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: 10 Jan 2019

Reviewer: Masami Arai

Reviewer's report:

In this revised version, the article has been more refines. But there is still room for improvement.

Major points

1. Family tree is still incomplete and confused.
   1) Two or three sibling lines are very close and it is too difficult to their relationship. Relationship lines should be drawn more separately to distinguish siblings per family.
   2) Individual numbers in each generation should not be described in each symbols. For example, ③ means 3 females in the sibling. To describe a female like 3○ is recommended.

2. You confirmed that out of 19 women who you performed genetic analysis of BRCA2, only 4 women had deleterious mutation in BRCA2. Therefore you should show the remaining 15 women with wild type in BRCA2 as E- in Fig. 1.

3. I understand why you performed sequence analysis for negative control. But cancer free healthy control are usually influenced by their age because breast cancer is adult onset disease. You should at least show the data of average age of control group and non-familial breast cancer patients group. Furthermore, what is the purpose of analysis BRCA2 mutation for breast cancer patients without family history? What do you reveal by this analysis?

If you found the same mutation in BRCA2 in sporadic breast cancer group, what do you conclude in this article?

4. You should comment whether there were family member with Fanconi anemia like symptom or early death, or not. If impossible, the sentence "it is unclear that whether there were family member with Fanconi anemia like symptom or early death, or not" should be inserted in Discussion.

5. What is the clinical and pathological characteristics of 15 recruited subjects described in line 181 which suddenly appeared in Discussion. Materials in this article consisted of 15 full sequence analysis materials (Are subjects patients with family history of breast cancer?), 48
healthy control and 48 breast cancer patients without family history. Is this right? Please clarify materials (subjects). What is the results of BRCA1/2 genetic analysis in the remaining 14 cases?

Or you should make this article case report for novel BRCA2 deleterious mutation, not to refer to remaining 14 cases.

Minor points

1. SBR in line 89 is not so commonly used in histological grade. This term should be listed in Abbreviations in page 12.

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

No

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:

Acceptable

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