DATA SHEET

Infinium™ Global Screening Array with Cytogenetics-24 v1.0 BeadChip

A powerful efficient assay for genome-wide cytogenetic analysis

- Updated, disease-focused content for discovery and validation studies
- 700K genome-wide probes with high marker-to-exon coverage for robust cytogenetic analysis
- 80K supplemental markers selected to enhance cytogenetic assay performance
- Compatible with powerful cytogenetic analysis software for data analysis and visualization

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Introduction

Cytogenetic variation is a cause of numerous disorders, including cancers, developmental conditions, and fetal anomalies. Properly designed single nucleotide polymorphism (SNP) arrays can be used to assess chromosome structure, copy number, and segregation in a rapid and affordable assay. These microarrays offer researchers valuable insights into genetic mechanisms of human health and disease.

The Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip (Figure 1) is an easy-to-use solution for investigating cytogenetic variability. To create this BeadChip, 80K cytogenetic-informative markers were added to 620K proven disease-targeted markers from the Infinium Global Screening Array-24 v3.0 (Table 1). Analysis for the Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip can be performed with various software packages, including NxClinical (BioDiscovery) software, a focused solution for investigating chromosomal aneuploidies.

Table 1: Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip information

Feature	Description
Species	Human
Total number of markers[a]	~700K
Number of samples per BeadChip	24
DNA input requirement	200 ng
SNP replicates	15
Number of SNPs needed to call CNV	10
Assay chemistry	Infinium HTS
Instrument support	iScan™ System
Maximum iScan System sample throughput	~5760 samples/week
Scan time per sample[b]	~1.5 minutes

a. Total number of markers includes ~620K included in the Infinium Global Screening Array-24 plus ~80K cytogenetic-specific content
b. Approximate values, scan times, and maximum throughput will vary depending on laboratory and system configurations

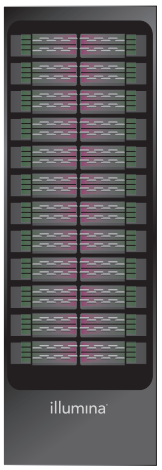

Figure 1: Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip—The BeadChip is built on the trusted 24-sample Infinium HTS platform with ~700K markers to create a versatile cytogenetic screening tool.

Optimized global and high-value content

The Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip builds on the success of the consortium version of the Infinium Global Screening Array-24 v3.0 that has been widely adopted by a community of human disease researchers, healthcare networks, consumer genomics companies, and genomic service providers. The multiethnic, genome-wide backbone includes variants from key scientific databases (Figure 2).[1-5]

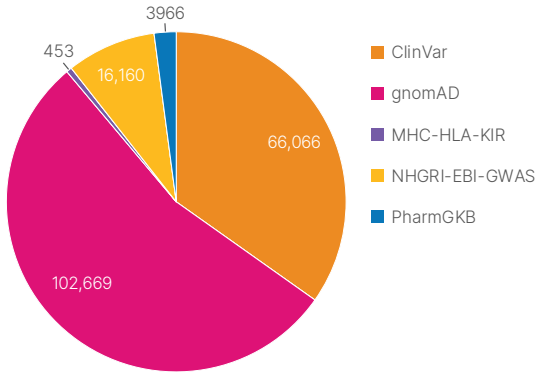

Figure 2: Clinical research content—BeadChip backbone content was expertly selected from reference scientific databases to create a highly informative assay for human health and disease research applications.

Extensive coverage of disease-associated variants

The extensive backbone content on the Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip supports multiple applications, including validation of disease associations, risk profiling, preemptive screening research, and pharmacogenomics studies. Variant selection includes pathology classifications based on National Center for Biotechnology Information (NCBI) ClinVar and American College of Medical Genetics (ACMG) ClinVar annotations (Figure 3A).[1,6] The BeadChip contains extensive coverage of phenotypes and disease classifications based on ClinVar andNational Human Genome Research Institute and European Bioinformatics Institute (NH GRI-EB I) genome wide association studies (GWAS) database catalog (Figure 3B).[4]

Focused content for cytogenetic research and discovery

To enable cytogenetic applications, the Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip is supplemented with 80K probes with a high marker-to-exon ratio categorized into four cytogenetic application tiers (Table 2). Content was carefully chosen through a cytogenetic consortium to create an exceptional tool for genome-wide copy number variation (CNV) coverage for oncology, prenatal, postnatal, reproductive health, and genetic disease research.

