ThermoFisher SCIENTIFIC

Agrigenomics

Enabling effective breeding programs with verified beef and dairy content

Axiom Bovine v3 array includes coverage of copy number variants and economically associated traits

The Applied Biosystems™ Axiom™ Bovine Genotyping v3 Array (available in 384HT and mini 96-well formats) is a third-generation bovine array on the Axiom genotyping platform. The content on the array was selected by key opinion leaders to enable effective breeding, and it was further enhanced with the inclusion of additional single-nucleotide polymorphisms (SNPs) and copy number variations (CNVs) for dairy evaluation, genomic selection, and parentage verification for routine use.

Highlights

Added informative content: The array includes 64,000 SNPs providing high coverage for *Bos taurus* and *Bos indicus* breeds.

Recessive traits: Inclusion of content associated with deleterious recessive traits for herd management.

CNV detection: Inclusion of probes in CNV regions to assess the impact of CNVs on bovine health and production traits.

CDCB verified markers: Inclusion of 44,887 markers that have been verified in collaboration with the Council on Dairy Cattle Breeding (CDCB).

Markers for fertility traits and traceability: The array includes markers on the Y and mitochondrial chromosomes to determine reproductive efficiency, fertility, and traceability.

applied biosystems

Enhanced array content and multiple formats

The content on the Axiom Bovine v3 array was selected in collaboration with key opinion leaders in the bovine community. A majority of the SNPs were chosen for uniform genomic coverage across the *Bos taurus* and *Bos indicus* breeds. The array has over 48,000 markers in common with the Applied Biosystems™ Axiom™ Bovine Genotyping v1 Array, including content that has a high call rate across multiple sample types. The array offers flexibility in sample processing with availability in both Axiom 384HT and mini 96-well formats. The Axiom 384HT format is automation friendly, while the Axiom mini 96-well format uses a manual assay for sample processing, allowing laboratories to choose a workflow that is suitable for their needs. The manufacturing technology of Axiom genotyping arrays produces 100% fidelity; all markers designed on the Axiom Bovine v3 array are present in every manufacturing batch. This helps ensure that all markers important in cattle breeding and parentage analysis are available on every array.

Coverage of relevant diseases and traits of economic importance

All core and extended parentage markers recommended by the International Society of Animal Genetics [1] are included on the array for parentage verification.

The array also includes SNPs that have been shown to have an impact on various diseases and conditions. The various traits covered in the array include polled variety, milk-specific traits, metabolic disorders, muscle function, and limb and ocular diseases [2].

Additionally, the array includes over 500 markers on the Y chromosome to offer the unique capability of prediction of specific haplotypes that are associated with embryo loss or perinatal mortality in cattle. Such markers are valuable for identifying the haplotypes or alleles, which can be transmitted from the parent to the offspring.

Haplotypes are also often used to characterize coat color, polledness, and genetic conditions in a variety of dairy breeds. Several haplotype tests for multiple breeds such as Ayrshire, Brown Swiss, Holstein, and Jersey are on the array and listed in Table 1.

Table 1. List of haplotypes for each breed.

Breed	Haplotype
Ayrshire	AH1
Brown Swiss	BH1, BH2, BHD, BHP, BHW
Holstein	HDR, HH1, HH2, HH3, HH4, HH5, HHP
Jersey	JH1, JHP

The Axiom Bovine v3 array includes several hundred SNPs that are optimized for short tandem repeat (STR) imputation. The inclusion of these SNPs will enable the transition from STR-based testing to array-based testing, providing access to relevant content in cattle breeding while maintaining the ability to extract information through imputation associated with STRs [3].

CNVs are an important source of genetic variation in cattle, which is often associated with complex traits. The Axiom Bovine v3 array contains copy number markers for osteopetrosis and brachyspina. These can provide a useful resource for assessing the impact of CNVs on bovine health and production traits. Copy number analysis can be performed using Applied Biosystems™ Axiom™ Analysis Suite 4.0 software or higher.

The Axiom Bovine v3 array includes over 13,000 *Bos indicus* SNPs that have been shown to have a high minor allele frequency (MAF >0.35) across Nellore, India.

