New Test Kit for Haemophilia A and Von Willebrand Disease

Prelims: Current events of national and international importance

Why in News?

The National Institute of Immunohaematology has indigenously developed a cost-effective point-of-care test kit for the early diagnosis of Haemophilia A, and Von Willebrand Disease.

- The test offers a promising alternative to the current standard of care, which relies on complex and costly diagnostic procedures limited to very few tertiary facilities in India.
- The World Federation for Haemophilia has shown interest in procuring these tests for deployment in countries where the disease is prevalent.
- Haemophilia A and VWD are the two most common inherited bleeding disorders.

Hemophilia A

- It is also known as ***factor VIII deficiency***, is a genetic bleeding disorder caused by a deficiency or malfunction of clotting factor VIII.
- This deficiency leads to prolonged bleeding, either spontaneously or after injuries or surgery, because the blood doesn't clot properly.
- It is typically inherited in an X-linked recessive pattern, primarily affecting males.
- **Symptoms -** Symptoms can range from mild to severe, depending on the level of factor VIII in the blood.
- Mild cases may only cause prolonged bleeding after surgery or trauma, while severe cases can result in spontaneous bleeding into joints, muscles, or internal organs.
- **Treatment -** Typically involves replacing the missing clotting factor VIII, either through infusions of factor VIII concentrates or newer therapies like emicizumab.

Von Willebrand Factor (VWF)

- VWF is a *protein in the blood* that plays a crucial role in blood clotting.
- It acts like a "glue" that helps platelets (small blood cells) stick to each other and the damaged blood vessel wall, forming a plug to stop bleeding.
- **Symptoms** - Prolonged bleeding, Easy bruising, Frequent nosebleeds, Heavy menstrual periods.
- **Types of VWD -** Type 1 is the most common and typically mild, while Type 2 has subtypes with varying degrees of severity, and Type 3 is the rarest and most severe.
- **Inheritance -**VWD is typically inherited, meaning it is passed down from parents to children through genes.
- Most cases are inherited in an autosomal dominant pattern, meaning only one copy of the affected gene from either parent is enough to cause the disease.
- **Treatment - Desmopressin (DDAVP) -** This medication can help release stored VWF from the body.

- **Replacement therapy -** In some cases, purified VWF or a combination of VWF and factor VIII (another clotting factor) may be infused.
- **Other medications -** Antifibrinolytic agents may be used to help prevent or slow down bleeding.
- **Prevalence in India** - India is estimated to have 1.5 lakh people with haemophilia, but only about 27,000 are officially diagnosed
- Incidence of haemophilia A is 1 in 5,000 male births. VWD is known to affect 1% of the general population.
- The VWD is an autosomal disease. This means both males and females can get it equally. It is *not linked to gender like haemophilia.*
- Haemophilia A and Von Willebrand Disease (VWD) represent significant public health challenges in India due to underdiagnosis and limited access to diagnostic facilities.

Reference

Economic Times | Test Kit for Haemophilia A, Von Willebrand Disease