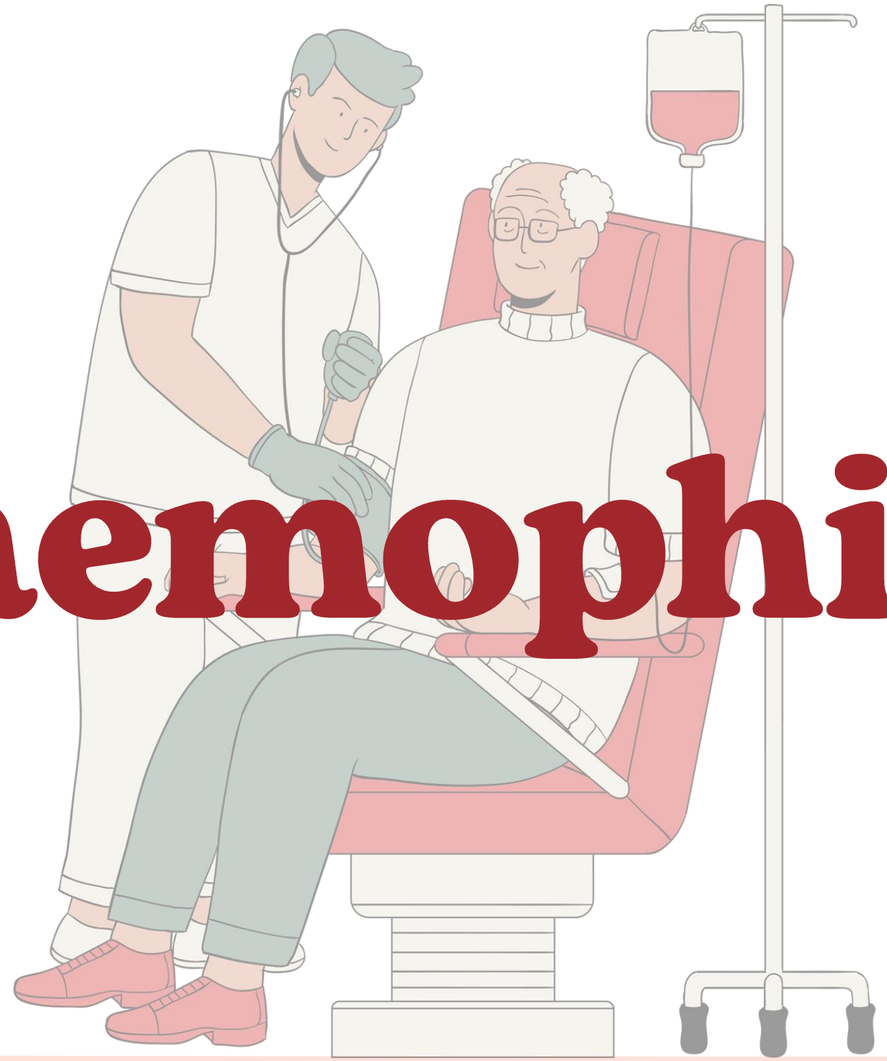
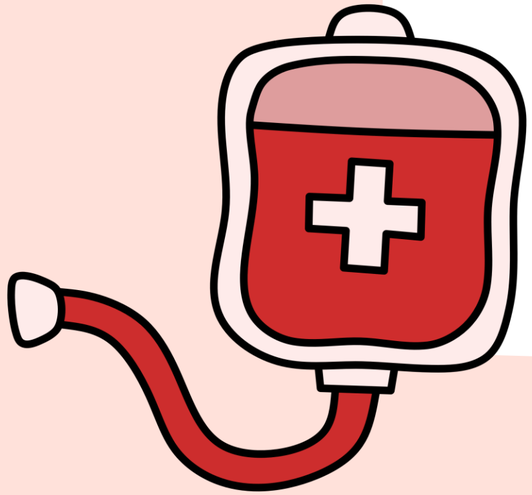
haemophilia





By :
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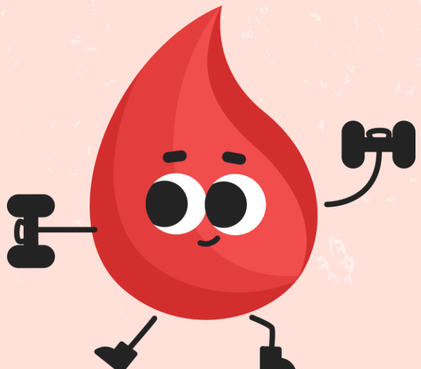
Supervisor :
Dr / Elsaeid El-Sherbiny

