Reviewer's report

Title: Unusual presentation of Lynch Syndrome

Version: 1 Date: 8 March 2009

Reviewer: Mef Nilbert

Reviewer's report:

Yu et al. present a case history describing an HNPCC family in which an individual developed 4 metachronous cancer not readily linked to HNPCC. Though it represents a sporadic case, it highlights the broad phenotypic spectrum of HNPCC and if thereby of interest, particularly to clinicians within the field. The format is acceptable, though the Discussion section could be shortened in line with the contribution being a case report.

REVISIONS NEEDED BUT CAN BE TRUSTED TO BE PERFORMED BY THE AUTHORS

INTRODUCTION. The risk of colorectal cancer (up to 80%) could be modified to a range and could included a more recent reference based on larger HNPCC cohorts (in which the risk indeed is lower).

CASE REPORT.

The description synchronous leiomyosarcoma is not used in sarcoma terminology. What is meant? A primary tumor with a satellite metastasis? Independent sarcomas is utterly rare.

Was MSI/immunostaining performed in the adenosquamous cervical cancer (in the sister)? These data could be relevant for its potential link to HNPCC.

The MSI status of the sarcoma should be defined in relation to positive markers and as low/high.

DISCUSSION

Soft tissue sarcomas have indeed been reported in more than 2 families (see Nilbert et al., Fam Ca, 2009 e-publ).

REFERENCES

#27 needs to be completed or presented as unpublished data.

FIGURES

Fig. 2 does not contain A-D (as indicted in the legends).

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Declaration of competing interests:
No competing interests apply.