illumina®

Infinium® Multi-Ethnic AMR/AFR BeadChip

A cost-effective array for understanding complex disease in Hispanic and African American populations.

Introduction

The Infinium Multi-Ethnic AMR/AFR 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 AMR/AFR BeadChip enables identification of genetic associations with common and rare traits, providing insight across Hispanic and African American 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 AMR/AFR BeadChip using tagging strategies with the power to perform more effective association studies in Hispanic and African American 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 AMR/AFR 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 Hispanic and African American populations and impute variants in a vast number of subpopulations.

The Infinium Multi-Ethnic AMR/AFR BeadChip contains the following content:

- Highly informative Hispanic and African American genome-wide tag SNPs from the Infinium HumanCore-24 BeadChip
- African Diaspora Consortium Power Chip content identified through sequencing of 692 individuals by CAAPA
- 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 AMR/AFR BeadChip Product Information

Feature	Description		
Total No. of Markers	1,430,141		
Capacity for Custom Bead Types	245,000		
No. Samples per BeadChip	8		
DNA Input Requirement	200 ng		
Assay Chemistry	Infinium® LCG		
Instrument Support	iScan® or HiScan® System		
Sample Throughput[a]	~ 1067 samples/week		
Scan Time per Sample	iScan System 11.3 min	HiScan System 6.5 min	
Data Performance		**Value[b]**	**Product Specification**
Call Rate		99.87%	> 99% avg.
Reproducibility		99.99%	> 99.9%
Log R Deviation		0.10	< 0.30
Spacing	**Mean**	**Median**	**90th %[c]**
Spacing (kb)	2.05	0.89	5.17

a. 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 prepared by methods other than standard Illumina protocols are excluded.

Table 2: Imputation Accuracy for Hispanic and African Populations from 1kGP at Different MAF Thresholds

	Minor Allele Frequency (MAF) Threshold		
Population[a]	**0.5–1%**	**1–5%**	**≥ 5%**
AFR	77.1%	89.0%	95.7%
AMR	81.3%	89.1%	96.8%

a. AFR: African; AMR: Ad-mixed American.[1]

Table 3: LD Mean r^2 for Hispanic and African Populations from 1kGP at Different MAF Thresholds

	Minor Allele Frequency (MAF) Threshold		
Population[a]	**0.5–1%**	**1–5%**	**≥ 5%**
AFR	0.162	0.406	0.664
AMR	0.414	0.593	0.830

a. AFR: African; AMR: Ad-mixed American.[1]

Table 4: Multi-Ethnic AMR/AFR BeadChip Marker Information

Marker Category			
Exonic Markers	381,196		
Intronic Markers	499,952		
Nonsense Markers	10,587		
Missense Markers	318,693		
Synonymous Markers	18,163		
Mitochondrial Markers	645		
Indels	11,779		
Sex Chromosomes	X 39,165	Y 2360	PAR / Homologous 3753

Ordering Information

Infinium Multi-Ethnic AMR/AFR-8 v1.0 Kit	Catalog No.
16 samples	20001090
48 samples	20001091
96 samples	20001092
384 samples	20001093
Infinium Multi-Ethnic AMR/AFR-8+ v1.0 Kit[a]	**Catalog No.**
16 samples	20001094
48 samples	20001095
96 samples	20001096
384 samples	20001097

a. Enabled for additional custom content.

Learn More

To learn more about the Infinium Multi-Ethnic AMR/AFR v1.0 BeadChip and other Illumina genotyping products and services, visit www.illumina.com/techniques/popular-applications/genotyping.html

References

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

Table 5: Multi-Ethnic AMR/AFR BeadChip High-Value Content

Content	No. of Markers	Research Application / Note
ADME Core and Extended Genes[a]	14,981	Drug metabolism and excretion
ADME Core and Extended Genes[a] +/- 10 kb	18,114	Drug metabolism and excretion (+ regulatory regions)
APOE	75	Cardiovascular disease, Alzheimer's disease, immunoregulation, and cognition
Blood Phenotype Genes	2667	Blood phenotypes
COSMIC Genes[a]	754,633	Somatic mutations in cancer
GO CVS Genes[a]	200,973	Cardiovascular conditions
Database of Genomic Variants	1,120,149	Genomic structural variation
eQTLs	6025	Genomic loci regulating mRNA expression levels
Fingerprint SNPs	474	Human identification
HLA Genes	833	Disease defense, transplant rejection, and autoimmune disorders
Extended MHC[a]	16,050	Disease defense, transplant rejection, and autoimmune disorders
KIR[a]	117	Autoimmune disorders and disease defense
Neanderthal SNPs	1705	Neanderthal ancestry and human population migration
NHGRI GWAS Catalog[a]	10,053	Markers from published genome-wide association studies
RefSeq 3' UTRs	34,398	3' untranslated regions of known genes
RefSeq 5' UTRs	23,818	5' untranslated regions of known genes
RefSeq All UTRs	56,579	All untranslated regions of known genes
RefSeq	830,329	All known genes
RefSeq +/-10 kb	933,485	All known genes +/- 10 kb to include regulatory regions
RefSeq Promoters	33,503	2 kb 5' of all known genes to include promoter regions
RefSeq Splice Regions	4947	Variants at splice sites in all known genes

a. ADME: absorption, distribution, metabolism, and excretion; COSMIC: catalog of somatic mutations in cancer[2]; GO CVS: Gene Ontology annotation of the cardiovascular system[3]; 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.

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

For Research Use Only. Not for use in diagnostic procedures.

illumina®

AGAATGATAACAGTAACACACTTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGCTACCGTCTTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAAC
TCAACGTACCGTAACGAACGTATCATTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGCTACCGTGCAACGACGAAAAGAATGATAACAGTAACACACTTCTGTTAACCTT
CGACGAAAAGAATGATAACAGTAACACACTTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGCTACC
AACGTACCATTAAGAGCTACCGTGCAACAGTAACACACTTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGCTACCGTGCAACGACGAAAAGAATGATAA
AGAATGATAACAGTAACACACTTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGCTACCGTCTTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAAC
GATTACTTGATCCACTGATTCAACGTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGCTTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAGCAACGAGC
CGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGTCTGTTAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAGAGCTACCGTGCAACGAAAAGAATGATAACAGTA