Hemophilia

Hemophilia is a group of disorders of hemostasis most often caused by a genetic mutation that leads to a deficiency of specific coagulation proteins or factors. The most common types of hemophilia are hemophilia A (factor VIII deficiency), B (factor IX deficiency or Christmas disease), and C (factor XI). The inheritance pattern of hemophilia A and B is X-linked recessive, which means that males are primarily affected. Hemophilia C is inherited in an autosomal recessive pattern and disproportionately affects Ashkenazi Jews. Symptoms of hemophiila include prolonged bleeding, pain, and hemarthrosis. Interventions include replacement of deficient clotting factors, desmopressin acetate (DDAVP), antifibrinolytic therapy, and analgesics. Genetic counseling and preventing injury are important considerations for patients with this bleeding diathesis.



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Cause/Mechanism

Genetic Mutation

Genetic Mutant

Hemophilia is most often caused by a genetic mutation in the F8 (if hemophilia A) or F9 (if hemophilia B) gene on the X chromosome. This leads to deficiencies of clotting factors, which increases the risk of bleeding. While hemophilia A and B are inherited in an X-linked recessive fashion, hemophilia C has autosomal recessive inheritance and is more prevalent in the Ashkenazi Jewish population.

Assessment

Prolonged Bleeding

Prolonged Bleeding

The lack of normally functioning coagulation factors in patients with hemophilia increases the risk of delayed and excessive bleeding. Minor trauma like small cuts may lead to persistent bleeding. Trauma to the oral mucosa caused by hard toothbrushes or dental extractions may lead to hemorrhage. Head injuries significantly increase the patient's risk of intracranial bleeding.

Pain

Pain-bolt

The deficiency in clotting factors causes uncontrollable bleeding that may contribute to hematoma formation. Hematomas may compress nerves and cause pain, paresthesia, or paralysis in the affected areas.

Hemarthrosis

He-man-roses

The lack of properly functioning clotting factors may lead to hemarthrosis or excessive bleeding into the joints. Commonly affected joints include knees, elbows, shoulders, hips, and ankles. The inflammation caused by bleeding leads to swelling as fluid fills the areas surrounding the joints. As hemarthrosis progresses, the patient may develop joint deformity that may advance to crippling.

Interventions

Clotting Factor Replacement Therapy

Clog Replacement

Clotting factor replacement therapy is initiated to treat acute bleeding episodes in patients with hemophilia. Replacement therapy is also used prophylactically and administered prior to invasive procedures such as surgery or dental care.

DDAVP (Desmopressin Acetate)

Designated Driver Vase-present

Desmopressin acetate (DDAVP) is a synthetic analog of vasopressin used to stimulate the production of factors VIII from endothelial cells, which is helpful for patients with hemophilia A. This mechanism of action decreases bleeding time. The onset of the drug is 30 minutes with a duration of 12 hours. Closely monitor the patient for signs of reaction and repeat doses as necessary.

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Antifibrinolytic Therapy

Ant-tie-fiber-lights

Antifibrinolytic therapy enhances fibrin stability in secondary hemostasis. The drugs are used to stabilize clots in areas of increased fibrinolysis, such as the oral cavity. Examples of antifibrinolytic medications include tranexamic acid (Cyklokapron) and aminocaproic acid. Antifibrinolytic therapy is especially beneficial in patients with epistaxis and menorrhagia.

Analgesics

A-nail-Jay-Z

Analgesics may be administered to help decrease pain and joint discomfort in patients with hemophilia. Ice packs may also help relieve joint discomfort. Examples of appropriate analgesics include acetaminophen and codeine. Do not administer aspirin or other NSAIDs with antiplatelet activity in patients with hemophilia, since the medication further increases their risk of bleeding.

Considerations

Genetic Counseling

Gene Counselor

Since a large percentage of individuals with hemophilia live into adulthood, genetic counseling is recommended to help address reproductive concerns of passing on genes responsible for defective coagulation factors. Carrier screening is encouraged in patients with a family history of hemophilia.

Prevent Injury

Preventing Injury

Teaching the patient measures to prevent injury is critical in individuals with hemophilia. Instruct the patient to prevent trauma and avoid the use of hard-bristled toothbrushes while performing daily oral hygiene. If the patient wants to participate in recreational activity, recommend noncontact sports, such as golf or swimming to prevent trauma. Instruct the patient to wear gloves to prevent cuts or abrasions during household chores. In case of an accident, the patient should wear a Medic Alert tag to inform healthcare professionals about their hemophilic condition.

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