Reviewer's report

Title: Five recurrent BRCA1/2 mutations are responsible for cancer predisposition in the majority of Slovenian breast cancer families

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Reviewer: Jan Lubinski

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The manuscript entitled „Five recurrent BRCA1/2 mutations are responsible for cancer predisposition in the majority of Slovenian breast cancer families presents spectrum of BRCA1/2 mutations in Slovenian families with familial aggregations of breast/ovarian cancers. The authors demonstrate very interesting results.

Five highly recurrent specific mutations were identified (1806C>T, 300T>G, 300T>A, 5382insC in the BRCA1 gene and IVS16-2A>G in the BRCA2 gene).

They observed an exceptionally high frequency of 4 different pathogenic missense mutations, all affecting one of the cryptic cysteine residues of the BRCA1 Ring Finger domain.

The results will contribute to a growing literature concerning the roles of BRCA1/2 mutations in breast/ovarian cancer predisposition.

Major Comments

The study group is mixed and not well defined. Probands were selected according „liberal intake criteria”.

Authors should clearly summarize study group.

Minor Comments

DGGE and especially PTT are methods with mutation detection sensitivity below 100%. This question should be discussed.

BRCA1 mutations 5382insC and 300T/G (detected by authors in 12 Slovenian families) are common Slavic ancestry mutations (many papers from Czech Republic, Lithuania, Poland, Slovakia, Russia …..) and should be properly presented in discussion.

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 declare that I have no competing interests