

EquineSNP50 Genotyping BeadChip

The EquineSNP50 Genotyping BeadChip features more than 54,000 evenly spaced and validated SNPs derived from the EquCab2 assembly. This 12-sample Infinium® BeadChip presents a cost-effective and high-quality genotyping solution for equine research.

INTRODUCTION

The EquineSNP50 Genotyping BeadChip was developed in collaboration with the International Equine Genome Mapping Workshop¹ and the Morris Animal Foundation's Equine Genome Consortium². This BeadChip features 54,602 evenly distributed SNPs derived from the EquCab2.0 SNP collection compiled by the Broad Institute. As the first genome-wide genotyping array for *Equus caballus*, the EquineSNP50 BeadChip supports a broad range of whole-genome genotyping applications for equine research. Designed to enable identification of genes and mutations that contribute to traits of interest in all major horse breeds, this BeadChip offers a powerful platform for

EQUINESNP50 BEADCHIP HIGHLIGHTS

- **Uniform Coverage:** High-density, evenly distributed coverage across the entire genome
- **High Throughput:** 12 samples per BeadChip, 54,602 SNP assays per sample
- **Streamlined Workflow:** PCR-free protocol
- **Low Sample Input:** 200 ng per sample
- **Integrated Data Analysis:** BeadStudio Software

improving horse breeding programs. More, it enables the development of new diagnostic and therapeutic approaches to promote equine health and welfare.

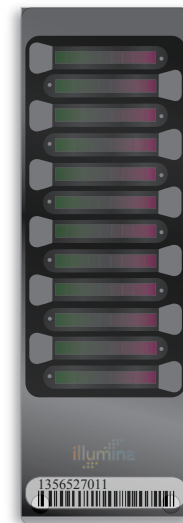
POWERFUL ASSAY TECHNOLOGY

The multi-sample Equine SNP Genotyping BeadChip is powered by Illumina's Infinium® II Assay. This revolutionary assay delivers the industry's highest call rates, allows for flexible content deployment, and enables detection and measurement of copy number variation. The assay's PCR-free single-tube sample preparation^{3,4} significantly reduces labor and potential sample handling errors. Illumina's multi-sample format further reduces experimental variability and overall project cost by allowing researchers to interrogate up to 12 samples in parallel.

CONTENT DESIGN

Equine BeadChip content is derived from data generated by the Broad Institute's Equine Genome Sequencing Project⁵. Researchers at the Broad Institute sequenced seven equine breeds that represent modern and ancestral horses. They discovered more than 940,000 SNPs across Arabian, Andalusian, Akhal-teke, Icelandic, Standardbred, Thoroughbred, and Quarter horse breeds. From this data set, the EquineSNP50

FIGURE 1: EQUINESNP50 BEADCHIP



The EquineSNP50 BeadChip contains 54,602 SNP assays per sample and allows for the interrogation of 12 samples in parallel.

BeadChip developers designed probes that target a subset of uniformly distributed SNPs spanning the entire horse genome. Using this strategy, Illumina and collaborators have developed the industry's most powerful tool for discovering the genetic basis of phenotypic variation in the modern horse.

COMPLETE GENOMIC COVERAGE

The 54,602 evenly spaced SNPs offer comprehensive coverage, enabling confident identification and map-

ping of quantitative trait loci. With an average probe spacing of 43.2kb across all 31 autosomes (Figure 2), the EquineSNP50 BeadChip provides more than sufficient marker information to support whole-genome-enabled selection.

BeadChip markers were validated in 15 modern horse breeds and shown to have an average minor allele frequency of 0.21 across all of the breeds tested. Over 53,500 probes target polymorphic loci, including 2,254 loci that appear to have an adjacent or third polymorphism such as a deletion or third allele.

ANALYSIS SIMPLIFIED

Illumina's BeadStudio analysis

software is a powerful and user-friendly tool for analyzing data generated by the EquineSNP50 Genotyping BeadChip. The Genotyping Module provides automated genotype calling and sample clustering. Powerful visualization tools and quality-control features enable graphical output of data and reports on reproducibility and Mendelian consistency.

As with all of Illumina's standard products, an optional Laboratory Information Management System (LIMS) and robotic automation are available to accurately and efficiently track samples throughout analysis.

