

Reviewer's report

Title: Contribution of the PALB2 c.2323C>T [p. Q775X] founder mutation in well-defined breast and/or ovarian cancer families and unselected ovarian cancer cases of French Canadian descent

Version: 1 **Date:** 22 October 2012

Reviewer: Christos Kroupis

Reviewer's report:

Minor Essential Revisions

This well-written manuscript is trying first to investigate the link of PALB2 gene mutations to ovarian cancer and then, to establish p.Q775X in the panel of recurrent mutations to be tested in the French Canadian population (and especially those from the Montreal area).

1. Authors should stress in the Discussion area the need for haplotype analysis in the future for mutation carriers in order to establish it as a founder mutation.
2. The pedigree of the severely-hit family F1469 is very interesting and readers would like to know more precisely whether this family has been BRCA-analyzed by Myriad or only for the common French-Canadian mutations (it is not mentioned anywhere in the text). Also, about the testing for these common BRCA mutations: have the samples been checked for six or five mutations (or more as mentioned in the Tonin et al, 1998 paper)? If the number is the same for all samples, probably it would be of value to add this info throughout the text (abstract included).

Other minor or discretionary revisions

1. For the sake of readers, please rearrange citations in a consecutive way e.g. [19, 20, 23, 21, 16, 22, 24] should be [16, 19-24].
2. In page 6 line 9, add next to the abbreviation LMP: (Low Malignant Potential).
3. For ovarian cancer staging, please add FIGO (and/or a citation) as this acronym is more easily recognized.
4. In p. 5 L. 11 remove Table 1 as it is mentioned twice. Next line, please correct to ...index "cases"were recruited...
5. In p.6 L.1 please add for clarity ...remaining 26 HBC and 14 HBOC cancer families were found...
6. In p. 9 L9, please add the BRCA2 variant; E2003K
7. In p. 9 L14 correct to "p.Q775X" carrier...
8. In p. 10, L. 21 suggest to change to "none of these two high-grade serous....."

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I have no financial competing interests. I work also in this field and the interesting findings provided in this manuscript could potentially assist my research work in the future.