Reviewer’s report

Title: Molecular Analysis of a Large Novel Deletion Causing α+-Thalassemia

Version: 0 Date: 14 Nov 2018

Reviewer: Maria Sonati

Reviewer's report:

This manuscript reports a new alpha-thalassemic deletion that removes about 6.9 kb of DNA including the alpha-2 gene, and therefore classified as alpha+. In our evaluation, it deserves publication after a major review. Firstly, the text must be fully revised by a native English speaker. In the Abstract, the specifications of the hematological analyzer and the electrophoresis equipment, as well as their results, are not necessary - the most important is to describe how the detection and characterization of the novel deletion was carried out. It is sufficient to inform that it was an adult male patient of Chinese origin with Hb H disease. On the other hand, there is no mention of the MLPA and DNA sequencing, both used in the detection and characterization of the deletion. In the Background (lines 46 to 49), the authors should assume that this is a case of Hb H disease. In Methods, references are missing in lines 23 (gap-PCR), 27 (PCR-RDB), 31 (common beta-thalassemia mutations in Chinese) and 42 (deletion breakpoints). In the Results, the authors should assume that it is Hb H disease (not a suspect). If there is no relationship of consanguinity between the patient and his wife, rather than informing his hematological data, it would be important to inform those of his parents, children and siblings if possible. Was the complete familial analysis not done? Table 1 should be supplemented with the other hematological values: RBC, Hb, Hct, reticulocyte counts, etc. (does the patient have no hemolytic anemia?), as well as with family data, if possible. It needs a title. Figure 2 is not necessary. In the Discussion, references are missed; the second and third paragraphs are repeats of what is already stated in the Methods and may be summarized to a few sentences. Finally, the Discussion could be enriched by comparing this case with those whose Hb H disease is caused by the association of the alpha0 allele with the -alpha3.7 and alpha4.2 alleles, the most common.

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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