Author's response to reviews

Title: Type I Gaucher disease with exophthalmos and pulmonary arteriovenous malformation

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Author's response to reviews:

Dear reviewer,
We really appreciate your comments on this article. The following are our replies to your suggestions. We have also revised our manuscript accordingly.

Comment 1: Modify the title since our patient may not be necessarily a "variant"
Reply: We agree with you and have deleted "a more clinical severe variant?". The title has changed to "Type I Gaucher disease with exophthalmos and pulmonary arteriovenous malformation".

Comment 2: Don't use the term "Gaucher patient".
Reply: This unjustified term, which was once used in the line 12 of background section, has been replaced by "patient with...... Gaucher disease".

Comment 3: Some references are less good choices.
Reply: We have revised our references with more appropriate and updated articles.

Comment 4: Discuss further about the relation between pulmonary hypertension/pulmonary AVM and ERT.
Reply: We have added more discussion about this issue. We think that pulmonary AVM and pulmonary hypertension are basically independent events. Hence these two different diseases should be discussed separately. As to her pulmonary hypertension, it is quite possible that it might be aggravated or even triggered by ERT. However, since there is no progression of pulmonary hypertension at echocardiographic follow-up (by measuring her TR velocity and pulmonary acceleration time) and she did feel much more energetic and vigorous after ERT, we still keep ERT for this patient so far. As to the pulmonary AVM, we prefer that it should have been present before ERT. It came to medical attention only when the disease course had been modified by ERT, which cleared the Gaucher cell in lung and thus unmasked the presence of AVM. Details are further described in the revised manuscript.

Comment 5: Phenotype-genotype correlation doesn't seem to be existent.
Reply: We are in complete accord with your opinion. Actually it is our belief that there must be some other epigenetic factors that influence the phenotypes among our index patients and her siblings. Perhaps we did not make it clear enough in previous manuscript to state our thought. We have revised this section.

Comment 6: Revise the compartmentation of discussion section.
Reply: We have revised the discussion section according to your suggestion.