Reviewer’s report

Title: Exon sequencing of the alpha-2-globin gene for the differential diagnosis of central cyanosis in newborns: a case report

Version: 1 Date: 06 May 2019

Reviewer: Reviewer 2

Reviewer's report:

PEER REVIEWER ASSESSMENTS:

RELEVANCE - Does this case report make a contribution to medical knowledge, have educational value, or highlight the need for a change in clinical practice or diagnostic/prognostic approaches?

Yes, this report contributes to medical knowledge

CASE DESCRIPTION - Are the details of the case sufficiently well described to understand the patient's symptoms and course of treatment?

No - there are minor issues

DIAGNOSIS/INTERPRETATION - Based on the facts presented, are the diagnosis, interpretation, and course of treatment medically sound?

Yes, the work described is medically sound

DISCUSSION OF THE CASE - Does the discussion appropriately analyse the importance of the findings and their relevance to future understanding of disease processes, diagnosis or treatment? Has an adequate literature review pertinent to the case been included?

No - there are minor issues

OVERALL MANUSCRIPT POTENTIAL - Could an appropriately REVISED version of this work represent a technically sound contribution?

Probably - with minor revisions
PEER REVIEWER COMMENTS:

GENERAL COMMENTS: Interesting case report and well written manuscript. However, authors should clarify how this case report is different from previously reported case reports. Also authors mentioned they will review the literature on similar cases however, discussion has not been organized that way.

REQUESTED REVISIONS:

* Recommend authors to explain in more details what makes this case different from previously reported cases.

* If authors want to review available literature, I recommend summarizing previously reported cases in a table including important information such as year of publication, disease presentation, diagnosis, etc.

* In the method section, please clarify what diagnostic tests were performed to rule out cardiac and pulmonary disorders.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

No

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript
Quality of written English
Please indicate the quality of language in the manuscript:

Acceptable

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Please complete a declaration of competing interests, considering the following questions:

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