Author's response to reviews

Title: Clinical characterisation and mutation spectrum in Swedish adenomatous polyposis families

Authors:

Gunilla Kanter Smoler (gunilla.kantersmoler@astrazeneca.com)
Kaisa Fritzell (kaisa.fritzell@karolinska.se)
Anna Rohlin (anna.rohlin@vgregion.se)
Yvonne Engwall (yvonne.engwall@vgregion.se)
Birgitta Hallberg (birgitta.j.hallberg@vgregion.se)
Annika Bergman (annika.bergman@clingen.gu.se)
Johan Meuller (johan.meuller@astrazeneca.com)
Henrik Gronberg (henrik.gronberg@ki.se)
Per Karlsson (per.karlsson@oncology.gu.se)
Jan Bjork (jan.bjork@karolinska.se)
Margareta Nordling (margareta.nordling@obgyn.gu.se)

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Author's response to reviews: see over
Dear Editor

We have revised our manuscript (MS ID: 1663264206185915) in accordance with the suggestions made by the referees in their review and we hereby re-submit our revised version of our manuscript entitled:

"Clinical characterisation and mutation spectrum in Swedish adenomatous polyposis families"

by authors Gunilla Kanter Smoler, Kaisa Fritzell, Anna Rohlin, Yvonne Engwall, Birgitta Hallberg, Annika Bergman, Johan Meuller, Henrik Gronberg, Per Karlsson, Jan Bjork and Margareta Nordling

We hope that we have been able to respond satisfactorily to all of the reviewer’s comments.

Yours sincerely,

Margareta Nordling, Associate Professor

We have made the following changes (point by point response to the reviewer’s comments;
**Point by point response to reviewer’s comments (080306):**

**Discretionary Revisions:**

1. The reason that we calculated the frequency of APC *de novo* mutation to be 16% and not 12.3% is that in 17 of the 81 patients we had no data on whether the mutation was *de novo* or not (see Table 1). Thus the number of patients with inheritance data was calculated to be 81-17=64 and as we had 10 patients with verified *de novo* mutation the *de novo* mutation frequency was calculated to be 10/64=15.6 or 16%.

2. It was 3 patients with *de novo* mutations that were examined for mosaicism. We have corrected this number in the Discussion, page 18.

3. We agree that the mosaic mutation was probably detected by heteroduplex formation. With our method SSCP/HD we get a combined SSCP and HD (HD=Heteroduplex) detection. We have clarified that the aberrant band probably originated from heteroduplex formation on page 12.

4. Revised as suggested.

5. The word phenotype was missing. Corrected.

6. Corrected. See point 2 above.

7. There was a mix-up of word in this sentence that has now been corrected.

8. The page and volume of this reference has now been added.