Reviewer's report

Title: First evidence of a CHEK2 duplication involved in cancer predisposition in an Italian family with hereditary breast cancer

Version: 1 Date: 16 April 2014

Reviewer: Marta Santisteban

Reviewer's report:

The manuscript describes evidence of a CHECK2 duplication involved in a cancer predisposition in an Italian family with hereditary breast cancer. My comments are related to major compulsory revisions only.

1) I do not understand why the genetic test is performed firstable in an healthy woman and not in a patient with breast cancer (mother or aunts).

2) I understand it is not possible to perform hte genetic test in the family founder. However, I recommend to perform the genetic test in IV-1 member.

2) Why do not search for mutations in p53 or RAD51 genes before the CHECK2?

I agree and recommend to perform MLPA in all these genetic studies in

Level of interest: An article of limited interest

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

None to all of the questions above