

hemophilia is an inherited interference with blood coagulation and there are numerous hemophilia types, each involving deficiency of a different blood coagulation . the two most common of the disorder are factor VIII deficiency (hemophilia A or Classic hemophilia) which accounts for about 75% of all cases and factor IX deficiency (hemophilia B or Christmas disease).

Pathophysiology Of Hemophilia A

the basic defect of hemophilia A is deficiency of factor VIII anti-hemophilic factor (AHF), which is produced by the liver and is necessary for the formation of thromoplasin in phase 1 of blood coagulation, less AHF the more severe of the disease. Individuals with hemophilia have two or three factors required for coagulation, vascular and platelets therefore they may bleed for longer period but not in a faster rate

- The most common sites of hemorrhage into the joint are knees, elbows, ankles, shoulders, wrists, and hips. Much of strain of everyday movement, these susceptible to knocks and crippling deformities occurs after repeated bleeding over several years

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Inheritance of hemophilia

Hemophilia affects males almost exclusively and is found in all populations, it is passed on by women who are usually mere carriers but have no bleeding problems. About one third of the people with hemophilia have no family history of disorder (.The genes which control factor VIII and factor IX production are both located in the X chromosome, as males have only one X chromosome, the synthesis of factor VIII or factor IX will be deficient, if the relevant gene is defective, a person with hemophilia passes his abnormal X chromosome on to all of his daughters and his Y chromosome which is normal will be passed on to his sons.

Females with one defective factor VIII gene are carriers of this trait and 50% of the male offspring of female carriers have the disease and 50% of their female offspring are carriers, all females' children of males with hemophilia are carriers of the trait

The diagnostic consultation

1-. The clinical history is crucial in the diagnosis of bleeding disorders and a family “tree” should be constructed in any case where there is a history of bleeding in other family members, and those particular aspects to look out for are: bleeding or lack of it after dental extraction, circumcision, tonsillectomy and child birth trauma

2-laboratory finding; persons with factor VIII deficiency have a prolonged PTT, the bleeding time PT is usually normal

Clinical manifestation and severity of hemophilia

hemophilia may be mild or severe, depending on the level of factor VIII coagulant activity, which is usually diagnosed in childhood. :

1- hemorrhage from any trauma, loss of deciduous teeth, circumcision, cuts, epistaxis and injections.

2-bleeding anywhere from or in the body, excessive bruising even from a slight injury, such as a fall, subcutaneous and intramuscularly hemorrhage,

3- hemoarthrosis, bleeding into joint cavities, especially the knee, ankle and elbows,

4-hematomas, pain, swelling and limited motion, and spontaneous hematuria.

A patient with factor VIII level <1% of normal has severe bleeding characterized by frequent spontaneous bleeding episodes involving skin, mucous membrane, joints, muscles and viscera

In contrast, patient with mild factor VIII deficiency 5-40% factor VIII activity, bleed only at time of trauma or surgery, and those with moderate factor VIII deficiency 1-5% factor VIII activity, have intermediate bleeding manifestation. The most crippling aspect of factor VIII deficiency is the tendency to develop recurrent hemoarthrosis, which insite cause joint destruction

Management

1-the main line of management of hemophilia is the prevention of bleeding prompt treatment to limit further tissue damage.

2-Bleeding must be controlled by the demonstration of factor VIII, this may be supplied by fresh whole blood or by a concentration of factor VIII

1 ml of normal plasma contains 1 unit of factor VIII, because the plasma volume is about 45 ml/kg, it is necessary to infuse 45 units/kg of factor VIII to increase its level in the hemophiliac recipient from 1-100% (0-100% units/dc), a dose of, 25-50 units/kg of factor VIII is usually given to raise the recipient level to 50-100% (50-100%units/dc) of normal, because the half-life of factor VIII in the plasma is about 8-12 hr, repeated infusions can be given, as necessary, to maintain the desired level of activity.

Complications of the disease

1-multiple hemarthroses resulting in severe joint deformities and contractures.

2-Hemorrhages into the elbows, knee and ankles cause pain and swelling and limit the movement of the joint , these may be induced by relatively minor trauma but often appear to be spontaneous, repeated hemorrhage may produce degenerative changes ultimately ,affixed unusable joint Life-threatening bleeding of the hemophilic patient is caused by bleeding in vital structures central nervous system (CNS),upper airway or by exsanguinations(external gastrointestinal, or iliopsoas hemorrhage).Life-threatening hemorrhage requires replacement therapy to achieve a level equal to that of normal plasma(100 ui/dl or 100%)

Complications of the therapy

1-infection with blood born viruses HIV, hepatitis C, and hepatitis B).

2-.The appearance of inhibitor to clotting factors is more common in patient with hemophilia A and is rare in patient with hemophilia B and the repeated exposure to exogenously administered factor eight (FVIII) results in the development of inhibitors to FVIII .