Reviewer’s report

Title: Saliva Samples are a Viable Alternative to Blood Samples as a source of DNA for High Throughput Genotyping.

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Reviewer: Katherine Mitsouras

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The manuscript entitled “Saliva Samples are a Viable Alternative to Blood Samples as a Source of DNA for High Throughput Genotyping” by Abraham et al compares the performance of paired saliva and blood samples using two different genotyping platforms, the Illumina HumanHap300 BeadChip arrays and the Taqman assay. Previous studies in dogs and humans have systematically assessed the performance of paired blood and saliva samples and found that saliva samples are a suitable alternative to blood DNA and yield high quality genotyping data [1, 2]. The present manuscript extends these previously published studies by using a much larger sample size of paired samples (79 paired blood/ saliva samples) and by assessing the performance of the paired samples on two different genotyping platforms that utilize different experimental techniques. The authors demonstrate that both the genotype call rates and the genotype concordance for the paired samples were >97% for both genotyping platforms, therefore DNA extracted from saliva yields high quality genotyping data and is ideally suited for large scale epidemiological studies and clinical trials.

Major compulsory revisions

1) In table 1, the authors need to include a column showing the mean and range for the A260/A280 ratio for blood and saliva-derived DNA. The A260/A280 ratio is a measure of DNA purity and should be reported in order to make meaningful comparisons of the quality of DNA extracted from blood and saliva. Similar studies comparing the quality and performance of DNA extracted from different sources (blood, saliva, buccal swabs) typically include the A260/A280 ratio [1, 3-6].

2) On page 8 the authors state that “A valid concern, prior to this study, was that saliva may contain significant proportions of DNA from oral bacteria and/ or food. If this had been the case, the true concentration of human DNA added to each assay would have been below the picogreen calculated concentration and overall rates would have been proportionately reduced.” DNA Genotek reports that the median bacterial content in DNA samples purified using the Oragene# self-collection kit is 11.8%, therefore the vast majority of DNA is of human origin (http://www.dnagenotek.com/US/pdf/PD-WP-011.pdf). The authors should revise the above statement to incorporate the report on the proportion of bacterial DNA provided by the kit manufacturer.
3) In the last sentence of the second paragraph of the background (page 4), the authors discuss a previous study by Bahlo et al. The study assessed the performance of saliva and blood-derived DNA samples on the Illumina genotyping platform, by measuring the concordance of the Genotype Call Rate and Illumina GenCall Score between the paired samples [2]. In reference to this study, the authors state “however no comparison was made of the quality of the data obtained.” This statement is unclear, vague and confusing and needs to be modified to better convey the authors’ point.

Minor essential revisions
1) The authors need to provide table and figure legends.
2) Figure 1 needs to be bigger in order for the axis labels and data labels to be legible.
3) “DNAgenotek” should be replaced with “DNA Genotek”, which is the way the company name is shown on the company website (http://www.dnagenotek.com/US/company/overview.html).
4) The authors should specify which Gen-Probe genomic DNA extraction kit they used for purification of DNA from blood.
5) There is a typo in the last sentence of the second paragraph on page 4.
6) In the methods section entitled “Saliva DNA extraction” (page 5), the authors state that after collection saliva samples were stored prior to DNA extraction. The authors need to state how long saliva samples were stored and the storage conditions.
7) The last sentence of the methods section entitled “Taqman Genotyping” (page 6) states that the Taqman assays used in the manuscript were chosen because they were shown to work well in other research projects. The authors need to either provide a reference for this statement or indicate that the data is not shown.
8) Although this manuscript focuses on human samples, a previous study in dogs compared the performance of paired saliva and blood-derived DNA in dogs using the Illumina Infinium platform and demonstrated that saliva DNA is suitable for high-throughput genotyping studies [1]. The authors should state this in their manuscript, as well as cite the canine study.

Discretionary revisions
“Additional table” should be changed to “supplemental table” (methods section entitled “Taqman genotyping”) on page 6.

References


Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests