CASE STUDY

How Do You Treat a Rare And Aggressive Ovarian Cancer?

Quick Summary

- Samiran Kaur*, an 53- year-old patient was diagnosed with small cell carcinoma of the ovary (neuroendocrine type).
- Genetic analysis of the tumor tissue led to the identification of mutations in the *PIK3CA* and *PTEN* genes.
- Everolimus was recommended as a therapeutic option for her.
- Genetic analyses can help to identify targeted therapies for the treatment of rare cancers.

Introduction

The most common form of ovarian cancer is the epithelial type - the cancer that develops from the tissue that covers the ovaries. Typically, 90% of all ovarian cancers are the epithelial type. The other classes of ovarian cancer are germ cell carcinoma, stromal cell carcinoma and small cell carcinoma of the ovary. Small cell ovarian cancers are really rare and make up just a tiny fraction (0.1%) of the total ovarian cancer cases. Usually, this type of cancer is evident in young women. An unusual case of small cell ovarian carcinoma in a 53-year-old woman was referred to Strand Life Sciences for genetic analyses.

Patient Profile

Samiran Kaur, 53- years-old, put her symptoms down to menopause, in the first few months when she experienced persistent bloating and irregular menstrual cycles. Pain in the pelvic area and feeling constantly full were also some of her health complaints. She also had bouts of indigestion and paradoxically, constipation at times as well. When the symptoms persisted, she consulted her general physician, who, in turn, referred her to Dr. Sewanti Limaye. Dr. Limaye is a prominent oncologist in Mumbai. She suspected a case of ovarian cancer and advised some medical investigations.

Results from these investigations showed that Samiran Kaur was suffering from ovarian cancer. Surprisingly, Samiran's ovarian cancer was a small cell ovarian cancer- one of the rare subtypes of ovarian cancer.

In this sub-category, Samiran's cancer was a neuroendocrine tumor.

Since this type of cancer was not usually seen in patients over 50 years of age, Dr. Limaye also advised Samiran Kaur to send a sample of her biopsy for genetic analysis. Genetic analysis of the tumor was expected to provide some directions for choice of therapeutic drugs.

Name changed to protect patient privacy

Results of Genetic Testing

The StrandAdvantage 48-gene somatic cancer test was prescribed for the analysis of Samiran Kaur's cancer. This test is designed to identify mutations in 48 genes that are associated with most cancers.

Therapy	Relevant Markers	Approved Indications	Trials
Everolimus	*PIK3CA* ^{E542K},*PTEN* ^{N69fs} *PTEN* ^{S10C}	Pancreatic Neuroendocrine Tumor, Gastrointestinal Neuroendocrine Tumor, Lung Neuroendocrine Tumor, Kidney Cancedr, Astrocytoma, Hormone receptor-positive HER2-negative Breast Cancer	NCT01827384, NCT02029001

In Samiran's case, one mutation in the *PIK3CA* gene- *PIK3CA*^{E542K}, and two mutations in the *PTEN* gene - *PTEN* ^{N69fs} and *PTEN* ^{S10C} - were detected in the tumor sample.

Therapeutic Options

Small cell ovarian cancer can be treated with platinum-based chemotherapy agents. In addition to using time-tested therapies, genetic analysis of these rare tumors is also advised, in order to guide choice of targeted therapeutic drugs (Foulkes et al. 2016).

In Samiran Kaur's case, the identified mutations in the *PIK3CA* and *PTEN* genes are predicted to create protein versions that can enhance cell growth *(PIK3CA)* and lose growth suppressing activity *(PTEN)*, respectively. These two genes are engaged in a cellular signaling pathway that can be inhibited by inactivating another protein- mammalian target of rapamycin-mTOR. This cellular pathway has been known to be active in neuroendocrine tumors (Cingarlini et al. 2012).

Everolimus is a drug that can be used to inhibit the growth of cells when genes like *PIK3CA* and *PTEN* are mutated (Dillon & Miller 2014).

This drug was recommended for treatment of Samiran Kaur's small cell carcinoma of the ovary - neuroendocrine type.

Conclusions

- Samiran Kaur, aged 53 years, was diagnosed with small cell carcinoma of the ovary - neuroendocrine type.
- Mutations in the *PIK3CA* and *PTEN* genes were identified from the tissue biopsy using the StrandAdvantage 48-gene test.
- The identification of these mutations allowed for the recommendation of Everolimus as a targeted therapy drug for her treatment.
- Genetic analysis helped to increase therapeutic options for this patient, in addition to platinum-based chemotherapy.

StrandAdvantage 48-gene Test

The StrandAdvantage 48-gene Test detects alterations in hot spot regions of 48 genes and reports on those that have possible therapeutic implications.

References

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