

Infinium™ OmniExpressExome-8 v1.6 BeadChip

Discover novel trait and disease associations with functional exonic content.

Overview

The Infinium OmniExpressExome-8 v1.6 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 OmniExpressExome-8 v1.6 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 OmniExpressExome-8 v1.6 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 OmniExpressExome-8 v1.6 BeadChip provides a comprehensive DNA analysis solution.



Figure 1: The Infinium OmniExpressExome-8 v1.6 BeadChip—The Infinium OmniExpressExome-8 v1.6 BeadChip provides excellent coverage of common variants and functional exonic content from the 1000 Genomes Project.

Table 1: Product information

Feature	Description		
Species	Human		
Total number of markers	962,215		
Capacity for custom bead types	30,000		
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 ^a	~ 960 samples/week		
Scan time per sample	iScan System	HiScan System	
	4.6 min	3.5 min	
Data performance	Value ^b	Product Specification ^d	
Call rate	99.88%	> 99% avg.	
Reproducibility	99.99%	> 99.9%	
Log R deviation	0.09	< 0.30 ^c	
Spacing			
Spacing (kb)	Mean	Median	90th% ^e
	3.02	1.36	7.57

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 331 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 $\geq 1\%$	MAF $\geq 2.5\%$	MAF $\geq 5\%$
AFR	0.28	0.35	0.43
AMR	0.54	0.65	0.72
EAS	0.67	0.74	0.78
EUR	0.63	0.71	0.77
SAS	0.58	0.68	0.73

- 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	566,140		
RefSeq genes +/- 10 kb	635,492		
RefSeq exons	275,744		
RefSeq promoter regions	28,880		
ADME ^b genes	17,807		
ADME genes +/- 10 kb	21,403		
ADME exons	6149		
MHC	11,513		
COSMIC ^c genes	527,784		
Genes in Gene Ontology ^d	141,808		
Nonsense markers ^e	5323		
Missense markers ^e	224,927		
Synonymous markers ^e	20,259		
Silent markers ^f	25,120		
Mitochondrial markers ^f	208		
Indels ^f	140		
Sex chromosomes ^f	X	Y	PAR/homologous
	22,783	1448	819

- 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.ucsc.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 OmniExpressExome-8 v1.6 Kit	Catalog no.
16 samples	20024676
48 samples	20024677
96 samples	20024678
384 samples	20024679
Infinium OmniExpressExome-8+ v1.6 Kit ^a	Catalog no.
16 samples	20024680
48 samples	20024681
96 samples	20024682
384 samples	20024683
a. Enabled for additional custom content.	b. Enabled for additional custom content.

Learn more

To learn more about the Infinium OmniExpressExome-8 v1.6 BeadChip and other Illumina genotyping products and services, visit www.illumina.com/genotyping

References

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