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HumanExome BeadChips

Access > 240,000 exonic variants to uncover biologically significant associations.

Overview

Illumina HumanExome 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 > 240,000 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 four 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 Illumina Exome 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, and can accommodate an additional 200K custom markers. Researchers can use this array to obtain new insights from previously genotyped cohorts, or run new studies focused on identifying functionally relevant associations.

Combine Illumina Omni Microarrays and HumanExome Content

On a single array, researchers can combine the novel, functional exonic content of the HumanExome BeadChip with the pre-optimized tag SNPs of any Illumina Omni microarray. These combined BeadChips allow researchers to gain insight into whole-genome variation while maximizing coverage of functional exonic SNPs.

HumanOmniExpressExome BeadChip

Along with the > 240,000 functional exonic markers, the 8-sample HumanOmniExpressExome BeadChip features > 700,000 genome-wide markers that provide coverage of common variants at > 5% minor allele frequency (MAF). Researchers can also add up to 30,000 custom markers.

HumanOmni2.5Exome BeadChip

The 8-sample HumanOmni2.5Exome BeadChip combines > 2.5 million tag SNPs from the HapMap and 1000 Genomes Project (> 2.5% MAF) with > 240,000 exonic markers.

HumanExome BeadChip Content Specifications

Marker Categories	Values
Total markers	> 240,000
Number of unique RefSeq entries covered by at least one probe	> 20,000
Nonsynonymous SNPs (NCBI)	219,621
SNPs in splice sites	10,675
Stop variants	5,637
SNPs in promoter regions	7,012
SNPs in extended MHC region	5,158
GWAS tag markers*	4,761
HLA tags	2,061
Ancestry informative markers	3,468
Identity by descent markers	3,369
X / Y / mitochondrial	470 / 101 / 177
Indels	180
Variation Captured† (r^2 > 0.8)	**Fraction**
MAF > 5.0%	0.10
MAF > 2.5%	0.096
MAF > 1.0%	0.088

*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].

HumanOmni5Exome BeadChip

The 4-sample HumanOmni5Exome BeadChip features > 4.3 million tag SNPs in addition to > 240,000 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. The Omni5Exome BeadChip can be further customized with the addition of up to 200,000 custom markers.

* Product names denoted with a "+" represent products that can accept custom beadtypes.

Ordering Information

Product	Catalog No.
Infinium HumanExome v1.1 BeadChips	
Infinium HumanExome v1.1 DNA Analysis BeadChip Kit (48 Samples)	WG-353-1101
Infinium HumanExome v1.1 DNA Analysis BeadChip Kit (288 Samples)	WG-353-1102
Infinium HumanExome v1.1 DNA Analysis BeadChip Kit (1,152 Samples)	WG-353-1103
Infinium HumanExome+ v1.1 DNA Analysis BeadChip Kit (48 Samples)	WG-353-1105
Infinium HumanExome+ v1.1 DNA Analysis BeadChip Kit (288 Samples)	WG-353-1106
Infinium HumanExome+ v1.1 DNA Analysis BeadChip Kit (1,152 Samples)	WG-353-1107
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

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 HumanOmni2.5Exome BeadChips	
Infinium HumanOmni2.5Exome DNA Analysis Kit (16 sample)	WG-311-2541
Infinium HumanOmni2.5Exome DNA Analysis Kit (48 sample)	WG-311-2542
Infinium HumanOmni2.5Exome DNA Analysis Kit (96 sample)	WG-311-2543
Infinium HumanOmni2.5Exome DNA Analysis Kit (384 sample)	WG-311-2544

Summary

A large set of exonic markers identified by leading genetic researchers is available on four 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, HumanOmni2.5Exome, and HumanOmni5Exome BeadChips can be used to maximize exonic coverage for new genotyping studies. Custom markers can be added to Exome BeadChips to tailor studies for specific populations or phenotypes of interest.

For more information, visit www.illumina.com/exome.

References

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

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Pub. No. 370-2011-020 Current as of 16 March 2012

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