Reviewer's report

Title: A de novo complete BRCA1 gene deletion identified in a Spanish woman with early bilateral breast cancer.

Version: 1 Date: 24 August 2011

Reviewer: Laura Papi

Reviewer's report:

Comments to the Author

Garcia-Casado and colleagues report the first case of a “de novo” BRCA1 gene deletion in a Spanish patient without family history for breast and ovarian cancer. The information reported is new and interesting.

• Major Compulsory Revisions
None

• Minor Essential Revisions
1) Abstract, Result section: replace “…. VAT1 locus to the beginning of gene…” by “…. VAT1 locus to the beginning of NBR1 gene….”.
2) Methods, Patients section: specify the age of onset of breast cancers also in the second paragraph.
3) Methods, Mutation analysis of BRCA1 and BRCA2 section: The mutation nomenclature must follow also the HGVS format (http://www.hgvs.org/mutnomen/); moreover, the appropriate GenBank reference sequence and version number for both genes studied should be given.
4) Results, Mutation analysis of BRCA1 and BRCA2: replace “….change was confirmed not to affect splicing,” by “….change was predicted not to affect splicing.”. Maybe, Authors would like to add results of another bioinformatics tool to predict splicing signals (i.e. Human Splicing Finder, URL:http://www.umd.be/HSF/)
5) Discussion: MIM numbers of NBR2 and ZFPM2 are missing.
6) Discussion, ZFMP2 amplification paragraph: replace “…and hence it would be prone to accumulate genetic…” by “ .. and we may speculate that it would be prone to accumulate genetic….”. I do not agree that the paper of Tirkkonen et al (1997) support the finding of a mosaic amplification of ZFPM2 in the Spanish patient. Tirkkonen et al (1997) found 8q gains in BRCA1-associated tumors as well as in BRCA2-associated and sporadic breast cancers supporting the view that 8q gains are common in breast cancers independently by the presence of a BRCA1/2 germinal mutation.

• Discretionary Revisions
1) Abstract, Conclusion section: replace “….in hereditary breast and ovarian cancer families…” by “..in young breast cancer patients without family history, as well as in hereditary breast and ovarian cancer families, …”

**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 declare that I have no competing interests'