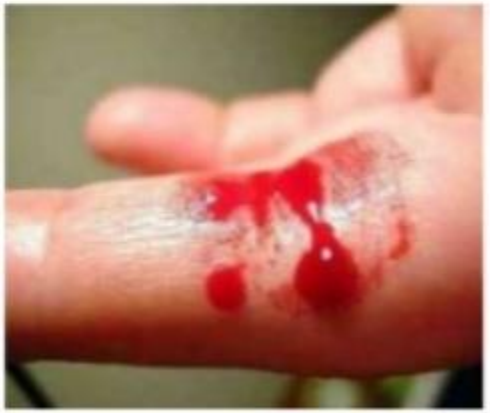
# SUBMITTED TO:
## *Lusine sahakyan*

*SUBMITTED BY:-**NIKITA JAIN***

# HEMOPHILIA

## *Royal Disease*

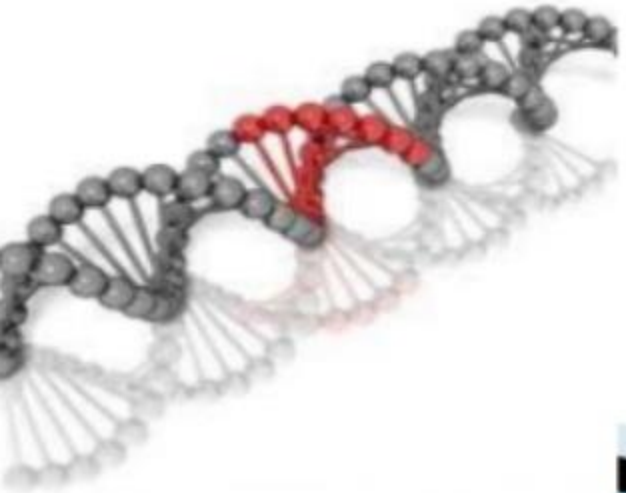


# History

Victoria and Albert with Alfred, Bertie, Alice, Vicky and Helena

# 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.**

# INCIDENCE

- 1 per 5,000 male births
- 1 per 10,000 population

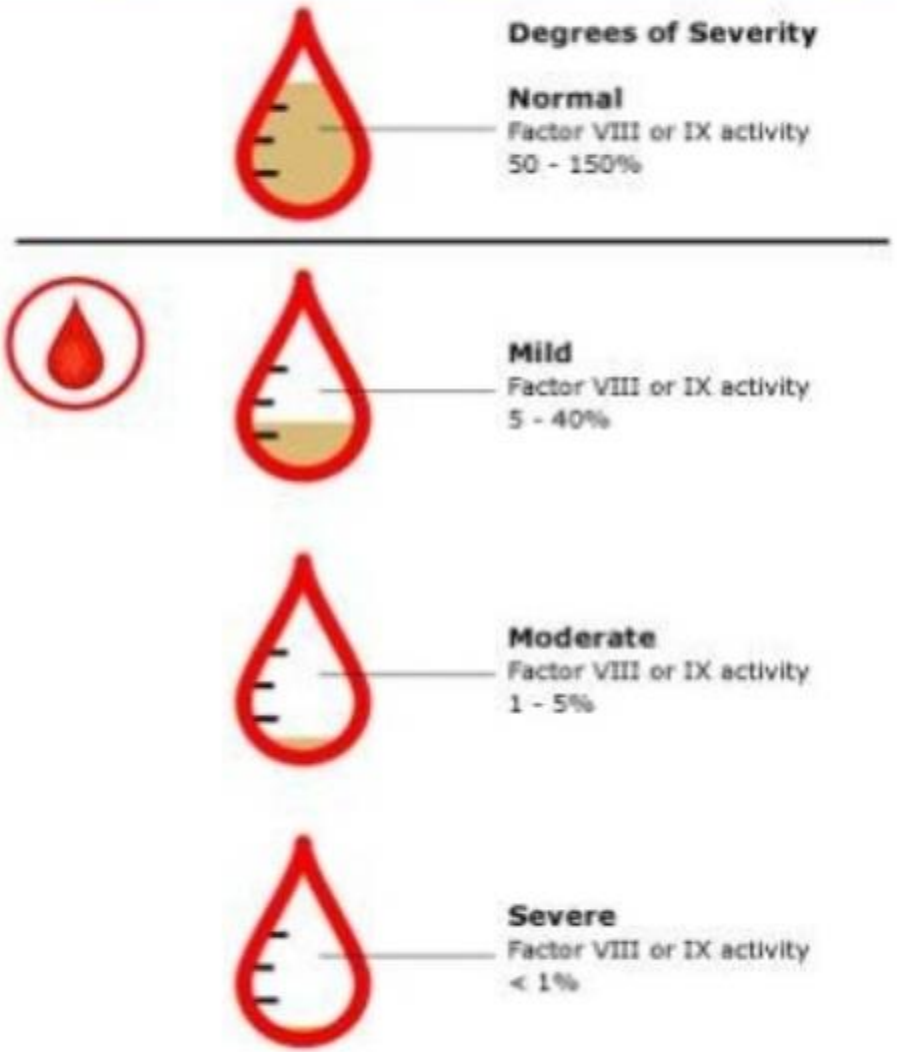
- 85 % - F VIII deficiency
- 10- 15 % - F IX deficiency
- Haemophilia Ratio

