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News: Hemophilia A

- Recently, the Union Minister of Science & Technology addressing the National Science Day 2024 programme stated that India conducted its first human clinical trial of gene therapy for hemophilia A (FVIII deficiency) at Christian Medical College (CMC) Vellore.

Hemophilia A

- Hemophilia is a group of rare bleeding disorders caused by a congenital deficiency in specific clotting factors. The most prevalent form is Hemophilia A.
- Hemophilia A results from a deficiency in a crucial blood clotting protein known as factor VIII.
- Due to this deficiency, individuals experience prolonged bleeding after injuries, as their blood takes longer to clot than usual.

Causes

- It is primarily inherited (genetic) and follows an X-linked recessive pattern, meaning the gene responsible for factor VIII production is located on the X chromosome.
- Males have one X and one Y chromosome, while females have two X chromosomes.
- If a male inherits an X chromosome with the defective gene from his mother, he will have hemophilia A.
- Females with one defective copy typically do not experience symptoms because the other X chromosome usually provides enough factor VIII.
- However, females can have hemophilia A if they inherit two defective copies, one from each parent (much less common).

Symptoms

- The severity of hemophilia A varies depending on the level of factor VIII activity in the blood.

Common symptoms can include

- Easy bruising and excessive bleeding from minor injuries (cuts, scrapes)

- Bleeding in the joints (especially knees, elbows, and ankles), causing pain, swelling, and stiffness.
- Bleeding after surgery or dental procedures.

Treatment

- The treatment involves replacing the missing blood clotting factor so that the blood can clot properly. This is typically done by injecting treatment products, called clotting factor concentrates, into a person's vein.

The two main types of clotting factor concentrates available are:

- **Plasma-derived Factor Concentrates**: Derived from human plasma, which is the liquid component of blood containing various proteins, including clotting factors.
- **Recombinant Factor Concentrates**: Introduced in 1992, recombinant factor concentrates are genetically engineered using DNA technology and do not rely on human plasma.
- They are free from plasma or albumin, eliminating the risk of transmitting bloodborne viruses.
- However, gene therapy is now gaining prominence.

- In recent trials, they used a new method that involves using a special type of virus called a lentiviral vector to insert a gene that produces FVIII into the patient's own stem cells.
- These modified stem cells then produce FVIII when they develop into specific types of blood cells.

Acquired Hemophilia A

- While Hemophilia A is typically inherited, it can also be acquired later in life as a result of auto-antibodies targeting factor VIII.
- This condition, known as acquired hemophilia A, is rare and differs from the congenital form in its onset and progression.