Efficient analysis of bovine genotypes for researching herd fitness, breeding choices, and dairy production

The Applied Biosystems™ Axiom™ SwiftArray™ Bovine 384F solution is a comprehensive platform for high-throughput analysis of selected bovine genetic variants. The solution includes Applied Biosystems™ SwiftArray™ Assay reagents, along with Applied Biosystems™ Axiom™ microarrays in a 384-peg format with flexible sample throughput options. The array content was developed in collaboration with key opinion leaders in bovine husbandry to support effective breeding strategies. Additional single-nucleotide polymorphisms (SNPs) and copy number variation (CNV) markers have been incorporated to enhance genomic selection, parentage verification, and identification of economically important production traits for routine genotyping applications.

Highlights

- **Comprehensive genomic coverage:** Includes approximately 64,000 SNPs providing extensive coverage for both *Bos taurus* and *Bos indicus* breeds.
- **Recessive trait screening:** Contains markers associated with deleterious recessive traits to support herd management.
- **CNV detection:** Includes probes targeting CNV regions for researching their impact on bovine health and productivity traits.
- **CDCB-verified markers:** Incorporates 44,887 markers developed in collaboration with the Council on Dairy Cattle Breeding (CDCB).
- **Fertility and traceability markers:** Includes markers on the Y chromosome and mitochondrial genome to research reproductive efficiency, investigate fertility, and lineage traceability.
- **High-throughput workflow:** Capable of generating up to 393 million SNP determinations for 6,144 samples in five working days using a single Applied Biosystems™ SwiftArrayStudio™ system.

Table 1. Content present on the Axiom SwiftArray Bovine 384F Microarray

	Number of probesets
Total bovine markers	64,720
CDCB verified markers	44,887
Bos taurus markers	51,442
Bos indicus markers	13,278

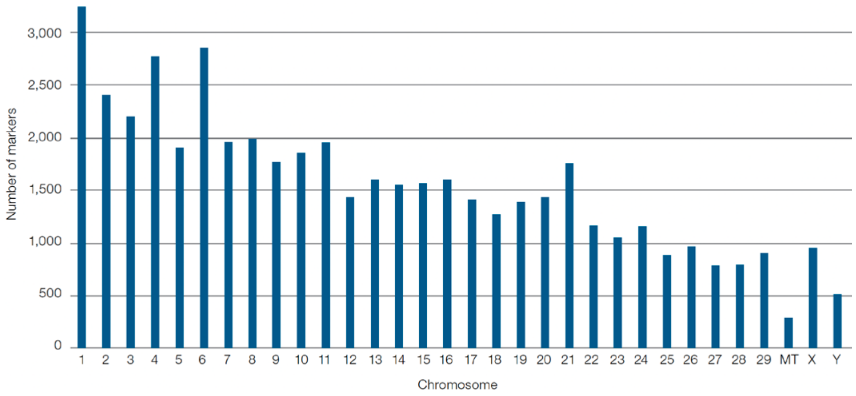

Figure 1. Distribution of markers on the Axiom SwiftArray Bovine 384F Microarray across the bovine genome.

Enhanced array content

The Applied Biosystems Axiom SwiftArray Bovine 384F array was developed in collaboration with experts in bovine genetics and husbandry. Most SNPs were selected for uniform genomic coverage across both *Bos taurus* and *Bos indicus* breeds (Figure 1). The array contains 44,887 markers developed in collaboration with CDCB and includes more than 13,000 *Bos indicus* SNPs with demonstrated high minor allele frequency (MAF >0.35) in Nellore cattle from India (Table 1).

Breeds and phenotypes covered

Different breeds have been selected for specific traits such as milk production, meat quality, fertility, or climate adaptability, and thus establishing breed identity is important in bovine husbandry. The array includes ancestry informative markers for multiple breeds, including Ayrshire, Brown Swiss, Holstein, Jersey, and others (Table 2). The panel also contains loci associated with breed-specific lethal traits, economically important production traits, and indicators of overall animal well-being. These features help producers research breeding decisions, maintain genetic diversity, and improve market value by helping ensure animals meet industry and consumer standards.

