BIOLOGY SEM-1

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TOPIC : Haemophilia

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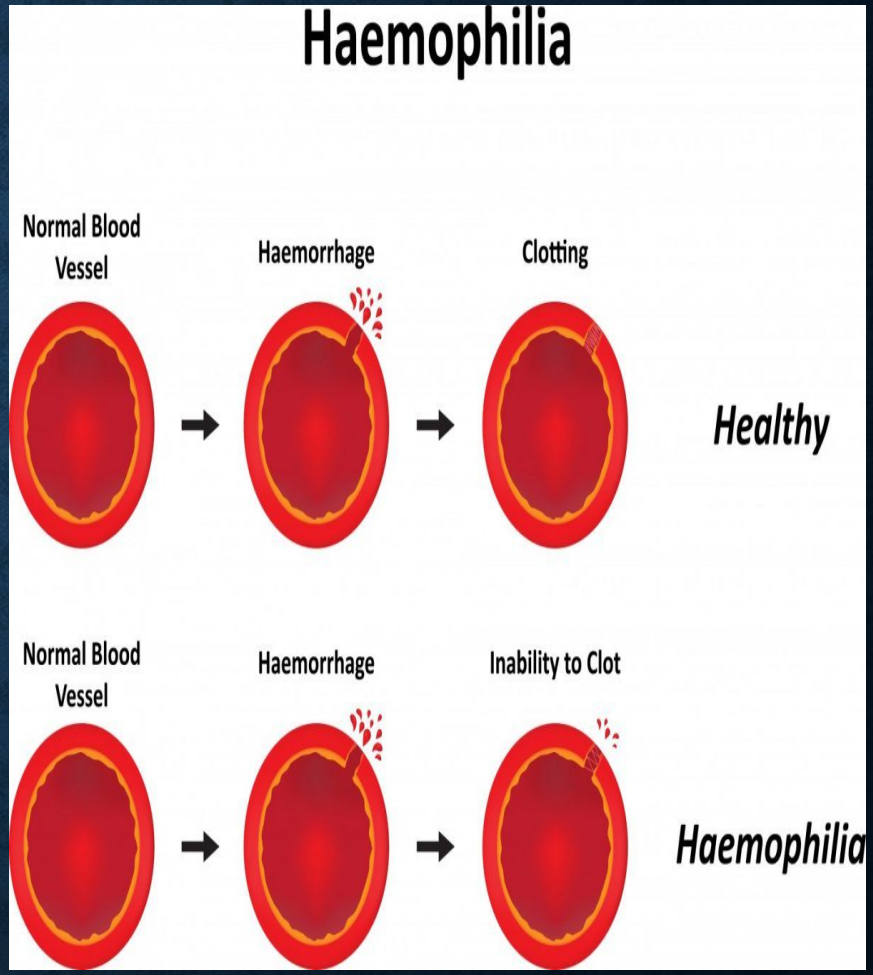
HAEMOPHILIA

Haemophilia is mostly inherited genetic disorder that impairs the body's ability to make blood clots, a process needed to stop bleeding. This results in people bleeding for a longer time after an injury, easy bruising, and an increased risk of bleeding inside joints or the brain.



TYPES OF HAEMOPHILIA

There are several types of haemophilia like :

- Haemophilia A
- Haemophilia B
- Haemophilia C
- *Parahaemophilia*
- *Acquired haemophilia A*
- *Acquired haemophilia B*

- Haemophilia A:

It is a recessive X-linked genetic disorder resulting in a deficiency of functional clotting Factor VIII.

- Haemophilia B:

It is also a recessive X-linked genetic disorder involving a lack of functional clotting Factor IX.

- Haemophilia C:

It is an autosomal genetic disorder involving a lack of functional clotting Factor XI. Haemophilia C is not completely recessive, as heterozygous individuals also show increased bleeding.

- Parahaemophilia:

The type of haemophilia known as *parahaemophilia* is a mild and rare form and is due to a deficiency in factor V. This type can be inherited or acquired.

