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## Endometrial adenocarcinoma: A genetic focus review

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Endometrial Cancer (EC) occurs worldwide with 36,000 new cases and 6,000 deaths per year. Patients with an advanced disease have a long-term survival of less than 50%. The mean age of the patients is around 63 years.

It is known that the incidence of cervical cancer in Mexico is 7,800 new cases per year. Endometrial carcinoma ranks second among gynecological cancers, after cervical cancer.

Abnormal uterine bleeding is the most common finding in endometrial adenocarcinoma. Patients with advanced disease have a clinical presentation similar to that of ovarian cancer with abdominal-pelvic pain, abdominal distention, early satiety and changes in defecation habits.

### Genetic Alterations in the Development of Endometrial Cancer

#### PTEN

One of the genes that is the most frequently altered is PTEN. When this happens it can lead to the development of adenocarcinoma. It plays an important role in the PI3K-PTEN-AKT-mTOR pathway. Therefore, if PTEN is lost or altered, aberrant cell growth will result. It has been observed that PTEN mutation is not enough for endometrial cancer to develop. Other genes participating in this alteration are KRA5 (15% to 30%),  $\beta$ -catenin/CTNNB1 (14% to 44%), PIK3CA (26% to 36%), PAX 2 (77%) and microsatellite repair factors (20% to 45%).

#### PIK3CA and PIK3R1

PIK3CA and PIK3R1 mutations are usually co-found with PTEN abnormalities, these genes encode for the catalytic and regulatory subunits of P13K. It has been suggested that these mutations have synergistic or additive effects to the alterations of PTEN.

#### Microsatellite instability (IMS)

Microsatellites are short sequences of polymorphic DNA. When errors occur in microsatellite repair, it leads to wrong base pairing, thus forming microsatellite instability. In endometrioid adenocarcinomas, IMS has been found in up to 45% of cases.

In the future, a panel of biomarkers with PTEN, P53, ARID1, etc. may be essential for diagnosis of endometrial carcinoma in order to know the prognosis of such patients.

#### References

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### **Biography**

Norma Estela Herrera González has been trained in molecular biology and immunology. Her passion is to investigate about the role of those copies of genes that acquire inactivating mutations and the role of pathways relevant in carcinogenesis.

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