DATA SHEET

Infinium™ Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip

A powerful, pan-ethnic solution for carrier screening studies

- Updated content includes 45K ACMG-recommended carrier screening markers for broad disorder coverage
- Comprehensive coverage enables high detection rates for difficult-to-call disorders
- Flexible Infinium workflow integrates with Igentify Analyze software for simplified data analysis

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Introduction

Autosomal recessive and X-linked disorders are estimated to occur in up to 1 in 175 pregnancies.[1] Carrier screening can identify individuals at risk of passing on a recessive genetic disorder to their children. In approximately 2.5% of screened couples, both parents will be identified as carrying pathogenic variants in the same gene, resulting in a 1 in 4 chance of having an affected pregnancy.[2] Available ethnic-based carrier screening modalities are inherently biased in carrier identification and have the potential to miss heritable genetic variation in ethnically diverse populations.[3] Expanded carrier screening, however, enables researchers to adopt a more population-neutral approach to identify carriers, thereby promoting equity and inclusion. The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip is a high-performance microarray research solution that provides a cost-effective and scalable workflow to support pan-ethnic expanded carrier screening studies (Figure 1, Table 1).

The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip features updated content, including key carrier screening markers recommended by the American College of Medical Genetics (ACMG),[4] for comprehensive pan-ethnic, genome-wide coverage. Researchers can consolidate multiple workflows into a single automation-compatible assay to maximize throughput while significantly reducing hands-on time. The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip includes access to the Igentify Analyze software, which can detect both sequence and structural variation, resulting in high detection rates and low residual risk for key inherited disorders.

Optimized content for expanded carrier screening

Ethnic-based carrier screening products are estimated to identify only about 30% of the disorders included on expanded carrier panels.[3] The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip contains ~1.87M markers for comprehensive genome-wide coverage. These markers include 45K targeted carrier screening variants covering 602 carrier screening–specific genes, selected in partnership with Igentify, on the Infinium Global Diversity Array-8 v1.0 genome-wide backbone (Table 2, Table 3).

Figure 1: Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip—Comprehensive carrier screening analysis for up to 8 samples built on the trusted Infinium assay platform. The BeadChip includes 45K carefully selected markers for expanded carrier screening and dedicated software analysis tools for pan-ethnic, expanded disease coverage in a single assay.

Table 1: Infinium Global Diversity Array Carrier Screening Content-8 v2.0 BeadChip overview

Feature	Description
Species	Human
Total number of markers[a]	~1.87M
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	3–5 minutes

a. Content includes ~1.8M markers in the genome-wide backbone from Infinium Global Diversity Array-8 v1.0 plus 45K targeted carrier screening markers.
b. Approximate values, scan times, and maximum throughput will vary depending on laboratory and system configurations.

Table 2: Infinium Global Diversity Array Carrier Screening Content-8 v2.0 BeadChip high-value content

Content	No. of markers[a]	Research application/notes
RefSeq hg19 genes	1,067,594	All known genes
RefSeq hg19 ± 10 kb	1,191,774	Regulatory regions[b]
RefSeq Promoters	45,656	2 kb upstream to include promoter regions
ADME Exonic	18,423	Drug absorption, distribution, metabolism, and excretion
ADME hg19 genes	33,076	
ADME hg19 ± 10 kb	37,952	Includes regulatory regions
HLA markers	16,826	Disease defense, transplant rejection, and autoimmune disorders
HLA hg19 genes	1246	
MHC markers	21,828	
COSMIC hg19 genes	1,049,266	Somatic mutations in cancer
GO hg19 genes	343,108	Gene ontology annotation

a. The number of markers for each category is subject to change.
b. Of all known genes.

Abbreviations: ADME, absorption, distribution, metabolism, and excretion; COSMIC, catalog of somatic mutations in cancer; GO, gene ontology database; hg19, human genome version 19; HLA, human leukocyte antigen; MHC, major histocompatibility complex; RefSeq, National Center for Biotechnology Information Reference Sequence Database.

Table 3: Marker information

Marker category	No. of markers[a]
Exonic markers[a]	562,192
Nonsense markers[b]	37,157
Missense markers[b]	407,150
Synonymous markers[b]	33,527
Silent SNPs[b]	37,296
Mitochondrial DNA markers[b]	1298
Indels[c]	53,467
Chr X markers[c]	58,931
Chr Y markers[c]	5211
PAR/homologous markers[c]	1867

a. RefSeq, NCBI Reference Sequence Database, ncbi.nlm.nih.gov/refseq/.
b. Compared against the UCSC Genome Browser, genome.ucsc.edu/.
c. NCBI Genome Reference Consortium, Version GRCh37 (hg19), ncbi.nlm.nih.gov/assembly/GCF_000001405.13/.

