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Hemophilia: Types, causes, diagnoses, and therapy

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Abstract
Hemophilia (hereditary bleeding) is the result of a genetic mutation in the X chromosome, a deficiency in platelet protein, or one of the clotting factors in it is defective, as the deficiency of factor VIII represents 85% and factor IX represents 15%, the causes of it may be either genetic or the result of a mutation in the chromosome, whose symptoms are internal or external bleeding. Symptoms of hemophilia include severe bleeding, bruising, or bleeding after surgery, and they vary in their severity to mild, moderate and severe, and from their types according to the severity of the injury, to hemophilia A, which is the most common, and hemophilia, which occurs as a result of a deficiency of factor IX B, and acquired hemophilia, which begins after puberty and the cause of which is unknown. They are diagnosed through family medical history, clinical examination, and blood analysis to measure levels of clotting factors such as factor measurement and genetic testing. Among the complications of the disease are bleeding in the brain, damage to the guillotines, and deep internal bleeding, and advances in the treatment of hemophilia were noticeable at the beginning of the third millennium, there are effective treatments that can help control symptoms and prevent complications, including preventive therapy, alternative therapy, or fibrin patches.

Keywords: Hemophilia, X chromosome, GPIB, factor VIII

Introduction

Hemophilia: Also known as hemophilia or hereditary bleeding [1], is a rare genetic bleeding disorder in which one of the clotting factors in the blood is defective, or the so-called Bernard Sullivan syndrome is a rare hereditary recessive bleeding disease characterized by a deficiency in platelet protein Ib (GpIb), proteins that work together to stop bleeding and vary in severity according to clotting factor [2], which is the receptor of von Wille brand factor, which plays a role in clotting [3, 4].

Symptoms of hemophilia include internal or external bleeding. The genes responsible for producing these factors are transferred to the X chromosome, where males carrying the XY gene are affected, and if a male inherits an X chromosome carrying the defective gene, he will develop hemophilia because he does not have another X chromosome to replace the defective gene, while females carrying the XX chromosome , they can be infected if they inherit two copies of the defective gene (one from the father and the other from the mother who suffers from hemophilia, this is rare and sometimes they are carriers of the disease if they inherit one copy of the defective gene and a normal copy, in which case they do not show symptoms of hemophilia or are only mild symptoms. But sometimes due to random disruption of one of the XX chromosomes, it can be severely affected, especially with menstrual bleeding (HA). Or factor VIII (hemophilia A) deficiency may account for 85% of cases, while factor IX deficiency (hemophilia B) may account for 15% of cases [6, 5].

Methods for screening for viral pathogens that contaminate blood and blood products have improved despite these advances, but bleeding and adherence to treatment regimens is still a problem for many patients. These are inherited bleeding disorders caused by partial deficiency. Hemophilia is a lifelong condition, but there are effective treatments to control, prevent and prevent its symptoms. Treatment of hemophilia should be provided by a specialized team at the Hemophilia Treatment Center, and treatment is specific to each case. If a person has hemophilia, their clotting factor levels are lower than normal [7].

Causes of hemophilia

- In most cases, hemophilia is hereditary, in which defective genes are passed from parents to children.
- Hemophilia is a mutation or change in one of the genes responsible for producing the clotting factors needed to form a blood clot [8]
- (Symptoms of hemophilia according to their severity

Hemophilia Light	▪ Easy, long-lasting bleeding after minor cuts may cause bruising ▪ You may experience prolonged bleeding after tooth extraction, surgery, or medical procedures that cause a skin injury or a serious accident. ▪ Females with hemophilia may experience heavy bleeding during menstruation or childbirth ▪ Otherwise, you may experience bleeding problems (recurrent perineitis) that sometimes require medical attention
Moderate hemophilia	▪ After minor wound bleeding, they can bruise easily and last a long time ▪ Bleeding problems after minor injuries, such as sports injuries ▪ Bleeding problems after surgery, medical or dental procedures that cut the skin, severe injuries, or accidents. ▪ Females are more likely to bleed due to heavy menstruation ▪ A person may sometimes experience bleeding without knowing the cause
Severe hemophilia	▪ You are likely to bruise easily and bleed for a long time after severe injuries ▪ Bleeding often occurs in joints, muscles and soft tissues ("spontaneous bleeding"), as well as after surgery, or wounds that cause deep cuts and injuries to the skin including minor bumps or injuries. ▪ Females at risk of heavy menstrual bleeding (heavy menstrual cycles) and during childbirth

How is hemophilia diagnosed?

