

# Infinium™ Canine HTS Genotyping BeadChip

More than 170,000 evenly spaced and validated SNPs derived from the CanFam4.0 assembly.

- Achieve exceptional data quality with > 99% average call rates and 99.99% reproducibility
- Access a simplified, PCR-free protocol powered by the Infinium HTS assay
- Interrogate up to 24 samples in parallel in a single assay with a high-throughput workflow



## Introduction

Dogs are arguably man's greatest invention,<sup>1</sup> originally domesticated from grey wolves as long as 100,000 years ago. Selective breeding over the last few centuries has led to an incredible amount of biological diversity among modern domestic dog breeds, as they display a wide range of physical traits and behaviors.<sup>2</sup> However, breeding methods and recent bottlenecks experienced in the domestic dog have unintentionally resulted in a high prevalence of specific diseases, including cancers, heart disease, epilepsy, and hip dysplasia.<sup>2</sup> Since humans and canines share a number of common diseases, the domestic dog has been an increasingly important model for understanding the genetic component of human disease.<sup>3</sup> Recent discoveries have also shown that selective breeding has led to large haplotype blocks being carried into the canine genomes. These large genomic regions are ideal for canine genome-wide association studies (GWAS), where far fewer markers are required to identify genetic disease associations compared human studies. The unique breeding history of the domestic dog offers the research community a powerful opportunity for unraveling the genetic basis of disease, behavioral and morphological traits, and mammalian variation.

In 2005, researchers at the Broad Institute completed the first assembly of the canine genome, CanFam2.0. The Canine Genome Sequencing Project produced a high-quality draft sequence of a female boxer named Tasha. The assembly of Tasha's genome was used to compare sequence data of other key breeds.<sup>2</sup> From these efforts over 2.5 million high quality single nucleotide polymorphisms (SNPs) have been identified. Subsequently, the Dog Genome Sequencing Consortium used next-generation sequencing (NGS) technologies to update the canine genome assembly from CanFam2.0 to CanFam4.0.<sup>4</sup>

The Infinium Canine HTS Genotyping BeadChip contains more than 170,000 markers placed on the CanFam4.0 reference sequence. Illumina developed the Infinium Canine HTS Genotyping BeadChip based on legacy content on the CanineHD Genotyping BeadChip, which was launched in collaboration with the LUPA Consortium and other partners, such as the Broad Institute. Featuring highly polymorphic SNP content and providing uniform genomic coverage, the Infinium Canine HTS BeadChip enables the interrogation of genetic variation in any domestic dog breed. Importantly, this BeadChip presents an average of greater than 70 markers per megabase

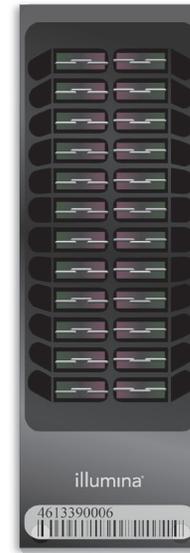


Figure 1: The Infinium Canine HTS Genotyping BeadChip—The BeadChip features more than 170,000 evenly spaced SNPs across the entire dog genome.

(Mb), providing ample SNP density for robust within-breed association and copy number variation (CNV) studies. In addition, across-breed haplotypes in dogs range between 10–100kb, suggesting that genetic risk factors may be shared across breeds. The dense marker set increases the ability to detect across-breed trait and risk factor signals. The combination of the Illumina Infinium assay, unconstrained locus selection, and a high-throughput format presents a comprehensive solution for whole-genome studies in the domestic dog.

## SNP selection strategy

The SNP content featured on the Infinium Canine HTS BeadChip features proven content from the CanineHD Beadchip, including the Dog Genome Project 2.5 million SNP set and 1696 SNPs identified using a hybridization-based targeted resequencing method of SNP discovery within gaps in the SNP map. SNPs were selected from a diverse set of breeds and picked to represent as many different breeds as possible, while maintaining coverage of ~70 SNPs per Mb. In addition, the Infinium Canine HTS BeadChip includes more than 5000 newly discovered SNPs associated with canine parentage, trait, diseases and more.<sup>5-7</sup> This content selection strategy empowers confident disease mapping in all dog breeds, as can be seen by the successful validation in 26 diverse breeds

(data not shown). On top of the validation performed on the CanineHD BeadChip base, the Infinium Canine HTS BeadChip shows an average call rate of 99.80% across breeds calculated from testing 288 canine samples across regions, including the Americas, Europe, and Asia (Table 1).

## High-quality data

The Infinium Canine HTS BeadChip is powered by the proven Infinium HD assay, providing high call rates, flexible content, and precise CNV detection. The SNPs on the Infinium Canine HTS BeadChip were subjected to rigorous functional testing across dog breeds to ensure strong performance using the Infinium HTS assay. GWAS are successful, in part, due to high call rates and accurately called genotypes. Since complex disease traits often have relatively small gene effects, potential associations may be missed if the assayed SNP in linkage disequilibrium with the disease SNP has a low call rate or incorrect genotype call. Illumina ensures that every Infinium Canine HTS BeadChip offers > 99% call rate on samples meeting the recommended input requirements. This includes samples that were collected from buccal swabs.

Internal validation of the Infinium Canine HTS BeadChip as performed by Illumina during its development (Table 1). Loci were identified and retained that appear to have an adjacent or third polymorphism (eg, deletion or third allele) among the breeds sampled. Although these loci yield lower call rates when compared to the other loci on the panel, they were retained because they may provide biologically relevant information for traits of interest and for future improvements in the genome build.

Table 1: Performance data and specifications

Parameter	Percent	Product specification
Average call rate	99.80%	> 99%
Reproducibility	> 99.9%	> 99.9%
Mendelian inconsistencies	0.002%	< 0.01%

Results based on 288 DNA samples.

## Illumina solutions for genotyping

The high-quality data and low per-sample cost of the Infinium Canine HTS BeadChip are part of the powerful Illumina Whole-Genome Genotyping Solution. In addition, optional automation and the Laboratory Information Management System (LIMS) lower costs by eliminating errors associated with manual processing. Illumina genotyping products can be accessed via Illumina Lab Services or one's own iScan™ System. Illumina solutions provide industry-leading levels of accuracy, flexibility, and affordability.

## Learn more

[Illumina genotyping solutions](#)

## Ordering information

Product	Description	Catalog no.
Infinium Canine HTS Kit (48 samples)	Each package contains two BeadChips and reagents for processing 48 samples.	20103711
Infinium Canine HTS Kit (288 samples)	Each package contains 12 BeadChips and reagents for processing 288 samples.	20103712
Infinium Canine HTS Kit (1152 samples)	Each package contains 48 BeadChips and reagents for processing 1152 samples.	20103713
Canine HTS FastTrack Service Project	The Illumina FastTrack Service requires a minimum of 94 samples and 1.5 µg of DNA for each sample.	FT-310-1236

Each Infinium Canine HTS Genotyping BeadChip can process 24 samples in parallel and assay 172,035 markers per sample.

## References

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1.800.809.4566 toll-free (US) | +1.858.202.4566 tel  
techsupport@illumina.com | www.illumina.com

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