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: Joachim Teichmann

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The aim of study was the description of genotype – phenotype correlation concerning to extracolonic manifestation of familial adenomatous polyposis. The question is well defined by the author’s.

Patients: Therefore, 11 families were recruited. The APC – gene mutations were identified.


Results: Nilbert et co-workers present novel APC mutations. There is a link between mutations and extracolonic manifestations e.g. thyroid cancer and FAP. A second main message demonstrates the need to consider attenuated FAP also among elderly patients with colon cancer!!!

The presentation of data is performed perfectly in Table 1.

Ergo: The discussion included a lot of references, is well structured. The conclusions were ballanced. There were no limitations of study design.

Level of interest: An article of outstanding merit and interest 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'