- *Acquired haemophilia A*:

It A non-genetic form of haemophilia is caused by autoantibodies against factor VIII and so is known as *acquired haemophilia A*.It is a rare but potentially life-threatening bleeding disorder caused by the development of autoantibodies (inhibitors) directed against plasma coagulation factors. Acquired haemophilia can be associated with cancers, autoimmune disorders and following childbirth

- Acquired haemophlia B:

Acquired hemophilia B is caused by the body's production of antibodies against its own factor IX protein. The factor IX antibodies destroy circulating factor IX in the blood causing bleeding symptoms. Acquired hemophilia B is extremely rare; most cases of acquired hemophilia are in those with hemophilia A

- Hemophilia A is most common. It occurs in about one in 5,000 male births; annually about 400 babies are born with hemophilia A.
- Hemophilia B is the second most common type of hemophilia. Hemophilia B occurs in about one in 25,000 male births and affects about 3,300 people in the United States.

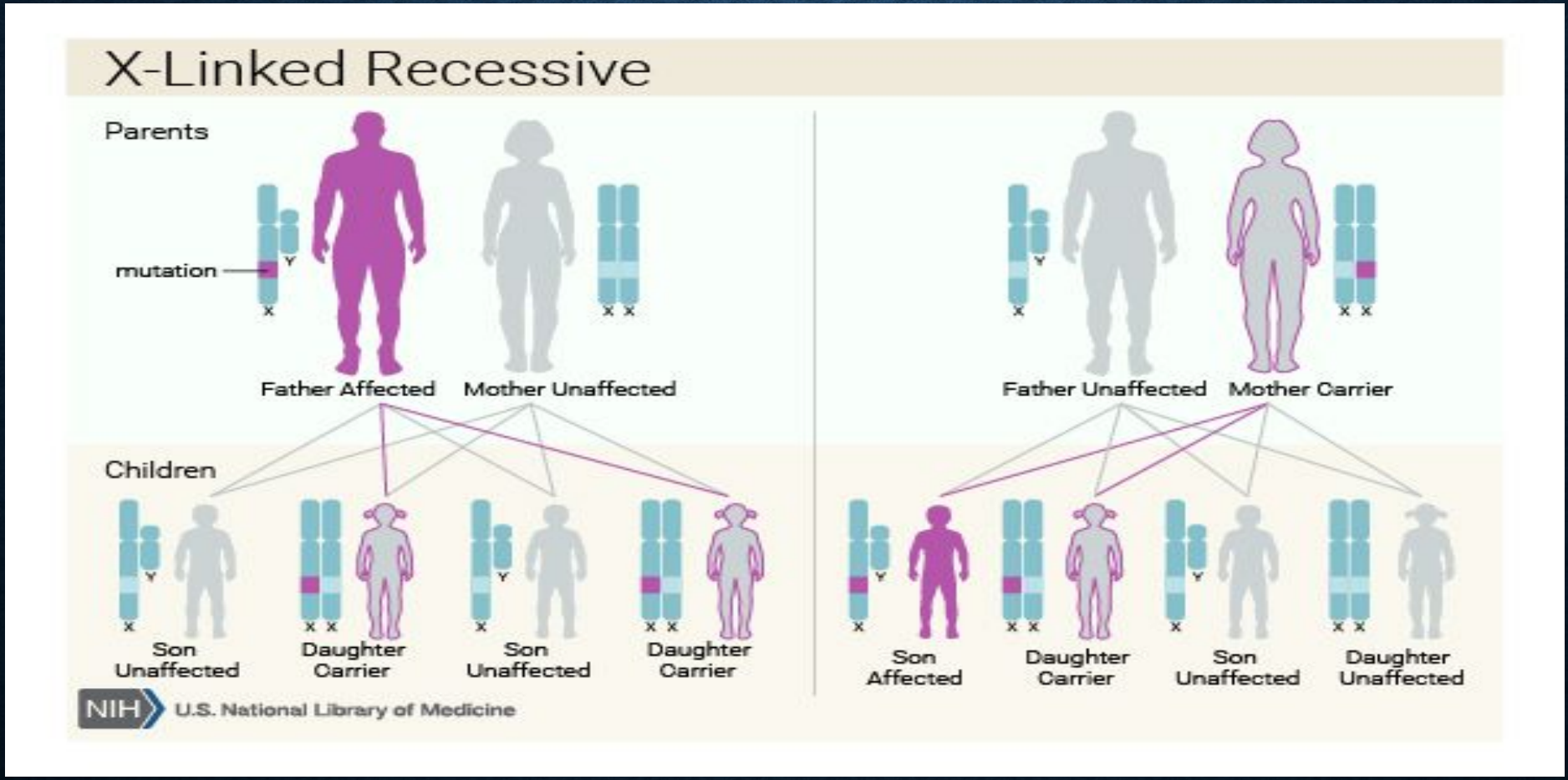
CAUSES