  A: B= 7:1

Organizing services for haemophilia in developing countries with few resources is a formidable task. There is wide variation in haemophilia care and management between developing and developed countries. In a country such as India, with one billion people and with a wide diversity in cultural, educational and financial conditions, educating people about such diseases is difficult and will take a long time to have an effect on attitudes

| | | |
|---|---|---|
| India | 78,314 | 1,065,070,607[2] |

# Severity of hemophilia



# Genetics of Hemophilia

- Hemophilia is an inherited X-linked genetic disorder.
- Our bodies have 12 clotting factors that work together in blood clotting and they are present on X chromosome. Having too little of factors VIII,IX or XI cause hemophilia.
- A person with hemophilia will only lack one factor not the three.

# Types of Hemophilia

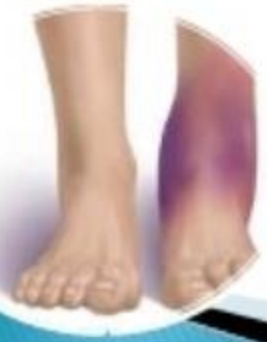
| A | B | C |
|---|---|---|
| It is the most common type of hemophilia. (Severe) | It is the second most common type of hemophilia. (Moderate) | It is a mild form of hemophilia. (Mild) |
| It is also known as factor VIII deficiency or classic hemophilia. | It was originally named "Christmas disease".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

- A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons. It is inherited in zig-zag manner.
- A male is “affected” if he has the abnormal gene on his X-chromosome.
- A female is a “carrier” of hemophilia if she has the abnormal gene on one of her X chromosomes. Even though she doesn’t have the condition, she can pass the gene on to her children.

# 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

# CAUSES OF HEMOPHILIA

- Lack of formation of prothrombin activator

1. Deficiency of factor VIII,IX,XI

- It is caused due to genetic mutation.