Abbreviations: Indel, insertion/deletion; NCBI, National Center for Biotechnology Information, PAR, pseudoautosomal region; SNP, single nucleotide polymorphisms, UCSC, University of California Santa Cruz.

This high-value content provides 97% coverage of genes for disorders with a carrier frequency of 1 in 200 or higher. This includes all Tier 1-3 ACMG-recommended carrier screening genes, with the exception of *FMR1* and *FXN*. As a result, the Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip enables high detection rates and low residual risk with comprehensive coverage of important inherited disorders (Table 4).

Exceptional coverage of exonic content

The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip includes enhanced tagging in exonic regions and enriched coverage to map genome-wide association study (GWAS) loci with previously identified disease or trait associations with precision. More than 400,000 markers of exome content were gathered from 36,000 individuals of diverse ethnic groups, including African Americans, Hispanics, Pacific Islanders, East Asians, and individuals of mixed ancestry. The Global Diversity Array also features diverse exonic content from the ExAC database,[5] including both cross-population and population-specific markers with either functionality or strong evidence for association (Table 5).

Table 4: Examples of high detection rates across disorders and ancestries[a]

Detection rates for ACMG Tier 1 screening	
Cystic fibrosis	~99%
Spinal muscular atrophy	~90%
Detection rates for Tay Sachs disease across ancestries	
African American	97%
Ashkenazi Jewish	98%
Asian	97%
Caucasian	97%
General population	97%
Detection rates across disorders in African American ethnicities	
Alpha thalassemia	90%
Beta hemoglobinopathies	99%
Canavan disease	99%
Congenital adrenal hyperplasia	50%
Cystic fibrosis	99%
Fanconi anemia, complementation group C	98%
Galactosemia	99%
Gaucher disease	90%
Medium-chain acyl-CoA dehydrogenase deficiency	99%

a. Data presented here are for representative purposes only, the list of disorder and ancestries is not exhaustive. Data on file for ~300 disorders with detection rates.[6]

Table 5: Exonic coverage across populations

Population(s)[a,b]	No. of markers
NFE	346,340
EAS	146,281
AMR	272,178
AFR	257,690
SAS	224,431
NEF/EAS/AMR/AFR/SAS	69,432

a. internationalgenome.org/category/population.
b. Based on gnomAD, gnomad.broadinstitute.org/.

Abbreviations: NFE, non-Finnish European; EAS, East Asian; AMR, admixed American; AFR, African; SAS, South Asian.

Sequence and structural variation detected in a single assay

Common inherited conditions may be caused by more than one type of genetic variation, including sequence and structural variants (Figure 2). Multiple assays, including PCR, digital PCR (dPCR), multiplex-ligation dependent probe amplification (MLPA), and microarrays may be needed to detect each type of variation in a single sample. The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip consolidates multiple workflows into a single assay that detects sequence and structural variants, reducing turnaround time[6] and increasing laboratory output.

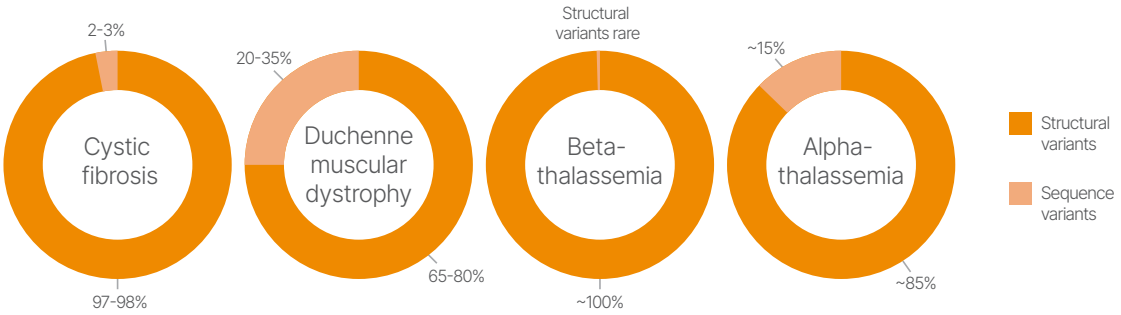

Figure 2: Examples of variable contribution of structural and sequence variation in inherited conditions—Sequence and structural variation underlying complex heritable conditions typically requires multiple assays for accurate detection. The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip consolidates multiple workflows into a single assay for reduced turnaround time and fast results, covering sequence and structural variants for ~300 inherited disorders.[6]

In addition, the Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip can be combined with content for several other research applications, including cytogenetics, polygenic risk score, pharmacogenomics, whole-genome sequencing quality control, cancer research, and more, creating a powerful all-in-one tool for clinical research.

