EAHAD

Certified center: European Association for Haemophilia and Allied Disorders (EAHAD)

CONTENTS

IMPRINT
Dr. med. Kai Severin
Cologne Haemophilia and Von Willebrand Centre
Aachener Str. 313, 50931 Cologne
Status: September 2026

Design: C. Harbarth • www.designplus.de

01 WELCOME TO THE COLOGNE HAEMOPHILIA AND VON WILLEBRAND CENTRE

About Us

We are a specialized center in Cologne for the diagnosis and treatment of haemophilia, von Willebrand disease (VWD), rare coagulation factor deficiencies and platelet disorders. We are certified by EAHAD (European Association for Haemophilia and Allied Disorders) as a Haemophilia Treatment Centre (EHTC).

Our focus is on the diagnosis and treatment of all bleeding disorders. We aim to provide comprehensive care and support you with all medical issues.

The Cologne Haemophilia Centre was founded with the aim of providing excellent medical care to people with haemophilia and other congenital or acquired bleeding disorders. We work on an interdisciplinary basis and provide a network of specialists covering medical treatment, physiotherapy and psychosocial support.

Our Goal

We want you to be able to lead an uncomplicated life despite the challenges of living with a bleeding disorder. Our focus is on your quality of life, the prevention of complications and support with any problems you may encounter in everyday life.

Our Team

Our team consists of experienced specialist physicians, nurses and therapists who specialize in the treatment of haemophilia. We work closely with other medical institutions and research centers to remain up to date with the latest scientific developments.

Contacts & Cooperation Partners

Internal Medicine – Haematology / Transfusion Medicine

Diagnosis and treatment of bleeding disorders

> **Dr. med. Kai Severin** (Head)
Specialist in Internal Medicine, Haematology and Oncology, Haemostaseology, Psychotherapy

> **Prof. Dr. med. Ralf Karger, MSc**
Specialist in Transfusion Medicine, Haemostaseology, Clinical Epidemiology

Orthopaedics

Treatment of joint and muscle damage

> **Prof. Dr. med. Andreas Strauss**
Orthopaedics, University Hospital Bonn

Physiotherapy

To promote mobility and quality of life

> **Fabienne Schönenborn**
Physiotherapist, “Beweglichmacher”, Cologne

Psychosocial Support

Advice and support for patients and relatives

> **Dipl.-Psych. Birgitt Hein-Nau**

Cooperation Partners

Various cooperation partners in the fields of dentistry, surgery, gynaecology, gastroenterology, infectious diseases, and paediatrics and adolescent medicine.

Services

Individual Treatment Plans
We develop a tailored treatment plan for each patient based on their individual needs.

Prophylaxis with Coagulation Factors and Antibodies
Through regular administration of coagulation factors, preferably as part of medically supervised home treatment, we help prevent bleeding and long-term damage.

Preparation of Replacement-Therapy Plans
In the event of surgery or other procedures, we prepare a supportive treatment plan to prevent bleeding complications.

Pain Management
If you experience pain due to joint or muscle damage, we offer effective solutions for relief.

Psychosocial Counseling
We help you cope with the challenges of living with haemophilia – both as a patient and as a relative.

Emergency Care – Treatment of Acute Bleeding
We are available around the clock in acute bleeding situations.
A stock of all commonly used factor products is available.

Diagnosis and Treatment
Early diagnosis and individualized treatment are crucial for the quality of life of people with haemophilia. Our diagnostic services include state-of-the-art laboratory techniques and genetic testing to determine your bleeding disorder precisely. Based on these findings, we develop a treatment plan tailored to your needs and life situation.

02 IMPORTANT INFORMATION FOR PATIENTS

Contact and Appointments

If you have any questions or would like to make an appointment, we are available to you at any time. You can reach us by telephone, email or via our online contact form:
www.gerinnung-koeln.de | info@gerinnung-koeln.de

Adress
Cologne Haemophilia and Von Willebrand Centre,
Aachener Str. 313, 50931 Cologne

Telephone

Appointments:	0221 99515-0
Haemophilia organization:	0176 21940868
Emergency Dr. Severin:	0178 2043413

Opening hours

Monday, Tuesday, Thursday	08:00 – 16:00 Uhr
Wednesday, Friday	08:00 – 13:00 Uhr

Saturday, Sunday and public holidays closed
In an emergency: 0178 2043413

Further information

Emergency Number

For acute bleeding situations, we have a 24/7 emergency number:
0178 2043413

Coagulation Factor Therapy

A stock of all commonly used products is available. Through our cooperating specialist pharmacies, all relevant medications can be ordered and delivered at short notice. Of course, medications can also be obtained through your local pharmacy.

