**Reviewer's report**

**Title:** Molecular Epidemiology and Hematologic Characterization of δβ-thalassemia and Hereditary Persistence of Fetal Hemoglobin in 125661 Families of Greater Guangzhou Area, the metropolis of Southern China

**Version:** 0  **Date:** 16 Dec 2019

**Reviewer:** Merlin Crossley

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This is an interesting paper that presents a survey of haemoglobinopathy mutations in a region of southern China. The work is of high quality. The results are interesting and are likely to be clinically helpful, in terms of enhancing diagnosis and genetic counselling. In particular the observation that the Italian ndHPFH is common in this region is new. In addition the analysis of novel deletions and characterisation of mutations that involve the foetal globin genes, including deletion of one of the genes, or triplication, is also very interesting.

I have only a few minor comments.

1. The paper would be enhanced by an illustration of the b-globin locus showing Chinese, Taiwanese, Hb Lepore and the SEA deletions. These are the main ones found, as shown in the pie chart, but many readers may benefit from seeing how they affect the locus.

2. Would it be possible to present a table with the main genotypes shown in the pie chart and the levels of HbF and HbA2?

3. In the final page of the manuscript it is stated that "Using K562 cells and HUDEP-2 cells, Gabriella E revealed that mutations at -200 of the γ-globin promoter disrupt ZBTB7A for HbF repressor binding" but not reference is given. I think this refers to work by Gabriella E Martyn et al.?

**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.

I am able to assess the statistics

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Please indicate the quality of language in the manuscript:

Acceptable

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