

# Infinium® Multi-Ethnic EUR/EAS/SAS BeadChip

A cost-effective array for understanding complex disease in European, East Asian, and South Asian populations.

#### Introduction

The Infinium Multi-Ethnic EUR/EAS/SAS BeadChip harnesses content from Phase 3 of the 1000 Genomes Project (1kGP)¹, Consortium on Asthma among African-ancestry Populations in the Americas (CAAPA), Population Architecture using Genomics and Epidemiology (PAGE), T2D-Genes Consortium, OMIM, ClinVar, ACMG, carrier screening panels, and other resources to create a multipurpose, multiethnic array. With > 1.4 million expertly selected markers, the Infinium Multi-Ethnic EUR/EAS/SAS BeadChip enables identification of genetic associations with both common and rare traits providing insight across European, East Asian, and South Asian populations to epidemiologists, health care researchers, population geneticists, and genomic researchers (Table 1–5).

## **Maximized Imputation Accuracy**

Consortium partners developed content for the Infinium Multi-Ethnic EUR/EAS/SAS BeadChip using tagging strategies with the power to perform more effective association studies in European, East Asian, and South Asian populations. The novel algorithm selects population-specific and transethnic tag SNPs that maximize imputation accuracy, as imputation has become a standard practice in the interpretation of genotyping data and allows for more accurate statistical inference of genotypes not directly genotyped.

### **Expert-Selected Content**

The Infinium Multi-Ethnic EUR/EAS/SAS BeadChip combines expertly selected markers and content from the most popular Illumina commercial arrays with the most current genomic information. Researchers can detect both common and rare variants across European, East Asian, and South Asian populations and impute variants in a vast number of subpopulations.

The Infinium Multi-Ethnic EUR/EAS/SAS BeadChip contains the following content:

- Highly informative European, East Asian, and South Asian genomewide tag SNPs from the Infinium HumanCore-24 BeadChip
- Genome-wide coverage for diverse populations selected by PAGE using a new cross-population tagging strategy
- Total exonic content of > 380,000 markers including the Infinium Exome-24 BeadChip content and Multiethnic exome content designed by PAGE
- Over 17,000 variants chosen to be relevant to clinical and pharmacogenetic studies and 23,000 hand-curated variants picked for functional, immunological, oncological, ancestry, and forensic applications

Table 1: Multi-Ethnic EUR/EAS/SAS BeadChip Product Information

Feature		Description		
Total No. of Markers		1,475,140		
Capacity for Custom Markers		245,000		
No. Samples per BeadChip		8		
DNA Input		200 ng		
Assay Chemistry		Infinium® LCG		
Instrument Support		iScan® or HiScan® System		
Sample Throughput <sup>a</sup>		~ 1067 samples/week		
Scan Time per Sample		iScan System 11.3 min	HiScan 6.5 min	System
Data Performance		Value <sup>b</sup>	Product S	Specification
Call Rate		99.87%	> 99% avç	g.
Reproducibility		99.99%	> 99.9%	
Log R Deviation		0.10	< 0.30	
Spacing	Mean	Media	n	90 <sup>th</sup> % <sup>c</sup>
Spacing (kb)	1.98	0.92		4.94

- Estimated sample throughput based on use of 1 HiScan System, 1 AutoLoader 2.x, 1
  Tecan robot, and a 5-day work week.
- b. Values are derived from genotyping 708 HapMap reference samples.
- c. Value expected for typical projects using standard Illumina protocols. Tumor samples and samples prepared by methods other than standard Illumina protocols are excluded.