- The genes that regulate the production of factors VIII and IX are found on the X chromosome only. Hemophilia is caused by mutations in either the factor VIII or factor IX genes on the X chromosome.
- If a woman carries the abnormal gene on one of her X chromosomes (females have a pair of X chromosomes), she will not have hemophilia herself, but she will be a carrier of the disorder. That means that she can pass the gene for hemophilia on to her children. There is a 50% chance that any of her sons will inherit the gene and will be born with hemophilia. There is also a 50% chance that any of her daughters will be carriers of the gene without having hemophilia themselves.

- It is very rare for a girl to be born with hemophilia, but it can happen if the father has hemophilia *and* the mother carries the gene for hemophilia. The daughter will then have the abnormal gene on both of her X chromosomes.
- In about 20% of all cases of hemophilia, the disorder is caused by a spontaneous gene mutation. In such cases there is no family history of abnormal bleeding.

GENETICS OF HAEMOPHILIA

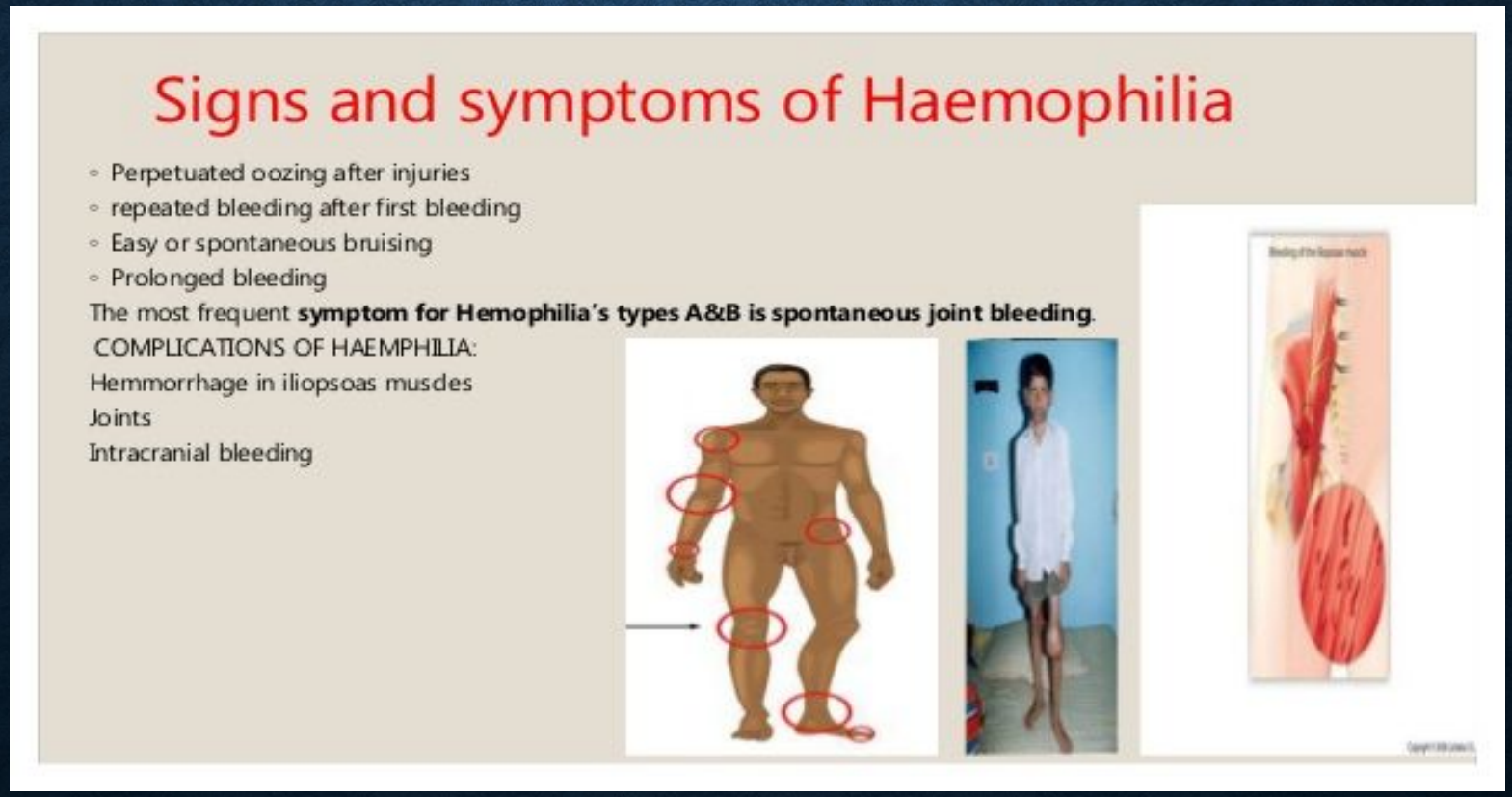
- Haemophilia A and B is characterised by X – linked recessive inheritance



