illumina®

BovineSNP50 v3 BeadChip

Featuring 53,218 evenly spaced and strategically placed SNPs across the bovine genome.

Highlights

- **Excellent call rates and accuracy**
  > 99% average call rates and 99.9% reproducibility
- **Comprehensive and uniform coverage**
  Evenly distributed polymorphic SNPs with a median spacing of 37.4 kb
- **Simple workflow**
  PCR- and ligation-free protocol
- **High-throughput format**
  Up to 24 samples can be interrogated in parallel

Introduction

In collaboration with the United States Department of Agriculture (USDA) Agricultural Research Service (ARS), the University of Missouri, and the University of Alberta, Illumina has developed the third generation of the BovineSNP50 v3 BeadChip (Figure 1). This high-density, genome-wide genotyping array (Table 1) features 53,218 informative single nucleotide polymorphism (SNP) probes that uniformly span the entire bovine genome to enable interrogation of genetic variation in cattle. The BeadChip empowers applications such as genome-wide–enabled selection, identification of quantitative trait loci, evaluation of genetic merit of individuals, and comparative genetic studies with imputation power in beef and dairy cattle.

Using the iScan™ System, integrated analysis software, and the Infinium high-throughput screening (HTS) assay, this 24-sample BeadChip provides exceptionally high call rates, allows for flexible content deployment, and enables the detection and measurement of copy number variation. In addition, PCR-free, single-tube sample preparation[3,4] significantly reduces labor and potential sample-handling errors.

BovineSNP50 v3 BeadChip content

High-value content was derived from publicly available sources such as the *Bos taurus* (bovine) reference genome,[1] and the Bovine Genome Consortia.[2] Illumina scientists and collaborators developed an informative and high-density SNP genotyping microarray that could be used to investigate genetic variation in any cattle breed. More than 12,000 probes were designed to target validated common SNPs (MAF ≥ 0.05) described by the Bovine HapMap Consortium. BeadChip developers also mined publicly available resources for common SNPs including Btau and whole-genome shotgun reads assembled by researchers at the Baylor College of Medicine[5]. Additional content sources include parentage markers identified by researchers from the US Meat Animal Research Center and Clay Center.[6] Content also includes SNPs identified by researchers at the USDA ARS through the comparison of Holstein bacterial artificial chromosome (BAC) sequence data[7] to the bovine genome assembly (Table 2).

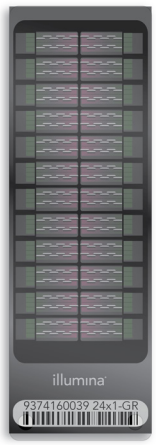

Figure 1: BovineSNP50 v3 BeadChip—The BovineSNP50 v3 BeadChip features 53,218 evenly spaced SNPs across the entire bovine genome.

Table 1: Product information[a]

Feature	Description
Species	*Bos taurus* (bovine)
Total number of markers	53,218
Capacity for custom bead types	600,000
Number of samples per BeadChip	24
DNA input requirement	200 ng genomic DNA
Assay chemistry	Infinium HTS
Instrument support	iScan System
Maximum iScan System sample throughput[a]	~5760 samples/week
Scan time per sample	30 minutes

a. Approximate values, scan times, and maximum throughput may vary depending on laboratory and system configurations.

More than 24,000 SNP probes featured on the BovineSNP50 BeadChip target novel SNP loci discovered by sequencing three pooled populations of economically important beef and dairy cattle. Illumina scientists and collaborators discovered more than 62,000 putative SNPs by deeply sequencing approximately 2% of the *Bos taurus taurus* genome[8]. More than 23,800 SNPs derived from this novel data source were chosen based on their spacing, expected minor allele frequency (MAF, and Infinium HTS Assay performance. In addition, BeadChip developers selected loci that target the largest unmapped contigs to ensure comprehensive coverage. All 53,218 SNP probes on the BovineSNP50 v3 BeadChip have been validated in 19 common beef and dairy breeds (Table 3).

The BeadChip targets evenly distributed SNPs that are polymorphic across the breeds tested, provides an average probe spacing of 50.6 kb and a median spacing of 37.4 kb, and presents an average MAF of 0.25 across all loci. Current research on linkage disequilibrium (LD) in

multiple breeds of cattle suggests haplotype blocks of approximately 70 kb on average.[9] This indicates that the resolution offered by the BovineSNP50 v3 BeadChip is well within that of LD in cattle. More than 53,000 SNP probes on the BovineSNP50 v3 BeadChip map to UMD 3.0, the most current bovine reference genome assembly.

Table 2: BovineSNP50 BeadChip content sources

Source	BovineSNP50 v1 Probes	BovineSNP50 v2 Probes	BovineSNP50 v3 Probes
Novel SNPs derived from sequencing common cattle breeds using the Illumina GenomeAnalyzer	23,840	24,181	22,299
Bovine HapMap Data Set	12,298	12,342	11,607
Btau Assembly SNPs	9361	9404	9086
Whole-Genome Shotgun Reads[a]	5808	6038	5485
Holstein BAC Sequence Data	1409	1411	1238
Parentage[b]	116	120	200
Other[c]	1169	1113	3384
Total	54,001	54,609	53,218

a. Reads derived from 6 cows/breeds (Norwegian Red, Holstein, Brahman, Angus, Jersey, and Limousin) compared against Btau2.0.
b. Highly curated parentage markers that include 30 duplicates.
c. Includes common SNPs validated by the Institute for Food and Agricultural Sciences Alberta and INRA and the French International Institute of Agriculture.

