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: 11 Dec 2018

Reviewer: John Waye

Reviewer's report:

The authors repeatedly refer to the "Xmn-1 locus". This is a SNP that should be described according to the gene and nucleotide change, specifically HBG2:c.-211C>T.

Page 3, lines 38-55: This can be omitted from this section since all of this data is contained in Table 1.

Page 5, line 26: RFLP not RFPL

Page 5, line 39: The correct HGVS nomenclature is HBB:c.82G>T, p.Ala28Ser (not Ala27Ser). HGVS counts the ATG initiation codon as codon 1, whereas the traditional hemoglobinopathy nomenclature begins with the first amino acid.

Page 7, line 23: The danger is failing to detect beta-thalassemia carriers, as opposed to misdiagnosing beta-thalassemia carriers.

Should include their most recent study of beta-thalassemia mutations in Syria (Hematology 2018; 23:697-704).
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.

Unable to assess

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

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