Reviewer's report


Version: 2 Date: 07 Feb 2018

Reviewer: Patricia Canto

Reviewer's report:

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This is a case presentation, in which the authors described an 11OHD patient with a precocious pseudopuberty and hypokalemia hypertension, who harbored a chimeric CYP11B2/CYP11B1.

This is a novel case; only I have some comments:

It is important that the authors put the electronic direction or a reference in relation How they performed the bioinformatics and molecular dynamics simulation of the mutant protein. Besides, in discussion section, the authors not discuss about their functional finding of the mutant protein (p.L340P) and how the combination of the p.L340P and a chimeric CYP11B2/CYP11B1 gene could influence the genotype of the proband, since the the chimera also was find in his mother, uncle and grand father (I know this is an autosomal recessive disease) and these relatives not saw a diseases o might be, they had some kind of disturbed in 17OHP, androstenedione, ACTH, etc, or this chimera don´t have impact in their disease?

This is important since the authors refer that the both the chimera and p.L340P contributors to pathogenicity.
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.
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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Please indicate the quality of language in the manuscript:
Acceptable

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