Infinium® CytoSNP-850K BeadChip

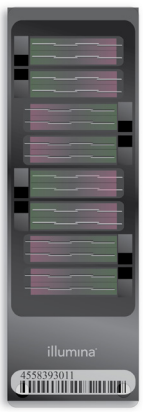
The Infinium CytoSNP-850K BeadChip provides the most comprehensive coverage of cytogenomic-relevant genes for constitutional and cancer applications. To demonstrate the superior performance of the CytoSNP-850K BeadChip, Illumina is offering a no-cost proof of principle study.

Highlights

- **Increased Performance and Detection Sensitivity**
  Over 850,000 SNPs with 15× redundancy
- **Current Content**
  Based on recent input from the international cytogenomics community and peer-reviewed literature
- **Suited for Constitutional and Cancer Applications**
  Enriched coverage for 3,262 dosage-sensitive genes providing robust performance across a broad range of sample types, including formalin-fixed paraffin-embedded (FFPE) tissues
- **True Gene-Level Coverage with an Exon-Centric Design**
  Constitutional coverage: contains updates and content evolution from the International Collaboration for Clinical Genomics (ICCG)[1]

  Cancer coverage: includes all Cancer Cytogenomics Microarray Consortium (CCMC) genes[2]
- **Higher Detection Sensitivity for Low-Level Mosaics**
  Long 50-mer SNP probes offer the highest signal-to-noise ratios of any arrays, facilitating greater detection sensitivity

Figure 1: Infinium CytoSNP-850K BeadChip



The eight-sample CytoSNP-850K BeadChip provides the most cytogenomic-relevant genes for constitutional and cancer studies with content from ICCG[1], CCMC[2], and Sanger.

Introduction

Structural variability is a substantial source of genetic variation that often has a major influence on phenotype. Accurate profiling of chromosomal aberrations, such as amplifications, deletions, rearrangements, and copy-neutral loss of heterozygosity (LOH) events, is crucial for studies associated with cancer and congenital disorders. To address this need, Illumina is introducing the Infinium CytoSNP-850K BeadChip.

The CytoSNP-850K BeadChip is the first SNP-based array to leverage the latest input on cytogenetic-relevant genes from the international community for constitutional and cancer applications. The gene list was defined based on content from the International Collaboration for Clinical Genomics (ICCG)[1] (formerly known as the International Standard of Cytogenomics Array (ISCA) consortium), the Cancer Cytogenetics Microarray Consortium (CCMC)[2], and Sanger, providing a comprehensive view of cytogenomic activity.

Inclusive Coverage

The CytoSNP-850K BeadChip contains 850,000 empirically selected single nucleotide polymorphisms (SNPs) spanning the entire genome with enriched coverage for 3,262 genes of known cytogenetics relevance in both constitutional and cancer applications. The high density of SNPs combined with an intelligent design based on the latest updates from peer-reviewed literature enables the detection of more relevant chromosomal aberrations relative to any other array technologies.

For greater detection sensitivity, the CytoSNP-850K BeadChip takes advantage of Infinium assay technology, which uses 50-mer SNP probes to generate higher signal-to-noise ratios than other commercially available arrays. This facilitates detection of low-level mosaics and more accurate breakpoint estimation for both copy number variations (CNVs) and absence of heterozygosity (AOH). Because each SNP is represented in at least 15× redundancy, CNV and AOH calls are more accurate across the genome. Only ten consecutive probes are necessary to make accurate CNV calls, demonstrating the high performance level of the Infinium assay.

Proof of Principle Study

To demonstrate the superior performance of the new CytoSNP-850K BeadChip, Illumina is offering researchers an eight-sample proof of principle (POP) study at no charge. This will enable you to see how the array performs using your own samples. In exchange, we simply ask that you consider adopting the CytoSNP-850K BeadChip for your cytogenomic applications.

Study Overview

The information below provides an overview of the POP study and process. For details, visit www.illumina.com/POPstudy.

Process

1. Contact your local Cytogenomics Specialist to discuss the details of your submission.
2. Fill out the Sample Submission Form and email it with the subject line “re: CytoSNP-850K BeadChip” to cytogenomics@illumina.com.
3. Select and prepare samples (see guidelines below). Send samples to Illumina/BlueGnome. Details are on the Submission Form available at www.illumina.com/POPstudy.
4. Review results with your local Cytogenomics Specialist.

Sample Guidelines

- Select samples representing a wide range of different chromosomal aberrations for both CNVs and AOH
- Include mosaic samples below 20%, samples representing uniparental isodisomy, and samples with unclear CNV calls on alternative microarray platforms to get the most out of your POP study
- Submit samples extracted from whole blood, cell line, saliva, bone marrow, and amniocytes. FFPE, fixed pellet, or blood spot samples will NOT be accepted
- Provide at least 1 µg (2 µg preferred) high-quality DNA for each sample:
  - Concentration: 50 ng/µl
  - Non-degraded: approximately 90% of the DNA > 10 Kb in size, as assessed on a 1% agarose gel
  - Inhibitor-free: examples include high concentrations of heme or chelating agents (i.e., EDTA); sample should be salt-free
  - Purity: $OD_{260/280}$ between 1.8 and 2.0
- Additional details provided with the Submission Form

Learn More

The CytoSNP-850K BeadChip provides the most comprehensive coverage of cytogenetically relevant genes on a proven platform, helping researchers find valuable information that may be missed by other technologies. Experience it for yourself when you participate in the no-cost POP study. Contact your local Cytogenomics Specialist to learn more today.

References

1. International Collaboration for Clinical Genomics (iccg.org)
2. Cancer Cytogenomics Microarray Consortium (www.cancergenomics.org)

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