Reviewer's report

Title: The T1048I mutation in ATP7A gene causes an unusual Menkes disease presentation

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Reviewer: Lisbeth B Møller

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The paper by Gregoria Leon-Garcia et al., describe a new mutation the p.T104I mutation in the ATP7A in a patient with Menkes disease. The paper is interesting because it shows that treatment might be useful.

I have the following minor comments

1) The language might not be 100% correct concerning English Grammar. It is easy to understand but for example is is correct to say:
   In abstract, case presentation. The treatment of the patient with copper-histidinate began late, until the age of 18 months (until?) …and with the conserved of some partial ATP7A activity (conserved??). In conclusion: therefore it is essential a detailed newborn screening as a mechanism for early detection and treatment of Menkes disease…should it be: therefore a detailed newborn screening is essential for early detection and treatment of Menkes disease
   It would be nice if a person with English as first language could read and correct the paper.

2) Nomenclature missense mutations are described by a p. before the mutation, please correct

3) Please include the mutation at the c.level also c.XXXXT>C

4) Figure 1 is very compact. In Figure C I don’t think it is necessary to include the mother. And if included the nucleotide should not be an T but C/T (heterozygous)

Major comments

5) It is interesting if the mutated protein still contains activity, and of cause the paper would improve if this was proven by complementation in yeast. It is still only speculative.
   In the result it is written: Despite the fact of (that?) T1048 does not form H-bonds with the ATP . How is this known??

6) What is the IQ of this patient is he going to a special school? Does he have
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

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests' below.