- The genes associated with these conditions are located on the X chromosome, which is one of the two sex chromosomes . In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, it is very rare for females to have hemophilia. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons

SYMPTOMS

- The major symptom is bleeding, either prolonged external bleeding or bruising after minor trauma or for no apparent reason. Symptoms vary depending on whether the patient has the mild, moderate, or severe form of the disorder:



- In severe hemophilia, unprovoked (spontaneous) bleeding episodes occur often.
- In moderate hemophilia, prolonged bleeding tends to occur after a more significant injury.
- In mild hemophilia, a patient might have unusual bleeding, but only after a major injury, surgery, or trauma.
- People with hemophilia may have any type of internal bleeding, but most often it occurs in the muscles and joints, such as the elbows, knees, hips, shoulders and ankles. There may be no pain at first, but if the bleeding continues, the joint may become hot to the touch, swollen, and painful to move.

- Repeated bleeding into the joints and muscles over time may cause permanent damage, such as joint deformity and reduced mobility.
- Bleeding in the brain is a very serious problem for those with severe hemophilia. It may be life-threatening. Get medical help immediately if you have signs of bleeding, such as:
- Changes in behavior, Excessive sleepiness, Headache that will not go away, Neck pain, Double vision, Vomiting, Convulsions or seizures.

DIAGNOSIS

- For people with a family history of hemophilia, it's possible to determine during pregnancy if the fetus is affected by hemophilia. However, the testing poses some risks to the fetus. Discuss the benefits and risks of testing with your doctor.
- In children and adults, a blood test can reveal a clotting-factor deficiency. Depending on the severity of the deficiency, hemophilia symptoms can first arise at various ages.
- Severe cases of hemophilia usually are diagnosed within the first year of life. Mild forms may not be apparent until adulthood. Some people first learn that they have hemophilia after they bleed excessively during a surgical procedure

- People who have 5%-30% of the normal amount of clotting factors in their blood have mild hemophilia.
- People with 1%-5% of the normal level of clotting factors have moderate hemophilia.
- People with less than 1% of the normal clotting factors have severe hemophilia.
- Your doctor may request that other family members have a Factor VIII level drawn to determine if they are affected. In some cases genetic testing may be necessary.

TREATMENT

- Several different types of clotting factors are associated with different varieties of hemophilia. The main treatment for severe hemophilia involves receiving replacement of the specific clotting factor that you need through a tube placed in a vein.
- This replacement therapy can be given to combat a bleeding episode that's in progress. It can also be administered on a regular schedule at home to help prevent bleeding episodes. Some people receive continuous replacement therapy.
- Replacement clotting factor can be made from donated blood. Similar products, called recombinant clotting factors, are manufactured in a laboratory and aren't made from human blood.

