Author’s response to reviews

Title: A novel TAB2 nonsense mutation (p.S149X) causing autosomal dominant congenital heart defects: a case report of a Chinese family

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Author’s response to reviews:

Dear Editors,

Many thanks for handling our manuscript. We appreciate the editor’s suggestion and the following is our revision with respect to the comments.

Major concerns of the editor:

1. Please include an “Abbreviations” section after the “Conclusions” section of your manuscript where you list all of the abbreviations used in the manuscript. All abbreviations should still be defined in the text at first use.
   Answer:
   An “Abbreviations” section was added after the “Discussion and conclusions” section. All abbreviations were defined in the text at first use.

2. In the consent to publish section, it is mentioned that the patients provided "signed consents for the
permission of the anonymous use of their clinical information and sequencing data”, however, much of the data included in the manuscript would make this not anonymous. Consent for publication refers to consent for the publication of identifying images or other personal or clinical details of participants that compromise anonymity. Please clarify if written informed consent was obtained for this.

Answer:
The adult participants were reinterviewed. They provided signed consents for the permission of using their clinical information and sequencing data for research purposes and/or scientific publications. The “Consent for publication” section was rewritten as followings:
“The enrolled adults provided signed consents for the permission of using their clinical information and sequencing data for research purposes and/or scientific publications. For the participants under 16 years old, written informed consent was obtained from their parents. For the participant died at adult stage (I:2), written informed consent was obtained from her daughters.”

3. Please clarify in the “Consent to publish” section if consent was obtained from the next of kin in cases where the patient died.

Answer:
For the participant died at adult stage (I:2), written informed consent was obtained from her daughters. The “Consent for publication” section was rewritten as followings:
“The enrolled adults provided signed consents for the permission of using their clinical information and sequencing data for research purposes and/or scientific publications. For the participants under 16 years old, written informed consent was obtained from their parents. For the participant died at adult stage (I:2), written informed consent was obtained from her daughters.”

4. Please remove the CARE checklist and Declarations file from the manuscript.

Answer:
The CARE checklist and Declarations files were removed from the manuscript.

5. At this stage, please upload your manuscript as a single, final, clean version that does not contain any tracked changes, comments, highlights, strikethroughs or text in different colours. All relevant tables/figures/additional files should also be clean versions. Figures (and additional files) should remain uploaded as separate files. Please ensure that all figures, tables and additional-supplementary files are cited within the text.

Answer:
The manuscript, the relevant table and figures were uploaded as clean versions and separate files. The figures and tables are cited within the text.

Thank you very much for your attention and consideration. If any parts of the manuscript need further improvement, please inform us for revision. I'm looking forward to hearing from you soon.

Sincerely yours,

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