

Response to Reviewers

Reviewer 1

Name: Mohammad Al-Haggar

Review Date: 2018-07-16 23:54

Specific Comments To Authors: Differentiate between names of gene and protein (keep MARS throughout the article in *Italic font*). Are there any symptoms in the parents transmitting the mutants? IF no, are there any phenotype genotype correlation in these mutation forms. Keep number < 9 in letters.

Scientific Quality: Grade B (Very good)

Language Quality: Grade B (Minor language polishing)

Conclusion:Accept (General priority)

Specific Comments To Authors (File):

Response:

1. First of all, we express our deepest gratitude towards thoughtful suggestions by Dr. Mohammad Al-Haggar.
2. We changed gene names to italic font format all throughout the manuscript as long as the name was mentioned as a gene, not a protein.
3. Parents were both healthy, and did not have any symptoms related to the disease phenotype. We also added information about the parental health status on page 6,

and 7 of the manuscript file.

4. We added the following information related to genotype-phenotype correlation on page 10: “All eight mutations from European ILLD cases were located in the Rossmann fold of MARS protein. However, only one out of four mutations from Chinese cases carried mutation in the Rossmann fold domain, and significantly different from European ILLD case (Fisher's exact=0.018) (Figure 2B). Our case also suggested that, severe mutations may lead to more organ/system involvement and severe outcome.”
5. We changed all numbers less than 9 within the manuscript to the letter format.
6. We also did some language polishing to make the manuscript more readable.

Reviewer 2

Name: Anonymous

Review Date: 2018-07-08 12:27

Specific Comments To Authors: Dear Editor, I reviewed the manuscript titled “Novel MARS variants/phenotypes in interstitial lung and liver disease, a case report”. In the literature, patients who have liver and pulmonary disorders with MARS mutation published before. Since this case has a novel truncating mutation and an insertion of a single aminoacid, and the originality of this case is related with these genetic changes, this manuscript is suitable for publication in a Journal mainly related with genetics. My

comments are listed below: 1. Case: “....at the age of 5-months without abilities of crawling..”. Babies can not crawling at the age of 5 months. 2. I think it is critical to present prothrombin time and INR values of such a patient with severe liver disease. 3.

Some grammatical errors “Trigleceryde”, “Aacetabular”, “hemoglobulin”, “platelete”

Scientific Quality: Grade D (Fair)

Language Quality: Grade B (Minor language polishing)

Conclusion:Rejection

Specific Comments To Authors (File):

Response:

1. First of all, thanks so much for the reviewer for constructive suggestions on improving the manuscript quality.
2. We removed the word “crawling” when describing the patient’s developmental milestones on page 5.
3. We added information on blood coagulation profiles, including prothrombin time and INR values, on Table 1 and Table 3.
4. We corrected grammatical errors such as “Trigleceryde”(Table 1), “Aacetabular”(Figure 1 caption), “hemoglobulin”(Table 1), “platelete”(Table 1).

Reviewer 3

Name: Anonymous

Review Date: 2018-07-16 12:55

Specific Comments To Authors: Major comments Abuduxikuer K et al. submitted a case report of MARS variants in a patient with ILLD. The submitted manuscript has been suit for publication in the current version. First, the principal finding of the study was that authors performed Whole exome sequencing and found a truncating variant (c.2158C>T/p.Gln720Stop) together with a novel tri-nucleotide insertion (c.893_894insTCG that caused the insertion of an arginine on the amino acid position of 299) in MARS gene. Second, the case report encompasses an interesting topic for general readers in World Journal of Gastroenterology. Third, the submitted manuscript has been suit for publication in the current version.

Scientific Quality: Grade B (Very good)

Language Quality: Grade A (Priority publishing)

Conclusion:Accept (General priority)

Specific Comments To Authors (File):

Response:

1. We would like to thank the reviewer for accepting our manuscript, as well as making kind suggestions for improvement.