DATA SHEET

Infinium™ CytoSNP-850K v1.3 BeadChip

Comprehensive coverage of cytogenomic-relevant genes for constitutional and cancer research applications

- Designed based on input from the international cytogenomics community and peer-reviewed literature
- Includes ~ 850K SNPs with 15× redundancy and enriched coverage for 3262 dosage-sensitive genes
- Offers high signal-to-noise ratios using long 50-mer SNP probes for high target specificity
- Delivers reproducible data with a broad range of sample types, including FFPE samples

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Introduction

Structural and numerical genetic variations are known to influence the presentation of a phenotype. Accurate profiling of chromosomal aberrations, such as duplications, deletions, unbalanced rearrangements, and copy-neutral absence of heterozygosity (AOH) events, is crucial for studies associated with cancer and genetic disease. The Infinium CytoSNP-850K v1.3 BeadChip (Figure 1) uses proven Infinium assay chemistry to offer the sensitivity and broad coverage needed to understand the impact of these genetic variants.

The Infinium CytoSNP-850K v1.3 BeadChip includes input on cytogenetic-relevant genes from the international community for constitutional and cancer research applications. The gene list contains updated content from the International Collaboration for Clinical Genomics (ICCG)[1] and the Cancer Cytogenomics Microarray Consortium (CCMC),[2] providing a comprehensive view of cytogenomic activity.

The Infinium CytoSNP-850K v1.3 BeadChip provides robust performance across a broad range of sample types, including formalin-fixed, paraffin-embedded (FFPE) tissue samples. The processed arrays can be scanned using the iScan™ System, HiScan™ System, or NextSeq™ 550 System with high reproducibility (Table 1). For exceptional data analysis, CytoSNP- 850K BeadChips are compatible with the BlueFuse™ Multi Software.

Inclusive coverage

The Infinium CytoSNP-850K v1.3 BeadChip contains ~ 850K empirically selected single nucleotide polymorphisms (SNPs) spanning across the genome. This high density of SNPs enables high-resolution analysis for discovery of meaningful chromosomal aberrations (Table 2). Intelligent design, based on the latest updates from peer-reviewed literature, provides enriched coverage for 3262 genes of known relevance in both constitutional and cancer research applications (Table 3).

For greater sensitivity, the Infinium CytoSNP-850K v1.3 BeadChip takes advantage of proven Infinium assay technology. The use of 50-mer SNP probes promotes high specificity to the target sequence. This facilitates enhanced identification of low-level mosaics[3] and accurate breakpoint estimation for both copy number variations (CNVs) and AOH. High 15× bead redundancy increases the signal-to-noise ratio (SNR), facilitating identification of CNV and AOH calls with high confidence. Accurate CNV calls can be made with as few as 10 consecutive probes, demonstrating the high performance level of the Infinium assay.

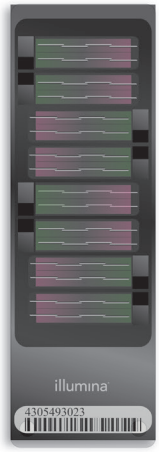

Figure 1: Infinium CytoSNP-850K v1.3 BeadChip—The 8-sample Infinium CytoSNP-850K v1.3 BeadChip provides cytogenomic-relevant genes for constitutional and cancer studies with content from ICCG and CCMC.

Powerful analysis software

BlueFuse Multi Software uses optimized, validated algorithms to detect copy number change and AOH automatically, enabling rapid, accurate molecular cytogenetics analysis. The software offers rich genomic annotation, a centralized database of historical case findings, and comprehensive reporting of identified variants to facilitate data interpretation. Developed in coordination with the clinical genomics community, the intuitive BlueFuse Multi Software interface provides an integrated framework for analyzing data for molecular cytogenetic applications.

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Table 1: Infinium CytoSNP-850K BeadChip product information

Feature	Description		
Species	Human		
No. of samples per BeadChip	8		
DNA input requirement	200 ng		
Assay chemistry	Infinium HD Super		
SNP replicates	15×		
No. of SNPs to call CNV	10		
Instrument support	**iScan System**	**NextSeq 550 System**	
Total no. of markers	848,511	848,511	
Sample throughput per week	960	128	
Scan time per sample	5 min	5 min	
Data performance	**iScan System**	**NextSeq 550 System**	**Product specification**
Call rate	99.87%	99.91%	> 98%
Reproducibility	99.99%	99.99%	>99%
Log R deviation	0.1005	0.11	< 0.20

Table 2: Infinium CytoSNP-850K v1.3 BeadChip coverage

Probe spacing	Average distance
Targeted region probe spacing	~ 1 kb
Targeted region resolution	~ 10 kb
Genomic backbone probe spacing	~ 5 kb
Overall probe spacing	~ 1.8 kb
Overall effective resolution	~ 18 kb

Table 3: Infinium CytoSNP-850K v1.3 BeadChip marker information[a]

Marker categories[b]	iScan System
Total no. of markers	848,511
RefSeq genes	451,304
RefSeq +/- 10 kb	522,349
ADME genes	14,147
ADME +/- 10 kb	17,419
COSMIC genes	419,775
HLA markers	5126
HLA genes	294
GO genes	139,256
Exonic regions	70,851
Promoter regions	24,891
X chromosome markers	29,837
Y chormosome markers	1229
PAR/homologous markers	984

a. Values are obtained from the Assay manifest file. Variations are due to different manifests/product files required to process the BeadChip for each instrument.
b. Compared against the human genome issue hg-19 reference genome.
Abbreviations: ADME, adsorption, distribution, metabolism, excretion; COSMIC, catalog of somatic mutations in cancer; MHC, major histocompatibility complex; HLA, human leukocyte antigen; PAR, pseudoautosomal region

Learn more

Infinium CytoSNP-850K v1.3 BeadChip, illumina.com/techniques/popular-applications/cytogenomics.html

BlueFuse Multi Software, illumina.com/clinical/clinical_informatics/bluefuse.html

Ordering information

Product	Catalog no.
Infinium CytoSNP-850K v1.3 BeadChip Kit (8 samples)	20025643
Infinium CytoSNP-850K v1.3 BeadChip Kit (16 samples)	20025644
Infinium CytoSNP-850K v1.3 BeadChip Kit (48 samples)	20025645
Infinium CytoSNP-850K v1.3 BeadChip Kit (96 samples)	20025646

References

1. Clinical Genome Resource. www.clinicalgenome.org. Published July 2018. Accessed December 12, 2022.
2. Cancer Genomics Consortium. www.cancergenomics.org. Published July 2018. Accessed December 12, 2022.
3. Conlin LK, Thiel BD, Bonnemann CG, et al. Mechanisms of mosaicism, chimerism and uniparental disomy identified by single nucleotide polymorphism array analysis. *Hum Mol Genet*. 2010;19(7):1263–1275. doi: 10.1093/hmg/ddq003.

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M-GL-01507 v1.0