DATA SHEET

Infinium™ Global Diversity Array with Cytogenetics-8 v1.0 BeadChip

A powerful efficient assay for genome-wide cytogenetic analysis

- Updated, disease-focused content for discovery and validation studies
- Exonic coverage of more than 4800 cytogenetic-relevant genes with 1.8M genome-wide probes
- 160K carefully selected SNP probes to enhance cytogenetic performance

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Introduction

Cytogenetic variation is a potential cause of a broad range of disorders, including cancers, developmental conditions, and fetal anomalies. Investigation of cytogenetic variation through the analysis of chromosome structure, copy number, and segregation provides valuable insights into genetic disorders and human health. The Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip is an easy-to-use solution for cytogenetic studies built on proven Infinium assay technology.

The BeadChip features carefully selected genotyping content from the Infinium Global Diversity Array-8 v1.0 plus supplemental exonic coverage of cytogenetic-relevant genes (Table 1, Figure 1). The carefully selected BeadChip content is analyzed optimally with NxClinical (BioDiscovery) analysis software to create a complete solution for rapid and cost-effective cytogenetic research.

Table 1: Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip information

Feature	Description
Species	Human
Total number of markers[a]	~1.8M
Number of samples per BeadChip	8
DNA input requirement	200 ng
SNP replicates	15
Number of SNPs needed to call CNV	10
Assay chemistry	Infinium LCG
Instrument support	iScan™ System
Maximum iScan System sample throughput[b]	~1728 samples/week
Scan time per sample[b]	3-5 minutes

a. The total number of markers includes ~1.6M genome wide backbone from Infinium Global Diversity Array-8 v1.0 plus 160k cytogenetic-specific content.
b. Approximate values, scan times, and maximum throughput will vary depending on laboratory and system configurations.

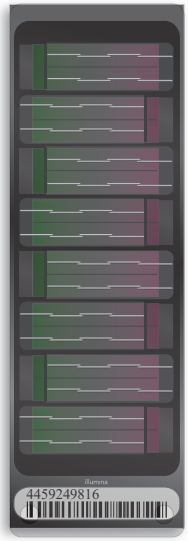

Figure 1: Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip—Economic cytogenetic analysis for up to 8 samples built on the trusted Infinium LCG platform. The BeadChip includes ~1.8M carefully selected markers and dedicated software analysis tools for research applications.

Content optimized for cytogenetic research and discovery

The Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip backbone contains ~1.6M current disease-focused markers sourced from the Infinium Global Diversity Array-8 v1.0 BeadChip—the array chosen by the National Institutes of Health for the All of Us research program. To ensure optimal cytogenetic performance, this backbone is supplemented with 160K cytogenetic-informative markers to create an assay that provides exceptional genome-wide copy number variation (CNV) analysis for oncology, reproductive health, prenatal, postnatal, and genetic disease applications. This supplemental marker content is divided into four cytogenetic application tiers, according to the research areas they support (Table 2). Probe spacing on the array is further optimized to ensure optimal coverage of key genes with spacing that supports CNV analysis, when compared to other commercially available arrays (Figure 2).

Table 2: Cytogenetic application tiers included on the Infinium Global Diversity Array with Cytogenetics-8 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[1]	409	6214	0.83 kb	5	> 99%	> 99%
2	DDG2P[a]: developmental disorders gene to phenotype, genes associated with cancer[2]	1254	18,353	0.89 kb	4	> 99%	> 99%
3	Input from cytogenetics consortia Mendeliome Panel	2766	36,840	0.97 kb	3	> 99%	> 60%
4	OMIM Morbid Genes not otherwise tiered[3]	456	5434	1.09 kb	3	> 99%	> 60%
	Total	4885	66,841				

a. DDG2P: developmental disorders gene to phenotype.