Patient Organizations

We can put you in contact with patient organizations and also regularly offer information events.

Interessengemeinschaft Hämophiler e.V.	www.igh.info
Deutsche Hämophiliegesellschaft	www.dhg.de

Cooperation with the Haemophilia Centre at University Hospital Bonn

There is close cooperation with University Hospital Bonn, Institute for Experimental Haematology and Transfusion Medicine, Venusberg Campus 1, 53127 Bonn – Haemophilia Centre.
Telephone: 0228 28715188

Close Cooperation and Networking with Hospitals and Medical Practices in the Region

here is close and effective cooperation with physicians in private practice, all departments of hospitals in Cologne and the surrounding area, and all departments of University Hospital Cologne.

Cooperation with the “Beweglichmacher” Physiotherapy Practices

The experienced physiotherapists at Beweglichmacher support us and our patients: **www.beweglichmacher.de**

Standorte:

Cologne Aachener Str.
Aachener Straße 60-62
50674 Cologne
0221 25 27 23

Cologne Weiden
Bunzlauer Straße 1
50858 Cologne
02234 4303398

Erftstadt
Otto Hahn Allee 14A
50374 Erftstadt
02235 6858366

Cologne Riehler Straße
Riehler Straße 36
50668 Cologne
0221 80008041

Cologne Breite Str.
Breite Straße 80-90
50667 Cologne
0221 57070676

Hürth
Beseler Straße 1
50354 Hürth
02233 377 80 50

Frechen
Augustinusstraße 11C
50226 Frechen
02234 9898428

They have particular expertise in joint changes resulting from congenital bleeding disorders (haemophilia and VWD, especially type 3), but also provide expert support for all other issues. You are, of course, free to choose your own physiotherapy practice.

Physiotherapy prescriptions are issued by us without difficulty as part of individualized treatment plans.

03 CLINICAL CONDITIONS

HAEMOPHILIA A

What is Haemophilia A?

Haemophilia A is a congenital, X-linked inherited bleeding disorder characterized by a deficiency of coagulation factor VIII (8).
Since factor VIII plays a central role in blood coagulation, a deficiency results in an increased risk of bleeding, particularly after injuries or surgery.

Symptoms of Haemophilia A

- Frequent and unexplained bruising (haematomas)
- Prolonged bleeding after injuries or minor cuts
- Pain and swelling in joints (especially the knees, elbows and ankles) due to internal bleeding
- Spontaneous bleeding, especially into muscles or joints, even without an identifiable external injury
- Severe bleeding after surgical procedures or dental treatment

Severe forms are generally diagnosed during childhood.

Severity of Haemophilia A

> Severe:
Factor VIII levels below 1% of normal; severe spontaneous bleeding (particularly into joints) or bleeding following minor injuries.

> Moderate:
Factor VIII levels between 1% and 5%; bleeding after major injuries or surgery. Spontaneous joint bleeds are less common.

> Mild:
Factor VIII levels between 5% and 40%; bleeding occurs mainly after severe injuries or surgery.

Treatment of Haemophilia A

Treatment is primarily based on regular administration of factor VIII concentrate to replace the deficient coagulation factor. Other therapeutic options include, for example, anti-TFPI antibodies[1] or factor VIII mimetics[2].
The standard approach is coagulation factor prophylaxis, which aims to prevent spontaneous bleeding.

[1] Anti-TFPI antibodies are therapeutic monoclonal antibodies that block Tissue Factor Pathway Inhibitor (TFPI), thereby strengthening blood coagulation in haemophilia.

[2] Factor VIII mimetics are bispecific monoclonal antibodies. They bind simultaneously to factors IXa and X to initiate the normal coagulation cascade and thereby protect against bleeding.