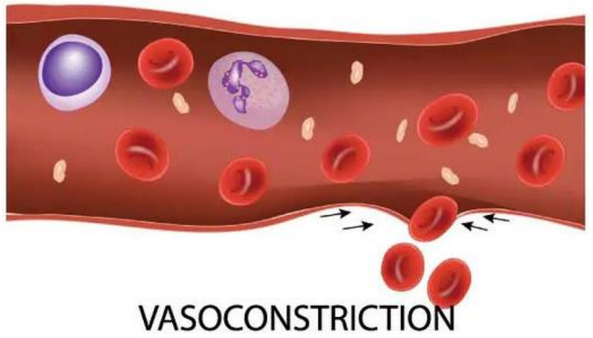
haemophilia

blood

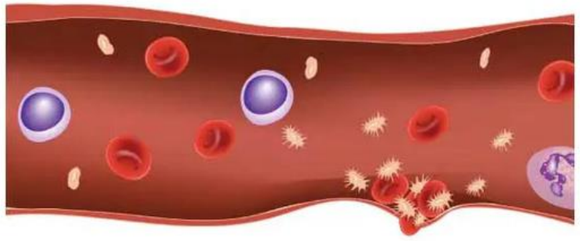
love

due to hemostasis process is impaired

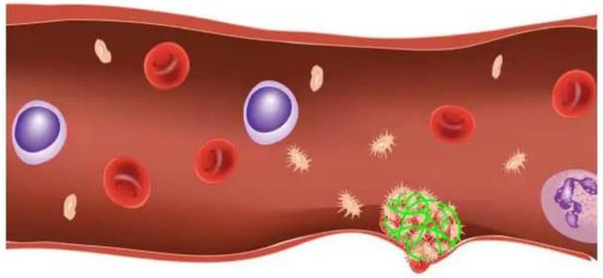




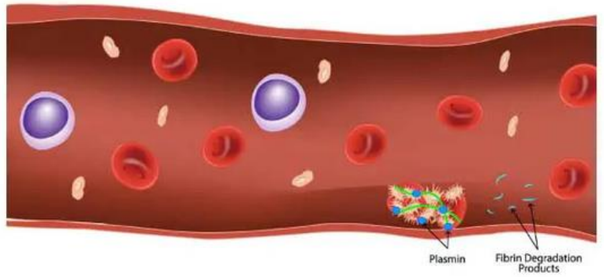