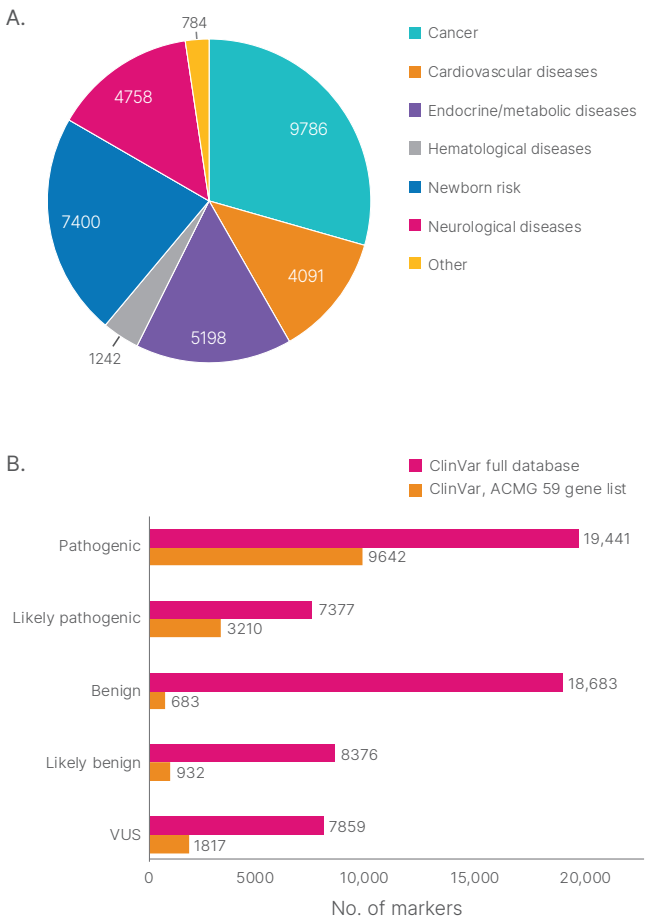

Figure 3: Broad coverage of disease categories—(A) Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip disease research content by category within the ClinVar database. Variant counts are subject to change. (B) Variants sorted by range of pathology classifications according to ACMG ClinVar annotations; VUS, variant of unknown significance.

Table 2: Cytogenetic application tiers included on the Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip

Application tier	Description	Genes in tier	Exons in tier	Average probe spacing	Median probes/exon	Exons with ≥ 1 probes	Exons with ≥ 3 probes
1	ClinGen pathogenic/likely pathogenic, haploinsufficient, and triploinsufficient[7]	409	6214	1.25 kb	5	> 99%	> 99%
2	DDG2P Genes associated with cancer[8]	1254	18,353	2.00 kb	4	> 99%	> 99%
3	Input from cytogenetics consortia Mendeliome Panel	2766	36,840	2.45 kb	3	> 99%	> 60%
4	OMIM Morbid Genes not otherwise tiered[9]	456	5434	2.82 kb	3	> 99%	> 60%
	Total	4885	66,841				

a. DDG2P: developmental disorders gene to phenotype

Optimized coverage and spacing for cytogenetic applications

Using SNP analysis to identify cytogenetic features requires careful selection of covered variants and sufficient spacing of markers across the target regions. The Infinium Global Screening Array with Cytogenetics-24 v1.0 was developed to ensure optimal coverage of key genes with spacing that supports CNV analysis when compared to other commercially available arrays (Figure 4).

QC markers for sample tracking

The marker backbone on the Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip also includes ~10K quality control (QC) markers. The QC marker content enables important sample tracking functions, including ancestry determination and stratification, and facilitates higher throughput studies (Figure 5).

