Microarrays

Application guide for the SwiftArrayStudio Microarray Analyzer

In this document, we show:

- Innovative features of the SwiftArrayStudio Microarray Analyzer
- Advances in the Axiom SwiftArray Assay that speed data production
- Concordance with data from the Axiom 2.0 assay
- A high-throughput workflow using the Multidrop Combi Reagent Dispenser
- Automation with the NIMBUS Target Preparation Instrument
- Use of the SwiftArrayStudio analyzer for companion animal genotyping

Introduction

DNA microarrays have played a central role in advancing genomic analysis across a wide range of applications, including human health research, model organism studies, and agricultural breeding. By enabling large-scale interrogation of genetic variation and gene expression, microarrays have become foundational tools in both basic and applied research, as well as in clinical and translational laboratories. Their versatility, scalability, and efficiency continue to make DNA microarrays a cornerstone technology for innovation, driving deeper understanding of the genomic factors underlying health, disease, and biological diversity.

Applied Biosystems™ instruments are part of a long history of innovation in genetic analysis technologies. In response to increasing demand for speed and throughput in microarray workflows, we developed the Applied Biosystems™ SwiftArrayStudio™ microarray solution. This integrated platform combines a rapid target preparation workflow—completed within a single day—with an instrument that automates array processing, including hybridization, washing, and scanning, enabling faster generation of high-quality data at scale. In this document, we introduce the instrument and assay, and illustrate a few of the applications that can be performed on the SwiftArrayStudio platform.

SwiftArrayStudio Microarray Analyzer—engineered for reliability

The Applied Biosystems™ SwiftArrayStudio™ Microarray Analyzer was designed from the bottom up for rapid acquisition of microarray data. Some of the benefits of the instrument are described here. For more information, see the SwiftArrayStudio Microarray Analyzer **data sheet**.

Usability and efficiency

Many of the key features of the SwiftArrayStudio Microarray Analyzer advance usability and efficiency. These features make setting up, using, and maintaining the system straightforward.

- Components required to run array plates are contained within a single unit. No external power supply, light source, or computer workstation is required.
- The SwiftArrayStudio Microarray Analyzer provides automated array processing. All on-instrument steps are facilitated by newly developed room temperature–stable reagents.
- Runs are set up and monitored on the instrument using a novel touchscreen-driven graphical user interface (Figure 1).
- The instrument is networkable, allowing remote monitoring of runs, and can be used in conjunction with a laboratory information management system (LIMS).

Reliability

The SwiftArrayStudio Microarray Analyzer has been designed to provide a robust and reliable platform that maximizes laboratory uptime. In addition to helping keep the data flowing, these innovations help minimize service visits.

- A novel auto-teach capability substantially reduces positioning calibration errors.
- An internal multiwavelength LED light source, designed to last for the lifetime of the instrument, eliminates the need for periodic replacement of the light source.
- On-platform placement of four 1 L ergonomic wash bottles with simple snap-in mounting helps minimize the amount of fluid tubing required to run the instrument.

Speed

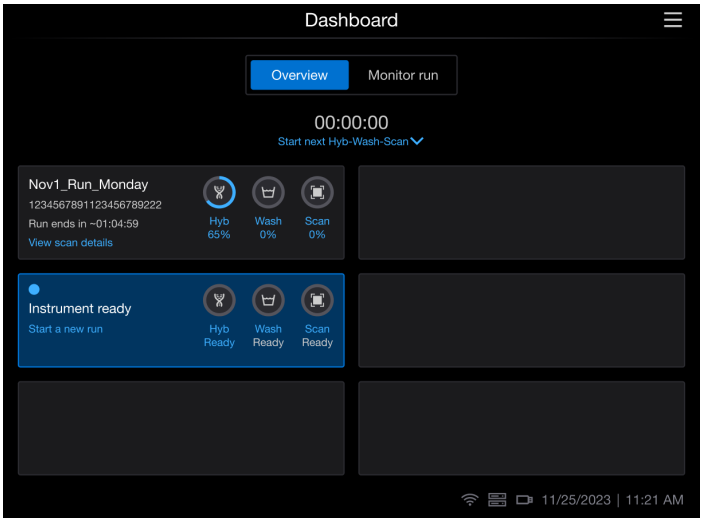
The SwiftArrayStudio Microarray Analyzer has incorporated internal features that improve the rate of array plate processing. These changes increase the speed at which data can be collected from samples.

- Improved autofocus algorithms during scanning enable fast calculation of the optimal plane for rapid image acquisition.
- The LED light source eliminates filter switching between wavelengths, further increasing the speed of image acquisition.
- A single plate can be processed without manual intervention in about 21 hours.

Support

The SwiftArrayStudio Microarray Analyzer was designed to be easy to use, but also easy to service when needed. These designs take advantage of the Thermo Fisher Scientific global service and support team.

- Service and maintenance are simplified by making extensive use of modular FRUs (field replaceable units).
- When connected to a network, a novel on-instrument Smart Help function enables remote troubleshooting, log file transfer, and support team engagement.
- Other software packages automate calibration and system testing, eliminating manual tuning and operator biases from field service engineers during preventive maintenance.



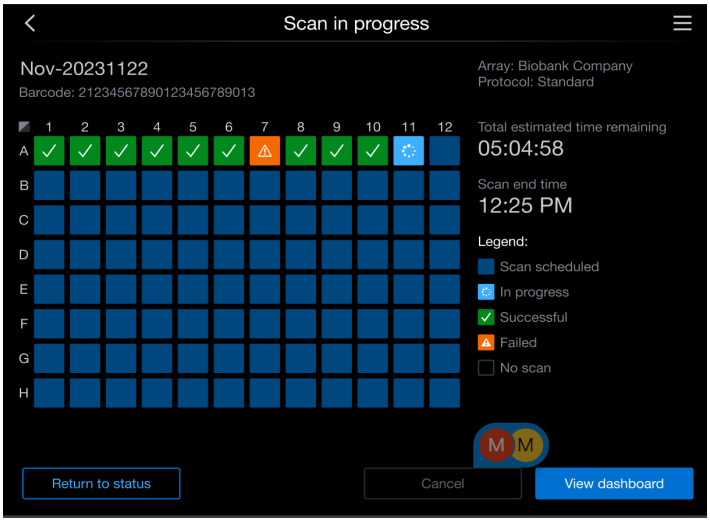


Figure 1. The SwiftArrayStudio Microarray Analyzer is controlled through an intuitive graphical user interface. This facilitates setting up, monitoring, and troubleshooting data collection from Axiom microarrays.