Dairy evaluations

The Axiom Bovine v3 array contains 44,887 markers that have been verified in collaboration with the CDCB, including the 1,000 CDCB core parentage markers. Using the array, dairy breeders can consolidate parentage verification and dairy evaluations in a single report. **The CDCB Export Tool** is a companion software application to Axiom Analysis Suite software that enables the export and direct upload of Axiom array genotype data to the CDCB database.

Automated data analysis and seamless integration with existing bioinformatics pipelines

The array leverages Axiom™ chemistry, and samples can be processed in three days from extracted DNA to data. The Axiom platform has been demonstrated to work on a variety of sample types including ear tissue, hair bulb, and dried blood spots. Axiom Analysis Suite software enables automated analysis and SNP classification of the data, as well as automated analysis of markers located on the Y chromosome. The software is integrated with algorithms for fixed-region copy number analysis and *de novo* copy number analysis.

The data obtained from this array can also be transformed into long format with the use of the **Applied Biosystems™ Axiom™ Long Format Export (AxLE) Tool**. This tool formats genotype data from Axiom arrays using the top (TOP) and bottom (BOT) designations based on the polymorphism itself or the contextual surrounding sequence. The tool also designates the A/B allele, enabling users to easily correlate genotype calls made at present to prior research.

Table 2 provides information on the bovine content included on the various bovine arrays from Thermo Fisher Scientific, including the Applied Biosystems™ Axiom™ Genome-Wide BOS 1 Bovine Array (Axiom BOS 1) and the Axiom™ Bovine Genotyping Arrays versions 1 through 3 (Axiom Bovine v1, Axion Bovine v2, and Axion Bovine v3). The coverage of markers per chromosome on the Axiom Bovine v3 array is shown in Figure 1.

Table 2. SNP overlap among several Axiom bovine genotyping arrays.

	Axiom BOS 1	Axiom Bovine v1	Axiom Bovine v2	Axiom Bovine v3
Axiom BOS 1	648,851	22,989	38,403	35,357
Axiom Bovine v1	22,989	51,988	49,877	48,878
Axiom Bovine v2	38,403	49,877	67,641	63,692
Axiom Bovine v3	35,357	48,878	63,692	63,988