Hemophilia is usually diagnosed by

- **Medical and family history:** genetic factors in the blood and measurement of the worker's activity level. With a factor 8 activity test, hemophilia is diagnosed by measuring the worker's activity level. These tests can be facilitated through hemophilia testing centers
- **Clinical examination:** to look for signs of bleeding and bruising.
- **Blood tests:** to measure the levels of different clotting factors and evaluate the blood's ability to clot. Two types of laboratory tests can be performed on hemophilia carriers: factor measurement and genetic testing [9].

Categories of Hemophilia

Hemophilia is divided by the level of clotting factor

Hemophilia A: or classical, which is the most common type. This type of hemophilia affects about 1 in 5,000 people. It is an X-linked disease caused by a deficiency in coagulation factor 8 [10].

Hemophilia B: caused by a deficiency in clotting factor 9) is the least common type, affecting 1/3000 people, and types A and B are the most common in the Arab world. [11]. It is a disorder that easily causes bruising and bleeding due to the inheritance of the mutation in the factor IX gene, leading to a deficiency of factor IX and is less common than factor VIII deficiency (hemophilia A) [12].

Hemophilia was identified B First as a characteristic disease entity in 1952 [13]. It is also known by the same name Christmas sickness [14]. It is named after Steven Christmas, the first patient described as having hemophilia B

Hemophilia c is a rare type of injury caused by a deficiency of coagulation factor 11(XI) [15].

Acquired hemophilia

It usually begins in adulthood and has an unknown cause, and is characterized by abnormal bleeding in the skin, muscles or other tissues due to genetic mutations.

Complications of hemophilia A

There are several complications that occur to a hemophilia patient

- Deep internal bleeding can cause bleeding within the deep muscle to swell the limbs and may cause pressure on the nerves that leads to numbness and pain in organs and tissues and may be life-threatening [16]. World Federation of Hemophilia Guidelines and Third Edition
- **Joint damage:** Frequent bleeding in the joints can lead to chronic arthritis and disability.
- **Bleeding in the brain:** It can cause severe headaches, vomiting, cramping, paralysis, and even death.
- Development of clotting factor inhibitors in some cases, the body can produce antibodies (inhibitors) that prevent the clotting factors given as a treatment from working effectively

Psychotherapy

Advances in the treatment of hemophilia were notable in the first twenty years of the third millennium, but innovation began with the description of plasma fractionation in 1946. Over the past years, life expectancy, quality of life expectancy and quality of life for people with hemophilia have improved dramatically, and their lives are now closer to their unaffected peers [17]. Additionally, there are effective treatments that can help control symptoms and prevent complications

The first thing to start with is first aid for minor wounds. Generally, such as mild pressure of bleeding. You can also use cold compresses for small areas of bleeding under the skin. Poppers can be used to slow light bleeding inside the mouth. Other treatments include

1. Alternative therapy, which involves injecting a hypocoagulation factor into the patient's blood, is given when bleeding episodes occur or on a regular basis to prevent them
2. Preventive treatment to prevent bleeding Coagulation factor is injected regularly
3. Fibrin patches. Especially useful in dental procedures. It can be applied directly to wounds to promote coagulation and healing.
4. Other treatments such as desmopressin, which can help release clotting factor VIII in some mild hemophilia, tranexamic acid medications to prevent blood clots, or you may use a medication imchizumab (Himlibra). This is the latest medication that helps prevent bleeding episodes in people with hemophilia A [18, 19].
5. Newer and more modern therapies, such as gene therapy and drugs that mimic clotting factors, are being developed and used.
6. Physiotherapy: It can relieve symptoms if the joints are damaged by internal bleeding.
7. Surgical treatment in severe cases may require surgical intervention

Ways to live with hemophilia

- It is important to recognize when bleeding occurs early, and to make sure that clotting factors are given before joint damage occurs.
- Treatment of joint bleeding where the patient is provided with clotting factors as soon as possible. Under the supervision of a specialized medical team.

Be careful of bleeding while extracting teeth.

- Before undergoing any surgical procedure, be sure to consult your doctor to take the necessary precautions.
- Prevent children with hemophilia from tampering with equipment and tools at home and elsewhere to prevent injuries and bleeding.
- Reduce injuries caused by playing by protecting your knees and elbows and wearing protective helmets.

Make sure to use seat belts in high chairs, car seats, and strollers to protect against falls.

- After consulting a doctor to determine the appropriate type of exercise and avoid activities that may lead to injury and bleeding (such as football, hockey, and wrestling), children can practice regular physical activity to maintain muscle flexibility and strengthen joints.
- Parents should know how to perform tests for their children and identify the causes of bleeding.
- Ensure that their child is wearing a hemophilia bracelet.
- Avoid taking certain medications, some of which affect the ability of the blood to clot. (such as aspirin),

- Teaching the patient how to recognize and treat the signs of the disease

For treatment: Alternative therapy: involves injecting weak clotting factors into the body to compensate for the deficiency [19, 20].

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