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Infinium® Omni5-4 v1.2 BeadChip

Powerful content combined with exceptional genome-wide coverage.

Overview

The Infinium Omni5-4 v1.2 BeadChip (Figure 1) delivers a comprehensive genome-wide backbone of content with high coverage across world superpopulations. This high-density array provides exceptional coverage of common, intermediate, and rare single nucleotide polymorphisms (SNPs) and harnesses tag SNPs selected from the International HapMap project[1] and 1000 Genomes Project (1000G)[2] (Tables 1–5). Using the proven iScan® System, integrated analysis software, and the Infinium LCG Assay, this 4-sample BeadChip offers optimized content for whole-genome genotyping and copy number variation (CNV) applications (Tables 6, 7). The Infinium Omni5-4 v1.2 BeadChip is compatible with the Infinium FFPE QC Kit and the Infinium HD FFPE DNA Restore Kit, enabling genotyping of formalin-fixed, paraffin-embedded (FFPE) samples. The Infinium Omni5-4 v1.2 Kit includes convenient kit packaging containing reagents and BeadChips for amplifying, fragmenting, hybridizing, labeling, and detecting genetic variants using the streamlined Infinium PCR-free protocol.

Figure 1: Infinium Omni5-4 v1.2 BeadChip—The Infinium Omni5-4 v1.2 BeadChip features over 4.3 million markers, providing exceptional coverage of common, intermediate, and rare targets.

Table 1: Product Information

Feature	Description		
Species	Human		
Total Number of Markers	4,327,108		
Number of Samples per BeadChip	4		
DNA Input Requirement	400 ng		
Assay Chemistry	Infinium LCG Quad		
Instrument Support	iScan or HiScan® System		
Sample Throughput[a]	~ 544 samples/week		
Scan Time per Sample	iScan System 38 min	HiScan System 15 min	
Data Performance	**Value[b]**	**Product Specification**	
Call Rate	99.9%	> 99% avg.	
Reproducibility	99.99%	> 99.9%	
Log R Deviation	0.13	< 0.30[c]	
Spacing			
Spacing (kb)	Mean 0.68	Median 0.36	90th%[c] 1.56

a. Estimate assumes 2 iScan Systems, 1 AutoLoader 2.x, 3 Tecan robots, and a 5-day work week.
b. Values are derived from genotyping 308 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 (Aggregate r^2) from 1000G[a]

Population[b]	Imputation Accuracy			
	MAF ≥ 5%	MAF ≥ 1%	MAF 1–5%	MAF 0.5–1%
AFR	0.98	0.96	0.94	0.85
AMR	0.98	0.96	0.94	0.89
EAS	0.97	0.94	0.83	0.59
EUR	0.98	0.97	0.94	0.83
SAS	0.98	0.95	0.88	0.69

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: MAF, minor allele frequency.

Table 3: Number of Markers Imputed at $r^2 \geq 0.80$ from 1000G[a]

Population[b]	Number of Markers Imputed at $r^2 \geq 0.80$ (% of Total Markers)			
	MAF ≥ 5%	MAF ≥ 1%	MAF 1–5%	MAF 0.5–1%
AFR	8.3 M (96%)	13.6 M (91%)	5.3 M (84%)	3.2 M (93%)
AMR	6.1 M (96%)	10.6 M (95%)	4.6 M (94%)	0.06 M (85%)
EAS	5.3 M (95%)	7.2 M (90%)	2.0 M (78%)	0.37 M (85%)
EUR	6.0 M (97%)	8.9 M (95%)	3.1 M (92%)	0.69 M (95%)
SAS	6.0 M (96%)	8.9 M (92%)	2.9 M (84%)	0.55 M (90%)

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.

Table 4: LD $r^2 \geq 0.80$ from 1000G[a]

Population[b]	LD Coverage ($r^2 \geq 0.80$)			
	MAF ≥ 5%	MAF ≥ 1%	MAF 1–5%	MAF 0.5–1%
AFR	0.74	0.60	0.41	0.17
AMR	0.90	0.82	0.70	0.57
EAS	0.91	0.82	0.55	0.25
EUR	0.93	0.89	0.80	0.57
SAS	0.91	0.81	0.60	0.34

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.