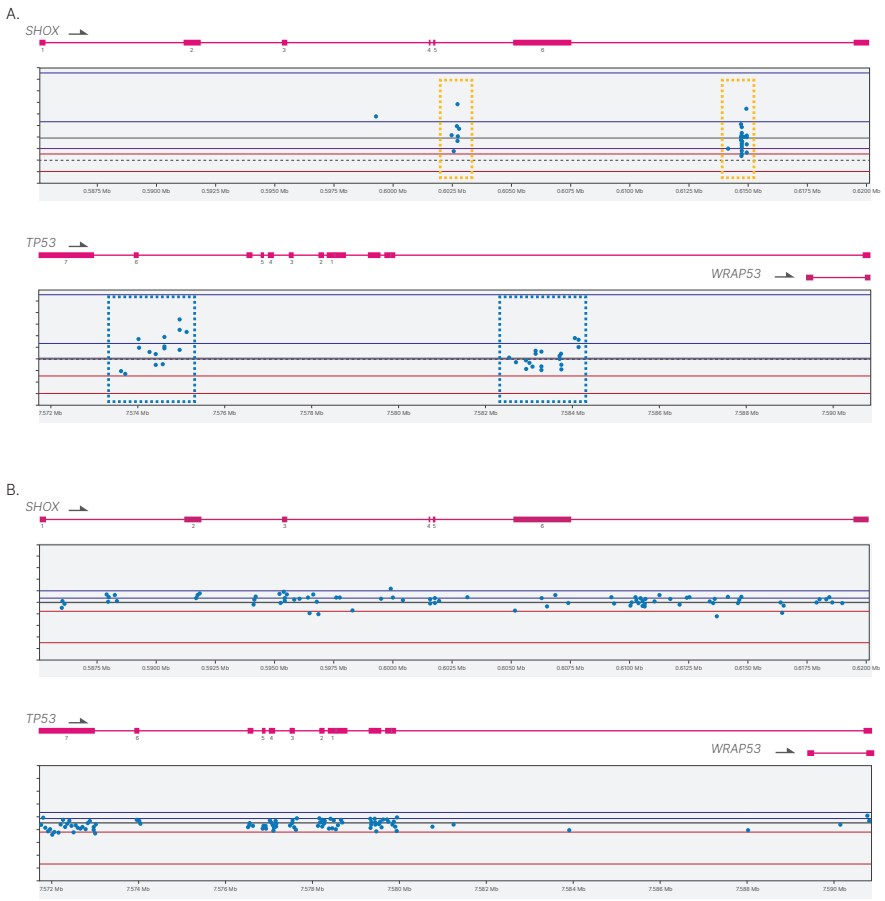

Figure 2: Example of cytogenetic variant coverage vs. similar cytogenetic array solution—(A) Other commercially available cytogenetics array with dense probe clusters (dashed gold boxes) resulting in poor spacing and probes in intronic and noncritical regions (dashed blue boxes), (B) The supplemental content in Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip is designed to ensure proper spacing, with focus on exonic regions, and high-value variant coverage in key genes that support CNV analysis.

Updated and relevant disease research content

The Infinium Global Diversity Array with Cytogenetics-8 v1.0 backbone is built on a strong foundation of disease research content. Clinical databases, such as ClinVar, are constantly evolving as new variants are added and variant designations change to "Pathogenic" or "Likely pathogenic." The BeadChip provides updated coverage of many of these high-value variants contained within annotated databases. Variants included on the array consist of markers with known disease association based on ClinVar,[4] the Pharmacogenomics Knowledgebase (PharmGKB),[5] and the National Human Genome Research Institute and European Bioinformatics Institute (NHGRI-EBI) genome-wide association studies (GWAS) database (Figure 3).[6]

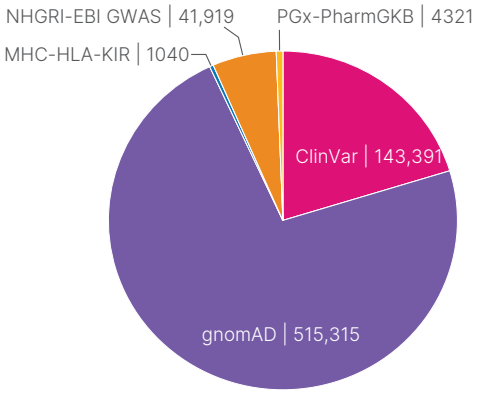

Figure 3: Clinical research content—Content was expertly selected from scientifically recognized databases to create a highly informative array for clinical research applications. Variant counts may be subject to change.

