# Infinium™ HumanCytoSNP-12 v2.1 BeadChip

A scalable, proven solution for genome-wide detection of genomic variants.

**Highlights**

- **High-Throughput, Scalable Solution**
  12-sample format enables low per sample cost compared to other methods
- **Proven Performance**
  Widely used Infinium HD assay generates high-sensitivity
- **Comprehensive Data Analysis**
  Intuitive BlueFuse® Multi Software streamlines copy number assessment and results reporting

## Introduction

Structural variability is a substantial source of genetic variation that can influence phenotypic changes, including growth and development. Accurate genome-wide profiling of chromosomal abnormalities, including amplifications, deletions, and rearrangements, is crucial for cancer and congenital studies. Traditional molecular cytogenomic methods for assessing chromosomal abnormalities enable copy number visualization, but often at low resolution and without an assessment of the allelic contribution. As a result, data from these methods can be inconsistent, and work-intensive as experimental procedures are repeatedly performed. The BeadChip (Figure 1) overcomes these challenges and offers a faster way to analyze multiple samples for chromosomal aberrations at higher resolution.

## High-Throughput, Scalable Solution

Using a 12-sample format, the proven Infinium HD assay, and user friendly analysis software, the BeadChip offers a scalable, reliable solution for cytogenomic studies, yielding accurate data and confident results. Reduced handling, more efficient scanning, and higher density assays contribute to higher sample throughput so studies are finished faster.

### One Assay, Multiple Applications

Using the BeadChip, researchers can process samples from various sources, with various purposes, in the same assay. The array offers SNP coverage across the entire genome, making it appropriate for use with prenatal, postnatal, and cancer samples. In addition, the BeadChip can be used with FFPE samples. By using a single array across multiple applications, cytogenetic laboratories can increase efficiency and scalability, offering more assays without purchasing and learning new technologies.

### Streamlined Workflow

Infinium HD BeadChips require only 200 ng DNA per sample, providing opportunities to use more limited sample sources. The simple, streamlined Infinium HD assay workflow requires minimal hands-on time and can be completed in just three days. Scanning is performed with the iScan™ or NextSeq™ 550 Systems. The iScan System uses advanced optics for high-resolution detection and high-throughput readout of assay results. The NextSeq 550 System combines array scanning capabilities with a robust sequencing system, maximizing applications while minimizing instrument costs.

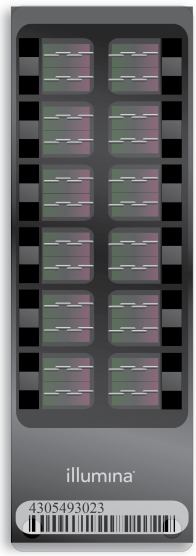


**Figure 1: The BeadChip —** The BeadChip offers a proven solution for efficient, accurate cytogenomic studies.

## Proven Performance

Taking advantage of the widely used Infinium HD assay, the BeadChip provides high, genome-wide single nucleotide polymorphism (SNP) coverage for analysis of structural variation. Generated data can be used to characterize common congenital disorders and profile reported copy number variation (CNV) regions, while providing dense coverage for novel CNV discovery. In fact, the clinical utility of the BeadChip has been demonstrated in numerous publications.[1,2]

### The Power of SNPs

SNP genotyping data offers significant advantages for cytogenomic analysis. High 15× bead redundancy increases the signal to noise (SNR), allowing for call rates typically > 99%. These features contribute to industry-leading accuracy and copy number data with lower noise, enabling copy number calls with as few as 10 contiguous SNPs. Every data point on the BeadChip combines B allele frequency and intensity (LogR), increasing the confidence and clarity of results. Using this information, absence of heterogeneity (AOH) up to ~3 MB

**Table 1: BeadChip Comprehensive Coverage**

| Feature | Description |
|---|---|
| **Overview** | |
| No. Markers per Sample | 299,140 |
| No. Samples per BeadChip | 12 |
| DNA Input (per sample) | 200 ng |
| Scan Time per Sample[a] | 3 min |
| **Minor Allele Frequency[b]** | |
| CEU / CHB + JPT / YRI (Median) | 0.21 / 0.19 / 0.19 |
| **Spacing (kb)** | |
| Mean / Median | 9.7 / 6.2 |
| 90th %ile Largest Gap | 18.7 |
| Resolution (targeted / overall) | ~ 62 kb / ~ 72 kb |
| **Marker Categories** | |
| Markers Within 10 kb of a RefSeq Gene | 148,967 |
| Non-Synonymous SNPs[c] | 2420 |
| MHC[d] / ADME[e] / Indel SNPs | 760 / 2388 / 0 |
| Sex Chromosome (X / Y / PAR Loci) | 15,400 /2972 /770 |
| Mitochondrial SNPs | 0 |