Trusted, high-quality assay

The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip uses trusted Infinium assay chemistry to deliver the same high-quality, reproducible data (Table 6) that Illumina genotyping arrays have provided for over a decade. 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 Carrier Screening Content-8 v2.0 BeadChip.

High-throughput workflow

The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip uses the 8-sample 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 quickly (Figure 3). For flexible throughput processing, the automation-compatible Infinium assay provides the capability to run up to 1728 samples per week using a single iScan™ System.

Table 6: Data performance

Performance metric	Value[a]	Product specification[b]
Call rate	99.61%	> 99% avg
Reproducibility	99.99%	> 99.90%
LogR deviation	0.1448	< 0.30 avg[c]

a. Values are derived from genotyping 2228 HapMap reference samples.
b. Excludes Y chromosome markers for female samples.
c. Values expected for typical projects using standard Illumina protocols. Tumor samples and samples prepared by nonstandard protocols are excluded.

Powerful analysis pipeline

Data from the Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip are analyzed optimally with Igentify Analyze software, a premier reporting software designed to make sample review fast, accurate, and comprehensive. The Igentify Analyze software offers industry-standard algorithms, admin controls, and an integrated audit trail to ensure analysis integrity and accuracy. The easy-to-use software leverages current clinical research databases to support variant annotations and provides phenotype-associated variant ranking. Using specialized algorithms, the Igentify Analyze software enables calling of structural and difficult-to-call variants. Following data analysis, the Igentify Analyze software includes an array of data visualization tools to help organize and present results.

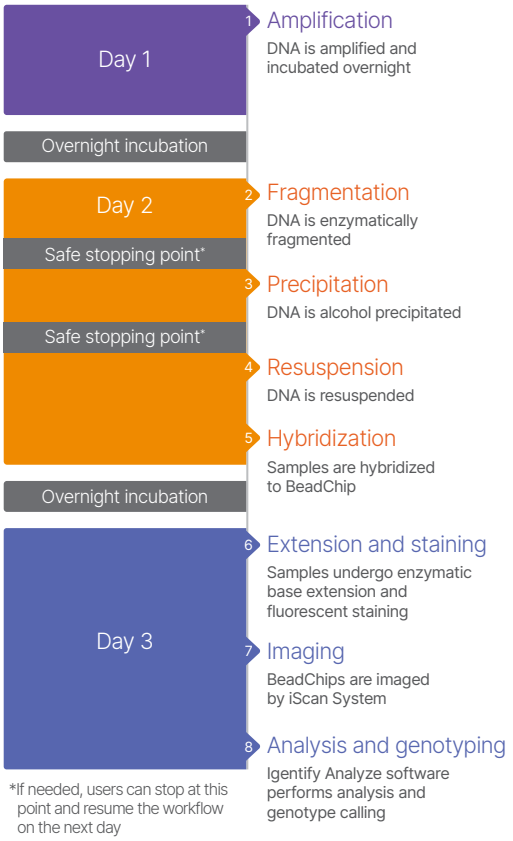

Figure 3: Infinium LCG workflow—The Infinium assay uses a rapid three-day workflow requiring minimal hands-on time.

Summary

The Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip is a comprehensive, pan-ethnic solution for expanded carrier screening. High-value content including 45K carrier screening markers on a 1.8M genome-wide backbone provides broad coverage across common inherited conditions. The simple Infinium workflow combined with the easy-to-use and customizable Igentify Analyze software enables researchers to reduce turnaround time while accessing high detection rates for historically difficult-to-call disorders.

Learn more

Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip, illumina.com/products/by-type/microarray-kits/infinium-global-diversity-array-carrier-screening

Infinium Global Diversity Array, illumina.com/products/by-type/microarray-kits/infinium-global-diversity

Igentify Analyze software, igentify.com/products/igentify-analyze/

Ordering information

Product	Catalog no.
Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip (16 samples)	20076135
Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip (48 samples)	20076136
Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip (96 samples)	20076137
Infinium Global Diversity Array with Carrier Screening Content-8 v2.0 BeadChip (384 samples)	20076138

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2. Haque IS, Lazarin GA, Wapner RJ. Prenatal Carrier Screening. *JAMA*. 2016;316(24):2675-2676. doi:10.1001/jama.2016.17401
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6. Data on file. Illumina, Inc. 2022.

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1.800.809.4566 toll-free (US) | +1.858.202.4566 tel
techsupport@illumina.com | www.illumina.com

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