Reviewer's report

Title: Prevalence of the BRCA1 founder mutation c.5266dup in Brazilian individuals at-risk for the Hereditary Breast and Ovarian Cancer Syndrome

Version: 1 Date: 26 September 2011

Reviewer: Pål Møller

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One word-wide BRCA1 founder mutation (c.5266) is allegedly reported twice in Brazil. The report validate this finding and report a prevalence of 5% in a highly selected set of patients. The cohort was not defined with respect to family history nor ethnicity besides the statement that they were not Ashkenazi. The cohort was tested for the three Ashkenazi mutations only, and the report followingly have no bearing on the possibile existence of additional frequent Brazilian mutations.

The mns uses too many words to describe breast cancer and inherited breast cancer in general, and without proper references (it is not documented that 5-10% of breast cancers are hereditary, and such a figure may have no meaning without reference to population examined).

An important information is given in last sentence in first section in Discussion: BRCA testing is not available in Brazil besides for a few who may pay privately.

As cost is dramatically dropping, and as results of full sequencing may be difficult to interpret and above all so in a distinct population where normal variation is unknown, to me the conclusion of this study should be that prevalent mutations should be looked for by cheap tests in all immigrant populations the last 500 years, and the test panel should include the founder mutations in the populations the immigrants were coming from. This will give no difficult answers to interpret, an the reported figure of 5% prevalence of one such mutation in the cohort examined, is actually high and indicates that there is much more to gain through this path.

Secondary, there is an obvious need to search for founder mutation(s) in the native Brazilian populations.

To me, the reported finding is interesting, and I would suggest the mns to revised as indicated.

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 conflict of interest.