The Axiom SwiftArray Assay—speed to results

To meet the growing demand for speed in high-throughput microarray analyses, we recently developed the Applied Biosystems™ Axiom™ SwiftArray™ Assay. The assay includes a target preparation workflow that can be completed in a single workday. When run on the SwiftArrayStudio Microarray Analyzer, the assay can provide data on an Applied Biosystems™ Axiom™ high-throughput array plate after an overnight hybridization and scan (Figure 2). The workflow reduces the sample-to-answer time from 5 days to 2 days, allowing researchers to gather and interpret genetic data to keep pace with their needs.

The Axiom SwiftArray Assay and workflow for target preparation is briefly described here (Figure 2). For in-depth details and protocols, see the Axiom SwiftArray Assay **user guide**.

- Genomic DNA is first chemically denatured to create single-stranded templates. After neutralization, the DNA is subjected to whole-genome amplification (WGA, 150 min).
- The samples are then digested under controlled conditions to produce fragments averaging 25–125 bp.
- The samples are concentrated by isopropanol precipitation and resuspended in hybridization buffer.
- The quality and quantity of the recovered DNA can be checked here by absorbance spectrophotometry and gel electrophoresis.
- Samples are transferred to a hybridization tray and loaded into the SwiftArrayStudio Microarray Analyzer, together with all the reagents necessary for automated staining and scanning.
- The array processing is initiated and completed within about 21 hours, when data are ready for secondary (interpretive) analysis.

Concordance of data generated with the Axiom 2.0 assay and GeneTitan instrument

A key requirement when introducing a system is to ensure that data quality is comparable to, or better than, that of legacy systems. To assess the concordance of data generated with the SwiftArrayStudio solution relative to existing data, we analyzed 96 genomic DNA samples obtained from the Coriell International HapMap Project sample set. These samples were processed in two independent runs using the Axiom SwiftArray Assay on the SwiftArrayStudio instrument. For comparison, the same samples were processed in two independent runs using the Axiom 2.0 assay on the GeneTitan MC instrument. The same human genotyping microarray type was used on both platforms.

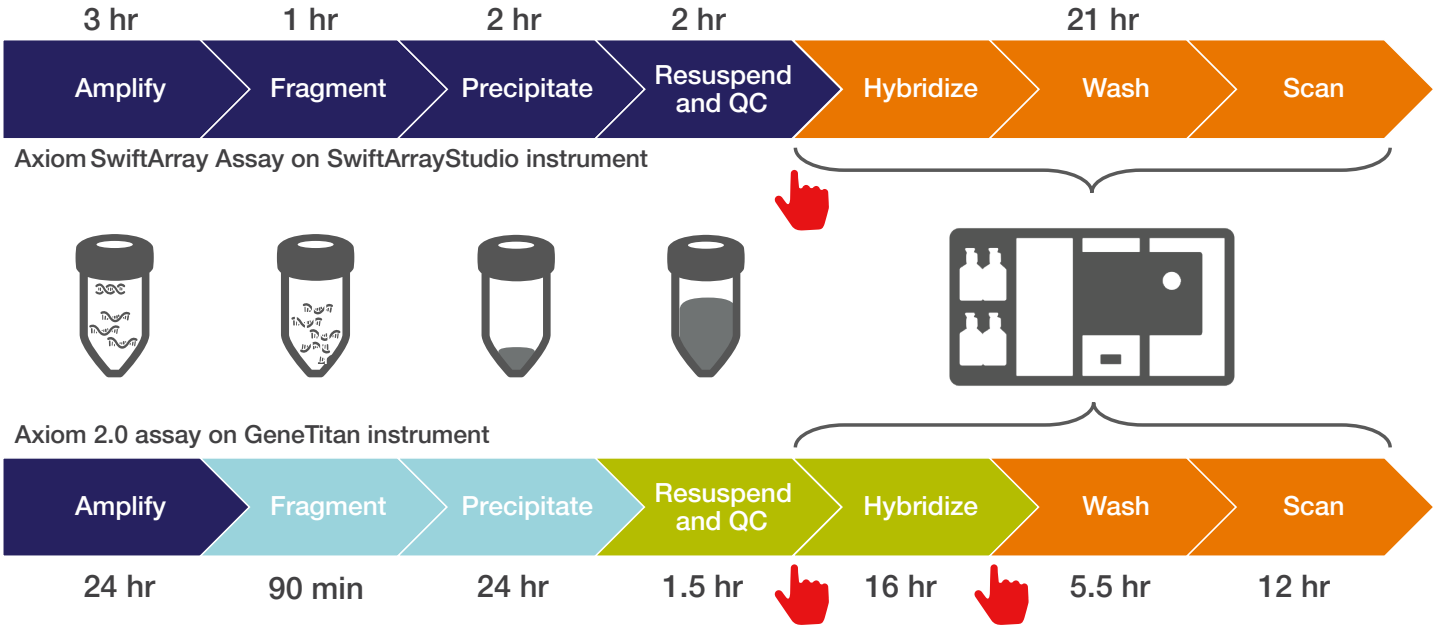


Figure 2. Introducing the Axiom SwiftArray Assay. (Top) The template preparation steps for the Axiom SwiftArray Assay can be completed in about 8 hours in a single working day. On-instrument, autonomous array processing can generate data within 24 hours of loading. Overall, a full set of genotypes and copy number calls from an Axiom array plate can be generated in less than 48 hours. (Bottom) In contrast, the Applied Biosystems™ Axiom™ 2.0 assay with array processing on the Applied Biosystems™ GeneTitan™ Multi-Channel (MC) Fast Scan Instrument requires a total of 5 days to generate data. Different colors represent workflow steps on different days; hand icons represent touchpoints on the instrument, where plates and reagents are loaded or switched out.

For each marker, genotype calls generated by each platform were compared against known reference genotypes. Across the full probe set, overall concordance was 99.96% for the Axiom 2.0 assay on the GeneTitan MC instrument and 99.92% for the Axiom SwiftArray Assay on the SwiftArrayStudio instrument (Table 1).

Table 1. Comparison of sample QC metrics and the concordance of calls made for all probesets and for pharmacogenomics probesets.

	Axiom SwiftArray Assay on the SwiftArrayStudio instrument	Axiom 2.0 assay on the GeneTitan MC instrument
Dish QC (DQC)	0.972	0.979
Average QC call rate (%)	99.68	99.40
Average sample concordance* (%)	99.921	99.957
Pharmacogenomics probeset concordance (%)	99.939	99.941

* Concordance is defined as calls that match known truth for all samples, as determined by next-generation sequencing (NGS). For Dish QC definition, please see the Axiom Analysis Suite v4.0.1 user guide.

On both platforms, 736,563 probes demonstrated 100% concordance with the known genotypes of the samples. These values include no-calls and probesets known to be suboptimized (Figure 3A). When analysis was restricted to a subset of probesets relevant to pharmacogenomic (PGx) research, concordance was 99.4% on both platforms (Figure 3B). Collectively, these results indicate that genotyping performance on the SwiftArrayStudio platform closely matches that of the legacy platform.

