

HAEMOPHILIA

For Class– B.Pharmacy 2nd Semester

Subject– Pathophysiology (BP204T)

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Definition



- ▶ **Hemophilia** is a coagulation disorder arising from a genetic defect of the **X chromosome**.
- ▶ Any of several hereditary blood-coagulation disorders in which the blood fails to clot normally because of a deficiency or abnormality of one of the clotting factors.
- ▶ **Hemophilia, a recessive trait associated with the X-chromosome, mostly occurred in males.**



CAUSES OF HEMOPHILIA

- ▶ Lack of formation of prothrombin activator
 1. Deficiency of factor VIII,IX,XI
 - ▶ It is caused due to genetic mutation.
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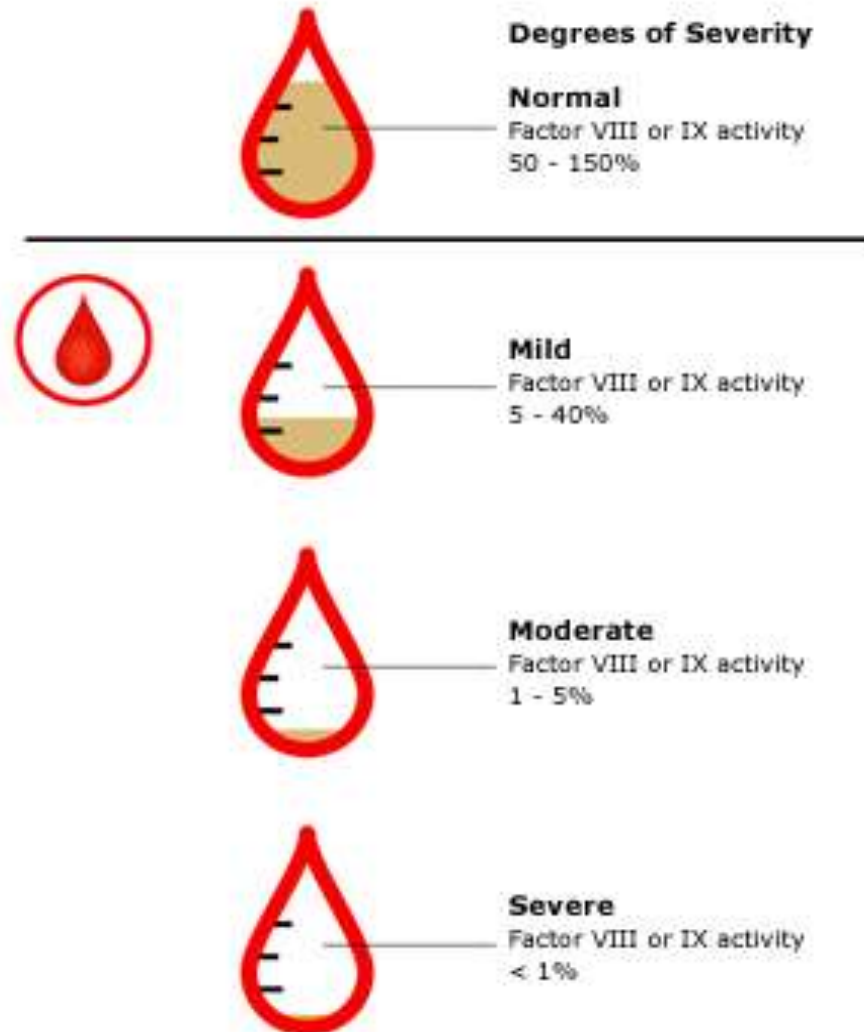
Types of Hemophilia

A	B	C
<p>It is the most common type of hemophilia.</p> <p>(Severe)</p>	<p>It is the second most common type of hemophilia.</p> <p>(Moderate)</p>	<p>It is a mild form of hemophilia.</p> <p>(Mild)</p>
<p>It is also known as factor VIII deficiency or classic hemophilia.</p>	<p>It was originally named "Christmas disease". Caused by factor IX deficiency</p>	<p>Deficiency of factor XI.</p>

INCIDENCE

- ▶ 1 per 5,000 male births
- ▶ 1 per 10,000 population
- ▶ 85 % - F VIII deficiency
- ▶ 10- 15 % - F IX deficiency
- ▶ Haemophilia Ratio
A: B= 7:1

Severity of hemophilia



General Symptoms



Bleeding into joints/muscle
causes pain and swelling



Frequent nose bleeds and
abnormal bleeding after injury or
surgery




Blood found in urine and easy
bruising

Signs and Symptoms

External bleeding may include:

- ▶ Bleeding in the mouth from a cut or bite or from cutting or losing a tooth.
- ▶ Nosebleeds for no obvious reason.
- ▶ Heavy bleeding from a minor cut.
- ▶ Bleeding from a cut that resumes after stopping for a short time.