HAEMOPHILIA B

What is Haemophilia B?

Haemophilia B, also known as “Christmas disease”[1], is a bleeding disorder similar to haemophilia A, but it is caused by a deficiency of coagulation factor IX (9). This disorder is also X-linked and predominantly affects males. Factor IX deficiency results in an increased risk of bleeding and symptoms similar to those of haemophilia A.

[1] Named after Stephen Christmas, who was the first to describe factor IX deficiency (haemophilia B) in a scientific journal in 1952.

Symptoms of Haemophilia B

- Frequent bruising and unusually prolonged bleeding
- Joint bleeding, particularly in large joints such as the knees, elbows and hips
- Spontaneous bleeding without an external cause
- Bleeding after surgery and dental treatment

Schweregrade der Hämophilie B

> Severe:
Factor VIII levels below Factor IX levels below 1%; spontaneous bleeding (particularly into joints) or bleeding following minimal injury.

> Moderate:
Factor IX levels between 1% and 5%; bleeding occurs after major injuries. Spontaneous joint bleeds are less common.

> Mild:
Factor IX levels between 5% and 40%; bleeding occurs after more severe injuries or surgery.

Treatment of Haemophilia B

Treatment includes administration of **factor IX concentrate** to correct the deficiency and control bleeding. Alternatively, anti-TFPI antibodies[1] can be administered subcutaneously using a pen.

Here too, regular prophylaxis plays a key role in preventing joint and muscle damage caused by recurrent bleeding.

[1] Anti-TFPI antibodies are therapeutic monoclonal antibodies that block Tissue Factor Pathway Inhibitor (TFPI), thereby strengthening blood coagulation in haemophilia.

VON WILLEBRAND DISEASE

What is Von Willebrand Disease?

Von Willebrand disease (VWD) is the most common inherited bleeding disorder and affects both men and women. It is caused by a deficiency or dysfunction of von Willebrand factor (vWF). This factor is important for blood coagulation and supports the binding and transport of platelets to the vessel wall as well as the activation of factor VIII.

Symptoms of Von Willebrand Disease

- Frequent and prolonged nosebleeds
- Frequent bleeding from the gums, especially during dental hygiene
- Excessive bleeding from small wounds or injuries
- Menstrual bleeding that is difficult to stop (menorrhagia)
- Bruising without an identifiable cause
- Less common but also severe bleeding, for example after surgery or accidents

Severity of Von Willebrand Disease

> Typ 1
Mild deficiency of von Willebrand factor; bleeding is usually mild to moderate.

> Typ 2
Dysfunction of von Willebrand factor; usually a moderate bleeding tendency.

> Typ 3
Severe deficiency of von Willebrand factor; symptoms resemble those of haemophilia and very severe bleeding may occur.

Diagnosis of Von Willebrand Disease

Diagnosis is based on a combination of clinical observations and specific laboratory tests:

1. Bleeding history
A detailed family history and consideration of accompanying diseases or medications are essential for diagnosis.

- Factor VIII level: Since vWF is the transport protein for factor VIII, factor VIII levels are usually also low in vWF deficiency.
- A vWF antigen (vWF:Ag) test measures the level of von Willebrand factor in the blood. A low level indicates acquired von Willebrand disease.
- vWF activity test (formerly ristocetin cofactor test): a laboratory value used to measure the function of von Willebrand factor (VWF) in the blood.

Treatment of Von Willebrand Disease

Treatment depends on the severity of the disease. In mild forms, desmopressin therapy is generally sufficient for prophylaxis during surgery or procedures. Desmopressin is a substance that stimulates the release of vWF from stores in the body.
In most cases, however, vWF is directly replaced using a plasma-derived or recombinant von Willebrand factor concentrate (see also page 21).
Another important component of treatment is the antifibrinolytic agent tranexamic acid, which is usually administered orally and, if necessary, intravenously.

COMMON FEATURES & DIFFERENCES

Similarities

- All three disorders lead to prolonged bleeding time and an increased tendency to bleed.
- All are inherited to varying degrees; haemophilia A and B are X-linked[1], while von Willebrand disease is inherited in an autosomal manner.
- Treatment of all three disorders is based on replacement of missing or defective coagulation factors, monoclonal antibodies or modulatory therapies.