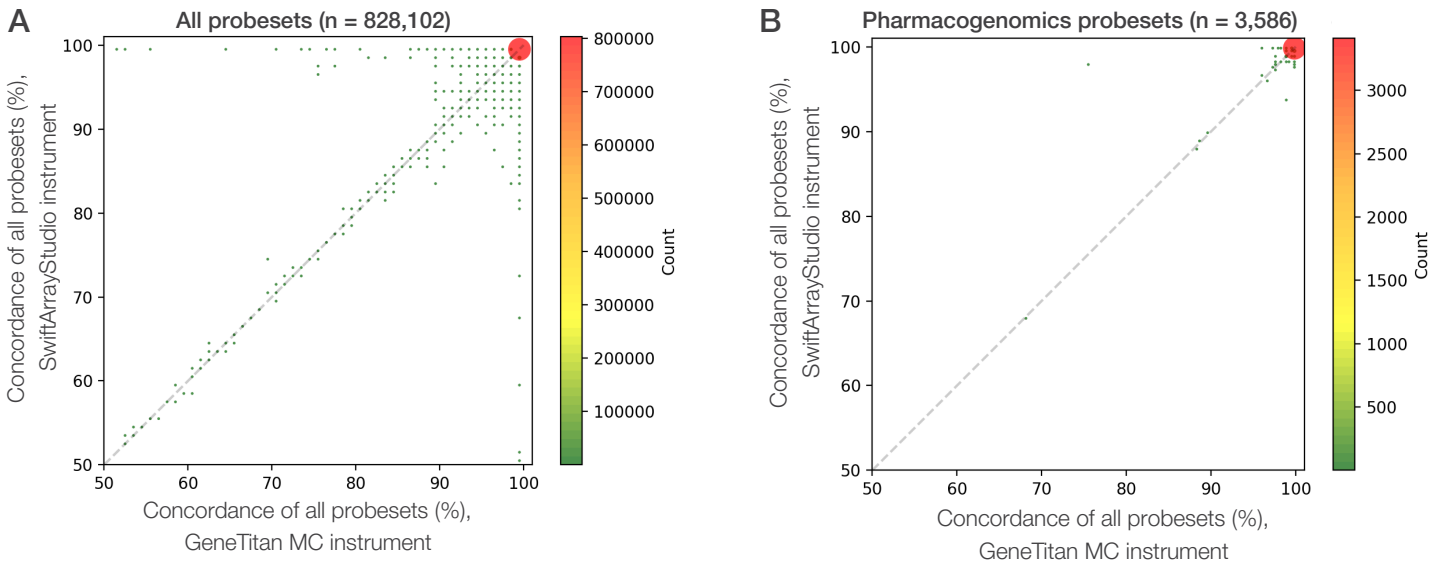


Figure 3. Concordance of genotyping calls made with Axiom SwiftArray Assay and Axiom 2.0 assay workflows. Values for each probeset as measured with the Axiom 2.0 assay on the GeneTitan MC instrument (x-axis) and the Axiom SwiftArray Assay on the SwiftArrayStudio instrument (y-axis) are shown. The number of probesets with a given genotype is colored as indicated on the scale on the right. Each point is a single measurement, with the exception of the red dot (not to scale), which represents the overwhelming majority that have 100% concordance. Many of the probesets with less than 100% concordance fall on the dashed diagonal line, indicating they are equally discordant on the two platforms.

The array design also includes markers for copy number determination at loci relevant to pharmacogenomics, hemoglobinopathies, and other conditions important for clinical research. Copy number concordance at these loci was evaluated using a similar analytical approach. As observed for genotyping, copy number performance was nearly identical between the two platforms (Table 2). These analyses demonstrate that high-quality genotype and copy number data can be generated within 30 hours of initiating target preparation. Furthermore, when compared with the standard Axiom 2.0 assay workflow, the SwiftArrayStudio platform offers excellent concordance for both genotyping and copy number calling. These findings show that the SwiftArrayStudio solution achieves data quality comparable to the Axiom 2.0 assay while reducing turnaround time by more than half. For more details, see the **application note**.

Table 2. Concordance of copy number calls made by Axiom SwiftArray Assay and Axiom 2.0 assay workflows.

	Axiom SwiftArray Assay on the SwiftArrayStudio instrument	Axiom 2.0 assay on the GeneTitan MC instrument
Copy number 0 sensitivity*	99.5	99.5
Copy number 0 specificity*	100.0	100.0
Fixed-region concordance	100.0	100.0
Fixed-region median PPV**	100.0	100.0
Fixed-region median sensitivity	100.0	100.0

* For the purposes of this table, sensitivity is #TP/(#TP + #FN) and specificity is #TN/(#TN + #FP), where #TP (true positive) is number of CN = 0 (homozygous deletions) and confirmed with truth; #FN (false negative) is a call that should be CN = 0, but was not called CN = 0; #TN (true negative) is a sample that is CN >0 and is confirmed with truth; and #FP (false positive) is called as CN = 0 but is not supported by truth.

** PPV = positive predictive value, defined here for fixed regions as #TP/(#TP + #FP). Fixed-region calls are those where copy number is greater than 0.

Multidrop reagent dispenser facilitates high-throughput applications

High-throughput laboratories require scalable workflows that support processing large numbers of samples prior to microarray hybridization. We previously developed a **high-throughput workflow** leveraging the Thermo Scientific™ Multidrop™ Combi Reagent Dispenser to prepare up to eight 96-array plates using the Axiom 2.0 assay workflow. This approach was subsequently adapted for use with the target preparation steps used for the SwiftArrayStudio Microarray Analyzer. In this high-throughput configuration, key steps—including denaturation, amplification, fragmentation, precipitation, and resuspension—are performed with the assistance of Multidrop reagent dispensers and automated plate sealers (Figure 4).

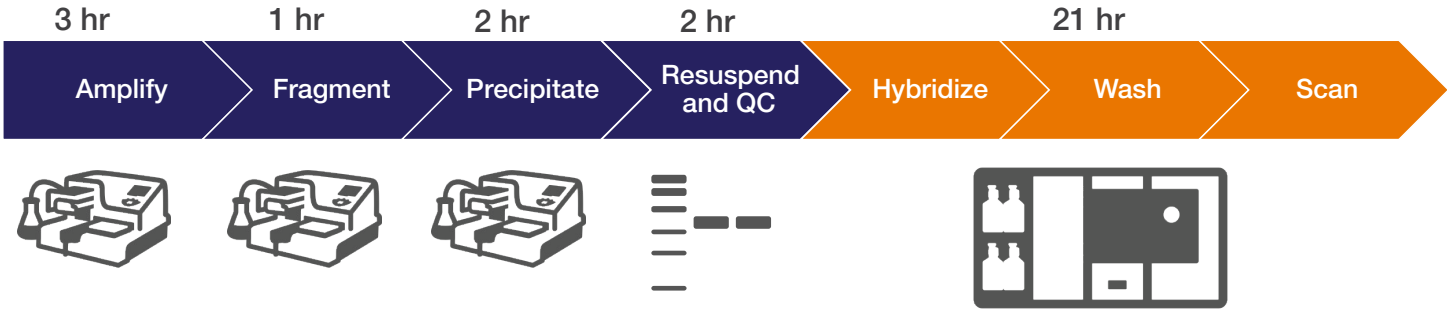


Figure 4. Illustration of the Axiom SwiftArray Assay workflow using Multidrop reagent dispensers. Reagents for the amplification, fragmentation, and precipitation steps are added using Multidrop instruments. After QC of the samples, they are loaded onto the SwiftArrayStudio Microarray Analyzer for automated array processing.

