Reviewer's report

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

Version: 1 Date: 04 Dec 2018

Reviewer: Umit Yavuz Malkan

Reviewer's report:

1. Do you believe the case report is authentic?

Yes/No

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:

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

Yes/No

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:

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

Comments:

9. Additional comments for the author(s)?
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Please indicate how interesting you found the manuscript:

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Quality of written English
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Acceptable

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