

Responses to reviewers

Reviewer #1:

Scientific Quality: Grade A (Excellent)

Language Quality: Grade B (Minor language polishing)

Conclusion: Accept (General priority)

Specific Comments to Authors: Report: Townes-Brocks syndrome with adult renal impairment in a Chinese family: A case report This is an interesting case report, however, the manuscript would benefit from the following corrections/amendments:

1. Please mention the genetic findings in the Abstract.

Response: This information was including in "CASE SUMMARY".

2. Please provide OMIM number for each malformation mentioned in the text.

Response: This information was added in the text, Page 2, INTRODUCTION

3. Methods: Please give the detail of Whole exome sequencing and filtration strategy.

Response: We thanks for the constructive suggestion. As the performance of WES is very regular, a reference is added.

4. Please give the allele frequency of the detected variant.

Response: This variant is not included in the known databases. Therefore, the frequency is unknown.

5. There is discrepancy in the report. In the Abstract and Results, the author mention '....a heterozygous variant of SALL1..' where as in legend to figure '.....Figure 2 Identification of the compound heterozygous variants in the family....'.

Response: We are sorry for the mistake. This is revised.

6. Fig. 2. Please give the Sanger of reference /unaffected subjects.

Response: The figure was revised.

7. Legend to figure 2. The statement is not correct '.....Affected family members are denoted in black.'

Response: The text was revised.

8. Pedigree. Please mention who underwent molecular study.

Response: This information was added.

Reviewer #2:

Scientific Quality: Grade B (Very good)

Language Quality: Grade A (Priority publishing)

Conclusion: Accept (High priority)

Specific Comments to Authors: Overall, this manuscript presents innovative ideas clearly and effectively. The structure is well-organized, and the writing is clear and concise. The author has done an excellent job of explaining a complex technical process in an easy-to-understand way. However, to improve the manuscript, the author should ensure that the references cited in the introduction and related work section are thoroughly addressed in the reference section. Additionally, the introduction should provide an extended version of the abstract, with

elaboration on the key points and supportive ideas and references. Lastly, the conclusion section needs revision to provide a more insightful and comprehensive summary of the manuscript. Finally, the author should ensure that all references are properly formatted according to the relevant rules.

Response: We thanks for the constructive suggestion. The references have re-organized. The abstract and conclusion are also revised according the suggestion.