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Infinium™ CytoSNP-850K v1.2 BeadChip

The Infinium CytoSNP-850K v1.2 BeadChip provides comprehensive coverage of cytogenomic-relevant genes for constitutional and cancer applications.

Highlights

- **Current content for constitutional and cancer studies**
  Intelligent design based on input from the international cytogenomics community and peer-reviewed literature
- **Dynamic performance and broad coverage**
  Nearly 850,000 SNPs with 15× redundancy and enriched coverage for 3262 dosage-sensitive genes
- **High analytical sensitivity for low-level mosaics**
  Long 50-mer SNP probes offer high signal-to-noise ratios for high target specificity
- **Reproducible data with challenging samples**
  Broad compatibility with a range of sample types, including formalin-fixed, paraffin-embedded (FFPE) tissues

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 v1.2 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 v1.2 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 v1.2 BeadChip provides robust performance across a broad range of sample types, including 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.

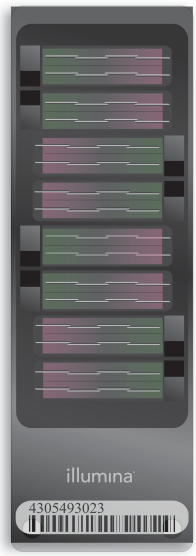

Figure 1: The Infinium CytoSNP-850K v1.2 BeadChip—The 8-sample Infinium CytoSNP-850K v1.2 BeadChip provides cytogenomic-relevant genes for consitutional and cancer studies with content from ICCG and CCMC.

Table 1: Infinium CytoSNP-850K v1.2 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/HiScan System**	**NextSeq 550 System**	
Total no. of markers[a]	846,819	844,855	
Sample throughput per week	960[b]	128[c]	
Scan time per sample	5 min	5 min	
Data Performance	**iScan/HiScan 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
CytoSNP-850K v1.2 legacy overlap with v1.1	99.00%	98.93%	

a. Final number of SNPs following analysis with GenTrain clustering algorithm for the Assay_C manifest file.
b. With automation, 24 BeadChips/batch, 2 batches/FTE/Tecan per week.
c. VWith 8 BeadChips/batch, 2 batches/FTE/Tecan per week.

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

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

Inclusive Coverage

The Infinium CytoSNP-850K v1.2 BeadChip contains ~ 850,000 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 v1.2 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.

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 3: Infinium CytoSNP-850K v1.2 BeadChip marker information

Marker Categories[b]	No. of Markers[a]	
	iScan/HiScan System	NextSeq 550 System
Total No. of markers	846,819	844,855
RefSeq genes	448,802	448,802
RefSeq +/- 10 kb	519,617	519,617
ADME genes	14,086	14,086
ADME +/- 10 kb	17,338	17,338
COSMIC genes	417,466	417,466
HLA markers	5046	5046
HLA genes	272	272
MHC markers	7572	7572
Exonic	68,503	68,503
Promoter regions	24,776	24,776
X chromosome markers	29,861	29,861
Y chromosome markers	1123	1123
PAR/Homologous markers	982	982

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-37 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

To learn more about the Infinium CytoSNP-850K v1.2 BeadChip and other Illumina genotyping products and services, visit www.illumina.com/applications/cytogenomics/kits.html

To learn more about BlueFuse Multi Software, visit www.illumina.com/clinical/clinical_informatics/bluefuse.html

Ordering Information

Product	Catalog No.
Infinium CytoSNP-850K v1.2 BeadChip Kit (8 samples)	20025643
Infinium CytoSNP-850K v1.2 BeadChip Kit (16 samples)	20025644
Infinium CytoSNP-850K v1.2 BeadChip Kit (48 samples)	20025645
Infinium CytoSNP-850K v1.2 BeadChip Kit (96 samples)	20025646

References

1. Clinical Genome Resource. www.clinicalgenome.org. Accessed July 2018.
2. Cancer Genomics Consortium. www.cancergenomics.org. Accessed July 2018.
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.

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