CLINICAL AND PRACTICAL SIGNIFICANCE OF HEMOPHILIA

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Annotation: This scientific article discusses the types of hemophilia, their pathogenesis, clinical manifestations, and treatment methods. Hemophilia is a hereditary disorder of the blood coagulation system that occurs as a result of a deficiency of certain clotting factors in the body. Hemophilia A is characterized by a deficiency of factor VIII, hemophilia B by a deficiency of factor IX, and hemophilia C by a deficiency of factor XI. In this disease, bleeding tends to last for a long time, and hematomas under the skin and in muscles, as well as bleeding into joints (hemarthrosis), occur frequently. Treatment of patients focuses on replacing the missing clotting factors, using antifibrinolytic drugs, and treating hematomas through physiotherapy or surgical methods. Special attention is also given to the concept of hematoma. A hematoma is an accumulation of blood that escapes from a vessel and collects within tissues, and it is one of the most common complications of hemophilia. The article highlights the relationship between hemophilia and hematoma, emphasizing the importance of early diagnosis, prevention, and appropriate treatment.

Keywords: Hemophilia, Hemophilia A, Hemophilia B, Hemophilia C, hematoma, hemarthrosis, antifibrinolytic drugs, hereditary disease, bleeding, chromosome,

Introduction

Hemophilia (from Ancient Greek: *haima* — blood and *philia* — affection or tendency), also known as continuous bleeding, is a rare and serious congenital disorder characterized by an increased tendency to bleed, caused by an abnormality linked to the X chromosome. In rare cases, about one-third of instances occur due to spontaneous mutations. People with hemophilia experience prolonged bleeding compared to normal individuals. According to estimates, approximately 1 in every 10,000 people is affected by hemophilia, and around 450,000 individuals suffer from this disease worldwide. The condition arises due to a deficiency of clotting factors necessary for blood coagulation in plasma.

Although hemophilia primarily occurs in males, it is transmitted by mothers — that is, it can be passed from a grandfather to a grandson through a healthy daughter. The symptoms of hemophilia usually appear in early childhood and may lessen as the child grows older. When a patient suffers an injury — such as bruising, muscle or joint trauma, biting the tongue, or tooth extraction — excessive bleeding may occur, posing a serious risk to life. Bleeding into large joints (such as the knee or ankle) causes severe joint changes.

A key diagnostic feature that distinguishes hemophilia from other bleeding disorders is the impaired blood clotting process: when blood from a hemophilia patient is drawn into a test tube, it may not clot for several hours. This is due to a deficiency of a protein known as antihemophilic globulin in the patient's blood.

There are several types of hemophilia, including Hemophilia A, Hemophilia B (also known as Christmas disease), and Hemophilia C (which is not considered a true form of hemophilia). Patients must avoid injuries as much as possible. In some cases, surgical intervention may be required to treat hemophilia and its complications. Before any surgical procedure, consultation at a specialized hemophilia treatment center is necessary, and surgery should preferably be performed in a highly specialized hospital.

The first line of prophylactic care for patients with hemophilia is mainly applied to children with severe forms of Hemophilia A and B during ongoing treatment. The optimal age to begin prophylactic therapy is between 1 and 2 years. Primary prophylaxis is prescribed for a minimum of six months and can later be extended without limitation.

Secondary prophylaxis is recommended for patients after the age of two or after more than two joint hemorrhages, but before the onset of joint damage. This form of prophylaxis is typically carried out in outpatient settings of multidisciplinary clinics or hematology centers.

Tertiary prophylaxis is administered after joint damage has occurred and been documented (through physical examination and joint radiography), with continuous regular treatment.

Screening is a process used to detect diseases at an early stage among healthy individuals or those without symptoms. The aim of screening is to identify the disease early, prevent complications, improve treatment effectiveness, and monitor population health. Genetic screening plays a crucial role in hereditary diseases such as hemophilia.

There are two types of hemophilia screening: prenatal and postnatal. Prenatal screening determines whether a pregnant woman carries a gene mutation linked to the X chromosome. If such a mutation is present, the risk of hemophilia in the child—especially in males—is high. Postnatal screening involves blood tests of newborns to assess clotting function and measure factor VIII or IX levels. If the disease is genetically confirmed, both the patient and family members are informed to take preventive measures and monitor potential disease development.