Differences

- **Haemophilia A and B** involve a deficiency or dysfunction of a single factor (VIII and IX, respectively), whereas von Willebrand disease involves a problem with an important factor for blood coagulation and platelet aggregation.
- **Von Willebrand** disease occurs in both sexes, whereas haemophilia A and B predominantly affect males.

[1] In X-linked inheritance, the patient's mother transmits the genetic alteration affecting factor VIII or IX. However, spontaneous mutations occur in up to 30% of cases.

ACQUIRED VON WILLEBRAND DISEASE

What is Acquired Von Willebrand Disease (aVWD)?

Acquired von Willebrand disease (aVWD) is a rare but serious bleeding disorder caused by dysfunction or deficiency of von Willebrand factor (vWF) in the blood. Unlike congenital von Willebrand disease (VWD), which is genetically inherited, the acquired form develops during a person's lifetime and is not genetic.

Acquired von Willebrand disease is an autoimmune disorder in which the body produces antibodies against von Willebrand factor that either inactivate vWF or impair its function. Increased breakdown of von Willebrand factor may also occur, significantly disrupting blood coagulation.

What is von Willebrand Factor?

Von Willebrand factor (vWF) is an important component of blood coagulation. It ensures that platelets adhere to damaged blood vessels and simultaneously forms a bond with factor VIII, an important coagulation factor that activates the coagulation cascade.
Without sufficient functional vWF, platelets cannot efficiently perform their role in blood coagulation, resulting in an increased tendency to bleed.

Causes of Acquired Von Willebrand Disease

Acquired von Willebrand disease is often associated with other underlying diseases that trigger an autoimmune response. Common causes include:

1. Malignant diseases (cancer)

In particular, lymphomas, leukaemias and solid tumors (e.g. carcinomas) can lead to acquired von Willebrand disease. In cancer patients, blood coagulation is often affected by the tumor disease or its treatment.

2. Autoimmune diseases

Autoimmune diseases such as systemic lupus erythematosus (SLE) or rheumatoid arthritis can promote the development of antibodies against vWF.

3. Cardiovascular diseases

Patients with severe heart disease, particularly those with mechanical heart valves, may develop acquired von Willebrand disease due to platelet activation and changes in vWF.

4. Drug-induced forms

In some cases, acquired von Willebrand disease may be triggered by medications such as phenytoin, heparin or valproic acid.

5. Infections

Viral diseases or bacterial infections can trigger acquired von Willebrand disease.

Symptoms of Acquired Von Willebrand Disease

Symptoms of acquired von Willebrand disease are similar to those of the **congenital form**, although their severity and type can vary considerably.
Common symptoms include:

- Frequent bleeding into the skin (haematomas).
- Nosebleeds and gum bleeding, particularly without an identifiable trigger.
- Prolonged menstrual bleeding (menorrhagia).
- Prolonged bleeding after minor injuries or surgical procedures.
- Severe bleeding after major surgery or trauma.
- Spontaneous bleeding, particularly internal bleeding such as joint or gastrointestinal bleeding.
- Fatigue or pallor, which may indicate recurrent blood loss.

Diagnosis of Von Willebrand Disease

Diagnosis is based on a combination of clinical observations and specific laboratory tests:

1. Bleeding history

A detailed family history and consideration of accompanying diseases or medications are essential for diagnosis.

- Factor VIII level: Since vWF is the transport protein for factor VIII, factor VIII levels are usually also low in vWF deficiency.
- A vWF antigen (vWF:Ag) test measures the level of von Willebrand factor in the blood. A low level indicates acquired von Willebrand disease.
- vWF activity test (formerly ristocetin cofactor test): a laboratory value used to measure the function of von Willebrand factor (VWF) in the blood.

2. Specific Antibody Tests (in Acquired VWD)

Detection of antibodies against von Willebrand factor is crucial for diagnosing the acquired form. These antibodies block the function of vWF and may impair its activity.