VASOCONSTRICTION



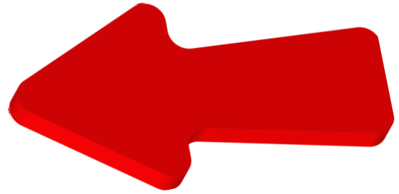
PRIMARY HEMOSTASIS

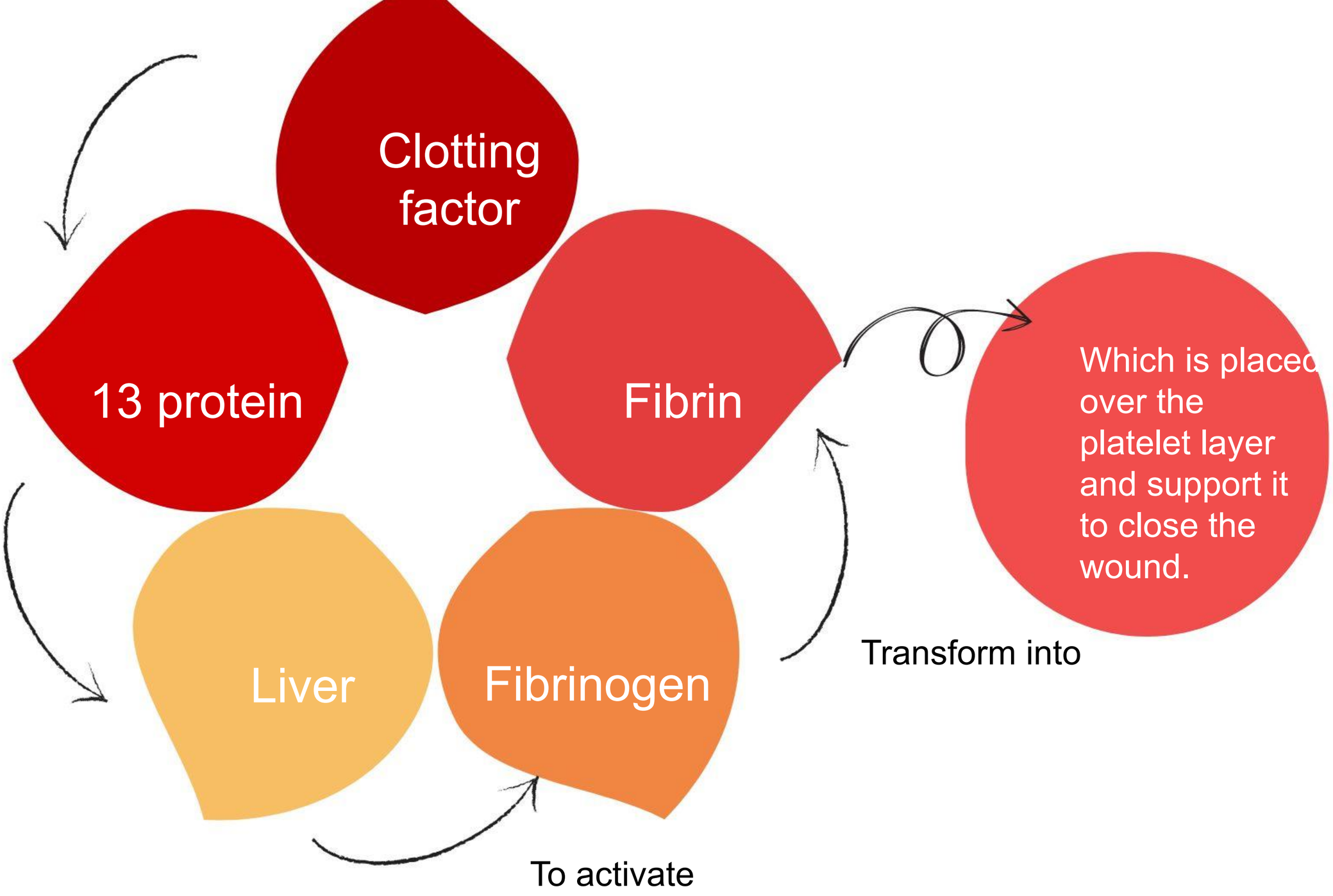


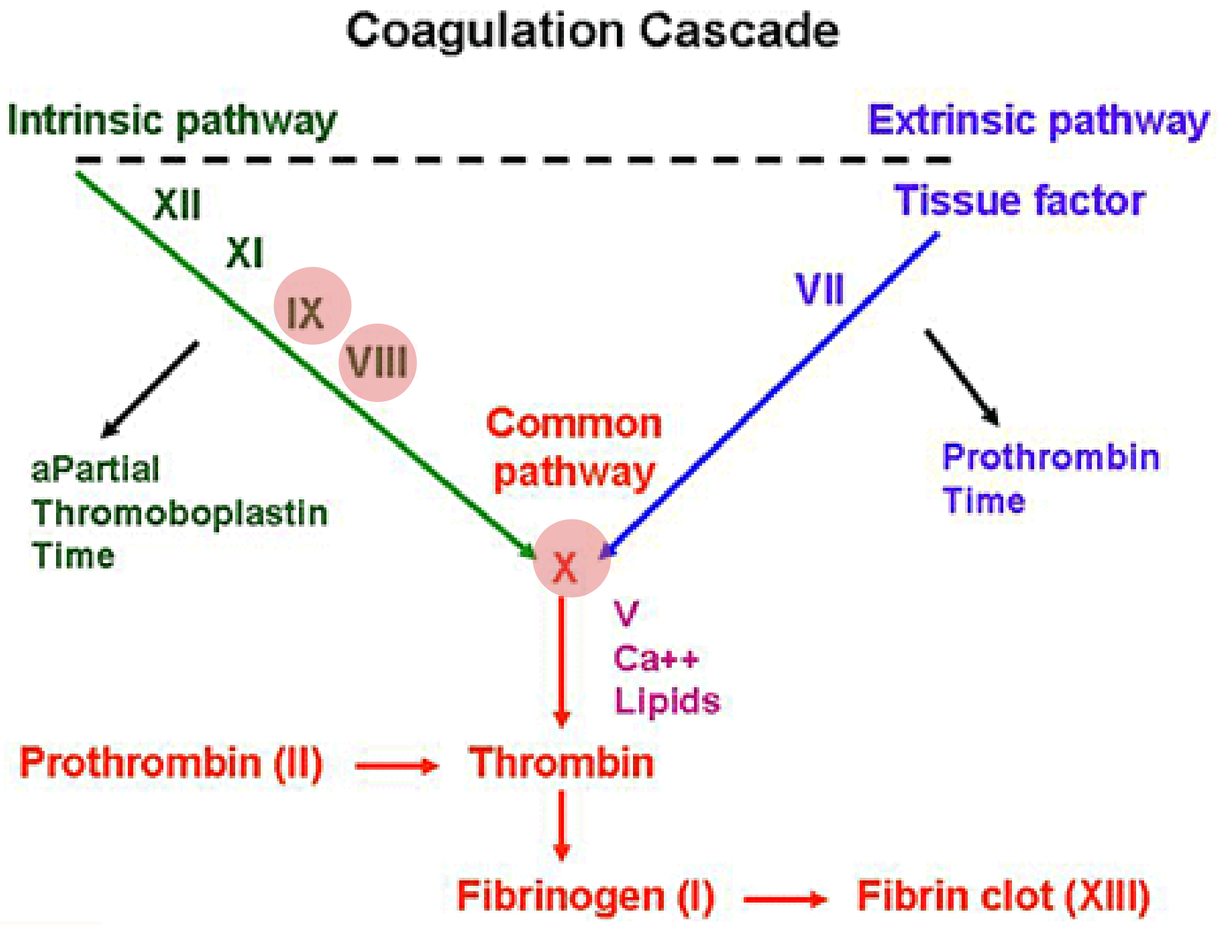
SECONDARY HEMOSTASIS



FIBRINOLYSIS




Clotting factor
13 protein
Fibrin
