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Annual Congress on

ONCOLOGY AND BIOMARKERS SUMMIT

November 27-28, 2017 | Atlanta, USA

Next generation sequencing and immuno-histochemistry profiling identify numerous biomarkers for personalized therapy of endometrioid endometrial carcinoma

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Endometrial cancer (EC) is the most common cancer of the female reproductive tract. In the current study, we were presented with a case of premenopausal woman suffering from EC and having a cancer family history from both paternal and maternal sides, an observation that suggests the presence of germline mutations. The main aim was to accurately classify our case of EC into a subtype, then, to report the associated genetic alterations and protein bio-markers. Furthermore, we aimed to develop individualized treatment strategies designed for maximum effectiveness. Multiple profiling technologies, including immunohistochemistry (IHC), next-generation sequencing (NGS) and chromogenic in situ hybridization (CISH) were used. Forty-four genes including proto-onco and tumor suppressor genes were sequenced to identify causal mutations, in total, 8 mutations in 5 genes were reported (phosphatase and tensin homolog, phosphatidylinositol-4, 5-bisphosphate 3-kinase, catalytic subunit alpha, etc.). Chromogenic in situ hybridization did not show any gene duplications or deletions. In addition, immuno-histochemistry analysis revealed altered levels of programmed death (PD-1) protein biomarker. Since the tumor was positive for PD-1, pembrolizumab (monoclonal) treatment followed Everolimus. Interestingly, the affected individual responded positively after 5 cycles of treatment (over 24 weeks) and tumor size decreased in size from 7 cm x 4.4 cm x 10.5 cm to 6.5 cm x 3 cm x 7.5 cm. Our results have deciphered genetic and protein biomarkers that might be implicated in the aetiology of endometrial cancer. Furthermore, it has established the guideline for a personalized treatment targeting the altered gene products.

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