3. Multimer Analysis

vWF multimer analysis is a specialized blood test used to examine the structure of von Willebrand factor (vWF) and precisely classify different forms of von Willebrand syndrome (VWS), an inherited or acquired bleeding tendency. It distinguishes quantitative defects (vWF deficiency) from qualitative defects (structural or functional abnormalities of the protein). The analysis is essential for identifying rare or complex subtypes of von Willebrand syndrome (such as types 2A, 2B, 2M or 2N), which differ in their electrophoretic patterns.

4. Collagen Binding Activity (CBA)

Collagen binding activity (CBA) assesses the functionality of von Willebrand factor, specifically its ability to bind to subendothelial collagen. The VWF:CBA test is helpful in distinguishing between different types of VWD in patients suspected of having type 2A, 2B or 2M.

5. Determination of Blood Group

The concentration of vWF in the blood depends on the patient's blood group. Concentration and activity levels are physiologically often significantly lower in patients with blood group O than in people with blood group A, B or AB.

Treatment of Acquired Von Willebrand Disease

Treatment of acquired von Willebrand disease depends on disease severity, **bleeding risk** and the underlying cause. Several therapeutic approaches are available:

1. Treatment of the underlying cause (in acquired VWD)

- In patients with an autoimmune disease or malignancy, treatment often focuses on the underlying disease. In cancer, chemotherapy or immunotherapy may be necessary.
- In heart disease, adjustment of medication may be required.

2. Replacement therapy with von Willebrand factor

- Factor VIII concentrate (which also contains von Willebrand factor) is often administered for prophylaxis or treatment of severe bleeding. A genetically engineered pure VWF product is available.

3. Desmopressin (DDAVP)

- In some cases, desmopressin can be used to stimulate the release of vWF from vessel walls, which may improve blood coagulation.

4. Immunosuppressive therapy (in acquired VWD)

- If the disorder is caused by autoimmune processes, immunosuppressants such as corticosteroids, rituximab or azathioprine may be required to suppress antibody production.

ACQUIRED HAEMOPHILIA

What is Acquired Haemophilia?

Acquired haemophilia is an autoimmune disorder in which the body produces antibodies directed against coagulation factors, particularly factor VIII. These antibodies, also called inhibitors, prevent the normal function of the affected coagulation factor and reduce the blood's ability to clot, resulting in unusually severe and prolonged bleeding.

Unlike congenital haemophilia, which is present from birth and inherited, acquired haemophilia usually develops suddenly and can affect people of any age, including those without a previous illness or family history.

Causes of Acquired Haemophilia

The exact causes of inhibitor formation are not yet fully understood. However, several **associated risk factors** and underlying diseases may promote the development of acquired haemophilia

- Autoimmune diseases: In diseases such as rheumatoid arthritis or systemic lupus erythematosus, the immune system may mistakenly attack the body's own coagulation factors.
- Malignant diseases: Acquired haemophilia can occur particularly in cancer, especially blood cancers such as leukaemia or lymphoma.
- Medication: Certain medications, such as penicillin or heparin, can rarely trigger acquired haemophilia.
- Pregnancy: In very rare cases, acquired haemophilia can occur during pregnancy, particularly in association with postpartum complications.
- Infections: In rare cases, a viral infection (e.g. hepatitis or HIV) may act as a trigger.

Symptoms of Acquired Haemophilia

Symptoms resemble those of **congenital haemophilia** but are often more severe and can develop very rapidly. Common symptoms include:

- Spontaneous bleeding: particularly extensive haematomas that develop without an identifiable external injury.
- Prolonged bleeding: bleeding after minor injuries, surgery or even injections may last much longer than usual.
- Joint bleeding: this often occurs in large joints such as the knees, elbows or shoulders, causing pain and restricted movement.
- Gastrointestinal bleeding: this may present as blood in the stool or black, tarry stool (melaena).
- Blood in the urine: red or pink urine may indicate bleeding in the urinary tract.
- Less common symptoms: in very severe cases, internal bleeding into vital organs may occur and can be life-threatening.

Treatment of Acquired Haemophilia

Treatment of acquired haemophilia (autoantibodies against factor VIII) is based on two main pillars: acute haemostasis (bypassing therapy) and immediate immunosuppression to eliminate the inhibitor.

