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: 27 February 2013

Reviewer: Ramunas Janavicius

Reviewer's report:

In the manuscript authors delineate BRCA1/2 mutational profile in breast/ovarian cancer families from Asturias, ~1 mln. inhabitants harbouring relatively isolated region in Northern Spain. This is important contribution to the overall BRCA1/2 mutational profile in Europe. However, some points should be addressed before the acceptance should be considered.

Major Compulsory Revisions

1. The title is too long. It would be appropriate to leave the first sentence.
2. The newest HGVS nomenclature version (v2.0) should be used for mutations (e.g. “*” instead of “X” for stop codons)
3. The proportion (in percentages) of recurrent mutations should be presented more clearly. Please also mention, that future haplotype analysis of identified recurrent frequent mutations for the more specific delineation of founder effect is necessary.
4. Table 3. BRCA1 mutation was found in mucinous ovarian cancer type – this is very unusual, because it is generally regarded that mucinous OC is not associated with BRCA+ status. The decoding/pathology data should be critically reviewed.
5. The UVS class according international classification system should be assigned for the reported variants.

Minor Essential Revisions

1. there are typos prevalent throughout the text (e.g. “en”, “pb” etc.), the words should be consistent (e.g. “Exons” to “exons”)

Level of interest: An article of importance in its field

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