Reviewer's report

Title: Mutational analysis of BRCA1 and BRCA2 in hereditary breast and ovarian cancer families from Asturias (Northern Spain). Identification and geographical distribution of eight novel pathogenic mutations

Version: 1 Date: 11 March 2013

Reviewer: Rupninder Sandhu

Reviewer's report:

Minor Essential Revisions
1) Abstract: Title Conclusions is spelled wrong,
2) In Conclusions of the Abstract, the last sentence needs revision...something like.. 'Our findings may help design a first step.....'
3) First paragraph in Background needs references at the end of second sentence that ends in 'certain genes'.
4) References 2 and 3 do not substantiate the claims made in the sentence in terms of proportion, the authors need to provide numerical proportion supported by references instead of using the term large proportion
5) Last sentence in first paragraph needs editing, maybe authors can modify to something like, Testing.....genes can make it feasible to identify....'
6) Patient and Methods section- First paragraph contains the abbreviation HUCA that is not explained before (except in author affiliations). Authors need ti add the expanded form in the text at least once before using the abbreviated form
7) Second paragraph in study population sub-section of Patient and Methods section- D. One ......and one OR more, the R in OR is missing, the authors need to add that.
8) Second paragraph in study population sub-section of Patient and Methods section- H. This criteria is a little vague to understand what authors mean by term 'families close to fulfill any of the above criteria. It will be helpful if authors can be more specific.
9) Last paragraph of study population sub-section of Patient and Methods section-third line starting with 'Samples were.....member, it will be helpful to specify here what samples were taken whether tissue, blood.....etc.
10) Large genomic..... BRCA2-sub-section of Patient and Methods - The authors talk about normalizing the data to two control samples. However, there is no mention of details regarding control samples in terms of what they consisted of, how they were collected etc. The authors should include the details for control samples in parallel to the details of test samples.
11) Results and Discussion-First section (first paragraph)-the authors claim that highest mutation rate in BRCA1 was observed in certain groups but do not talk
about Group Ga that had 50% according to their observations in Table 1. The authors need to integrate that finding in their results.

12) Results and Discussion-First section (first paragraph) -- the authors do not discuss the results of BRCA2 at all (except for in last sentence in association with one patient). The authors should make observations/analyze parallel to BRCA1 here to make the manuscript stronger.

13) Results and Discussion-(second paragraph)- the link to BIC does not work. The authors need to provide a correct link.

14) Results and Discussion-(seventh paragraph in the sub-section BRCA1 large genomic deletion) The authors are missing important information in third sentence 'The deletion was....with and with...' without which it is not possible to understand the sentence. The authors should provide that information to make it understandable.

15) Results and Discussion-(tenth paragraph in the sub-section Deleterious BRCA2 mutations) The data is a little hard to understand the way it is presented, it is highly suggested that the authors revise it to make it more clear. Same paragraph has a grammatical error after reference 34 i is inserted, the sentence should read 'was identified' instead of 'was identify'. The authors should correct that.

16) Results and Discussion-(twelfth paragraph in the sub-section Unclassified variants)- Third sentence reads 'We could......was death......', death word needs to be replaced by 'dead or deceased.

17) Conclusions section-first paragraph -second sentence- should end as 8 of them being novel, the authors need to revise this.

18) Table 6 is not referenced at all in the text. The authors should either remove it or reference it appropriately.

Discretionary Revisions

1) In the acknowledgement section, it is usually not customary to add 'to' after 'thank' but it is the authors' discretion

Level of interest: An article of limited interest

Quality of written English: Needs some language corrections before being published

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