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: 27 August 2012

Reviewer: Thierry van de Wetering

Reviewer's report:

Major Compulsory Revisions

The subject of the article is very interesting. As mentioned in the article no one has connected NBCCS with CALS, and this could be the first article.

Only to be sure if this could be connected successfully with each other, 1 patient is not enough. The frequency of CALS is quite high, while the NBCCS is lower. I would suggest too mark this article as a case report and mention in the discussion that in the future this connection will be investigated by your research team or by other people.

In the article is mentioned that the patient was misplaced in the group of NF-1. This shows the importance of setting-up different criteria to diagnosis which disease the patient has. In this case genetic analysis was not really necessary if the doctors would have held to the specific criteria of NF-1 and NBCCS. The genetic analyses gave an extra support what kind of disease the patient has. This extra support is not only concerning the diseases NF-1 or NBCCS but also on other hereditary cancer syndromes. The title suggests that also other hereditary cancer syndromes will be reported. No other patient with CALS and/or NBCCS was mentioned which could be related to other hereditary cancer syndromes, for example like McCune-Albright syndrome or Leopard Syndrome. I would suggest to change the title (for example delete the last part of the title).

In this report is written about a mutation in the PTCH1 gene and that molecular analysis was performed. Please tell us which type of analysis was used, for example RFLP, ASA-PCR, sequencing, RealTime-PCR, etc. Was only one molecular analysis used or more? How often was the blood taken and analyzed for verification of the mutation? Was this mutation analyzed on DNA or cDNA? In other words the section material and methods is not complete. If you have already an article where you describe the methods, please refer to.

Minor Essential Revisions

None

Level of interest: An article of outstanding merit and interest in its field

Quality of written English: Acceptable
Declaration of competing interests:

I declare that I have no competing interests