Author's response to reviews

Title: A novel germline PALB2 deletion in Polish breast and ovarian cancer patients

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Author's response to reviews: see over
To
Melissa Norton, MD
Editor-in-Chief of BMC Medical Genetics


Dear Dr Norton,

We have made some corrections according to the editors' suggestions received in the last e-mail. Enclosed we are sending the revised version of the manuscript, as well as the response to the BMC Series Editorial Production Team.

We could not open the attached referee comment, however, we did not receive any response from the Editorial Office related to this matter. Thus, probably there were no further comments.

Sincerely yours
Jolanta Kupryjańczyk MD, PhD
The BMC Series Editorial Production Team Comments and responses:

Formatting changes to be made:

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Major revisions (we require the author to make these changes)

Affiliations: Each affiliation should be written in full, in the following format: Department, Institute, (Street), City, (Postal code), Country. This may mean repeating institutional addresses if the authors are from different departments within an institution.

Done.

Tables: the first table referenced in the manuscript text should be labelled Table 1. The second, Table 2, etc. The tables should be provided in this same order at the end of the manuscript file. Every table provided at the end of the manuscript file must be referred to in the text.

The tables are numbered according to their appearance in the text. The table 2 that appeared before the table 1 in Methods section was removed from a text.

It was (Methods, Molecular genetic methods, page 7):

Sequence variants of interest were further studied in larger groups of ovarian carcinomas and in control blood samples (Table 2).

We have corrected it as follows:

Sequence variants of interest were further studied in larger groups of ovarian carcinomas and in control blood samples (see: Results).

Minor revisions (we can make these changes for you, although it will speed up publication of your manuscript if you do them while making the major changes above)

Materials: Please include this in the Methods section.

Done. We have changed a title "Material" on the "Subject", as a subtitle of Methods (page 5), and we have added a subtitle "Molecular genetic methods" (page 7)

Contribution: Please rename this section 'Authors' contributions'.

Done.

Figures: Please ensure all mention of figures is consistent (it should be Figure of Fig but not a mixture of both)

We have changed the Fig to a Figure in a text (page 11, paragraphs 2 and 3; and page 12, paragraph 5)
Typography: Please take this opportunity to check your manuscript for any typographical errors and to make any final corrections or revisions. This is the final proofing stage for your manuscript, and you will not be able to make any changes after acceptance.

Done.

We have also corrected other minor errors:

1) We have changed all special Polish letters in the authors' and institutions' name for its English equivalents.

2) We have specified the exons where BRCA1 mutations were analyzed

   It was (Methods, Subjects, Ovarian cancer patients, page 6):
   
   \textit{BRCA1} mutational status (exons 2, 5 and 11) had been previously determined in the majority of ovarian carcinomas (n=243), and 39 (16\%) were positive [18].

   We have corrected it as follows:
   
   \textit{BRCA1} mutational status (exons 2, 5, 11 \textbf{and} 20) had been previously determined in the majority of ovarian carcinomas (n=243), and 39 (16\%) were positive [18].

   It was (Methods, Subjects, Breast cancer patients, page 6):
   
   \textit{BRCA1} mutations had been previously detected for the purpose of counselling in 75 of 982 breast cancer patients (8.3\%).

   We have corrected it as follows:
   
   \textit{BRCA1} mutations (exons 2, 5, 11 \textbf{and} 20) had been previously detected for the purpose of counselling in 75 of 982 breast cancer patients (8.3\%).

3) We have replaced abbreviation "no" for a "number"

   It was (Results, c.509-510delGA, Ovarian cancer patients, page 11):
   
   The analysis of sequencing diagrams (Figure 1) of tumor sample \textbf{no} 375 (about 30\% of stromal cell contamination) and blood DNA of the same patient suggests the loss of heterozygosity at this locus.

   We have corrected it as follows:
   
   The analysis of sequencing diagrams (Figure 1) of tumor sample \textbf{number} 375 (about 30\% of stromal cell contamination) and blood DNA of the same patient suggests the loss of heterozygosity at this locus.

4) We have specified JK contribution in Authors' Contribution section

   It was (Authors' Contribution section, page 17):
   
   JK characterized the ovarian cancer material, analyzed immunohistochemical stainings, participated in the design and coordination of the study, critically reviewed the original and revised manuscript.

   We have corrected it as follows:
JK characterized the ovarian cancer material, analyzed immunohistochemical stainings, participated in the design and coordination of the study, critically reviewed and drafted the manuscript.

5) We have corrected some typographical errors in table 2 and 4

It was (Table 2, 4th column, 12th row):
4.7% (4/70)
We have corrected it as follows:
5.7% (4/70)

It was (Table 2, 5th column, 11th row):
4.5% (16/326)
We have corrected it as follows:
4.9% (16/326)

It was (Table 4, 6th column, 1st row):
23,717034
We have corrected it as follows:
23,717,034