

HumanExome BeadChips

Access > 250,000 exonic variants to uncover biologically significant associations.

Overview

Illumina Human Exome BeadChips deliver unparalleled coverage of putative functional exonic variants selected from over 12,000 individual exome and whole-genome sequences. Markers were identified through a close collaboration with leading geneticists with the goal of developing an extensive catalog of exome variants. The exonic content consists of > 250,000 markers representing diverse populations—including European, African, Chinese, and Hispanic individuals—and a range of common conditions, such as type 2 diabetes, cancer, metabolic, and psychiatric disorders.

Content is available on three individual microarrays to support a variety of study types aimed at uncovering functionally relevant disease associations. In addition, researchers can include additional custom markers on any of the BeadChips to target specific regions of the genome with higher density, focus on populations of interest, or incorporate selected disease-related variants.

Infinium® HumanExome BeadChip

The Infinium HumanExome BeadChip provides the exonic content on a 12-sample array format. It delivers focused coverage of exonic regions, but does not include coverage outside of coding regions. Researchers can use this array to obtain new insights from previously genotyped cohorts, or run new studies focused on identifying functionally relevant associations.

HumanOmniExpressExome BeadChip

Along with the > 250,000 exonic markers, the HumanOmniExpressExome BeadChip features > 700,000 genome-wide markers that provide coverage of variants at > 5% minor allele frequency. The array maximizes coverage of coding variants within genes for genome-wide association studies (GWAS) focused on common variation.

HumanOmni5Exome BeadChip

The four-sample HumanOmni5Exome BeadChip features > 4.3 million tag SNPs in addition to the > 250,000 exonic markers. This array delivers extensive coverage of common and rare variants (> 1% MAF) across the human genome, along with unprecedented coverage of coding variants within genes, providing the highest likelihood for obtaining new insight to potential disease pathways.

Summary

A large set of exonic markers identified by leading genetic researchers is available on three unique microarrays to support a variety of study objectives. Researchers can use the HumanExome BeadChip to obtain new insights for previously genotyped cohorts. The HumanOmniExpressExome and HumanOmni5Exome BeadChips can be used to maximize exonic coverage for new genotyping studies. Custom markers can be added to all three beadchips to tailor studies for specific populations or phenotypes of interest.

Human Exome Content Specifications*

Marker Categories	Values
Total markers	> 250,000
SNPs within RefSeq genes	258,304
SNPs within 10 kb of RefSeq genes	260,575
Number of unique refseq entries covered by at least one probe	22,754
Average number of probes per refseq entry	15
SNPs within 333 known ADME genes	5,572
SNPs within 10 kb of 333 known ADME genes	6,136
Nonsynonymous SNPs (NCBI)	232,125
SNPs in coding regions (includes UTR)	246,139
SNPs in Splice Sites	12,459
SNPs in exon splice regions	55,376
SNPs in uRNA target regions	14,116
SNPs in promoter regions	7,012
SNPs in ECR regions	128,039
SNPs in extended MHC region	5,158
GWAS Tag Markers†	5,325
HLA Tags	2,459
Ancestry informative markers	3,468
Identity by descent markers	3,369
X / Y / mitochondrial	470 / 180 / 245
Indels	180
Variation Captured‡ (r² > 0.8)	Fraction
MAF > 5.0%	0.10
MAF > 2.5%	0.096
MAF > 1.0%	0.088

* This product is not yet available. Pre-release specifications are subject to change.

† Disease-associated tag markers identified from recent GWAS.

‡ The exome content provides focused coverage of exonic regions and does not provide broad genomic coverage outside of coding regions. To see specifications for genome-wide content on the HumanOmniExpressExome or HumanOmni5Exome BeadChips, please see the HumanOmniExpress or HumanOmni5 BeadChip data sheets, respectively^{1,2}.

Ordering Information

Product	Catalog No.
Infinium HumanExome BeadChips	
Infinium HumanExome DNA Analysis BeadChip Kit (48 Samples)	WG-353-1001
Infinium HumanExome DNA Analysis BeadChip Kit (288 Samples)	WG-353-1002
Infinium HumanExome DNA Analysis BeadChip Kit (1,152 Samples)	WG-353-1003
Infinium HumanExome+ DNA Analysis BeadChip Kit (48 Samples)	WG-353-1005
Infinium HumanExome+ DNA Analysis BeadChip Kit (288 Samples)	WG-353-1006
Infinium HumanExome+ DNA Analysis BeadChip Kit (1,152 Samples)	WG-353-1007
Infinium HumanExome+ Custom BeadChip	WG-353-1009
Infinium HumanOmniExpressExome BeadChips	
Infinium OmniExpressExome DNA Analysis Kit (16 reactions)	WG-350-2206
Infinium OmniExpressExome DNA Analysis Kit (48 reactions)	WG-350-2207
Infinium OmniExpressExome DNA Analysis Kit (96 reactions)	WG-350-2208
Infinium OmniExpressExome DNA Analysis Kit (384 reactions)	WG-350-2209
Infinium OmniExpressExome+ DNA Analysis Kit (16 samples)	WG-353-2210
Infinium OmniExpressExome+ DNA Analysis Kit (48 samples)	WG-353-2211
Infinium OmniExpressExome+ DNA Analysis Kit (96 samples)	WG-353-2212
Infinium OmniExpressExome+ DNA Analysis Kit (384 samples)	WG-353-2213
OmniExpressExome+ Custom BeadChip	WG-353-1010

Ordering Information, Continued

Product	Catalog No.
Infinium HumanOmni5Exome BeadChips	
Infinium HumanOmni5Exome DNA Analysis Kit (16 sample)	WG-311-5010
Infinium HumanOmni5Exome DNA Analysis Kit (48 sample)	WG-311-5011
Infinium HumanOmni5Exome DNA Analysis Kit (96 sample)	WG-311-5012
Infinium HumanOmni5Exome DNA Analysis Kit (384 sample)	WG-311-5013
Infinium HumanOmni5Exome+ DNA Analysis Kit (16 sample)	WG-311-5014
Infinium HumanOmni5Exome+ DNA Analysis Kit (48 sample)	WG-311-5015
Infinium HumanOmni5Exome+ DNA Analysis Kit (96 sample)	WG-311-5016
Infinium HumanOmni5Exome+ DNA Analysis Kit (384 sample)	WG-311-5017
Infinium HumanOmni5Exome+ Custom BeadChip	WG-311-5018

References

- HumanOmniExpress BeadChip data sheet: http://www.illumina.com/documents/products/datasheets/datasheet_human_omni_express.pdf
- HumanOmni5 BeadChip data sheet: http://www.illumina.com/documents/products/datasheets/datasheet_omni5.pdf

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AGAATGATAACAGTAAACACACTTCTGTAACTTAAGATTACTTGTCCACTGATTCAACGTACCGTAAACGAAAGCGTATCAATTGAGACTAAATATTAACGTACCCATTAAAGAGCTACCGTTCCTTGTAACTTAAGATTACTTGTATCCACTGATTCAAC
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