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 consume time and laboratory resources and negatively impact the integrity of research studies.

Standard procedures for sample traceability and quality assurance 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 genetic 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 uses integrated 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 high-quality samples using the 96 SNP array. The workflow samples are loaded on an IFC and combined automatically in the Juno system. The IFC is subsequently moved to the Biomark HD system to complete profiling.

Prepare samples and assay 

Transfer samples and assay to the IFC. 

Process the IFC using the automated Juno™ system. 

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

Analyze results.

Figure 1. The Advanta Sample ID workflow, from sample preparation through data analysis. 96 samples and the 96-assay panel are loaded on an IFC, and combined automatically in the Juno system. The IFC is subsequently moved to the Biomark HD system for thermal cycling and imaging. The genotyping run is then analyzed using Fluidigm SNP Genotyping Analysis software.

Advanta Sample ID Genotyping Panel provides an accurate, reproducible fingerprint

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 incorrectly curated samples can improve research quality and reduce costs of superfluous testing of poor-quality samples.

Results

SNP profiling uniquely identifies individuals

Genetic sex discrepancy can signal a contamination event.

Sample | Gender Call by Software | Sample | Gender Call by Software
--- | --- | --- | ---
F1 | Male | M1-1% T3 | Male
F1-1% M3 | Female | M1-1% T3 | Male
F1-10% M3 | No call/100% male | M1-10% T3 | Male
F2 | Female | M2-100% T3 | Male
F2-10% M3 | Female | M2-100% T3 | Male
F2-5% M3 | No call | M2-5% T3 | Male
F2-10% M3 | Klinefelter male | M2-10% T3 | Male
Figure 5a: Sex chromosome analysis detects ≥5% contamination

Sex chromosome analysis detects ≥5% contamination

- **Gender Call by Software**: For Research Use Only. Not for use in diagnostic procedures.

SNP genotyping can identify low-quality samples

- **qPCR concentration analysis of samples**: For Research Use Only. Not for use in diagnostic procedures.

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 incorrectly curated samples can improve research quality and reduce costs of superfluous testing of poor-quality samples.