Internal bleeding may include:


- ▶ Blood in the urine (from bleeding in the kidneys or bladder).
 - ▶ Blood in the stool (from bleeding in the intestines or stomach).
 - ▶ Large bruises (from bleeding into the large muscles of the body).
- 

Signs and Symptoms

Bleeding in the Joints

- ▶ Bleeding in the knees, elbows, or other joints is another common form of internal bleeding in people who have hemophilia.
- ▶ The bleeding causes tightness in the joint with no real pain or any visible signs of bleeding. The joint then becomes swollen, hot to touch, and painful to bend.

Bleeding in the Brain

- ▶ Long-lasting, painful headaches or neck pain or stiffness
 - ▶ Sudden weakness or clumsiness of the arms or legs or problems walking
 - ▶ Double vision
 - ▶ Convulsions or seizures
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Injury Occurs

- 1 Injury to blood vessel results in bleeding.



- 2 Vessel constricts and clotting factors are activated.



Normal

- 3 Along with other substances, clotting factor VIII causes a strong platelet plug to form.



- 4 A stable fibrin clot forms over the platelet plug as a final seal on the injury, and the bleeding stops.



Hemophilia

- 3 Lack of clotting factor VIII causes a weak platelet plug to form.



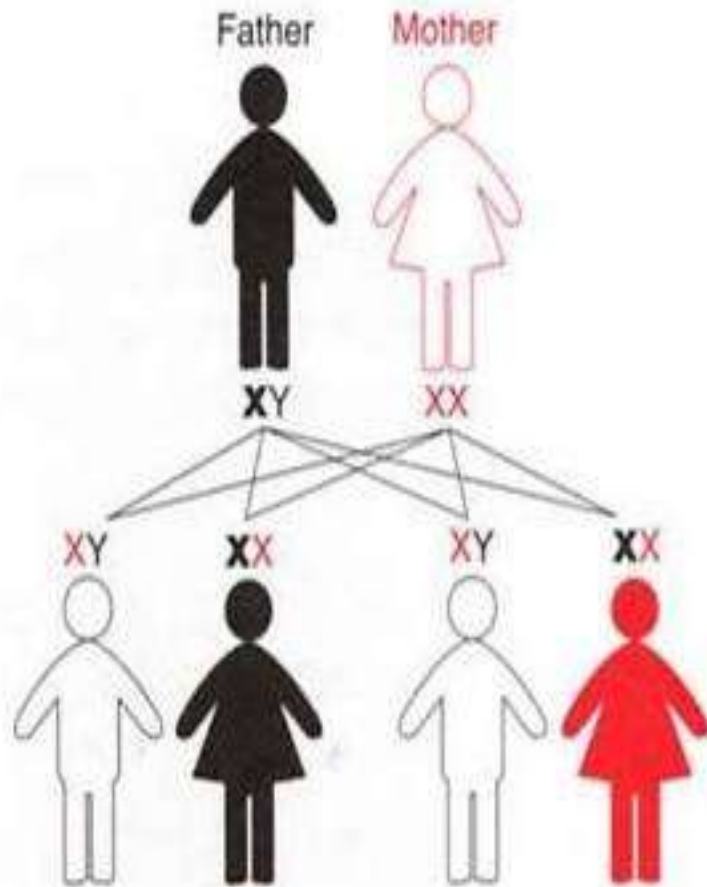
- 4 Incomplete and/or delayed fibrin clot allows bleeding to continue.