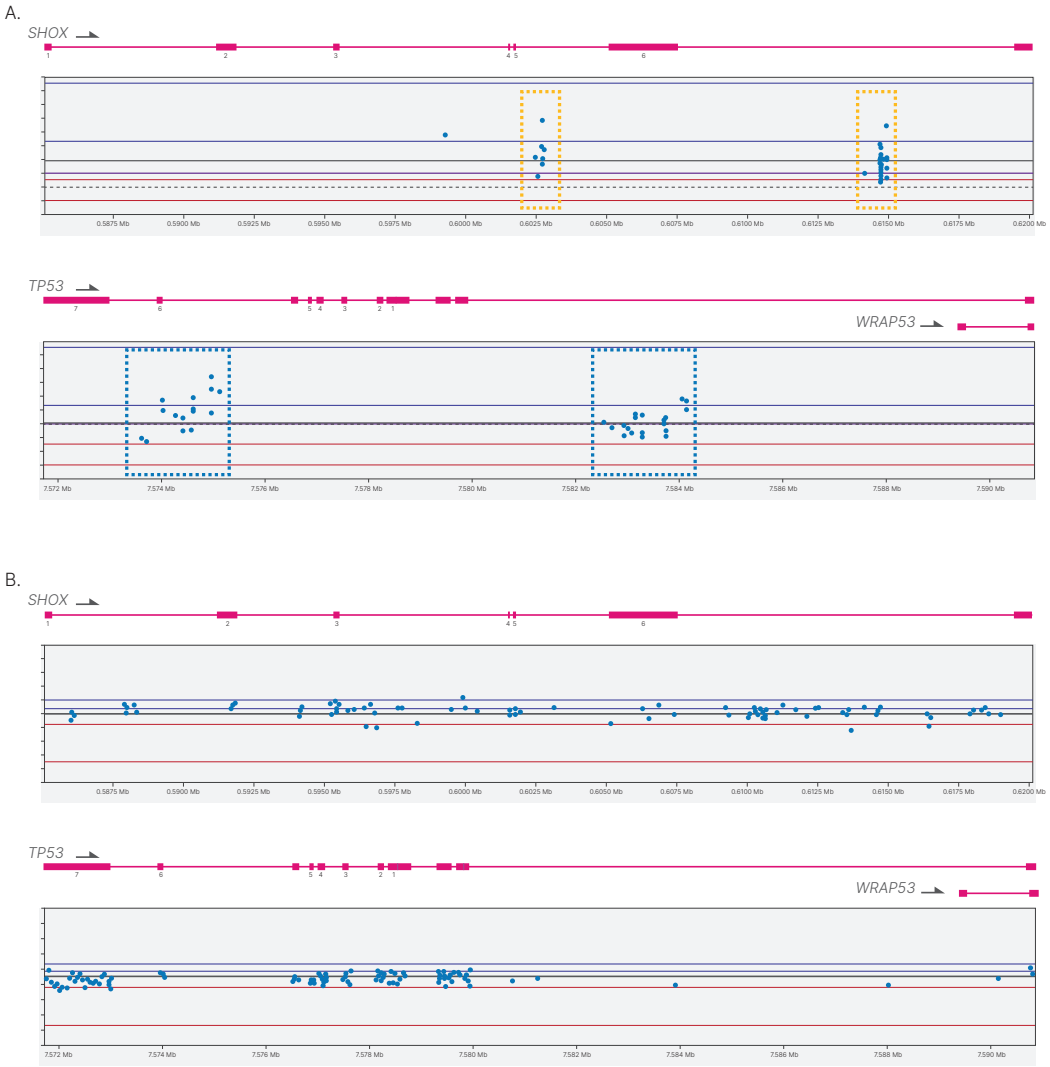

Figure 4: Cytogenetic variant coverage vs. similar cytogenetic array solution—(A) Other commercially available cytogenetics array with dense probe clusters (dashed gold boxes) and poor spacing and coverage in intronic and noncritical regions (dashed blue boxes), (B) Supplemental content in Infinium Global Screening Array with Cytogenetics-24 v1.0 designed with improved spacing and focus on high-exonic, high-value coverage of key genes supporting gene structure and copy number analysis.

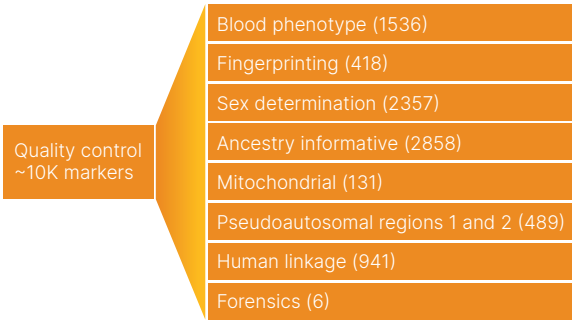

Figure 5: QC content by category—The BeadChip contains ~10K markers enabling various sample tracking functions such as sex determination, continental ancestry, human linkage, and more.

Trusted, high-quality assay

The Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip uses trusted Infinium assay chemistry to deliver the same level of high-quality, reproducible results that Illumina genotyping arrays have provided for over a decade (Table 3). In addition, the high signal-to-noise ratio inherent with individual genotyping calls in the Infinium assay provides the data quality necessary for the genome-wide CNV analysis featured on the array. The BeadChip is also compatible with the Infinium FFPE QC and DNA Restoration Kit, enabling genotyping of formalin-fixed, paraffin-embedded (FFPE) samples.

