Reviewer's report

**Title:** Diagnostic and Pathogenetic role of Cafe-au-lait macules in Nevoid Basal Cell Carcinoma Syndrome and other hereditary cancer syndromes

**Version:** 1  **Date:** 1 September 2012

**Reviewer:** Ketty Peris

**Reviewer's report:**

Minor essential revisions
- In Figure 1, one last column with signs and symptoms of internal organs is appreciated
- KCOT is already abbreviated when initially mentioned in Case presentation (page 3)
- In the context of previously reported PTCH1 mutations in NBCCS, how common is the PTCH1 germline mutation they found in their patients? It seems that description of this novel mutation is in press by the same authors. If this is the case it should be better specified in the discussion.
- In the conclusion section, it should be specified which are the major and minor criteria for the diagnosis of NBCCS

**Level of interest:** An article whose findings are important to those with closely related research interests

**Quality of written English:** Needs some language corrections before being published

**Declaration of competing interests:**

I have no competing interests.