INHERITANCE

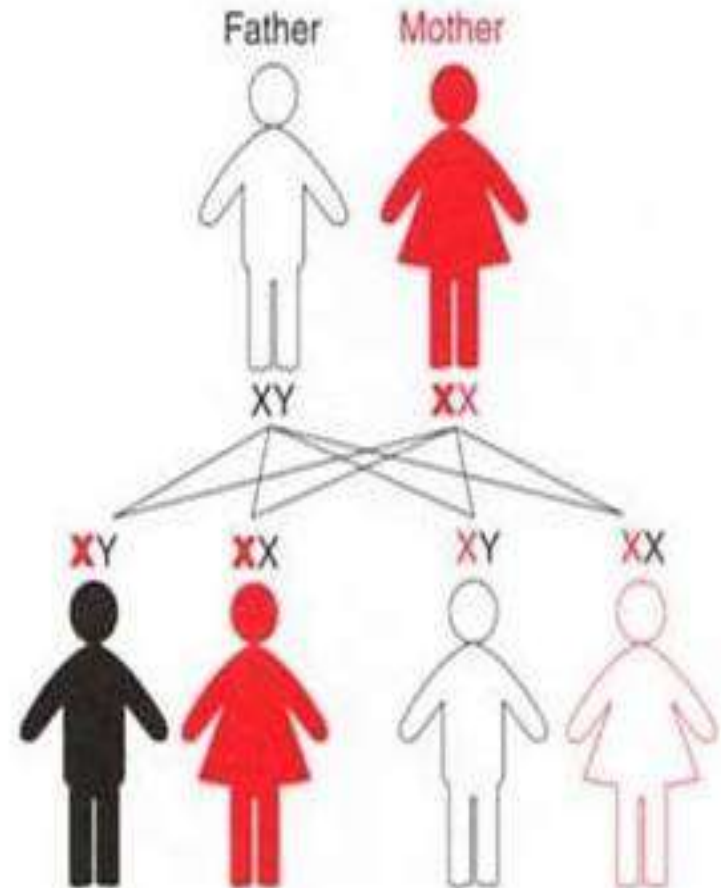
- ▶ Father with Haemophilia:
Daughters are carriers Sons are normal.
- ▶ Mother with haemophilia gene (carrier)
Sons 50:50 normal or affected
Daughters 50:50 normal or carriers.

When the father has haemophilia
and the mother is unaffected



None of the sons will have haemophilia.
All of the daughters will carry the haemophilia gene.

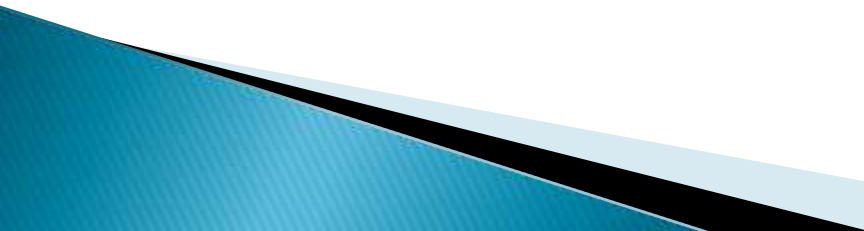
When the mother carries the haemophilia
gene and the father is unaffected



There is a 50% chance at each birth
that a son will have haemophilia.
There is a 50% chance at each birth that
a daughter will carry the haemophilia gene.

Diagnosis

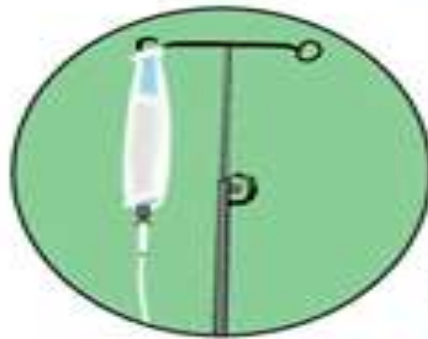
Prenatal testing

- ▶ If a pregnant woman has a history of hemophilia, a hemophilia gene test can be done during pregnancy. A sample of placenta is removed from the uterus and tested. This test is known as a CVS (chorionic villus sampling) test.
 - ▶ **Blood test**
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How to Deal With Hemophilia



Treatment



Treatment

Clotting factor concentrates

- ▶ Plasma-derived clotting factors - prepared from the plasma of donated human blood.
- ▶ Recombinant clotting factors

