Reviewer’s report

Title: Desmoid tumors complicating Familial Adenomatous Polyposis: a meta-analysis of the mutation spectrum of at-risk individuals

Version: 2 Date: 29 January 2015

Reviewer: Heather Hampel

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Nice manuscript addressing genotype-phenotype correlations between desmoids tumors and mutations in the APC gene. First study to normalize the findings by comparing to the frequency of all mutations in various regions of APC.

Major comment:
Why no mention of codons 1256-1303 throughout the manuscript when the table appears to show a relative risk of 2.0 for this region that is almost as statistically significant (if I'm reading the table right) as codons 543-713.

Regarding surveillance for desmoids, at what age would you begin annual abdominal ultrasound? How would you screen for extra-abdominal desmoids given the high frequency you found in this meta-analysis?

Minor comment:
I'm not sure if this is just a problem with the upload but there were multiple instances throughout the manuscript where two words appeared not to have a space between them (e.g. line 52 "mutationswith" and line 55 "ofAPC" and line 124 "51unique". I stopped keeping track because there were too many. Please check that this is not occurring in the original.

Tables/Figures are uploaded twice and there are formatting problems (Figure 2, y axis "Percentage of" is on top of the %es.)