HEMOPHILIA

DEFINITION

- Hemophilia is a rare disorder in which your blood doesn't clot normally because it lacks sufficient blood-clotting proteins (clotting factors).

TYPES

- There are three main types of hemophilia

- ❑ HEMOPHILIA A
- ❑ HEMOPHILIA B
- ❑ ACQUIRED HEMOPHILIA

Plasma clotting factors

- I – Fibrinogen
- II – Prothrombin
- III – Thromboplastin
- IV – Calcium
- V – Proaccelerin
- VI – Not in use
- VII – Prothrombin conversion accelerator
- VIII – Anti hemophilic globulin
- IX – Plasma thromboplastin component
- X – Stuart factor
- XI – Plasma thromboplastin antecedent
- XII – Hageman factor
- XIII – Fibrin stabilizing factor

Hemophilia A

- Hemophilia A, also called factor VIII (FVIII) deficiency or classic hemophilia, is a genetic disorder caused by missing or defective factor VIII, a clotting protein. Although it is passed down from parents to children

Hemophilia B

- Hemophilia B, also called factor IX (FIX) deficiency or Christmas disease, is a genetic disorder caused by missing or defective factor IX, a clotting protein. Although it is passed down from parents to children

ACQUIRED HEMOPHILIA

- **Acquired hemophilia** is a rare autoimmune disorder characterized by bleeding that occurs in patients with a personal and family history negative for hemorrhages.
- Autoimmune disorders occur when the body's immune system mistakenly attacks healthy cells or tissue.

CAUSES

- Hemophilia is inherited. However, about 30 percent of people with hemophilia have no family history of the disorder. In these people hemophilia is caused by a genetic change (spontaneous mutation).

Clinical manifestations

- **Signs of external bleeding**
- Slow, persistent, prolonged bleeding from minor trauma and small cuts
- Uncontrollable hemorrhage after dental extraction
- Epistaxis
- GI bleeding from ulcers and gastritis
- Hematuria from GU trauma
- Neurologic signs such as pain and paralysis
- Hemarthrosis

- Excessive bleeding following surgery
- Bleeding from cuts that resumes after stopping for a short time
- Blood in the urine

- **Bleeding in the joints**

bleeding in the knees, elbows or other joints is another form of internal bleeding in people who have hemophilia.

- At first the bleeding cause tightness in the joints with no real pain or any visible signs of bleeding
- Then the joint become swollen, hot to touch, and painful to bend.

- Bleeding in the brain

internal bleeding in the brain is a very serious complication of hemophilia. The symptoms are,

1. Long lasting head aches
2. Repeated vomiting
3. Sleeplessness
4. Changes in behaviour
5. Sudden weakness

- 6. slurred speech or other speaking difficulties
- 7. double vision
- 8. convulsions or seizures

DIAGNOSIS

- Prenatal testing:

if a pregnant woman has a history of hemophilia, a hemophilia gene testing can be done during pregnancy.

A sample of placenta is removed from the uterus and tested. The test is known as CVS (Chorionic Villus Sampling) test.

- Blood studies

if the physician suspect the child for hemophilia, a blood test can be determine whether the patient has hemophilia A or B.

❖ A normal platelet count, normal PT, and prolonged aPTT are the characteristics of hemophilia A and B

- Specific test for blood clotting factors can then be performed to meassure factor VIII or factor IX level to confirm the diagnosis.

MANAGEMENT

- **Treatment with replacement therapy:**

The main treatment of hemophilia is called replacement therapy.

- Concentrates of clotting factor VIII and IX are slowly dripped or injected in the vein.

- Clotting factors concentrates can be made from two different ways

1. Plasma derived clotting factors

clotting factor concentrates can be made from the plasma of donated human blood that has been treated to prevent the spread of disease.

2. Recombinant clotting factors:

- The first generation of Recombinant products use animal products in the culture medium and human albumin added as a stabilizer.
- Second generation products used animal derived materials in the culture medium but do not have added albumin, instead of that use sucrose or other non-human derived materials as a stabilizer.

Other managements.....

- **Desmopressin**

- desmopressin (DDAVP) is a man made hormone used to treat people who have mild to moderate hemophilia A.

- it is not used to treat hemophilia B and severe hemophilia A

- it is usually given as injections or nasal sprays.

- **Antifibrinolytic medicines**

- it include trenaxamic acid and aminocaporic acid.

- they are usually given as pills and they help to keep the blood clot from breaking down.

- **Gene therapy**

  - researches are going to correct the faulty genes that causes hemophilia.

- **Analgesics**

  - pain relievers may prescribed to relieve pain.

  - aspirin and NSAID are not using

  - naproxen and ibuprofen are used

- **RICE**

Rest, Ice, Compression and Elevation is recommended by the professionals for joint bleeding.

it also helps to reduce the swelling and tissue damage