Infinium™ Omni2.5Exome-8 v1.4 BeadChip

Comprehensive coverage and functional exonic content for genotyping, GWAS, and CNV analysis.

Overview

The Infinium Omni2.5Exome-8 v1.4 BeadChip (Figure 1) delivers comprehensive coverage of common, rare, and exonic single nucleotide polymorphism (SNP) content from the 1000 Genomes Project,[1] providing maximum genomic information of diverse world populations (Table 1 and Table 2). With combined markers from the Infinium Omni2.5-8 and Infinium Exome-24 BeadChips, the Infinium Omni2.5Exome-8 v1.4 BeadChip is a powerful tool for next-generation genotyping and genome-wide association studies (GWAS) (Table 3). Using the proven HiScan™ or iScan™ System and integrated analysis software, the 8-sample Infinium Omni2.5Exome-8 v1.4 BeadChip offers high throughput, optimized tag SNPs, functional exonic content, and fully supported copy-number variation (CNV) analysis. Combined with convenient packaging and a streamlined PCR-free protocol, the Infinium Omni2.5Exome-8 v1.4 BeadChip provides a comprehensive DNA analysis solution.

Figure 1: The Infinium Omni2.5Exome-8 v1.4 BeadChip— The Infinium Omni2.5Exome-8 v1.4 BeadChip supports rapid, cost-effective GWAS studies with coverage of common, rare, and functional variants from the 1000 Genomes Project.

Table 1: Product information

Feature	Description		
Species	Human		
Total number of markers	2,619,927		
Number of samples per BeadChip	8 Samples		
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[d]**	
Call rate	99.8%	> 99% avg.	
Reproducibility	99.99%	> 99.9%	
Log R deviation	0.11	< 0.30[c]	
Spacing			
Spacing (kb)	Mean 1.11	Median 0.56	90th%[c] 2.63

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 458 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: LD $r^2 \geq 0.80$ from 1000G[a] at various MAF thresholds

1000G population[b]	LD coverage ($r^2 \geq 0.80$)		
	MAF ≥ 1%	MAF ≥ 2.5%	MAF ≥ 5%
AFR	0.54	0.64	0.69
AMR	0.74	0.82	0.86
EAS	0.79	0.86	0.89
EUR	0.79	0.86	0.89
SAS	0.75	0.83	0.87

a. Compared against Phase 3, version 5 of the 1000 Genomes Project (1000G). www.1000genomes.org. Accessed July 2016.
b. See www.1000genomes.org/category/frequently-asked-questions/population

Abbreviations: LD, linkage disequilibrium; MAF, minor allele frequency; AFR, African; AMR, Ad Mixed American; EAS, East Asian; EUR, European; SAS, South Asian.

Table 3: Marker information

Marker categories			No. of markers
RefSeq[a] genes			1,337,616
RefSeq exons			363,435
RefSeq promoter regions			57,433
ADME[b] genes			23,745
Extended MHC			22,480
COSMIC[c] genes			1,190,730
Genes in Gene Ontology[d]			309,642
Nonsense markers[e]			5506
Missense markers[e]			237,687
Synonymous markers[e]			41,606
Silent markers[f]			78,673
Mitochondrial markers[f]			400
Indels[f]			172
Sex chromosomes[f]	X 58,301	Y 2464	PAR/homologous 2710

a. RefSeq - NCBI Reference Sequence Database. www.ncbi.nlm.nih.gov/refseq. Accessed September 2016.
b. PharmaADME Gene List. www.pharmaadme.org. Accessed August 2014.
c. 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.
f. NCBI Genome Reference Consortium, Version GRCh37. www.ncbi.nlm.nih.gov/grc/human. Accessed July 2016.

Abbreviations: indel: insertion/deletion; PAR: pseudoautosomal region.

Ordering information

Infinium Omni2.5Exome-8 v1.4 Kit	Catalog no.
16 samples	20024552
48 samples	20024553
96 samples	20024554
384 samples	20024555

Learn more

To learn more about the Infinium Omni2.5Exome-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.

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