Table 2: Imputation Accuracy for European, East Asian, and South Asian Populations from 1kGP at Different MAF Thresholds

	Minor Allele Frequency (MAF) Threshold		
Population <sup>b</sup>	0.5–1%	1–5%	≥ 5%
EAS	57.3%	82.4%	96.3%
EUR	70.2%	87.9%	97.1%
SAS	61.5%	84.7%	96.5%
a. EAS: East Asian; EUR: European; SAS: South Asian.1			

Table 3: LD Mean r<sup>2</sup> for European, East Asian, and South Asian Populations from 1kGP at Different MAF Thresholds

	Minor Allele Frequency (MAF) Threshold		
Population <sup>a</sup>	0.5–1%	1–5%	≥ 5%
EAS	0.300	0.636	0.873
EUR	0.307	0.616`	0.870
SAS	0.302	0.609	0.854
a. EAS: East Asian; EUR: European; SAS: South Asian.1			

Table 4: Multi-Ethnic EUR/EAS/SAS BeadChip Marker Information

Marker Category		No. of Mar	kers
Exonic Markers		381,752	
Intronic Markers		522,294	
Nonsense Markers		10,826	
Missense Markers		318,990	
Synonymous Markers		18,228	
Mitochondrial Markers		645	
Indels		11,871	
Sex Chromosomes	X 39,507	Y 1598	PAR / Homologous 265

# **Ordering Information**

Infinium Multi-Ethnic EUR/EAS/SAS-8 v1.0 Kit	Catalog No.
16 samples	20001100
48 samples	20001101
96 samples	20001102
384 samples	20001103
Infinium Multi-Ethnic EUR/EAS/SAS-8+ v1.0 Kita	Catalog No.
16 samples	20001104
48 samples	20001105
96 samples	20001106
384 samples	20001107
a. Enabled for additional custom content.	

#### Learn More

To learn more about the Infinium Multi-Ethnic EUR/EAS/SAS v1.0 BeadChip, visit

www.illumina.com/techniques/popular-applications/genotyping.html

#### References

- 1. 1000 Genomes (www.1000genomes.org). Accessed July 21, 2015.
- COSMIC: Catalogue of somatic mutations in cancer (http://cancer.sanger. ac.uk/cosmic). Accessed July 21, 2015.
- Gene Ontology Consortium (www.geneontology.org). Accessed July 21, 2015.

Table 5: Multi-Ethnic EUR/EAS/SAS BeadChip High-Value Content

Content	No. of Markers	Research Application / Note
ADME Core and Extended Genes <sup>a</sup>	14,342	Drug metabolism and excretion
ADME Core and Extended Genes <sup>a</sup> +/- 10 kb	17,598	Drug metabolism and excretion (+ regulatory regions)
APOE	73	Cardiovascular disease, Alzheimer's disease, immunoregulation, and cognition
Blood Phenotype Genes	2754	Blood phenotypes
COSMIC Genes <sup>a</sup>	774,774	Somatic mutations in cancer
GO CVS Genesa	231,232	Cardiovascular conditions
Database of Genomic Variants	1,155,179	Genomic structural variation
eQTLs	6091	Genomic loci regulating mRNA expression levels
Fingerprint SNPs	475	Human identification
HLA Genes	846	Disease defense, transplant rejection, and autoimmune disorders
Extended MHC <sup>a</sup>	16,499	Disease defense, transplant rejection, and autoimmune disorders
KIRª	120	Autoimmune disorders and disease defense
Neanderthal SNPs	1816	Neanderthal ancestry and human population migration
NHGRI GWAS Catalog <sup>a</sup>	10,024	Markers from published genome- wide association studies
RefSeq 3' UTRs	34,396	3' untranslated regions of known genes
RefSeq 5' UTRs	23,912	5' untranslated regions of known genes
RefSeq All UTRs	56,624	All untranslated regions of known genes
RefSeq	852,999	All known genes
RefSeq +/-10 kb	959,082	All known genes +/- 10 kb to include regulatory regions
RefSeq Promoters	33,940	2 kb 5' of all known genes to include promoter regions
RefSeq Splice Regions	5087	Variants at splice sites in all known genes

a. ADME: absorption, distribution, metabolism, and excretion; COSMIC: catalog of somatic mutations in cancer<sup>2</sup>; GO CVS: Gene Ontology annotation of the cardiovascular system<sup>2</sup>; eQTL: expression quantitative trait loci; HLA: human leukocyte antigen; KIR: killer cell Ig-like receptor; MHC: major histocompatibility complex; NHGRI: national human genome research institute.

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