Which is placed over the platelet layer and support it to close the wound.
Transform into
Liver
Fibrinogen
To activate



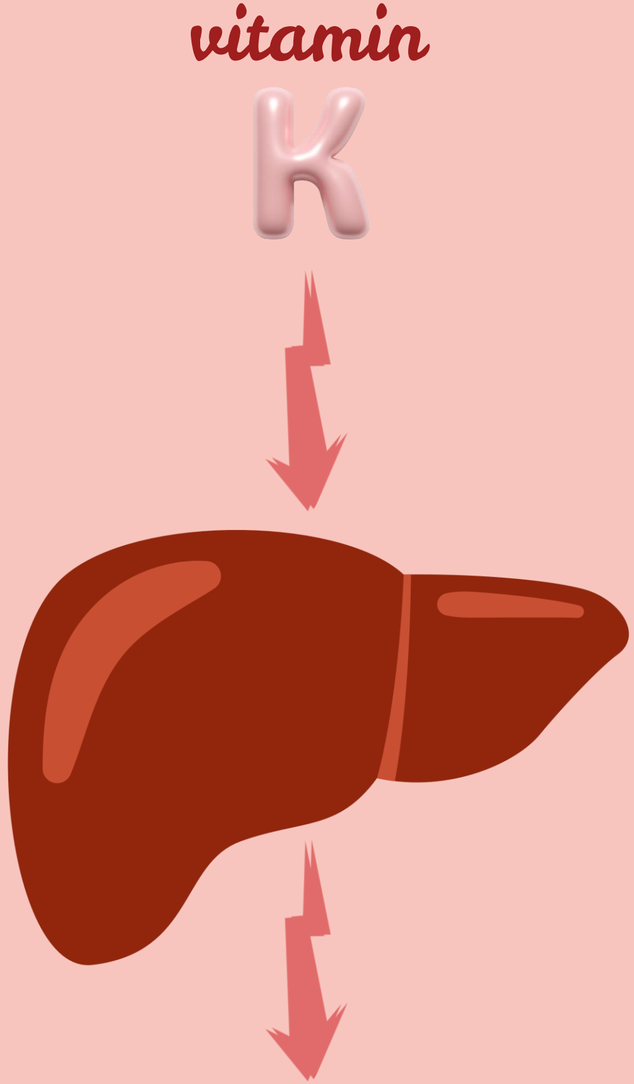
VIII **Haemophilia A**

IX **Haemophilia B**

XI **Haemophilia c**

Haemophilia (acquired)

