Author’s response to reviews

Title: Identification of two novel COL10A1 heterozygous mutations in two Chinese pedigrees with Schmid-type metaphyseal chondrodysplasia

Authors:
Lingchi Kong (13122230023@163.com)
Li Shi (593635386@qq.com)
Wenbo Wang (259525450@qq.com)
Rongtai Zuo (zuorongtai@163.com)
Mengwei Wang (1617704772@qq.com)
Qinglin Kang (orthokang@163.com)

Version: 3 Date: 04 Dec 2019

Author’s response to reviews:

Dear Editor,

Thanks for your letter dated December 4th. We were pleased to receive a minor revision decision, which enabled us to further improve the manuscript. We appreciate you and reviewers for the time and efforts that you have put into reviewing the previous version of the manuscript.

Appended to this letter are our point-by-point responses to the comments raised by editor and reviewer. The comments were reproduced and our responses were given directly afterward. We hope that this version is acceptable for publication in “BMC Medical Genetics”. Should you have any question, please feel free to contact me. Thank you!

Yours sincerely,

Qing-Lin Kang

E-mail: orthokang@163.com
Assistant Editor Comments:

1. Consent for publication

Currently, the statement in your “Consent for publication” section of your declarations is incorrect. 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.

Response: Thanks for your comment. In this section, we have declared that written informed consent for publication of clinical details and clinical images was obtained from all of the adult participates and the legal guardians of any participant under the age of 18 (Line 324 to 325, Page 15).

2. Role of funding body

Please provide more details regarding the specific role of the funding body in your study. If the funding body only provided the financial means to allow the authors to carry out the study, please state “The funding body played no role in the design of the study and collection, analysis, and interpretation of data and in writing the manuscript” and include this statement in the Funding section of your manuscript.

However, if the funding body took part in the design of the study and collection, analysis, and interpretation of data, and the writing of the manuscript, this must be stated in the Competing Interests section of your manuscript.

Response: Thanks for your comment. In this section, we have explained that the funding body played no role in the design of the study and collection, analysis, and interpretation of data and in writing the manuscript (Line 310 to 312, Page 15).

3. Availability of data and materials

Please note that BMC mandates data deposition for the following:

• Protein sequences
• DNA and RNA sequences
• DNA and RNA sequencing data
• Genetic polymorphisms
• Linked genotype and phenotype data
• Macromolecular structure
• Microarray data (must be MIAME compliant)
• Crystallographic data for small molecules

As such, we ask that you please provide the relevant accession numbers if your data has been deposited into a data bank or the appropriate web links if the data has been uploaded into a repository. If the data has not yet been deposited, please do so and provide the relevant information needed to access it in the ‘Availability of data and materials’ section in your manuscript. Please ensure all data is already released and that any accession numbers/web links are in the ‘Availability of Data and Materials’ in their final form, and that all links and accessions allow access to public data.

Your data availability statement can take the following form: “The datasets generated and/or analysed during the current study are available in the [NAME] repository, [PERSISTENT WEB LINK OR ACCESSION NUMBER TO DATASETS]”.

Response: Thanks for your guidance. As for availability of data and materials, necessary data relevant to this study are included in the article and supplementary files, and the other data we consider may be used for further study. When all work is finished and published, all relevant data will be available in the BMC public datasets. If any researchers would like to access it before turning to public, they could contact the corresponding author by e-mail, and we will be pleased to provide it.

Therefore, we declared that “all data generated and analyzed during the current study are available from the corresponding author on reasonable request” in this section (Line 315 to 316, Page 15).

4. Supplementary files

As per our submission guidelines, we also require that all additional files be individually cited in the text in sequence in a manuscript. Your supplementary files are currently not cited in your manuscript. Please ensure that they are appropriately cited within the main body of text in your manuscript. Any items which are not cited may be deleted by our production department upon publication.
Response: Thanks for your kind comment. All additional files have been individually cited in the manuscript in sequence (Line 69, Page 4; Line 103, Page 5; Line 120, Page 6; Line 145, Page 7; Line 234, Page 11).

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. Should you wish to respond to these revision requests, please put your responses to the reviewers'/editors’ comments in the Response to Reviewers box in Editorial Manager. Please do not upload a separate letter.

Response: We have well prepared these materials for publication according to the requirements. Thank you!

Reviewer reports:

Franco Cammarata-Scalisi (Reviewer 2):

I only have one comment to modify the term "irregular dominance" by incomplete dominance.

Response: Thanks for this comment. We have replaced the term “irregular dominance” by “incomplete dominance” throughout the manuscript.