Author’s response to reviews

Title: Case Report: A synonymous VHL mutation (c.414A>G, p.Pro138Pro) causes pathogenic familial hemangioblastoma through dysregulated splicing

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Author’s response to reviews:

Assistant Editor Comments:

1. Author names

Please provide the full names of each of the authors on the title page of your manuscript and in the submission system.

Please note that the order when referring to an author in the submission system and on the title page of your manuscript should be [first name][middle name, if applicable][last name].

Au: We confirm the accuracy of the names.

2. Consent to Participate

In your “Ethical approval and consent to participate” section, please clarify whether informed consent to participate, written or verbal, was obtained from all of the participants in the study and clearly state this in your manuscript.
Please note that in the case of minors, which refers to individuals younger than the age of 16, consent to participate must be obtained from their parents or legal guardians. As such, we ask that in your “Ethics approval and consent to participate” section, to clarify whether informed consent to participate, written or verbal, was obtained from the parents or legal guardians of any participant under the age of 16 and clearly state this in your manuscript.

If consent was verbal, please state the reason and whether the ethics committee approved this procedure. If the need for consent was waived by an IRB or is deemed unnecessary according to national regulations, please clearly state this, including the name of the IRB or a reference to the relevant legislation.

Au: We have revised the statement to the following sentences: “Written informed consent to participate was obtained from all of the participants in the study. Written informed consent to participate was obtained from the parents of the participants under the age of 16.” (Declarations, line 227-229, page 11-12)

3. Consent for publication

Currently, the statement in your “Consent for publication” section of your declarations is incorrect. Consent for publication refers to consent for the publication of identifying images or other personal or clinical details of participants that compromise anonymity.

As such, we ask that in your “Consent for publication” section, to please clarify whether written informed consent for publication of clinical details and/or clinical images was obtained from the all of the participants.

Please note that in the case of minors, which refers to individuals younger than the age of 18, consent for publication must be obtained from their parents or legal guardians. As such, we ask that in your “Consent for publication” section, to please clarify whether written informed consent for publication of clinical details and/or clinical images was obtained from the parents or legal guardians of any participant under the age of 18.

Au: We have revised the statement to the following sentences: “Written informed consent for publication of clinical details and clinical images was obtained from the all of the participants. Written informed consent for publication of clinical details and clinical images was obtained from the parents of any participant under the age of 18.” (Declarations, line 232-234, page 12)

4. Availability of data and materials

a) Please amend the statement in the section: “The data and materials generated and analyzed during the current study are not publically available (because the data and materials relate to human research participants, it may not be appropriate to make them publically available) but are available upon reasonable request with fulfillment of Materials Transfer Agreement and in a format compliant with Health Insurance Portability and Accountability Act (HIPAA).” to "The
datasets generated and analysed during the current study are not publicly available because it is possible that individual privacy could be compromised but are available upon reasonable request with fulfillment of Materials Transfer Agreement and in a format compliant with Health Insurance Portability and Accountability Act (HIPAA).

b) Please include further details in the Availability of data and materials section of your manuscript in regard to who should be contacted and their contact details or where to apply for permissions to obtain access to the raw data.

Au: a) Change was made as instruction (Declarations, line 237-240, page 12)

b) The following sentence was added “To request the datasets, please contact the corresponding authors (K.H. khal达尔@nd.edu or X.L. xlu@nd.edu).” (Declarations, line 240-241, page 12)

5. Authors’ contributions

Please consider the list of authors as it currently stands with reference to our guidelines regarding qualification for authorship (http://www.biomedcentral.com/submissions/editorial-policies#authorship).

Currently, the contributions of author CZ does not automatically qualify them for authorship. In the section “Authors’ contributions”, please provide further clarifications on their contributions, and see our guidelines for authorship below.

An 'author' is generally considered to be someone who has made substantive intellectual contributions to a published study. Authors are expected to fulfil the criteria below (adapted from McNutt et al., Proceedings of the National Academy of Sciences, Feb 2018, 201715374; DOI: 10.1073/pnas.1715374115; licensed under CC BY 4.0):

Each author is expected to have made substantial contributions to the conception OR design of the work; OR the acquisition, analysis, OR interpretation of data; OR the creation of new software used in the work; OR have drafted the work or substantively revised it AND to have approved the submitted version (and any substantially modified version that involves the author's contribution to the study); AND to have agreed both to be personally accountable for the author's own contributions and to ensure that questions related to the accuracy or integrity of any part of the work, even ones in which the author was not personally involved, are appropriately investigated, resolved, and the resolution documented in the literature.

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Au: We are sorry not to specify CZ’s role more accurately in the previous manuscript. In the revised manuscript, we modified this sentence to justify the authorship: “FL, BC, MSA, CZ, KH and XL drafted the manuscript and substantively revised it.” (Authors' contributions, line 262-263, page 13)

6. Figure 3

We note that you have included an in-text citation to “Fig. 3C” and “Fig. 3D” however these figures are absent from your manuscript. If you wish to include these citations, please upload the figures into your manuscript and into the file inventory. However, if you do not wish to include these figures in your manuscript, please remove the in-text citations corresponding to “Fig. 3C” and “Fig. 3D” on page 10 of your manuscript.

Au: Sorry about that. We have removed the in-text citations corresponding to “Fig. 3C” and “Fig. 3D” on page 10.

7. Remove attachments

Please remove the additional file entitled ‘CARE form.pdf’ from your manuscript and from the file inventory as it is no longer needed at this stage.

Au: We followed the instruction to exclude the ‘CARE form.pdf’ in the revision submission.

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Au: We followed the instructions.