Other therapies may include:

- **Desmopressin.** In some forms of mild hemophilia, this hormone can stimulate your body to release more clotting factor. It can be injected slowly into a vein or provided as a nasal spray.
- **Clot-preserving medications.** These medications help prevent clots from breaking down.
- **Fibrin sealants.** These medications can be applied directly to wound sites to promote clotting and healing. Fibrin sealants are especially useful in dental therapy.
- **Physical therapy.** It can ease signs and symptoms if internal bleeding has damaged your joints. If internal bleeding has caused severe damage, you may need surgery.

COMPLICATIONS

- Severe complications are much more common in cases of severe and moderate haemophilia. Complications may arise from the disease itself or from its treatment:
- **Deep internal bleeding**, e.g. deep-muscle bleeding, leading to swelling, numbness or pain of a limb.
- **Joint damage** from haemarthrosis (haemophilic arthropathy), potentially with severe pain, disfigurement, and even destruction of the joint and development of debilitating arthritis.
- **Transfusion transmitted infection** from blood transfusions that are given as treatment.

- **Adverse reactions** to clotting factor treatment, including the development of an immune inhibitor which renders factor replacement less effective.
- **Intracranial haemorrhage** is a serious medical emergency caused by the buildup of pressure inside the skull. It can cause disorientation, nausea, loss of consciousness, brain damage and death.

- **First aid for minor cuts.** Using pressure and a bandage will generally take care of the bleeding. For small areas of bleeding beneath the skin, use an ice pack. Ice pops can be used to slow down minor bleeding in the mouth.
- **Vaccinations.** Although blood products are screened, it's still possible for people who rely on them to contract diseases. If you have hemophilia, consider receiving immunization against hepatitis A and B.

LIVING WITH HAEMOPHILIA

- With treatment, most people with haemophilia can live a normal life.

However, patients should:

- avoid contact sports, such as rugby
- be careful taking other medicines – some can affect your blood's ability to clot, such as aspirin and ibuprofen
- take care of your teeth and gums and have regular check-ups at the dentist
- Looking after your teeth and gums helps you avoid problems such as gum disease, which can cause bleeding. Most non-surgical dental treatment can be done at a general dental surgery.

- Your care team at the hospital can give you advice about surgical dental procedures, such as having a tooth removed, and further information and advice about living with haemophilia.

QUESTIONS

- Gracy[1]: -Explain Replacement therapy for Haemophilia.

  -How replacement clotting factor made for treatment?

- Keerthana[2]: -What is clotting factor and how much a health person should own?

  -Explain the mutation process leads to Haemophilia.

- Karmshil[3]: -Define Parahaemophilia.

  -Which is the most common type of Haemophilia? why?

- Sakhi[4]: -Define inheritance of Haemophilia.

  -Explain the complications did Haemophilic patients face during treatment.

- Aswin[5]: -Define Haemophilia.

  -Explain the proper first aid procedure if a Haemophilic women got a minor cut.

- Teena[6]: -Define Acquired Haemophilia A and B.

  -Haemophilia can only occur via inheritance, express your opinion.

- Nidhi[7]: -Can both male and female can act as carrier for Haemophilia? Explain.

  -List out the precautions to live with Haemophilia.
- Amit[8]: -Which is the most preferable diagnosis method for Haemophilia?Explain.

  -List the vaccinations preferences for the Haemophilic patients and why?
- Vikram[9]: -What is the main reason for the cause of Haemophilia?

  - If a carrier women marries a affected man then show the probability of their children.
- Aishwarya[10]: -What are the types of Hemophilia?

  -Why Haemophilic males are higher that females ?
- Shankar[11]: -Point out the non functioning factor for Haemophilia A, B, C.

  -Haemophilia is curable or not, express your view.
- Ekta[13]: -Types of therapies you know to treat Haemophilia.

  -Surgery for the Haemophilic patient is possible are not ?Express your opinion.

THANK YOU FOR YOUR