a. Scan times are approximations based on the iScan System
b. Based on HapMap ref 24
c. Based on RefSeq and Ensembl databases
d. Based on de Bakker PI, McVean G, Sabeti PC, et al. A high-resolution HLA and SNPhaplotype map for disease association studies in the extended human MHC. NatGenet. 2006;38(10):1166–72
e. Within 10 kb of 333 known ADME-related genes

can be resolved. Furthermore, certain types of structural lesions, such as. consanguinity, are only detectable using SNP genotyping. Low-level mosaicism is also amenable to study using SNP markers.

### Optimized for Cytogenetics

The BeadChip is optimized to detect cytogenetic abnormalities most relevant to human disease.[3] Content includes ~300,000 SNPs targeting regions shown to be important for cytogenetic analysis (Table 1). The result is dense coverage of ~250 disease regions, including subtelomeric regions, pericentromeric regions, and sex chromosomes, commonly screened in cytogenetics labs. Sufficient SNP coverage is provided to determine dosage sensitivity of > 800 genes.

### High-Quality Data

The BeadChips undergo rigorous functional testing to ensure strong and reproducible performance (Table 2). Data from the BeadChip show strong reproducibility (> 99.9%) and concordance with the International HapMap Project (> 99.2%). Additionally, this BeadChip provides precise copy number metrics with low overall noise levels, allowing reliable detection of CNVs with as few as 10 contiguous markers.

## Comprehensive Data Analysis

To support the study of cytogenomic-relevant genes, Illumina offers BlueFuse Multi Analysis Software. The easy-to-use graphical interface was designed with input from the cytogenomics community, providing analysts with greater control over data interpretation. BlueFuse Multi Software offers automated batch import from the primary scanner files, data processing and segmentation using validated algorithms, and automatic report generation for fast sample turnaround time and confident results reporting.

**Table 2: BeadChip High Data Quality**

| Parameter | Value from Reference Samples | Product Specification[a] |
|---|---|---|
| **Call Frequency** | 99.71% | > 99% average |
| **Reproducibility** | 100.00% | > 99.9% |
| **LogR Ratio[b]** | 0.10 | < 0.20 |
| **B Allele Frequency[b]** | 0.03 | < 0.040 |

a. Values expected for typical projects, excluding tumor samples or samples prepared not following standard Illumina protocols
b. Excludes sex chromosomes, mtDNA, and intensity-only loci. Heterozygotes only

## Summary

Combining a 12-sample format, the proven Infinium assay, and comprehensive BlueFuse Multi Analysis Software, the BeadChip offers a scalable solution for superior detection in cytogenomic studies.

## Learn More

To learn more about the BeadChip, visit www.illumina.com/clinical/reproductive-genetic-health.html

Learn more about BlueFuse Multi Software for molecular cytogenetics at www.illumina.com/clinical/clinical_informatics/bluefuse.html

## Ordering Information

| Product | Catalog Number |
|---|---|
| **HumanCytoSNP-12 v2.1 BeadChip (12 samples)** | WG-320-2101 |
| **HumanCytoSNP-12 v2.1 BeadChip (24 samples)** | WG-320-2102 |
| **HumanCytoSNP-12 v2.1 BeadChip (48 samples)** | WG-320-2103 |
| **HumanCytoSNP-12 v2.1 BeadChip (288 samples)** | WG-320-2104 |
| **HumanCytoSNP-12 v2.1 BeadChip (1152 samples)** | WG-320-2105 |
| **HumanCytoSNP-FFPE (24 samples)** | WG-321-1003 |
| **HumanCytoSNP-FFPE (48 samples)** | WG-321-1004 |
| **iScan System** | SY-101-1001 |
| **NextSeq 550 System** | SY-415-1002 |

## References


1. Srebniak MI, Boter M, Oudesluijs GO, et al. Genomic SNP array as a gold standard for prenatal diagnosis of foetal ultrasound abnormalities. *Mol Cytogenet*. 2012;5:14.
2. Data on file. Illumina, Inc. 2016.
3. Bejjani BA, Shaffer LG. Clinical utility of contemporary molecular cytogenetics. *Annu Rev Genomics Hum Genet*. 2008;9:71-86.


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