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: 27 May 2018

Reviewer: Mirjana Kocova

Reviewer's report:

Manuscript titled "A Chinese patient with 11β-hydroxylase deficiency due to novel compound heterozygous mutation in CYP11B1 gene: a case report" describes a poorly treated young patient with 11β-hydroxylase deficiency, who was subsequently molecularly confirmed to carry novel mutations as compound heterozygote. After the diagnosis the therapy was appropriately changed however iatrogenic Cushing syndrome was additionally diagnosed.

Generally, the text should be shortened, written in correct English, focused upon the main findings, especially when the laboratory results are presented (Table gives enough information)

Case presentation: What was happening between 12 months and 6 years of age? The growth should be given in SDS (>97%o is very vague determination), and this child was extremely tall at 6 years, but short as an adult, present it in SDS scores. Several statements in the text are confusing, e.g. no data are available on bone age, so advanced bone age should be considered assumption and addressed in discussion only, not in the case report.

Since all laboratory values are given in the table, referring in the text should be shortened to the most important values.

Both of the novel mutations are partial gene deletions with a different transcriptional effect, thus, using the term "missing mutation" is incorrect and should be avoided.

English language corrections by a native speaker are necessary for the clarity and precision of the statements.

Specific comment

The last sentence in the introduction should be rephrased:

Suggestion: In this study, we reviewed the clinical data of a 19-year-old Chinese patient with classic 11β-OHD and conducted a genetic analysis confirming a novel 75 compound heterozygous mutation in the CYP11B1 gene
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?
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Yes

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Quality of written English
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Not suitable for publication unless extensively edited

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