

Endometriosis – India's First Genome Study

Prelims: Current events of national and international importance | Disease

Why in News?

India's first genome-wide study of endometriosis has identified 21 genetic regions potentially linked to the disease in Indian women.

- **Endometriosis** - It is a chronic condition in which **tissue similar to the lining of the uterus (endometrium) grows outside the uterus**.
- This condition leads to inflammation and the formation of scar tissue.
- It can develop at any time from **menarche to menopause**, regardless of ethnic or social background.
- It can also affect **transgender men and non-binary menstruating individuals**.
- Globally, it affects approximately 10 percent of women of reproductive age, or about 190 million individuals.

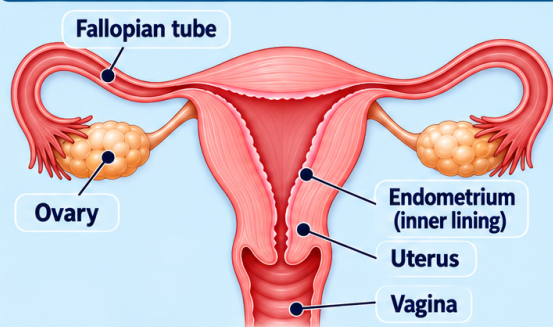
Endometriosis

When endometrial-like tissue grows outside the uterus

Not just period pain

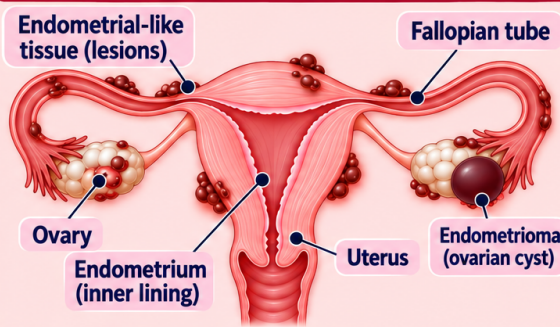


Normal Female Reproductive System



In a healthy system, the endometrium lines the uterus and sheds during menstruation.

Endometriosis



In endometriosis, tissue similar to the uterine lining grows outside the uterus, causing inflammation, pain and sometimes infertility.



Common cause of chronic pelvic pain in women.



Tissue responds to hormonal changes during the menstrual cycle.



Can affect fertility.



Often underdiagnosed; requires clinical evaluation and imaging.

- **Treatment** - No definitive cure currently exists.
- **Common symptoms**
 - Severe menstrual/period pain

- Chronic pelvic pain
- Pain associated with menstruation
- Infertility
- Sometimes other pain-related symptoms
- **Potential Future Applications**
- Study provides evidence that **genetic susceptibility may contribute to endometriosis**.
- The findings may contribute to research in the following areas:
- **Disease mechanisms** - Advancing understanding of the biological pathways involved in endometriosis.
- **Earlier recognition** - Facilitating the identification of women who may require closer clinical attention.
- **Non-invasive diagnostics** - Supporting the development of diagnostic approaches that could reduce reliance on invasive procedures.
- **Personalised treatment** - Enabling the customization of treatment strategies based on individual biological or genetic profiles.

Endometriosis Clinical and Genetic Research in India (ECGRI)

- The initiative was established to develop a comprehensive clinical and genomic dataset of Indian women diagnosed with endometriosis.
- **Key Features**
- The study involved collaboration among **18 centres across India**.
- Participants included women with **surgically confirmed diagnoses of endometriosis**.
- Control participants were also included for comparative analysis.
- The dataset integrates the following components:
 - Clinical information
 - Genomic data
 - Biorepository

Reference

[Indian Express | Endometriosis Could Be Genetic](#)