Affymetrix® Axiom® Array performance using ORAcollect®•DNA samples prepared with prepIT®•Q2A: A new direct-to-assay method eliminating extraction, quantification and normalization

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Introduction

The growing awareness of personalized medicine, popularity of pharmacogenetic testing and continued research in disease and population studies has increased the number of genomic samples collected and analyzed year over year. The non-invasive nature of DNA collection using ORAcollect®•DNA from saliva has increased access to donors across the globe, contributing to the growth of genomic testing. The increased demand for genomic testing has challenged laboratories to process more samples on a daily basis however routine handling, such as DNA extraction, quantification and normalization, adds considerable time and cost to the laboratory workflow, often limiting the number of samples that can be processed in a single day.

prepIT®•Q2A reagent enables a rapid, liquid based removal of inhibitors found in ORAcollect•DNA saliva samples, and eliminates the need to perform DNA extraction, quantification and normalization prior to genotyping. The reduced workflow and compatibility on automated liquid handlers allows prepIT•Q2A to be used inline with genotyping array preparation, making it ideal for laboratories wanting to scale up their sample throughput — from saliva sample direct to assay.

This study evaluates the performance of prepIT•Q2A prepared ORAcollect•DNA from saliva samples on the Affymetrix® Axiom® Genotyping Array platform using QIAGEN® QIAamp® DNA extracted DNA of matched samples as a control.

Materials and methods

Sample collection: Saliva samples were collected from 20 healthy donors using ORAcollect•DNA devices. These samples were pre-screened for DNA yield, and 12 of these samples were selected to represent the range of DNA yield observed in a typical population.

DNA preparation: DNA was prepared from ORAcollect•DNA samples by prepIT•Q2A and QIAGEN QIAamp DNA Mini. A comparison of the steps involved in each protocol is outlined in Figure 1.

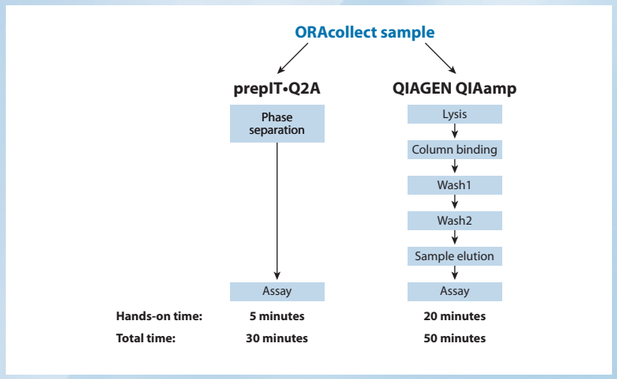

***Figure 1**: Workflow comparison of prepIT•Q2A preparation vs. QIAGEN extraction.*

Affymetrix array: An Axiom DTC array examining 795,261 SNPs was used. All samples were directly input into the array workflow without normalization. Processing was performed according to the manufacturer's instructions and analysis was performed using the Axiom Analysis Suite software.

Results

Call rates of paired samples processed by QIAGEN QIAamp and prepIT•Q2A are shown in Table 1. Call rates were greater than 98.7% in all samples tested, and no difference was observed between the two DNA preparation methods. Concordance rates for each paired sample was greater than 99.0% as shown in Figure 2.

prepIT•Q2A samples resulted in similar DNA concentrations as paired QIAGEN QIAamp samples. When the impact of DNA input on call rates was examined (Figure 3), no trend was observed. This indicates that, even at low inputs, samples were of sufficient quality to generate acceptable call rates, and sample normalization was not required.

Table 1: Call rates by preparation method

Donor number	QIAGEN QIAamp	prepIT•Q2A
1	99.032	99.298
2	99.183	99.219
3	99.08	99.042
4	98.983	98.97
5	99.031	99.02
6	99.342	99.163
7	99.235	99.286
8	99.261	98.725
9	98.878	99.274
10	99.144	98.764
11	99.139	99.228
12	99.101	99.101
Average	**99.117**	**99.091**
Minimum	**98.878**	**98.725**
Maximum	**99.342**	**99.298**

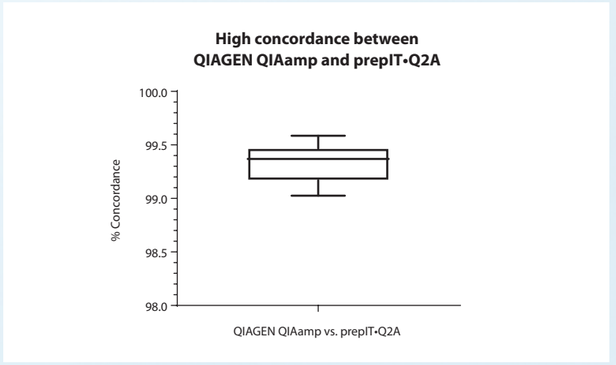

***Figure 2**: % Concordance (# of concordant SNPs/Called SNPs) between QIAGEN QIAamp and prepIT•Q2A paired samples. Whiskers indicate minimum and maximum values.*

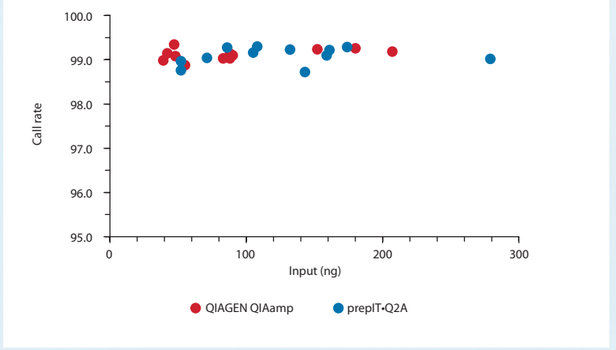

***Figure 3**: Call rate vs. DNA input by sample preparation method.*

Conclusion

prepIT•Q2A prepared ORAcollect•DNA saliva samples generated high quality call rates on the Affymetrix Axiom array and performed as well as QIAGEN QIAamp extracted samples. Sample normalization was not required to generate these results.

Thus, prepIT•Q2A offers a fast and effective solution helping laboratories that perform genotyping overcome sample throughput challenges to meet the growing demands of genomic testing.

DNA genotek

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Patent (www.dnagenotek.com/legalnotices) MK-01200 Issue 1/2019-04

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