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Long-term survival of patients with mismatch repair-deficient, high-stage ovarian clear cell carcinoma

[1,2]Colin JR Stewart, [3,4]David DL Bowtell, [2]Dorota A Doherty, [2,5]Yee C Leung.

[1]Department of Pathology, King Edward Memorial Hospital, Perth, Western Australia, [2]School for Women's and Infants' Health, University of Western Australia, [3]Peter MacCallum Cancer Centre, East Melbourne, Victoria 3002, Australia, [4]Department of Biochemistry and Molecular Biology, University of Melbourne, Parkville, Victoria 3052, Australia, and [5]Department of Gynaecological Oncology, King Edward Memorial Hospital, Perth, Western Australia.

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Correspondence:

Dr C Stewart, Department of Histopathology, King Edward Memorial Hospital, Bagot Road, Perth, Western Australia 6008.

Telephone: 08 9340 2715

Fax: 08 9340 2636

Email: colin.stewart@health.wa.gov.au

ABSTRACT

Aims: Gynaecological cancer patients with germline mutations appear to have a better prognosis than those with sporadic malignancies. Following the observation of long-term survival in a patient with stage III ovarian clear cell carcinoma (CCC) and possible Lynch syndrome (LS), DNA mismatch repair (MMR) protein immunohistochemistry was performed in a series of high-stage CCC and correlated with patient outcomes.

Methods and Results: Thirty-two consecutive cases of stage III/ IV ovarian CCCs accessioned between 1992 and 2015 were examined. The tumours from two patients (6%), including the index case, showed loss of MSH2/MSH6 expression while MLH1/PMS2 staining was retained. The index patient subsequently developed colonic and rectal carcinomas that were also MSH2/MSH6 deficient while the second patient had a genetically confirmed germline *MSH2* mutation. All other tumours showed retained expression of the four MMR proteins. The two patients with MMR protein-deficient tumours were alive 160 months and 124 months following surgery whereas the median survival of patients with MMR protein-intact CCCs was 11.8 months (75^{th} and 25^{th} percentiles of 8.1 months and 39.3 months, respectively), with 21 patients deceased due to tumour.

Conclusions: Larger studies are required but high-stage, MMR protein-deficient CCCs may have a relatively favourable prognosis.