To demonstrate the throughput capacity and reproducibility of this workflow, four replicate plates of Coriell reference samples were prepared using Multidrop reagent dispensers configured for target preparation steps of the Axiom SwiftArray Assay. The plates were then processed with a human genotyping microarray on the SwiftArrayStudio Microarray Analyzer. Average sample concordance relative to known genotype truth was calculated across multiple allele classes, including all alleles, major homozygous alleles, heterozygous alleles, and minor homozygous alleles (Figure 5). Concordance exceeded 99% across all allele classes. In addition, concordance was evaluated for each individual sample across the four replicate plates (Figure 6). When each of the 96 samples was compared across replicates, the average concordance was 99.7%.

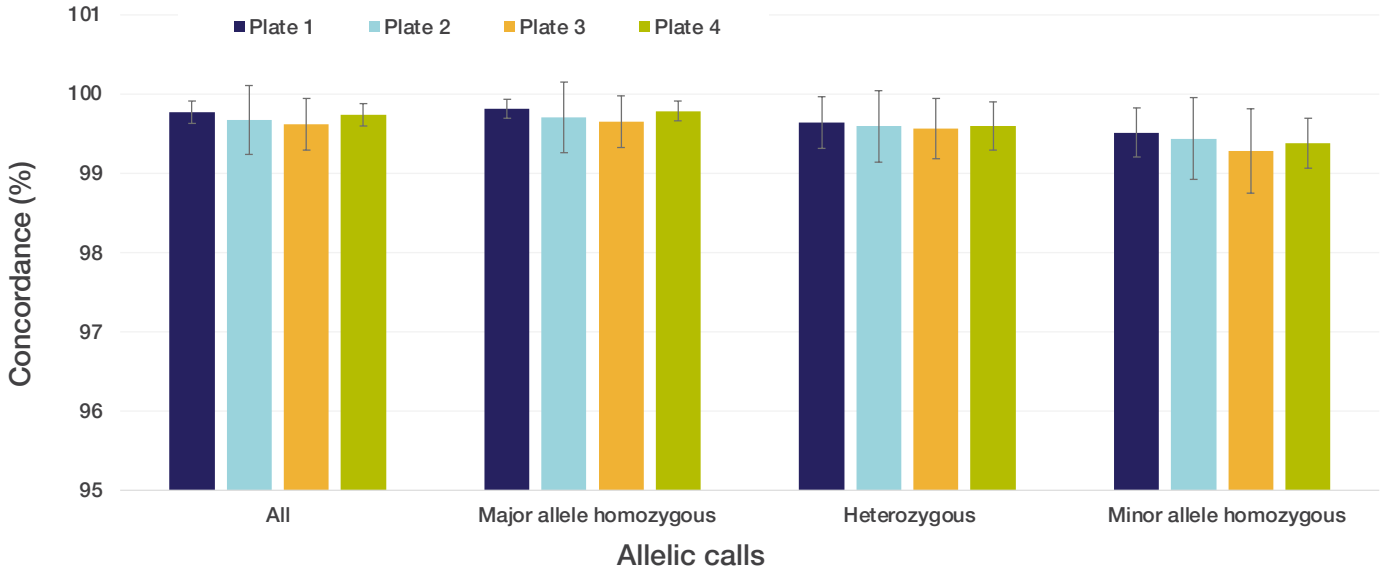


Figure 5. Concordance of allele calls on four replicates of 96-format array plates, processed using the Axiom SwiftArray Assay with reagent dispensing via the Multidrop Combi Reagent Dispenser. Shown are the concordances of all calls, homozygous for major allele calls, heterozygous calls, and homozygous for minor allele calls. Concordance with known genotype in each sample was determined by NGS across more than 837,000 probesets.

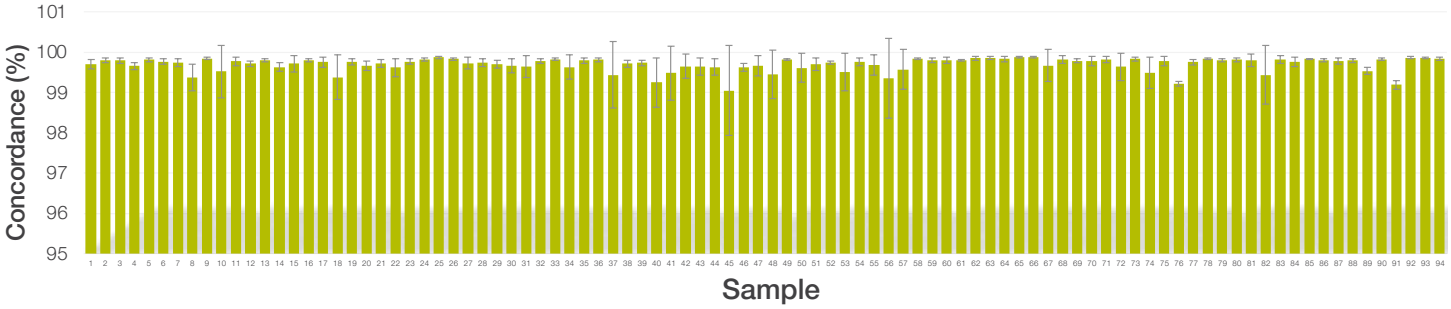


Figure 6. Concordance of all allele calls on four replicates of each sample on separate array plates, processed using the Axiom SwiftArray Assay and automated reagent dispensing. Note that although there is some sample-to-sample variance, in each case the concordance is greater than 99%. Samples with large error bars had one concordance call that was lower than the other three.

High concordance was also observed for copy number variation measurements. For each copy number state evaluated (CN = 0, 1, 2, or 3), sensitivity and specificity were calculated for each plate replicate using Applied Biosystems™ Axiom™ Analysis Suite Software (Figure 7). For CN = 0, 1, and 2, sensitivity exceeded 98.8% and specificity exceeded 99.9%. Sensitivity for CN = 3 was modestly lower (average 97.5% across replicates), likely reflecting the smaller number of CN = 3 events; however, specificity remained high (>99.9%). Importantly, performance was highly consistent across all four plates, with nearly identical sensitivity and specificity values.