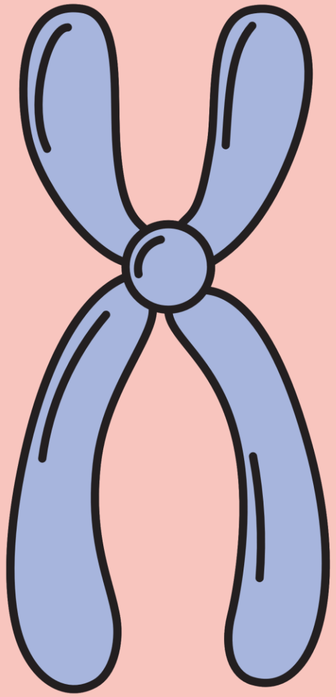
1. It appears suddenly during a person's life, not from birth.
2. Common causes include:
3. Autoimmune diseases: where the immune system attacks clotting factors.
4. Certain medications: such as anticoagulants.
5. Tumors: especially those affecting the bone marrow.
6. Liver diseases: such as cirrhosis,due to : (clotting factors are produced by the liver)
7. Vitamin K deficiency, which is responsible for activating clotting factors.



Haemophilia (Hereditary)

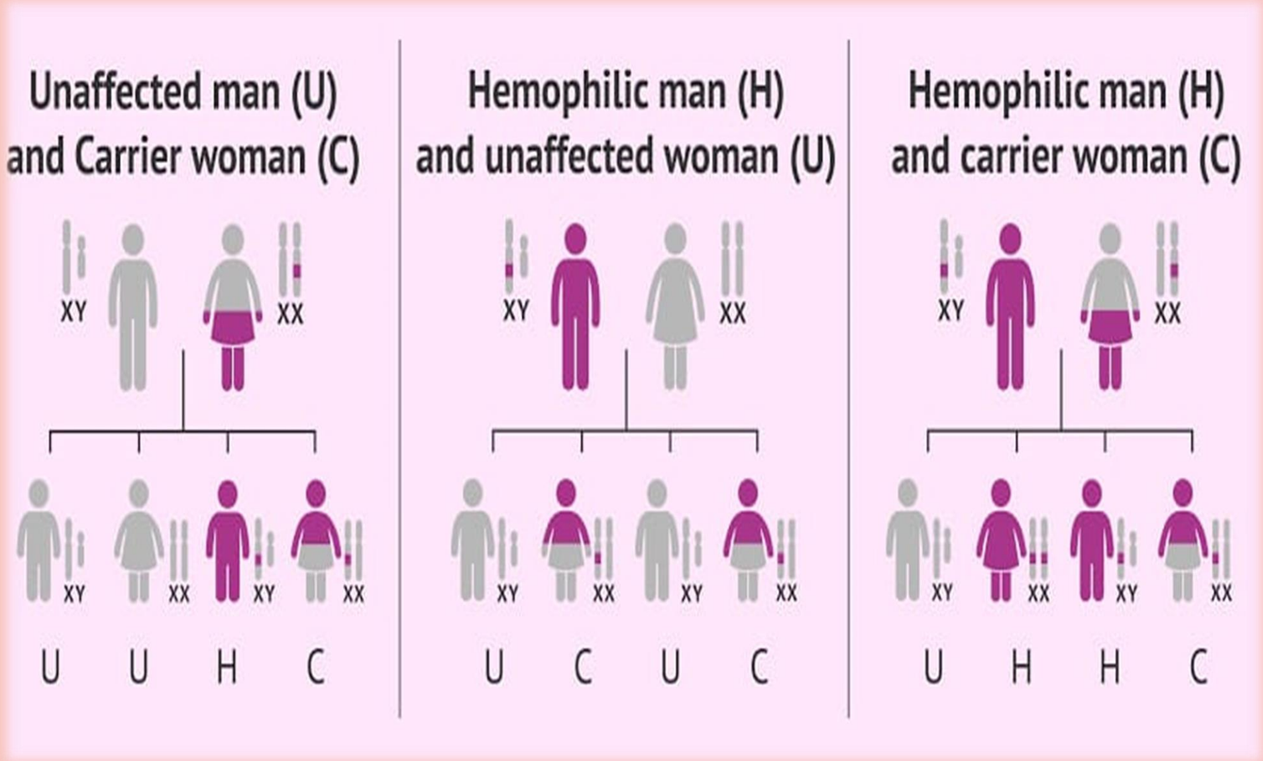
- Hemophilia is genetically transmitted and occurs due to a deficiency or defect in one of the factors.