A factor VIII mimetic is often used as bridging therapy. The aim is to stop production of autoantibodies as quickly as possible. Corticosteroids (e.g. prednisolone) are usually used first, often in combination with cyclophosphamide. If treatment fails, the monoclonal antibody rituximab may also be used.
In some cases, acquired haemophilia occurs without an identifiable underlying disease or trigger.
Less common but also severe bleeding may occur, for example after surgery or accidents.

RARE COAGULATION FACTOR DEFICIENCIES

What are Rare Coagulation Factor Deficiencies?

Rare factor deficiencies are bleeding disorders caused by the absence or deficiency of specific coagulation factors. These disorders are much rarer than haemophilia A and B and involve different coagulation factors.

Symptoms are similar to those of haemophilia but may vary depending on the affected factor. The following is an overview of some of the most important rare factor deficiencies.

1. Haemophilia C (Factor XI Deficiency)

Was ist Hämophilie C?

Haemophilia C is a very rare bleeding disorder caused by a deficiency of factor XI.

It is usually inherited in an autosomal recessive manner, meaning that both parents must carry the defective gene for the disorder to occur. Unlike haemophilia A and B, it affects both men and women.

Factor XI deficiency impairs the coagulation cascade, resulting in prolonged bleeding.

Symptoms

- Frequent bleeding after surgery or injuries, but no spontaneous joint bleeding as in haemophilia A or B.
- Prolonged bleeding after surgical procedures or tooth extractions.
- Milder symptoms compared with haemophilia A and B, which is why the condition is often not discovered until adulthood.

Treatment

- For surgery or severe bleeding, treatment is usually with human fresh frozen plasma (FFP), as specific purified factor XI concentrates are not established as standard finished products in Germany.
- In less severe cases, medications such as desmopressin may be used.

2. Factor VII Deficiency

What is Factor VII Deficiency?

Factor VII deficiency is a rare autosomal recessive disorder that impairs blood coagulation. Factor VII is essential for initiating the coagulation cascade. The disorder can range from mild to severe depending on the amount of factor VII in the blood.

Symptoms

- Frequent nosebleeds and gum bleeding
- Haematomas after minor injuries
- In severe cases, internal bleeding, joint bleeding or gastrointestinal bleeding may occur.

Treatment

Factor VII concentrate is used to treat and prevent bleeding. For prophylaxis during surgery or in the event of bleeding, a plasma-derived or activated recombinant factor VII preparation (FVIIa) may be used.

3. Factor X Deficiency

What is Factor X Deficiency?

Factor X deficiency is a very rare autosomal recessive disorder in which insufficient amounts of coagulation factor X are present. The deficiency impairs the coagulation-promoting cascade, resulting in an increased tendency to bleed.

Symptoms

- Haematomas, nosebleeds and gum bleeding
- Severe bleeding after surgical procedures
- Spontaneous joint bleeding is rare but may occur in severe cases.

Treatment

Specific factor X concentrates or prothrombin complex concentrate (PCC; PPSB) are used to replace the coagulation factor and control bleeding.

4. Factor V Deficiency (Owren Syndrome)

What is Factor V Deficiency?

Factor V deficiency, also known as Owren syndrome, is a rare bleeding disorder in which factor V is present in too small an amount or in an inactive form. Factor V is a cofactor in the coagulation cascade that works with other factors to promote blood clotting.

Symptoms

- Prolonged bleeding after minor injuries or surgery
- Haematomas and nosebleeds
- In severe cases, internal bleeding or joint bleeding may also occur.

Treatment

A factor V concentrate for the treatment of bleeding is not available, so treatment must be given with fresh frozen plasma. However, fresh frozen plasma contains relatively low amounts of coagulation factors (1 mL of fresh frozen plasma contains 1 unit of coagulation factor).
Occasionally, a combined factor V and factor VIII deficiency is present.

5. Factor XIII Deficiency (Fibrin Stabilization)

What is Factor XIII Deficiency?

Factor XIII plays a crucial role in cross-linking fibrin, which ensures that the blood clot remains stable and is not dissolved. Factor XIII deficiency results in unstable clot formation, which can cause a clot to break down rapidly after bleeding.
Factor XIII is not detected by the global coagulation tests Quick and aPTT and is therefore often overlooked.

