Author’s response to reviews

Title: Identification of genetic variants for clinical management of familial colorectal tumors

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Re: MGTC-D-17-00172

Dear Editor,

Thank you for the positive and constructive reviews of the manuscript above. We have revised the manuscript in line with the reviewers’ suggestions and we respond to reviewers as follows:
Reviewer#1

- Comments:

I would only suggest the authors add some clinical comments if they wish, as it would strengthen the conclusions.

Response: We agree and have included a text on the topic (Result section, page 9).

Reviewer#2

- Comments:

The authors have addressed the comments made in the first review. Nevertheless, when reviewing the current evidence about the clinical validity of the only CHEK2 deleterious variant identified in this small cohort of 34 patients at risk of inherited CRC, there is not enough evidence to support an association between this germline finding and the CRC susceptibility. In my opinion, the authors remain too strong in their conclusions, as their percentages are based in only one case. I'd suggest to increase the study population, or perform a case-control study to assess the association of this variant with CRC susceptibility.

Response: We agree, and we have therefore modified the phrasing. We now mention this aspect in the Discussion. (Abstract section, page 2; Discussion section, page 9-10; Conclusion section, page 11).

With these alterations, we hope that the manuscript may be acceptable for publication in BMC Medical Genetics.

On behalf of the authors

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