Reviewer's report

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

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Reviewer: David Wallon

Reviewer's report:

This manuscript describes a classical case of a rare genetical disease: cerebrotendinous xanthomatosis. It reports two novel mutations in CYP27A1 gene.

However several points are not suitable

1°) The main objective of this Ms is to prove the interest of sequencing the CYP27A1 gene in asymptomatic family members. However, the member most at risk, the brother noted as II3, could not be genetically tested. In fact he was the only one who could illustrate your diagnosis approach. The other asymptomatic members of this family only show the recessive characteristic of CTX.

2°) I do not think that spindle-shaped lipid crystal clefts in xanthomas are pathognomonic lesions for CTX (familial hypercholesterolemia).

Minor Essential Revisions

1°) what about serum cholestanol analysis ? an other way for screening CTX in a family..

2°) I am not sure that figure 2 is useful

3°) The Ms needs help with English and a second attentive reading. Different mistakes or misspellings can be found all along the Ms. To illustrate this point:

  Eg. (1) Abstract (Result): the family memebers
  Eg. (2) « She became retardated » => she presented cognitive impairements
  Eg. (3) figure 1 “leisions”

...

Level of interest: An article of limited interest

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

Statistical review: Yes, and I have assessed the statistics in my report.
Declaration of competing interests:

I declare that I have no competing interests.