Reviewer's report

Title: 2 Novel Deletions of the Sterol 27-hydroxylase Gene in A Chinese Family with cerebrotendinous xanthomatosis

Version: 1 Date: 18 March 2011

Reviewer: Antonio Federico

Reviewer's report:

The manuscript “2 Novel Deletions of the Sterol 27-hydroxylase Gene in a Chinese Family with Cerebrotendinous Xanthomatosis” by Tian Di and Zhang Zaiqiang is interesting but not very well written. The use of a professional editor, to revise the numerous spelling mistakes and style is suggested.

The study reported a Chinese patient with a clinical and genetic diagnosis of CTX. The CTX patient carried two novel CYP27A1 gene deletions. I find that the experimental approach is reasonable but methods description can be improved.

I do have only minor comments or suggestions. All changes are essential for publication.

Minor Essential Revisions.
1. There are many bad spelling. Please write “Cerebrotendinous Xanthomatosis” initials in capital letters (always capital letters or always minuscule letters). Title: change “in A Chinese” to “in a Chinese”; Abstract, Objectives: Abstract, Conclusion: change “family members and desciding” to “family members and deciding”; Abstract, Results: change “family memebers” to “family members”. Introduction, line 3: change “9 Exons” to “9 exons”; figure 3 legend: change “were not availble” to “were not available”.
Please check for other errors.
2. Running title may be shortened for example to “2 Novel CYP27A1 Gene Deletions in a Chinese Family.”
3. Use the correct nomenclature for the mutations (c.73delG and c.369_375delGTACCCA) and add the result of these mutations (only for example: p.Trp26Cys). You can see it at http://www.hgvs.org/mutnomen/.
4. Figure 1 legend: re-order alphabetically from A to D.
5. Case presentation: add normal value for serum concentration of cholesterol, HDL and LDL. Have you measured cholestanol?
6. Genetic analysis, first line: informed consent should be obtained from the family members.
7. Genetic analysis, last line: Add State and City of Invitrogen
company.
8. Some corrections are necessary in the figure 2 legend to read better. Please consider to change to: “Electropherogram section showing exon 1 deletion (c.73delG)...”.
9. Figure 2 legend: the last phrase should be written better or at least differently.
10. The heading Discussion is incorrect: change “Dicussion” to “Discussion”.
11. Discussion: “As her father is the carrier….from her mother”- this sentence could be erased.
12. Discussion, last line: “we are grateful…us technique” - add this sentence in acknowledgement.