- F8 & F9 exist on x chromosome ,meaning that males are typically affected because they have only one X chromosome, while females with two X chromosomes are often asymptomatic carriers. The genetic mutation responsible for hemophilia can be inherited from a carrier mother or result from a spontaneous mutation



- men who have a defective copy of the FVIII or IX gene on their X chromosome will pass on a normal Y chromosome to alltheir male children and an abnormal X chromosome to all their female daughters, their sons will be unaffected, and all of their female daughters will be carriers .

- For female carriers, at each birth, there is a 50% chance of transmitting the disorder to their sons and a 50% chance that female daughters



- the classification of hemophilia severity is directly linked to the amount of clotting factors present in the blood.

  - Severe Hemophilia: Characterized by very low levels of clotting factors (less than 1% of normal levels).

(Individuals with severe hemophilia experience spontaneous bleeding episodes, particularly into joints and muscles.)

  - Moderate Hemophilia: Clotting factor levels range from 1% to 5% of normal levels.

(Bleeding typically occurs after minor injuries or trauma, and spontaneous bleeding may occur occasionally.)

  - Mild Hemophilia: Clotting factor levels range from 5% to 30% of normal levels.

(Bleeding usually only occurs after significant trauma, such as surgery or major injury. Some individuals with mild hemophilia may not experience any bleeding problems.)

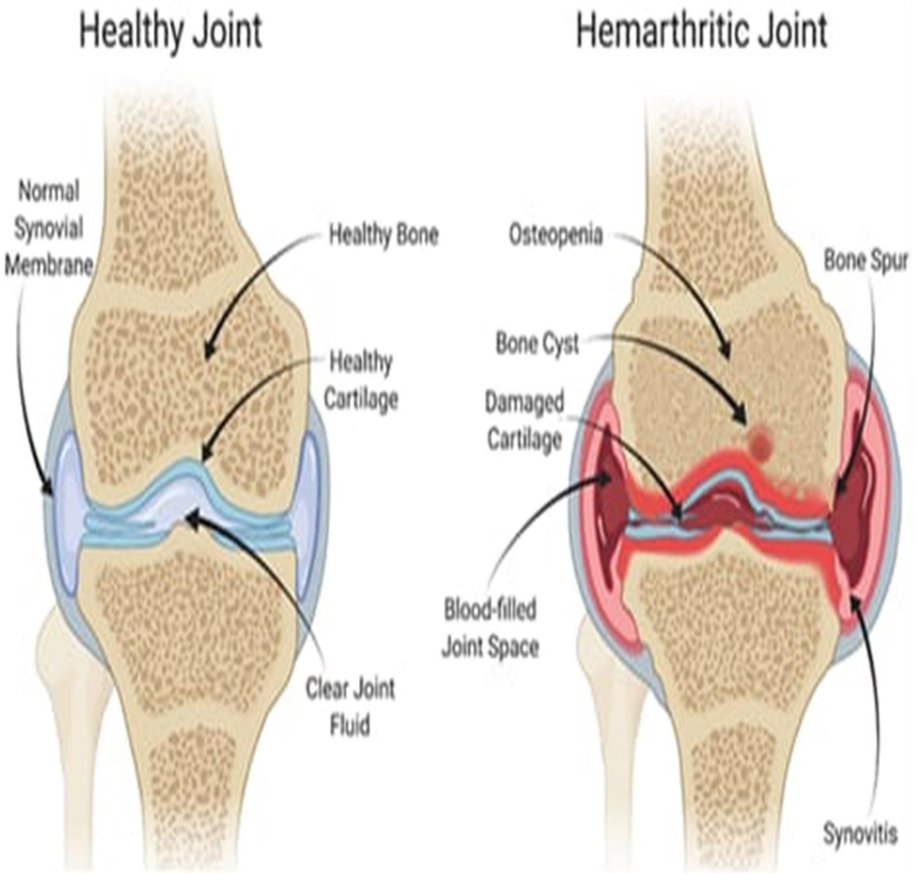
symptoms of hemophilia:

