

Mutations

1. HPV & Female Genital Diseases

- High risk HPV (especially HPV 16 and 18) usually integrate into the host genome and express large amounts of certain viral proteins called E6 and E7 proteins, which block or inactivate tumor suppressor genes p53 and RB, respectively.

2. Endometrial carcinoma

Endometrioid carcinoma

- Mutation in DNA mismatch repair gene
- Mutation in PTEN

Serous carcinoma

- Mutations in the p53 tumor suppressor gene

3. Ovarian Epithelial Neoplasms

Sporadic cases:

- BRCA 1 and 2 → 10%
- p53 → 50% -most common-
- HER 2/NEU over-expression → 35%
- K-RAS protein over-expression → 30%

Familial cases:

- BRCA1 and 2

Serous tumors

- BRAF and K-RAS mutations → Borderline & low grade serous carcinomas
- p53 and BRCA1 mutations → High-grade serous carcinomas

4. Tubal malignancies

Serous tubal intraepithelial carcinoma (STIC)

- TP53 (90%)
- Increased in women with BRCA mutations

4. Breast cancer

- BRCA1 and BRCA2
- HER2 amplification
- TP53
- PTEN