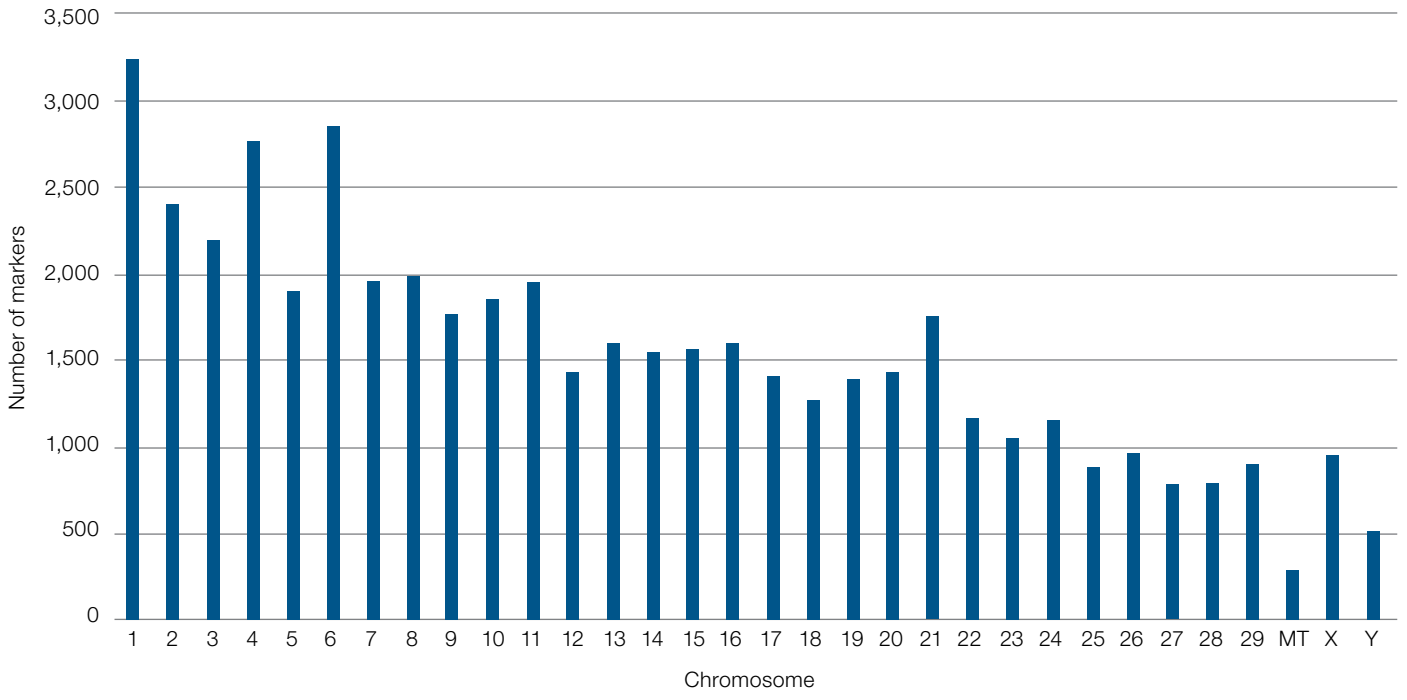

Figure 1. Axiom array marker coverage across the bovine genome.

Some of the traits and disease conditions available on the Axiom Bovine v3 array are listed in Table 3.

Table 3. Examples of traits and disease conditions available on the Axiom Bovine array.

Trait or disease condition	Trait type
Holstein dominant red coat	Color
MC1R_Ed	Color
PMEL17 64G_A	Color
Polled	Beneficial
Maple syrup urine disease (MSUD)	Lethal
Citrullinemia	Lethal
Deficiency of uridine monophosphate synthase (DUMPS)	Lethal-embryo
Calpain 1 CAPN1_530	Meat
Calpastain CAST_282	Meat
Calpastain CAST_2870	Meat
Calpastain CAST_2959	Meat
Acyl-CoA:diacylglycerol acyltransferase	Milk
Beta casein 118	Milk
Beta casein 151	Milk
Beta casein 154	Milk
Beta casein 322	Milk
Beta casein 363	Milk
Beta casein 411	Milk
Beta casein 500	Milk
Kappa casein 92	Milk
Kappa casein 342	Milk
Kappa casein 352	Milk
Kappa casein 373	Milk
Bovine leukocyte adhesion deficiency (BLAD)	Unwanted
Crooked tail syndrome CTA_AG	Unwanted
Crooked tail syndrome CTS_C>T	Unwanted
Chediak-Higashi syndrome	Unwanted
Spinal muscular atrophy	Unwanted

Ordering information

Product	Description	Cat. No.
Axiom Bovine Genotyping v3 Array (384HT format)	Contains one plate with 384 arrays; reagents and GeneTitan Multi-Channel consumables must be quoted separately	551089
Axiom 2.0 384HT Reagent Kit	Includes all reagents (except isopropanol) for processing 384 DNA samples	902245
Axiom 2.0 384HT GeneTitan Consumables Kit	Contains all GeneTitan Multi-Channel Instrument consumables required to process one array plate	902234
Axiom Bovine Genotyping v3 Array (mini-96 format)	Contains one plate with 96 arrays; reagents and GeneTitan Multi-Channel consumables must be quoted separately	551090
Axiom 2.0 Assay Mini 96 Reagent Kit	Includes all reagents (except isopropanol) for processing samples for two mini 96-array plates	903013
Axiom 2.0 Assay Mini 96 Manual Target Preparation Consumables Kit	Includes all consumables for processing samples for four mini 96-array plates	902986
Axiom 384HT High-Volume Consumables Kit	Contains all GeneTitan Multi-Channel Instrument consumables required to process samples for five 384-array plates or five mini 96-array plates	902629

References

1. International Society for Animal Genetics. https://www.isag.us/committees.asp?autotry=true&ULnotkn=true.
2. 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.
3. McClure MC, Sonstegard TS, Wiggans GR 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.

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