Versatile 96-SNP Genotyping Panel Enables DNA Fingerprinting and Sample Integrity Assessments

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Introduction

Biorepositories provide access to high-quality, curated samples for basic and clinical research purposes. Sample degradation, misidentification and contamination are significant risks to the integrity of banked samples. Distribution of such samples can waste time and laboratory resources and negatively impact the integrity of research studies.

Standard procedures for sample traceability and quality assessment have been employed by biorepositories for many years, including but not limited to barcode labeling, LIMS tracking and DNA quantification. Implementing a DNA fingerprinting method in the biorepository workflow provides more informative quality assessment tools and a direct assessment of sample molecular identity.

The Advanta Sample ID Genotyping Panel is a 96-SNP (single-nucleotide polymorphism) assay that generates a sample-specific DNA fingerprint and supports multiple quality assessments of research specimens throughout the sample journey. Developed for use with the Biomark™ HD system and based on Fluidigm microfluidics technology, the workflow integrates fluidic circuits (IFCs) to precisely combine multiple reactions at nanoliter volumes. In this poster, we demonstrate the utility of the Advanta Sample ID Genotyping Panel as a sample identity and quality assessment tool.

Advanta Sample ID Genotyping Panel content and workflow

The Advanta Sample ID Genotyping Panel facilitates DNA sample quality control (QC) and tracking of human samples through the use of 96 SNPs. 80 SNPs are located in exonic regions and 16 SNPs are located in intronic and non-coding regions. The workflow uses Fluidigm microfluidics technology to complete profiling of up to 96 samples per run in four hours.

Prepare samples and assays.

Transfer samples to the IFC.

Process the IFC using the automated Juno™ system.

Perform real-time PCR and data acquisition using Biomark HD.

Analyze results.

Results

SNP profiling uniquely identifies individuals

96 sample call map of some of genotyping runs

Figure 2a. 96 samples were run on a 96.96 Dynamic Array™ IFC and sequenced on the Biomark HD system. Samples include one autologous typing with 17 reactions. All samples generated unique SNP profiles to duplicate profiles. Below is a call map review of 96.96 Dynamic Array™ IFC. Samples are represented in rows and indexed SNP calls in columns.

Sex chromosome analysis detects ≥5% contamination

Genomic sex discrepancy can signal a contamination event.

Sample | Gender Call by Software | Sample Size | Gender Call by Software
<table>
<thead>
<tr>
<th></th>
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<tbody>
<tr>
<td>T1</td>
<td>Male</td>
<td>T1</td>
</tr>
<tr>
<td>T2</td>
<td>Female</td>
<td>T2</td>
</tr>
<tr>
<td>T3</td>
<td>Klinefelter male</td>
<td>T3</td>
</tr>
<tr>
<td>T4</td>
<td>Klinefelter male</td>
<td>T4</td>
</tr>
</tbody>
</table>

Figure 3a. Two male (M1 & M2) and two female (F1 & F2) samples were tested on a 96.96 Dynamic Array IFC and Juno™ 96.96 Genotyping IFC using the Juno and Biomark HD workflow. Duplicate genotypes were generated with 96.96 Dynamic, Array™ IFC. Parental gender determines which chromosome will be used for gender identification.

Figure 3b. Left scatter plot of X chromosome analysis detects ≥5% M3 and female M3.

Sex chromosome analysis detects ≥5% contamination

Figure 4a. Each DNA sample was incubated at 95 °C for 15 min in a 1.5 mL tube and 0.2 µL β-actin primers and probes were added to the 96-well plate. Beta-actin controls are included to control DNA integrity. Amplification was performed using the Fluidigm Biomark HD system with the 96.96 Dynamic Array™ IFC. Samples were incubated at 95 °C for 15 min and then 40 cycles of 95 °C for 1.5 min and 60 °C for 1 min.

SNP genotyping can identify low-quality samples

SNP call rate of 100% identity

Figure 4b. Quantities for 200 bp and 60 bp qPCR concentration analysis of samples show the unique SNP profiles generated for 13 individuals from the CEPH pedigree (father, mother and offspring). P-values and qPCR concentration analysis of samples show the unique SNP profiles generated for 13 individuals from the CEPH pedigree (father, mother and offspring). P-values and qPCR concentration analysis of samples show the unique SNP profiles generated for 13 individuals from the CEPH pedigree (father, mother and offspring). P-values and qPCR concentration analysis of samples show the unique SNP profiles generated for 13 individuals from the CEPH pedigree (father, mother and offspring). P-values and qPCR concentration analysis of samples show the unique SNP profiles generated for 13 individuals from the CEPH pedigree (father, mother and offspring). P-values and qPCR concentration analysis of samples show the unique SNP profiles generated for 13 individuals from the CEPH pedigree (father, mother and offspring).

Conclusion

The Advanta Sample ID Genotyping Panel generates a sample-specific DNA fingerprint that can be differentiated from even closely related samples. It can identify degraded samples or those that have been mixed up or contaminated. Early detection of poor-quality, contaminated or improperly curated samples can improve research quality and reduce costs of superfluous testing of poor-quality samples.

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