# 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.

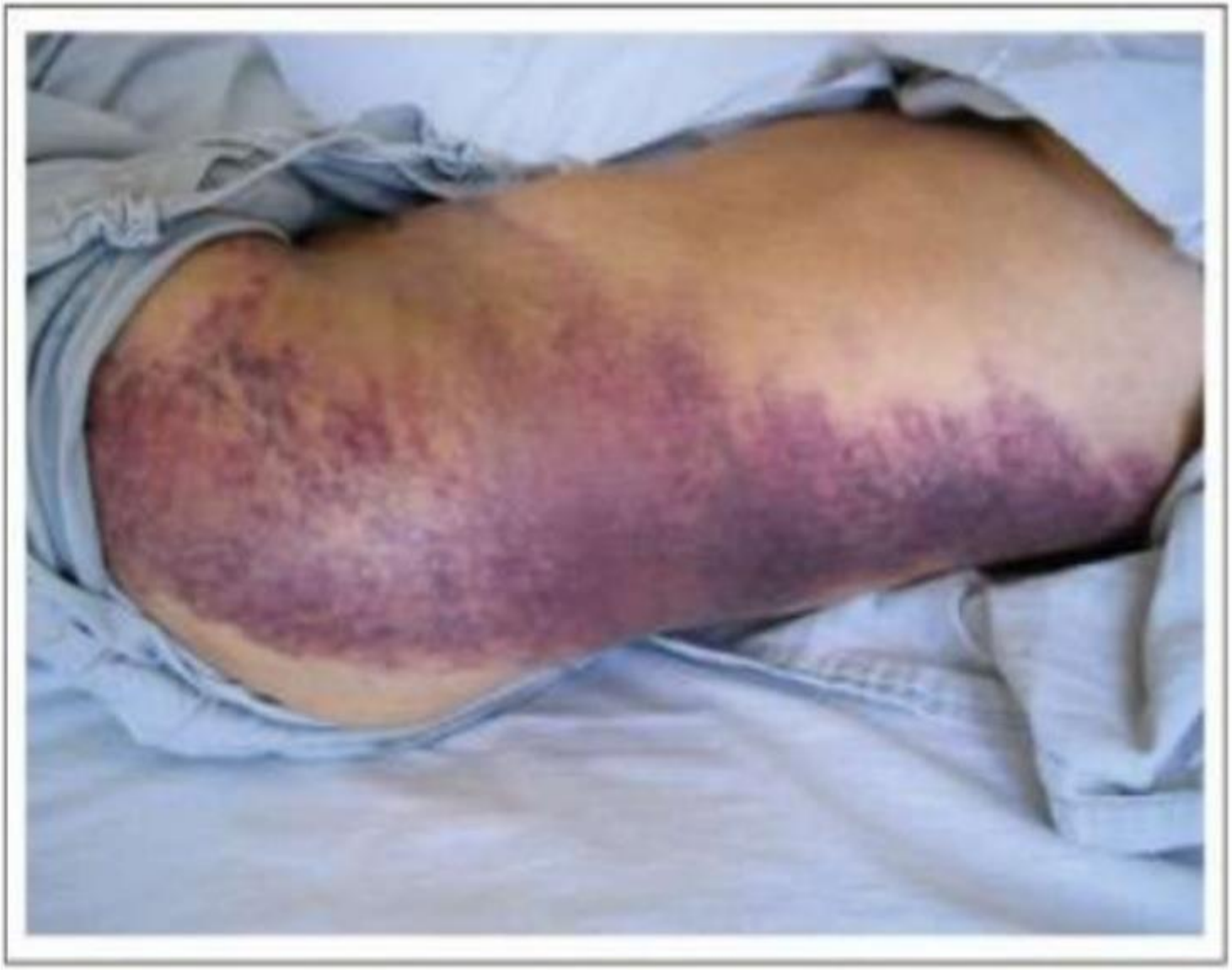


**Figure 2.** Lower limb ecchimosis.

Knowledge of genetics lets us make the following statements about hemophilia:

- Very rarely, a girl is born with hemophilia. This can happen if her father has hemophilia and her mother is a carrier.
- Females who are carriers usually have enough clotting factors from their one normal X chromosome to prevent serious bleeding problems.
- Some males with the disorder are born to mothers who aren't carriers. In these cases, a mutation (random change) occurs in the gene.

