Reviewer’s report

Title: Identification of compound heterozygous patients with Primary Hyperoxaluria Type 1: Clinical Evaluations and In Silico Investigations

Version: 0 Date: 01 Jan 2017

Reviewer: Neveen Soliman

Reviewer's report:

An interesting study by Kanoun and colleagues to report the identification of AGXT gene compound heterozygous mutations in two PH1 non-consanguineous families. They also come across the clinical characterization of the four affected patients from the above unrelated two families. That being said the authors need to elaborate more on clinical phenotyping of the reported patients.

Below are further comments/concerns to be addressed:

- The article needs thorough English linguistic review

- Abstract: Page 4 Line 52 "Two patients were compound heterozygous for the c.731T>C, c.32C>T, c.1020A>G and c.33_34insC and presented the disease with recurrent urinary tract infection" better changed to "with clinical presentation of " or "and presented clinically with". This applies for many parts of the manuscript.

- Clinical Severity evaluation of the compound heterozygous studied patients. (This is supposed to be heading not a sentence so no dot at the end of it). This section needs more elaboration as to deep clinical phenotyping of the compound heterozygous studied patients. Linguistic editing is strongly recommended in this part of the manuscript.

- More detailed information are needed as to the value of in silico investigation in the current study and its link to clinical phenotypes of the studied patients/families.
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.

Unable to assess

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.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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