illumina®

BovineSNP50 Genotyping BeadChip

Featuring 53,714 evenly spaced and strategically placed SNP probes that span 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

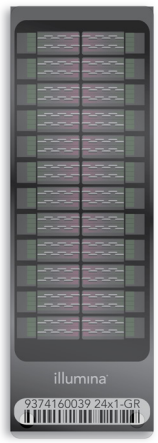

Figure 1: BovineSNP50 BeadChip—The BovineSNP50 BeadChip features 53,714 evenly spaced SNPs across the entire bovine genome.

Introduction

Illumina, in collaboration with the USDA ARS, the University of Missouri, and the University of Alberta, has developed the third generation of a proven high-density, genome-wide genotyping array for the interrogation of genetic variation in cattle. The BovineSNP50 v3 BeadChip (Figure 1) features 53,714 informative SNP probes that uniformly span the entire bovine genome. BovineSNP50 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.

More than half of the probes on this BeadChip are designed to target SNPs discovered using proven Illumina sequencing by synthesis (SBS) chemistry. Additional high-value content is derived from publicly available sources such as the bovine reference genome, Btau[1], and the Bovine HapMap Consortium data set[2]. The BovineSNP50 BeadChip covers common SNPs validated in economically important beef and dairy cattle breed types and presents an average minor allele frequency (MAF) of 0.25 across all loci. Importantly, this BeadChip offers uniform coverage with an average probe spacing of 50.6 kb to provide more than sufficient SNP density for robust genome-association studies in cattle.

The BovineSNP50 BeadChip is a multisample genotyping panel powered by the Illumina Infinium® HTS Assay. This revolutionary assay 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. Illumina offers multisample format that further reduces experimental variability and overall project cost by allowing researchers to interrogate up to 24 samples in parallel.

The BovineSNP50 BeadChip features a combination of proprietary assay technologies, unconstrained locus selection, and high-throughput format that offers a comprehensive solution for whole-genome studies in beef and dairy cattle.

BovineSNP50 v3 BeadChip Content

Illumina scientists and collaborators set out to develop 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 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 chromosomes (BAC) sequence data[7] to the bovine genome assembly (Table 1).

More than 24,000 SNP probes featured on the BovineSNP50 BeadChip target novel SNP loci discovered by sequencing 3 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 MAF, and Infinium HTS Assay performance. In addition, BeadChip developers selected loci that target the largest unmapped contigs to ensure comprehensive coverage. All 53,714 SNP probes on the BovineSNP50 BeadChip have been validated in 19 common beef and dairy breeds (Table 3). This product targets evenly distributed SNPs that are polymorphic across the breeds tested and provides an average probe spacing of 50.6 kb and a median spacing of 37.4 kb. Current research on linkage disequilibrium (LD) in multiple breeds of

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

Table 1: 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 53,714 SNP assays 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 with the SNP of interest has a low call rate or incorrect genotype call. Illumina ensures that every BovineSNP50 BeadChip offers > 99% call rate.

Table 2 shows the outstanding results from internal validation of the BovineSNP50 v3 BeadChip content using samples provided by the Bovine HapMap Consortium. Product developers identified and retained 4,290 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 BeadChip demonstrates the commitment by Illumina to provide the most innovative tools for research and production needs to support the livestock industry.

Solutions for Genotyping

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

Table 2: BovineSNP50 v3 Performance and Specifications

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 3: BovineSNP50 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. MAF > 0.05
b. Across all 53,714 loci

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Ordering Information

Catalog No.	Product	Description
20000766	INFBovineSNP50-24 v3 BeadChip Kit (for 48 samples)	Each package contains 2 BeadChips, along with reagents for amplifying, fragmenting, hybridizing, labeling, and detecting 48 DNA samples
20000767	INFBovineSNP50-24 v3 BeadChip Kit (for 288 samples)	Each package contains 12 BeadChips, along with reagents for amplifying, fragmenting, hybridizing, labeling, and detecting 288 DNA samples
20000768	INFBovineSNP50-24 v3 BeadChip Kit (for 1152 samples)	Each package contains 48 BeadChips, along with reagents for amplifying, fragmenting, hybridizing, labeling, and detecting 1152 DNA samples
20000769	INFBovineSNP50-24+ v3 BeadChip Kit (for 48 samples)	Each package contains 2 BeadChips, and allows researchers to include up to 600,000 additional custom probes per sample for targeted studies
20000830	INFBovineSNP50-24+ v3 BeadChip Kit (for 288 samples)	Each package contains 12 BeadChips, and allows researchers to include up to 600,000 additional custom probes per sample for targeted studies
20000831	INFBovineSNP50-24+ v3 BeadChip Kit (for 1152 samples)	Each package contains 48 BeadChips, and allows researchers to include up to 600,000 additional custom probes per sample for targeted studies
Each BovineSNP50 v3 DNA Analysis BeadChip can process 24 samples and analyze approximately 53,000 loci.		

Product Summary

Developed in collaboration with leading bovine researchers, the BovineSNP50 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.

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