Symptoms

- Recurrent bleeding and difficulty with wound healing
- Bleeding into the skin and recurrent haematomas
- Spontaneous bleeding into joints, muscles and internal organs may occur in more severe cases.

Treatment

- Factor XIII concentrates are used to treat bleeding.
- If unavailable, fresh frozen plasma or cryoprecipitate[1] may also be used because these products contain factor XIII.

[1] Cryoprecipitate is a blood product derived from plasma and produced by slowly thawing frozen fresh plasma. Cryoprecipitate contains fibrinogen, factor VIII, factor XIII and von Willebrand factor (vWF).

6. Hageman Factor Deficiency (Factor XII Deficiency)Was ist der

What is Hageman Factor Deficiency?

Factor XII (Hageman factor) is part of the intrinsic coagulation cascade. However, deficiency of this factor does not normally result in increased bleeding.
Factor XII deficiency is often discovered incidentally during coagulation testing because patients generally remain asymptomatic.

Symptoms

- Almost always asymptomatic, as factor XII deficiency does not cause significant bleeding problems.
- Factor XII deficiency causes a prolonged aPTT, which often leads to further investigation before surgery.

Treatment

- Specific treatment is generally not required.

Rare coagulation factor deficiencies are a group of bleeding disorders caused by the absence or deficiency of specific coagulation factors. Symptoms are often similar to those of haemophilia, but different factors and treatment approaches are involved. Treatment is generally based on replacement therapy with specific coagulation factors or plasma, depending on the individual disorder.

Because these disorders are very rare, diagnosis and treatment require close cooperation with specialized haemostaseologists at specialized centers. With correct diagnosis and appropriate treatment, patients with these rare disorders can lead a normal life.

THROMBOCYTOPATHIES

What are thrombocytopathies?

Thrombocytopathies are a group of blood clotting disorders characterised by a dysfunction of the thrombocytes (platelets). Unlike thrombocytopenia (where the platelet count in the blood is too low), in thrombocytopathies the platelet count is normal or even elevated, but the function of the platelets is impaired. This dysfunction leads to an increased tendency to bleed, as the platelets are unable to perform their role in haemostasis and wound healing effectively. Thrombocytopathies can be either congenital or acquired. Below you will find a detailed description of the most important thrombocytopathies, their causes, symptoms and treatment options.

1. Congenital thrombocytopathies

A) Glanzmann Thrombasthenia

What is Glanzmann Thrombasthenia?

Glanzmann thrombasthenia is a rare autosomal recessive inherited disorder characterized by dysfunction of integrin glycoproteins on the surface of platelets, particularly GPIIb/IIIa. These integrins are important for platelet aggregation (the clumping of platelets) in order to form blood clots.
Due to this dysfunction, platelets cannot aggregate properly, resulting in delayed or inadequate haemostasis.

Symptoms

- Spontaneous bleeding, particularly haematomas and nosebleeds
- Prolonged menstrual bleeding (menorrhagia)
- Bleeding after minor injuries, injections or tooth extractions
- Severe bleeding after surgical procedures
- Joint bleeding is rare but can occur if the condition is not treated appropriately

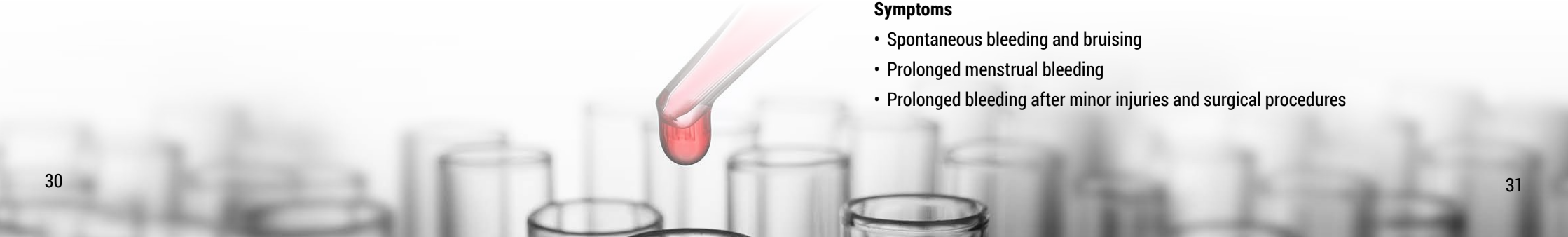
Treatment

- The most important therapeutic option is administration of activated factor VII (FVIIa).
- Platelet concentrates should be avoided whenever possible because of the risk of antibody formation.
- Desmopressin may be helpful in some patients because it stimulates the release of platelet granules.