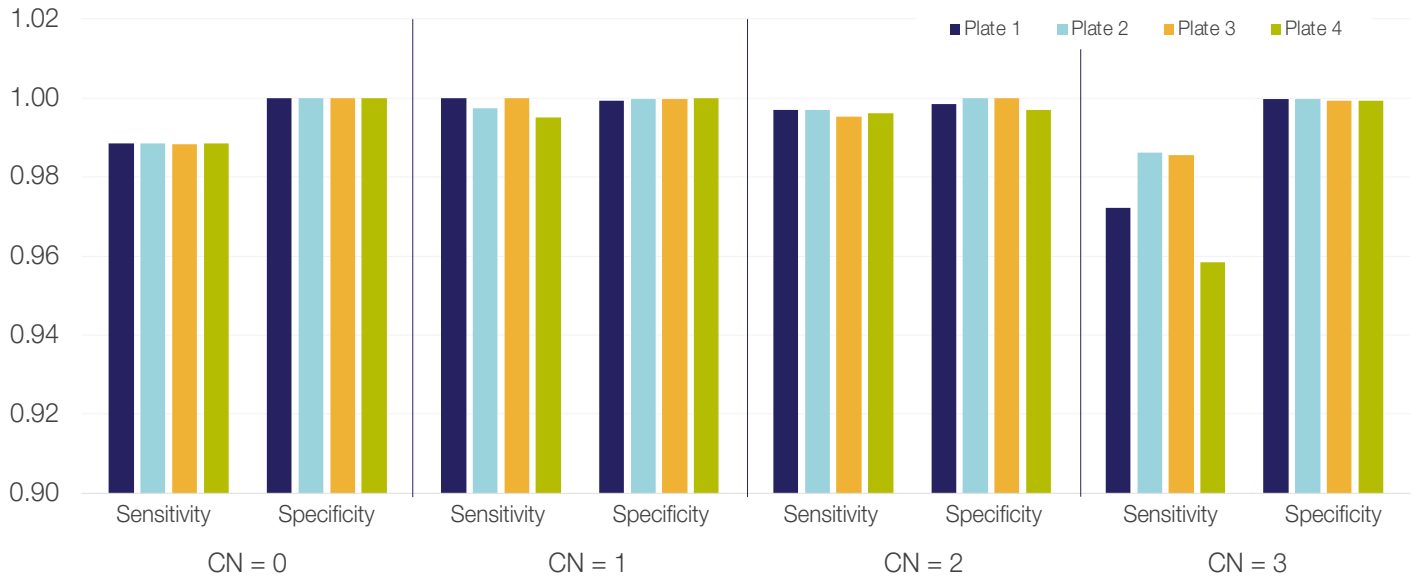
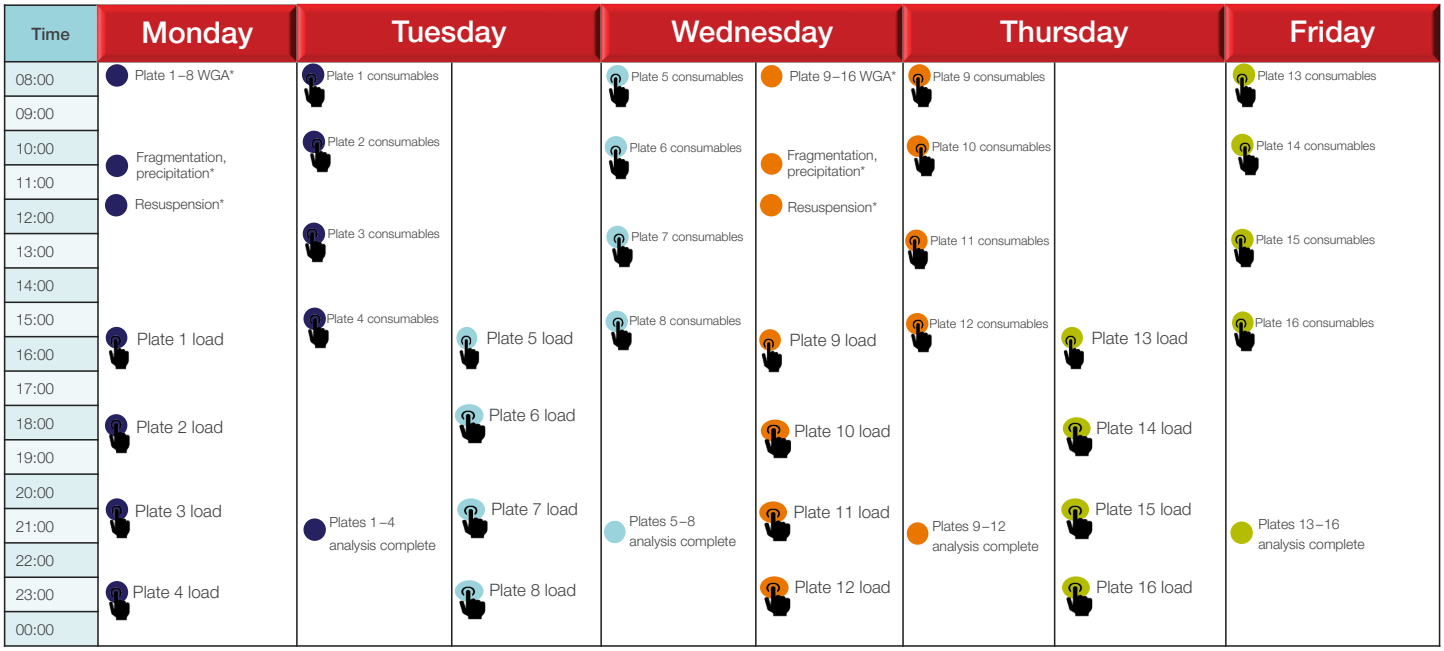


Figure 7. Concordance of copy number calls made on four replicate plates prepared with the Axiom SwiftArray Assay and automated reagent dispensing. For the purpose of this figure, sensitivity is #TP/(#TP + #FN) and specificity is #TN/(#TN + #FP), where #TP is an expected copy number that has been confirmed with the known copy number; #FN is a missed call of the correct copy number; #TN is a call that was accurately called as not having the right copy number; and #FP is a copy number that was not supported by the known copy number.

