List of Responses

Dear Editors and Reviewers:

Thank you for your letter and for the reviewers' comments concerning our manuscript entitled "Geleophysic dysplasia caused by a mutation in FBN1: a case report and review of the literature" (ID: 62415). Those comments are all valuable and very helpful for revising and improving our paper, as well as the important guiding significance to our researches. We have studied comments carefully and have made correction which we hope meet with approval. The main corrections in the paper and the responds to the reviewer's comments are as flowing:

Responds to the reviewer's comments:

Reviewer #1:

1. Response to comment: Mention the OMIM number with each malformation described in the text.

Response: We have mention the OMIM number with each malformation described in the text according to the Reviewer's comments.

2. Response to comment: Omit the mention of acromicric dysplasia (AD) and Weill-Marchesani syndrome (WMS) in the introduction and Abstract.

Response: We have omit the mention of AD and WMS in the "Introduction" and "Abstract" section according to the Reviewer's comments. We introduce AD and WMS in the "Discussion" section instead.

3. Response to comment: Report the origin, basic demographics and

ethnicity of the subject

Response: We have described that of the subject in the “Personal and family history” section according to the Reviewer’s comments.

4. Response to comment: Mention the parental parameters/symptoms and consanguinity.

Response: We have mentioned the parental parameters/symptoms and consanguinity in the “Whole-exome sequencing” section according to the Reviewer’s comments.

5. Response to comment: Table 1. There are many typos in the table. Almost all the terms used in first column are problematic.

Response: We are very sorry for our negligence of that, and have made correction in Table 1 according to the Reviewer’s comments.

6. Response to comment: It is not clear who is Paitent 1 and Patient 2 in Table 1.

Response: We have mention the original reports of Paitent 1 and Patient 2 in Table 1 according to the Reviewer’s suggestion.

7. Response to comment: It is not clear what is the relationship of respiratory tract infections (RRTIs) with GD. The authors may like to put light on this.

Response: The reviewer’ statement is correct in that. However, we wish to research the relationship of RRTIs with GD in the near future and will publish it at a later time.

8. Response to comment: The mutation detected in this patient is already reported，the authors need to explicitly mention this in the text.

Response: As the Reviewer suggested, we have mentioned that at the line 4-5 of the third paragraph in the "Discussion" section.

And special thanks to you for your good comments.

Reviewer #2:

If the relatives of this child have been investiged as well.

Response: We also underwent the whole exome sequencing of the child's consanguineous parents, and did not find the same mutation.

Science editor: The "Case Presentation" section was not written according to the Guidelines for Manuscript Preparation. Please re-write the "Case Presentation" section according to the Guidelines and Requirements for Manuscript Revision.

Response: We are very sorry for our negligence of that, and have re-write the "Case Presentation" section according to the Guidelines for Manuscript Preparation.

Other changes:

1. Title, the statements of "Geleophysic dysplasia caused by a mutation in *FBN1*: case report and review of the literature" were corrected as "Geleophysic dysplasia caused by a mutation in *FBN1*: a case report and review of the literature"

We tried our best to improve the manuscript and made some changes in the manuscript. These changes will not influence the content and framework of the paper. We appreciate for Editors/Reviewers' warm work earnestly, and hope that the correction will meet with approval.

Once again, thank you very much for your comments and suggestions.