B) Bernard-Soulier Syndrome

What is Bernard-Soulier Syndrome?

Bernard-Soulier syndrome is an autosomal recessive inherited disorder caused by a deficiency or dysfunction of glycoprotein GPIb-IX-V on the surface of platelets. This glycoprotein is responsible for platelet adhesion to the vessel wall following injury. Without functional GPIb, platelets cannot adequately perform their ability to aggregate and adhere to damaged blood vessels.



Symptoms

- Spontaneous bleeding such as nosebleeds (epistaxis), gum bleeding and haematomas
- Prolonged bleeding after minor injuries or surgery
- Severe postoperative bleeding
- Joint bleeding is less common than in other platelet disorders, but may occur in severe cases

Treatment

- Platelet concentrates are the main treatment.
- Desmopressin may be used in some patients to promote platelet aggregation.
- In severe bleeding, supportive coagulation treatment with recombinant factor VIIa may be used.

C) Storage Pool Disease (SPD)

What is Storage Pool Disease?

Storage pool disease is a group of disorders in which the granules in platelets (stores of substances such as ADP, serotonin and calcium) do not function properly or cannot be released. These granules are responsible for platelet aggregation and the secretion of signaling substances that are important for blood coagulation.
There are two main types: primary and secondary storage pool disease, depending on whether the cause is inherited or acquired.

Symptoms

- Spontaneous bleeding and bruising
- Prolonged menstrual bleeding
- Prolonged bleeding after minor injuries and surgical procedures

Treatment

- Platelet concentrates are used to help in severe bleeding.
- Desmopressin may help in some cases.
- The antifibrinolytic agent tranexamic acid is an important basic therapy. It reversibly blocks the binding sites of plasminogen, thereby preventing its activation to plasmin and stopping premature breakdown of fibrin. The already formed blood clot remains stabilized.

2. Acquired thrombocytopathies

A) Drug-induced thrombocytopathies

What is a Drug-induced thrombocytopathies?

One of the most common acquired platelet disorders is caused by medications such as aspirin (acetylsalicylic acid) or clopidogrel. These medications block platelet aggregation receptors or cyclooxygenase (COX) enzymes responsible for the formation of thromboxane A2, a potent activator of platelet aggregation.

These medications are frequently used for thrombosis prophylaxis and the treatment of cardiovascular diseases.

Symptoms

- Frequent minor bleeding such as nosebleeds or gum bleeding
- Prolonged bleeding after surgery
- Bruising after minimal injury

Treatment

If necessary, medication may be reduced or discontinued depending on the underlying disease if the platelet disorder causes clinical symptoms.

B) Liver Disease and Platelet Disorders

What is the Connection Between Liver Disease and Platelet Disorders?

Liver diseases, particularly cirrhotic changes, can cause secondary platelet dysfunction. This is because the liver not only produces coagulation factors but also influences platelet activation and adhesion. In addition, thrombocytopenia (a low platelet count) caused by splenomegaly (enlargement of the spleen) in liver cirrhosis can impair platelet function.

Symptoms

- Skin bleeding, gum bleeding or haematomas
- Prolonged bleeding after injections or dental treatment
- In severe cases, internal bleeding may also occur

Treatment

- Treatment of the liver disease to improve platelet function
- Platelet concentrates may help stabilize blood coagulation

Conclusion

Platelet disorders are a group of bleeding disorders caused by impaired platelet function. These disorders can be congenital or acquired and generally lead to an increased tendency to bleed due to inadequate platelet function. Treatment depends on the underlying cause and often includes platelet concentrates, medication management and, in some cases, coagulation factor therapies.

04 NOTES

hämophilie von-willebrand zentrum köln

www.gerinnung-koeln.de