The combination of reagent dispensing on the Multidrop instrument and efficient target preparation with the Axiom SwiftArray Assay and workflow enables rapid generation of large-scale genotyping data. Using this workflow, targets for up to 8 array plates can be prepared and ready for hybridization on the SwiftArrayStudio Microarray Analyzer within a single day. By staggering plate loading on the SwiftArrayStudio instrument, data from these 8 plates can be generated within approximately 73 hours. Moreover, multiple runs can be initiated within the same week, increasing throughput to as many as 16 plates (1,536 samples using 96-format Axiom arrays; approximately 1.28 billion data points; Figure 8). In addition, prepared targets can undergo quality control (QC) in batches and be stored for up to 10 days, allowing plates that meet QC criteria to be processed on the SwiftArrayStudio Microarray Analyzer as needed.



* Step performed with the Multidrop Combi Reagent Dispenser.

Figure 8. Illustration of a staggered workflow (load later) to process 16 96-sample plates (total of 1,526 samples) in one week with only two touchpoints per plate. Multidrop reagent dispensers can facilitate the WGA, fragmentation and precipitation, and resuspension steps on 8 plates. After resuspension, the hybridization-ready plates can be stored at –20°C until loading onto the SwiftArrayStudio instrument for automated hybridization, washing, and scanning. Hand icons indicate interaction with the SwiftArrayStudio instrument.

Integration of the Multidrop Combi Reagent Dispenser with the Axiom SwiftArray Assay supports high-throughput genotyping by automating many of the fluid-handling steps required for the SwiftArrayStudio Microarray Analyzer. This combined solution enables laboratories to scale target preparation, improve operator ergonomics, and reduce pipetting variability, while maintaining confidence in the generation of high-quality data within a shortened turnaround time. For more details and a complete protocol, see the **application note**.

Automation with liquid-handling robots reduces manual steps

When processing large sample volumes, translational and clinical researchers must ensure that all liquid-handling steps are performed with a high degree of accuracy, as even minor errors can substantially affect downstream results. Consequently, many laboratories are adopting automated liquid-handling platforms to reduce the risk of human error. Automation minimizes errors associated with manual pipetting, such as inaccurate volume transfer or inadvertent omission of critical steps, and reduces run-to-run variability introduced by complex, repetitive manual workflows. In addition, automation allows laboratory personnel to focus on other tasks while instruments are operating, thereby improving overall laboratory efficiency and reproducibility.

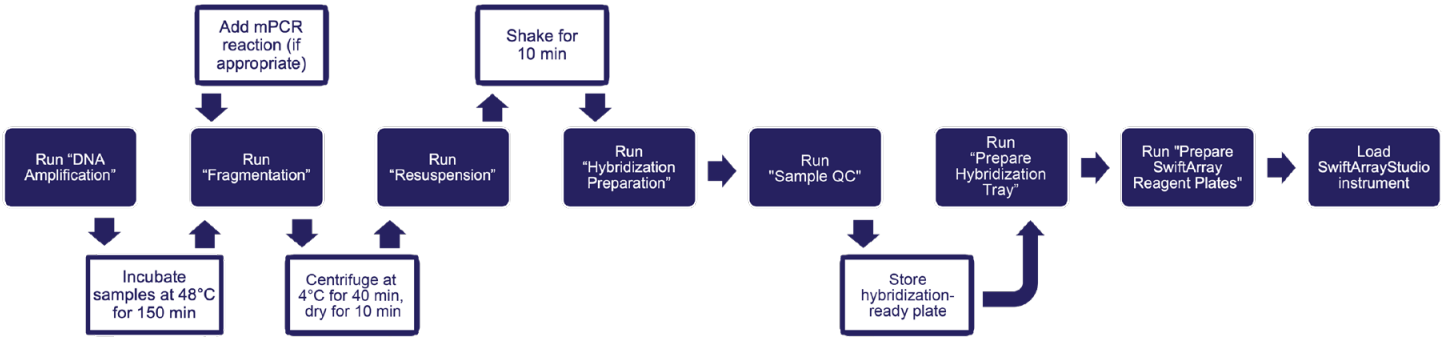


Figure 9. Workflow for running the Axiom SwiftArray Assay on the NIMBUS instrument. Each filled box represents a separate sub-method of the automated protocol on the NIMBUS instrument. Unfilled boxes are steps run off-instrument. Note that we recommend that the "Prepare Hybridization Tray" and "Prepare SwiftArray Reagent Plates" sub-methods be performed off-instrument. Overall, the process takes about 5 hours.

Thermo Fisher Scientific previously developed an **automated target preparation protocol** for the Axiom 2.0 assay on the Applied Biosystems™ NIMBUS® Target Preparation Instrument. Developed by Hamilton Robotics, the NIMBUS instrument is a compact, automated high-speed multichannel pipetting robot that is widely recognized for its reliable pipetting performance and durability. We adapted the previous protocol to support the target preparation steps of the Axiom SwiftArray Assay. The resulting workflow divides the Axiom SwiftArray Assay into seven discrete sub-methods. In brief, samples and consumables are loaded onto the NIMBUS instrument, and the amplification sub-method is initiated (Figure 9). Upon completion, samples are either transferred to the on-instrument 48°C heat block or placed in a 48°C oven for 150 minutes. The amplified samples are then returned to the instrument for execution of the fragmentation sub-method. Following fragmentation, samples are centrifuged and dried off-instrument. Reagents for pellet resuspension are added during the third sub-method, followed by off-instrument shaking. The fourth sub-method prepares plates for hybridization. These hybridization-ready plates can serve as the source for pre-loading quality control of samples (fifth sub-method) or be stored until needed. Finally, plates destined for loading onto the SwiftArrayStudio Microarray Analyzer are prepared either manually (off-instrument) or using the sixth and seventh sub-methods. The complete target preparation workflow requires approximately 6 hours, with minimal hands-on time limited to transferring plates between steps and loading or unloading the NIMBUS instrument between sub-methods.

To demonstrate the utility of the automated target preparation workflow for comprehensive pharmacogenomic coverage, we analyzed 96 well-characterized human genomic DNA reference samples from the Coriell Research Institute (part of the Applied Biosystems™ PharmacoScan™ Training Kit, 96-format, Cat. No. 913027). The Applied Biosystems™ Axiom™ PharmacoPro™ SwiftArray™ Kit (Cat. No. 952647), which includes the Applied Biosystems™ Axiom™ PharmacoPro™ Array and Axiom™ SwiftArray™ reagents, were used for the analysis. Workflow accuracy was assessed by comparing genotype calls from two replicate plates against sequencing-based reference genotypes (Figure 10). For plate 1, the average concordance across all samples was 99.85%, while plate 2 showed an average concordance of 99.83% (median concordance for both plates was 99.86%). Reproducibility was further evaluated by performing pairwise comparisons of concordance across loci for each sample between the two plates. The resulting measurements were highly consistent, with an average concordance of 99.996%. Although concordance decreased slightly with lower call rates, no sample exhibited a call rate below 99.43%, and overall data quality remained high.