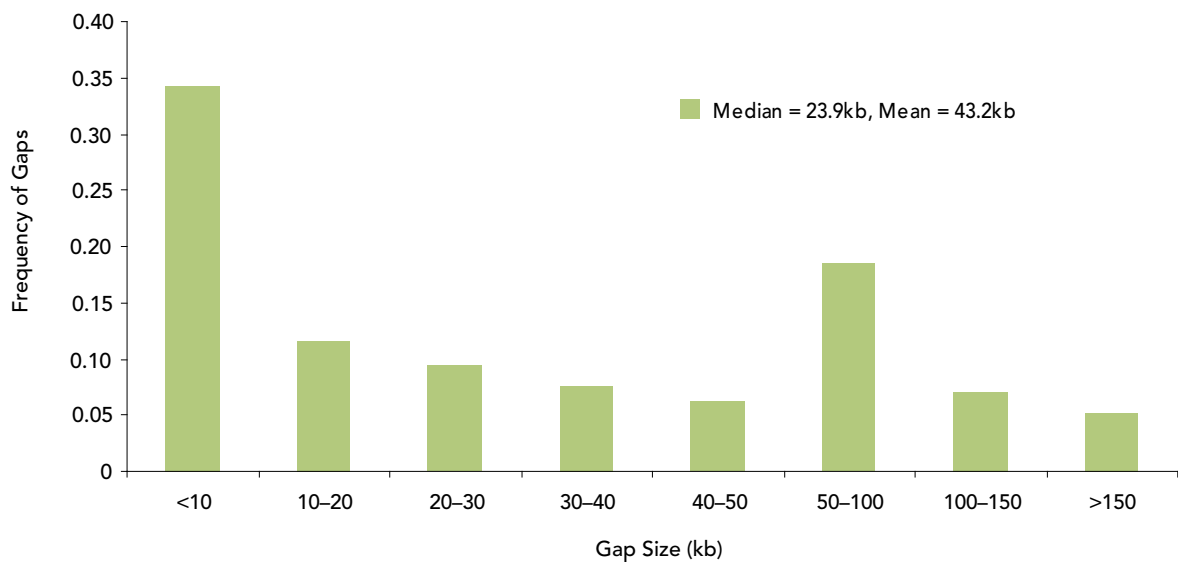
Researchers can genotype their samples using the illumina high-

throughput iScan™ System, submit samples to Illumina's FastTrack Genotyping Service, or assess Illumina's genotyping products through the Illumina Customer Sample Evaluation (CSE) program.

INFINIUM GENOTYPING SOLUTIONS

The combination of Illumina proprietary assay technologies and flexible content deployment provides the most comprehensive solution available for genome-wide genotyping. Like all Infinium products, the EquineSNP50 BeadChip delivers the industry's highest data quality, allowing researchers to confidently pursue the fastest path to discovery.

FIGURE 2: EQUINESNP50 BEADCHIP MARKER SPACING



The EquineSNP50 BeadChip offers more than sufficient marker density for confident identification and mapping of quantitative trait loci in modern horse breeds.

TABLE 1: EQUINESNP50 BEADCHIP PERFORMANCE DATA AND SPECIFICATIONS

PARAMETER	RESULTS*	PRODUCT SPECIFICATION
Average Call Rate†	99.54%	> 99%
Reproducibility	100%	> 99%
Mendelian Inconsistencies	0.01%	< 0.1%

*Results based on 372 DNA samples including 8 replicates and 15 trios from fifteen breeds. See Table 2 for the list of breeds tested.

†Excludes Hokkaido samples. Average call rate including Hokkaido samples is 99.41%.

TABLE 2: BREEDS USED TO EVALUATE EQUINESNP50 BEADCHIP

BREED	# OF SAMPLES	CALL RATE (%)	POLYMORPHIC LOCI (MAF > 0.05)	AVERAGE MAF
Andalusian	20	99.50	47,549	0.20
Arabian	24	99.50	47,176	0.21
Belgian	24	99.51	44,303	0.18
Franches Montagnes	22	99.54	47,144	0.20
French Trotter	18	99.56	46,961	0.21
Hanoverians	20	99.55	48,362	0.23
Icelandic	18	99.51	43,032	0.18
Mongolian	21	99.06	49,729	0.20
Norwegian Fjord	22	99.46	43,227	0.18
Quarter Horse	48	99.58	52,052	0.23
Saddlebred	23	99.52	47,503	0.21
Standardbred	20	99.55	45,715	0.20
Swiss Warmblood	19	99.57	49,349	0.23
Thoroughbred	44	99.59	47,648	0.22
Hokkaido	13	95.85	40,127	0.16
Total	356	99.54*/99.41	53,524†	0.23

*Call rate excludes Hokkaido breed.

† Includes 2,254 loci that appear to have an adjacent or third polymorphism (e.g., deletion or third allele).

ORDERING INFORMATION

CATALOG NO.	PRODUCT	DESCRIPTION
WG-402-1001	EquineSNP50 Genotyping Kit	Each package contains four BeadChips and reagents for processing 48 samples.
WG-402-1002	EquineSNP50 Genotyping Kit	Each package contains 24 BeadChips and reagents for processing 288 samples.
WG-402-1003	EquineSNP50 Genotyping Kit	Each package contains 96 BeadChips and reagents for processing 1152 samples.
FT-20-116	EquineSNP50 FastTrack Service Project	Illumina's FastTrack Services requires a minimum of 94 samples and 1.5 µg of DNA for each sample.

The EquineSNP50 Genotyping BeadChip can process 12 samples in parallel and genotype > 54,000 loci per sample.

ADDITIONAL INFORMATION

To learn more about Illumina's solutions for agricultural research, please visit www.illumina.com/agriculture or contact us at the address below.

REFERENCES

- (1) <http://www.uky.edu/Ag/Horsemap/>
- (2) www.morrisanimalfoundation.org/equine
- (3) Gunderson KL, Steemers FJ, Lee G, Mendoza LG, Chee MS (2005) A genome-wide scalable SNP genotyping assay using microarray technology. *Nat Genet* 37(5): 549-554.
- (4) Steemers FJ, Weihua Chang W, Lee G, Barker DL, Shen R, et al. (2006) Whole-genome genotyping with the single-base extension assay. *Nat Methods* 3(1): 31-33.
- (5) <http://www.broad.mit.edu/mammals/horse/>

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