Reviewer’s report

Title: BRCA1 Mutations in Women with Familial or Early-Onset Breast Cancer in Estonia

Version: 1 Date: 16 October 2009

Reviewer: Arvids Irmejs

Reviewer’s report:

- Major Compulsory Revisions (which the author must respond to before a decision on publication can be reached)
  NO
- Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)
  NO
- Discretionary Revisions (which are recommendations for improvement but which the author can choose to ignore)
  1. Discussion and comparison with Finnish BRCA1 studies results is lacking (Ethnically closest neighbours!!!)
  2. Criteria for familial cases are very unspecific and therefore this is very heterogeneous group of individuals/families. Very low percentage of pathogenic mutations confirms the low specificity of selection criteria. The same percentage of clinical mutations probably will be detected also in consecutive breast cancer group.
  3. BRCA2 gene testing study could be done in young breast cancer group and in families with 3 or more cases of breast/ovarian cancer, in order to increase knowledge about hereditary cancer etiology in Estonia, because Estonia is geographically very close to Finland, where there are BRCA2 gene founder mutations confirmed.

Level of interest: An article whose findings are important to those with closely related research interests

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