**Reviewer’s report**

**Title:** Splicing analysis of CYP11B1 mutation in a family affected with 11β-hydroxylase deficiency: case report

**Version:** 0  **Date:** 05 Apr 2016

**Reviewer:** Emma Webb

**Reviewer's report:**

General comments:

This paper describes the phenotype of two siblings with 11ß-OHD. The phenotype is that of classic 11ß-OHD. They have performed functional studies which give more information on the pathogenic mechanism of disease in the IVS7+IG>A mutations.

In general there are many grammatical errors (some highlighted below) and the paper does not always read fluently. The phenotype description could be shortened.

In detail

**Abstract:**

Line 31 A 46,XX sister-amend to read A 46,XX female.

Line 32: her 46,XY brother presented with sexual precocity-change to precocious puberty

The abstract does not flow that well. Move sentence regarding mutation analysis (line 38) to after the line on Sanger sequencing (line 34).
Conclusion: Unexciting—could be improved

Main paper

Line 58—sexual precocity—change to precocious puberty

Line 62—cosyntropin—ACTH stimulation

Line 78—At that time—what time

Lines 83–87—put all information into a table with baseline and stimulated values. This will be much clearer and reduce length of text.

Line 86—change levels to concentrations (on all occasions)

Line 91—after molecularly confirmed diagnosis—Treatment should be started on the basis of no response to ACTH stimulation

Line 95—what is masculinization (precocious puberty). Was hypertension not identified when the patient presented as opposed to the patient presenting with hypertension.

Line 99—height gain accelerated—describe as change in growth velocity centile/height SDS.

Was urine steroid profile performed?
Line 168-change an to a

Lines 169-171-repeat of introduction

Line 177-at 2 years old not in infancy

Lines 209-211-rewrite as message not clear but would be the main point of getting this data published.

**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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