CLOVES Syndrome

Prelims: *Current events of national and international importance| General science*

Why in News?

Recently, Researchers believed that PIK3CA causes CLOVES Syndrome by mutations in a gene order.

- **CLOVES Syndrome –** Congenital, Lipomatous, Overgrowth, Vascular malformations, Epidermal nevi and Spinal/skeletal anomalies/Scoliosis.
- It is a ***rare overgrowth genetic disorder*** caused by mutations in a gene as a part of a larger group of similar disorders called PIK3CA-related overgrowth spectrum or PROS.
- This gene makes one of the proteins in an enzyme called PI3K.
- Mutations in the PIK3CA gene may cause the PI3K enzyme to become overactive, which may cause cancer cells to grow.
- These found in many types of cancer, including cancers of the breast, lung, ovary, stomach, brain, colon, and rectum.
- **1st discovered –** 1867.
- **Risk factors –** PIK3CA gene are believed to occur spontaneously in the womb.
- It is not hereditary and cannot be passed on. At present, there are no known risk factors.
- Mutations in the gene result in two sets of cells in the body (one set with the mutation and one set without) and the mutated cells lead to the formation of abnormal tissue.
- **Vulnerables -** It affects males and females equally regardless of their race or ethnicity.
- **Symptoms –** Babies are born with this condition, can range from mild to severe and may vary from child to child include:
    - Soft masses of fatty tissue on the abdomen, back, sides and buttocks.
    - Vascular or blood vessel anomalies such as dilated veins that may be seen in the chest, arms and legs and that may pose a risk of blood clots.
    - Large and wide hands or feet, with large gaps between fingers and toes, ‘port wine stain’ birthmarks.
    - Spinal problems such as scoliosis or a tethered cord.
    - Kidney problems, skin abnormalities such as raised bumps, and sometimes, intestinal problems and asymmetry in growth.
- **Diagnoses –** A detailed physical examination, medical history and through various imaging tests such as MRIs, CT scans, ultrasounds and X-rays.
- A confirmation of the diagnosis can be made with *molecular genetic testing for the mutation.*

- **Treatments –** There is no cure as yet, usually involves a multidisciplinary team of specialists.
- **Scenario in India -** A few cases of CLOVES syndrome have been documented in India.

Reference

The Hindu| CLOVES Syndrome