# MANAGMENT

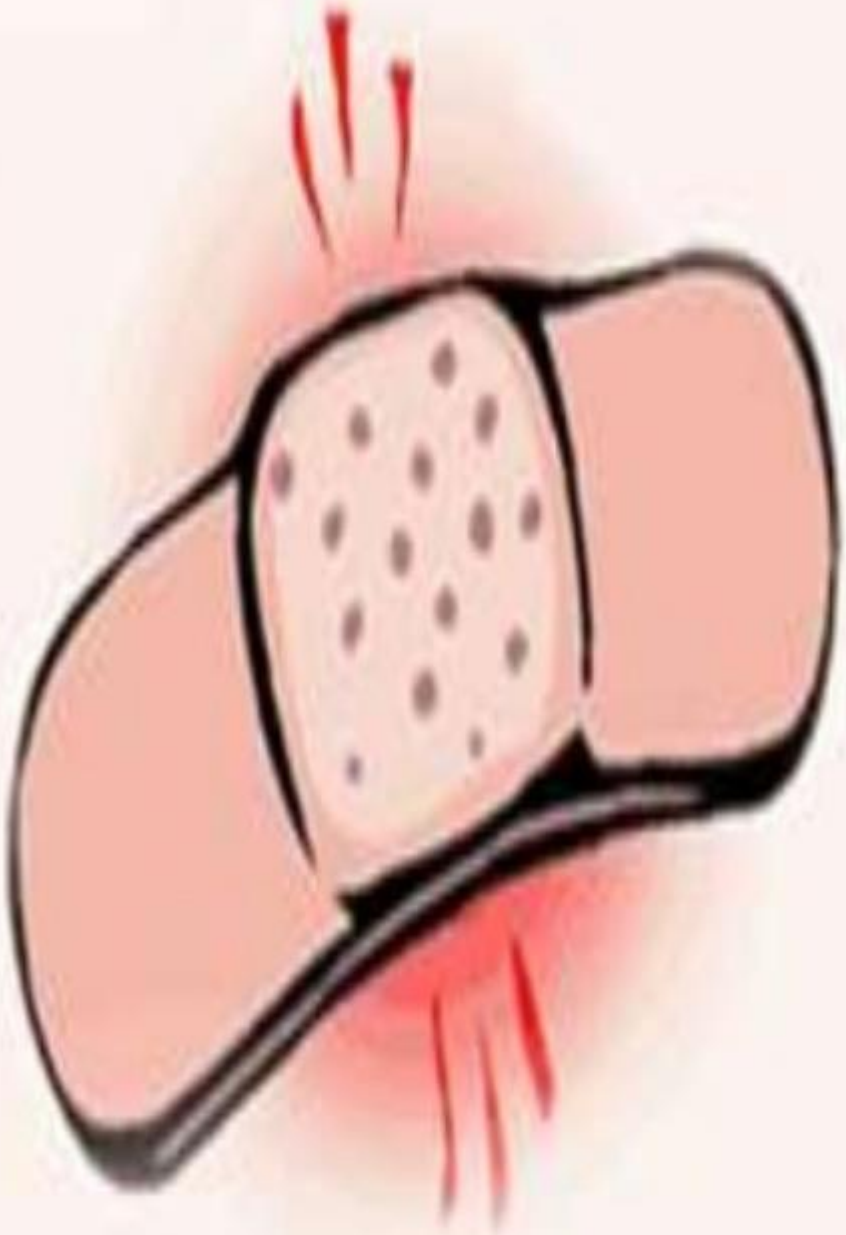
Hemophilia isn't curable , but treatment can prevent crippling deformities and prolong life expectancy. Correct treatment quickly stops bleeding by increasing plasma levels of the deficient clotting factor. This helps to prevent disabling deformities due to repeated bleeding into muscles and joints .

