Reviewer’s report

Title: Novel BRCA2 pathogenic variant c.5219T>G p.(Leu1740Ter) in a consanguineous Senegalese family with hereditary breast cancer

Version: 1 Date: 28 Mar 2019

Reviewer: Florentia Fostira

Reviewer's report:

The authors have adhered to suggestions, to a large extent, but still there are multiple minor errors. I have made an effort to collect them.

Abstract, line 43: Rephrase to: We report a BRCA2 pathogenic variant in a …

Abstract, line 50: add the word novel before nonsense.

Gene names are always shown in Italics. Please change to BRCA2, BRCA2 an BRCA1/2 throughout the manuscript, including the title.

Lines 65-66: breast cancer risk is associated to BRCA1/2 pathogenic variants, not the genes per se. Please rephrase. Consequently, after rephrasing omit lines 67 and 68

Line 81: Here should be replaced with herein.

Line 82: Add family before history.

Methods-Line 84: Probably it is better to change the subtitle to Case presentation, as you are mostly referring to the single family/case.

The healthy cohort described in Methods is different from the one described in the Abstract. What is valid? Add a paragraph describing specifically your control group.

Methods: Paragraph describing BRCA1&2 sequencing is too detailed, describing amplicons, etc and I don’t feel that this is necessary.

Lines 115-116: Rephrase; Maybe DNA was of poor quality?

Line 118: There is no need to declare that there were no BRCA1 pathogenic variants. Omit the sentence.

Lines 118-120: Rephrase to: A BRCA2 pathogenic variant, namely…. Lines 121-122 should be transferred in this sentence.

Lines 128: Please rephrase. What is causal elements?
Discussion, line 141: Diagnosis before the age of 55 years is not younger since median age at
diagnosis in European populations is 52 years. Maybe you meant something else?

Lines 156-157. Please rephrase. Maybe: Pathogenic variants involving the particular codon
haven't been reported in SSA populations?

Lines 158-161. Please merge the two sentences to make your point.

Lines 184-189: This whole paragraph should be transferred to the Results section. Please
proofread it, as well.

Lines 190-194: This paragraph is not well-written and it doesn't make any sense. Please re-write
it and transfer it before the paragraph starting in line 176.

Line 196: Switch new to novel.

Line 197: Please add family before history.

In Figure Legends: Figure 2 - omit reverse strand.

Pedigree format - I am not sure if this is going to look good in the printed version.

**Are the methods appropriate and well described?**
If not, please specify what is required in your comments to the authors.

Yes

**Does the work include the necessary controls?**
If not, please specify which controls are required in your comments to the authors.

Yes

**Are the conclusions drawn adequately supported by the data shown?**
If not, please explain in your comments to the authors.

Yes

**Are you able to assess any statistics in the manuscript or would you recommend an
additional statistical review?**
If an additional statistical review is recommended, please specify what aspects require further
assessment in your comments to the editors.

Not relevant to this manuscript
Quality of written English
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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