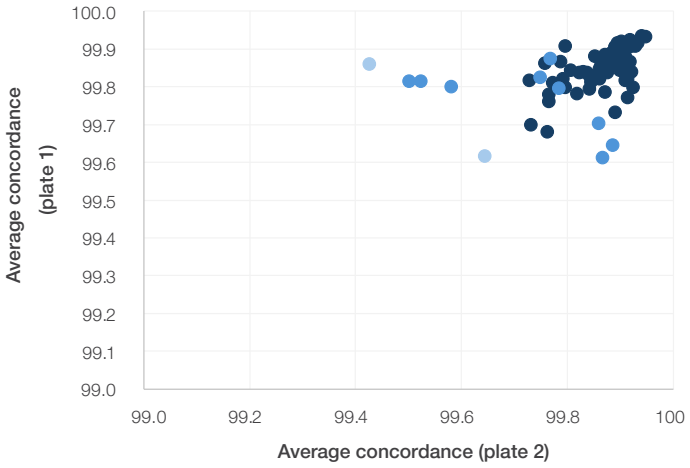


Figure 10. Concordance with known truth of genotyping calls made with the Axiom SwiftArray Assay with target preparation on the NIMBUS instrument. All loci, including those queried by multiplex PCR (mPCR), were compared against known truth. The concordance of each sample correlates extremely well across the two replicates (R^2 >0.999). Data points are colored according to total call rate (CR): dark blue = CR >99.9%, medium blue = CR >99.75%, light blue = CR >99.5%.

Accuracy and reproducibility of copy number variant (CNV) calls were also evaluated relative to known reference values. For accuracy, sensitivity and specificity were calculated for each CNV region interrogated by the array. Average sensitivity was 98.8% for both plates, while average specificity was 99.6%. Reproducibility was assessed by correlating the number of samples assigned to each copy number class at each locus between the two plates. The resulting correlation coefficient was 0.9963, indicating highly consistent CNV classification across replicates.

One of the advantages of automation is that the liquid handler can be used to process more samples when it would otherwise be idle (Figure 11). For example, three hybridization-ready plates (288 samples) can be prepared in a single day with normal work shifts. One of these can be loaded into the SwiftArrayStudio Microarray Analyzer; a second plate can be loaded in a longer workday. Plates that are made hybridization-ready using the NIMBUS instrument can be stored at –20°C for up to 10 days before analyzing on the SwiftArrayStudio Microarray Analyzer.

Scenario 1: trays prepared manually

	Run 1	Run 2	Run 3
9:00	DNA Amplification		
9:15			
9:30			
9:45	Sample incubation at 48°C	DNA Amplification	
10:00			
10:15			
10:30		Sample incubation at 48°C	DNA Amplification
10:45			
11:00			
11:15			Sample incubation at 48°C
11:30			
11:45			
12:00			
12:15			
12:30	Fragmentation		
12:45			
13:00			
13:15	Centrifugation at 4°C and drying	Fragmentation	
13:30			
13:45			
14:00		Centrifugation at 4°C and drying	Fragmentation
14:15			
14:30			
14:45	Resuspension		Centrifugation at 4°C and drying
15:00	Hybridization Preparation		
15:15		Resuspension	
15:30	Sample QC		
15:45			Resuspension
16:00	Tray preparation	Hybridization Preparation	
16:15	Load analyzer	Sample QC	
16:30			Hybridization Preparation
16:45			Sample QC
17:00			Run later
17:15			
17:30			
17:45			
18:00			
18:15			
18:30		Tray preparation	
18:45		Load analyzer	
19:00			
19:15			
19:30			
19:45			
20:00			
20:15			

Scenario 2: trays prepared on-instrument

	Run 1	Run 2	Run 3
9:00	DNA Amplification		
9:15			
9:30			
9:45	Sample incubation at 48°C	DNA Amplification	
10:00			
10:15			
10:30		Sample incubation at 48°C	DNA Amplification
10:45			
11:00			
11:15			Sample incubation at 48°C
11:30			
11:45			
12:00			
12:15			
12:30	Fragmentation		
12:45			
13:00			
13:15	Centrifugation at 4°C and drying	Fragmentation	
13:30			
13:45			
14:00		Centrifugation at 4°C and drying	Fragmentation
14:15			
14:30			
14:45	Resuspension		Centrifugation at 4°C and drying
15:00	Hybridization Preparation		
15:15		Resuspension	
15:30	Sample QC		
15:45			Resuspension
16:00	Prepare Hybridization Tray		
16:15	Prepare SwiftArray Reagent Plates		
16:30			
16:45	Load analyzer	Hybridization Preparation	
17:00		Sample QC	
17:15			Hybridization Preparation
17:30			Sample QC
17:45			Run later
18:00			
18:15			
18:30		Prepare Hybridization Tray	
18:45		Prepare SwiftArray Reagent Plates	
19:00			
19:15		Load analyzer	
19:30			
19:45			
20:00			
20:15			

Scenario 3: full run on-instrument

	Run 1	Run 2
9:00	DNA Amplification	
9:15		
9:30		
9:45	Sample incubation at 48°C	
10:00		
10:15		
10:30		
10:45		
11:00		
11:15		
11:30		
11:45		
12:00		
12:15	Fragmentation	
12:30		
12:45		
13:00	Centrifugation at 4°C and drying	
13:15		
13:30		
13:45		
14:00	Resuspension	
14:15		DNA Amplification
14:30		
14:45		
15:00		Sample incubation at 48°C
15:15		
15:30		
15:45		
16:00		
16:15		
16:30		
16:45		
17:00		
17:15		
17:30		Fragmentation
17:45		
18:00		
18:15	Hybridization Preparation	Centrifugation at 4°C and drying
18:30	Sample QC	
18:45	Prepare Hybridization Tray	
19:00	Prepare SwiftArray Reagent Plates	
19:15		
19:30		Resuspension
19:45	Load analyzer	Hybridization Preparation
20:00		Sample QC
20:15		Run later

Figure 11. Scenarios for multiple runs on a single NIMBUS instrument. In scenario 1, staggering the runs on instrument can allow up to three plates of 96 samples each to be made ready for hybridization and quality analysis. By manually preparing the stain and ligation trays (tray preparation), two array plates can be run 2.5 hours apart. In scenario 2, the stain and ligation trays are prepared on the instrument (Prepare Hybridization Tray and Prepare SwiftArray Reagent Plates). Again, up to three sets of 96 samples can be made ready, but in a slightly longer workday. In scenario 3, all steps, including genome amplification, are run on the instrument. This requires the least amount of active monitoring, but only results in two sets of 96 samples, ready for hybridization, in a long workday. Other scenarios not indicated here are also possible. Filled boxes represent steps performed on-instrument, and unfilled boxes represent steps performed off-instrument. The green, yellow, blue, and red colors align with cap colors used in the Applied Biosystems™ Axiom™ SwiftArray™ Reagent Kit at each sub-method. For the Sample QC and Prepare Hybridization Tray sub-methods (gray), all materials are user-supplied.

