Reviewer’s report

Title: Broad phenotypic spectrum in familial adenomatous polyposis; from early onset and severe phenotypes to late onset of attenuated polyposis with the first manifestation at age 72

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Reviewer: Janusz Limon

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Review
Nilbert M. at al.: „Broad phenotypic spectrum in familial adenomatous polyposis: from early onset and severe phenotypes to late onset attenuated polyposis with the first manifestation at age 72”

This is an interesting well written manuscript concerning a very important clinical and molecular aspects of the FAP syndrome, namely the relationship between the molecular findings and clinical features. The paper reviews the literature well. I have no objection of any importance.

This manuscript should be accepted after discretionary revisions.

Minor Essential Revision.

1. Abstract. Authors stated “10 novel APC gene mutation were identified in 11 individuals/families”. Do they revealed the novel mutations only?

2. Abstract. It is necessary do delete “u” in the description of the insertion c.1582_insGC.

3. Material and Methods. There is no information: how many patients were analysed?

4. Material and Methods. Authors used the MLPA technique – in which cases? Cases A37M and A40M?

5. Results. It is not clear for me: are the data regarding the mean age 32(14-72) related to the patients with mutation only?

6. Results. “two sisters with an insertion” - my suggestion is to add the code of these patients: A46- 1F and 2F (Tab. 1). The same with “woman without family history of colorectal polyps” - A30/F

7. Discussion. “two individuals with whole-gene APC deletions” – to add the code of these patients: A37M and A40M.