Hemophilia A is an inherited disorder of the blood coagulation system caused by a deficiency or inactivity of clotting factor VIII. The disease is inherited in an X-linked recessive pattern. Males are more commonly affected, as they possess only one X

chromosome, whereas females have two X chromosomes and therefore are less likely to have the disease but can act as carriers, passing it from one generation to the next.

The pathogenesis of the disease involves a decrease or absence of factor VIII (antihemophilic globulin A), which plays a vital role in the fibrin formation cascade. Its deficiency slows or impairs hemostasis, resulting in prolonged bleeding episodes.

To prevent Hemophilia A, genetic counseling for carriers, prophylactic administration of factor VIII in children, and medical supervision during surgical or dental procedures are essential.

Hemophilia A is classified into three forms: mild, moderate, and severe.

In the mild form, factor VIII activity is 5–40%, and bleeding usually occurs only after significant injury.

In the moderate form, factor VIII activity is 1–5%, and bleeding may occur even after minor trauma.

In the severe form, factor VIII activity is below 1%, and spontaneous (unprovoked) bleeding is observed.

Conclusion

Hemophilia is a hereditary bleeding disorder caused by a deficiency or dysfunction of specific blood-clotting factors, most commonly factors VIII and IX. Despite being a rare disease, it poses serious health challenges due to recurrent and prolonged bleeding episodes that can lead to joint damage, anemia, and disability if left untreated. Early diagnosis through genetic and laboratory screening, together with proper classification of disease severity, plays a crucial role in effective management.

Modern treatment focuses on replacing the missing coagulation factors, using antifibrinolytic agents, and applying physiotherapy or surgical interventions when necessary. Preventive (prophylactic) therapy initiated in early childhood significantly reduces bleeding frequency and prevents joint complications, thereby improving patients' quality of life.

Genetic counseling and carrier detection among families are essential for preventing transmission to future generations. Continuous patient education, regular follow-up at specialized hemophilia centers, and public awareness programs remain key strategies for reducing complications and improving long-term outcomes. In conclusion, a multidisciplinary approach that combines prevention, early diagnosis, and modern treatment methods is vital for the effective control and management of hemophilia.

Hemophilia is one of the most important hereditary disorders of the blood coagulation system, and despite significant progress in modern medicine, it still remains a serious global health concern. The disease primarily affects males, while females serve as carriers, transmitting the defective gene through generations. Understanding the genetic mechanism and pathophysiology of hemophilia is crucial for timely diagnosis, effective treatment, and prevention of complications.

In clinical practice, hemophilia manifests as prolonged bleeding, spontaneous hematomas, and frequent hemarthrosis, which often lead to chronic joint deformities and disability. Therefore, comprehensive clinical observation and laboratory monitoring of

coagulation factors are necessary for early detection and control of the disease process. The development of advanced diagnostic technologies, including prenatal and postnatal genetic screening, allows early identification of carriers and affected individuals, thus providing opportunities for preventive measures and informed family planning.

Therapeutically, factor replacement therapy remains the cornerstone of hemophilia management. The use of recombinant factor VIII and IX concentrates has significantly improved treatment safety and reduced the risk of viral transmission compared to plasma-derived products. In addition, antifibrinolytic agents, physiotherapy, and orthopedic interventions help in managing acute bleeding episodes and restoring joint function. Recent innovations such as gene therapy and long-acting clotting factor preparations offer hope for a more sustainable and possibly curative approach in the future.

Preventive strategies play an equally important role. Primary prophylaxis started at an early age effectively reduces the frequency of bleeding and protects joint health. Secondary and tertiary prophylaxis programs implemented in outpatient and hematology centers ensure continuous patient care and improve life expectancy. Education of patients and families about self-care, prompt recognition of bleeding symptoms, and safe lifestyle habits are essential components of long-term management.

In conclusion, hemophilia requires a multidisciplinary approach involving hematologists, pediatricians, surgeons, physiotherapists, and genetic counselors. Continuous research, public awareness, and accessibility of factor replacement therapy are vital for reducing complications and improving the quality of life of hemophilia patients. With the advancement of genetic medicine and personalized therapy, there is real hope that future generations will be able to live free from the severe consequences of this hereditary disease.

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