Author’s response to reviews

Title: The additional genetic diagnosis of homozygous Sickle Cell Disease in a patient with Waardenburg-Shah Syndrome: A Case Report

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Author’s response to reviews:

November 29, 2018

Professor Michael Kidd

Editor

Journal of Medical Case Reports

RE: JMCR-D-18-00652

"The additional genetic diagnosis of homozygous Sickle Cell Disease in a patient with Waardenburg-Shah Syndrome: A Case Report"

Dear Professor Kidd

We thank the editors and reviewers for a very thorough review of this manuscript.

Please see the following responses to comments and queries raised. We make reference in particular to reviewer two.
1. Please supply the full work postal address for the corresponding author.

Please note the full work postal address of the corresponding author is as follows:

Dr Angela E Rankine-Mullings
Sickle Cell Unit
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Jamaica

PAVANKUMAR TANDRA, MBBS (Reviewer 1) Accept
1. Do you believe the case report is authentic? YES
   Yes/No
   Authors’ response: We note this response and thank the reviewer for comments received.

2. Do you have any ethical concerns? Please consider if local Institutional Review Board approval or ethical approval was obtained (if appropriate) and if the patient (or their parent or guardian in the case of children under 18) gave written, informed consent to publish this case and any accompanying images. A statement to this effect should appear in the manuscript.

   Comments: I DONT HAVE ANY ETHICAL CONCERNS.
   Authors’ response: We note this response and thank the reviewer for comments received.

3. Does the Introduction explain the relevance of the case to the medical literature? YES
   Yes/No
   Authors’ response: We note this response and thank the reviewer for comments received.
4. Does the article report the following information? Where information is missing, please specify.

YES TO ALL BELOW QUESTIONS.

a. The relevant patient information, including:
   - De-identified demographic information (age, gender, ethnicity)
   - Main symptoms of the patient
   - Medical, family and psychosocial history
   - Relevant past interventions and their outcomes

b. The relevant physical examination findings

c. Important dates and times in this case (if appropriate, organized as a timeline via a figure or table); if specific dates could lead to patient identification, consider including time relevant to initial presentation, i.e. initial presentation at T = 0, follow up at T = 1 month.

d. Diagnostic assessments, including:
   - Diagnostic methods
   - Challenges (e.g., financial, language/cultural)
   - Reasoning and prognostic characteristics (e.g., staging), where applicable

e. Types and mechanism of intervention

f. A summary of the clinical course of all follow-up visits

Comments:

Authors’ response: We note this response and thank the reviewer for comments received.

5. Is the interpretation (discussion and conclusion) well balanced and supported by the case presented? YES
Comments:

Authors’ response: We note this response and thank the reviewer for comments received.

6. Is the anonymity of the patient protected? Please consider any identifying information in images such as facial features or nametags, whether the patient is named etc. If not, please detail below. YES

Yes/No

Authors’ response: We note this response and thank the reviewer for comments received.

7. Is the Abstract representative of the case presented? YES

Comments:

Authors’ response: We note this response and thank the reviewer for comments received.

8. Does the case represent a useful contribution to the medical literature?

Comments: VERY WELL WRITTEN CASE AND IT IS A USEFUL CONTRIBUTION TO THE EXISTING LITERATURE.

Authors’ response: We thank the reviewer for these comments.

9. Additional comments for the author(s)?

Umit Yavuz Malkan (Reviewer 2) Minor Revision

1. Do you believe the case report is authentic?

Yes/No

Y

Authors’ response: We note this response and thank the reviewer for comments received
2. Do you have any ethical concerns? Please consider if local Institutional Review Board approval or ethical approval was obtained (if appropriate) and if the patient (or their parent or guardian in the case of children under 18) gave written, informed consent to publish this case and any accompanying images. A statement to this effect should appear in the manuscript.

Comments: consent should be taken from patient

Authors’ comments: Thank you for your comment. Please see the section of the manuscript submitted entitled “Ethics approval and consent to participate.” (Page 9 lines 167-170)

Please note that this patient is deceased hence consent was obtained from the patient’s nearest relative who was listed in the medical notes of this patient as the next of kin. We have revised this section in the manuscript to make this clearer.

3. Does the Introduction explain the relevance of the case to the medical literature?

Yes/No

y

Authors’ response: We note this response and thank the reviewer for comments received.

4. Does the article report the following information? Where information is missing, please specify.

a. The relevant patient information, including:

- De-identified demographic information (age, gender, ethnicity)
- Main symptoms of the patient
- Medical, family and psychosocial history
- Relevant past interventions and their outcomes

b. The relevant physical examination findings
c. Important dates and times in this case (if appropriate, organized as a timeline via a figure or table); if specific dates could lead to patient identification, consider including time relevant to initial presentation, i.e. initial presentation at T = 0, follow up at T = 1 month.

d. Diagnostic assessments, including:
   - Diagnostic methods
   - Challenges (e.g., financial, language/cultural)
   - Reasoning and prognostic characteristics (e.g., staging), where applicable

e. Types and mechanism of intervention

f. A summary of the clinical course of all follow-up visits

Comments:

5. Is the interpretation (discussion and conclusion) well balanced and supported by the case presented?

Comments:

6. Is the anonymity of the patient protected? Please consider any identifying information in images such as facial features or nametags, whether the patient is named etc. If not, please detail below.

Yes/No

7. Is the Abstract representative of the case presented?

Comments: Y

Authors’ response: We note this response and thank the reviewer for comments received.

8. Does the case represent a useful contribution to the medical literature?
Comments: Y

Authors’ response: We note this response and thank the reviewer for comments received.

9. Additional comments for the author(s)? Y

Authors’ response: We note the reviewer’s comments and thank the reviewer for comments received.

aisha alosail, md (Reviewer 3) Accept

Thanks a lot for allowing me to review this interesting case

It was really interesting In my country sickle cell is very common and this case report was the second I saw

Well done

Corresponding author’s response: We appreciate the comments received and thank the reviewer for communicating this to us.

Author Decision Letter Minor revision

Sincerely

Angela E Rankine-Mullings
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Sickle Cell Unit
Caribbean Institute for Research
University of the West Indies