Table 5: LD Mean r^2 from 1000G[a]

Population[b]	LD Coverage (Mean r^2)			
	MAF ≥ 5%	MAF ≥ 1%	MAF 1–5%	MAF 0.5–1%
AFR	0.85	0.74	0.59	0.31
AMR	0.93	0.89	0.81	0.71
EAS	0.94	0.85	0.61	0.30
EUR	0.95	0.93	0.87	0.69
SAS	0.94	0.86	0.69	0.42

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

Table 6: Marker Information

Marker Categories			Number of Markers[a]
Exonic Markers[a]			329,595
Intronic Markers[a]			1,856,136
Nonsense Markers[b]			1008
Missense Markers[b]			75,197
Synonymous Markers[b]			67,324
Silent Markers[b]			185,859
Mitochondrial Markers[c]			209
Indels[c]			4638
Sex Chromosomes[c]	X 113,536	Y 2445	PAR/Homologous 5263

a. RefSeq - NCBI Reference Sequence Database. www.ncbi.nlm.nih.gov/refseq. Accessed September 2016.
b. Compared against the University of California, Santa Cruz (UCSC) Genome Browser. genome.ucsc.edu. Accessed July 2016.
c. NCBI Genome Reference Consortium.Version GRCh37, released February 27, 2009. www.ncbi.nlm.nih.gov/grc/human. Accessed July 2016.

Abbreviations: PAR, pseudoautosomal region.

Table 7: High-Value Content

Content	Number of Markers	Research Application/Note
ADME Core and Extended Genes[3]	32,451	Drug metabolism and excretion
ADME Core and Extended Genes[3] +/- 10 kb	41,773	Drug metabolism and excretion (plus regulatory regions)
APOE[4]	12	Cardiovascular disease, Alzheimer's disease, immunoregulation, and cognition
Blood Phenotype Genes[5]	2962	Blood phenotypes
COSMIC[6] Genes	1,855,606	Somatic mutations in cancer
GO[7] CVS Genes	481,329	Cardiovascular conditions
Database of Genomic Variants[8]	3,351,570	Genomic structural variation
eQTLs[9]	13,319	Genomic loci regulating mRNA expression levels
Fingerprint SNPs[10]	598	Human identification
HLA Genes[4]	2443	Disease defense, transplant rejection, and autoimmune disorders
Extended MHC*[11]	52,335	Disease defense, transplant rejection, and autoimmune disorders
KIR Genes[4]	125	Autoimmune disorders and disease defense
Neanderthal SNPs[12]	5143	Neanderthal ancestry and human population migration
NHGRI GWAS Catalog[13]	9037	Markers from published genome-wide association studies
RefSeq[14] 3' UTRs	159,792	3' untranslated regions of known genes
RefSeq 5' UTRs	44,451	5' untranslated regions of known genes
RefSeq All UTRs	197,594	All untranslated regions of known genes
RefSeq	2,107,849	All known genes
RefSeq +/- 10 kb	2,485,221	All known genes plus regulatory regions
RefSeq Promoters	94,355	2 kb upstream of all known genes to include promoter regions
RefSeq Splice Regions	5320	Variants at splice sites in all known genes

*Extended MHC is a ~ 8 Mb region.

Abbreviations: ADME, absorption, distribution, metabolism, and excretion; APOE, apolipoprotein E; COSMIC, catalog of somatic mutations in cancer; GO CVS, gene ontology annotation of the cardiovascular system; eQTL, expression quantitative trait loci; HLA, human leukocyte antigen; KIR, killer cell immunoglobin-like receptor; MHC, major histocompatibility complex; NHGRI, national human genome research institute; GWAS, genome-wide association study; UTR, untranslated region; RefSeq, reference sequence.

Ordering Information

Infinium Omni5-4 v1.2 BeadChip Kit	Catalog No.
16 Samples	20005150
48 Samples	20005151
96 Samples	20005152
384 Samples	20005153

Learn More

To learn more about the Infinium Omni5-4 v1.2 BeadChip and other Illumina genotyping products and services, visit www.illumina.com/applications/genotyping.html.

References

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14. NCBI Reference Sequence Database. www.ncbi.nlm.nih.gov/refseq. Accessed July 2016.

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