

HEMOPHILIA



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By

Ms. RUPALI RUPASMITA ROUT
ASSISTANT PROFESSOR

INTRODUCTION:

Hemophilia is a genetic disorder that affects blood clotting. It is passed down from parents to their children through their genes. People with hemophilia have a deficiency or absence of certain clotting factors in their blood, which makes them bleed more and for longer periods of time.

TYPES:

There are two main types of hemophilia:

Hemophilia A: This is the more common type, which is caused by a deficiency in clotting factor VIII.

Hemophilia B: This type is less common and is caused by a deficiency in clotting factor IX.

CAUSES/ETIOLOGY:

- Genetic or mutation in gene.
- Hemophilia A is caused by a mutation in the F8 gene, which provides instructions for making clotting factor VIII.
- Hemophilia B is caused by a mutation in the F9 gene, which provides instructions for making clotting factor IX.

PATHOPHYSIOLOGY:

- Hemophilia is a genetic disorder that is caused by a mutation in one of the genes that provides instructions for making the clotting factors needed for blood to clot properly. These genes are responsible for producing proteins that work together to form clots in response to bleeding.
- When an injury or cut occurs, the body typically responds by forming a blood clot to stop the bleeding. The clotting process involves a complex series of chemical reactions in which various clotting factors interact to form a fibrin clot, which helps to seal the damaged blood vessel.

- In people with hemophilia, mutation in the F8 gene, which provides instructions for making clotting factor VIII and mutation in the F9 gene, which provides instructions for making clotting factor IX occurs.
- The deficient clotting factor impairs the formation of the fibrin clot, which makes it more difficult to stop bleeding.
- This can lead to prolonged bleeding after injuries or surgeries, and can even result in spontaneous bleeding into the joints, muscles, and other tissues.

SIGN & SYMPTOMS/CLINICAL MANIFESTATION:

- **Excessive bleeding:** People with hemophilia may experience excessive bleeding from minor cuts, injuries, or surgeries. Bleeding can be prolonged and difficult to stop.
- **Bruising:** People with hemophilia may develop large bruises or hematomas from minor injuries or activities.
- **Joint pain and swelling:** Repeated bleeding into the joints can cause pain, swelling, and stiffness, which can limit mobility and lead to joint damage.
- **Muscle pain and swelling:** Repeated bleeding into the muscles can cause pain, swelling, and tightness, which can limit movement.
- **Nosebleeds:** People with hemophilia may experience frequent and prolonged nosebleeds.
- **Blood in urine or stool:** Hemophilia can cause bleeding in the digestive tract or urinary system, which can result in blood in the stool or urine.
- **Fatigue:** Chronic bleeding can lead to anemia and fatigue.

DIAGNOSIS:

Medical history & family history: The doctor will ask about the patient's family history of bleeding disorders, as hemophilia is an inherited disorder. They will also ask about any episodes of bleeding or bruising and whether they occurred after an injury or without any apparent cause.

Physical examination: The doctor will perform a physical exam to look for signs of bleeding or bruising, particularly in the joints and muscles.

Blood tests: Blood tests can measure the levels of clotting factors in the blood and can help diagnose the type and severity of hemophilia. The most common tests used to diagnose hemophilia are the activated partial thromboplastin time (aPTT) and factor VIII or factor IX activity tests.

Genetic testing: Genetic testing can confirm the diagnosis of hemophilia and identify the specific gene mutation responsible for the disorder.

PHARMACOLOGICAL MANAGEMENT:

The main treatment for hemophilia is replacement therapy, which involves replacing the deficient clotting factor with a clotting factor concentrate. This helps to stop bleeding and prevent complications associated with hemophilia.

There are two main types of clotting factor concentrates used to treat hemophilia:

- **Factor VIII concentrate:** This is used to treat hemophilia A, which is caused by a deficiency in clotting factor VIII.
- **Factor IX concentrate:** This is used to treat hemophilia B, which is caused by a deficiency in clotting factor IX.

Desmopressin: This medication stimulates the release of stored clotting factor VIII and can be used to treat mild to moderate cases of hemophilia A.

Antifibrinolytics: These medications help to prevent the breakdown of clots and can be used to manage bleeding in certain situations.

Immune tolerance therapy: This is a specialized treatment used to manage inhibitors, which are antibodies that can develop in response to clotting factor replacement therapy and reduce its effectiveness.

NON PHARMACOLOGICAL MANAGEMENT/PREVENTION:

- **Early diagnosis:** Early diagnosis of hemophilia can help prevent complications and improve outcomes. If there is a family history of hemophilia or if someone is experiencing symptoms of the condition, they should seek medical attention promptly.
- **Regular check-ups:** Regular check-ups with a hematologist or other medical specialist can help ensure that people with hemophilia receive appropriate care and treatment.
- **Clotting factor replacement therapy:** Regular replacement therapy with clotting factor concentrates can help prevent bleeding episodes and reduce the risk of complications associated with hemophilia.
- **Exercise and physical therapy:** Regular exercise and physical therapy can help maintain joint mobility and prevent joint damage in people with hemophilia.
- **Avoiding certain medications:** Some medications, such as aspirin and nonsteroidal anti-inflammatory drugs (NSAIDs), can increase the risk of bleeding in people with hemophilia and should be avoided unless recommended by a doctor.
- **Wearing protective gear:** People with hemophilia should wear protective gear, such as helmets and knee pads, when participating in sports or other activities that carry a risk of injury.
- **Genetic counselling:** Genetic counselling can help families understand the inheritance pattern of hemophilia and make informed decisions about family planning.

