Reviewer’s report

Title: Validation of a digital identification tool for individuals at risk for hereditary cancer syndromes

Version: 0 Date: 20 Jul 2018

Reviewer: Florentia Fostira

Reviewer's report:

The authors describe a digital ID tool developed in order to accurately identify high-risk individuals in the context of BRCA-related cancers and Lynch syndrome, based on NCCN criteria. In the retrospective analysis, 197 pedigrees have been evaluated by both the digital tool and a certified genetic counselor, leading to 100% and 99.47% concordance in the high-risk and low-risk group, respectively.

This is a well-executed approach that can be potentially really helpful in identifying individuals that are at high risk for cancer diagnoses and are possibly missed. In general, this is a well-written paper with a good structure, throughout.

I only have some minor comments:

(a) In the abstract and in the main text, apart from HBOC and Lynch syndromes, polyposis syndrome is also mentioned as a potential syndrome that can be evaluated via the digital tool, but no cases were actually evaluated. Therefore, this should be removed throughout the text. The authors can report in the discussion that this can be also approached by the tool, but they had no cases in this study.

(b) I feel that there should be emphasized that the tool cannot stand alone and that it should be used by healthcare providers at least with moderate genetics background. This should be discussed and be clear enough.

(c) The authors mention a number of online tools; their actual use in clinical practice is debatable. The one that stand out from those is PREMM. I think that there should be some comparison/comment in the discussion about that.

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