Reviewer’s report

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

Version: 0 Date: 29 Oct 2018

Reviewer: Florentia Fostira

Reviewer's report:

The authors have been able to make significant changes to the manuscript. At this point, the article is easy to read through, while the language is much improved.

The majority of my corrections (2-7) involve the discussion part, which has some flaws language-wise and doesn't flow smoothly:

1. Table 1 is unnecessary and does not add any value to the information provided.

2. The first paragraph of the discussion is weak and needs to be rephrased, e.g. type of cancer instead of cancer in the first line, triple negative histopathology instead of tumors, etc. Also, characteristics do not apply.

3. In the second paragraph of the discussion: breast cancer risk is associated to pathogenic BRCA1/2 variants and not to the genes per se, moderate genes not minor, TP53 not P53. Also, remove the word dominant as this refers to type of inheritance and not the type of variants.

4. In the third paragraph: remove brackets form table 2. I do not understand the point of the sentence "Any pathogenic variant at this position has not yet been reported in SSA 158 populations". Please amend.

5. The following sentences should be rephrased: "Meanwhile other pathogenic variants surrounding this position of the BRCA2 gene have been reported in the ClinVar database. These pathogenic variants lead to stop codons or frameshift at amino acid 1740 or 1741 of BRCA2 protein (Table 3)". I think what you are trying to say is that other pathogenic variants have been identified in adjacent nucleotides and/or amino acids and these are summarized in Table 3.

6. Line 181: correct to family, also correct to BRCA1/2 genetic testing (BRCA genes testing).

7. The comment in lines 182 and 183 should be removed, as it has not been mentioned throughout.

8. Close-up should leave some future perspective.
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.

Unable to assess

Are the conclusions drawn adequately supported by the data shown?
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Yes

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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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