Although this study utilized the Axiom PharmacoPro Array, the automated script used for the NIMBUS instrument is broadly applicable to other 96-format Axiom arrays in combination with the SwiftArrayStudio instrument. Collectively, this workflow demonstrates how automation can enhance data accuracy, reproducibility, and laboratory efficiency when high-throughput genotyping applications are required. For more information on using the NIMBUS instrument together with the SwiftArrayStudio solution, see the **application note**.

SwiftArrayStudio solution for nonhuman genotyping applications

Our animal companions are an essential part of our lives. Because they mean so much to us, we want to understand as much as possible about their health. Advances in genotyping help identify deleterious alleles that can be important for understanding breeds and for supporting veterinary care research. Thermo Fisher Scientific recently released **high-throughput genotyping microarrays for feline and canine genome analysis**. These arrays include over 650,000 markers that support the study of genetic variation, trait-associated regions, and breed composition in feline and canine genomes.

To demonstrate the applicability of the SwiftArrayStudio solution to companion animal genotyping, we evaluated its performance using canine and feline samples. To reflect real-world sample collection conditions, buccal swab samples were obtained from colleagues' cats and dogs using Thermo Scientific™ GenoTube™ Livestock Swabs. Genomic DNA was purified from the swabs using a modified protocol of the Invitrogen™ RecoverAll™ Total Nucleic Acid Isolation Kit for FFPE. For each sample, up to 100 ng of DNA was processed using the manual Axiom SwiftArray Assay workflow, and each sample set was analyzed in triplicate. For samples in which 100 ng of input DNA was not available, the maximum allowable input volume (20 µL) was used.

Overall, high-quality genotyping data were generated for the majority of samples (Table 3). Among the canine samples, five failed the DQC metric, corresponding to approximately a 93% pass rate; samples that passed exhibited an average call rate of 99%. Notably, this group included several samples with substantially less than the recommended 100 ng DNA input. Similar results were observed for feline samples, with seven samples failing DQC (approximately 92% pass rate) and passing samples achieving an average call rate of 99.5%.

Table 3. Performance of the SwiftArrayStudio solution for feline and canine microarrays.*

	Feline	Canine
Total number of markers	662,069	712,332
Number of input samples	288	287
Samples with Dish QC (DQC) ≥0.82	93.40%	93.73%
DQC median	0.997	0.999
Average passing QC call rate	99.54%	98.96%
Average sample call rate	99.69%	99.51%
Percent of markers with high performance in dataset	97.20%	93.80%
Average sample reproducibility	99.99%	99.93%
Gender calls		
Female	136	110
Male	118	120
Gender call rate	100%	100%
Displayed values in Axiom Analysis Suite Software		
PolyHighResolution	89.29%	69.83%
NoMinorHom	5.59%	12.79%
MonoHighResolution	2.30%	11.15%

* Values shown are the average of three triplicate plates for analysis of buccal swabs of pet DNA. For more details about the metrics, see the Axiom Analysis Suite v4.0.1 **user guide**.

Assessment of genotypic clustering quality showed that 91.5% of canine markers were classified as polymorphic high-resolution, no minor homozygotes, or monoallelic high-resolution, while 95.4% of feline markers fell into these informative clustering categories. These results demonstrate that the SwiftArrayStudio solution can generate robust and reliable genotyping data from noninvasive buccal samples collected under typical field conditions in companion animals.

These results facilitated the identification of breeds and traits in the animals. A representation of the most common breed loci identified in the example genomic DNA from dogs is shown in Table 4, and a more detailed genotypic output from selected cat traits is shown in Table 5. Note that for both the canine and feline arrays, more than 640,000 markers were analyzed; the results shown here are a small fraction of the total collected. For more information on the feline and canine microarrays, visit our companion animal genotyping **web page**.

Table 4. Top 20 dog breeds represented in colleagues' pets.

Breed*	Number
Toy poodle	12
Golden retriever	11
Vizsla	9
American cocker spaniel	8
East-European shepherd	8
American Staffordshire terrier	7
Chihuahua	7
German shepherd	7
Labrador retriever	7
Poodle	7
English shepherd	5
Siberian husky	5
Staffordshire bull terrier	5
Akbash	4
Central Asian shepherd	4
Italian greyhound	4
Australian cattle dog	3
Blue Gascony griffon	3
Bull terrier	3
English cocker spaniel	3

* Most breed results are due to mixtures of characteristic loci detected in the dogs, not necessarily pure breeds.

Table 5. Tally of selected cat traits in a collection of 92 cats.

Trait*	Gene	No. of reference homozygous	No. of heterozygous	No. of alternate homozygous
Burmese coat color pattern	*TYR*	90	1	1
Ticked	*DKK4*	85	4	3
Polydactyly	*LMBR1*	90	2	0
Sphynx hairless	*KRT71*	70	19	1
Chocolate	*TYRP1*	85	7	0
Coat color, dilute	*MLPH*	35	33	24
Long hair	*FGF5*	64	25	3
Long hair	*FGF5*	49	32	11
Blotched tabby	*LVRN*	22	41	28
Coat color, non-agouti (black)	*ASIP*	8	35	49
Coat color, salmiak	*KIT*	0	0	0
Curly coat, Devon rex	*KRT71*	66	12	10
Curly coat, Ural rex	*LIPH*	92	0	0
Coat color, russet	*MC1R*	92	0	0
Coat color, copal	*MC1R*	92	0	0
Tail, short	*TBXT*	92	0	0
Tail, short	*TBXT*	91	1	0
Munchkin standard	*UGDH*	0	0	0

* Some traits are listed twice; this due to the two different alleles of the same gene being examined. Note that this is a subset of the more than 640,000 genotypes analyzed on the array.

Conclusions

Thermo Fisher Scientific is a global leader in developing genetic analysis solutions. The SwiftArrayStudio Microarray Analyzer represents our latest innovation in microarray analysis technologies and was designed for ease of use and reliability. As part of the SwiftArrayStudio solution, the Axiom SwiftArray Assay streamlines the workflow, reducing the time needed for target preparation from 3 days to a single day. The latest sequencing information is incorporated into modern Axiom microarrays, including the latest human, feline, and canine arrays. These innovations provide genotyping performance that is comparable to that of previous versions of these tools. The SwiftArrayStudio solution also offers flexibility in throughput. For users requiring single-plate processing options, it offers a rapid and easy-to-use workflow. For users who need high-throughput options, it is compatible with high-capacity and automated liquid-handling instruments. The SwiftArrayStudio solution is therefore poised to provide basic, translational, and clinical researchers with tools that can generate high-quality genotyping information rapidly, accurately, and reliably.

Find out more at **thermofisher.com/swiftarraystudio**