Desmopressin (DDAVP)(for hemophilia A)

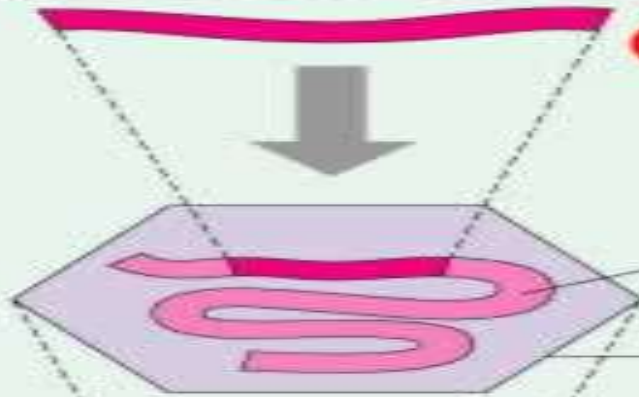
- ▶ This medication is a synthetic hormone which encourages the body to produce more of its own Factor VIII.

RICE (Rest, Ice, Compression, Elevation)

- ▶ RICE is a treatment many health care professionals recommend for joint bleeds. It also reduces swelling and tissue damage when used together with clotting factor concentrates.

**Cloned gene
(normal allele)**

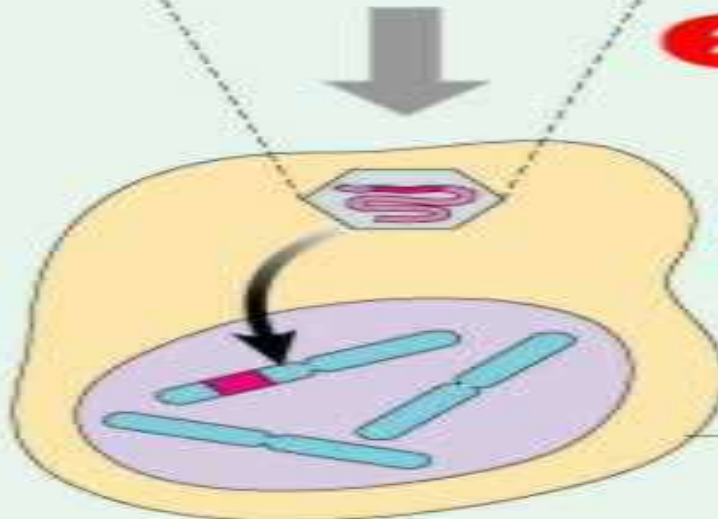
**1 Insert
normal gene
into virus**



**Viral nucleic
acid**

Retrovirus

**2 Infect bone
marrow cell
with virus**



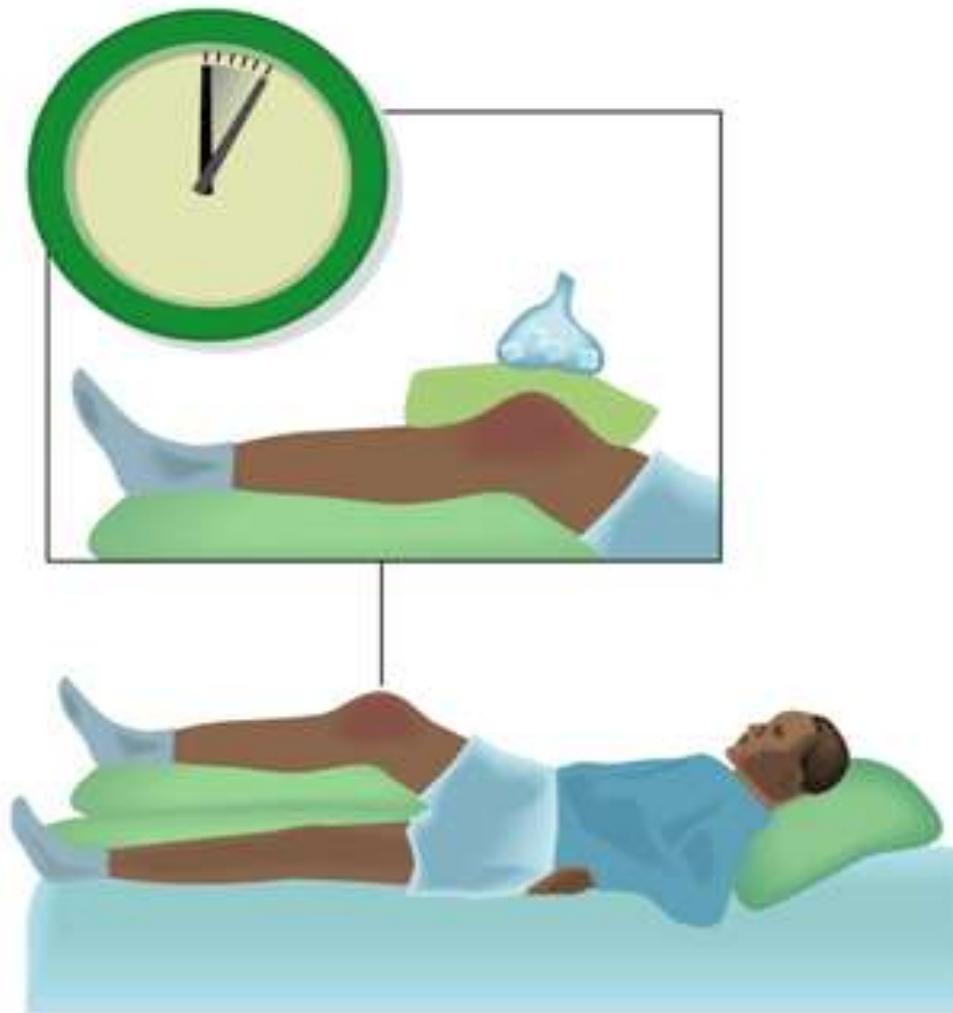
**3 Viral DNA
inserts
into
chromosome**

**Bone marrow
cell from patient**

**4 Inject cells
into patient**



RICE (Rest, Ice, Compression, Elevation)



Treatment

Administering clotting factor concentrates

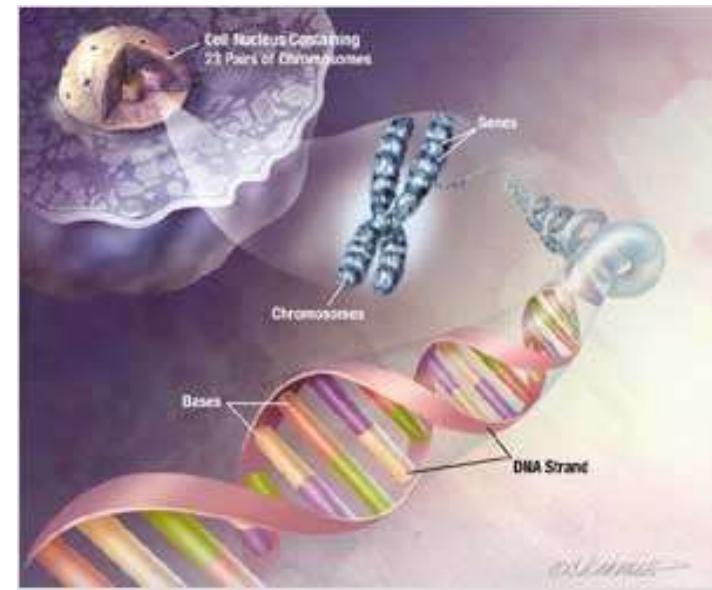
- ▶ The medication is injected into a vein - generally in the back of the hand or at the crook of the elbow.

Storing treatment

- ▶ Factor concentrates should usually be stored in a refrigerator but are stable at room temperature for quite long periods. They should not be frozen as this may damage the vials or syringes.

Gene Therapy

- ▶ Researchers are trying to develop ways to correct the defective gene's that cause hemophilia
- ▶ Such as gene therapy hasn't yet developed to the point that its an accepted treatment
- ▶ Researchers continue to test gene therapies for hemophilia in clinical trails



PREVENTION

Avoid IM injections



Avoid contact sports



PREVENTION

Control Bleeding Episodes

- Local measures: apply direct pressure; elevate or ice compress
- Epistaxis sit up lean forward



PREVENTION

Prevent joint degeneration

- Immobilize joint during acute bleeding
- Progressive exercise
- Avoid prolong immobility

