
Peer Review File

Article information: <http://dx.doi.org/10.21037/tp-20-138>.

Review Comments

1.This manuscript reported a compelling case which found a novel mitochondrial DNA deletion of Pearson syndrome in an infant who presented with hepatomegaly, liver dysfunction, anaemia and lactic acidosis. Such a finding would no doubt fill our knowledge gap in Pearson syndrome, making the manuscript potentially publishable at Translational Paediatrics. However, there are some concerns.

The present title is obscure. It fails to point out the finding. And phrasing like "aetiology exploration" makes itself unconfident. Please refine the title and add vital information into it, including the novel deletion finding.

Reply: We have made modifications on the title accordingly.

2.The abstract is wordy. Besides, what's unique in this case is not well highlighted.

Reply: We have rewritten this part according to the Reviewer's suggestion, as marked in red in the revised version.

3.Introduction lacks essential background information, including known mtDNA deletion, the golden diagnosis criteria of Pearson syndrome, how many cases have reported, how many of those are among infants etc.

Reply: We have added these information into the introduction.

4.Draw a timeline to outline the whole process. Make sure it stands alone.

Reply: The whole process and laboratory indices over time in the patient were showed in the table 1.

5.Ease dosage is required for a medication.

Reply: We added dosage in the Case presentation.

6.Add one separate paragraph to discuss both strengths and limitations about this case report.

Reply: We have added a paragraph to discuss both strengths and limitations about this case report.

7.The take-away lesson is unclear. It is not just about that it enriches the variant spectrum but also how such findings indicate for clinical practice and potential hypothesis for the aetiology of Pearson syndrome.

Reply: We have rewritten the Conclusion according to the Reviewer's suggestion. Revised portion have marked in red in the paper.

8.Title: replace 'a patient' with 'an infant'

Reply: We have made modifications on the title accordingly.

9.Abstract: add information regarding the finding of the novel deletion of mitochondrial in this case.

Reply: We have modified this part according to the Reviewer's suggestion, as marked in red in the revised version (see Page 2, line 24 to 26).

10.Introduction: add information regarding how this case report differs from the previously reported 52 cases.

Reply: We have modified our text as advised (see Page 3, line 56 to 58).

11.Figure: draw a timeline that outlines the key information in a time-based manner. You could find an example of a timeline from our sister journal TLCR: <http://tlcr.amegroups.com/article/view/38743/html>

Reply: We added a timeline of the patient. (See Page 13, Figure legend, Figure 2).

12.One last concern, in the timeline, please further add what's found in addition to what's done.

Reply: We have modified this part according to the Reviewer's suggestion (See Page 13, Figure legend, Figure 2).