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Infinium® Omni5Exome-4 v1.3 BeadChip

Powerful content combined with exceptional genome-wide coverage.

Overview

The Infinium Omni5Exome-4 v1.3 BeadChip (Figure 1) delivers a comprehensive genome-wide backbone combined with putative functional exonic variants selected from over 12,000 individual human exome and whole-genome sequences. This high-density array provides exceptional coverage of common, intermediate, and rare single nucleotide polymorphisms (SNPs) and harnesses powerful tag SNPs selected from the International HapMap[1] and 1000 Genomes Projects (1000G)[2] (Tables 1–5). Using the proven iScan® System, integrated analysis software, and Infinium LCG Assay, this 4-sample BeadChip provides optimized content for whole-genome genotyping, exonic content, and copy number variation (CNV) for various research applications (Tables 6, 7).

The Infinium Omni5Exome-4 v1.3 Kit is compatible with the Infinium FFPE QC Kit and Infinium HD FFPE DNA Restore Kit, enabling genotyping of formalin-fixed, paraffin-embedded (FFPE) samples. The Infinium Omni5Exome-4 v1.3 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.

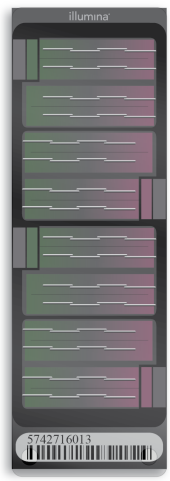

Figure 1: Infinium Omni5Exome-4 v1.3 BeadChip—The Infinium Omni5Exome-4 v1.3 BeadChip features over 4.5 million markers, providing exceptional coverage of common, intermediate, and rare targets.

Table 1: Product Information

Feature	Description		
Species	Human		
Total Number of Markers	4,559,465		
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	0.14	< 0.30[c]	
Spacing			
Spacing (kb)	Mean 0.64	Median 0.33	90th%[c] 1.50

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	0.60
EUR	0.98	0.97	0.94	0.84
SAS	0.98	0.95	0.89	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 ≥ 0.80 from 1000G[a]

Population[b]	Number of Markers Imputed at r^2 ≥ 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%)	5.4 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	0.42	0.18
AMR	0.90	0.83	0.70	0.58
EAS	0.91	0.82	0.55	0.25
EUR	0.93	0.89	0.81	0.57
SAS	0.91	0.81	0.61	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	0.60	0.32
AMR	0.93	0.89	0.81	0.72
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	0.43

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 6: Marker Information

Marker Categories			Number of Markers[a]
Exonic Markers[a]			544,817
Intronic Markers[a]			1,886,632
Nonsense Markers[b]			6040
Missense Markers[b]			275,657
Synonymous Markers[b]			76,120
Silent Markers[b]			186,900
Mitochondrial Markers[b]			408
Indels[c]			4766
Sex Chromosomes[c]	X 118,204	Y 2574	PAR/Homologous 5364

a. RefSeq - NCBI Reference Sequence Database. www.ncbi.nlm.nih.gov/refseq. Accessed July 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 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]	36,557	Drug metabolism and excretion
ADME Core and Extended Genes +/- 10 kb	46,461	Drug metabolism and excretion (plus regulatory regions)
APOE[4]	14	Cardiovascular disease, Alzheimer's disease, immunoregulation, and cognition
Blood Phenotype Genes[5]	3466	Blood phenotypes
COSMIC[6] Genes	2,070,366	Somatic mutations in cancer
GO[7] CVS Genes	537,409	Cardiovascular conditions
Database of Genomic Variants[8]	3,536,075	Genomic structural variation
eQTLs[9]	14,002	Genomic loci regulating mRNA expression levels
Fingerprint SNPs[10]	873	Human identification
HLA Genes[4]	2675	Disease defense, transplant rejection, and autoimmune disorders
Extended MHC*[11]	56,387	Disease defense, transplant rejection, and autoimmune disorders
KIR Genes[4]	143	Autoimmune disorders and disease defense
Neanderthal SNPs[12]	5390	Neanderthal ancestry and human population migration
NHGRI GWAS Catalog[13]	13,582	Markers from published genome-wide association studies
RefSeq[14] 3' UTRs	170,415	3' untranslated regions of known genes
RefSeq 5' UTRs	55,162	5' untranslated regions of known genes
RefSeq All UTRs	218,360	All untranslated regions of known genes
RefSeq	2,331,552	All known genes
RefSeq +/- 10 kb	2,710,974	All known genes plus regulatory regions
RefSeq Promoters	100,332	2 kb upstream of all known genes to include promoter regions
RefSeq Splice Regions	7348	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 immunoglobulin-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 Omni5Exome-4 v1.3 Kit	Catalog No.
16 Samples	20005140
48 Samples	20005141
96 Samples	20005142
384 Samples	20005143

Learn More

To learn more about the Infinium Omni5Exome-4 v1.3 BeadChip and other Illumina genotyping products and services, visit www.illumina.com/applications/genotyping.html.

References

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