Reviewer's report

Title: A Chinese patient with 11β-hydroxylase deficiency due to novel compound heterozygous mutation in CYP11B1 gene: a case report

Version: 1 Date: 01 Feb 2018

Reviewer: Maja Stojiljkovic

Reviewer's report:

The authors described one patient with 11β-hydroxylase deficiency and reported two novel mutations in the CYP11B1 gene. Functional studies assessing effect of novel mutations have not been performed.

Surprisingly, the authors are incorrectly using basic genetic terms. The term "missing mutation" is used instead "frameshift mutation" and the term "codon" was used where should be said "nucleotide". Also, the term "compound heterozygosity" is incorrectly used in the title and through the manuscript.

Line 139 - It is stated that primers covering RECQL4 gene were used to sequence CYP11B1 gene.

How primers were designed? The accession number of the sequence used to design primers must be provided. How have you ensured that primers do not amplify highly homologous aldosterone synthase CYP11B2 gene?

Line 154 - "…we also identified two polymorphic loci in the patient's CYP11B1 gene" - please specify what kind of polymorphic loci… or silent genetic variants have you identified.

Line 164 - It is incorrect to say "A novel compound heterozygous mutation was found", it should be stated: "the patient is a compound heterozygous"… "two novel mutations in CYP11B1 gene were found"

Please carefully consult HGVS nomenclature in order to correctly describe novel mutations at the protein level (regarding premature termination and abolishing of stop codon).

Figure 3 (B) - it is advisable to repeat DNA sequencing and obtain better picture.

Figure 4 should be moved to Supplementary material.
Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

No

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.

Not relevant to this manuscript

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

Not suitable for publication unless extensively edited

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