Reviewer's report

Title: Hb Knossos (HBB: c.82G>T), β-globin CD 5 (-CT) (HBB: c.17_18delCT) and δ-globin CD 59 (-A) (HBD: c.179delA) mutations in a Syrian patient with β-thalassemia intermedia

Version: 1 Date: 04 Dec 2018

Reviewer: Maria de Fatima Sonati

Reviewer's report:

The manuscript describes a rare and interesting case of hemoglobinopathies which is worthy of publication. However, some points can be improved to make it more suitable to the reader. The title could be a bit more concise: "Hb Knossos (HBB:c.82G-T), β-globin CD 5 (-CT) (HBB:c.17_18delCT) and δ-globin CD 59 (-A) (HBD:c.179delA) mutations in a Syrian patient with β-thalassemia intermedia". In addition to this nomenclature, the one recommended by the HGVS should be mentioned at least once in the text. In both the Abstract and the Background, it should be clarified what is thalassemia intermedia and what are its most frequent causes. In Case Presentation, both in the Abstract and in the text, it would be important to include the patient's age and clinical presentation, as well as his history of blood transfusions (never? Once? Sometimes? Under what circumstances?). The concomitant presence of deletional α-thalassemia should be excluded and this should be mentioned in both the Abstract and the text. At the end of the Conclusions, in the Abstract, the authors declare that "different δ-thalassemia gene mutation must be screened to avoid the misdiagnosis of β-thalassemia disease". On what basis should this be done? Please complete this sentence. In line 55 of page 2 (Background), the authors declare that "Hb A2 increased more than 3.2% in a typical ...". The cut-off value depends on the method used, please make this clear. In line 8 of page 3 (Background), the statement "However, homozygous δβ-thalassemia may give ... with mild anemia" requires a specific reference or should be excluded. In Case Presentation (page 3), there is no reference to Table 1. Hematological data could be summarized as they are detailed in this table. The methods could also be summarized, provided it has the proper references. Sequences of the primers, for example, if not original, can only be indicated by references. Please inform about the investigation of deletional α-thalassemia. Discussions and Conclusions may also be more concise. Figure 1 is unnecessary. References are Ok but need standardization. English can be improved.
Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

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:

Needs some language corrections before being published

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