

# Infinium<sup>™</sup> OmniZhongHua-8 v1.4 BeadChip

Exceptional coverage of common, intermediate, and rare variants specific to Chinese populations.

## Overview

The Infinium OmniZhongHua-8 v1.4 BeadChip (Figure 1) delivers exceptional coverage of common, intermediate, and rare variants found within Chinese populations for genome-wide association studies (GWAS). Optimized tag single nucleotide polymorphism (SNP) content from all three HapMap phases and the 1000 Genomes Project (1kGP)<sup>1</sup> has been strategically selected to create a population-based array for the discovery of novel disease- and trait-associatons in Chinese populations (Table 1 and Table 2). Using the proven Infinium HD assay, the HiScan™ or iScan™ System, and integrated analysis software, the Infinium OmniZhongHua-8 v1.4 BeadChip combines exceptional data quality with high sample throughput for comprehensive DNA analysis solution.

# Comprehensive Coverage

The Infinium OmniZhongHua-8 v1.4 BeadChip provides coverage of 77% of common variation (minor allele frequency (MAF) > 5%), 73% of intermediate variation (MAF > 2.5%), and 65% of rare variation (MAF > 1%) in the Chinese population at  $r2 \ge 0.8$ . This powerful chip provides greater coverage of intermediate and rare variation than the competing CHB array. It also offers equivalent coverage of common variation, making it the ideal starting point for Chinese population GWAS studies (Figure 2).



Figure 1: The Infinium OmniZhongHua-8 v1.4 BeadChip—The Infinium OmniZhongHua-8 v1.4 BeadChip provides exceptional coverage of common, intermediate, and rare SNP variants specific to Chinese populations as defined by the 1000 Genomes Project.

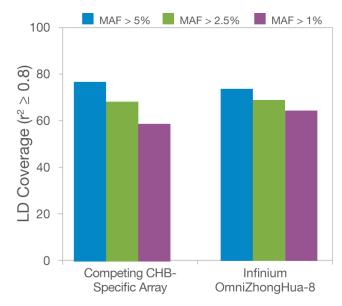


Figure 2: Array coverage comparison in Chinese populations — Coverage calculations based on known common and rare Chinese population variants from the International HapMap Project and 1000 Genomes Project.

## **Proven Infinium Quality**

The whole-genome Infinium HD assay underlies the power of the Omni family of microarrays. Genetic researchers worldwide use the Infinium HD assay for disease research, amassing a vast publication record. The Infinium OmniZhongHua-8 v1.4 BeadChip enables profiling of > 1,175,489 markers per sample. The assay is deployed with BeadArray technology from Illumina, delivering exceptionally high data quality for call rates (average > 99%), reproducibility (> 99.9%), and low sample repeat rates. High signal-to-noise ratios and low overall noise levels allow for precise, reliable calls and copy-number analyses.

Table 1: Product information

Feature	Description	Description			
Species	Human				
Total number of markers	1,175,489				
Number of samples per BeadChip	8 Samples				
DNA input requirement	200 ng				
Assay chemistry	Infinium HD Super				
Instrument support	iScan or HiScan System				
Sample throughput <sup>a</sup>	~ 960 samples/week				
Scan time per sample	iScan System	HiScan System	1		
	~7.5 min	~7.5 min			
LD coverage (r <sup>2</sup> ≥ 0.80)	1kGP MAF > 5%	1kGP MAF > 2.5%	1kGP MAF > 1%		
CHB	0.77	0.73	0.65		
Data performance	Value <sup>b</sup>	Product Specification <sup>d</sup>			
Call rate	99.7%	> 99% avg.			
Reproducibility	99.99%	> 99.9%			
Log R deviation	0.11	< 0.30°			
Spacing					
Spacing (kb)	Mean	Median 90th% <sup>c</sup>			
	2.48	1.30 5.86			

- a. Estimate assumes 1 iScan System, 1 AutoLoader 2.x, 2 Tecan robots, and a 5-day work week.
- b. Values are derived from genotyping 326 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.
- d. Excludes Y chromosome markers for female samples.

#### Table 2: Marker information

Marker categories			No. of markers
RefSeq <sup>a</sup> genes			535,135
RefSeq genes +/- 10 kb			645,834
RefSeq exons			80,956
RefSeq promoter regions			30,627
ADME <sup>b</sup> genes			15,446
ADME genes +/- 10 kb			19,425
ADME exons			2160
MHC			16,246
COSMIC° genes			486,889
Genes in Gene Ontology <sup>d</sup>			130,264
Nonsense markerse			262
Missense markers <sup>e</sup>			22,370
Synonymous markers <sup>e</sup>			22,603
Silent markers <sup>f</sup>			35,712
Mitochondrial markersf			112
Indels <sup>f</sup>			39
Sex chromosomes <sup>f</sup>	X	Υ	PAR/homologous
	30,157	2221	1970

- RefSeq NCBI Reference Sequence Database.
  www.ncbi.nlm.nih.gov/refseq. Accessed September 2016.
- b. PharmaADME Gene List. www.pharmaadme.org. Accessed August 2014.
- Catalog of somatic mutations in cancer. cancer.sanger.uk/cosmic. Accessed July 2016.
- d. Gene Ontology Consortium. www.geneontology.org. Accessed July 2016.
- e. Compared against the University of California, Santa Cruz (UCSC) Genome Browser. genome.ucsd.edu. Accessed August 2014.
- NCBI Genome Reference Consortium, Version GRCh37. www.ncbi.nlm.nih.gov/grc/human. Accessed July 2016.

see www.illumina.com/company/legal.html. Pub. No. 370-2011-010-B QB #

 ${\color{blue} \textbf{Abbreviations: indel: insertion/deletion; PAR: pseudoautosomal region.} \\$ 

# Ordering information

Infinium OmniZhongHua-8 v1.4 Kit	Catalog no.
16 samples	20024684
48 samples	20024685
96 samples	20024686
384 samples	20024687

### Learn more

To learn more about the Infinium OmniZhongHua-8 v1.4 BeadChip and other Illumina genotyping products and services, visit www.illumina.com/genotyping

#### References

1. 1000 Genomes Project, www.1000genomes.org. Accessed April 2014.



© 2018 Illumina, Inc. All rights reserved. All trademarks are the property of Illumina, Inc. or their respective owners. For specific trademark information,