Reviewer’s report

Title: Long-term follow-up in a Chinese child with congenital lipoid adrenal hyperplasia due to a StAR gene mutation

Version: 1 Date: 07 Sep 2018

Reviewer: Lin Lu

Reviewer’s report:

Dear Author:

The paper "Case Report: Long-term follow-up in a Chinese child with congenital lipoid adrenal hyperplasia due to a StAR gene mutation" has been reviewed. There are rare reports for the long term follow-up of CLAH. However, there are many unclear details that the author should clarify.

1. Page 2, 45th line, and page 3, 48th line, should the "OMIN" be a mistake? OMIM is well known, but not "OMIN".

2. Page 3, 51th line, there is a sentence said "but there are only 17 confirmed cases of CLAH in Chinese patients". We believe it should be presented as published or reported cases, not confirmed cases since some cases didn't get published. Pls be careful for the described words you used.

3. Page 3, 53-54th line, there is a sentence "All affected individuals are phenotypically female regardless of their gonadal sex [1]." Same to the above opinion, the author should be careful for the descriptions. As classic CLAH, the phenotype must be female whatever the sex chromosome is. But for nonclassic CLAH, the phenotype may have variations. Pls modify this sentence properly.

4. Page 3 64-66th line, the sentence "However, her compliance was poor, and she was not followed up regularly. There were several episodes of adrenal crisis before 13 years of age. No adverse events were recorded". Pls give detailed information about whether poor compliance is also indicated poor treatment compliance.

5. Page 3 68th line, The sentence "The patient was alert", What does alert mean? Pls give detailed information or modify the description properly.

6. Page 3 68th line, The sentence "her height, weight and body mass index (BMI) were 148.7 m (-1.3 SDS), 35.3 kg (-1.5 SDS) and 15.96 kg/m2 ". Pls correct the height, the meter is not the correct unit.

7. Page 3 71th line, the sentence "Normal external female genitalia were observed with a normal clitoris and vaginal orifice." Pls delete the word "normal" since the patient is a 46 XY DSD.
8. In addition to the correction of the above opinion. In this paper the clinical manifestation, physical examination were listed separately. This kind of description was a bit messy. Pls give the information chronologically. The important lab value also should be given in the context in addition to the tables of lab data.

9. The condition of birth was recommended to list before the medical history chronologically.

10. Although the gene sequencing were done in the patient and her father, it should also be done to her two sisters to confirm whether there is carrier.

11. The significant point is a small adrenal for the patients. But the discussion about that is very limited in the paper. Could the author search some literatures to figure out whether there is reasonable speculation.

12. Pls add the treatment details in the table 1.

13. Since the cases of CLAH usually has hypogonadism, the bone age was behind the actual age for most of the cases. But this patient has the age matched bone age. Could the author give some pictures to show the bone age?

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.

No

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

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