Reviewer's report

Title: A Novel Germline ARMC5 Mutation in a Patient with Bilateral Macronodular Adrenal Hyperplasia: a case report

Version: 1 Date: 20 Feb 2018

Reviewer: Sulman Basit

Reviewer's report:
Comments to the Author

Subject: A Novel Germline ARMC5 Mutation in a Patient with Bilateral Macronodular Adrenal Hyperplasia: a case report

Authors have reported a novel sequence variant in the ARMC5 gene. Generally the manuscript is well written. Following minor corrections/suggestions may be incorporated to facilitate acceptance of the manuscript for publication.

DNA extraction and sample collection from the tumor tissue has not been described. Benign tissue from tumor containing organ may contain un-mutated genome. What procedure was used to avoid this?

I would suggest Sanger sequencing of all exons of ARMC5 gene in DNA isolated from carefully selected tumor. Please note that ARMC5 is a small gene with only 4 exons.

MAH in this case is corticotropin dependent or corticotropin independent? Please elaborate.

Several typographic errors are present throughout the text.
For example;
* page 6, line 1, change "wide-type" to "wild-type".
* Page 9, last paragraph, 4th line, change "AMC5" to "ARMC5".

The authors need to show the genomic position of the variant and also give the frequency of the variant in normal population from ExAc/gnomAD database.
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

Quality of written English
Please indicate the quality of language in the manuscript:
Acceptable

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