The Omni Family of Microarrays

Omni microarrays are the path to the future of genome-wide association studies, soon offering up to five million markers per sample and extensive coverage of new variants identified by the 1,000 Genomes Project.

INTRODUCTION

Genome-wide association studies (GWAS) have identified hundreds of common genetic variants associated with human diseases and provided valuable insights into the genetic architecture behind many diseases. Building on this success, the 1,000 Genomes Project (1KGP) is expected to dramatically expand the catalog of human genetic variants for the next generation of GWAS, as the project will seek to identify nearly all variants that exist at any appreciable frequency.

THE 2010 GWAS PRODUCT ROADMAP

In preparation for the new information that will emerge from 1KGP, Illumina has outlined a product roadmap for the Omni family of microarrays that will offer researchers quick access to the newly identified markers as they are released into the public domain. Soon providing up to five million markers per sample and broad coverage of rare variants, Omni microarrays are the path to the future of GWAS.

Illumina is developing these arrays in collaboration with leading researchers from 1KGP to select the highest-value markers. New Omni arrays will be released incrementally, and will include coverage of the latest available variants. These arrays will allow Omni customers to add supplemental content to their ongoing GWAS and progressively build up to the full five million variants.

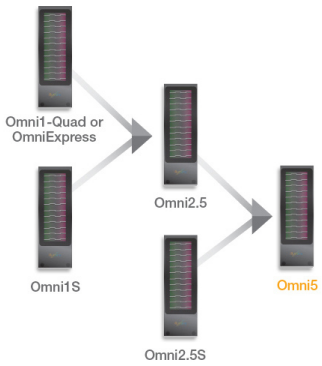
As shown in Figure 1, a customer carrying out a GWAS using the HumanOmni1-Quad (Omni1-Quad) or HumanOmniExpress (OmniExpress) BeadChip will have access to more than 1 million additional markers when the Omni1S BeadChip is released, increasing their total data set to as high as 2.5 million markers. These 2.5 million markers will also be available on the subsequent Omni2.5 BeadChip. The additional 1 million-plus markers on the Omni1S will include novel data from the first releases of 1KGP, covering minor allele frequencies as low as ~2.5%.

The Omni2.5S BeadChip will provide an additional 2.5 million markers to increase the total data set to as high as five million markers. These additional 2.5 million markers will include novel data from 1KGP, covering minor allele frequencies as low as ~1%. The complete five million marker set will be released on the Omni5 BeadChip.

GENOME-WIDE COVERAGE

Omni microarrays offer unparalleled genomic coverage using intelligently selected tag SNPs that maximize the likelihood of finding true associations for a given phenotype. The power of a tag SNP approach stems from the inherent correlation among markers that form haplotype blocks, allowing one highly correlated marker to serve as a proxy for a number of others across the genome. This approach allows for the broadest selection of maximally informative markers, resulting in the best genome-wide coverage of both common and rare variants.

FIGURE 1: OMNI MICROARRAYS



Starting with the Omni1-Quad or OmniExpress BeadChip, researchers can progressively build up to the full five million variants.

1KGP data will enable researchers to explore the role of rare variation in human traits and diseases. Omni microarrays will feature comprehensive coverage of newly discovered rare variants as they are made available to the scientific community, enabling novel discoveries and a more complete understanding of the role of variation in human health and diseases.

Structural variation, including copy number variants (CNVs) and copy neutral variants (inversions and translocations), is a significant contributor to the genetic basis of human disease. Dense genome-wide coverage on Omni microarrays, coupled with the sensitive Infinium® HD assay, offer researchers a powerful tool for structural variation analysis.

TABLE 1: THE OMNI FAMILY OF MICROARRAYS

	OMNI-EXPRESS	OMNI1-QUAD	OMNI1S	OMNI2.5	OMNI2.5S	OMNI5
Number of Loci	> 700K	> 1M	> 1M	~2.5M	~2.5M	~5M
Assay	Infinium HD	Infinium HD	Infinium HD	Infinium HD	Infinium HD	Infinium HD
Content Source	HapMap Phases 1–3	HapMap Phases 1–3 and 1KGP	1KGP	HapMap Phases 1–3 and 1KGP	1KGP	HapMap Phases 1–3 and 1KGP
MAF Range	> 5%	> 5%	> 2.5%	> 2.5%	> 1%	> 1%
Compatibility	Base tag SNP content	Base tag SNP content and high-value additions such as CNV regions	New 1KGP content (MAF > 2.5%)	All content from Omni1 and Omni1S	New rarer 1KGP content (MAF > 1%)	All content from Omni2.5 and Omni2.5S
CNV Capabilities	Yes	Yes	Yes	Yes	Yes	Yes
Analysis Software	GenomeStudio	GenomeStudio	GenomeStudio	GenomeStudio	GenomeStudio	GenomeStudio
Scanner	iScan (5 min/sample)	iScan (13 min/sample) or BeadArray™ Reader	iScan	iScan	iScan	iScan
Call Rate	> 99%*	> 99%	> 99%*	> 99%*	> 99%*	> 99%*
Reproducibility	> 99.9%*	> 99.9%	> 99.9%*	> 99.9%*	> 99.9%*	> 99.9%*

* These averages are estimated before final Gentrain; final number will vary slightly

The Infinium HD Assay delivers very high signal-to-noise ratios and low overall noise levels, which are ideal for precise structural analysis. For both genotype calling and structural variation analysis, Omni microarrays provide a single solution for any course of genetic research.

POWERFUL GENOTYPING PLATFORM

The arrays are delivered on the proven Infinium genotyping platform, which has provided researchers the fastest time to publication, and catalyzed many revolutionary discoveries in disease research. The assay consistently produces industry-leading call rates (> 99%) and the highest reproducibility (> 99.9%) available (*Table 1*).

For large-scale projects, Illumina-supported LIMS and robotic systems for sample tracking are available to optimize project management and maximize sample throughput.

SIMPLIFIED DATA ANALYSIS

Illumina's GenomeStudio® software offers the ability to jointly analyze SNPs and CNV probes for downstream analysis, and includes links to numerous third-party plug-ins for both genotyping and CNV analysis.

SUMMARY

Omni microarrays will soon allow access to over five million variants per sample with extensive coverage of new variants identified by 1KGP. Backed by the powerful Infinium genotyping platform, Omni microarrays will drive the next generation of discoveries for GWAS.

ADDITIONAL INFORMATION

For additional information on the Omni family of microarrays, visit www.illumina.com/gwas.

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