DATA SHEET

Infinium™ CytoSNP-850K v1.4 BeadChip

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

- Incorporates 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 50mer 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 v1.4 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 v1.4 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 v1.4 BeadChip provides robust performance across a broad range of sample types, including formalin-fixed, paraffin-embedded (FFPE) tissue samples. Processed arrays can be scanned with high reproducibility using an iScan™ System or NextSeq™ 550 System (Table 1). For exceptional data analysis, CytoSNP-850K BeadChips are compatible with BlueFuse™ Multi Software.

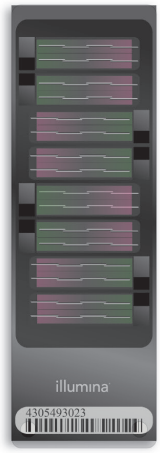

Figure 1: Infinium CytoSNP-850K v1.4 BeadChip—The 8-sample Infinium CytoSNP-850K v1.4 BeadChip provides cytogenomic-relevant genes for constitutional and cancer studies with content from ICCG and CCMC.

Inclusive coverage

The Infinium CytoSNP-850K v1.4 BeadChip contains ~850K empirically selected single nucleotide polymorphisms (SNPs) spanning the genome (Table 2 and Table 3). This high density of SNPs enables high-resolution analysis for discovery of meaningful chromosomal aberrations. 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.

For greater sensitivity, the Infinium CytoSNP-850K v1.4 BeadChip takes advantage of proven Infinium assay technology. The use of 50mer 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, 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.

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.

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	848,902	848,902	
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	99.89%	99.90%	> 98%
Reproducibility	99.99%	99.99%	> 99%
Log R deviation	0.0929	0.1035	< 0.20

Table 2: Infinium CytoSNP-850K v1.4 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 v1.4 BeadChip marker information[a]

Marker categories[b]	No. of markers (iScan System)
Total no. of markers	848,902
RefSeq genes	467,422
RefSeq +/- 10 kb	541,515
ADME genes	15,153
ADME +/- 10 kb	18,590
COSMIC genes	418,131
HLA markers	5145
HLA genes	276
GO genes	137,873
Exonic regions	68,801
Promoter regions	26,814
X chromosome markers	29,894
Y chormosome markers	1197
PAR/homologous markers	728

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 hg19 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

Cytogenomics

Infinium CytoSNP-850K v1.4 BeadChip

BlueFuse Multi Software

Ordering information

Product	Catalog no.
Infinium CytoSNP-850K v1.4 BeadChip Kit (8 samples)	20103480
Infinium CytoSNP-850K v1.4 BeadChip Kit (16 samples)	20103481
Infinium CytoSNP-850K v1.4 BeadChip Kit (48 samples)	20103482
Infinium CytoSNP-850K v1.4 BeadChip Kit (96 samples)	20103483

References

1. Clinical Genome Resource. Welcome to ClinGen. www.clinicalgenome.org. Updated November 21, 2023. Accessed November 29, 2023.
2. Cancer Genomics Consortium. Home - Cancer Genomics Consortium. www.cancergenomics.org. Accessed November 29, 2023.
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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