Reviewer's report

Title: Multiplex SNaPshot for detection of BRCA1/2 founder mutations in Spanish and Spanish related breast/ovarian cancer families

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Reviewer: Lilian Jara

Reviewer's report:

1. The focus of the paper is relatively new and is well defined
2. The methods are well described, with sufficient details to replicate their work
3. This paper describes a new technic for the detection of BRCA1/2 founder mutations in breast/ovarian cancer families. The technic named PCR multiplex SNaPshot could be applied to recurrent and founder mutations using a single reaction per patient sample. Nevertheless, it is not a cost-effective analysis as the authors claim, because it needs four different fluorochromes to assay each base and controlled extension primers size. Also it needs a system of capillary electrophoresis and this method is only applicable in populations where founder mutations exist. Therefore, it is not useful in admixed population where the number of high frequency mutations decreases and the range of mutations that need to be included in the pre-screening increases. The last situation is observed in Latin-American populations and in this populations this method is difficult to apply and it is precise in this populations where a cost-effective assay is needed.

4. Yes
5. In the discussion not sufficient data are considered about the number of founder mutations detected in Latin-American population. For example in Chile this method could not be applied due to the great number of detected mutations and the unreported founded mutations, Chilean data are available in the literature as well as are in the Brazilian population.

6. Yes
7. In summary, this paper is well written, but despite that the authors report that they are describing a new technic, it does not differ significantly from others. Also it is necessary to consider that recurrent and founder mutations are specific of each population, therefore in each particular case it is very time consuming in standardizing the system of PCR multiplex, moreover this protocols frequently fail in time. Also it is necessary to have a system of capillary electrophoresis. I think that the article is not acceptable because it is not something really new and it is not applicable to many populations.