Reviewer's report

Title: Rapid detection of PAH gene mutations in Chinese people

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Reviewer: Guy Lenk

Reviewer's report:

The paper is concerning the detection of PAH mutations utilizing a multiplex PCR system followed by reverse dot hybridization. It employs a panel of 9 mutations that are described as being the most common in the Chinese population. The motivation for this research was to develop a sensitive, specific and cost-effective technique to detect these changes.

Although this seems to be a sound scientific undertaking, the language in the paper struggles to convey to the reader the ideas presented. There are many minor examples throughout that need to be addressed to meet the rigor and quality of this journal, but additionally the technical descriptions of the experiments and results themselves is lacking. Two specific examples follow:

P5; line 55 - "If all or most of the wild-type control spots are not colored, the test fails, and it needs to be retested after the analysis of the cause." All or most is very vague. What is the number? If half the control spots were to fail would that be considered a valid test?

Section 2.3 (Evaluation of Methodology) - This section states that of the 200 test samples with known mutations this method detected all with "100% specificity and accuracy". That is an impressive result, and should be expanded upon. How many of each variant were tested? How was the "blinding analysis" mentioned carried out? Since these tests require the ability to determine if a spots intensity is sufficient, were these assessed by multiple individuals? What was the state of the control spots in these assays?

In general much more data and more description of the actual analysis of the hybridization technology needs to be included. This paper also could benefit from a more extensive statistical analysis of the results.
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.

I recommend additional statistical review

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Please indicate the quality of language in the manuscript:

Not suitable for publication unless extensively edited

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