Reviewer’s report

Title: Clinical and genetic characterization of hereditary breast cancer in a Chinese population

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Reviewer: Rodney Scott

Reviewer's report:

This report assess the prevalence of a variety of germ line variants in two populations, one that has a family history of breast cancer and a personal history of disease the other a population of health women who come from families with a history of breast cancer. The control population cannot be described as a control population as it is heavily biased in favour of genetic predispositions to breast cancer and as such should not be used for this study as it over-estimates the true incidence of causative variants in the population. Much focus is put on the presence of MUYTH variants and little emphasis is placed on the recessive nature of disease associated with this gene. Overall, as it stands the study is poorly conceived and requires significant change.

All germline changes revealed by panel sequencing should be termed germ line causative variants (if they are unequivocally so) and use of the 5-tier rating system should be incorporated into the manuscript. The authors have too few TNBC patients to make any meaningful statement about causative variant prevalence and as such should not attempt to state that the frequency of BRCA mutations in this group of patients is so high.

Level of interest
Please indicate how interesting you found the manuscript:

An article of importance in its field

Quality of written English
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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