- Excessive bleeding after injuries: Even small wounds can cause severe and prolonged bleeding.
- Large and easy-to-form bruises: Even a slight blow can cause large and painful bruises.
- Frequent nosebleeds: Bleeding may continue for a long time and be difficult to control.
- Bleeding gums: Gums may bleed easily while brushing teeth or after a dental injury.
- Blood in the urine or stool: This may be a sign of internal bleeding.
- Bleeding after surgery or dental procedures.
- Internal bleeding in internal organs.
- Bleeding in the brain (rare but serious).

symptoms of hemophilia:

- Spontaneous bleeding: Bleeding may occur for no apparent reason, especially in the joints and muscles.
  - Bleeding: This is the main factor that leads to joint damage.
  - Inflammation: The body responds to bleeding by releasing chemicals that cause

  (inflammation. This inflammation increases tissue damage and leads to the formation of scar tissue.)
  - Cartilage damage: Cartilage is a flexible tissue that covers the ends of bones in a joint and allows for smooth movement. Bleeding and inflammation lead to cartilage damage and erosion.
  - Bone damage: Repeated bleeding causes damage to the bones under the cartilage, leading to joint weakness.
  - Hematoma formation: Blood may collect inside the joint to form a hard mass called a hematoma. Hemophilia's effect on joints
  - Joint stiffness: Cartilage damage can lead to joint stiffness and difficulty moving.
  - Joint deformity: In advanced cases, joint damage can lead to deformity.
  - Disability: Joint damage can lead to disability and difficulty performing daily activities.



diagnoses

- Family history: Having a family history of hemophilia.
- Physical examination:
  - Bruising: Having large, easy-to-form bruises.
  - Bleeding: Looking for any signs of internal or external bleeding.
- Laboratory tests:
  - A test to determine how much factor IX & Factor VIII is present in the body
  - An activated partial thromboplastin time test to detect how fast the blood clots
  - A Prothrombin time test to detect how quickly the blood clots.
  - A Fibrinogen test to determine the body's ability to form clot

treatments

- Replacement of the missing clotting factor: If a person has a deficiency in one of the clotting factors (such as factor VIII or IX), it is given by injection with formulas containing this factor to compensate for the deficiency and improve the blood's ability to clot.
- Medications that affect blood clotting: Some medications such as "desmopressin" can be used to stimulate the release of some factors from the body to improve clotting.
- Gene therapy: In some cases, gene therapies are being developed that aim to correct the genetic factors that cause hemophilia, but they are still in the experimental stages.

Gene therapy

- It is a modern medical technique that aims to treat diseases by modifying genes within the body's cells.

Gene therapy can include adding a new gene, correcting a defective gene, or even removing or replacing missing or damaged genes

- Techniques used in gene therapy:

  - Viral vectors: Modified viruses are used to deliver the new gene into cells. (Viruses are the most common option as they can effectively carry the genetic material to the target cells.)
  - Non-viral vectors: Methods such as injecting the gene directly into cells or using technologies such as nanoparticles can be used to deliver genes into cells.

How gene therapy for hemophilia is done ??

- Stem cell extraction:

Blood cells, such as hematopoietic stem cells, are taken from the patient.

- Introduction of a healthy gene: The healthy gene that codes for the missing factor (such as factor VIII or factor IX) is introduced into the patient's cells using gene editing techniques such as CRISPR-Cas9 or viral vectors.
- Transplantation of the modified cells: After the genes are modified, the modified stem cells are returned to the patient, where the modified cells travel to the bone marrow and begin to produce the missing clotting factor naturally.

Advantages:

- Long-term treatment: Gene therapy may provide a permanent cure for hemophilia, as the body can begin to produce the missing factor naturally.
- Reduced need for ongoing treatments: Hemophilia patients usually need continuous injections of the missing factors, but gene therapy may significantly reduce this need.

Challenges and risks:

- Efficacy and side effects: Gene therapy is still in the clinical research stages, and more trials are needed to ensure its long-term effectiveness and safety.
- Cost: Gene therapy requires advanced technologies, making it very expensive. Immunological
- risks: There is a possibility of immune reactions to the modified cells or viral vectors, which can lead to complications .

Thank you