Table 2. Dairy breeds and phenotypic traits covered on the Axiom SwiftArray Bovine 384F Microarray

Breed-specific Markers	Number of probesets	Trait-specific markers	Number of probesets
Ayrshire	12	Color	10
Brown Swiss	182	Meat	19
Guernsey	21	Milk	62
Holstein	246	Polled	2
Hereford White Face	578	Lethal	265
Jersey	207	Undesirable	55
		Beneficial	359

Table 3. Copy number variants covered on the Axiom SwiftArray Bovine 384F Microarray

Copy number variant examined	Number of probesets
CNV Total – Non-lethal	578
CNV Total – Lethal	506
White Face	777
Brachyspina	428
Osteopetrosis	487

Copy number variants

Copy number variants are an important source of genetic variation associated with complex traits in cattle. The Axiom SwiftArray Bovine 384F array contains more than 1,000 CNV probes (Table 3), including markers associated with coat coloration, osteopetrosis, and brachyspina. Integrated CNV analysis within Applied Biosystems™ Axiom™ Analysis Suite 6.0 software and higher enables efficient assessment of CNVs relevant to bovine health and economically important traits.

Coverage of pathogenic markers

The Axiom SwiftArray Bovine 384F array supports the research of numerous congenital and inherited diseases, including metabolic disorders, muscular disorders, developmental abnormalities, and ocular diseases (Reference [1] and Table 4). Additionally, the array includes more than 500 Y-chromosome markers, enabling prediction of haplotypes associated with embryo loss or perinatal mortality in cattle populations.

Table 4. Pathologies covered on the Axiom SwiftArray Bovine 384F Microarray

Pathogenic marker	Pathogenic marker	Pathogenic marker
Alpha Mannosidosis	Congenital hypotrichosis (Rat-tail Syndrome)	Maple Syrup Urine
Arachnomelia	Congenital Muscular Dystonia 1	Mulefoot
Axonopathy	Congenital Muscular Dystonia 2	Neuronal Ceroid Lipofuscinosis
Beta Mannosidosis	Congenital Myoclonus	Osteopetrosis
Bovine Leukocyte Adhesion Deficiency	Crooked Tail Syndrome	Paunch Calf Syndrome
Brachyspina	Deficiency of Uridine Monophosphate Synthase	Protoporphyria
Bulldog Dwarfism	Developmental Duplication	Pseudomyotonia
Cardiomyopathy and Woolly Haircoat Syndrome	Dystrophic Epidermolysis Bullosa	Growth Retardation
Chediak Higashi Syndrome	Factor X1 Deficiency	Spinal Muscular Atrophy
Citrullinaemia	Hypotrichosis	Thrombopathia
Complex Vertebral Malformation	Infectious Bovine Keratoconjunctivitis	

Table 5. Ancestry and inheritance markers on the Axiom SwiftArray Bovine 384F Microarray

Parentage tracking	No. of markers
ISAG Parentage	246
STR Imputation	1062
Y SNPs	569
Mitochondrial SNPs	*292*

Ancestry and inheritance markers

Parentage tracking and informed mating decisions are critical requirements for herd genotyping. The International Society of Animal Genetics (ISAG) has recommended a standard set of bovine markers for inheritance tracking. All core and extended ISAG parentage markers are included on the array for reliable parentage verification [2]. The Axiom SwiftArray Bovine 384F array also includes a large set of SNPs optimized for short tandem repeat (STR) imputation, supporting the transition from STR-based testing to array-based testing while maintaining compatibility with historical STR datasets [3].

Throughput

The Axiom™ 384HT format is designed for automation-friendly, high-throughput processing. Thermo Scientific™ MultiDrop™ Combi Reagent Dispensers can be integrated with the **Axiom™ SwiftArray™ Assay workflow and the Applied Biosystems™ SwiftArrayStudio™ Microarray Analyzer**. The workflow includes target preparation that can be completed within a single workday, followed by automated hybridization, washing, and scanning on the SwiftArrayStudio™ system, generating genotype results within approximately 38 hours. By combining automated reagent dispensing with streamlined processing, the overall sample-to-answer timeline can be reduced from five days to two days (Figure 2). Staggered loading on the SwiftArrayStudio™ instrument enables processing of up to 16 plates per five-day workweek. Prepared samples may also be stored at –20°C for future processing as needed.

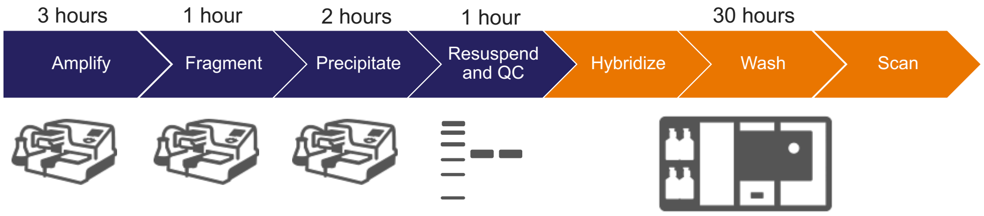

Figure 2. Illustration of the SwiftArray Assay workflow using MultiDrop Reagent Dispensers. Reagents for the amplification, fragmentation, and precipitation steps are added using the MultiDrop instruments. After resuspension and QC of the samples, they are loaded onto the SwiftArrayStudio instrument for automated array processing.