Exceptional coverage of disease-associated variants

Along with support for cytogenetic analysis, the Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip supports validation of a wide range of disease associations, risk profiling, preemptive screening research, and pharmacogenomics studies. Disease-associated variants have been selected from the NHGRI-EBI GWAS database to cover a broad range of phenotypes and disease classifications (Figure 4).[6] This content provides a powerful opportunity for researchers interested in studying diverse populations to test and validate associations previously found in European populations.

Additionally, the BeadChip content covers a range of pathology classifications based on ClinVar and American College of Medical Genetics (ACMG) annotations (Figure 5A).[7] There is also extensive coverage of phenotypes and disease classifications based on the ClinVar database (Figure 5B).[4] This representation of disease categories and classifications ensures the flexibility of the Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip for research and discovery.

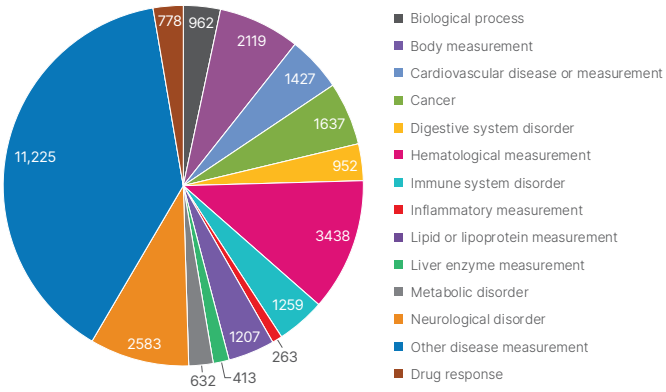

Figure 4: NHGRI-EBI disease categories—Clinical research content includes markers across a broad range of disease categories based on the NHGRI database.

QC markers for sample tracking

The Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip includes ~10K quality control (QC) markers. This QC marker content enables important sample tracking functions, ancestry determination, and stratification to facilitate higher throughput studies (Figure 6).

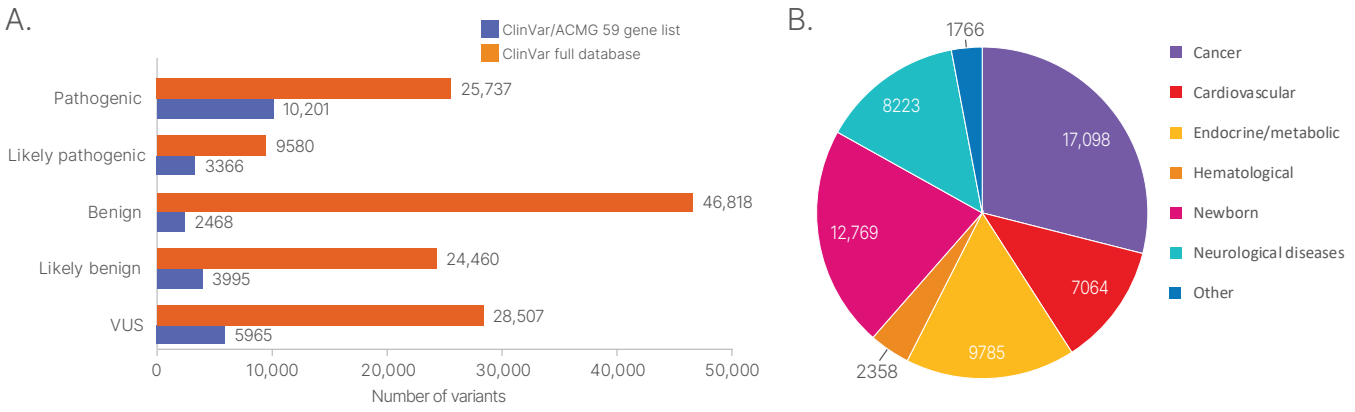

Figure 5: Broad coverage of disease categories—(A) Variants sorted by range of pathology classifications according to ClinVar American College of Medical Genetics (ACMG) annotations; VUS, variant of unknown significance. (B) Global Diversity Array clinical research content by category within the ClinVar database.

