Reviewer's report

Title: A novel mutation of the StAR gene with congenital adrenal hyperplasia and its association with heterochromia iridis: a case report

Version: 0 Date: 29 Aug 2019

Reviewer: Lin Lu

Reviewer's report:

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This article describes a rare congenital adrenal hyperplasia caused by StAR gene deficiency, and a combination of heterochromia iridis. This is an interesting case, two rare diseases coexisted. But there are some major revisions needed to be modified.

1. In the description of the manifestation of the case, please add the level of renin together with aldosterone.

2. There are infrequent descriptions of endocrine-related symptoms in the article, which were important to differentiation diagnosis. For example, the gender of the external genitalia at birth, and the test results of gonadotropin, estradiol and testosterone should be supplied in the initial examination.

3. It is a great pity that the father and sister of the patient did not have a genetic test. Could the author try to complete the genetic test of families?

4. The special feature of this patient is that there was a new mutation in the StAR gene and the patient had normal puberty and regular periods. It is suggested that the content of the discussion section will be well organized, focusing on the functional changes caused by new mutations in the StAR gene, and secondly in the rare manifestations of heterochromia iridis combined with StAR gene defects.

5. There is a sentence, "If confirmed in additional cases, discolorization of the iris might be a useful phenotypical feature aiding in the diagnosis of severe forms of adrenal failure." The sentence needs to be carefully revised, and it is difficult to draw conclusions based on speculation alone.

6. Since the patient has a consanguineous parents, the details of the family history need to be given in detail. If there is similar manifestation or other patients in the family.
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?
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Unable to assess

Are the conclusions drawn adequately supported by the data shown?
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No

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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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