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An observational study of synchronous/metachronous tumours in microsatellite unstable/mismatch repair deficient endometrial carcinomas (2009-2015)

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ABSTRACT

Objective

Microsatellite instability (MSI) is one of the genetic abnormalities underlying endometrial carcinoma, especially endometrioid endometrial carcinoma (EEC). Microsatellite unstable endometrial carcinomas may be associated with multiple primary malignancies. We aimed to document the incidence and type of associated tumours in MSI/mismatch repair deficient (MMR-d) endometrial carcinomas at a single institution in South Africa.

Methods

The study assessed EECs for the period 2009-2015 at a Johannesburg hospital and followed on previous research undertaken in our department in which 66/145 MSI/MMR-d EECs were identified using immunohistochemistry and polymerase chain reaction (PCR). After ethical clearance was granted, using the microsatellite unstable/MMR-d endometrial carcinoma case-specific laboratory reference numbers, a retrospective search and cross-reference for associated histologically proven tumours, was performed.

Results

Three patients (4.5%) out of 66 confirmed MSI/MMR-d endometrial carcinoma cases had associated tumours, including an invasive squamous cell carcinoma of the cervix and mucinous breast carcinoma. One patient had two tumours, namely uterine leiomyosarcoma and rectal adenocarcinoma. The incidence of EEC patients with an associated tumour is significantly lower ($p=0.0045$) than that documented in the United States of America (USA).

Conclusion

Our study's incidence of associated tumours in MSI/MMR-d endometrial carcinoma patients was significantly lower than studies from the USA. The associated tumours in our study included traditional Lynch syndrome tumours such as colonic adenocarcinoma and demonstrated less commonly associated tumours, namely cervical carcinoma and uterine leiomyosarcoma. These findings highlight the need for further research in our population to assess risk factors and the true incidence of Lynch syndrome-associated tumours.