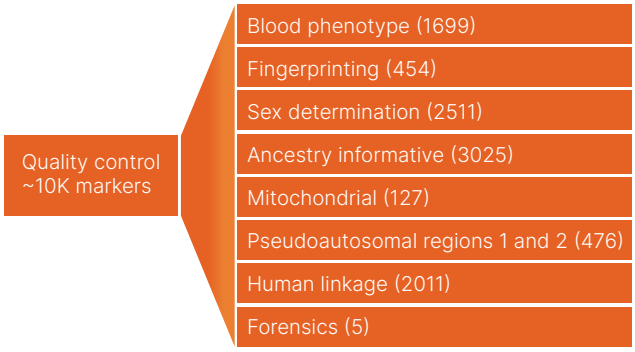

Figure 6: QC marker by category—QC variants on the array enable various capabilities for sample tracking such as sex determination, continental ancestry, human identification, and more.

Powerful analysis pipeline for cytogenetic research

The Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip provides data that is compatible with software analysis platforms. For labs looking for a complete cytogenetic workflow, the Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip is analyzed optimally with NxClinical (BioDiscovery) software, a premier cytogenetic software designed to make sample review fast, accurate, and comprehensive. The NxClinical software offers industry-standard algorithms, admin controls, and an integrated audit trail to ensure analysis integrity and accuracy. The easy-to-use software uses current clinical research databases to support variant annotations, and provides phenotype-associated variant ranking. Following data analysis, the NxClinical software includes an array of data visualization tools to help organize and present results.

Trusted, high-quality assay

The Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip uses trusted Infinium assay chemistry to deliver the same high-quality, reproducible data (Table 3) that Illumina genotyping arrays have provided for over a decade. The BeadChip is also compatible with the Infinium FFPE QC and DNA Restoration Kit, enabling genotyping of formalin-fixed, paraffin-embedded (FFPE) samples. In addition, the high signal-to-noise ratio inherent with the individual genotyping calls from the Infinium assay provides access to the genome-wide CNV calling featured on the Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip.

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]	
Spacing on tiers	**Mean**	**Median**	**90th percentile[c]**
Spacing (kb)	1.0 kb	0.5 kb	--
	Targeted	Backbone	
Resolution	~5 kb	~20 kb	

a. Values are derived from genotyping 2051 HapMap reference samples.
b. Excludes Y chromosome markers for female samples.
c. Based on results from the 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 Diversity Array with Cytogenetics-8 v1.0 BeadChip uses the proven 8-sample BeadChip format that enables laboratories to scale efficiently, as needed. The Infinium assay provides a three-day workflow that allows you to gather and report data quickly (Figure 7). For flexible throughput processing, the Infinium assay provides the capability to run up to 1728 samples per week using a single iScan™ System.

Figure 7: The Infinium platform provides a rapid three-day workflow with minimal hands-on time.

Ordering information

Product	Catalog no.
Infinium Global Diversity Array with Cytogenetics-8 v1.0 kit (8 samples)	20122861
Infinium Global Diversity Array with Cytogenetics-8 v1.0 kit (16 samples)	20066507
Infinium Global Diversity Array with Cytogenetics-8 v1.0 kit (48 samples)	20066508
Infinium Global Diversity Array with Cytogenetics-8 v1.0 kit (96 samples)	20066509
Infinium Global Diversity Array with Cytogenetics-8 v1.0 kit (384 samples)	20066510

Learn more

Infinium Global Diversity Array with Cytogenetics-8 v1.0 BeadChip

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