Performance

Thermo Fisher Scientific maintains rigorous performance standards for all microarray products, and the Axiom SwiftArray Bovine 384F microarray meets or exceeds these criteria. These arrays were extensively verified using 384 bovine samples across five replicate arrays. Genomic DNA samples were processed with three different lots of reagents, and data were collected on three different SwiftArrayStudio™ Microarray Analyzers. The **Axiom™ Analysis Suite software**, which automatically calculates quality-control metrics, including dishQC, QC call rates, and reproducibility measurements, was used to evaluate the performance. The arrays demonstrated high DishQC values, high QC call rates, 100% gender-call accuracy, and 100% sample reproducibility (Table 6 and Figure 3). In addition, CDCB marker call rates consistently exceeded 99.7%. CNVs are also automatically analyzed within Axiom™ Analysis Suite, enabling reliable detection of variants associated with researching important production and for fitness traits (Figure 4).

Table 6. Quality metrics obtained

Metric	Plate 1*	Plate 2	Plate 3	Plate 4	Plate 5
Percent passing DishQC and QC call rate	99.5	99.0	99.2	100.0	99.6
Mean QC call rate for samples passing DishQC and QC call rate	99.9	99.9	99.9	99.9	99.9
Percent samples with gender call	100	100	100	100	100
Mean sample reproducibility	100	100	100	100	100
Mean call rate for CDCB markers	99.9	99.9	99.8	99.9	99.9

* Plates 1-3 analyzed 384 samples, Plates 4-5 analyzed 288 samples

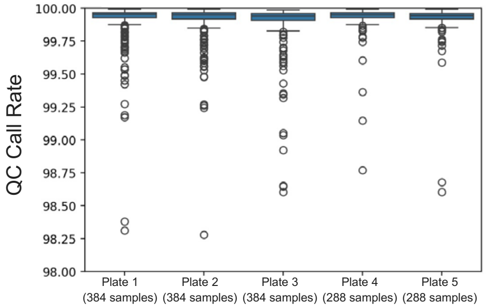

Figure 3. QC call rates on Axiom SwiftArray Bovine 384F microarrays, prepared with MultiDrop reagent dispensers and analyzed on SwiftArrayStudio Microarray Analyzer. Thresholds used for allele calling were defined in the default Best Practices workflow of Axiom Analysis Suite.

Figure 4. Copy number determination for four traits contained on the Axiom SwiftArray Bovine 384F microarray. Thresholds used for copy number calling were defined in the default Best Practices workflow of Axiom Analysis Suite.

Summary

The Axiom SwiftArray Bovine 384F Microarray and SwiftArrayStudio™ platform provide a robust and efficient solution for high-throughput bovine genotyping. The array includes 44,887 CDCB-verified markers and supports genotyping for both *Bos taurus* and *Bos indicus* cattle. The platform includes markers associated with deleterious recessive traits, CNVs, fertility, parentage, reproductive efficiency, and economically important production characteristics. Combined with automated sample preparation using the Thermo Fisher Scientific™ MultiDrop™ Combi Reagent Dispenser and data acquisition using the SwiftArrayStudio™ Microarray Analyzer, the workflow enables high-throughput processing of up to 6,144 samples and approximately 393 million SNP determinations in five days on a single system.

References

1. McClure M, McClure J (2016) Understanding genetics and complete genetic disease and trait definition. https://www.icbf.com/wp/wp-content/uploads/2014/06/Farmer-Genetic-Disease-and-Trait-Information-for-IDB-Genotyped-Animals-in-Ireland_9_20_16.pdf.
2. International Society for Animal Genetics. www.isag.us/committees.asp?autotry=true&ULnotkn=true.
3. McClure MC, et al. (2013) Imputation of microsatellite alleles from dense SNP genotypes for parentage verification across multiple Bos taurus and Bos indicus breeds. Front. Genet. 4:176. doi: 10.3389/fgene.2013.00176

Ordering information

Description	Cat. No
Axiom™ SwiftArray™ Bovine 384F microarray	551718
Axiom™ SwiftArray™ Reagent Kit	952691
Axiom™ SwiftArrayStudio™ Consumables Kit	952680

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