Reviewer’s report

Title: Mutation Spectrum in South American Lynch Syndrome Families

Version: 1 Date: 17 September 2013

Reviewer: Aung Ko Win

Reviewer’s report:

Major Compulsory Revisions

Abstract
- The abstract needs to be rewritten with more detail information.

Methods
- The details of patients/families selection and published data selection should be reported.
- Why did the centers genotype only on MLH1 and MSH2? Why not MSH6 and PMS2?
- Please add Statistical Analysis section.

Results
- What is the total number of patients/families eligible for genetic testing? (Is it 141 families in the first paragraph of Discussion?) What is the prevalence of mutation in patients/families who met AC-I/AC-II?
- That would be more clear if the authors can present the flow chart of selection of patients/number of eligible patients (how many met AC-I, how many met AC-II)/how many tested/how many observed etc…
- Of 87 patients/families, 69 were classified as disease-predisposing, 9 were VUS - what about the remainder 9?
- What cancer did the authors refer to in “The mean age at first diagnosis of…”?
- Present the mean age at first diagnosis with standard deviation for colorectal cancer (and endometrial cancer and other cancers separately) for MLH1 and MSH2 mutation carriers/families separately.
- Page 6, line 1: what does “recurrent” mean for 13 mutations?
- Page 6, line 2-10: These should be in introduction or discussion.

Discussion
- What are the limitations of the study?

Level of interest: An article of importance in its field

Quality of written English: Needs some language corrections before being published
Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests: None