High-quality data

The 53,218 SNP probes on the BovineSNP50 v3 BeadChip were subjected to rigorous functional testing to ensure strong performance using the Infinium HTS assay. Whole-genome association studies are successful, in part, due to high call rates and accurately called genotypes. Because complex traits often have relatively small gene effects, potential associations may be missed if the assayed SNP in linkage disequilibrium (LD) with the SNP of interest has a low call rate or incorrect genotype call. Illumina ensures that every BovineSNP50 v3 BeadChip offers > 99% call rate (Table 3).

Internal validation of content on the BovineSNP50 v3 BeadChip using samples provided by the Bovine HapMap Consortium showed outstanding results (Table 4). Product developers identified and retained 4290 loci that appear to have an adjacent or underlying deletion among the breeds sampled. These loci yield lower call rates when compared to the other loci on the panel. However, they were retained because they may provide biologically relevant information for traits of interest and for future improvements in the genome build.

The performance and content validation results clearly show the reliability and outstanding data quality the BovineSNP50 v3 BeadChip delivers. With this BeadChip, researchers can predict the genetic merit for phenotypes of interest and investigate the genetic basis of variation among a multitude of cattle breed types. The BovineSNP50 v3 BeadChip demonstrates the commitment by Illumina to provide innovative tools for research and production needs to support the livestock industry.

Table 3: BovineSNP50 v3 BeadChip performance

Parameter	Results	Product specification
Average Call Rate[a]	99.7%	> 99%
Reproducibility[b]	100%	> 99.9%
Mendelian inconsistencies	0.06%	< 0.1%

a. Based on 274 individuals from 17 major cattle breeds, 31 trios, and 1 replicate.
b. Based on 4 Holstein trios.

Table 4: BovineSNP50 v3 BeadChip content validation

Breed	Samples	Polymorphic loci[a]	Mean MAF	Median MAF[b]
Angus	22	41,209	0.22	0.23
Beefmaster	24	43,741	0.23	0.23
Bos indicus Gir	17	25,320	0.11	0.03
Bos indicus Nelore	5	22,422	0.1	0
Brahman	20	33,038	0.13	0.08
Charolais	14	42,685	0.23	0.21
Guernsey	21	37,313	0.2	0.19
Hereford	20	43,902	0.23	0.23
Holstein	22	41,913	0.23	0.23
Jersey	9	36,683	0.18	0.17
Limousin	10	42,732	0.21	0.2
N'Dama	6	29,032	0.14	0.08
Piedmontese	21	42,652	0.23	0.24
Red Angus	10	43,028	0.22	0.2
Romagnola	6	38,521	0.2	0.17
Santa Gertrudis	7	42,675	0.21	0.21
Sheko	9	36,582	0.17	0.17
Overall	274	47,919	0.26	0.26

a. Minor allele frequency (MAF) > 0.05
b. Across all 53,218 loci

Solutions for genotyping

An optional Laboratory Information Management System (LIMS) and robotic automation are available to track samples accurately and efficiently throughout the workflow. Researchers can genotype their own samples using the Illumina GenomeStudio DNA analysis software or FastTrack Genotyping Service.

Summary

Developed in collaboration with leading bovine researchers, the BovineSNP50 v3 BeadChip features more than 53,000 evenly spaced SNP probes that span the bovine genome. This 24-sample BeadChip presents a high-throughput, cost-effective solution for whole-genome studies in beef and dairy cattle.

Ordering information

BovineSNP50-24 v3 BeadChip Kit[a]	Description	Catalog no.
48 samples	Each package contains 2 BeadChips, along with reagents for amplifying, fragmenting, hybridizing, labeling, and detecting 48 DNA samples	20000766
288 samples	Each package contains 12 BeadChips, along with reagents for amplifying, fragmenting, hybridizing, labeling, and detecting 288 DNA samples	20000767
1152 samples	Each package contains 48 BeadChips, along with reagents for amplifying, fragmenting, hybridizing, labeling, and detecting 1152 DNA samples	20000768
BovineSNP50-24+ v3 BeadChip Kit[b]	**Description**	**Catalog no.**
48 samples	Each package contains 2 BeadChips, and allows researchers to include up to 600,000 additional custom probes per sample for targeted studies	20000769
288 samples	Each package contains 12 BeadChips, and allows researchers to include up to 600,000 additional custom probes per sample for targeted studies	20000830
1152 samples	Each package contains 48 BeadChips, and allows researchers to include up to 600,000 additional custom probes per sample for targeted studies	20000831

a. Each BeadChip can process 24 samples and analyze ~53,000 loci.
b. Enabled for custom content.

Learn more

Learn more about the BovineSNP50-24 v3.0 BeadChip at www.illumina.com/products/by-type/microarray-kits/bovine-snp50.html

References

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