Powerful analysis for cytogenetic research

The Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip is compatible with multiple array analysis software platforms. For labs also looking for a software solution, NxClinical (BioDiscovery) software is available for fast, accurate, and comprehensive sample analysis. NxClinical software offers industry-standard algorithms, admin controls, and an integrated audit trail to ensure analysis accuracy and integrity. The easy-to-use software uses current research databases to support variant annotations and provides phenotype-associated variant ranking. After data are analyzed, NxClinical software offers an array of data visualization tools to help organize and present results.

Table 3: Data performance and spacing

Data performance	Observed[a]	Product specification[b]	
Call rate	99.7%	> 99.0 avg	
Reproducibility	99.99%	> 99.90	
Log R deviation	0.12[c]	< 0.30 avg[d]	
	Mean	Median	90th percentile[c]
Probe spacing	4.0 kb	2.0 kb	--
	Targeted	Backbone	
Resolution	~10 kb	~25 kb	

a. Values are derived from genotyping 2051 HapMap reference samples
b. Excludes Y chromosome markers for female samples
c. Based on results from GenTrain sample set
d. Value expected for typical projects using standard Illumina protocols; tumor samples and samples prepared by nonstandard protocols are excluded

High-throughput workflow

The Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip uses the proven 24-sample Infinium HT BeadChip format that enables laboratories to scale efficiently, as needed. The Infinium assay provides a three-day workflow that allows researchers to gather and report data and generate results quickly (Figure 6). For flexible throughput processing, the Infinium assay provides the capability to run up to 5760 samples per week using a single iScan™ System.

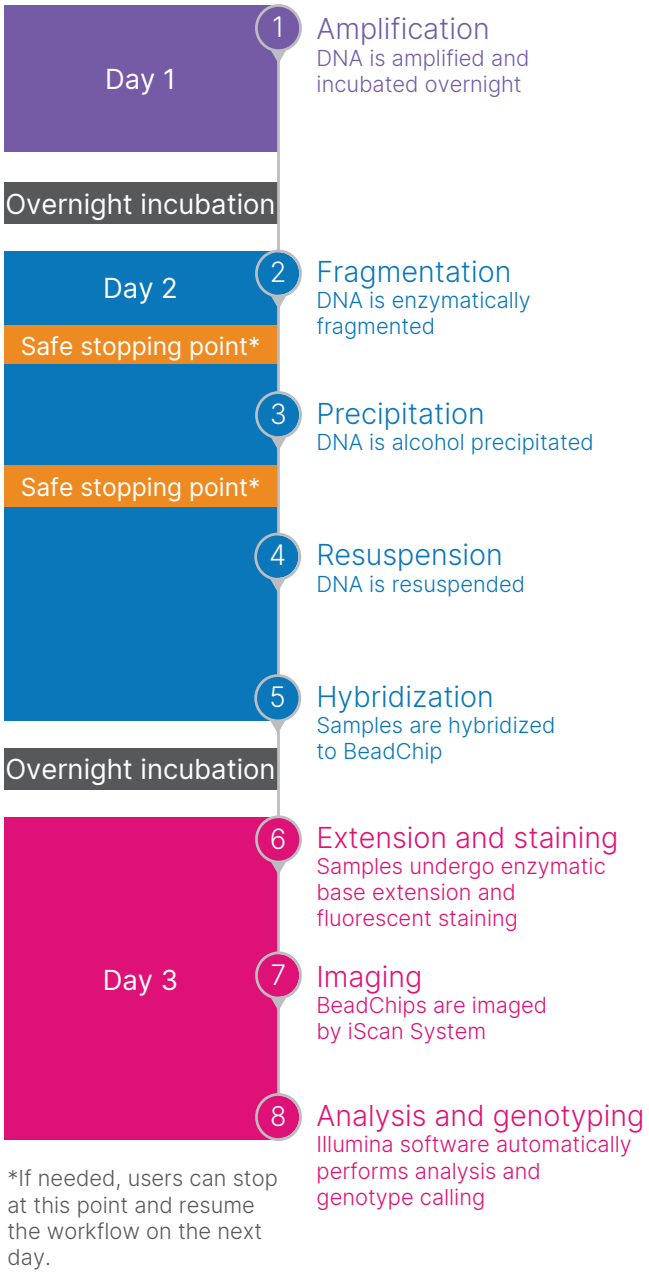

Figure 6: The Infinium 24-sample assay workflow—The Infinium HTS format provides a rapid three-day workflow with minimal hands-on time.

Ordering information

Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip	Catalog no.
48 samples	20066469
288 samples	20066470
1152 samples	20066471

Learn more

Infinium Global Screening Array with Cytogenetics-24 v1.0 BeadChip, https://www.illumina.com/products/by-type/clinical-research-products/infinium-global-screening-array-cytogenetics-24.html

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