# 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**

# How to Deal With Hemophilia



# Treatment

**Clotting factor concentrates**

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

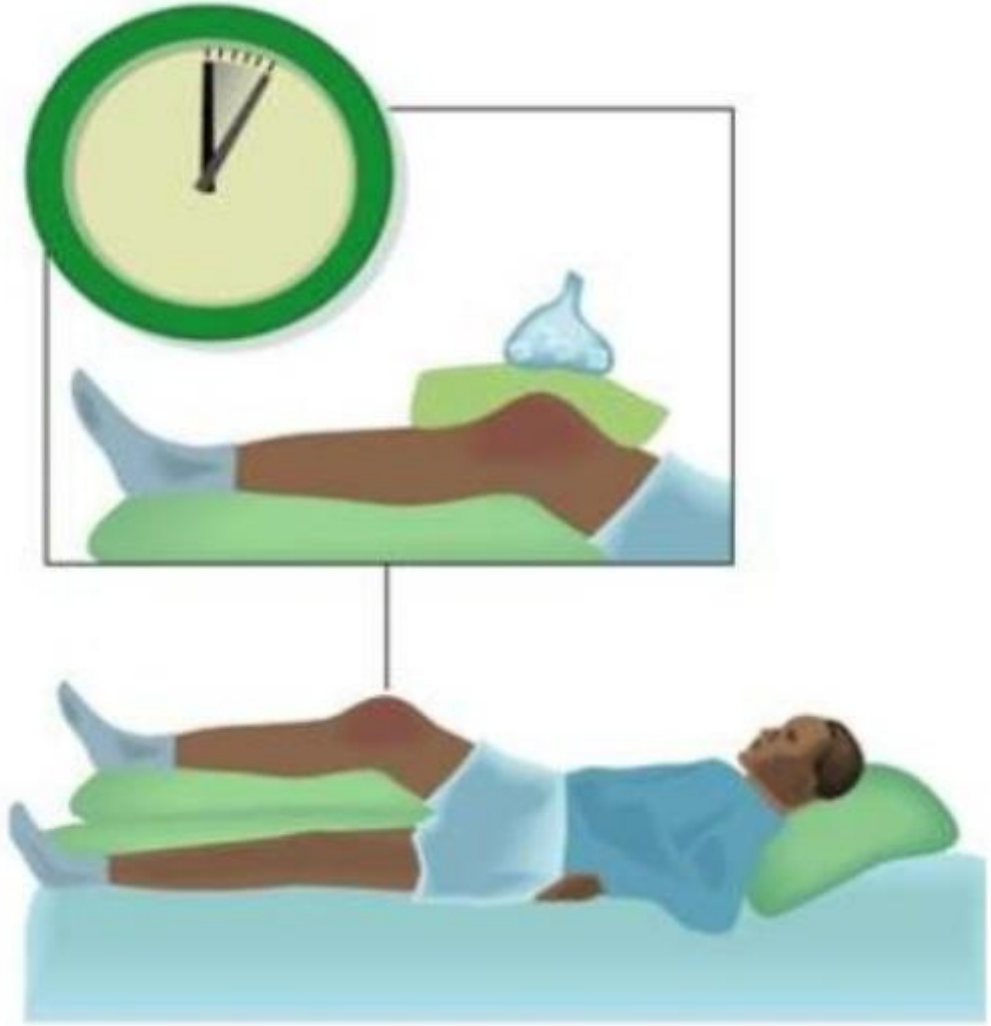
**Desmopressin (DDAVP)(for hemophilia A)**

- 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.

# 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**

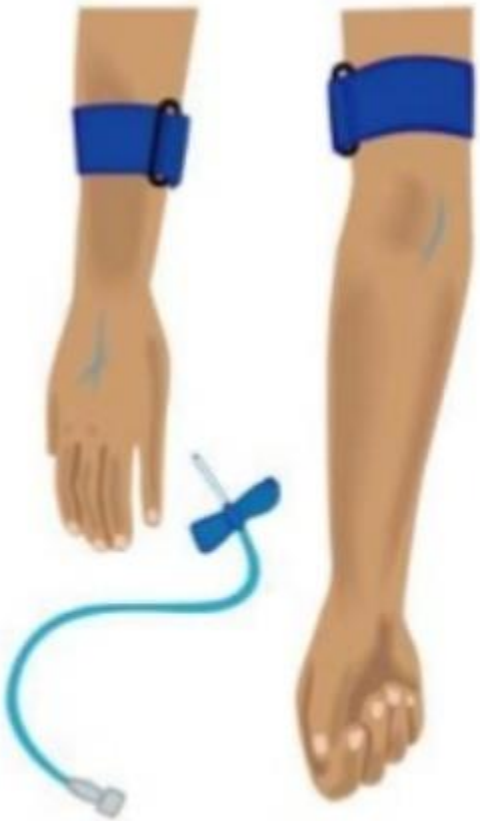
- **F**actor concentrates should usually be stored in a refrigerator but are stable at room temperature for quite long periods. They should not be frozen as this may damage the vials or syringes.

# 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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# Thank You


The End