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Comments on: del Valle J, Feliubadaló L and Lázaro C. Letter to the Editor

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To the Editor,

In this issue, Jesús del Valle, Lúdia Feliubadaló and Conxi Lázaro rightly noted that we made two nomenclature errors in our paper [1]. They are absolutely correct, in that we made cDNA nomenclature errors regarding the deletion of exon 22 in *BRCA1* and the deletion of exons 15 – 16 in *BRCA2* reported by del Valle et al. [2]. We inadvertently considered nucleotide one of both the NM_007294.2 and NM_000059.3 mRNA sequences as the first nucleotide for nomenclature of the aforementioned mutations, described in tables 1 and 2 of our paper [1]. The manner in which these mutations were designated by del Valle et al.,[2] is therefore correct. We would however like to stress that the genomic DNA nomenclature of these mutations, as indicated in our paper, remains correct.

We sincerely thank Jesús del Valle, Lúdia Feliubadaló and Conxi Lázaro for bringing these errors to our attention.

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2. del Valle J, Feliubadaló, L, Nadal M et al. (2009). Identification and comprehensive characterization of large genomic rearrangements in the *BRCA1* and *BRCA2* genes. *Breast Cancer Res Treat*. Doi: 10.1007/s10549-009-0613-9

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Dear Editor,

We have read with great interest the recent article “Large genomic rearrangements of the *BRCA1* and *BRCA2* genes: review of the literature and report of a novel *BRCA1* mutation” by Michelle D. Sluiter and Elizabeth J. van Rensburg, published online in your Journal [1]. We share the authors’ opinion that it is necessary to establish uniform mutation nomenclature to facilitate interpretation of results. We support the interest in thorough characterization (at the DNA and RNA level) of rearrangements, not only to investigate the mechanism underlying the mutation but also to distinguish between mutations affecting the same exons. Indeed, rearrangements that appear to be the same can have different biological impacts and even different clinical consequences.

However, there are a couple of points that we would like to clarify. In tables 1 and 2 of the Shuiter and van Rensburg paper, some mutations described in our article “Identification and comprehensive characterization of large genomic rearrangements in the *BRCA1* and *BRCA2* genes” [2] are reviewed. Shuiter and van Rensburg state that two of our described mutations are incorrectly named in our previous article, as follows:

- Deletion of exon 22 of *BRCA1* was reported by us as c.5333-198_5387del according to mRNA sequence NM_007294.2. Sluiter and Rensburg state that it should be named c.5533-198_5587del. We deduce that they consider the first nucleotide of NM_007294.2 to be nucleotide number 1 for mutation nomenclature. According to HGVS guidelines for mutation nomenclature, with a coding DNA reference sequence, nucleotide 1 is the A of the ATG-translation initiation codon (<http://www.hgvs.org/mutnomen/recs-DNA.html>). As such we maintain that our designation according to a coding DNA reference sequence is correct.

- Deletion of exons 15 and 16 of *BRCA2* was reported by us as c.7436-270_7805+691del according to mRNA sequence NM_000059.3. As in the first case, Sluiter and Rensburg state that the correct nomenclature of this mutation should in fact be c.7636-270_8032+691del. Furthermore, they have referred by mistake to NM_007294.2 (*BRCA1*) instead of NM_000059.3 (*BRCA2*). We maintain the argument outlined above, based on HGVS guidelines to support our designation.

We believe that it is very important to make these aspects of the nomenclature clear in order to follow HGVS guidelines.

REFERENCES

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2. Del Valle J, Feliubadalo L, Nadal M et al (2009). Identification and comprehensive characterization of large genomic rearrangements in the BRCA1 and BRCA2 genes. *Breast Cancer Res Treat.* doi: 10.1007/s10549-009-0613-9