Reviewer's report

Title: Three novel mutations in the ATP7B gene of unrelated Vietnamese patients with Wilson disease

Version: 1 Date: 26 Mar 2018

Reviewer: Panieh Terraf

Reviewer's report:

The case report describes three patients from three unrelated Vietnamese families with Wilson's disease. They report 5 mutations in the ATP7B gene, which according to authors three are novel.

Comments and Suggestions:

* Authors present His251Alafs*19 in exon 2 as a novel mutation. However, this mutation has previously been reported in a south Asian family. Please refer to gnomad.broadinstitute.org

* I would suggest for authors to include pictures of patient's eyes pointing out the Kayser-Fleischer rings.

* The normal range for serum ceruloplasmin is between 20-35 and not <20 as stated by authors. So please revise throughout paper and in table 1.

* It would be best if authors changed the normal 24h urinary copper from <100 to <60 since this is more commonly reported. Please change throughout manuscript and in table 1.

* PT stands for Prothrombin Time. Please revise throughout paper and change "prothromlin time" to "prothrombin time".

* In clinical features, authors should explain the term "insensible".
* The manuscript needs English proofreading. There are many grammatical errors throughout the paper.

* In line 5 of page 2, please change "unrealed" to "unrelated"

* In line 3 of page 4 authors might consider revising the sentence "will be helpful in developing effective treatment" to "will be helpful in efficiently diagnosing Wilson's disease and early therapeutic intervention for patients."

* Line 1, page 5 "QIAamp DNA Blood Mini preparation kit" should be changed to "QIAamp DNA Blood Midi preparation kit"

* In Line 21, page 6 the sentence is not very clear. Consider changing to "The genetic analysis in his mother showed that she had a heterozygous mutation in exon 14 (Phe1026Tyr) but did not have the mutation in intron 12".

* The mutation in intron 12 is known to affect the correct splicing of ATP7B mRNA not the incorrect splicing. Please revise in line 14 of page 7.

* Line 23 and 24 of page 7 are repeated in line 14 and 15 of the same page. There is no significance for that. Please only include one or the other.

* I think it would be good to change the phrase "located in the shear zone, which is affect mRNA splicing" of line 9 in page 8 to "located in the splice site, which can affect mRNA splicing".

* Line 16, page 9 the author's name is misspelled. Please change to Kalach et al.

* Line 17, page 9 please change to "acute liver failure".
Line 17, page 9 the sentence is vague and unclear. Please consider changing to "Yi et al. (2012) who established neural and hepatic differentiation platforms of patient-derived induced pluripotent stem cells, showed that liver and brain are the organs most affected"

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.
Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.
Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.
Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.
I am able to assess the statistics

Quality of written English
Please indicate the quality of language in the manuscript:
Not suitable for publication unless extensively edited

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