Author's response to reviews

Title: Mutation analysis of the AATF gene in breast cancer families

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Author's response to reviews: see over
Dear Editor,

Thank you very much for the opportunity to submit a slight revision of our manuscript entitled “Mutation analysis of the AATF gene in breast cancer families” by Haanpää et al., for your further consideration of publication as a research paper to BMC Cancer.

Below, we have now responded (highlighted with yellow in the manuscript text) to all requests and inquiries by the two reviewers of our paper, and therefore hope that the slightly revised manuscript now will be found suitable for publication.

Thank you again for your consideration.

Sincerely yours,

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RESPONSE TO THE REVIEWERS COMMENTS:

In the slightly revised manuscript, all requested and suggested improvements to the text as well as the added references have been highlighted with yellow.

Referee 1 (Csilla Szabo):

DR.1) As suggested we have strengthened the biological rationale for AATF.

MER.1) We have corrected this sentence as requested.

MER. 2, 3 and 5) These typographic errors have now been corrected.

MER. 4) As requested we have written out the abbreviation aa (amino acid).

MER 6) As requested we have corrected the format of the table.

MCR. 1) As requested we have re-worked the backgrounds 2nd paragraph, and added more details to describing AATF function with new appropriate references. This should make the text more comprehensible.

MCR. 2) The 17 mutation positive families refer here to the previously identified gene mutations (BRCA1, BRCA2, CHK2, TP53, RAD50, RAP80 or PALB2). We decided to include these 17 families into our study because we did not want to rule out potential modifier effects of AATF mutations. As this is the first comprehensive mutation screening of AATF gene in breast cancer patients, there is no previous knowledge what role AATF mutations could have in breast cancer predisposition (high, moderate or low penetrance, or a modifier effect). We have now made our text more clearly acknowledging this matter.

Referee 2:

MCR) The 17 mutation positive families refer here to the previously identified gene mutations (BRCA1, BRCA2, CHK2, TP53, RAD50, RAP80 or PALB2). We decided to include these 17 families into our study because we did not want to rule out potential modifier effects of AATF mutations. As this is the first comprehensive mutation screening of AATF gene in breast cancer patients, there is no previous knowledge what role AATF mutations could have in breast cancer predisposition (high, moderate or low penetrance, or a modifier effect). We have now made our text more clearly acknowledging this matter.

MER) As requested we have included data form GWAS into introduction.

DR) The text has been changed according to reviewer’s suggestions.